MEDICAL GENETICS

ELEMENTS OF MORFOLOGY

2015

Index

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EDITORIAL COMMENT

AMERICAN JOURNAL OF medical genetics PART A

Editorial Comment: Editor's Foreword to a Special Issue "Elements of Morphology: Standard Terminology"

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This Special Issue of *American Journal of Medical Genetics* comprises an Introduction and six articles on "The Elements of Morphology". I would suggest to our readership that these pieces represent a timely and substantial contribution to the field of medical genetics. These six articles provide recommendations for the description and definition of human phenotypic variations in the same way that the International Standing Committee on Human Cytogenetic Nomenclature accomplished this for human cytogenetics [ISCN, 2005] and the Nomenclature Working Group proposed the description of human sequence variations [den Dunnen and Antonarakis, 2001].

Taking up somewhat where an international working group on malformations left off in the 1980s [Spranger et al., 1982], the working group that authored the articles in this issue set out 4 years ago to develop consensus definitions and nomenclature for variations of phenotype. These articles are the direct result of that work.

The group consisted of over 30 clinical geneticists from the United States, Canada, Europe, and Australia. The process of developing these consensus definitions was time consuming and stringent: recruitment of committee chairs, formation of six working committees, drafting of assigned definitions, attendance at two meetings, hundreds of emails, perusal of over 400 figures, 22 months of revisions, and five web-based conference calls.

As Editor of the *Journal*, I would like to point out that the articles published herein are the final product of a unique peer-review process: All of the participants provided input for the wording of the definitions for each section. The committee chairs reviewed and critiqued the proposals of the other reports. The almost-final versions represented the consensus recommendations of this ongoing process of reviewing and editing. As a last step, four of the six committee chairs edited and reviewed the final manuscripts of each region (Head/Face, Periorbital, Ear, Nose/Philtrum, Lips/Mouth, and Hands/Feet) for internal consistency and style.

The *American Journal of Medical Genetics* is pleased to publish these articles, which will be indefinitely available on the Journal's web page at http://www.wileyinterscience.com/journal/ajmg.

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Am J Med Genet Part A 149A:1.

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RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Elements of Morphology: Introduction

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An international group of clinicians working in the field of dysmorphology has initiated the standardization of terms used to describe human morphology. The goals are to standardize these terms and reach consensus regarding their definitions. In this way, we will increase the utility of descriptions of the human phenotype and facilitate reliable comparisons of findings among patients. Discussions with other workers in dysmorphology and related fields, such as developmental biology and molecular genetics, will become more precise. Here we describe the general background of the project and the various issues we have tried to take into account in defining the terms. Published 2009 Wiley-Liss, Inc.†

Key words: nomenclature; definitions; morphology; dysmorphology; birth defects; malformations; minor anomalies; common variants

How to Cite this Article:
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INTRODUCTION

This issue of the journal contains six articles that describe the initial results of a project intended to develop accurate and clear definitions of terms for the craniofacies in general, the major components of the face, and the hands and feet [Allanson et al., 2009; Biesecker et al., 2009; Carey et al., 2009; Hall et al., 2009; Hennekam et al., 2009; Hunter et al., 2009]. These articles are the result of a significant amount of planning, organization, negotiation, review, and writing, while, at the same time, they are but a start.

Dysmorphology evolved from a small nucleus of clinicians in the 1950s into a recognized and widely practiced discipline, and more recently has incorporated translational research into developmental biology, molecular genetics, and metabolic medicine. The terms that clinicians use to describe a body part have gradually evolved in a haphazard and uncoordinated manner, and have not been critically reviewed. Clinicians and researchers have always made comparisons among patients and syndromes, and in the last decade it has become increasingly possible and necessary to use clinical data for studies of etiology and pathogenesis, epidemiology, the isolation of causative gene mutations, and for evaluation of interventions. Therefore, we need to have uniform and internationally accepted terms to describe the human phenotype.

ORGANIZATION OF THE PROJECT

Five years ago, after many informal discussions, we brought together an international group of clinicians working in the field of dysmorphology with the goal of standardizing this nomenclature. The participants were chosen for their knowledge of morphology, syndromes, and skills in syndrome delineation. Because we anticipated that the group would need to gather several times and we recognized financial constraints, we restricted the group to 34 individuals and had to accept that many excellent colleagues could not be invited to join the project.

Our original goal was to review, revise if necessary, and delineate definitions for all of the terms used in the London Dysmorphology Databases (n = 683). However, recognizing the enormity of the task, we chose to begin the project with the cranium, the face and its features, and the hands and feet—the parts of the body that are often used in dysmorphology to describe patients and delineate syndromes.

We set out to accomplish this work in six teams, each focused on a particular area of the human body (Table I). While much of our

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TABLE I. Nomenclature Subgroups and Their Members (the Chair of Each Group Is in Italics)

Head and Face: *Judith E. Allanson*, Chris Cunniff, Gene Hoyme, Julie McGaughran, Max Muenke, Giovanni Neri
Hands and Feet: *Leslie G. Biesecker*, Jon M. Aase, Carol Clericuzio, Fiorella Gurrieri, Karen Temple, Helga Toriello
Mouth: *John C. Carey*, M. Michael Cohen Jr, Cynthia Curry, Koen Devriendt, Lewis Holmes, Alain Verloes
Ear: *Alasdair G. W. Hunter*, Jaime Frias, Gabrielle Gillessen-Kaesbach, Ken Jones, Helen Hughes, Louise Wilson
Periorbital structures: *Bryan D. Hall*, John M. Graham Jr, Suzanne B. Cassidy, John M. Opitz
Nose and Philtrum: *Raoul C. M. Hennekam*, Valerie Cormier-Daire, Judith G. Hall, Karoly Méhes, Michael Patton, Roger Stevenson

work has been carried out in small groups using electronic communication, we have met as a large group on two occasions: in Bethesda in December 2005 (host: Leslie G. Biesecker) and Rome in November 2006 (hosts: Giovanni Neri and Fiorella Gurrieri) (Fig. 1). These face-to-face meetings were invaluable in expediting the process and allowing discussion of principles and initial points of disagreement, of which there were many.

ATTRIBUTES OF THE TERMS

For each feature we provide a preferred term and use a standard format to provide a definition and description of how to observe and measure (where possible) the feature. For many of the terms, both subjective and objective definitions of terms are provided. When an objective assessment can be made, this is always preferable to the subjective. A few terms have multiple alternative objective definitions. Typically, this reflects multiple sets of norms for a quantitative trait and, in such cases, each definition is equally valid.

Our aim is to formalize, unify, and standardize the approach to clinical assessment in the hope that it will become generally accepted and applied. For each term we have added relevant comments and synonyms, and indicated terms that should no longer be used. Most clinicians have at least a few terms of which they are inordinately fond. In spite of these personal biases, our goal is to make a universal language to describe morphology. We have avoided terms that indicate pathogenesis or an active process; we aspired to simply describe what can be observed.

We recognize that a number of terms in common usage may be considered pejorative in some or all cultures. We have created alternative descriptors in these cases, even if they are not in current common usage. The definitions are intended to be applied broadly, thus we avoided obscure terms, flowery language, etc. We have tried to apply a standard format that would be readily understandable by a medical student who has completed training in physical diagnosis.

In addition to standardized terms and definitions, we aimed to provide clear illustrations of each feature. We have used our joint

FIG. 1. Nomenclature group members present in November 2006 in Rome (Suzanne Cassidy was also present but could not be depicted on this picture). From left to the right are visible (first row) Helga Toriello, Cynthia Curry, Julie McGaughran, M Michael Cohen Jr, Louise Wilson, John Carey, Fiorella Gurrieri, Valerie Cormier-Daire, (second row) Jaime Frias, Giovanni Neri, Judith Allanson, Judith Hall, Karen Temple, Alain Verloes, (third row) Michael Patton, Alasdair Hunter, Gene Hoyme, Helen Hughes, John M Graham Jr, (fourth row) Roger Stevenson, Leslie Biesecker, Koen Devriendt, Bryan Hall, and Raoul Hennekam.

collection of pictures and the extensive Robert J. Gorlin slide collection which is the series of digitalized slides that Prof Gorlin gathered during his long career, available to one of us (RCMH). It is important that readers recognize that a given photograph may include multiple findings. Where possible, photographs have been cropped to avoid showing other signs and to emphasize one feature. We tried to show variable expression of the features, both mild and severe.

The terms are intended for clinical evaluation of the individual using observation, palpation, or simple morphometric techniques (such as measuring of inner canthal distance). We provide basic general principles for measuring each of the body parts, and indicate in the definitions if a reliable measurement is possible and if norms are available. However, extensive description of morphometric techniques is outside the scope of this effort and readers should consult authoritative sources for this information (Farkas L. *Anthropometry of the Head and Face in Medicine*. Elsevier, 1981; and Hall JG, Allanson JE, Gripp KW, Slavotinek AM. *Handbook of Physical Measurements*, 2nd Edition, OUP, 2007). We have referenced the most important standards for each of the body parts. The present project was not intended to generate new normative data, novel objective assessment techniques, or novel morphologic features. We acknowledge a tremendous lack of data on ethnic groups other than Caucasian, and we encourage efforts to address this deficiency.

The use of radiographs as part of morphologic assessments (such as a hand radiograph to distinguish a broad thumb from a bifid thumb) was controversial in the group. Although we decided not to use radiographic findings in the definitions and only use data obtained through surface examination, this issue will need to be re-evaluated.

We provide only limited and general anatomical background and have avoided more substantive explanations and reviews of what is known (or hypothesized) about the mechanism of the genesis of the physical findings. This is for several reasons. First, other excellent texts provide thorough discussions of this topic (Sadler TW, Langman J. *Langman's Medical Embryology*, 8th Edition, Lippincott Williams & Wilkins, 2000; Moore KL, Dalley AF. *Clinical Oriented Anatomy*, 5th Edition, Lippincott Williams & Wilkins, 2005). Second, our understanding of the genesis of an anatomical finding is likely to change over time, and we did not want the definitions to change should this occur. Third, the current debate about the proposed genesis of many of the findings could detract from the intended goals of this work.

We emphasize that the terms deliberately avoid any commentary about normal or abnormal status, even when this is obvious (such as a proboscis). This is because many morphological findings are commonly observed as isolated features in the normal population. There can also be a remarkable difference in frequency of a finding among various populations. The objectively determined findings are by definition abnormal in a normative, statistical sense. However, even in these cases, the individual should simply be considered to have the feature, and no judgment of normal or abnormal status is made.

We have elected not to provide differential diagnoses for the features as such lists could be very long or incomplete. Syndromes are typically diagnosed by combinations of features, and therefore differential diagnoses of single features are of limited utility. Readers should consult databases of medical dysmorphology for this information (*London Dysmorphology Databases*, version 1.0.14, 2008; *POSSUM* web version 2008).

Bundling is a word we have used to describe a term that represents two or more component findings. The term "large nose" can serve as an example of a bundled term as the term comprises several distinct features: prominent nose; wide nasal ridge; prominent nasal tip; and broad nasal base. We have eliminated most bundled terms as they often include presumptions of pathogenesis or association, which may or may not be correct. Bundling is also problematic because it can obscure component abnormalities and if individuals are described using both the single bundled term and the component terms, it can lead to confusion. A number of terms have remained bundled, as we felt that the utility of the bundled term outweighed the potential pitfalls of bundling. If a term is considered to be bundled, this is mentioned in the definition and the individual components are indicated in the description.

THE FUTURE OF THE TERMINOLOGY PROJECT

While the project thus far has taken more than 5 years, there is a long road ahead. The present series of papers should be considered a starting point, and we enthusiastically solicit input from colleagues within and beyond the field of dysmorphology on some of the difficult issues described here to correct, edit, modify, or extend the definitions. We acknowledge that we are a self-selected special interest group and do not claim to represent the international dysmorphology community. We want the definitions to be available and useful to a large community of clinicians and researchers. Thus, we plan to distribute them to other expert groups including dentists, ophthalmologists, dermatologists and other non-geneticists for comments in their area of expertise. Our intent is to use this input to refine the terms and definitions, add the body parts that we have not yet defined, and publish the complete group of terms needed to describe the human phenotype as a monograph. With time we hope to make them available in other languages.

We are presently working on adapting the working groups that established the definitions presented here into a permanent international nomenclature committee, such as the ones that exist in cytogenetics (International Standard of Cytogenetic Nomenclature) and molecular genetics (The Human Genome Variation Society nomenclature recommendations). Such a committee could periodically review the definitions and change them where necessary. Edward Sapir (1884–1939) is famous for his axiom "Language structures thought." We hope our joint effort will help to structure the language we use in dysmorphology.

ACKNOWLEDGMENTS

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RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Elements of Morphology: Standard Terminology for the Head and Face

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An international group of clinicians working in the field of dysmorphology has initiated the standardization of terms used to describe human morphology. The goals are to standardize these terms and reach consensus regarding their definitions. In this way, we will increase the utility of descriptions of the human phenotype and facilitate reliable comparisons of findings among patients. Discussions with other workers in dysmorphology and related fields, such as developmental biology and molecular genetics, will become more precise. Here we introduce the anatomy of the craniofacies and define and illustrate the terms that describe the major characteristics of the cranium and face. Published 2009 Wiley-Liss, Inc.†

Key words: nomenclature; definitions; anatomy; anthropometry; head; cranium; face; neck; chin; maxilla; mandible

How to Cite this Article:
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Am J Med Genet Part A 149A:6–28.

INTRODUCTION

General

This paper is part of a series of six papers defining the morphology of regions of the human body [Biesecker et al., 2008; Carey et al., 2008; Hall et al., 2008; Hennekam et al., 2008; Hunter et al., 2008]. The series is accompanied by an introductory article describing general aspects of this study [Allanson et al., 2008]. The reader is encouraged to consult the introduction when using the definitions. The definitions are listed alphabetically based on the physical feature, not the modifier. When a feature is indicated in the text in ***Bold-italics***, a definition is available either in this paper or one of the accompanying papers.

The appearance of facial morphology varies considerably with facial expression and movement, and depending on the position of the observer and observed person. When assessing a feature, the head of the observed person should be held in the Frankfurt horizontal, with the facial and neck muscles relaxed, eyes open, lips making gentle contact, and facial expression neutral. The face of the observer should be at the same height as the face of the observed person.

Anatomy of the Face and Cranium

Head shape and upper face shape are closely related to the shape of the bony skull. Figures 1 and 2 show the bony anatomy of the face. Many anthropological landmarks, bony and soft tissue, are illustrated in Figures 3 and 4.

The Anatomy of the Various Structures is Described in More Detail Below.

Cranium: The upper part of the skull consists of paired frontal and parietal bones and a single posterior occipital bone (Figs. 1 and 2). In early life these bones are separated by five major sutures (Figs. 1 and 2). Three, the coronal, lambdoidal and squamosal, are paired, and two, the sagittal and metopic, are single. Cranial growth normally occurs perpendicular to each of these major sutures.

Forehead: The part of the face above the eyebrows, below the hairline and between the temples. The paired frontalis muscles join in the midline and adhere to the superficial fascia over the frontal bone. These muscles effect forehead wrinkling or furrowing. They have no bony attachments, but inferiorly the fibres blend with the

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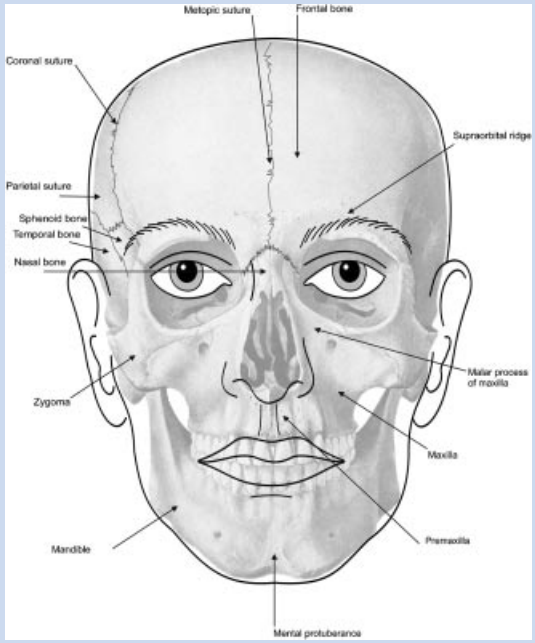

FIG. 1. An antero-posterior view of the cranium and face shows bony landmarks.

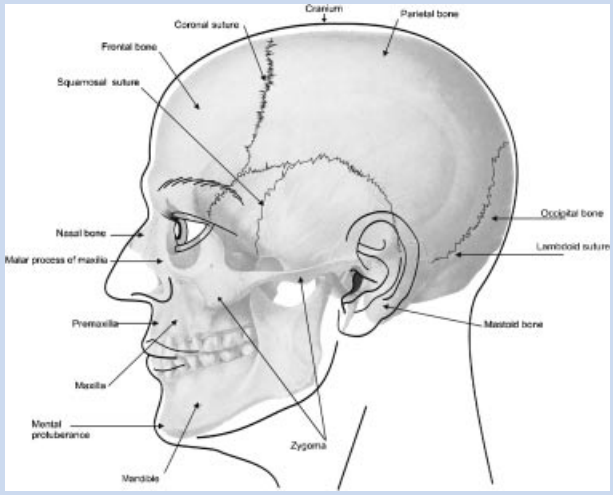

FIG. 2. A lateral view of the cranium and face shows bony landmarks.

muscles encircling the eyelids. From these attachments the fibers are directed upward, and join the galea aponeurotica below the coronal suture. The galea aponeurotica is a layer of dense fibrous tissue which covers the upper part of the cranium and attaches posteriorly to the occipital bone. It is closely connected to the integument by the firm, dense, fibro-fatty layer which forms the superficial fascia of the scalp. It cannot be wrinkled or furrowed because it does not contain muscle fibres. The anterior hairline is typically situated at the junction of frontalis muscle and galea aponeurotica.

Glabella: The most prominent point on the frontal bone above the root of the nose.

Supra-orbital ridge: The supraorbital portion of the frontal bones.

Midface: This is a region and not an anatomical term. It extends, superiorly, from the inferior orbital margin to, inferiorly, the level of nasal base. It is formed by the maxilla (upper jaw) and zygoma. Traditionally, the nose and premaxilla are not included in the midface.

Maxilla: These paired bones form, by their union, the upper jaw and contain the upper dentition. Each assists in forming the boundaries of three cavities—the palate, floor and lateral wall of the nose (frontal or malar process), and floor of the orbit. Each bone consists of a body and four processes—zygomatic, malar (frontal), alveolar and palatine.

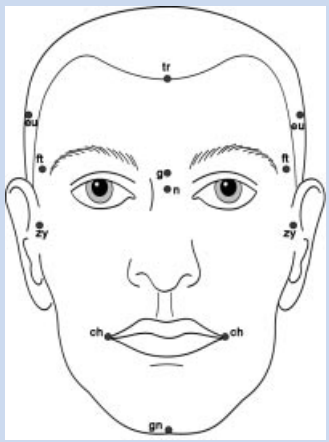

FIG. 3. Anthropological landmarks of the face, frontal view, which are described in this paper.

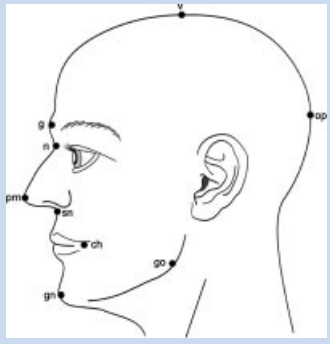

FIG. 4. Anthropological landmarks of the face, lateral view, which are described in this paper.

Malar process (syn. frontal process): The most medial and superior part of the maxilla. It forms the medial border of the inferior bony orbit, and is contiguous with the lateral boundary of the nasal bridge.

Zygoma: The part of the temporal bone of the skull that forms the prominence of the cheek. It is also known as the zygomatic bone or arch, the malar bone (creating confusion with the malar process of the maxilla), the cheek bone and the yoke bone. The zygomatic arch is composed of the malar process of the maxilla, medially, the zygoma, centrally, and the temporal bone, posterolaterally. It forms part of the part of the lateral wall and floor of the orbit.

Premaxilla: The part of the maxilla in which the 4 upper incisors develop, which forms the primary palate, and underlies the philtrum and upper lip.

Lower face: The part of the face between the mouth and the inferior point of the chin

Cheek: The soft tissues between the zygoma and mandible

Mandible: The lower jaw in which the lower teeth reside. It consists of a curved, horizontal portion, the body, and two perpendicular portions, the rami, which unite with the ends of the body nearly at right angles.

Chin: The inferior portion of the face lying inferior to the lower lip and including the central prominence of the lower jaw

Neck: The part of the body connecting the head with the shoulders

DEFINITIONS

CRANIUM

Acrocephaly: See ***Turricephaly***

Brachycephaly

Definition: Cephalic index greater than 81% (Fig. 5). *objective* OR

Apparently shortened anteroposterior dimension (length) of the head compared to width. *subjective*

FIG. 5. ***Brachycephaly*: The skull has a reduced antero-posterior dimension with the back of the head appearing to have reduced convexity.**

Comments: Cephalic index is the ratio of head width expressed as a percentage of head length. The normal range is 76–80.9%. Head length is measured between the glabella (the most prominent point on the frontal bone above the root of the nose) and the most prominent part of the occiput in the midline, using spreading calipers. Head width is measured between the most lateral points of the parietal bones on each side of the head, using spreading calipers. Cephalic index standards are derived from Caucasians and have limited relevance for other races and ethnicities. Current norms also have limited validity because of changes in infant sleeping position and consequent changes in head shape. New data should be developed. Brachycephaly is distinct from ***Flat occiput***, but both can be present in the same individual and should be coded separately.

Dolichocephaly

Definition: Cephalic index less than 76% (Fig. 6). *objective* OR

Apparently increased antero-posterior length of the head compared to width. *subjective*

Comments: Cephalic index is the ratio of head width expressed as a percentage of head length. The normal range is 76–80.9%. Head length is measured between the glabella (the most prominent point on the frontal bone above the root of the nose) and the most prominent part of the occiput in the midline, using spreading calipers. Head width is measured between the most lateral points of the parietal bones on each side of the head, using spreading calipers. Cephalic index standards are derived from Caucasians and have limited relevance for other races and ethnicities. Current norms have limited validity because of changes in infant sleeping position and consequent changes in head shape. New data should be developed. Dolichocephaly is distinct from ***Prominent occiput***, but both can be present in the same individual and should be coded separately. Scaphocephaly is a subtype of dolichocephaly where the anterior and posterior aspects of the cranial vault are pointed (boat-shaped).

FIG. 6. ***Dolichocephaly*. The skull has an increased antero-posterior dimension. Scaphocephaly is demonstrated on the right. Note that this subtype of dolichocephaly is "boat-shaped" with pointed anterior and posterior aspects of the cranial vault.**

Head circumference, enlarged: See ***Macrocephaly***

Head circumference, reduced: See ***Microcephaly***

Kleeblattschädel: See ***Skull, cloverleaf***

FIG. 7. *Macrocephaly*. Note the increased size of the cranium. Differences in size are difficult to appreciate but increased head size in this child is notable because of comparison with the smaller face.

Macrocephaly

Definition: Occipitofrontal (head) circumference greater than 97th centile compared to appropriate, age matched, sex-matched normal standards (Fig. 7). *objective* OR

Apparently increased size of the cranium. *subjective*

Comments: Head circumference is measured from just above the glabella (the most prominent point on the frontal bone above the root of the nose) to the most posterior prominent point of the occipital bone using a tape measure. Some standard charts are organized by centiles [Hall et al., 2007], others by standard deviations [Farkas, 1981]. It is important to add an indication of how far above the normal standard the head circumference is if an accurate assessment of this can be made. Macrocephaly is an absolute term. The term relative macrocephaly can be used when the head size centile exceeds the centile for height, for example, head size at the 75th centile with height at the 5th centile for age and sex.

Synonyms: Head circumference, enlarged; OFC, large.

Replaces: Macrocranium

Macrocranium: See ***Macrocephaly***

Microcephaly

Definition: Occipito-frontal (head) circumference (OFC) less than 3rd centile compared to appropriate, age matched, normal standards (Fig. 8). *objective* OR

Apparently decreased size of the cranium. *subjective*

Comments: Head circumference is measured from just above the glabella (the most prominent point on the frontal bone above the root of the nose) to the most posterior prominent point of the occipital bone using a tape measure. Some standard charts are organized by centiles [Hall et al., 2007], others by standard deviations [Farkas, 1981]. It is important to add an indication of how far below the normal standard the head circumference is if an accurate assessment of this can be made. Microcephaly is an absolute term. The term relative microcephaly can be used when the head size centile is less than the centile for height, for example, head size at the 3rd centile with height at the 75% for age and sex.

FIG. 8. *Microcephaly*. Decreased size of the cranium is accompanied by marked posterior sloping of the forehead.

Synonyms: Head circumference, reduced small; OFC, small.

Replaces: Microcranium

Microcranium: See ***Microcephaly***

Occiput, Flat

Definition: Reduced convexity of the occiput (posterior part of skull) (Fig. 9). *subjective*

Comments: Reduced convexity of the occiput gives an appearance of flattening. There are no objective measures for convexity of the occiput, and evaluation depends heavily on the experience of the observer. This finding may or may not be accompanied by ***Brachycephaly*** (which should be coded separately), and may be observed more frequently when an infant is placed to sleep on his/her back.

FIG. 9. *Occiput, flat*. There is reduced convexity of the occiput giving an appearance of flattening of the back of the skull.

Occiput, Prominent

Definition: Increased convexity of the occiput (posterior part of the skull) (Fig. 10). *subjective*

Comments: Increased convexity of the occiput gives an appearance of prominence. There are no objective measures for convexity of the occiput, and evaluation depends heavily on the experience of the observer. This finding may or may not be accompanied by ***Dolichocephaly***, but this should be coded separately.

FIG. 10. ***Occiput, prominent*****. The posterior part of the skull shows increased convexity.**

OFC, enlarged: See ***Macrocephaly***

OFC, reduced: See ***Microcephaly***

Oxycephaly: See ***Turricephaly***

Plagiocephaly

Definition: Asymmetric head shape, which is usually a combination of unilateral occipital flattening with ipsilateral frontal prominence, leading to rhomboid cranial shape (Fig. 11). *subjective*

Comments: Plagiocephaly may affect the posterior skull alone.

Scaphocephaly: See ***Dolichocephaly***

Skull, Cloverleaf

Definition: Trilobar skull configuration when viewed from the front or behind (Fig. 12). *subjective*

Synonym: Kleeblattschädel

Trigonocephaly

Definition: Wedge-shaped, or triangular head, with the apex of the triangle at the midline of the forehead and the base of the triangle at the occiput (Fig. 13). *subjective*

Comments: This shape should be assessed from above, with the examiner looking down on the head of the patient.

Turricephaly

Definition: Tall head relative to width and length (Fig. 14). *subjective*

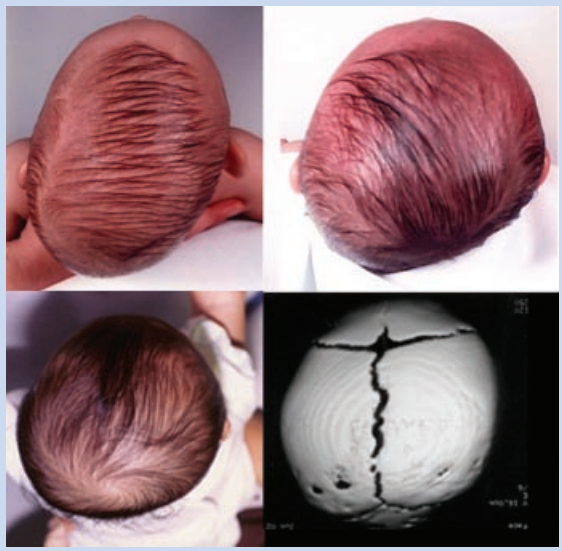

FIG. 11. ***Plagiocephaly*****. There is asymmetry of head shape: Note that one can see a combination of unilateral occipital flattening with ipsilateral frontal prominence, leading to rhomboid cranial shape or asymmetry of the posterior skull alone. These figures are kindly provided by John Graham Jr.**

FIG. 12. ***Skull, Cloverleaf*****. The skull has a trilobar configuration when viewed from the front or behind.**

Comments: This feature may have previously been considered to overlap with or include a tall forehead. *Turricephaly* is present when the head appears tall (subjective) and head length and width are reduced compared to normal age-related standards (objective). Head length is measured between the glabella (the most prominent point on the frontal bone above the root of the nose) and the most prominent part of the occiput in the midline, using spreading calipers. Head width is measured between the most lateral points of the parietal bones on each side of the head, using spreading calipers. The term acrocephaly (or oxycephaly) is used when there is turricephaly and the top of the skull assumes a cone shape.

FIG. 13. *Trigonocephaly*. Note the wedge-shaped, or triangular head, with the apex of the triangle at the midline of the forehead and the base of the triangle at the occiput.

FIG. 14. *Turricephaly*. The head is tall head relative to its width and length. The terms acrocephaly or oxycephaly are used when there is turricephaly and the top of the skull assumes a cone shape (lower image).

SCALP HAIR

Cowlick: See ***Hair, frontal upsweep***

Crown, double: See ***Hair whorl, abnormal position***

Frontal Balding

Definition: Absence of hair in the anterior midline and/or parietal areas (Fig. 15). *subjective*

FIG. 15. *Frontal balding*. Note the absence of hair in the anterior midline and/or parietal areas.

Hair, Frontal Upsweep

Definition: Upward and/or sideward growth of anterior hair (Fig. 16). *subjective*

Replaces: Cowlick, which may be considered pejorative.

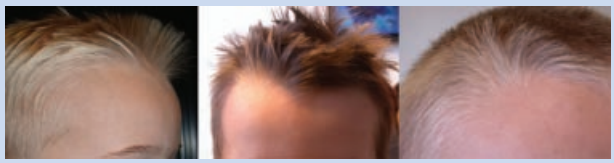

FIG. 16. *Hair, frontal upsweep*. Note the pattern of upward and sideward growth of anterior hair.

Hair Whorl, Abnormal Number

Definition: More than two clockwise hair whorls. *objective*

Comments: Most individuals have one clockwise hair whorl at a single point on the scalp lateral to the midline but close to the vertex of the skull. Five percent of the population has two whorls (Fig. 17). A double hair whorl is sometimes referred to as a double crown. In 10%, whorl direction is counter-clockwise.

FIG. 17. *Hair whorl, double*.

Hair Whorl, Abnormal Position

Definition: Hair growth from a single point on the scalp in any location other than lateral to the midline and close to the vertex of the skull (Fig. 18). *objective*

Comments: Placement of hair whorl should be described as parietal, vertex, eccentric, etc. In addition, the number of hair whorls should be noted. Five percent of the population has two whorls.

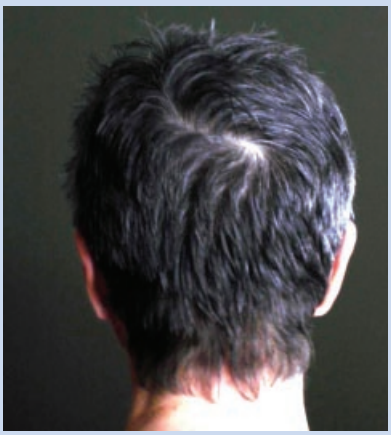

FIG. 18. ***Hair whorl, abnormal position*****. The hair whorl is positioned postero-inferiorly than its usual location lateral to the midline and close to the vertex of the skull.**

FIG. 19. ***Hairline, high anterior*****. The high anterior hairline contributes to an appearance of tall forehead.**

Hairline, High Anterior

Definition: Distance between the hairline (trichion) and the glabella (the most prominent point on the frontal bone above the root of the nose), in the midline, more than two SD above the mean (Fig. 19a,b). *objective* OR

Apparently increased distance between the hairline and the glabella. *subjective*

Comments: This measurement is carried out with sliding calipers [Farkas, 1981].

This feature gives the appearance of a tall forehead, and may or may not include reduction of hair in the temporal areas. This can be distinguished from male pattern baldness as the hairline is the superior boundary of the muscular forehead, which can be actively wrinkled, in contrast to the scalp where no wrinkling can occur. In addition, texture of the skin of the scalp differs from the texture of the skin over the forehead.

Synonym: Forehead, tall

Hairline, Low Anterior

Definition: Distance between the hairline (trichion) and the glabella (the most prominent point on the frontal bone above the root of the nose), in the midline, more than two SD below the mean (Fig. 20a,b). *objective* OR

FIG. 20. ***Hairline, low anterior*****. The low anterior hairline contributes to an appearance of short forehead.**

Apparently decreased distance between the hairline and the glabella. *subjective*

Comments: This measurement is carried out with sliding calipers [Farkas, 1981].

This feature gives the appearance of a short forehead. It is distinct from hirsutism of the forehead. In the latter, orientation of hair growth is lateral and texture and density of hair differs from scalp hair.

Synonym: Forehead, short

Hairline, Low Posterior

Definition: Hair on the neck extends more inferiorly than usual (Fig. 21). *subjective*

Comments: This feature is often seen in later childhood, as the neck lengthens, in an individual who was born with redundant nuchal skin, which should be assessed and coded separately.

FIG. 21. ***Hairline, low posterior***. Hair on the neck extends more inferiorly than usual, particularly in the lateral aspects.

Scalp Hair, Sparse

Definition: Decreased number of hairs per unit area (Fig. 22). *subjective*

Comments: Hypotrichosis should not be used as a synonym as, formally, it means underdevelopment of the hair. No normal values for number of hairs per unit area exist.

FIG. 22. ***Scalp hair, sparse***. Hair density is reduced giving a thinned appearance.

Replaces: Scalp hair, thinning; Scalp hair, thin

Scalp hair, thin: See ***Scalp hair, sparse***

Scalp hair, thinning: See ***Scalp hair, sparse***

Widow's Peak

Definition: Frontal hairline with bilateral arcs to a low point in the midline of the forehead (Fig. 23). *subjective*

Comments: The hair may need to be pulled back to recognize this feature. Historically, English widows in the 18th century wore a black hat, triangular in shape, with a point facing forward in the midline.

FIG. 23. ***Widow's peak***. Frontal hairline shows bilateral arcs to a low point in the midline of the forehead.

FACE

Brows, prominent: See ***Supraorbital ridges, prominent***

Brows, underdeveloped: See ***Supraorbital ridges, underdeveloped***

Face, Broad

Definition: Bizygomatic (upper face) and bigonial (lower face) width greater than 2 SD above the mean (Fig. 24). *objective* OR

An apparent increase in the width of the face. *subjective*

Comments: Objective measurement of upper facial width is made with spreading calipers. The tips of the calipers are passed over the zygomatic arches until maximum width is determined. Objective measurement of the lower face is made with spreading calipers, with the tips firmly pressed against the inferomedial surface of the angle of the mandible [Farkas, 1981]. Broad face is distinct from ***Round face***.

FIG. 24. *Face, broad*. An increased width of the upper and lower face.

Face, Coarse

Definition: Absence of fine and sharp appearance of brows, nose, lips, mouth and chin, usually because of rounded and heavy features or thickened skin with or without thickening of subcutaneous and bony tissues (Fig. 25). *subjective*

Comments: Note that this is a bundled term, which conveys an impression or gestalt. It is nonetheless a useful term. Note that the word coarse may be considered pejorative by some.

Synonyms: Lineaments, coarse; Features, coarse.

FIG. 25. *Face, coarse*. Facial features lack the usual fine and sharp appearance and are rounded and heavy with thickened skin, subcutaneous or bony tissues.

Face, elongated: See ***Face, long***

Face, Expressionless

This term, which is synonymous with dull expression or hypomimic face, is not defined here as it describes a functional not a structural feature (reduced facial movement leads to reduced crease formation).

Face, Flat

Definition: Absence of concavity or convexity of the face when viewed in profile (Fig. 26). *subjective*

Comments: A useful guide is to imagine that a line connecting the glabella to the anterior most part of the mandible touches the top of the philtrum where it meets the base of the columella. The glabella is the most prominent point on the frontal bone above the root of the nose. If the superior philtrum is anterior to this line the face is convex, if the superior philtrum is posterior to this line the face is concave.

FIG. 26. *Face, flat*. The profile of the face is flat with no concavity or convexity.

Face, Hypotonic

This term is to be deleted as it is a functional, not anatomic, descriptor. Also, it is not a unitary objective finding and instead represents a conclusion based on a number of features that are defined elsewhere. These include reduced nasolabial folds, laxity and drooping of lateral supraorbital skin and ptosis, mouth held open and reduced facial expression.

Face, Long

Definition: Facial height (length) is more than 2 SD above the mean (Fig. 27). *objective* OR

An apparent increase in the height (length) of the face. *subjective*

Comments: Objective measurement of face height is made with sliding calipers from the nasion, just above the depth of the nasal root, to the gnathion, the inferior border of the mandible, both in the midline [Farkas, 1981]. Long face is distinct from ***Narrow face***.

Synonym: Face, elongated

FIG. 27. *Face, long*. Height (length) of the face is increased in comparison to face width. Without actual measurement it can be difficult to decide whether increased height or reduced width is real.

Face, Narrow

Definition: Bizygomatic (upper face) and bigonial (lower face) width are more than 2 SD below the mean (Fig. 28). *objective* OR

An apparent reduction in the width of the upper and lower face. *subjective*

Comments: Objective measurement of upper facial width is made with spreading calipers. The tips of the calipers are passed over the zygomatic arches until maximum width is determined. Objective measurement of the lower face is made with spreading calipers, with the tips firmly pressed against the inferomedial surface of the angle of the mandible [Farkas, 1981]. Narrow face is distinct from ***Long face***.

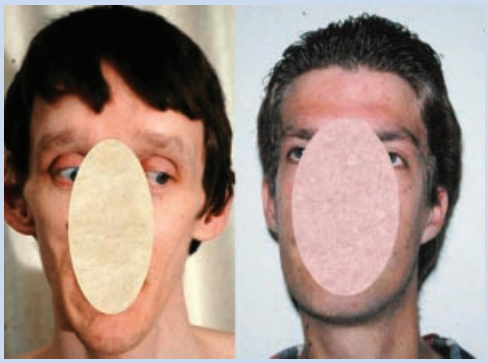

FIG. 28. *Face, narrow*. There is reduction in width of the upper and lower face. Without actual measurement it can be difficult to decide whether increased height or reduced width is real.

Face, Prematurely Aged

This term is to be deleted as it represents a conclusion based on a number of features, such as thin skin, more prominent subcutaneous venous patterning, lack of subcutaneous fat, excessive wrinkles, and pigmentary changes, that are defined elsewhere.

Face, Round

Definition: Facial appearance is more circular than usual, as viewed from the front (Fig. 29). *subjective*

Comment: While a ***Broad face*** may have rounded cheeks, a Round face appears to be as long as it is broad.

FIG. 29. *Face, round*. Facial appearance is more circular than usual.

Face, Short

Definition: Facial height (length) is more than 2 SD below the mean (Fig. 30). *objective* OR

An apparent decrease in the height (length) of the face. *subjective*

FIG. 30. *Face, short*. Decreased height (length) of the face is usually appreciated in comparison to face width and it may be difficult to decide whether reduced height or increased width is present without measurement.

Comments: Objective measurement of face height is made with sliding calipers from the nasion, just above the depth of the nasal root to the gnathion, the inferior border of the mandible, both in the midline [Farkas, 1981]. Short face is distinct from ***Wide face***.

Face, Small

This term is to be deleted as it represents a combination of two terms: ***Narrow face*** and ***Short face***. Both are defined elsewhere.

Face, Square

Definition: Facial contours, as viewed from the front, show a broad upper face/cranium and lower face/mandible, creating a square appearance (Fig. 31). *subjective*

Comments: Square face shape is usually related to increased lower facial width (a bigonial distance more than 2 SD above the mean). In ***Broad jaw*** the lower face is wider than the upper face.

FIG. 31. ***Face, square***. **The upper face/cranium and lower face/mandible are both broad, creating a square appearance.**

Face, Triangular

Definition: Facial contour, as viewed from the front, triangular in shape, with breadth at the temples and tapering to a narrow chin (Fig. 32). *subjective*

FIG. 32. ***Face, triangular***. **Facial contours are triangular in shape, with breadth at the temples tapering to a narrow chin.**

Comment: This feature is distinct from ***Narrow jaw*** where width of the midface is unchanged.

Features, coarse: See ***Face, coarse***

Lineaments, coarse: See ***Face, coarse***

FOREHEAD

Bitemporal narrowing: See ***Forehead, narrow***

Brows, prominent: See ***Supraorbital ridges, prominent***

Brows, underdeveloped: See ***Supraorbital ridges, underdeveloped***

Forehead, Broad

Definition: Width of the forehead or distance between the frontotemporales is more than 2 SD above the mean (Fig. 33). *objective* OR

Apparently increased distance between the two sides of the forehead. *subjective*

Comments: Frontotemporalis is a point lateral to the vertical component of the supraorbital ridge, where there is a hollowing. Spreading caliper tips are placed in the deepest part of that hollow [Farkas, 1981]. This term should not be confused with ***Prominent forehead***.

Synonym: Forehead, wide

FIG. 33. ***Forehead, broad***. **Note the increased distance between the two sides of the forehead.**

Forehead, bulging: See ***Forehead, prominent*** and ***Frontal bossing***

Forehead, high: See ***Hairline, high anterior***

Forehead, low: See ***Hairline, low anterior***

Forehead, Narrow

Definition: Width of the forehead or distance between the frontotemporales is more than two SD below the mean (Fig. 34). *objective* OR

Apparently narrow inter-temporal region. *subjective*

Comments: Frontotemporalis is a point lateral to the vertical component of the supraorbital ridge, where there is a hollowing. Spreading caliper tips are placed in the deepest part of that hollow [Farkas, 1981].

Synonyms: Bitemporal narrowing; Intertemporal narrowing

FIG. 34. ***Forehead, narrow*****. Note the decreased distance between the two sides of the forehead with narrowing at the temples.**

Forehead, Prominent

Definition: Forward prominence of the entire forehead, due to protrusion of the frontal bone (Fig. 35). *subjective*

Comments: This is not the same as ***Frontal bossing*** (see below).

Replaces: Forehead, bulging

FIG. 35. ***Forehead, prominent*****. The entire forehead is prominent due to protrusion of the frontal bone.**

Forehead, short: See ***Hairline, low anterior***

Forehead, Sloping

Definition: Inclination of the anterior surface of the forehead from the vertical more than 2 SD above the mean (Fig. 36). *objective* OR

Apparently excessive posterior sloping of the forehead in a lateral view. *subjective*

Comments: Measurement requires an angle meter, inclined on the anterior surface of the forehead, in the midline, along a line connecting the hairline to the glabella, compared to the vertical [Farkas, 1981] with the head held in the Frankfurt horizontal.

FIG. 36. ***Forehead, sloping*****. The anterior surface of the forehead slopes posteriorly in an excessive manner.**

Forehead, tall: See ***Hairline, high anterior***

Forehead, wide: See ***Forehead, broad***

Forehead Creases, Vertical

Definition: Vertical soft tissue creases in the midline of the forehead, often extending from the hairline to the brow, and seen with facial expression, or when the face is at rest (Fig. 37). *subjective*

FIG. 37. ***Forehead creases, vertical*****. Vertical soft tissue creases are noted in the midline of the forehead. These often extending from the hairline to the brow.**

Frontal Bossing

Definition: Bilateral bulging of the lateral frontal bone prominences with relative sparing of the midline (Fig. 38). *subjective*

Comments: This is not the same as ***Prominent forehead*** (see above)

Replaces: Forehead, bulging

FIG. 38. ***Frontal bossing*****. There is bilateral bulging of the lateral aspects of the forehead with relative sparing of the midline.**

Glabella, Depressed

Definition: Posterior positioning of the midline forehead between the supraorbital ridges (Fig. 39). *subjective*

Comments: The glabella is the area of the forehead in the midline between the supraorbital ridges, just above the nasal root. The term "depressed" used here is not meant to signify an active process.

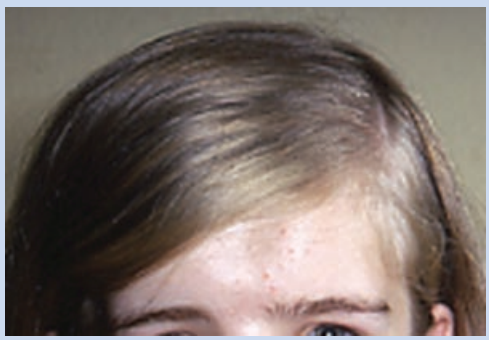

FIG. 39. ***Glabella, depressed*****. Note the depression of the midline forehead between the supraorbital ridges.**

Glabella, Prominent

Definition: Forward protrusion of the glabella (Fig. 40). *subjective*

Comments: The glabella is the area of the forehead in the midline between the supraorbital ridges, just above the nasal root.

Intertemporal narrowing: See ***Forehead, narrow***

FIG. 40. ***Glabella, prominent*****. Note prominence of the glabella, the area of the forehead in the midline between the supraorbital ridges, just above the nasal root.**

Metopic Depression

Definition: Linear vertical groove in the midline of the forehead, extending from hairline to glabella (Fig. 41). *subjective*

Comments: There is no underlying bony defect (metopism).

FIG. 41. ***Metopic depression*****. There is a linear vertical groove in the midline of the forehead, extending from hairline to glabella.**

Metopic Ridge, Prominent

Definition: Vertical bony ridge positioned in the midline of the forehead (Fig. 42). *subjective*

Comments: The ridge may extend from the hairline to the glabella or may be partial.

Synonym: Metopic suture, prominent

FIG. 42. *Metopic ridge, prominent*. Note the vertical bony ridge in the midline of the forehead.

Metopic suture, prominent: See ***Metopic ridge, prominent***

Supraorbital ridges, flattened: See ***Supraorbital ridges, underdeveloped***

Supraorbital ridges, hypoplastic: See ***Supraorbital ridges, underdeveloped***

Supraorbital ridges, hyperplastic: See ***Supraorbital ridges, prominent***

Supraorbital Ridges, Prominent

Definition: Greater than average forward and/or lateral protrusion of the supraorbital portion of the frontal bones (Fig. 43). *subjective*

Comments: The ridges need not be thickened or heavy to be prominent.

Replaces: Supraorbital ridges, hyperplastic; Brows, prominent

FIG. 43. *Supraorbital ridges, prominent*. The supraorbital portion of the frontal bones protrudes forward and laterally.

Supraorbital Ridges, Underdeveloped

Definition: Flatness of the supraorbital portion of the frontal bones (Fig. 44). *subjective*

Synonyms: Supraorbital ridges, flattened

Replaces: Supraorbital ridges, hypoplastic; Brows, underdeveloped

FIG. 44. *Supraorbital ridges, underdeveloped*. The supraorbital portion of the frontal bones is less prominent than usual.

MAXILLA AND MIDFACE

Cheekbone, flat: See ***Cheekbone, underdeveloped***

Cheekbone Prominence

Definition: Enlargement of the zygomatic process of the temporal bone of the skull, which forms the middle and lateral inferior orbital margin (Fig. 45). *subjective*

FIG. 45. *Cheekbone prominence*. The cheekbones overlying the zygoma of the temporal bone of the skull are more prominent than usual.

Synonym: Zygomatic prominence
Replaces: Zygomatic hyperplasia

Cheekbone Underdevelopment

Definition: Reduction in size of the zygomatic process of the temporal bone of the skull, which forms the middle and lateral inferior orbital margin (Fig. 46). *subjective*

Synonym: Cheekbone, flat; Zygomatic underdevelopment
Replaces: Zygomatic hypoplasia

FIG. 46. ***Cheekbone underdevelopment*****. The cheekbones overlying the zygoma of the temporal bone of the skull are less prominent than usual.**

Cheeks, Full

Definition: Increased prominence or roundness of the soft tissues between the zygomata and mandible (Fig. 47). *subjective*

FIG. 47. ***Cheeks, full*****. Note the increased prominence or roundness of the soft tissues between the cheekbones and mandible.**

Cheeks, Sunken

Definition: Lack or loss of the soft tissues between the zygomata and mandible (Fig. 48). *subjective*

Comments: This appearance is found more often in edentulous patients.

FIG. 48. ***Cheeks, sunken*****. Note the reduced prominence or fullness of the soft tissues between the cheekbones and mandible.**

Malar Flattening

Definition: Underdevelopment of the malar (frontal) process of the maxilla, appreciated in profile and/or by palpation (Fig. 49). *subjective*

Comments: The malar process is the most medial and superior portion of the maxilla, contiguous with the lateral boundary of the nasal bridge.

Replaces: Malar hypoplasia (surface examination cannot distinguish hypoplasia from hypotrophy)

FIG. 49. ***Malar flattening*****. Note the underdevelopment of bony tissues lateral to the nasal bridge extending from the inner corner of the eye to the medial aspect of the cheekbone.**

Malar hyperplasia: See ***Malar prominence***

Malar hypertrophy: See ***Malar prominence***

Malar hypoplasia: See ***Malar flattening***

Malar Prominence

Definition: Prominence of the malar (frontal) process of the maxilla and infraorbital area, appreciated in profile and from in front of the face (Fig. 50). *subjective*

Comments: The malar process is the most medial and superior portion of the maxilla, contiguous with the lateral boundary of the nasal bridge.

Replaces: Malar hypertrophy; malar hyperplasia

FIG. 50. *Malar prominence*. Note the prominence of bony tissues lateral to the nasal bridge extending from the inner corner of the eye to the medial aspect of the cheekbone.

Midface hyperplasia: See ***Midface prominence***

Midface hypertrophy: See ***Midface prominence***

Midface hypoplasia: See ***Midface retrusion***

Midface Prominence

Definition: Anterior positioning of the infraorbital and perialar regions, or increased convexity of the face, or increased nasolabial angle (Fig. 51). *subjective*

FIG. 51. *Midface prominence*. Note prominence of the infraorbital and perialar regions leading to more pronounced convexity of the face and increased nasolabial angle.

Comments: This term represents increased size of the maxilla (upper jaw) in length (increased midface height) or depth (midface prominence). In the presence of normal mandible size, maxillary prominence may give the appearance of retrognathia.

Replaces: Midface hyperplasia; Midface hypertrophy

Midface Retrusion

Definition: Posterior positioning and/or vertical shortening of the infraorbital and perialar regions, or increased concavity of the face and/or reduced nasolabial angle (Fig. 52). *subjective*

Comments: This term represents underdevelopment of the maxilla (upper jaw) in length (decreased midface height) or depth (retrusion of the maxilla). In the presence of normal mandible size, midface retrusion may give the appearance of prognathism. Caution should be used in making this assessment in edentulous patients. This is different from a ***Flat face***.

Replaces: Midface hypoplasia

FIG. 52. *Midface retrusion*. Note underdevelopment of the infra-orbital and peri-alar regions leading to more pronounced concavity of the face and reduced nasolabial angle. This gives the appearance of prognathia.

Nasolabial crease, hypoplastic: See ***Nasolabial fold, underdeveloped***

Nasolabial crease, prominent: See ***Nasolabial fold, prominent***

Nasolabial crease, underdeveloped: See ***Nasolabial fold, underdeveloped***

Nasolabial fold, hypoplastic: See ***Nasolabial fold, underdeveloped***

Nasolabial Fold, Prominent

Definition: Exaggerated bulkiness of the crease or fold of skin running from the lateral margin of the nose, where nasal base meets the skin of the face, to a point just lateral to the corner of the mouth (cheilion, or commissure) (Fig. 53). *subjective*

Comments: Increasing prominence with age is usual.

Synonym: Nasolabial crease, prominent

FIG. 53. ***Nasolabial fold, prominent*****. The crease or fold of skin running from the lateral margin of the nose, where nasal base meets the skin of the face, to a point just lateral to the corner of the mouth is more prominent than usual.**

Nasolabial Fold, Underdeveloped

Definition: Reduced bulkiness of the crease or fold of skin running from the lateral margin of the nose, where nasal base meets the skin of the face, to a point just lateral to the corner of the mouth (cheilion or commissure) (Fig. 54). *subjective*

Synonym: Nasolabial crease, underdeveloped

Replaces: Nasolabial crease, hypoplastic; Nasolabial fold, hypoplastic

FIG. 54. ***Nasolabial fold, underdeveloped*****. The crease or fold of skin running from the lateral margin of the nose, where nasal base meets the skin of the face, to a point just lateral to the corner of the mouth is less prominent than usual.**

Premaxillary hyperplasia: See ***Premaxillary prominence***

Premaxillary hypoplasia: See ***Premaxillary underdevelopment***

Premaxillary Prominence

Definition: Overdevelopment of the premaxilla (Fig. 55). *subjective*

Comments: As a consequence of prominence of the premaxilla, the overlying structures, the nose and philtrum, may appear prominent. There is increased convexity of the face and an increased nasolabial angle. In the presence of a normal sized mandible, retrognathia may be appreciated.

Replaces: Premaxillary hyperplasia

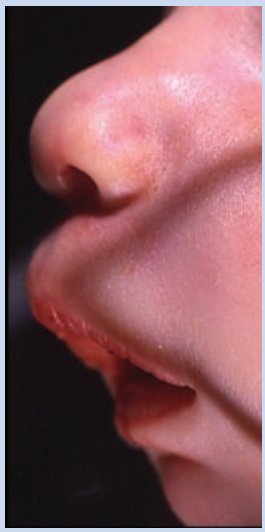

FIG. 55. ***Premaxillary prominence*****. Note increased convexity of the face and an increased nasolabial angle giving the impression of retrognathia.**

Premaxillary Underdevelopment

Definition: Reduction in size of the premaxilla (Fig. 56). *subjective*

Comments: As a consequence of underdevelopment of the premaxilla, the overlying structures, the nose and philtrum, may appear flattened. There is increased concavity of the face and a reduced nasolabial angle. In the presence of a normal sized mandible, prognathism may be appreciated.

Replaces: Premaxillary hypoplasia

FIG. 56. ***Premaxillary underdevelopment*****. Note decreased convexity of the face and nasolabial angle giving the impression of prognathia.**

Zygomatic hyperplasia: See ***Cheekbone prominence***

Zygomatic hypoplasia: See ***Cheekbone underdevelopment***

Zygomatic prominence: See ***Cheekbone prominence***

Zygomatic underdevelopment: ***See Cheekbone underdevelopment***

MANDIBLE

Chin, retruded: See ***Retrognathia***

Habsburg/Hapsburg chin: See ***Prognathism***

Habsburg/Hapsburg jaw: See ***Prognathism***

Jaw, Broad

Definition: Bigonial distance (lower facial width) more than 2 SD above the mean (Fig. 57). *objective* OR

Apparently increased width of the lower jaw (mandible) when viewed from the front. *subjective*

Comments: The lower jaw is measured from the right gonion to the left gonion using spreading calipers. The gonion is the point at the angle of the bony mandible where the ramus changes direction to become the body of the mandible. It faces inferiorly and medially and is best found by placing a finger over the outward facing angle and rolling the finger downwards and inwards [Farkas, 1981]. When a broad jaw is accompanied by a broad upper face the term ***Square face*** is used.

Synonyms: Jaw, wide; Mandible, broad; Mandible, wide; Lower face, broad; Lower face, wide

FIG. 57. ***Jaw, broad*****. Note increased width of the lower jaw (mandible).**

Jaw, Narrow

Definition: Bigonial distance (lower facial width) more than 2 SD below the mean (Fig. 58). *objective* OR

Apparently decreased width of the lower jaw (mandible) when viewed from the front. *subjective*

Comments: This dimension is measured from the right gonion to the left gonion using spreading calipers. The gonion is the point at the angle of the bony mandible where the ramus changes direction to become the body of the mandible. It faces inferiorly and medially and is best found by placing a finger over the outward facing angle and rolling the finger downwards and inwards [Farkas, 1981].

Synonyms: Mandible, narrow; Lower face, narrow

FIG. 58. ***Jaw, narrow*****. Note decreased width of the lower jaw (mandible).**

Jaw, small: See ***Micrognathia***

Jaw, wide: See ***Jaw, broad***

Lower face, broad: See ***Jaw, broad***

Lower face, narrow: See ***Jaw, narrow***

Lower face, wide: See ***Jaw, broad***

Mandible, broad: See ***Jaw, broad***

Mandible narrow: See ***Jaw, narrow***

Mandible, wide: See ***Jaw, wide***

Mandible, Cleft

Definition: Midline deficiency of the mandible and some or all overlying tissues (Fig. 59). *objective*

FIG. 59. ***Mandible, cleft*****. There is a complete midline deficiency of the mandible on the left and deficiency of overlying tissues on the right.**

Mandible, narrow: See ***Jaw, narrow***

Mandible, retruded: See ***Retrognathia***

Mandible, wide: See ***Jaw, broad***

Micrognathia

Definition: Apparently reduced length and width of the mandible when viewed from the front but not from the side (Fig. 60). *subjective*

Comments: This is a bundled term comprising shortening and narrowing of the mandible and chin. It is defined here as it is a term in common usage.

Synonyms: Micrognathism; Jaw, small

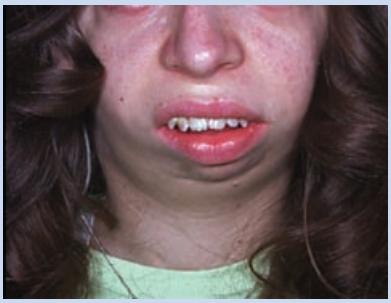

FIG. 60. ***Micrognathia*****. There is shortening and narrowing of the mandible and chin.**

Micrognathism: See ***Micrognathia***

Prognathia: See ***Prognathism***

Prognathism

Definition: Anterior protrusion of the mandibular alveolar ridge beyond the vertical plane of the maxillary alveolar ridge, best appreciated in profile (Fig. 61). *subjective*

Comments: The examiner must use judgment to distinguish a protruding mandible from an ***Underdeveloped premaxilla*** or ***Retruded midface***, either of which may be accompanied by class III malocclusion.

Synonym: Prognathia

Replaces: Habsburg chin; Hapsburg chin; Habsburg jaw; Hapsburg jaw

FIG. 61. ***Prognathism*****. There is anterior protrusion of the mandible such that the alveolar ridge extends beyond the vertical plane of the maxillary alveolar ridge.**

Retrognathia

Definition: Posteriorly positioned lower jaw, which is set back from the plane of the face when viewed from the side but not from the front (Fig. 62). *subjective*

Comments: This feature may be accompanied by micrognathia (microretrognathia) in which case both retrognathia and micrognathia should be specified and coded separately. Alternatively, it may be a mismatch in the relative position of the mandible and premaxilla, with normal mandibular length and premaxillary prominence and is accompanied by a class II malocclusion.

Synonyms: Chin, retruded; Mandible, retruded; Retrognathism

Retrognathism: See ***Retrognathia***

FIG. 62. ***Retrognathia*****. The lower jaw is set back from the plane of the face.**

CHIN

Chin, Broad

Definition: Increased width of the midpoint of the mandible (mental protuberance) and overlying soft tissue (Fig. 63). *subjective*

Comments: Micrognathia is a term that represents reduction in size of both length and width of the chin. By contrast, increases in chin size are coded separately as ***Tall chin*** and ***Broad chin***.

FIG. 63. ***Chin, broad*****. The midpoint of the mandible (mental protuberance) and overlying soft tissue is broader than usual.**

Chin, cleft: See ***Chin, vertical crease***

Chin Dimple

Definition: A persistent midline depression of the skin over the fat pad of the chin (Fig. 64). *subjective*

Comments: The borders of the depression are round.

FIG. 64. ***Chin dimple***. Note the midline depression of the skin over the fat pad of the chin.

Chin, Horizontal Crease

Definition: Horizontal crease or fold situated below the vermilion border of the lower lip and above the fatty pad of the chin, with the face at rest (Fig. 65). *subjective*

FIG. 65. ***Chin, horizontal crease***. Note the horizontal crease or fold situated below the vermilion border of the lower lip and above the fatty pad of the chin.

Chin, H-Shaped Crease

Definition: H-shaped crease in the fat pad of the chin (Fig. 66). *subjective*

Comments: The H-shape must be distinguished from ***Vertical crease of the chin***.

Synonym: Chin, H-shaped groove.

FIG. 66. ***Chin, H-shaped crease***. Note the H-shaped crease in the fat pad of the chin.

Chin, H-shaped groove: See ***Chin, H-shaped crease***

Chin, long: See ***Chin, tall***

Chin, Pointed

Definition: A marked tapering of the lower face to the chin (Fig. 67). *subjective*

Comments: The two rami of the mandible meet at an acute angle.

FIG. 67. ***Chin, pointed***. Note the marked tapering of the lower face to the chin with the two sides of the mandible meeting at an acute angle.

Chin, Short

Definition: Decreased vertical distance from the vermilion border of the lower lip to the inferior-most point of the chin (Fig. 68). *subjective*

Comments: ***Micrognathia*** is used when the chin is both short (vertical dimension) and narrow (horizontal dimension).

FIG. 68. *Chin, short*. Note the reduced vertical distance from the vermilion border of the lower lip to the inferior-most point of the chin.

Chin, Tall

Definition: Increased vertical distance from the vermillion border of the lower lip to the inferior-most point of the chin (Fig. 69). *subjective*

Synonym: Chin, long

FIG. 69. *Chin, tall*. Note the increased vertical distance from the vermillion border of the lower lip to the inferior-most point of the chin.

Chin, Vertical Crease

Definition: Vertical crease in the fat pad of the chin with the face at rest (Fig. 70). *subjective*

Comments: A vertical crease must be distinguished from an ***H-shaped crease of the chin***.

Replaces: Chin, cleft

FIG. 70. *Chin, vertical crease*. Note the vertical crease in the fat pad of the chin.

NECK

Bull-neck: See ***Neck, broad***

Neck, Broad

Definition: Increased width of the neck when viewed from the front or back (Fig. 71). *subjective*

Comments: Neck circumference may provide an objective measure of increase in width. Neck circumference is measured in a horizontal plane at the level of the most prominent portion of the thyroid cartilage, using a tape measure, with the head held erect and eyes facing forward. See ***Neck webbing*** for a related finding.

Synonyms: Neck, thick; Neck, wide

Replaces: Bull-neck

FIG. 71. *Neck, broad*. Note the increased width of the neck.

Neck, Long

Definition: Increased distance from the point where neck and shoulders meet to the inferior margin of the occipital bone (Fig. 72). *subjective*

Comments: No objective measure is available

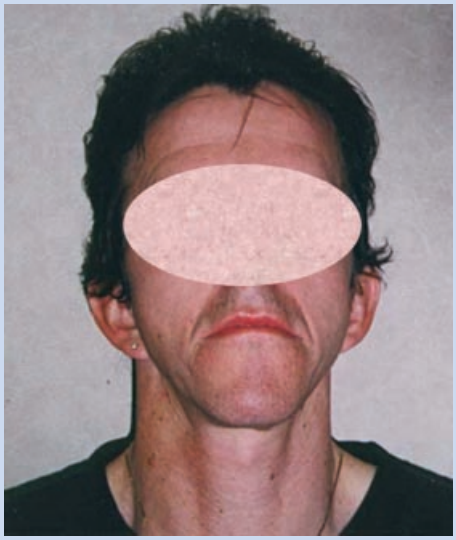

FIG. 72. ***Neck, long***. Note the increased distance from the point where neck and shoulders meet to the inferior margin of the occipital bone.

Neck, Short

Definition: Decreased distance from the point where neck and shoulders meet to the inferior margin of the occipital bone (Fig. 73). *subjective*

Comments: No objective measure is available

FIG. 73. ***Neck, short***. Note the decreased distance from the point where neck and shoulders meet to the inferior margin of the occipital bone.

Neck, thick: See ***Neck, broad***

Neck Webbing

Definition: A paravertically oriented fold of skin on the posterolateral aspect of the neck, usually extending from the mastoid region of the skull to the acromion, and best appreciated in frontal or posterior view (Fig. 74). *subjective*

Comments: This feature is often accompanied by a ***Low posterior hairline***, but this should be coded separately.

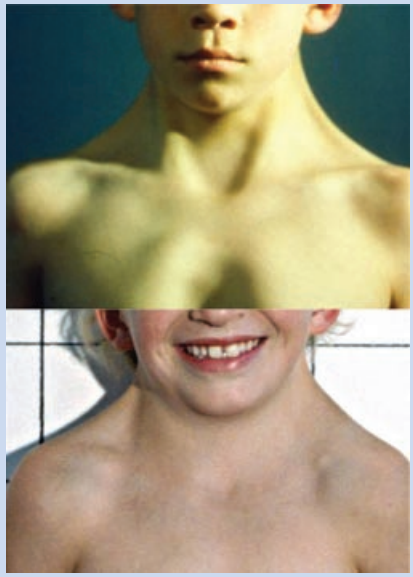

FIG. 74. ***Neck webbing***. Note the bilateral folds of skin on the posterolateral aspect of the neck.

Synonym: Pterygium colli

Neck, wide: See ***Neck, broad***

Nuchal Skin, Redundant

Definition: Excess skin around the neck, often lying in horizontal folds (Fig. 75). *subjective*

Comments: With age and increased vertical growth of the neck, excess nuchal skin may disappear and the neck may become broad or webbed. If the skin folds are vertical or paravertical, the term ***Neck webbing*** should be used.

FIG. 75. ***Nuchal skin, redundant***. Note the excess skin around the neck.

Pterygium colli: See ***Neck webbing***

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RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Elements of Morphology: Standard Terminology for the Periorbital Region

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An international group of clinicians working in the field of dysmorphology has initiated the standardization of terms used to describe human morphology. The goals are to standardize these terms and reach consensus regarding their definitions. In this way, we will increase the utility of descriptions of the human phenotype and facilitate reliable comparisons of findings among patients. Discussions with other workers in dysmorphology and related fields, such as developmental biology and molecular genetics, will become more precise. Here we introduce the anatomy of the periorbital area and define and illustrate the terms that describe the major characteristics of the periorbital area. © 2009 Wiley-Liss, Inc.

Key words: terminology; definitions; periorbital structures; eyebrows; eyelashes; eyelids; palpebrae; telecanthus; lacrimal glands

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INTRODUCTION

General

This article is part of a series of six articles defining the morphology of regions of the human body [Allanson et al., in press-b; Biesecker et al., in press; Carey et al., in press; Hennekam et al., in press; Hunter et al., in press]. The series is accompanied by an introductory article describing general aspects of this project [Allanson et al., in press-a]. The reader is encouraged to consult the introduction when using the definitions.

Anatomy

The general anatomy of the non-globe periorbital region is depicted in Figure 1. The definitions for the terms utilized in describing the features within this region are listed alphabetically. The anatomy of the various structures is described in more detail below.

Brow: The soft tissue at the junction of the frontalis and orbicularis oculi muscles, overlying the bony supraorbital ridge.

Eyebrow: The arch of hair on the brow (Fig. 2) [Goss, 1959]. The eyebrows usually extend further laterally than medially, in relation to the eye, and are wider and thicker medially. Based on observed localized abnormalities of the eyebrow, it is useful to divide the eyebrow into three parts: medial, middle (central), and lateral. The hairs of the medial part are oriented laterally, while those of the middle (central) part are oriented superolaterally. The transition between the middle and lateral parts is less frequently visible. Some syndromes have unique patterns of aberrations in one or more of these three areas. The eyebrow is sometimes referred to as the supercilium.

Eye spacing: There is wide variation in interorbital distance and in the placement of the canthi [Cohen et al., 1995]. A number of terms in this article address the nomenclature of these variations. Several of the terms are commonly confused (especially telecanthus and hypertelorism). Some of the variations are illustrated in Figure 3.

Eyelashes: Hairs that emanate from the margins of the eyelids [Goss, 1959].

Eyelid (syn. Blepharon, palpebra {plural: palpebrae}): A fold of skin and its subcutaneous components that covers the anterior globe. The upper lid is bounded by the soft tissue overlying the inferior border of the bony supraorbital ridge and inferiorly by the lid margin. The lower lid is bounded by the soft tissue overlying the infraorbital rim and superiorly by the lid margin. A

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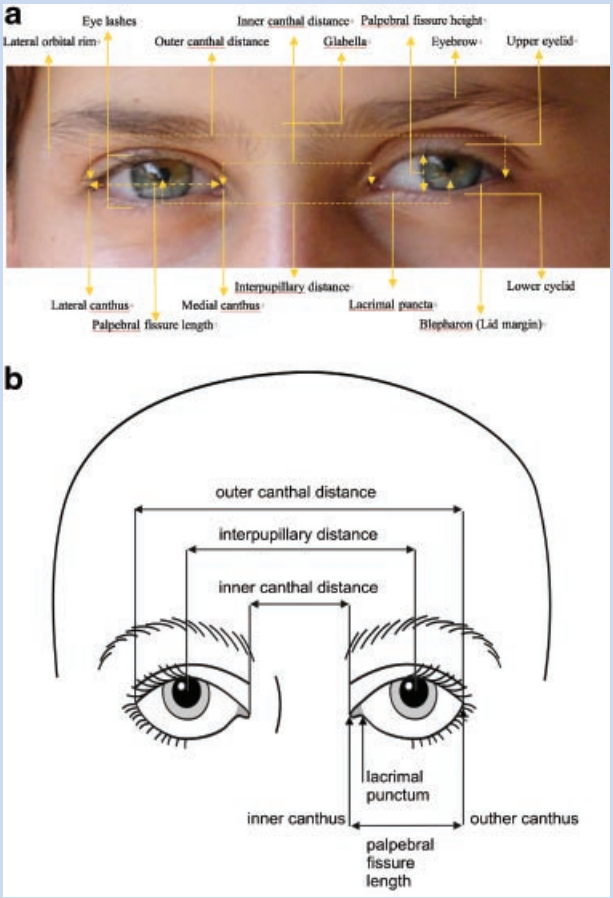

FIG. 1. Periorbital anatomy and terminology. A: A periorbital image with a number of landmarks indicated. B: A schematic diagram showing only the major periorbital features that are commonly measured (courtesy R. Hennekam, M.D.).

crescent-shaped crease on the upper eyelid represents the location of attachment of the levator palpebrae muscle to the orbicularis oculi muscle [Goss, 1959].

Lacrimal punctum {**plural: puncta**}: This structure represents the external aperture of the tear duct system. It can be absent,

FIG. 2. Typical eyebrow with mild arch, widening medially, and gradual thinning laterally.

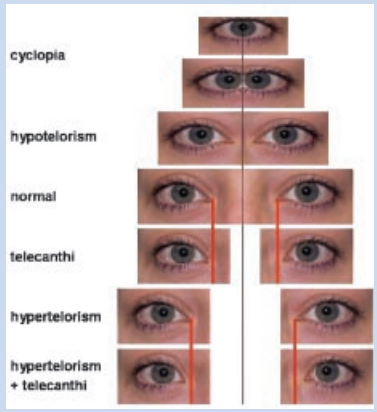

FIG. 3. Visual comparison of normal eye spacing and medial (inner) canthal position with *Closely spaced eyes*, *Widely spaced eyes* and *Telecanthus* (courtesy of R. Hennekam, M.D.). These images were artificially manipulated to simulate clinically observed variations.

malpositioned, or obstructed [Ogawa and Gonnering, 1991], and several terms below address these findings.

Palpebral fissure: When the eye is open the palpebral fissure is a scaphoid space or outline formed by eyelid margins. The palpebral fissure extends from the lateral canthus (outer canthus) to the medial canthus (inner canthus). Many factors (e.g., size, slant, eyelid architecture, ptosis) can contribute to configuration of the palpebral fissures [Hall et al., 2007].

Note that the plurality of the terms is variable. The default chosen is to specify the singular form of the term unless the term relates to a pair of structures and only makes sense in the plural form (e.g., ***Eyes, closely spaced***) or refers to a structure with many elements (e.g., ***Eyelashes, sparse***). The plurality of the terms was ignored when they were alphabetized and the terms were grouped together (e.g., "Eye. . ." and "Eyes. . ." are grouped together, and not interrupted by "Eyelashes. . .").

DEFINITIONS

Ablepharon

Definition: Absent eyelids (Fig. 4). *objective*

Comments: In ***Ablepharon*** the globe is continuously exposed [Stevens and Sargent, 2002]. It is arguable whether true aplasia of

FIG. 4. *Ablepharon*, or absent eyelids, in a patient who also has *Telecanthus* and *Downslanted palpebral fissures* (courtesy of C. Stevens, M.D.).

the eyelids exists, or whether this represents severe hypoplasia. Nevertheless, as it is difficult or impossible to make this distinction on clinical grounds, the term was felt to be useful.

Synonym: Absent eyelids

Ankyloblepharon

Definition: Partial fusion of the upper and lower eyelid margins by single or multiple bands of tissue (Fig. 5). *objective*

Comment: This term derives from Weiss et al. [1992]. A minimally expressed form, especially when located fully laterally, may be more difficult to ascertain and would be a subjective feature. A band may break and leave no evidence of its presence. Note that this term is distinct from ***Cryptophthalmos***.

Synonym: Eyelid synechiae; Ankyloblepharon filiforme adnatum

FIG. 5. ***Ankyloblepharon*, or partial fusion of the eyelids, in a patient who also has *Short palpebral fissures*.**

Ankyloblepharon filiforme adnatum: See ***Ankyloblepharon***

Antimongoloid slant: See ***Palpebral fissures, downslanted***

Atrichia: See ***Eyelashes, sparse***

Blepharochalasis

Definition: Lax, wrinkled, and baggy eyelid skin (Fig. 6). *subjective*

Comment: This finding is usually more apparent in the upper eyelid. The eyelid tissue thickness is usually also reduced. The feature is frequent in older persons [Held and Schneiderman, 1990].

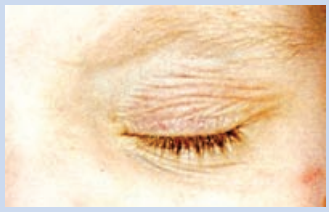

FIG. 6. ***Blepharochalasis*, wrinkled, thin eyelids, which in this patient is limited to the upper lids.**

Blepharophimosis

Definition: A fixed reduction in the vertical distance between the upper and lower eyelids with short palpebral fissures (Fig. 7). *subjective*

Comment: This term is derived from Cunniff et al. [1998]. It is an acknowledged bundled term. When palpebral fissures are severely shortened, they cannot be widely separated, actively or passively. ***Ptosis*** is the term to be used when the reduction in eyelid opening is not fixed but can be increased actively or passively. Blepharophimosis is often associated with ***Epicanthus inversus***.

FIG. 7. ***Blepharophimosis*, tight lids with short palpebral fissures giving a pseudoptosis. This patient also has *Epicanthus inversus*, which is often associated, but not required for the finding.**

Blepharoptosis: See ***Ptosis***

Ciliary trichomegaly: See ***Eyebrows, long***

Cryptophthalmos

Definition: Absent palpebral fissures, with skin passing continuously from the forehead or eyebrow onto the cheek (Fig. 8). *subjective*

Comments: This term is based on Saal et al. [1992]. This is an acknowledged bundled term, though the separate coding of the components (palpebral fissure absence; presence of eyelashes) was deemed impractical. This is typically associated with a rudimentary or small globe. Frequently, a tuft of hair accompanies the aberrant skin

FIG. 8. ***Cryptophthalmos* secondary to skin covering the lids and palpebral fissures (courtesy of John M. Graham, M.D.).**

Dystopia canthorum: See ***Telecanthus***

Ectropion

Definition: An outward turning (eversion) or rotation of the eyelid margin (Fig. 9). *subjective*

FIG. 9. *Ectropion of the lower lid*, or outward turned (everted) lower eyelid margins. See also Figure 34.

Comment: This term is derived from Cheng and Biglan [2002]. This finding is frequently associated with overexposure of the palpebral and scleral conjunctiva and cornea. It usually involves the lower eyelid. Modifiers such as "lower" and "lateral" may be applied as appropriate.

Entropion

Definition: An inward turning (inversion) of the eyelid margin (Fig. 10). *subjective*

Comment: The inward turned eyelid margin increases the potential for mechanical irritation of the eye by eyelashes [Cheng and Biglan, 2002]. This should be distinguished from ***Epiblepharon***.

FIG. 10. *Entropion*, or turned-in lower eyelids. This patient has congenital lymphedema, but this finding is not required for *Entropion* (Note that the illustration in Fig. 41 *Upper eyelid fullness*, is the maternal grandmother of the patient in this figure). The patient also has *Telecanthus* and *Widely spaced eyes*.

Epiblepharon

Definition: Redundant eyelid skin pressing the eyelashes against the cornea and/or conjunctiva (Fig. 11). *subjective*

Comment: This term is based on Lemke and Stasior [1981]. This should be distinguished from ***Entropion*** (see above).

FIG. 11. *Epiblepharon* in a young boy with folded-down upper eyelid skin, which is overlapping the palpebral aperture. He also has *Downslanted palpebral fissures*.

Epicanthal fold: See ***Epicanthus***

Epicanthus

Definition: A fold of skin starting above the medial aspect of the upper eyelid and arching downward to cover, pass in front of and lateral to the medial canthus (Fig. 12A). *subjective*

Comment: In extreme cases, the skin fold can start as high as the eyebrow [Hall et al., 2007]. This is called epicanthus superciliaris (Fig. 12B).

Synonym: Epicanthal fold; Epicanthus palpebralis

Epicanthus Inversus

Definition: A fold of skin starting at or just below the medial aspect of the lower lid and arching upward to cover, extend in front of and lateral to the medial canthus (Fig. 12C). *subjective*

Comment: For additional information on this finding, see [Oley and Baraitser, 1988].

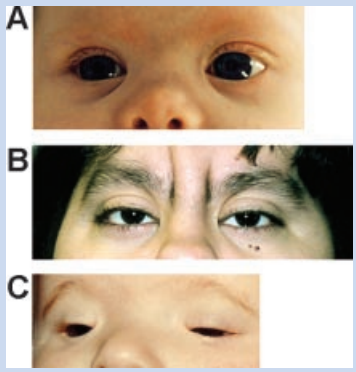

FIG. 12. A: *Epicanthus*, which comprises epicanthal folds coming from skin above the lid across and in front of medial canthi. B: *Epicanthus superciliaris*, which comprises more extensive epicanthal folds with their origins in the eyebrow. C: *Epicanthus inversus*, which is the reverse of epicanthal folds with its origin at or below the lower eyelid margin. See also Figure 7.

Eyes, Closely Spaced

Definition: Interpupillary distance more than 2 SD below the mean (Fig. 13). *objective* OR

Interpupillary distance below the 3rd centile. *objective* OR

The interpupillary distance appears to be decreased. *subjective*

Comment: The finding is measured according to Hall et al. [2007]. Note that the data from Hall et al. [2007] show SD for newborns and centiles for older patients. In the latter data, there is a continuing increase in this measurement between 14 and 15 years of age, so in the objective finding cannot be made using these norms in persons above 15 years of age. See Figure 3 for a gestalt of normal and abnormal eye spacing.

Synonym: Hypotelorism

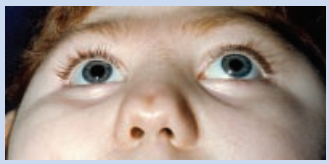

FIG. 13. ***Closely spaced eyes*, in a child. See also Figure 36.**

Eye, Deeply Set

Definition: An eye that is more deeply recessed into the plane of the face than is typical (Fig. 14). *subjective*

Comments: This finding should be distinguished from a prominent supraorbital ridge or inferior orbital margin. In ***Deeply set eyes***, the globe is recessed in comparison to the overall prominence of the face. There is no known objective measurement, and diagnosing this feature depends heavily on the experience of the observer.

Synonym: Sunken eyes

FIG. 14. ***Deeply set eyes* in a child.**

Eyes, Widely Spaced

Definition: Interpupillary distance more than 2 SD above the mean (newborns 27–41 weeks gestational age Fig. 15). *objective* OR

Interpupillary distance above the 97th centile (0–15 years of age). *objective* OR

The interpupillary distance appears to be increased. *subjective*

Comment: The finding is measured according to Hall et al. [2007]. Note that the data from Hall et al. [2007] show continuing increase in this measurement between 14 and 15 years of age, so this finding should only be made according to the subjective definition in persons above 15 years of age. It is important to distinguish between truly increased interpupillary distance and the apparently increased spacing that is caused by ***Telecanthus*** (see that entry for additional discussion). See Figure 3 for an illustration of variations of eye spacing.

Synonym: Hypertelorism

FIG. 15. **A boy with *Widely spaced eyes, subjective* but without *Telecanthus*, demonstrating the independence of these two features. See also Figures 10, 30, 33, and 40B.**

Eyebrow, Broad

Definition: Regional increase in width of the eyebrow (Fig. 16). *subjective*

Comment: broadening or flaring can be medial or lateral, and the term may be modified by appending one of these words to the term (see Fig. 16). Flaring is used to describe a widening with a change in direction of the hairs constituting the eyebrow, but these terms are considered synonymous here.

Synonym: Flared eyebrow

FIG. 16. ***Broad medial eyebrows* in a boy. In this patient, the modifier of medial is inserted into the primary term.**

Eyebrow, bushy: See ***Eyebrows, thick***

Eyebrow, Highly Arched

Definition: Increased height of the central portion of the eyebrow, forming a crescent, semicircular, or inverted U shape (Fig. 17). *subjective*

Comment: Most eyebrows have some arch with down turning medially and laterally. We know of no normative data for eyebrow arching. Identifying this feature is dependent on the experience of the observer. It may help to closely compare the shape/arching with that of siblings and parents.

FIG. 17. ***Highly arched eyebrows* in a young man. In this patient, the eyebrows are high-set and thin and they ride above the supraorbital rim, although these features are not required for the term to be used.**

Eyebrow, hirsutism of: See ***Eyebrow, thick***

Eyebrow, Horizontal

Definition: An eyebrow that extends straight across the brow, without curve (Fig. 18). *subjective*

Comment: Evaluation should be performed with the face at rest. Horizontal eyebrows are an uncommon finding.

Synonym: Straight eyebrows

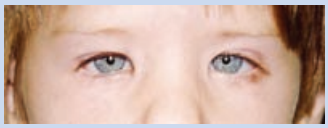

FIG. 18. ***Horizontal eyebrows***, which extend straight across from medial to lateral in this girl. See also Figure 19.

Eyebrow, hypertrichosis of: See ***Eyebrow, thick***

Eyebrow, Laterally Extended

Definition: An eyebrow that extends laterally beyond the orbital rim rather than turning gently downward at that location (Fig. 19). *subjective*

Comment: The degree of extension beyond the orbital rim considered abnormal has not been established. This feature is also uncommon.

FIG. 19. ***Laterally extended eyebrows*** in a teenage boy whose eyebrows extend far beyond the lateral orbital wall. He also has ***Horizontal eyebrows***, but this should be coded separately.

Eyebrow, Sparse

Definition: Decreased density/number and/or decreased diameter of eyebrow hairs (Fig. 20). *subjective*

Comment: Sparseness can be regional (medial, central, lateral) or total. These modifiers should be incorporated into the term, when appropriate.

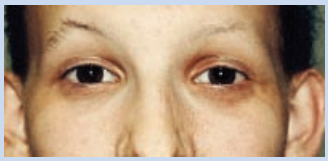

FIG. 20. ***Sparse eyebrows*** in a girl with other features of ectodermal dysplasia. See also Figure 25.

Synonym: Hypotrichosis of the eyebrow.

Eyebrow, straight: See ***Eyebrow, horizontal***

Eyebrow, Thick

Definition: Increased density/number and/or increased diameter of eyebrow hairs (Fig. 21). *subjective*

Comment: Thickness can be regional (medial, middle (central), lateral) or total. These modifiers should be incorporated into the term, when appropriate.

Synonym: Hypertrichosis of the eyebrow; Bushy eyebrow

FIG. 21. ***Thick eyebrows***, in a patient who also has ***Prominent eyelashes*** as well. This pre-teenage boy also shaves his facial hair.

Eyelashes, Absent

Definition: No eyelashes are present (Fig. 22). *objective*

Comment: This term is based on Ahmad et al. [1998]. Often this finding is congenital and associated with alopecia universalis, but this should be coded separately.

Synonym: Atrichia of eyelashes

FIG. 22. ***Absent eyelashes*** or atrichia or in an adult female.

Eyelashes, Long

Definition: Mid upper eyelash length >10 mm (Fig. 23). *objective*
OR

FIG. 23. ***Long eyelashes***, in a boy. Note that, in addition to their length, the eyelashes are unusually angled, although that feature is not required for the finding of ***Long eyelashes***. See also Figure 24.

Increased length of the eyelashes. *subjective*

Comment: Measurement should be done on the longest lashes, which are usually at the center of the lid. Normal values are 7.99 ± 1.05 mm in boys and 7.76 ± 1.03 mm in girls [Pucci et al., 2005].

Synonym: Ciliary trichomegaly.

Eyelashes, Prominent

Definition: Eyelashes that draw the attention of the viewer due to increased density and/or length and/or curl without meeting the criteria of trichomegaly (Fig. 24). *subjective*

Comment: This is admittedly a bundled term, but it may be useful in clinical practice.

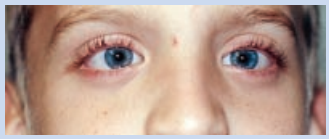

FIG. 24. ***Prominent eyelashes*** **in a boy. He also has the finding of** ***Long eyelashes*****, but that should be coded separately. See also Figure 21.**

Eyelashes, Sparse

Definition: Decreased density/number of eyelashes (Fig. 25). *subjective*

Comment: Sometimes ***Sparse eyelashes*** are abnormally formed eyelashes. The sparseness may be limited to one portion of the eyelid. We know of no normative data for ***Sparse eyelashes***.

Synonym: Hypotrichosis of eyelashes

FIG. 25. ***Sparse eyelashes*** **in a girl who also has some thinning of the lashes as well as** ***Sparse eyebrows*** **and sparse scalp hair. See also Figure 26.**

Eyelid, Cleft

Definition: A short discontinuity of the margin of the lower or upper eyelid (Fig. 26). *subjective*

Comment: The lateral segment of the lower eyelid is most commonly involved. As the milder forms of this finding are clearly subjective and no boundary of subjective and objective is defined, the term is considered subjective. The term "eyelid coloboma" has been replaced because the word "coloboma" should be used only for defects at the site of fusion of embryologic structures, which is not the case here. Modifiers to designate the location of the cleft may be added, such as "lower" and "lateral."

Synonym: Notched eyelid

Replaces: Eyelid coloboma

FIG. 26. ***Cleft lower, outer eyelid*** **in a child. Note that the modifiers of "lower" and "outer" are added to the term. Note that she also has** ***Sparse lower eyelashes*****.**

Eyelid, coloboma of: See ***Eyelid, cleft***

Eyelid, notched: See ***Eyelid, cleft***

Hypertelorism: See ***Eyes, widely spaced***

Hypotelorism: See ***Eyes, closely spaced***

Hypotrichosis: See ***Eyebrows, sparse*** or ***Eyelashes, sparse***

Infra-Orbital Crease

Definition: Skin crease extending from below the inner canthus laterally along the malar process of the maxilla and zygoma (Fig. 27). *subjective*

Comment: This feature is often found in the presence of hypoplasia of the malar process of the maxilla or zygoma, but this should be described separately. See ***Infra-orbital fold*** for a related term.

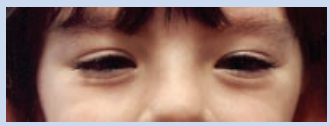

FIG. 27. ***Infra-orbital creases*** **in a girl who also has** ***Infraorbital folds*****, flat malar bones, and laterally protruding ears. See also Figure 33.**

Infra-Orbital Fold

Definition: Elevated ridge(s) of skin starting well below the medial aspect of the lower lid that curves gradually upward toward and/or across the nasal bridge (Fig. 28). *subjective*

Comments: This excludes ***Epicanthus inversus***. See ***Infra-orbital crease*** for a related term.

Lacrimal Punctum, Absence

Definition: No identifiable superior and/or inferior lacrimal punctum (Fig. 29). *objective*

Comments: The openings of the tear ducts are normally located at the medial margin of each eyelid. The opening on the lower eyelid

FIG. 28. *Infra-orbital folds* associated with upper facial edema (the latter is not required for the finding to be made). Note that these folds are oriented toward the lateral borders of the nasal bridge. See also Figure 27.

border is more visible than the opening on the upper eyelid border. Absence of the lacrimal punctum is uncommon [Ferreira et al., 2000] and although it is typically presumed to be caused by agenesis, we avoid causal mechanisms in this terminology. There was some disagreement as to whether Nasolacrimal duct obstruction should be included in this terminology. As that assessment is primarily functional and not anatomic, it was removed.

Replaces: Agenesis of the lacrimal punctum

FIG. 29. This child has *Absence of the lacrimal puncta*. Note that she also has *Short palpebral fissures, Downslanted palpebral fissures, Telecanthus*, and Nasolacrimal duct obstruction with erythematous and swollen lacrimal ducts below the medial canthi.

Lacrimal Punctum, Ectopic

Definition: Positioning of a lacrimal punctum other than at the medial margins of the eyelid (Fig. 30). *subjective*

Comment: The openings of the tear ducts are normally located at the medial margin of each eyelid. The opening on the lower eyelid border is more visible than the opening on the upper eyelid border. Ectopic positions can include the upper eyelid, nasal bridge, or inferior to medial aspect of the lower lid.

FIG. 30. The left eye shows an inferiorly placed medial canthus and *Ectopic lacrimal punctum* plus a defect in the left nostril.

Lagophthalmos

Definition: Inability to totally close the eyelids while awake, asleep, or both (Fig. 31). *subjective*

Comment: It may be an isolated finding or part of a syndrome [Korula et al., 1995] and can be associated with ***Ectropion. Lagophthalmos*** frequently results in chronic conjunctival and/or corneal irritation.

FIG. 31. This teenage girl has *Lagophthalmos*, which has caused her to have severe corneal and scleral irritation.

Mongoloid slant: See ***Palpebral fissure, upslanted***

Palpebral Fissure, Almond-Shaped

Definition: A shape created by an acute downward arching of the upper eyelid and upward arching of the lower eyelid, toward the medial canthus, which gives the outline of the palpebral fissures the configuration of an almond; thus, the maximum distance between the fissures is offset from, and medial to, the center point (Fig. 32). *subjective*

Comments: The almond configuration tends to dissipate with time as the surrounding tissues (e.g., eyelid, nasal bridge) grow.

FIG. 32. This shows a typical *Almond-shaped palpebral fissure* of the left eye. A comparison with the right eye illustrates the difference, that being the sharp descent of the upper medial canthal fold.

Palpebral Fissure, Downslanted

Definition: The palpebral fissure inclination is more than 2 SD below the mean for age (Fig. 33). *objective* OR

The inclination of the palpebral fissure is less than typical for age. *subjective*

Comments: The slant, or inclination, of the palpebral fissure is defined as the angle formed by two lines: an imaginary line that connects the lateral canthus and the medial canthus of each eye, and an imaginary horizontal line formed by the two medial canthi when the patient holds their head with the facial midline vertical, the head in a neutral vertical position (neither flexed nor extended) and the gaze forward [Farkas, 1994]. Palpebral fissure inclination norms are

FIG. 33. This boy has *Downslanted palpebral fissures, Widely spaced eyes, Proptosis,* and *Infra-orbital creases*. See also Figures 4, 11, 29, 38, and 39.

specified in Farkas [1994] for Caucasians (pg. 283) and for Chinese and African-American populations (limited data, pgs 342, and 349, respectively). Hall et al. [2007] only specifies norms for Caucasians between 6 and 16 years of age. Note that the mean inclination is slightly upslanting at all ages [Farkas, 1994; Hall et al., 2007]. Some features (e.g., ***Ptosis*** or ***Epicanthus***) may hinder palpebral fissure inclination assessment. Malar and/or zygomatic hypoplasia and ***Widely spaced eyes*** may be associated with a downward slant.

Replaces: Antimongoloid slant

Palpebral Fissure, Long

Definition: Distance between the medial and lateral canthi is more than 2 SD above the mean for age (Fig. 34). *objective* OR

Apparently increased length of the palpebral fissures. *subjective*

Comment: Measurement techniques and norms are as specified in Hall et al. [2007] and Farkas [1994]. The term "wide palpebral fissure" is discouraged because this term is vague with respect to which dimension (vertical vs. horizontal) it refers to, coupled with the popular definition of "wide-eyed," which refers to palpebral fissure height.

Replaces: Wide palpebral fissure

FIG. 34. *Long palpebral fissures*, and *Lateral ectropion* are obvious in this boy.

Palpebral Fissure, Short

Definition: Distance between the medial and lateral canthi is more than 2 SD below the mean for age (Fig. 35). *objective* OR

Apparently reduced length of the palpebral fissures. *subjective*

Comments: Measurement techniques and norms are as specified in Hall et al. [2007] and Farkas [1994]. These sources include more

FIG. 35. Young female with mildly to moderately *Short palpebral fissures*. Note some mild *Ptosis*. See also Figures 5 and 29.

complete data for Caucasians than for other groups. Decreased palpebral fissure length may be accompanied by a decreased vertical distance between the upper and lower eyelid, which gives the eyes a more slit-like appearance and may give the appearance of ***Ptosis***, but this should be coded separately if it is present. Severe shortening of the palpebral fissures leads to ***Blepharophimosis***.

Palpebral Fissure, Upslanted

Definition: The palpebral fissure inclination is more than 2 SD above the mean for age (Fig. 36). *objective* OR

The inclination of the palpebral fissure is greater than typical for age. *subjective*

Comments: The slant, or inclination, of the palpebral fissure is defined as the angle formed by two lines: an imaginary line that connects the lateral canthus and the medial canthus of each eye, and an imaginary horizontal line formed by the two medial canthi when the patient holds their head with the facial midline vertical, the head in a neutral vertical position (neither flexed nor extended) and the gaze forward [Farkas, 1994]. Palpebral fissure inclination norms are specified in Farkas [1994] for Caucasians (pg. 283) and for Chinese and African-American populations (limited data, pgs 342, and 349, respectively). Hall et al. [2007] only specifies norms for Caucasians between 6 and 16 years of age. Upslanted palpebral fissures may be associated with microcephaly, but this should be coded separately. Some features (e.g., ***Ptosis*** or ***Epicanthus***) may hinder palpebral fissure inclination assessment.

Replaces: Mongoloid slant

FIG. 36. *Upslanted palpebral fissures*, in a boy who also has *Closely spaced eyes*. See also Figure 41B.

Palpebral fissure, wide. See: ***Palpebral fissure, long***

Proptosis

Definition: An eye that is protruding anterior to the plane of the face to a greater extent than is typical (Fig. 37). *subjective*

FIG. 37. Bilateral *Proptosis*. This is a duplicate of Figure 33.

FIG. 39. A patient with *Synophrys* and *Downslanted palpebral fissures*.

Comments: This finding should be distinguished from underdevelopment of the supraorbital ridge or maxilla/zygoma. In ***Proptosis***, the globe is anteriorly protuberant in comparison to the overall plane of the face. There is no known objective measurement, and diagnosing this feature depends heavily on the experience of the observer.

Synonym: Prominent eyes

Ptosis

Definition: The upper eyelid lid margin is positioned 3 mm or more lower than usual and covers the superior portion of the iris (Fig. 38). *objective* OR

The upper lid margin obscures at least part of the pupil. *subjective*

Comment: True ptosis usually occurs in the presence of normal palpebral fissure length. Apparent ptosis is seen in ***Blepharophimosis*** and other causes of ***Short palpebral fissures***. Pseudoptosis occurs in the presence of severe zygomatic underdevelopment when the resulting dramatic downward eye slant pulls the upper eyelid diagonally across the globe.

Synonym: Blepharoptosis

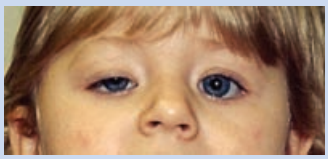

FIG. 38. *Ptosis* of the right eyelid. Note that the upper lid margin partially covers the right pupil (compare with normal left upper eyelid). She also has *Downslanted palpebral fissures*. See also Figure 35.

Synechiae: See ***Ankyloblepharon***

Synophrys

Definition: Meeting of the medial eyebrows in the midline (Fig. 39). *subjective*

Comment: Cosmetic hair removal or shaving may obscure this feature. It is controversial whether the medial eyebrows must meet in the midline to warrant this descriptor, as opposed to eyebrows that extend markedly toward the midline but do not meet.

Replaces: Unibrow

Telecanthus

Definition: Distance between the inner canthi more than 2 SD above the mean (Fig. 40). *objective* OR

Apparently increased distance between the inner canthi. *subjective*

Comments: Telecanthus may be present without (Fig. 40A) or with (Fig. 40B) ***Widely spaced eyes***. In the latter case, ***Widely spaced eyes*** should be coded separately. Inner canthal distance varies among ethnic groups. Norms are available for American Africans [Murphy and Laskin, 1990], Chinese [Wu et al., 2000], and Caucasians [Laestadius et al., 1969; Feingold and Bossert, 1974; Merlob et al., 1984; Evereklioglu et al., 2001]. In the presence of an ***Epicanthal fold***, ascertainment of inner canthal distance can be difficult.

Synonym: Dystopia canthorum

FIG. 40. A: *Telecanthus* with normal interpupillary distance. Note lack of adequate visualization of the sclera toward the medial canthi. B: *Telecanthus* with *Widely spaced eyes*. See also Figures 4, 10, 15, and 29.

Unibrow: See ***Synophrys***

Upper Eyelid, Fullness of

Definition: Swelling or distention of the upper eyelid (Fig. 41). *subjective*

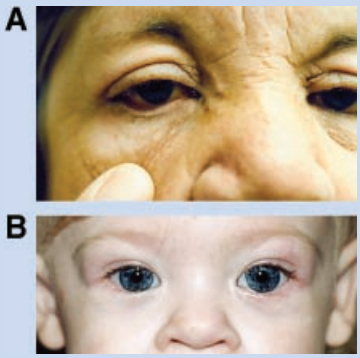

FIG. 41. A: Adult female with *Upper eyelid fullness*. In this case, the patient had eyelid edema which gives it fullness and which can invert the lids causing eye irritation. B: Localized *Upper lateral eyelid fullness* in a child who also has *Upslanted palpebral fissures* and stellate irides.

Comment: The swelling can be due to edema, fat or other depositions or inflammation. It is controversial whether there should be a separate term for fullness of the lateral upper eyelid.

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RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Elements of Morphology: Standard Terminology for the Ear

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An international group of clinicians working in the field of dysmorphology has initiated the standardization of terms used to describe human morphology. The goals are to standardize these terms and reach consensus regarding their definitions. In this way, we will increase the utility of descriptions of the human phenotype and facilitate reliable comparisons of findings among patients. Discussions with other workers in dysmorphology and related fields, such as developmental biology and molecular genetics, will become more precise. Here we introduce the anatomy of the ear and define and illustrate the terms that describe the major characteristics of the ear.

Key words: nomenclature; definitions; ear; anatomy; anthropometry

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INTRODUCTION

General

This paper is part of a series of six papers defining the morphology of regions of the human body [Allanson et al., 2009b; Biesecker et al., 2009; Carey et al., 2009; Hall et al., 2009; Hennekam et al., 2009]. The series is accompanied by an introductory article describing general aspects of this study [Allanson et al., 2009a]. The reader is encouraged to consult the introduction when using the definitions.

Anatomy of the Ear

The anatomy of the external ear, also known as the auricle or pinna, is complex [Hunter and Yotsuyanagi, 2005] and remarkably inaccurately described by most authors. The major landmarks of the external ear are depicted in Figure 1. The external ear consists of skin (with adnexa), cartilage, and six intrinsic muscles. The anatomy of the various components of the ear are described below, and illustrations are shown each time in the section describing the various features of the components (Figs. 3, 13, 18, 24, 28, 46, and 68).

Antihelix: A Y-shaped curved cartilaginous ridge arising from the antitragus and separating the concha, triangular fossa, and scapha (Fig. 3). The antihelix represents a folding of the conchal cartilage and it usually has similar prominence to a well-developed helix. The stem (the part below the bifurcation) of the normal antihelix is gently curved and branches about two thirds of the way along its course to form the broad fold of the superior (posterior) antihelical crus, and the more sharply folded inferior (anterior) crus. The inferior and superior crura of the antihelix can vary both in volume and degree of folding.

Antihelix, inferior crus: The lower cartilaginous ridge arising at the bifurcation of the antihelix that ends beneath the fold of the ascending helix, and separates the concha from the triangular fossa (Fig. 3). The inferior antihelical crus runs in an anterior and slightly superior direction, is usually sharply defined, and appears less variable than its superior counterpart. A synonym is anterior crus of the antihelix.

Antihelix, superior crus: The upper cartilaginous ridge arising at the bifurcation of the antihelix that separates the scapha from the triangular fossa (Fig. 3). The superior crus runs in a superior and slightly anterior direction and is usually less sharply folded than the

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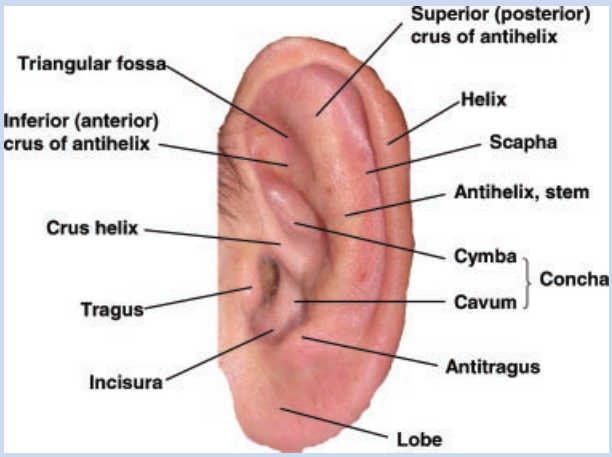

FIG. 1. Normal anatomy of the external ear.

lower portion and inferior crus. A synonym is the posterior crus of the antihelix.

Antitragus: The anterosuperior cartilaginous protrusion lying between the incisura and the origin of the antihelix (Fig. 13). The anterosuperior margin of the antitragus forms the posterior wall of the incisura.

Concha: The fossa bounded by the tragus, incisura, antitragus, antihelix, inferior crus of the antihelix, and root of the helix, into which opens the external auditory canal. It is usually bisected by the crus helix into the cymba superiorly and cavum inferiorly (Fig. 18).

Frankfurt horizontal: A plane connecting the lowest point on the lower margin of each orbit and highest point on the upper margin of the external auditory meatus [Farkas, 1981]. The Frankfurt horizontal or Frankfurt plane is used as the general horizontal plane of the head and as reference point for other planes and structures.

Helix: The outer rim of the ear that extends from the superior insertion of the ear on the scalp (root) to the termination of the cartilage at the earlobe (Fig. 28). The helix can be divided into three approximate parts: the *ascending* helix, which extends vertically from the root; the *superior* helix, which begins at the top of the ascending portion, extends horizontally and curves posteriorly to the site of Darwin tubercle (vide infra); the *descending* helix (sometimes called *posterior*), which begins inferior to Darwin tubercle and extends to the superior border of the earlobe. The lower portion of the posterior part is often non-cartilaginous. The border of the helix usually forms a rolled rim but the helix is highly variable in shape. Lange [1966] developed a graphic classification of folding variants (Fig. 29; modified from Lange [1966]). Figure 30 illustrates some variation in the helix observed among a small group of colleagues.

Helix, crus: The continuation of the anteroinferior ascending helix, which extends in a posteroinferior direction into the cavity of the concha above the external auditory meatus (Fig. 1). The average crus helix extends about one half to two thirds the distance across the concha. A synonym is crista helicis

Lobe: The soft, fleshy, inferior part of the pinna. It is bounded on its posterosuperior border by the end of the descending helix, on the anterosuperior border by the inferior border of the antitragus and superiorly by the incisura (Fig. 46). The earlobe is highly variable in size and in the degree of attachment of the anteroinferior portion to the face.

Scapha: The groove between the helix and the antihelix.

Tragus: A posterior, slightly inferior, protrusion of skin-covered cartilage, anterior to the auditory meatus. The inferoposterior margin of the tragus forms the anterior wall of the incisura (Fig. 68).

Triangular fossa: The concavity bounded by the superior and inferior crura of the antihelix and the ascending portion of the helix.

Anatomical Variation

Anomalies of the ear include quantitative traits and qualitative features of the entire ear, and of the individual components.

(1) Variation in size (macrotia; microtia; anotia).
(2) Variation in position (low-set ears; posterior angulation of the ear).
(3) Variations of the individual anatomical parts: antihelix; antitragus; concha; helix; lobe; scapha; tragus; triangular fossa.
(4) Named ear anomalies (crumpled ear; cryptotia; cupped ear; lop ear; preauricular and auricular pits; preauricular and auricular tags; preauricular ectopias; prominent ear; question mark ear; detachment of ascending helix; satyr ear; shell ear; Stahl ear).

The various features are listed alphabetically. If a feature is indicated in ***bold-italics***, the feature is listed and a definition is available. This can be in the present or one of the accompanying papers. The terms are alphabetized based on the physical feature, not the modifier.

The appearance of facial morphology varies considerably with the position of the observer and observed person, and facial movements. In assessing morphology, the head of the observed person should be held in the Frankfurt horizontal, with the facial and neck muscles relaxed, eyes open, lips making gentle contact, and a neutral facial expression. The face of the observer should be at the same height as the face of the observed person.

DEFINITIONS

Anotia

Definition: Complete absence of any auricular structures (Fig. 2). *objective*

Synonym: Ear, absent

Antihelical Shelf

Definition: Antihelix protrusion directed more anteriorly than laterally, forming a shelf overlying the posterior concha (Figs. 3–4). *subjective*

Comments: In marked cases this often appears to be associated with lack of lateral protrusion of the antihelix.

Synonym: Conchal shelf

FIG. 2. Anotia.

Antihelix, Absent

Definition: No discernible ridge between concha and triangular fossa, and helix (Fig. 5). *objective*

Comment: This finding is common in a ***Protruding*** and ***Cupped ear***, where the superior and inferior parts of the antihelix are often absent. This is distinct from partial absence of the antihelix as may occur in, for example, ***Underdeveloped inferior crus of the antihelix***.

Antihelix, Additional Crus

Definition: Supernumerary ridge or crus of the ear arising from the antihelix (Fig. 6). *objective*

Comment: The supernumerary crus usually emanates posteriorly from the antihelix at, or just above, the point of its bifurcation, but may have a different origin. In the former case, the finding is termed ***Stahl ear*** [Yamada and Fukuda, 1980].

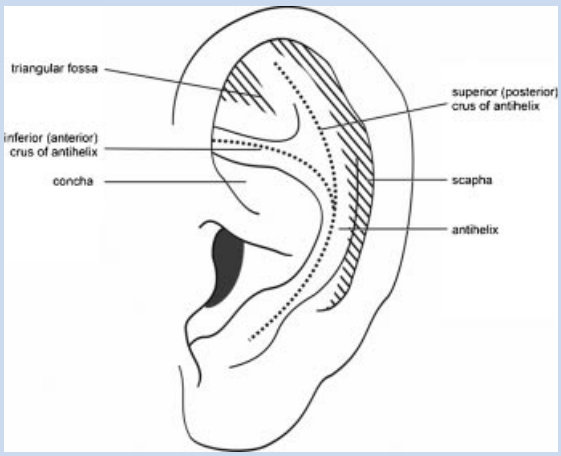

FIG. 3. Anatomy of antihelix.

FIG. 4. Antihelical shelf.

Antihelix, Angulated

Definition: Antihelical ridge that forms an acute angle between the antitragus and its bifurcation (stem) instead of a gently curving arc (Fig. 7). *subjective*

Antihelix, Inferior Crus, Broad

Definition: Increased width of the inferred cross-section of the inferior crus (Fig. 8c). *subjective*

Comment: This finding is highly variable, and the range is illustrated in Figure 8. The inferior crus is usually sharply folded giving a narrow profile.

Antihelix, inferior crus, hypoplastic: see ***Antihelix, inferior crus, underdeveloped***

Antihelix, inferior crus, hypotrophic: see ***Antihelix, inferior crus, underdeveloped***

Antihelix, inferior crus, hyperplastic: see ***Antihelix, inferior crus, prominent***

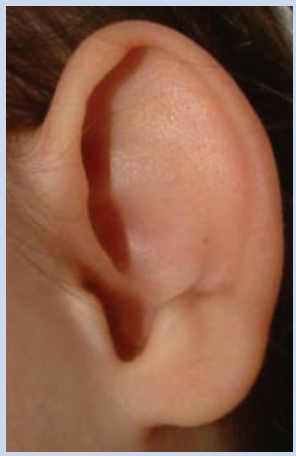

FIG. 5. Absent antihelix.

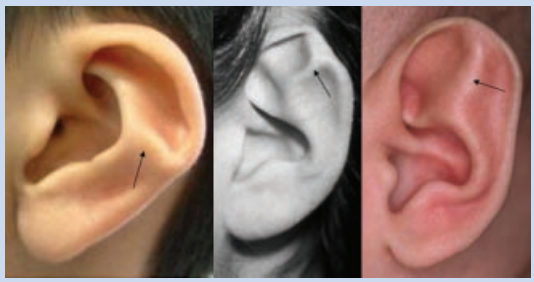

FIG. 6. Additional crus of antihelix (Stahl ear). The additional crus is indicated in each panel by an arrow.

FIG. 8. Variation in width of the inferior crus of the antihelix. a: Narrow. b: Average. c: Broad.

Antihelix, inferior crus, hypertrophic: see ***Antihelix, inferior crus, prominent***

Antihelix, Inferior Crus, Prominent

Definition: Increased protrusion of the inferior crus relative to the prominence of the antihelix stem (Fig. 9c). *subjective*

Comment: This finding is highly variable, and the range is illustrated in Figure 9.

Replaces: Inferior crus of antihelix, hyperplastic; inferior crus of antihelix, hypertrophic

Antihelix, Inferior Crus, Underdeveloped

Definition: Decreased protrusion of the inferior crus relative to the prominence of the antihelix stem (Fig. 9a). *subjective*

Comment: This finding is highly variable.

Replaces: Inferior crus of antihelix, hypotrophic; inferior crus of antihelix, hypoplastic

Antihelix, stem, hyperplastic: see ***Antihelix, stem, prominent***

Antihelix, stem, hypertrophic: see ***Antihelix, stem, prominent***

Antihelix, stem, hypoplastic: see ***Antihelix, stem, underdeveloped***

Antihelix, stem, hypotrophic: see ***Antihelix, stem, underdeveloped***

Antihelix, Stem, Prominent

Definition: Increased protrusion of the antihelical ridge, proximal to its bifurcation, relative to the prominence of the helix (Fig. 10d,e). *subjective*

Comments: This finding is highly variable, and the range is illustrated in Figure 10. The relative prominence is attributable to either increased volume of the cartilage and/or the acuteness of the folding angle. Interpretation of relative antihelical prominence may be difficult when the conchal anatomy is distorted, for example a ***Cupped ear***.

Replaces: Antihelix stem, hyperplastic; antihelix stem, hypertrophic

Antihelix, Stem, Serpiginous

Definition: Posterior curving of the antihelix from its origin at the antitragus, traveling initially almost perpendicular to the descending helix and obscuring some of the concha (Fig. 11). *subjective*

FIG. 7. Angulated antihelix. Note also the presence of a bifid lobe in the left panel.

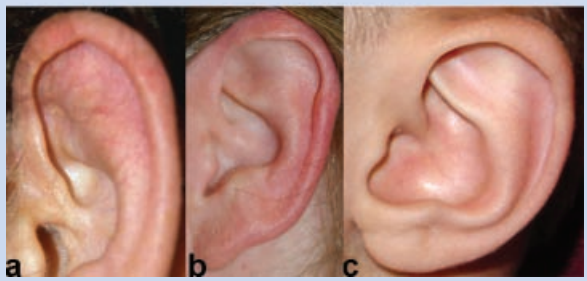

FIG. 9. Prominent and underdeveloped inferior crus of antihelix. a: Underdeveloped. b: Average. c: Prominent.

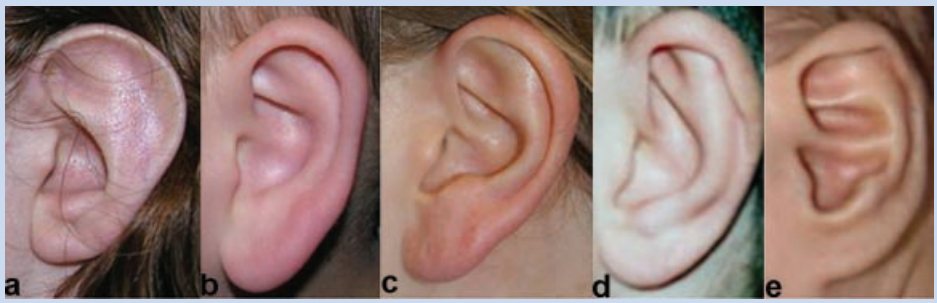

FIG. 10. Variation in protrusion of the antihelix stem. a: Marked underdevelopment. b: Mild underdevelopment. c: Average development. d: Mild prominence. e: Marked prominence.

Antihelix, Stem, Underdeveloped

Definition: Decreased protrusion of the antihelical ridge, proximal to its bifurcation, relative to the prominence of a normal helix (Fig. 10a,b). *subjective*

Comments: This finding is highly variable, and the range is illustrated in Figure 10. The degree of prominence is attributable to the volume of the cartilage and/or the acuteness of the folding angle. Interpretation of degree of antihelical prominence may be difficult when the conchal anatomy is distorted, for example a ***Cupped ear***.

Replaces: Antihelix stem, hypotrophic; antihelix stem, hypoplastic

Antihelix, superior crus, hyperplastic: see ***Antihelix, superior crus, prominent***

Antihelix, superior crus, hypertrophic: see ***Antihelix, superior crus, prominent***

Antihelix, superior crus, hypoplastic: see ***Antihelix, superior crus, underdeveloped***

Antihelix, superior crus, hypotrophic: see ***Antihelix, superior crus, underdeveloped***

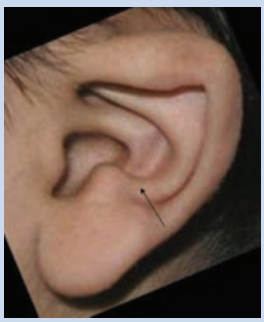

FIG. 11. Serpiginous stem.

Antihelix, Superior Crus, Prominent

Definition: Increased protrusion of the superior crus relative to the prominence of a normal antihelix stem (Fig. 12d,e). *subjective*

Comment: This finding is highly variable, and the range is illustrated in Figure 12. There may be an inverse relationship between the relative sizes of the superior and inferior crura, but these should be coded separately.

Replaces: Superior crus of antihelix, hypertrophic; superior crus of antihelix, hyperplastic

Antihelix, Superior Crus, Underdeveloped

Definition: Decreased protrusion of the superior crus relative to the prominence of a normal antihelix stem (Fig. 12a,b). *subjective*

Comment: This finding is highly variable, and the range is illustrated in Figure 12. There may be an inverse relationship between the relative size of the superior and inferior crura, but these should be assessed separately.

Replaces: Superior crus of antihelix, hypotrophic; superior crus of antihelix, hypoplastic

Antihelix, third crus: see ***Antihelix, additional crus*** and ***Stahl ear***

Antitragus, Absent

Definition: Absence of the anterosuperior prominence of the area between the bottom of the incisura and the inner margin of the antihelix (Figs. 13 and 14a,b [line 0]). *objective*

Comment: The size of the antitragus is highly variable, and the range is illustrated in Figure 14b [modified from Lange, 1966].

Antitragus, Bifid

Definition: Double rather than single peak of the antitragus (Fig. 15). *objective*

Synonym: Antitragus, double

Antitragus, double: see ***Antitragus, bifid***

Antitragus, enlarged: see ***Antitragus, prominent***

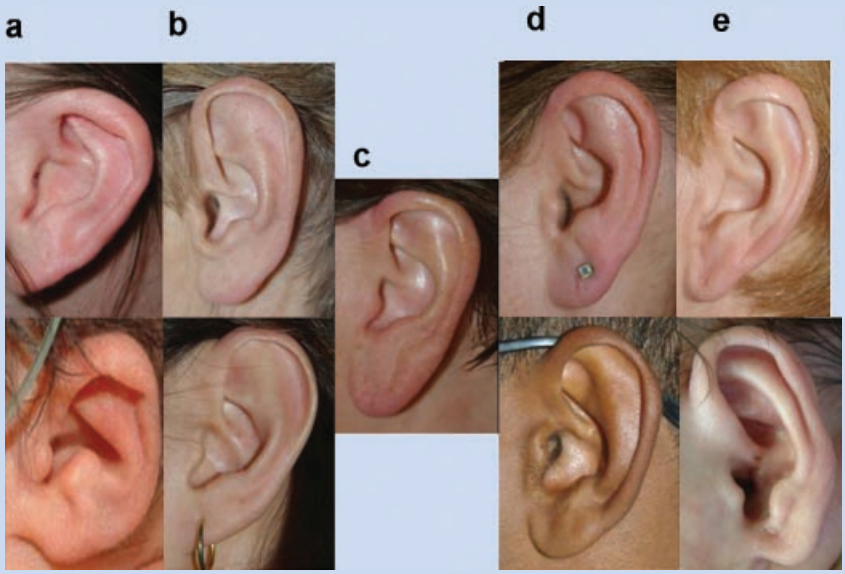

FIG. 12. Prominent and underdeveloped superior crus of antihelix. a: Absent, very indistinct. b: Slightly indistinct. c: Average. d: Slightly more distinct. e: Very distinct, sharp.

Antitragus, Everted

Definition: Positioning of the antitragus at an angle perpendicular to the plane of the ear (oriented away from the plane of the ear) (Fig. 16). *objective*

Comment: This is a common feature.

Antitragus, hyperplastic: see ***Antitragus, prominent***

Antitragus, hypertrophic: see ***Antitragus, prominent***

Antitragus, hypoplastic: see ***Antitragus, underdeveloped***

Antitragus, hypotrophic: see ***Antitragus, underdeveloped***

FIG. 13. Normal anatomy of antitragus.

Antitragus, Prominent

Definition: Increased anterosuperior prominence of the area between the bottom of the incisura and the inner margin of the antihelix (Figs. 14b [line 3], 17c). *subjective*

Comment: This finding is highly variable, and the range is illustrated in Figures 14 and 17.

Synonym: Antitragus, enlarged

Replaces: Antitragus, hyperplastic; antitragus, hypertrophic

Antitragus, small: see ***Antitragus, underdeveloped***

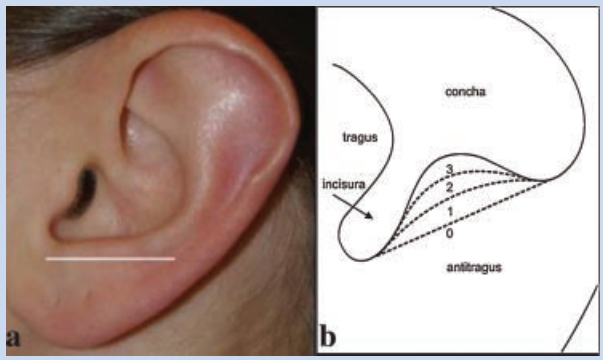

FIG. 14. Variation in size of the antitragus. a: Absent antitragus. b: The size of the antitragus is divided into 4°, of which 0 indicates absence, 1 is underdeveloped, 2 indicates the average size, and 3 indicates prominence (Fig. 14b adapted from Lange, 1966).

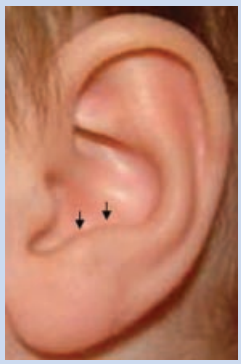

FIG. 15. Bifid antitragus.

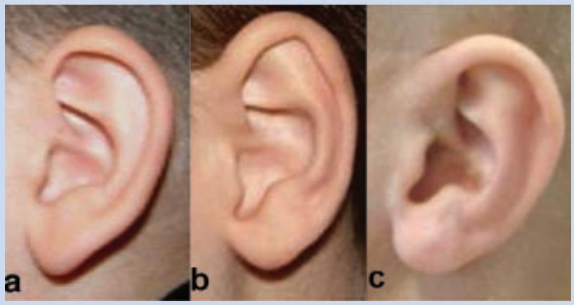

FIG. 17. Prominent and underdeveloped antitragus. a: Underdeveloped. b: Average. c: Prominent.

Antitragus, Underdeveloped

Definition: Reduction in the anterosuperior prominence of the area between the bottom of the incisura and the inner margin of the antihelix (Figs. 14b [line 1], 17a). *subjective*

Comment: This finding is highly variable, and the range is illustrated in Figures 14 and 17.

Synonym: Antitragus, small

Replaces: Antitragus, hypoplastic; antitragus, hypotrophic

Concha, Extra Fold

Definition: Folds or ridges within the concha that are distinct from the crus helix (Figs. 18–19). *objective*

Comment: These folds may occur in the absence of a well developed crus helix and can be distinguished by their separate origin and position.

Conchal shelf: see ***Antihelical shelf***

Constricted ear: see ***Question mark ear***

Cosman ear: see ***Question mark ear***

Cryptotia

Definition: Invagination of the superior part of the auricle under a fold of temporal skin (Fig. 20). *objective*

Comments: There are associated anomalies of the upper antihelix and crura. The upper one-third of the auricle is primarily affected and there is an inferomedial displacement of the ***Helical Darwin tubercle***. Two types are recognized: Type I, the antihelix and superior crus are reduced in size (Fig. 20a); Type II, it is the antihelix and inferior crus that are affected (Fig. 20b) [Hirose et al., 1985].

FIG. 16. Everted antitragus.

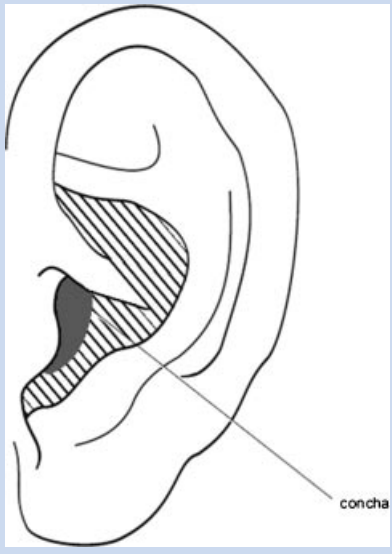

FIG. 18. Normal area of concha.

FIG. 19. Extra folds in the concha.

FIG. 21. Crumpled ear.

Ear, absent: see ***Anotia***

Ear, additional crus: see ***Antihelix, additional crus*** and ***Stahl ear***

Ear, bat: see ***Ear, protruding***

Ear, capuchin: see ***Ear, cupped***

Ear, cockleshell: see ***Microtia, second degree***

Ear, constricted helix type IV: see ***Microtia, second degree***

Ear, Crumpled

Definition: Distortion of the course of the normal folds of the ear and the appearance of supernumerary crura and folds (Fig. 21). *subjective*

Comments: This is distinct from ***Stahl ear*** and ***Shell ear***, with which the term has sometimes been conflated. The appearance often changes markedly after birth.

Ear, Cupped

Definition: Laterally protruding ear that lacks antihelical folding (including absence of inferior and superior crura) (Fig. 22). *subjective*

Replaces: Ear, Capuchin

Ear, cupped, severe/type III: see ***Microtia, second degree***

Ear, devil: see ***Satyr ear***

Ear, dog-bite: see ***Helix, underfolded***

Ear, dysplastic: The term "dysplastic" is no longer accepted as a descriptor for an ear with unusual morphology. Each specific anatomical component of the ear should be described when the ear is thought to be "abnormal" in appearance.

Ear, Focal Absence

Definition: Absence of a localized portion of the ear, when that cannot be described by a more precise term (e.g., absent ear lobe) (Fig. 23). *objective*

Comment: This definition is in the terminology set to acknowledge that there may be particular instances of absent structures not captured by the included terms. The specific affected area should be noted. For example, focal absence of the triangular fossa.

Ear, grade II dysplasia: see ***Microtia, second degree***

Ear, grade III dysplasia: see ***Microtia, third degree***

Ear, hypoplastic, group II: see ***Microtia, third degree***

Ear, long

Definition: Median longitudinal ear length greater than two SD above the mean (Fig. 24). *objective* OR

Apparently increased length of the ear. *subjective*

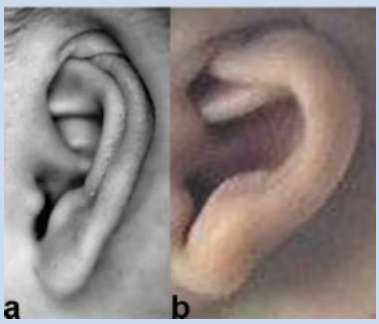

FIG. 20. Cryptotia. a. Type I; b. Type II.

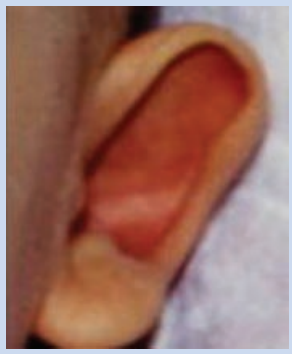

FIG. 22. Cupped ear.

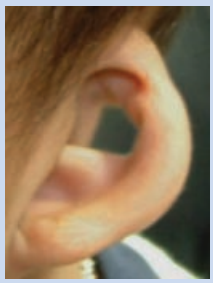

FIG. 23. Focal aplasia of ear.

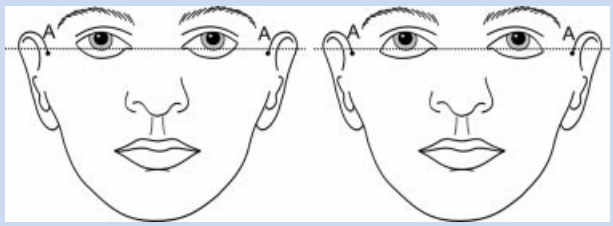

FIG. 25. Ear positioning. Please note that the position of the ears is normal in both graphs despite the difference in positioning of the outer canthi.

Comments: Ear length is determined by the maximal distance from the superior aspect to the inferior aspect of the external ear. Normal values are available from birth to 16 years of age [Feingold and Bossert, 1974; Hall et al., 2007] and birth to 18 years of age [Farkas, 1981] specific for sex. Adult values for length and width, separated by sex, are published from a sample of US Army personnel (Tech Report NATICK/TR-89/027, pp 89–90) but these are difficult to obtain. Adult Japanese data are also available [Itoh et al., 2001]. Both adult sets suggest ears increase in length into adulthood and ears in males are larger than those in females. Ears probably more often look large in relation to a small head than actually are large. For this reason we strongly support measurements in assessing ear length. Subjective assessments of ear length should only be used if unavoidable. We encourage recording ear width as well [Farkas, 1981]. In fact, the commonly used term ***Macrotia*** is a bundled term comprising increased length and width (surface area).

Ear, Low-Set

Definition: Upper insertion of the ear to the scalp below an imaginary horizontal passing through the inner canthi and extend that line posteriorly to the ear (Fig. 25). *objective*

Comments: The position of the ear can be determined in four different ways [Hall et al., 2007]. There is some controversy regarding objective assessments of ear position as methods do not place the ear with respect to a fixed plane [Hall et al., 2007]. The Frankfurt plane uses the position of the auditory meatus as a landmark and thus can not be used to assess ear position. If palpebral fissures run horizontal they can be used to guide the plane. The definition used here minimizes the problem whereby the position of the subject's head relative to the observer may influence the subjective impression of the position of the ears. Subjective assessment of ear position is unacceptable since it is unreliable and often confounded by changes in head shape, size and tilt or changes in ear anatomy, especially the superior portion.

Ear, mini: see ***Microtia, second degree***

Ear, peanut: see ***Microtia, third degree***

Ear, Posterior Angulation, Increased

Definition: Angle formed by the line perpendicular to the Frankfurt plane and the medial longitudinal axis of the ear (the two most remote points of the ear) greater than 2 SD above the mean for age (Fig. 26). *objective*

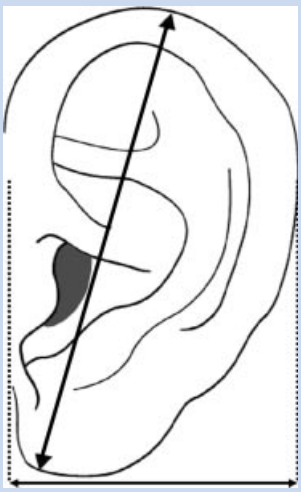

FIG. 24. Lines illustrating maximal longitudinal ear length and ear width.

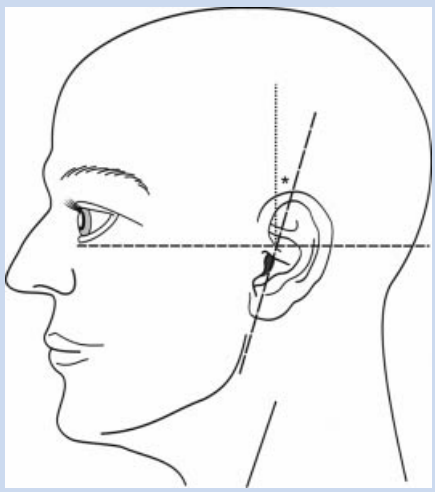

FIG. 26. Measurement of posterior rotation of the ear. Angle * is presently used to determine the degree of rotation.

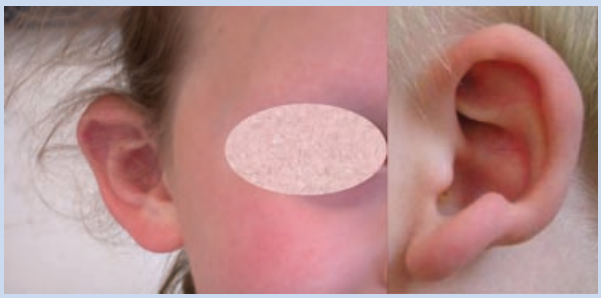

FIG. 27. Protruding ears.

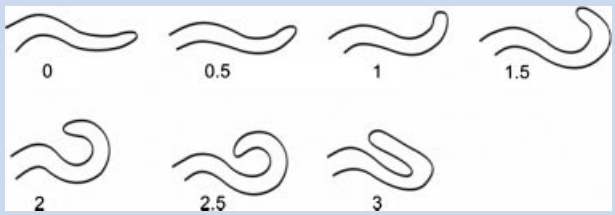

FIG. 29. Variations in folding of the helix in cross-section [adapted from Lange, 1966].

Comments: Normal values are available [Farkas, 1981; Hall et al., 2007]. The mean angle is near 20° used the Frankfurt plane is compromised if the ear is also low-set. Subjective assessment of ear rotation is unacceptable because it is unreliable and often confounded by changes in head position. Abnormalities of ear shape may make it difficult to reliably determine the medial longitudinal axis of the ear. In such cases, it is probably unwise to make an assessment of the rotational status of the ear.

Synonym: Ear, posteriorly rotated

Ear, posterior helical groove: see ***Helix, posterior pit***

Ear, posterior helical notch: see ***Helix, posterior pit***

Ear, posteriorly rotated: see ***Ear, posterior angulation, increased***

Ear, prominent: see ***Ear, protruding***

Ear, Protruding

Definition: Angle formed by the plane of the ear and the mastoid bone greater than the 97th centile for age (Fig. 27). *objective* OR

Outer edge of the helix more than 2 cm from the mastoid at the point of maximum distance. *objective*

Comments: Normal values are available [Farkas, 1981; Hall et al., 2007]. In mild cases the superior crus of the antihelix is deficient; in more severe cases the lack of normal folding may be more extensive, but these should be recorded separately. Ears that are protruding may give the appearance of increased size, but this should be assessed separately.

Synonym: Ear, prominent

Replaces: Ear, bat

Ear, shell: see ***Microtia, second degree***

FIG. 28. Normal anatomy of the helix. a: Ascending part. b: Superior part. c: Descending or posterior part.

FIG. 30. Variation in helix formation among the group of clinical geneticists involved in defining morphology.

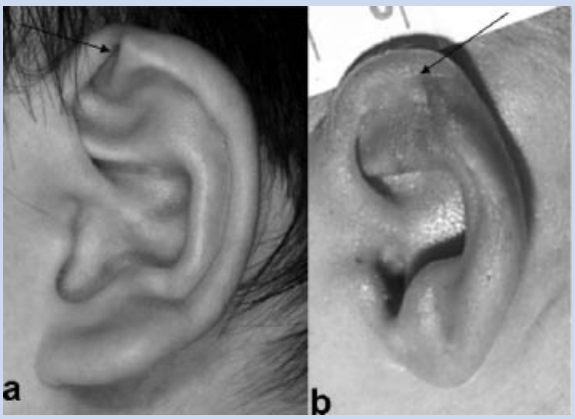

FIG. 31. Cleft helix. In b, the term notched helix is also used.

FIG. 33. Absent crus of helix.

Ear, Short

Definition: Median longitudinal ear length greater than 2 SD below the mean (Fig. 24). *objective* OR

Apparently decreased length of the ear. *subjective*

Comments: Ear length is determined by the maximal distance from the superior aspect to the inferior aspect of the external ear. Normal values are available from birth to 16 years of age [Feingold and Bossert, 1974; Hall et al., 2007] and birth to 18 years of age [Farkas, 1981] specific for sex. Adult values for length and width, separated by sex, are published from a sample of US Army personnel (Tech Report NATICK/TR-89/027, pp 89–90) but these are difficult to obtain. Adult Japanese data are also available [Itoh et al., 2001]. Both adult sets suggest ears increase in length into adulthood and ears in males are larger than those in females. Subjective assessment of the length of the ear is markedly influenced by the other craniofacial dimensions and easily distorted. For this reason we strongly support measurements in assessing ear length. Subjective assessments of shortness of the ear should only be used if unavoidable. The commonly used term ***Microtia*** is a bundled term comprising decreased length and width (surface area).

Ear, snail: see ***Microtia, second degree***

Helix, Cleft

Definition: Defect in the continuity of the helix, which may occur at any point along its length (Figs. 28, 31). *objective*

Comment: This may take the form of a sharp cleft as in Figure 31a, or a less well-demarcated area as in the outer upper portion of Figure 31b. This should be distinguished from a ***Question-mark ear***. If the defect or notch occurs at the junction of the superior and descending portions of the helix, it should be coded as ***Darwin notch of the helix***.

Synonym: Helix, notched

Helix, Crimped

Definition: Linear, circumferential indentation in the convexity of the outer surface of the helix (Fig. 32). *subjective*

Comment: The crimp is usually found in the middle third of the descending helix. The helix has the appearance of having been pinched or flattened along its posterior margin. The crimp may distort the free margin of the helix.

Synonym: Indented helix.

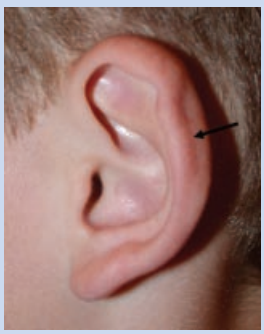

FIG. 32. Crimped helix.

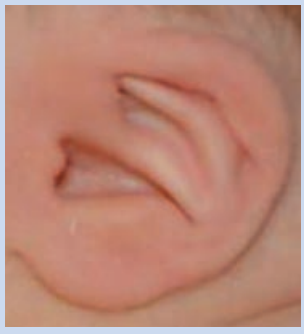

FIG. 34. Crus helix connected to antihelix.

FIG. 35. Expanded terminal portion of crus helix.

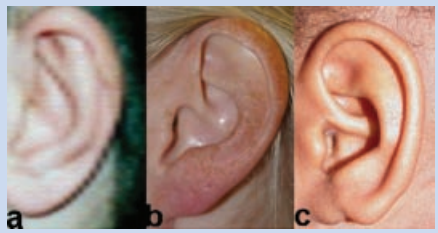

FIG. 37. Degree of development of the crus helix. a: Underdeveloped. b: Average. c: Prominent.

Helix, Crus, Absent

Definition: Continuum between the tragus and ascending helix, without any evidence of a posterior extension (crus) towards the concha (Fig. 33). *subjective*

Helix, Crus, Connected to Antihelix

Definition: Extension of the ridge of the crus helix across the ear and connection of the crus to the antihelix (Fig. 34). *objective*

Helix, Crus, Expanded Terminal Portion

Definition: Widening, rather than tapering, of the crus at its posterior border near the antihelix (Fig. 35). *subjective*

Helix, Crus, Horizontal

Definition: Main axis of the crus of the helix perpendicular to the medial longitudinal axis of the ear, instead of sloping inferoposteriorly (Fig. 36). *subjective*

Comment: The term ''railroad track sign'' has been used to describe prominent horizontal crus of the helix in combination with prominent and parallel inferior crus of the antihelix. It is preferable to simply describe each component separately.

Helix, crus, hyperplastic: see ***Helix, crus, prominent***

Helix, crus, hypertrophic: see ***Helix, crus, prominent***

Helix, crus, hypoplastic: see ***Helix, crus, underdeveloped***

Helix, crus, hypotrophic: see ***Helix, crus, underdeveloped***

Helix, Crus, Prominent

Definition: Development of the crus helix to the same degree as an average antihelix stem or helix (Fig. 37c). *subjective*

Comments: This finding is highly variable, and the range is illustrated in Figure 37. Judgment of prominence is highly subjective and may be influenced by the relative development of the other ear components. There appears to be a correlation between the length of the crus helix and its relative prominence, but these should be coded separately.

Replaces: Hypertrophic crus helix; hyperplastic crus helix

Helix, Crus, Serpiginous

Definition: Curving course of the crus of the helix, approaching or joining the antitragus (Fig. 38). *subjective*

Helix, Crus, Tragal Bridge

Definition: The anterior origin of the crus encompasses the superior margin of the tragus, the crus overrides the upper portion of the conchal cavum and ends at the antihelix (Fig. 39). *subjective*

Comment: The antihelix can also be anomalous, but this should be coded separately.

Helix, Crus, Underdeveloped

Definition: Flatter and/or shorter crus helix than average (Fig. 37a). *subjective*

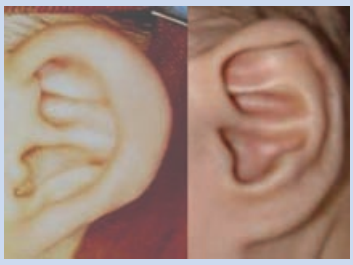

FIG. 36. Horizontal crus of helix.

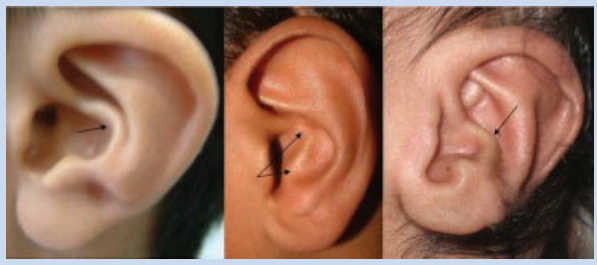

FIG. 38. Serpiginous crus of helix.

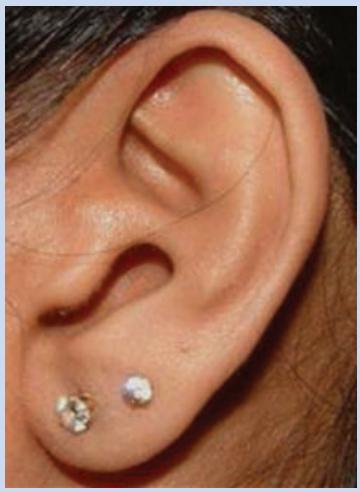

FIG. 39. Tragal bridge of crus of helix. Note the crus helix is also attached to the antihelix, which is under-developed in its lower portion.

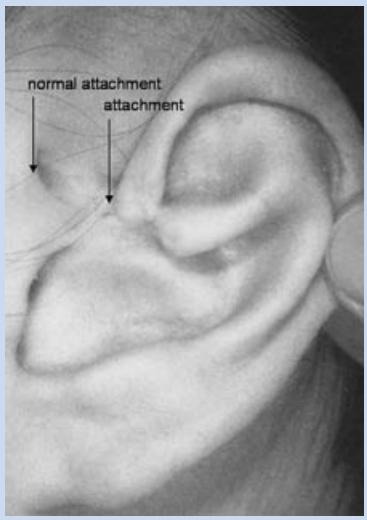

FIG. 41. Detachment of ascending part of helix (reprinted with permission from Park and Roh, 1999).

Comments: This finding is highly variable, and the range is illustrated in Figure 37. There appears to be a correlation between the length of the crus helix and its relative prominence.

Replaces: Hypoplastic crus helix; hypotrophy crus helix

Helix, Darwin Notch

Definition: Small defect of the helical fold that lies at the junction of the superior and descending portions of the helix (Fig. 40b). *objective*

Comment: Defects at other points along the helix are coded as ***Cleft helix***.

Helix, Darwin Tubercle

Definition: Small expansion of the helical fold at the junction of the superior and descending portions of the helix (Fig. 40a). *objective*

Helix, Discontinuous Ascending Root

Definition: Interruption between the ascending helix and the crus helix, allowing the ascending helix to be attached directly to the mastoid area (Fig. 41). *objective*

Comments: This is an uncommon feature.

Replaces: Helix, ascending, detachment

Synonym: Anomalous origin of ascending most of the helix.

Helix, indented: see ***Helix, crimped***

Helix, notched: see ***Helix, cleft***

Helix, overfolded

Definition: Excessive curling of the helix edge, whereby the free edge is parallel to the plane of the ear (Figs. 29 [example 3], 42). *subjective*

Comments: This is most often seen in the superior helix where it must be distinguished from a ***Lop ear*** (where the usual convexity of

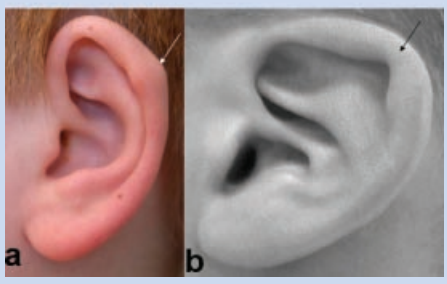

FIG. 40. Darwin tubercle (a) and Darwin notch (b).

FIG. 42. Overfolded helix.

FIG. 43. Pits in posterior helix.

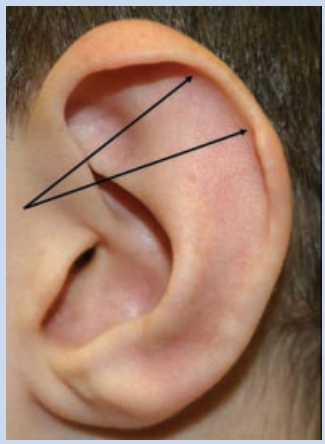

FIG. 45. Localized underdeveloped helix.

the posterior border of the ear is lost). Helix folding is highly variable, and the range is illustrated in Figures 29 and 30.

Helix, Posterior Pit

Definition: Permanent indentation on the posteromedial aspect of the helix that may be sharply or indistinctly delineated (Fig. 43). *objective*

Comment: They are usually linear to a narrow oval in shape and may be single or multiple [Prescott and Hennekam, 2007].

Replaces: Ear, posterior helical groove; ear, posterior helical notch

Helix, Squared Superior Portion

Definition: Flattening instead of curving or rounded superior helix, allowing the superior helix to run more horizontally than usual (Fig. 44). *subjective*

Comment: This is not to be confused with ***Lop ear*** or ***Satyr ear*** and may represent an underdevelopment of the upper third of the pinna. This is usually associated with a short ascending helix.

Helix, Underfolded

Definition: Underdevelopment of the helix that either affects the entire helix, or is localized (Fig. 45). *subjective*

Comment: Helix folding is highly variable, and the range is illustrated in Figures 29 and 30. To use this term, the affected area must be too long to be considered a ***Cleft helix***. Underdevelopment of part of the helix can lead to the impression that the scaphal area is enlarged.

Replaces: Ear, dog-bite

Lobe, Absent

Definition: Absence of fleshy non-cartilaginous tissue inferior to the tragus and incisura (Figs. 46 and 47). *objective*

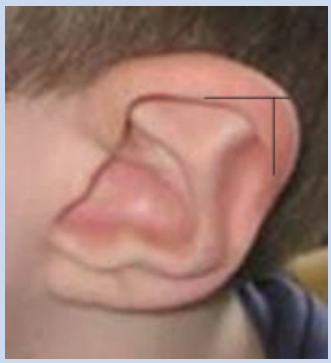

FIG. 44. Squared superior portion of helix.

FIG. 46. Normal anatomy of the lobe.

FIG. 47. Absent lobe.

FIG. 49. Attached lobe.

Comments: See ***Attached lobe*** for a finding that should be distinguished from ***Absent lobe***.

Lobe, Anterior Crease(s)

Definition: Sharply demarcated, typically linear and approximately horizontal, indentations in the outer surface of the ear lobe (Fig. 48). *subjective*

Comment: Shallow grooves or indentations are quite common, especially in large lobes. Ear lobe creases may arise postnatally [Chitayat et al., 1990]. ***Posterior helical pits*** can be a related finding but should be assessed and coded separately.

Lobe, Attached

Definition: Attachment of the lobe to the side of the face at the lowest point of the lobe without curving upward (Fig. 49). *objective*

Comment: The earlobe does not have a dependent portion.

Lobe, bifid: see ***Lobe, cleft***

Lobe, Cleft

Definition: Discontinuity in the convexity of the inferior margin of the lobe (Fig. 50). *objective*

Comment: The cleft is often more visible if the lobe is pulled forward or when seen from behind. Tears acquired from earrings should be distinguished.

Synonym: Bifid lobe; notched lobe

Lobe, fleshy: see ***Lobe, large***

Lobe, Forward Facing

Definition: Positioning of the anterior surface of the ear lobe in a more coronal plane than the remainder of the ear (Fig. 51). *subjective*

Comment: The lobe should be viewed from the front. This feature is distinct from the situation where the entire ear is forward facing and prominent (as shown in Fig. 27). The lobe normally lies more or less in the same plane as the remainder of the ear. This feature should be distinguished from ***Uplifted lobe***.

Lobe, hyperplastic: see ***Lobe, large***

Lobe, hypertrophic: see ***Lobe, large***

Lobe, hypoplastic: see ***Lobe, small***

Lobe, hypotrophic: see ***Lobe, small***

FIG. 48. Anterior creases in lobe.

FIG. 50. Cleft lobe.

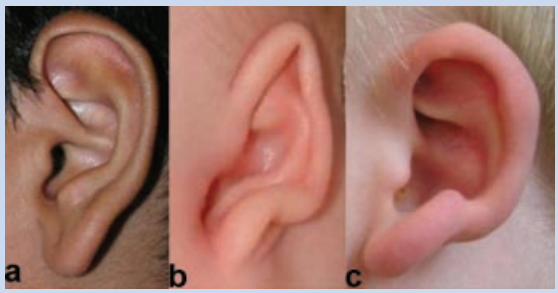

FIG. 51. Forward facing lobe. Note that in (c) the lobe is also uplifted.

Lobe, Large

Definition: Increased volume of the earlobe (Fig. 52d,e). *subjective*

Comments: All gradations in size of the earlobe may be seen from absent to clearly enlarged compared to average. This finding is highly variable, and the range is illustrated in Figure 52. Lobe size increases throughout adulthood [Ferrario et al., 1999; Itoh et al., 2001].

Replaces: Hyperplastic lobe; hypertrophic lobe; fleshy lobe

Lobe, notched: see ***Lobe, cleft***

Lobe, Small

Definition: Reduced volume of the earlobe (Fig. 52a,b). *subjective*

Comments: All gradations in size of the earlobe may be seen from absent to clearly enlarged compared to average. This finding is highly variable, and the range is illustrated in Figure 52.

Replaces: Hypoplastic lobe; hypotrophic lobe

Lobe, Uplifted

Definition: Lateral surface of ear lobe faces superiorly (Fig. 53). *subjective*

Synonym: ***Lobe, upturned***

Lobe, upturned: see ***Lobe, uplifted***

Lop Ear

Definition: Anterior and inferior folding of the upper portion of the ear that obliterates triangular fossa and scapha (Fig. 54). *subjective*

Comments: Mild forms are limited to the superior ear, more severe forms affect the superior and posterior ear. The concha may be excessively concave. This should be distinguished from an ***Overfolded helix*** where the external contour of the ear is normal.

Macrotia

Definition: Median longitudinal ear length greater than 2 SD above the mean and median ear width greater than 2 SD above the mean (Fig. 24). *objective* OR

Apparently increase in length and width of the pinna. *subjective*

Comment: This is acknowledged to be a bundled term but retained here because of its usefulness in practice. Ear length is determined by the maximal distance from the superior aspect to the inferior aspect of the external ear. Normal values are available from birth to 16 years of age [Feingold and Bossert, 1974; Hall et al., 2007] and birth to 18 years of age [Farkas, 1981] specific for sex. If only length is increased the term ***Long ear*** should be used. Normal values for ear width are available [Farkas, 1981].

Microtia, First Degree

The definitions of microtia below follow a widely used, surgically based, classification of ear anomalies outlined by Weerda [1988]. As

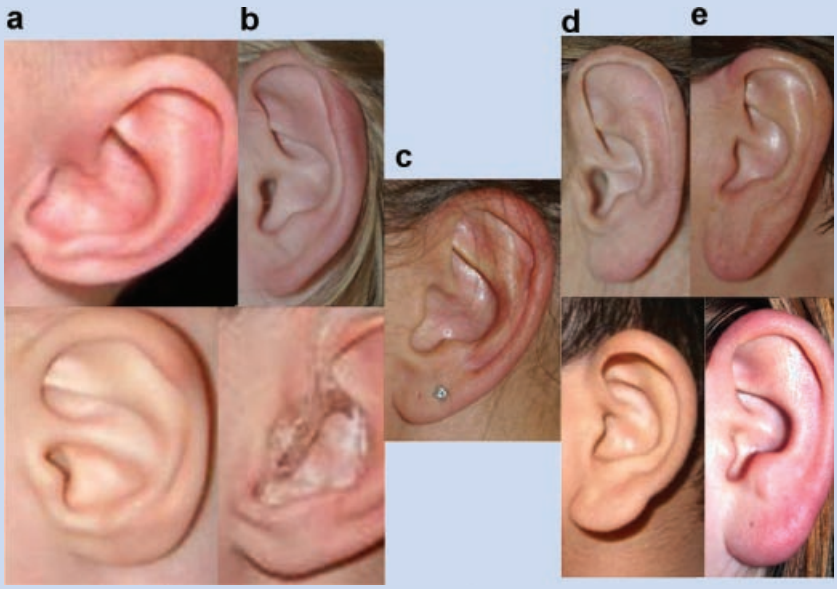

FIG. 52. Variations in volume of the earlobe. a: Very small. b: Small. c: Average. d: Large. e: Very large.

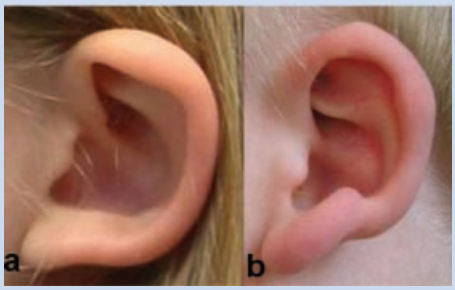

FIG. 53. Uplifted lobe; note that (b) is also forward-facing.

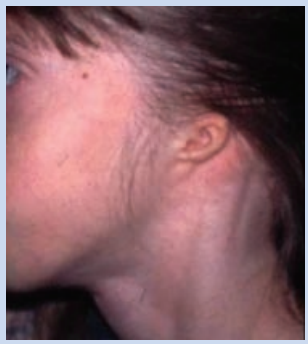

FIG. 55. Microtia, first degree.

microtia indicates at least both decreased length and width, and in more severe forms it includes abnormal shape of structures, all forms are acknowledged to be bundled terms, but are retained here because they are well established.

Definition: Presence of all the normal ear components and the median longitudinal length more than 2 SD below the mean (Fig. 55). *objective*

Comments: See ***Short ear*** for a discussion of altered ear length. A better assessment of size would include both length and width (i.e., an estimate of surface area). Normal values for width and length measurements are available [Farkas, 1981; Hall et al., 2007].

Microtia, Second Degree

Definition: Median longitudinal length of the ear more than 2 SD below the mean in the presence of some, but not all, parts of the normal ear (Fig. 56). *subjective*

Replaces: Ear, grade II dysplasia; ear, cupped severe, type III; ear, cockleshell; ear, constricted helix type IV; ear, snail; ear, shell; ear, mini

Microtia, Third Degree

Definition: Presence of some auricular structures, but none of these structures conform to recognized ear components (Fig. 57). *objective*

Comments: This malformation is commonly associated with atresia of the external canal, but that anomaly should be coded separately. Complete absence of the ear should be coded as ***Anotia***.

Replaces: Ear, grade III dysplasia; ear, hypoplastic, group II; ear, peanut

Pit, Auricular

Definition: Small indentation in the lower part of the ascending helix, concha, or in the crus helix (Fig. 58). *objective*

Comment: The location of the pits is the plane of fusion of the first branchial cleft [Wood-Jones and I-Chuan, 1933].

Pit, Preauricular

Definition: Small indentation anterior to the insertion of the ear (Fig. 59). *objective*

Comment: The location of these pits is the plane of fusion of the first branchial cleft [Wood-Jones and I-Chuan, 1933].

Polyotia: see ***Pretragial ectopia***

Pretragal duplication: see ***Pretragial ectopia***

Pretragal Ectopia

Definition: Variably shaped, cartilage-containing tissue anterior to the external auditory meatus (Fig. 60). *objective*

Comment: These structures are frequently complex and should be distinguished from ***Preauricular tags***. They may be difficult to distinguish from striated muscle hamartomas or ***Tragal duplications***. ***Pretragal ectopias*** often appear helix-like (Fig. 61a), and in such cases may be called ***Polyotia*** (Fig. 61c).

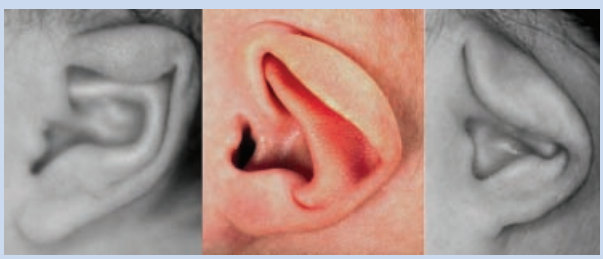

FIG. 54. Lop ear.

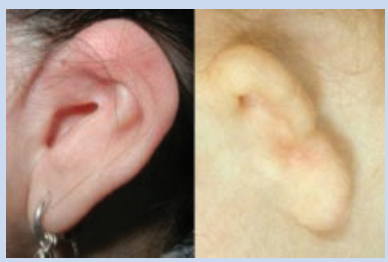

FIG. 56. Microtia, second degree.

FIG. 57. Microtia, third degree.

Synonyms: Pretragal duplication; polyotia
Replaces: Accessory tragus

Quelprud Nodule

Definition: Small cartilaginous prominence on the posterior concha (Fig. 61). *objective*

Comments: This is best visualized when the lobe is tilted anteriorly.

Question Mark Ear

Definition: Cleft between the helix and the lobe (Fig. 62). *subjective*

Comments: Relatively few cases have been reported [Priolo et al., 2000]. Variation from a small notch to complete separation of the helix from the lobe is noted, there may be unilateral or bilateral involvement. The lobe is relatively laterally recessed compared to the upper portion of the ear and the scapha may be absent. This is distinct from a ***Cleft helix*** where the cleft is within the helix.

Synonym: Cosman ear; constricted ear

Satyr Ear

Definition: Sharp pointed superior portion of the ear, with variable overfolding of the helix (Fig. 63). *subjective*

Comments: The satyr ear appears to have an abnormally small upper-lateral portion. More extensive underdevelopment continuing down to and including the lobe produces a more extreme anomaly that, unfortunately, has been called Devil ear.

Replaces: Ear, devil

FIG. 58. Auricular pits.

FIG. 59. Preauricular pits.

Shell Ear

Definition: Absence of the superior antihelical crus, and broadening of the inferior antihelical crus, which runs more horizontally with a sharper "take-off" from the helix than usual (Fig. 64). *subjective*

Comments: This is a bundled term, consisting of absence of the superior antihelical crus, broad inferior antihelical crus and abnormal orientation of the antihelix. As the term is much used in practice it is kept. In addition, the helix may show variable abnormalities of folding. The crus helix may be more protruding and extend further across the concha than usual.

Stahl Ear

Definition: Third crus arising at or above the normal bifurcation of the antihelix (Fig. 65). *objective*

Comments: The helix is often poorly formed. Four types have been recognized in the surgical literature [Yamada and Fukuda, 1980], but are not further delineated here.

Synonym: Antihelix, third crus; ear, additional crus

Tag, Auricular

Definition: Small protrusion within the pinna (Fig. 66). *objective*

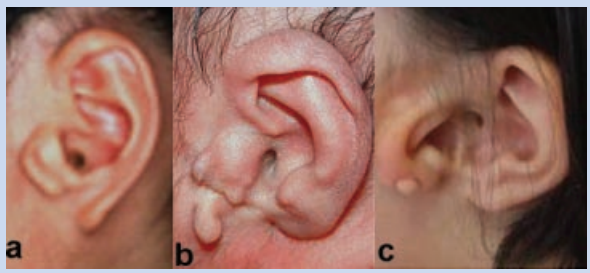

FIG. 60. Pretragial ectopias; in (a) note the resemblance of the ectopia to normal helix; in (c) there is clear duplication of hyoid components in this example of polyotia. Note the hair on part of the duplication in (b), which is typical of the infant helix but not the tragus. Note also that neither figure shows presence of a normal tragus. ((c) Courtesy of Dr Sergio B. De Sousa).

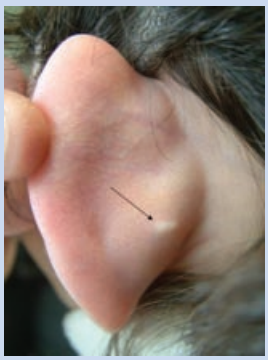

FIG. 61. Quelprud nodule.

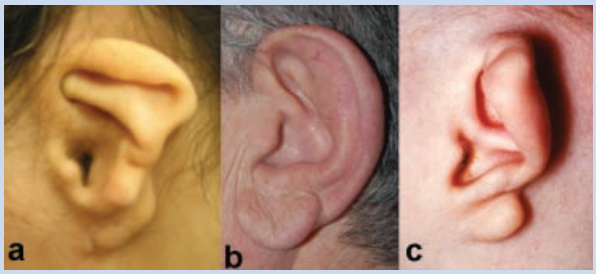

FIG. 62. Question mark ear in (a); (b) represents a minor form. c: Courtesy of Dr Alison Stewart.

Comment: The tag can be located on either side of the pinna.

Tag, Preauricular

Definition: Small non-cartilaginous protrusion anterior to the insertion of the ear (Fig. 67). *objective*

Comment: The location of these tags is the plane of fusion of the first branchial cleft [Wood-Jones and I-Chuan, 1933]. At times it can be a challenge to distinguish other pedunculated lesions in this area; specifically duplications of ear components, ***Pretragal ectopias***, or a striated muscle hamartoma. Preauricular tags usually lack hair, are limited to the plane of fusion, and do not contain striated muscle.

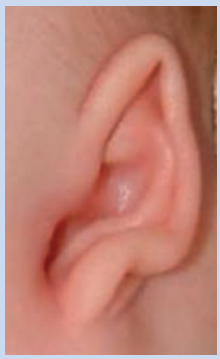

FIG. 63. Satyr ear.

FIG. 64. Shell ear.

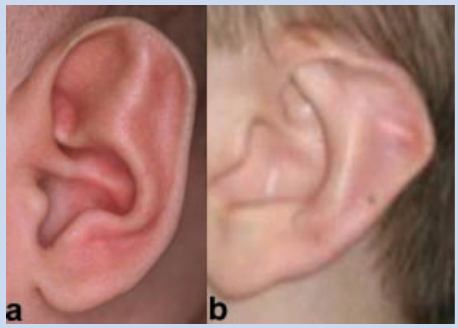

FIG. 65. Stahl ear.

Tragus, Absent

Definition: Lack of convexity or prominence of the contour of the ridge between the bottom of the incisura and the confluence of the ascending helix and crus helix (Figs. 68 and 69). *objective*

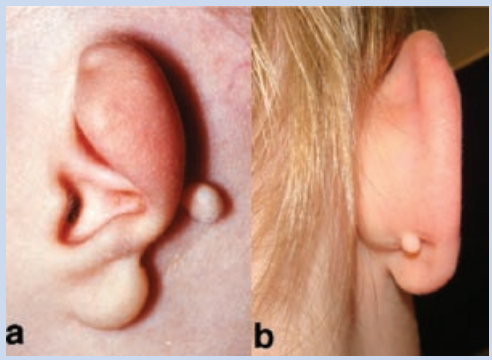

FIG. 66. Auricular tag. Both panels show a postauricular tag; (a) is in a question mark ear. a: courtesy of Dr Alison Stewart.

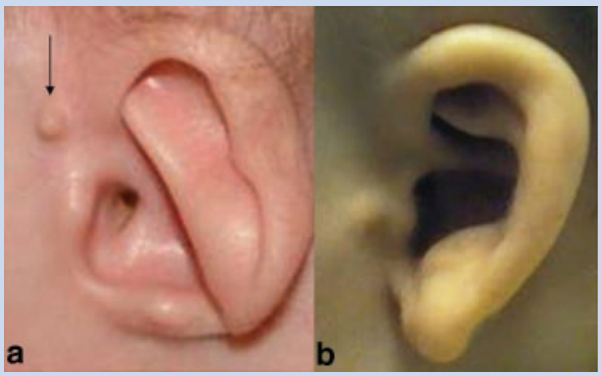

FIG. 67. Preauricular tags in (a) typical. In (b), the location outside the plane of mandibular-hyoid fusion suggests this is a duplicated tragus rather than a preauricular tag.

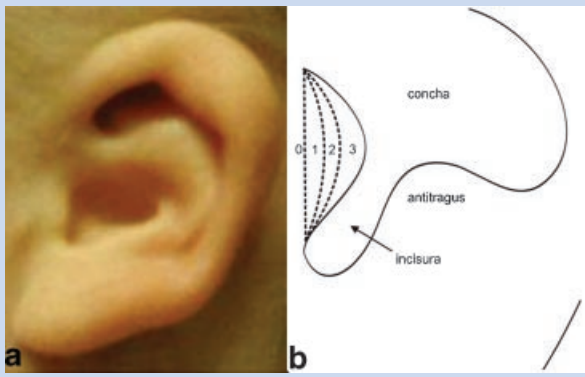

FIG. 69. Variability in the size of the tragus. a: Absent tragus. b: The size of the tragus is divided into 4°, of which 0 indicates absence, 1 is underdeveloped, 2 indicates the average size, and 3 indicates prominence (modified from Lange, 1966).

Comment: See Lange [1966]. This appears to be unusual in an otherwise normal ear, and is most often seen in microtia with atretic auditory meatus, but those findings should be coded separately.

Tragus, accessory: see ***Tragus, duplicated***

Tragus, Bifid

Definition: Increased height of the tragal ridge with a shallow indentation at the apex, giving the appearance of a double peak (Fig. 70). *objective*

Synonym: Tragus, notched

Tragus, Duplicated

Definition: A complete or partial duplication of the tragus; expected to lie anterior to the normal tragus (Fig. 67b). *objective*

Comment: It is unclear how often, or even whether, this feature which would represent a duplication of mandibular components, occurs. More common occurrences in this region would include preauricular tags and pretragial duplications of hyoid origin.

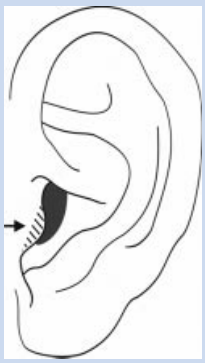

FIG. 68. Normal anatomy of the tragus.

Synonym: Accessory tragus

Tragus, enlarged: see ***Tragus prominent***

Tragus, hyperplastic: see ***Tragus prominent***

Tragus, hypertrophic: see ***Tragus prominent***

Tragus, hypoplastic: see ***Tragus underdeveloped***

Tragus, hypotrophic: see ***Tragus underdeveloped***

Tragus, notched: see ***Tragus, bifid***

Tragus, Prominent

Definition: Increase posterolateral protrusion of the tragus (Figs. 69 [line 3] and 71d). *subjective*

Comment: This finding is highly variable, and the range is illustrated in Figures 69 and 71.

Synonym: Enlarged tragus

FIG. 70. Bifid tragus.

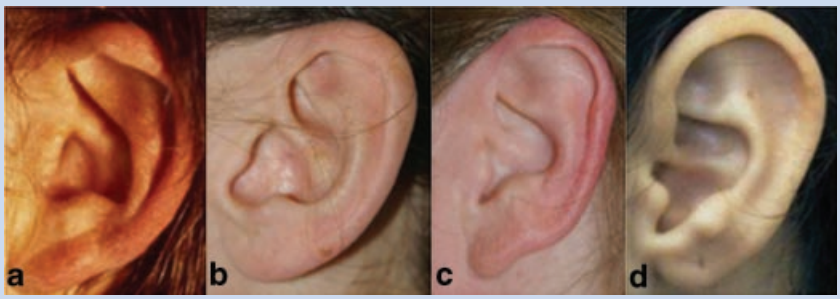

FIG. 71. Degree of prominence of the tragus. a: Very underdeveloped. b: Mildly underdeveloped. c: Average. d: Prominent.

Replaces: Hyperplastic tragus; hypertrophic tragus

Tragus, small: see ***Tragus underdeveloped***

Tragus, Underdeveloped

Definition: Decreased posterolateral protrusion of the tragus (Figs. 69b [line 1] and 71a,b). *subjective*

Comment: This finding is highly variable, and the range is illustrated in Figures 69 and 71.

Synonym: Small tragus

Replaces: Hypoplastic tragus; hypotrophic tragus

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RESEARCH ARTICLE

AMERICAN JOURNAL OF medical genetics PART A

Elements of Morphology: Standard Terminology for the Nose and Philtrum

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An international group of clinicians working in the field of dysmorphology has initiated the standardization of terms used to describe human morphology. The goals are to standardize these terms and reach consensus regarding their definitions. In this way, we will increase the utility of descriptions of the human phenotype and facilitate reliable comparisons of findings among patients. Discussions with other workers in dysmorphology and related fields, such as developmental biology and molecular genetics, will become more precise. Here we introduce the anatomy of the nose and philtrum, and define and illustrate the terms that describe the major characteristics of these body regions. © 2009 Wiley-Liss, Inc.

Key words: nomenclature; definitions; nose; philtrum; anatomy; anthropometry; morphology; dysmorphology

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INTRODUCTION

General

This paper is part of a series of six papers defining the morphology of regions of the human body [Allanson et al., 2009b; Biesecker et al., 2009; Carey et al., 2009; Hall et al., 2009; Hunter et al., 2009]. The series is accompanied by an introductory paper describing general aspects of this study [Allanson et al., 2009a]. The reader is encouraged to consult the introduction when using the definitions.

Anatomy of the Nose

The normal anatomy of the nose is shown in Figures 1–3: the various terms used for nasal structures and dimensions (Fig. 1), the cartilaginous components of the nose and terms for regions (Fig. 2), and in Figure 3 the cross section is shown.

Some anatomical landmarks deserve specific mention as these are not always used with standard meaning.

Nasal root: the most depressed, superior part of the nose along the nasal ridge.

Nasion: the midline point just superior to the nasal root overlying the naso-frontal suture.

Nasal bridge: A saddle-shaped area that includes the nasal root and the lateral aspects of the nose. It lies between the glabella and the inferior boundary of the nasal bone, and extends laterally to the inner canthi.

Nasal ridge: the midline prominence of the nose, extending from the nasal root to the tip (also called the dorsum of the nose).

Nasal base: an imaginary line between the most lateral points of the external inferior attachments of the alae nasi to the face.

Nasal tip: the junction of the inferior margin of the nasal ridge and the columella. Commonly, it is the part of the nose furthest from the plane of the face. In rare circumstances, such as markedly prominent and convex nasal profiles, other parts of the ridge may be further removed from the facial plane.

Ala: the tissue comprising the lateral boundary of the nose, inferiorly, surrounding the naris.

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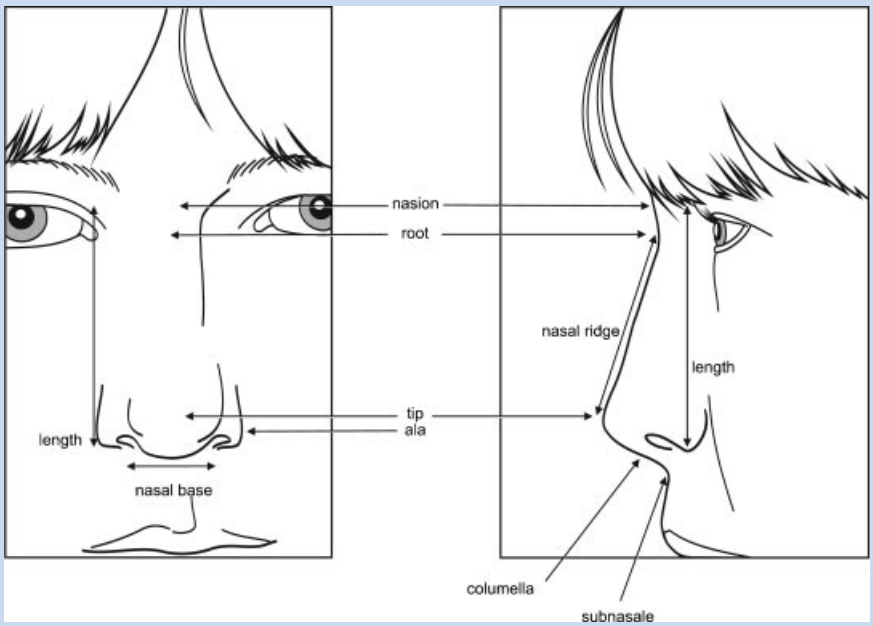

FIG. 1. Anatomy of the nose: Landmarks and distances.

Columella: the tissue that links the nasal tip to the nasal base, and separates the nares. It is the inferior margin of the nasal septum.

Measurements of the Nose

Measurements of the nose are possible using sliding calipers. The reliability of measurements using a tape measure is poor. Furthermore, the actual position of several of the landmarks may preclude accurate measurement. For example, if the nasal tip overhangs the upper lip, the position of subnasale is difficult to define [Hall et al., 2007]. Nasal length and width are the most common measurements taken in practice. A short description of how to measure each dimension is provided as the various terms are defined.

Growth of the nose does not end at puberty: the nose continues to increase in size with age. There are no normal standards for nasal size in adulthood.

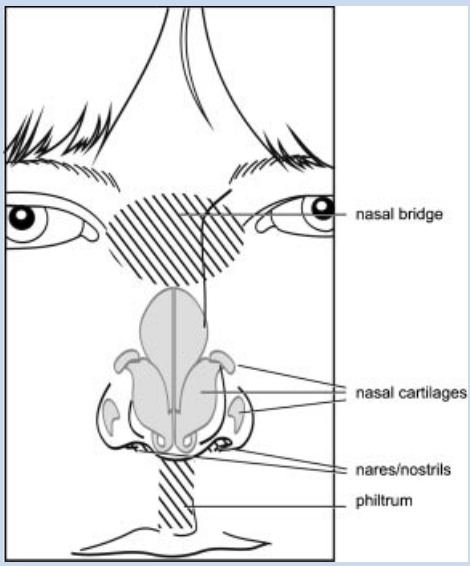

FIG. 2. Anatomy of the nose: Areas.

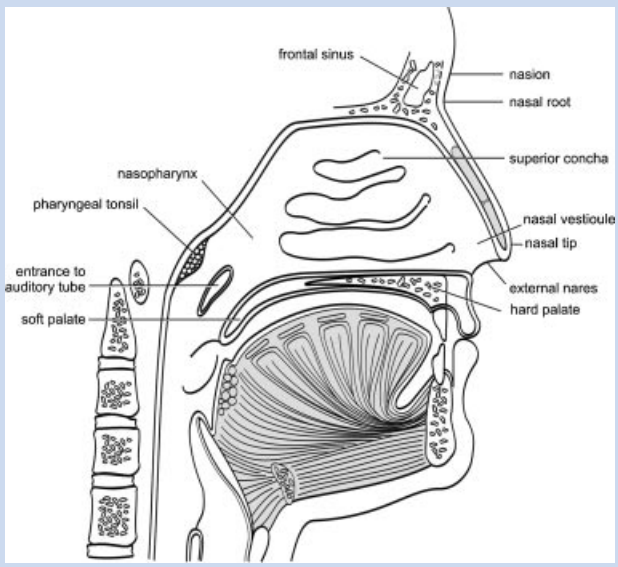

FIG. 3. Anatomy of the nose: Cross section.

The reader is referred to the *Handbook of Normal Physical Measurements* [Hall et al., 2007] for detailed descriptions of measurement techniques, additional nasal measurements not described here, and growth standards. Most are available for Caucasians of Northern European extraction only, and similar standards for other ethnicities are urgently needed.

Anatomical Variation

Anomalies of the nose may be classified into quantitative traits and qualitative features:

1. Variations in length: long; short
2. Variations in width: wide nose; narrow nose, broad nasal base; narrow nasal base; broad nasal tip; narrow nasal tip; wide nasal ridge; narrow nasal ridge; wide nasal bridge; narrow nasal bridge; broad columella.
3. Variations in length and width: prominent nose; absent nasal cartilage; absent nose.
4. Variations in shape or position: depressed nasal bridge; depressed nasal ridge; depressed nasal tip; bulbous nose; bifid nasal tip; bifid nose; overhanging nasal tip; deviated nasal tip; fullness of paranasal tissue; prominent nasal bridge; convex nasal ridge; concave nasal ridge; low insertion of the columella; low hanging columella; short columella; high insertion of the columella; thick ala nasi; underdeveloped ala nasi; cleft ala nasi; enlarged naris; narrow naris; single naris; proboscis; supernumerary naris; anteverted nares.

The various features are listed alphabetically. If a feature is indicated in ***bold-italics***, the feature is listed and a definition is available. This can be in the present or one of the accompanying papers. The terms are alphabetized based on the physical feature, not the modifier.

The appearance of facial morphology varies considerably with the position of the observer and observed person, and facial movements. In assessing morphology, the head of the observed person should be held in the Frankfurt horizontal, with the facial and neck muscles relaxed, eyes open, lips making gentle contact, and a neutral facial expression. The face of the observer should be at the same height as the face of the observed person.

DEFINITIONS

Ala Nasi, Cleft

Definition: Notch in the margin of the ala nasi (Fig. 4). *subjective*

Comments: The alae nasi are the lateral portions of the nose or the wings of the nostrils, which partly encircle the nostrils (nares). They are usually about the width of the columella, but vary greatly depending on the shape of the nostril. Note that individuals may be described as having a cleft ala nasi, but have a severely ***Underdeveloped ala nasi***. In an underdeveloped ala the continuity of the tissue encircling the nostril is undisturbed while in a cleft it is disrupted. The term coloboma has been replaced because a coloboma is a remnant of a physiologically occurring discontinuity of tissue; the alae nasi do not have such a discontinuity.

Synonym: Ala nasi, notched

FIG. 4. Cleft alae nasi. Please note the difference with an underdeveloped ala nasi in Figure 6. Left hand and middle panel courtesy of Dr. Jenneke van den Ende and Dr. Yolande van Bever.

Replaces: Ala nasi, coloboma

Ala nasi, coloboma: see ***Ala nasi, cleft***

Ala nasi, hypoplastic: see ***Ala nasi, underdeveloped***

Ala nasi, notched: see ***Ala nasi, cleft***

Ala Nasi, Thick

Definition: Increase in bulk of the ala nasi (Fig. 5). *subjective*

FIG. 5. Variation in thickening of the alae nasi: a, normal, b, mildly thickened, c, more expressed thickening, d, marked thickening. Note that also the extension of the alae towards the philtral ridges increases with increasing alar thickness.

Ala Nasi, Underdeveloped

Definition: Thinned, deficient, or excessively arched ala (Fig. 6). *subjective*

Comments: The alae nasi are the lateral portions of the nose or the wings of the nostrils, which partly encircle the nostrils (nares). They are usually about the width of the columella, but vary greatly depending on the shape of the nostril. Note that individuals may be described as having a ***Cleft ala nasi***, but have a severely underdeveloped ala nasi. In an underdeveloped ala the continuity of the tissue encircling the nostril is undisturbed while in a cleft it is disrupted.

Replaces: Ala nasi, hypoplastic

Columella below alae nasi: see ***Columella, low hanging***

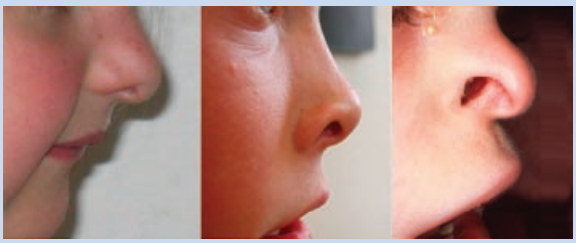

FIG. 6. Underdeveloped alae nasi. This feature is best assessed in a side view.

Columella, Broad

Definition: Increased width of the columella (Fig. 7). *subjective*

Comments: The columella should be viewed from below. This feature should be distinguished from ***Narrow nares*** although both may be present in a given individual.

Synonym: Columella, wide

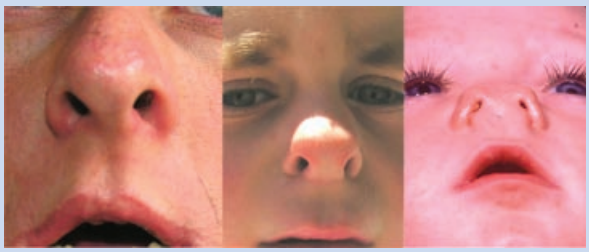

FIG. 7. Broad columella. Please note that in the right panel not only the columella is broad but there are also soft tissue swellings on each side of the columella.

Columella, High Insertion

Definition: Insertion of the posterior columella superior to the nasal base (Fig. 8). *objective*

Columella, low: see ***Columella, low hanging***

FIG. 8. High insertion of the columella. Note that it is usually impossible to determine whether the alar attachment to the face is lower than usual or the columella higher.

Columella, Low Hanging

Definition: Columella extending inferior to the level of the nasal base, when viewed from the side (Fig. 9). *subjective*

Comments: This feature may occur with or without low insertion of the columella. It may be confused with an ***Overhanging nasal tip*** which may co-occur but the two should be assessed and coded separately.

Synonym: Columella below alae nasi; Columella, low

Replaces: Columella, rounded

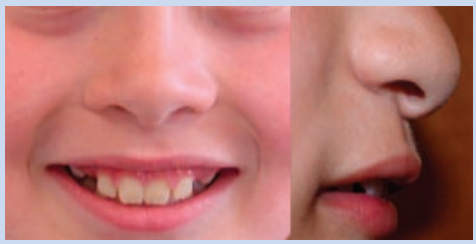

FIG. 9. Low hanging columella. Please note in the left panel that there is also a low insertion of the columella.

Columella, Low Insertion

Definition: Insertion of the posterior columella below the nasal base (Fig. 10). *objective*

Comments: This feature is different from a convex ***Low hanging columella*** that has a normal insertion. It may be associated with a ***Short philtrum***, but this should be assessed and coded separately. A low insertion is best appreciated when viewed from the side.

Columella, rounded: see ***Columella, low hanging***

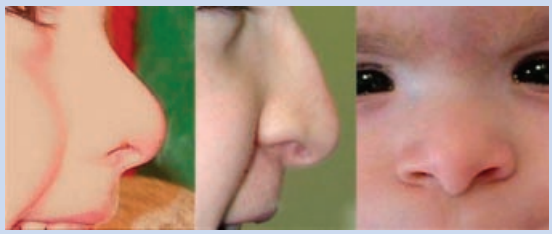

FIG. 10. Low insertion of the columella. Please note the difficulty to discern this from a low hanging columella in the frontal view in the right panel.

Columella, Short

Definition: Reduced distance from the anterior border of the naris to the subnasale (Fig. 11). *subjective*

Comment: This is often accompanied by a ***Depressed nasal tip*** but this should be assessed and coded separately. The term "Absent columella" has been deleted because a columella is thought to always be present, except in ***Single naris*** and ***Proboscis***.

Columella, wide: see ***Columella, broad***

Laterally built up nose: see ***Paranasal tissue, fullness***

FIG. 11. Short columella. Note that the two panels on the right show the same child, but only in the most right-hand panel the feature is clear.

Nares, Anteverted

Definition: Anteriorly-facing nostrils viewed with the head in the Frankfurt horizontal and the eyes of the observer level with the eyes of the subject (Fig. 12). *subjective*

Comments: The tip of the nose is upturned and is positioned superiorly to the nasal base, allowing the nares to be easily visualized from the front. With maturation and growth of the nasal ridge and tip, the nares usually become more downwardly directed.

Synonym: Nasal tip, upturned

Replaces: Pug nose

Naris, broad: see ***Naris, enlarged***

FIG. 12. Anteverted nares.

Naris, Enlarged

Definition: Increased aperture of the nostril (Fig. 13). *subjective*

Comments: The nostrils or nares are typically symmetric, wide openings. They should be assessed with the face at rest to avoid the effect of flared alae nasi that occurs with distress. Note that the nares change with age, from rounded in infancy to elongated at a later age.

Synonym: Naris, broad

Naris, flared: The term "flared naris" is not defined here as it is a functional characteristic (see ***Naris, enlarged***)

FIG. 13. Enlarged nares. Note subtleness of the enlargement in the left panel. The feature should be present with the face in a neutral position.

Naris, Narrow

Definition: Slender, slit-like aperture of the nostril (Fig. 14). *subjective*

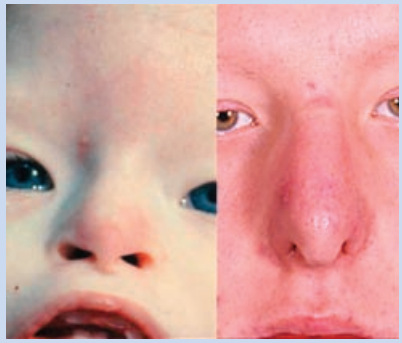

FIG. 14. Narrow nares. Note that the longest axis of the nares has a different direction in the two panels.

Comment: The shape of the nostrils has been classified into four main types by Paul Topinard (1830–1911) (Fig. 15). This classification is not in general use.

Synonym: Naris, thin; Naris, slit-like

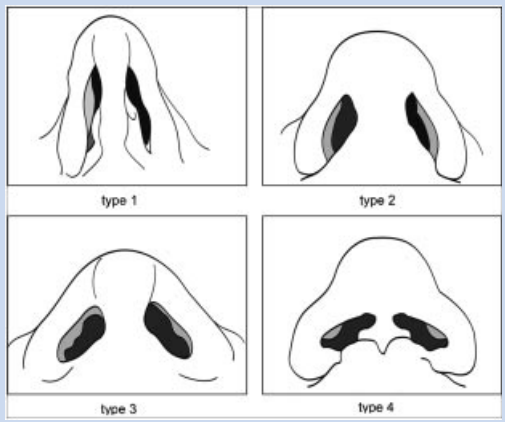

FIG. 15. Topinard classification of nostril shape. Type 2 is the most common type in the general population.

Naris, Single

Definition: One external opening of the nose (Fig. 16). *objective*

Comments: The single opening may occur in the midline or it can occur on one side, that is, it can be symmetric or asymmetric. Although the columella is invariably absent, this is implicit and does not need to be separately specified.

Naris, slit-like: see ***Naris, narrow***

FIG. 16. Single naris. Note positioning in the midline in left and middle panel, and on the left side in right panel. The patient in the right panel has a heminasal agenesis.

Naris, Supernumerary

Definition: More than two nares (Fig. 17). *objective*

Naris, thin: see ***Naris, narrow***

Nasal base, broad: see ***Nasal base, wide***

FIG. 17. Supernumerary nares. The right panel shows a completely duplicated nose.

Nasal Base, Narrow

Definition: Decreased distance between the attachments of the alae nasi to the face (Fig. 18). *subjective*

Comment: There is a marked difference in width of the nasal base depending on ethnic background

Nasal Base, Wide

Definition: Increased distance between the attachments of the alae nasi to the face (Fig. 19). *subjective*

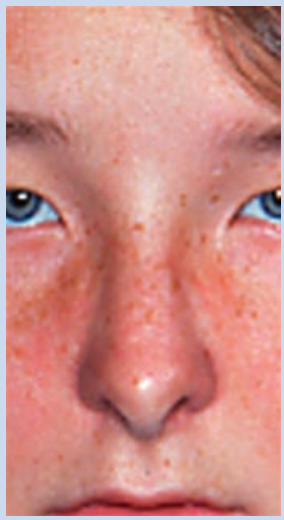

FIG. 18. Narrow nasal base.

FIG. 19. Wide nasal base. Please note that in the right panel the picture is taken slightly from above allowing the overhanging nasal tip to obscure almost completely the broad nasal base, so this would not be a useful figure to show this feature.

Comment: There is a marked difference in width of the nasal base depending on ethnic background

Synonym: Nasal base, broad

Nasal bridge, broad: see ***Nasal bridge, wide***

Nasal bridge, decreased protrusion: see ***Nasal bridge depressed***

Nasal Bridge, Depressed

Definition: Posterior positioning of the nasal root in relation to the overall facial profile for age (Fig. 20). *subjective*

Comments: The adjective "depressed" here does not indicate an active process but a status. A depressed nasal bridge can occur irrespective of the width of the nasal bridge, and the width should be assessed independently. In infancy, the nasal bridge is relatively more posterior than in the older person. The term depressed nasal

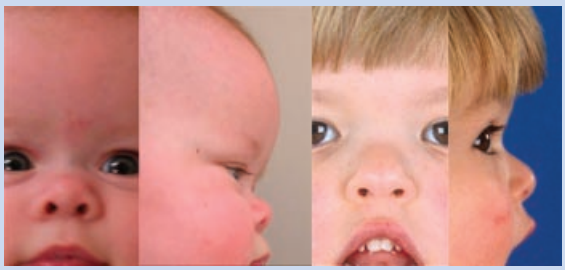

FIG. 20. Depressed nasal bridge. Although the nasal bridge in infants and toddlers is still at a more posterior position compared to older children and adults, the bridge can be tagged here depressed as it is more posterior compared to peers.

bridge should only be used when the bridge is more posterior than is typical for age and ethnic background.

Synonym: Protrusion of the nasal bridge, decreased; Nasal bridge, retruded; Nasal bridge, recessed; Nasal root, depressed; Nasal root, recessed

Replaces: Snub nose; Nasal bridge, low; Nasal bridge, flat

Nasal bridge, flat: see ***Nasal bridge, depressed***

Nasal bridge, high: see ***Nasal bridge, prominent***

Nasal bridge, low: see ***Nasal bridge, depressed***

Nasal Bridge, Narrow

Definition: Decreased width of the bony bridge of the nose (Fig. 21). *subjective*

Comments: The narrowness may be accompanied by a sharp, keel-shaped appearance. The nasal bridge may narrow with age.

Synonym: Nasal bridge, thin

FIG. 21. Narrow nasal bridge. Sometimes just the nasal bridge is narrow, like in the middle panel, and sometimes both nasal bridge and ridge are narrow, like in the right panel.

Nasal Bridge, Prominent

Definition: Anterior positioning of the nasal root in comparison to the usual positioning for age (Fig. 22). *subjective*

Comments: A prominent nasal bridge can occur irrespective of the width of the nasal bridge, and the width should be assessed

FIG. 22. Prominent nasal bridge. It can be difficult to determine whether in addition the eyes are deeply set.

separately. The nasal bridge becomes more prominent with age. Although the nasal root may be anteriorly placed without increasing the space between the eyes, prominence of the nasal bridge may be accompanied by ***Telecanthus*** or ocular ***Hypertelorism*** [Hall et al., 2009]. If such findings are present these should be coded separately. ***Deep-set eyes*** [Hall et al., 2009] may lead to the impression of a prominent nasal bridge, but this finding should be coded separately.

Synonym: Nasal bridge, high

Nasal bridge, recessed: see ***Nasal bridge, depressed***

Nasal bridge, retruded: see ***Nasal bridge, depressed***

Nasal bridge, thin: see ***Nasal bridge, narrow***

Nasal Bridge, Wide

Definition: Increased breadth of the nasal bridge (Fig. 23). *subjective*

Comments: Care should be taken to distinguish between increased width of bone and ***Fullness of paranasal tissue***. A wide nasal bridge should be distinguished from ***Telecanthus*** and ***Hypertelorism*** [Hall et al., 2009] (Fig. 2). A wide nasal bridge can be either prominent or depressed, which should be coded separately.

Synonym: Nasal bridge, broad

FIG. 23. Wide nasal bridge.

Nasal Cartilage, Absent

Definition: Lack of a palpable nasal cartilage (Fig. 24). *objective*

Comments: This feature may be accompanied by a deficiency of the nasal bone. Absence of the nasal cartilage may lead to a ***Depressed nasal tip***, which should be coded separately.

Nasal cartilages, separated: see ***Nasal tip, bifid***

FIG. 24. Absent nasal cartilage. This usually but not always goes along with holoprosencephaly.

Nasal ridge, broad: see ***Nasal ridge, wide***

Nasal Ridge, Concave

Definition: Nasal ridge curving posteriorly to an imaginary line that connects the nasal root and tip (Fig. 25). *objective*

Comments: Note the difference from ***Depressed nasal bridge***.

FIG. 25. Concave nasal ridge. Please note the concave nasal ridge goes along with anteverted nares in the left two panels but not in the right two.

Replaces: Saddle nose; Ski jump nose

Nasal Ridge, Convex

Definition: Nasal ridge curving anteriorly to an imaginary line that connects the nasal root and tip (Fig. 26). *objective*

Replaces: Nose, beaked; Nose, hooked

FIG. 26. Convex nasal profile. The nose appears often also prominent, and the columella low.

Nasal Ridge, Depressed

Definition: Posteriorly-placed nasal ridge (Fig. 27). *subjective*

Comments: The adjective "depressed" here does not indicate an active process but a status. The feature should be assessed in a profile view. This finding is typically associated with a ***Short columella***, but this should be assessed separately.

Synonym: Nasal ridge, retruded; Nasal ridge, recessed

Replaces: Nose, flat

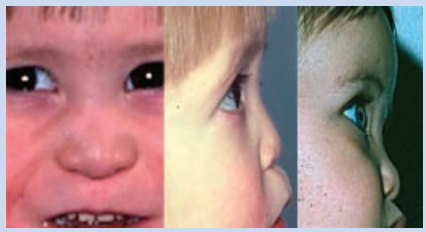

FIG. 27. Depressed nasal ridge. Please note that in addition the nasal bridge is flattened in each of the examples.

Nasal Ridge, Narrow

Definition: Decreased width of the nasal ridge (Fig. 28). *subjective*

Comments: A narrow nasal ridge may appear sharp, but this is not obligate. There is no objective measure of width.

Replaces: Nose, pinched

Nasal ridge, recessed: see ***Nasal ridge, depressed***

Nasal ridge, retruded: see ***Nasal ridge, depressed***

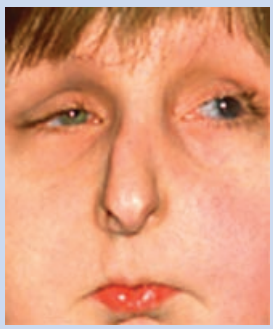

FIG. 28. Narrow nasal ridge. Note that there is also a narrow nasal bridge and narrow nasal base, so the nose in total is narrow.

Nasal Ridge, Wide

Definition: Increased width of the nasal ridge (Fig. 29). *subjective*

Comments: This feature should be assessed in a frontal view. There is no objective measure of width. This may give the impression of a ***Depressed nasal ridge***, but this should be assessed in profile and separately coded. Marked widening of the nasal ridge may be difficult to distinguish from a ***Bifid nose***.

FIG. 29. Wide nasal ridge. Note widening over the base, ridge and bridge in all three examples.

Synonym: Nasal ridge, broad

Nasal root, depressed: see ***Nasal bridge, depressed***

Nasal root, recessed: see ***Nasal bridge, depressed***

Nasal Tip, Bifid

Definition: Visually assessable vertical indentation, cleft, or depression of the nasal tip (Fig. 30). *subjective*

Comments: Note that this definition does not require that the tip is wide. The division should be visible at inspection and not just palpable, as a palpable bifid tip is usual. If a bifid nasal tip goes along with an indentation or cleft of the nasal ridge and the nasal bridge, this should be coded as ***Bifid nose***.

Replaces: Nasal cartilages, separated

FIG. 30. Bifid nasal tip. Please note than in the middle and right panel the nasal bridge, ridge and base are also broad.

Nasal Tip, Broad

Definition: Increase in width of the nasal tip (Fig. 31). *subjective*

Comments: Nasal tip width is assessed at the anterior junction of the alae and the tip. This is easier in persons with a somewhat squared shape of the nasal tip. This may be best viewed from the inferior aspect of the nose. No objective measures are available. See ***Bulbous nose*** for a related term.

Synonym: Nasal tip, wide

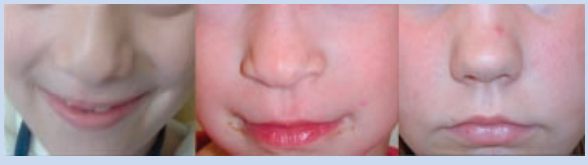

FIG. 31. Broad nasal tip.

Nasal tip, bulbous: see ***Nose, bulbous***

Nasal Tip, Depressed

Definition: Decreased distance from the nasal tip to the nasal base (Fig. 32). *subjective*

Comments: This often accompanies a ***Short columella***, ***Overhanging nasal tip***, and ***Underdeveloped nasal tip***, but these should be assessed and coded separately.

Synonym: Nasal tip, retruded; Nasal tip, recessed

FIG. 32. Depressed nasal tip. Please note that in the three pictures from a single patient in the lower panels, the right panel shows this best, and also note the presence of a short columella in the lower right panel and less in upper right panel.

Nasal Tip, Deviated

Definition: Nasal tip positioned to one side of the midline (Fig. 33). *subjective*

Comments: There is no specific minimal angle before the tip can be determined to be deviated. The assessment of a mild degree of deviation is highly dependant on the experience of the observer. A deviated nasal septum can accompany a deviated nasal tip.

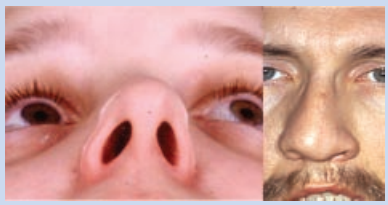

FIG. 33. Deviated nasal tip. Please note that a view from below the tip (left) shows this feature better than a frontal view (right).

Nasal Tip, Narrow

Definition: Decrease in width of the nasal tip (Fig. 34). *subjective*

Comments: Nasal tip width is assessed at the anterior junction of the alae and the tip. This is easier in persons with a somewhat squared shape of the nasal tip. This may be best viewed from the inferior aspect of the nose. No objective measures are available.

Replaces: Nasal tip, pinched

FIG. 34. Narrow nasal tip. Please note that in left panel the nasal bridge and ridge are broad and only the tip is narrow, while in right panel also the nasal bridge and ridge are narrow.

Nasal Tip, Overhanging

Definition: Positioning of the nasal tip inferior to the nasal base (Fig. 35). *subjective*

Comments: This finding is often associated with a long nasal ridge. It is best appreciated in profile. This may also coexist with a ***Depressed nasal tip*** and ***Low insertion of the columella*** and should be coded separately.

Nasal tip, pinched: see ***Nasal tip, narrow***

Nasal tip, recessed: see ***Nasal tip, depressed***

Nasal tip, retruded: see ***Nasal tip, depressed***

Nasal tip, upturned: see ***Nares, anteverted***

Nasal tip, wide: see ***Nasal tip, broad***

FIG. 35. Overhanging nasal tip. Please note that this feature should be evaluated from the side and can only be appreciated in a frontal view when very expressed as in the left panel.

Nose, Absent

Definition: Complete absence of all nasal structures (Fig. 36). *objective*

Nose, beaked: see ***Nasal ridge, convex***

FIG. 36. Absent nose.

Nose, Bifid

Definition: Visually assessable vertical indentation, cleft, or depression of the nasal bridge, ridge and tip (Fig. 37). *subjective*

Comments: This is a bundled term, but as it is useful in practice it is kept here. If it is only an indentation or cleft of the nasal tip, this should be coded as ***Bifid nasal tip***.

Nose, broad: see ***Nose, wide***

FIG. 37. Bifid nose.

Nose, Bulbous

Definition: Increased volume and globular shape of the anteroinferior aspect of the nose (Fig. 38). *subjective*

Comments: This is a bundled term, but as it is useful in practice it is kept here. This alteration of size and shape may be limited to the tip, but may involve the lower third of the nose. If only the width of the nasal tip is increased this should be coded as ***Broad nasal tip***.

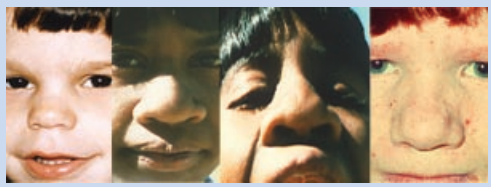

FIG. 38. Bulbous nose.

Replaces: Potato nose; Pear-shaped nose; Nasal tip, bulbous

Nose, flat: see ***Nasal ridge, depressed***

Nose, hooked: see ***Nasal ridge, convex***

Nose, Large: The term "large nose" has not been defined as it is a bundled term comprising several distinct features: prominent nose; wide nasal ridge; prominent nasal tip; and broad nasal base. It requires an assessment of volume that at present cannot be readily determined. A prominent nose is often mislabeled as a large nose.

Nose, Long

Definition: Distance from nasion to subnasale more than two SD above the mean (Fig. 39). *objective* OR

Apparently increased length from the nasal root to the nasal base. *subjective*

Comments: Normal values for length are available [Farkas, 1981; Zankl et al., 2002; Hall et al., 2007]. The length of the nose usually shows an apparent inverse relationship with the length of the philtrum. The nasion may be difficult to determine, in which case only the subjective assessment can be made. Sometimes a nose is well within normal limits in length when measured (objectively) but still appears large (subjectively). The infant's nose usually appears shorter in relation to facial length than does that of the adult [Farkas and Munro, 1987]. The nose continues to grow throughout life, contributing in adulthood to increases in length of the nasal ridge but not necessarily of the nasal length. There are no normal adult standards to determine this. Long nose is distinct from large nose.

Synonym: Increased nasal height

FIG. 39. Long nose. Please note that the length of the nose here is increased in an absolute sense. For an adequate subjective evaluation one needs the facial width and length for comparison.

Nose, Narrow

Definition: Interalar distance more than 2 SD below the mean for age (Fig. 40). *objective* OR

Apparently decreased width of the nasal base and alae. *subjective*

Comments: The nasal width is defined as the distance between the most lateral aspects of alae nasi. A narrow nose is often, but not necessarily, accompanied by a ***Narrow nasal base***. It may also be accompanied by decreased thickness of the alae, narrow nares, or a narrow columella. The term narrow nose is also used by many to denote decreases in not only the interalar distance but also the nasal tip, ridge and bridge. As such it is a bundled term. We do not favor this more general use of the term and instead recommend describing the individual components.

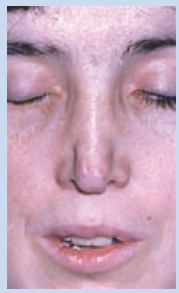

FIG. 40. Narrow nose.

Nose, Prominent

Definition: Distance between subnasale and pronasale more than two standard deviations above the mean (Fig. 41). *objective* OR

Apparently increased anterior protrusion of the nasal tip. *subjective*

Replaces: Nose, large

FIG. 41. Prominent nose. Please note that a prominent nose is best appreciated from the side, as the frontal pictures do not show the prominence well.

Nose, Short

Definition: Distance from nasion to subnasale more than two standard deviations below the mean (Fig. 42). *objective* OR

Apparently decreased length from the nasal root to the nasal tip. *subjective*

Comments: Normal values for length are available [Zankl et al., 2002; Hall et al., 2007]. The length of the nose usually shows a relation with the length of the philtrum. The nose and philtrum together occupy the central face zone; a change of length of one will usually be accompanied by a compensatory, opposite change in the length of the other. The nasion may be difficult to determine, in which case only the subjective assessment can be made. Sometimes a nose is well within normal limits in length when measured

FIG. 42. Short nose. Please note that the length of the nose here is decreased in an absolute sense. For an adequate subjective evaluation one needs the facial width and length for comparison.

(objectively) but still appears small (subjectively) due to a large (mid)facial length. The infant's nose usually appears shorter in relation to facial length than does that of the adult. The nose continues to grow throughout life, contributing in adulthood to increases in length of the nasal ridge but not necessarily of the nasal length. There are no normal adult standards to determine this.

Replaces: Nose, small (a small nose both has a decreased height and a decreased width)

Nose, small: see ***Nose, short***

Nose, Wide

Definition: Interalar distance more than 2 SD above the mean for age (Fig. 29). *objective* OR

Apparently increased width of the nasal base and alae. *subjective*

Comments: The nasal width is defined as the distance between the most lateral aspects of alae nasi. A broad nasal width is often, but not necessarily, accompanied by a ***Wide nasal base***. It may also be accompanied by increased thickness of the alae, enlarged nares, or a broad columella. The term wide nose is used by many to denote increases in not only the interalar distance but also the nasal tip, ridge and bridge. As such it is a bundled term. We do not favor this more general use of the term and instead recommend describing the individual components.

Synonym: Nose, broad

Paranasal Tissue, Fullness

Definition: Increased bulk of tissue alongside the nose (Fig. 43). *subjective*

Comments: The fullness can be caused by both bony and soft tissues. This is distinct from ***Wide nasal ridge*** and ***Wide nasal bridge***.

Replaces: Laterally built up nose

Pinched nose: see ***Nasal ridge, narrow***

Potato nose: see ***Nasal tip, bulbous***

Proboscis

Definition: a fleshy, tube-like structure usually located in the midline of the face or just to one side of the midline (Fig. 44). *objective*

FIG. 43. Fullness of paranasal tissue. Please note that the fullness is very subtle in the left panel.

FIG. 44. Proboscis.

Comments: A proboscis has a single cavity [McGrath, 1992]. It may be found in association with a malformed nose and absence of one or both nares.

Pug nose: see ***Nares, anteverted***

Saddle nose: see ***Nasal ridge, concave***

Ski jump nose: see ***Nasal ridge, concave***

Snub nose: see ***Nasal bridge, flattened***

PHILTRUM

Anatomy of the Philtrum

The philtrum (Greek: philtron = *love potion* [ancient Greeks considered the philtrum to be one of the most erogenous spots on the human body]) is a vertical groove in the midline portion of the upper lip bordered by two lateral ridges or pillars (Fig. 2). It lies between the base of the nose (*subnasale*) and the vermilion border (*labiale superius*), which is also designated as the nasolabial distance. The lower end of the groove and the ridges form the central portion of the Cupid's bow of the vermilion [see Carey et al., 2009].

Measurements of the Philtrum

Measurement of the nasolabial distance is inaccurate, because determination of short distances between soft tissue points is difficult and may be misleading [Méhes, 1988; Ward and Jamison, 1991]. Several surveys of normal values of philtral length in various population samples have been published. For details see a selection of such papers listed in Table I.

Ideally the philtral length is measured with sliding calipers, but in practice it is typically performed with a transparent ruler [Hall et al., 2007]. Measurements should be performed in a neutral position, since facial expression can alter the landmarks. In addition to

TABLE I. Normal Values for Philtrum Length (Selected Studies)

Age of subjects/method	Country	References
13–42 weeks of gestation/sonography	Israel	Gull et al. [2005]
28–42 weeks of gestation/caliper[a] or blunt compasses	Hungary	Méhes [1981]
27–41 weeks of gestation/caliper	Japan	Fok et al. [2003]
37–41 weeks of gestation/caliper	Japan	Tateishi and Kajii [1992]
1 month to 15 years/calliper	Japan	Igarashi and Kajii [1988]
1–18 years/caliper	North America	Farkas et al. [1992]
Birth to 14 years	USA	Feingold [2001]
Birth to 12 years/photo-anthropometry	Germany	Stengel-Rutkowski et al. [1984]
Birth to 97 years/rigid transparent caliper	Switzerland	Zankl et al. [2002]

[a]Caliper = sliding caliper in each case.

difficulties of measurement, significant ethnic, age and sex variations should also be considered [Stengel-Rutkowski et al., 1984].

Anatomic Variation

Characteristics of the philtrum may be classified into quantitative traits and qualitative features:

1. Variations in length: long; short
2. Variations in width: broad; narrow
3. Variations in depths: smooth; deep
4. Unusual appearance: tented; malaligned philtral ridges; midline raphe; midline sinus

DEFINITIONS

Philtral Ridges, Malaligned

Definition: Absence of the usual parallel position of philtral ridges (Fig. 45). *subjective*

Comments: Hajniš [1972] has described *downwards convergent*, *downwards divergent* (trapezoid or triangular), *convex* (ovoid), and *concave* philtral configurations. Any of these findings may be coded using the single term "malaligned philtral ridges."

FIG. 45. Malaligned philtral ridges. In the left panel an ovoid philtrum and in the right panel a trapezoid philtrum is shown.

Philtrum, Broad

Definition: Distance between the philtral ridges, measured just above the vermilion border, more than 2 SD above the mean (Fig. 46). *objective* OR

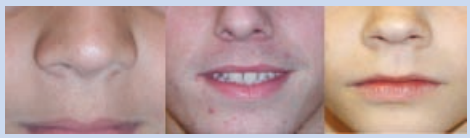

FIG. 46. Broad philtrum.

Apparently increased distance between the ridges of the philtrum. *subjective*

Comments: The mean width of the philtrum was found to be 7 mm in infants by Franz and Sokol [1971] and 9.7 mm in adults by Ward and Jamison [1991]. Measuring width of the philtrum is even more inaccurate than measuring length (vide supra). A broad philtrum may be associated with reduced ridge prominence or a shallow groove, a ***Smooth philtrum***, which should be assessed and coded separately. It may be found with a broad nasal septum.

Synonym: Philtrum, wide

Philtrum, Deep

Definition: Accentuated, prominent philtral ridges giving rise to an exaggerated groove in the midline between the nasal base and upper vermillion border (Fig. 47). *subjective*

Comments: Some have used the term depressed philtrum, suggesting the presence of normal philtral ridges with a deeper

FIG. 47. Deep philtrum.

groove, while admitting the difficulty in distinguishing this from prominent ridges with a normal philtral groove [Pashayan, 1983]. We are unaware of a truly deepened philtral groove with normal height of the ridges.

Synonym: Philtrum, prominent

Replaces: Philtrum, depressed

Philtrum, depressed: see ***Philtrum, deep***

Philtrum, flat: see ***Philtrum, smooth***

Philtrum, indistinct: see ***Philtrum, smooth***

Philtrum, Long

Definition: Distance between nasal base and midline upper lip vermilion border more than 2 SD above the mean (Fig. 48). *objective* OR

Apparently increased distance between nasal base and midline upper lip vermilion border. *subjective*

Comments: The length of the philtrum usually shows a relation with the length of the nose. The philtrum and nose together occupy the central face zone; a change of length of one will usually be accompanied by a compensatory, opposite change in the length of the other. Therefore a long philtrum often accompanies a ***Short nose***, but this is not always the case and should be assessed and coded separately.

FIG. 48. Long philtrum. Please note that the length of the philtrum here is increased in an absolute sense. For an adequate subjective evaluation one needs the nasal length for comparison.

Philtrum, Midline Raphe

Definition: Narrow ridge in the midline of the philtral groove (Fig. 49). *objective*

FIG. 49. Midline raphe of the philtrum.

Comment: The ridge may be very subtly elevated.

Philtrum, Midline Sinus

Definition: Pit in the midline of the philtral groove (Fig. 50). *objective*

Comments: Although congenital sinuses of the lips generally occur in the lower lip, in rare instances they may appear in the philtrum of the upper lip, commonly close to the insertion of the columella [Asahina et al., 1997].

FIG. 50. Midline sinus of the philtrum. The lower two panels courtesy of Dr. Alan Fryer.

Philtrum, Narrow

Definition: Distance between the philtral ridges, measured just above the vermilion border, more than 2 SD below the mean (Fig. 51). *objective* OR

Apparently decreased distance between the ridges of the philtrum. *subjective*

Philtrum prominent: see ***Philtrum, deep***

FIG. 51. Narrow philtrum.

Philtrum, Short

Definition: Distance between nasal base and midline upper lip vermilion border more than 2 SD below the mean (Fig. 52). *objective* OR

Apparently decreased distance between nasal base and midline upper lip vermilion border. *subjective*

FIG. 52. Short philtrum.

Comments: Usually the nasal columella inserts at the base of the nose. A ***Low insertion of the columella*** leads to shortening of the philtrum, but these should be coded separately. Using the insertion of the columella as the upper limit of the philtrum would then lead to a false shortening. An ***Everted upper lip*** often leads to the subjective appearance of a short philtrum but these should be coded separately.

Philtrum, simple: see ***Philtrum, smooth***

Philtrum, Smooth

Definition: Flat skin surface, with no ridge formation in the central region of the upper lip between the nasal base and upper vermilion border (Fig. 53). *subjective*

FIG. 53. Smooth philtrum. Please note that the facial movement in patient depicted in the lower panel gives the false impression of a smooth philtrum as becomes clear in the same patient with a neutral expression in the middle panel.

Comments: There is a spectrum of this finding from total absence of the philtral ridges to a some prominence of the ridges. The central groove varies from absent to shallow. Normal values for the frequency of smooth philtrum are available [Queisser-Luft et al., 2001]. Grading of the smoothness of the philtrum, used in the assessment of Fetal Alcohol Syndrome [Astley and Clarren, 1995] (Fig. 54), has been developed. This finding is greatly influenced by the facial expression, and care should be taken to evaluate the philtrum when the face is in a neutral position. A smooth philtrum can be associated with a ***Long philtrum***. However, the two findings should be coded separately.

Synonym: Philtrum, flat

Replaces: Philtrum, indistinct; Philtrum, simple

FIG. 54. Classification for the degree of smoothness in Caucasians (upper panels) and American Africans (lower panels) [Astley and Clarren, 1995]. In grade 4 and 5 the philtrum is considered smooth. Courtesy of Dr. Susan Asley.

Philtrum, Tented

Definition: Prominence of a triangular soft tissue area of the philtrum with the apex to the columella (Fig. 55). *subjective*

Comments: A tented philtrum may accompany a ***Smooth philtrum***. If present this should be coded separately.

Philtrum, wide: see ***Philtrum, broad***

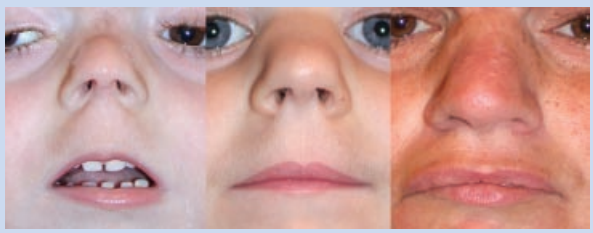

FIG. 55. Tented philtrum.

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RESEARCH ARTICLE

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Elements of Morphology: Standard Terminology for the Lips, Mouth, and Oral Region

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An international group of clinicians and scientists working in the field of dysmorphology has initiated the standardization of terms used to describe human morphology. The goals are to standardize these terms and reach consensus regarding their definitions. In this way, we will increase the utility of descriptions of the human phenotype and facilitate reliable comparisons of findings among patients. Discussions with other workers in dysmorphology and related fields, such as developmental biology and molecular genetics, will become more precise. Here we summarize the anatomy of the oral region and define and illustrate the terms that describe the major characteristics of the lips and mouth.

Key words: nomenclature; definitions; terminology; lips; mouth; anatomy; anthropometry

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INTRODUCTION

This paper is part of a series of six articles defining the morphology of regions of the human body [Biesecker et al., 2009; Hall et al., 2009; Hennekam et al., 2009; Hunter et al., 2009; Allanson et al., 2009b]. The series is accompanied by an introductory article describing the general aspects of this study and the principles used in establishing the definitions [Allanson et al., 2009a]. The reader is encouraged to consult the Introduction when using the definitions.

ANATOMY OF THE LIPS, MOUTH, AND ORAL REGION

General

The appearance of the lips varies with facial movement. Smiling and crying can alter dramatically the shape of the upper lip, as do pursing or pouting. Therefore, the lips must be assessed when the subject has a relaxed (neutral) face: the eyes are open, the lips make gentle contact, and the teeth are slightly separated. The neck, jaw, and facial muscles should not be stretched nor contracted, and the face should be positioned using the Frankfurt horizontal (a line joining the orbitale and the porion) [Farkas, 1981]. Here we define the anatomic features important in proposing the Definitions of the paper.

Anatomy

Lips: The structures that surround the oral aperture (Fig. 1). In the central region their superior border corresponds to the inferior margin of the base of the nose. Laterally, their limits follow the alar sulci and the upper and lower lips join at the oral commissures. The inferior limit of the lips in the central region is the mentolabial sulcus. Anatomically, the philtrum and its pillars are a part of the upper lip. The surface of the lip is comprised of four zones: hairy

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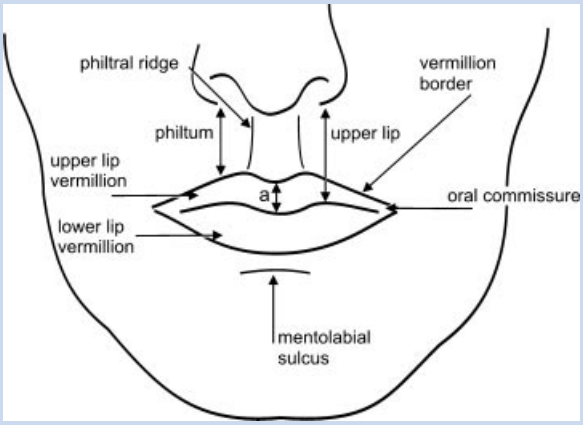

FIG. 1. This drawing depicts the major anatomic landmarks of the lips and mouth (see text).

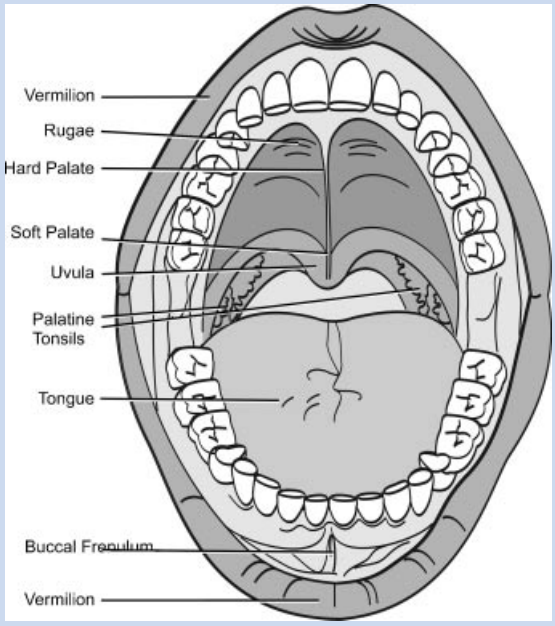

FIG. 2. This drawing depicts the important features of the oral cavity (see text).

skin, vermilion border, vermilion and oral mucosa. The normal shape of the lips varies with age, and is influenced by ethnicity.

Vermilion: The red part of the lips (Fig. 1). It is covered with a specialized stratified squamous epithelium, which is in continuity with the oral mucosa of the gingivolabial groove. Confusingly, the vermilion itself is also often referred to as "the lips."

Vermilion border: The rim of paler skin that demarcates the vermilion from the surrounding skin.

Cupid's bow: The contour of the line formed by the vermilion border of the upper lip. In a frontal view, this line resembles an archer's bow, which curves medially and superiorly from the commissures to the paramedian peaks located at the bases of the pillars of the philtrum (crista philtrae) with an inferior convexity lying between those peaks. The philtrum is the vertical groove in the midline of the upper lip bordered by these lateral pillars (ridges) [Hennekam et al., 2009].

Oral mucosa: Stratified squamous non-keratinized epithelium covering of the inner aspect of the oral cavity [Standing, 2005].

Mouth: The oral aperture that opens into the oral cavity proper [Standing, 2005]. The opening is bounded by the upper and lower vermilion. The cavity comprises the alveolar arches with gums and teeth, the hard and soft palate, and the tongue, anchored to the floor of the mouth (Fig. 2). The oral cavity leads into the oropharynx, bounded by the tonsillar pillars. Standards exist for measuring the length and height of the oral aperture [Farkas, 1981].

Oral commissure: The place where the lateral aspects of the vermilion of the upper and lower lips join. The cheilion is the anthropological landmark located at this site (see Fig. 1).

Labial fissure: Slit-like space between the lips; the oral vestibule.

Oral Cavity: The space bounded superiorly by the hard and soft palates, laterally by the alveolar processes of the maxillary bone, and inferiorly by the tongue (see Fig. 2).

Alveolar ridge: The U-shaped bony crests of the upper and lower jaw in which the teeth are situated.

Hard palate: Bony anterior two-thirds of the roof of the mouth separating the nasal cavity from the oral cavity. The boundary of the hard and soft palates can be determined by palpation.

Soft palate (*velum palatinum*): Posterior one third of the palate comprised of a fibromuscular fold of soft tissue suspended from the hard palate and separating the nasal and oral cavities.

Uvula: A conical projection of soft tissue extending inferiorly from the posterior edge of the middle of the soft palate (see Fig. 2).

Gingiva (gums): Dense fibrous tissue covered by mucous membrane overlying the alveolar ridge in which the teeth are situated.

Buccal frenulum: A thin fold of soft tissue extending from the gingiva of the mid-anterior alveolar ridge to the inner surface of the medial part of the upper or lower lip (see Fig. 2).

Lingual frenulum: A thin fold of soft tissue extending from the floor of the mouth to the base of the tongue.

Tongue: Muscular organ of deglutition, speech and taste covered with epithelium and bound to the floor of the mouth.

Teeth: Hard dental structures located on the alveolar ridges and situated in the gingiva. In humans, teeth have two stages, the primary (deciduous) and the secondary (permanent, adult).

LIPS: DEFINITIONS

Commissural Pit

Definition: Depression located at an oral commissure (Fig. 3). *objective*

Comments: This pit has no relationship to a ***Lip pit***.

Cupid's bow: see ***Cupid's bow, exaggerated***

Cupid's bow, accentuated: see ***Cupid's bow, exaggerated***

FIG. 3. *Commissural pit*. These are always located at the same position at the corners of the oral aperture.

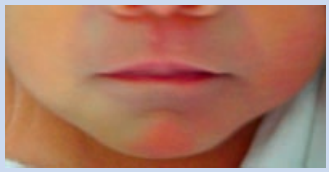

FIG. 4. *Absent Cupid's bow*. This feature is often associated with a thin vermilion of the upper lip as in this child.

Cupid's Bow, Absent

Definition: Lack of paramedian peaks and median notch of the upper lip vermilion (Fig. 4). *objective*

Comment: This bow is often absent in a ***Thin vermilion of the upper lip***, but that should be assessed separately. This finding is commonly associated with ***Smooth philtrum***, but that should be coded separately [Hennekam et al., 2009].

Cupid's Bow, Exaggerated

Definition: More pronounced paramedian peaks and median notch of the Cupid's bow (Fig. 5). *subjective*

Comment: This may be associated with a ***Deep philtrum***, [Hennekam et al., 2009] but that finding should be coded separately.

Synonym: Cupid's bow, accentuated

Replaces: Cupid's bow (used without adjective)

Lip fistula: see ***Lip pit***

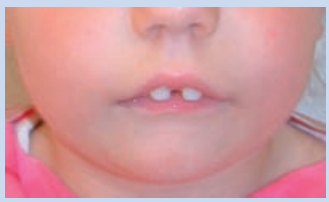

FIG. 5. *Exaggerated Cupid's bow*.

Lip Freckle

Definition: Increased focal pigmentation of the vermilion of the lips (Fig. 6). *subjective*

Comment: Lip freckles may be accompanied by ***Perioral hyperpigmentation***, but this should be assessed separately. Lentigo is commonly used as a synonym for freckle in reference to the vermilion, but these are distinct terms when referring to the skin.

Synonym: Lip lentigo

FIG. 6. Freckling of the vermilion of the lower lip.

Lip, coarse: see, ***Vermilion, upper lip, thick***

Lip, full: see ***Vermilion, upper lip, thin***

Lip, lentigo: see ***Lip freckle***

Lip, lower drooping: see ***Vermilion, lower lip, everted***

Lip, lower full: see ***Vermilion, lower lip, thick***

Lip, lower thick: see ***Vermilion, lower lip, thick***

Lip Pit

Definition: Depression located on the vermilion of the lower lip, usually paramedian (Fig. 7). *objective*

Comments: A lip pit may be connected by a fistula to mucous minor salivary glands in the lower lip. In addition, a lip pit may on occasion be seen with a surrounding tissue elevation (mound). Pits located at the labial commissure (cheilion) are distinct from lip pits; see ***Commissural pit***.

Synonym: Lip fistula

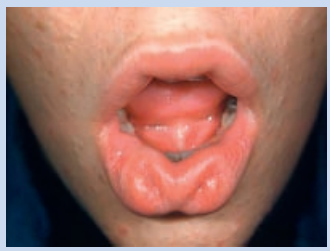

FIG. 7. Typical *lip pits*. Note: these are usually just lateral to the midline.

Lip, thick: see ***Vermilion, upper lip, thin*** Lip, upper thin: see ***Vermilion, upper lip, thin***

Mouth, tented: see ***Vermilion, upper lip, tented***

Nasolabial crease, hypoplastic: see ***Nasolabial fold, underdeveloped***

Nasolabial crease, prominent: see ***Nasolabial fold, prominent***

Nasolabial crease, underdeveloped: see ***Nasolabial fold, underdeveloped***

Nasolabial Fold, Prominent

Definition: Exaggerated bulkiness of the crease or fold of skin running from the lateral margin of the nose, where nasal base meets the skin of the face, to a point just lateral to the corner of the mouth (cheilion, or commissure) (Fig. 8). *subjective*

Comments: Increasing prominence with age is usual. See Allanson et al. [2009b].

Synonym: Nasolabial crease, prominent

FIG. 8. ***Prominent nasolabial fold.***

Nasolabial Fold, Underdeveloped

Definition: Reduced bulkiness of the crease or fold of skin running from the lateral margin of the nose, where nasal base meets the skin of the face, to a point just lateral to the corner of the mouth (cheilion or commissure) (Fig. 9). *subjective*

Comments: See Allanson et al. [2009b].

Synonym: Nasolabial crease, underdeveloped

Replaces: Nasolabial crease, hypoplastic; Nasolabial fold, hypoplastic

FIG. 9. ***Underdeveloped nasolabial fold.***

Perioral Hyperpigmentation

Definition: Increased pigmentation, either focal or generalized, of the skin surrounding the vermilion of the lips (Fig. 10). *subjective*

Comment: Periorbital hyperpigmentation may be accompanied by ***Lip freckles***, but this should be assessed separately.

Vermilion border, thin: see ***Vermilion, upper lip, thin***

FIG. 10. ***Perioral hyperpigmentation.***

Vermilion, Lower Lip, Everted

Definition: Inner aspect of the lower lip vermilion (normally apposing the teeth) visible in a frontal view (Fig. 11). *subjective*

Comments: In frontal view, with the face relaxed, the apparent height of the lower lip vermilion is excessive and the lower incisors may be visible. On profile view, the vermilion is more convex than usual. An everted lower lip may be viewed as "pouting," but this designation is a functional term.

Replaces: Drooping lower lip

FIG. 11. ***Everted vermilion of the lower lip*. The designation pouting lower lip is sometimes used but this is a functional term.**

Vermilion, Lower Lip, Thick

Definition: Height of the vermilion of the lower lip in the midline more than 2 SD above the mean (Fig. 12). *objective*

OR apparently increased height of the vermilion of the lower lip in the frontal view. *subjective*

Comments: Normal values for the height of the vermilion are available [Farkas, 1981] but measurements are not commonly used. Most clinicians determine this feature subjectively. The lower lip is

FIG. 12. *Thick vermilion of the lower lip.*

typically thicker than the upper one. The height of the vermilion of the lower lip varies among ethnic groups, and the vermilion should be compared to a population of same ethnic background. When the vermilion is thick, it is more convex and more everted than usual on profile view, but that should be assessed separately.

Replaces: Thick lower lip; Full lower lip

Vermilion, Lower Lip, Thin

Definition: Height of the vermilion of the medial part of the lower lip more than 2 SD below the mean (Fig. 13). *objective*

OR apparently reduced height of the vermilion of the lower lip in the frontal view. *subjective*

Comment: Normal values for the height of the vermilion are available [Farkas, 1981] but measurements are not commonly used. Most clinicians determine this feature subjectively. The height of the vermilion of the lower lip varies considerably among ethnic groups, and the vermilion should be compared to a population of same ethnic background. If the lower lip vermilion is thin, the inferior border of the vermilion is less curved, and on a profile view, the lower lip vermilion is less convex than usual.

FIG. 13. *Thin vermilion of the lower lip.*

Vermilion, Upper Lip, Everted

Definition: Inner aspect of the upper lip vermilion (normally apposing the teeth) visible in a frontal view (Fig. 14). *subjective*

Comments: In frontal view, with the face relaxed, the apparent height of the upper lip vermilion is excessive and the upper incisors may be visible. On profile view, the vermilion is more convex than usual. An everted upper lip may be associated with a short philtrum, and may be secondary to protruded upper teeth, but these should be assessed and described separately.

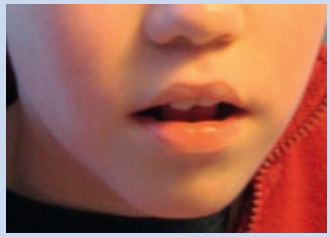

FIG. 14. *Everted vermilion of the upper lip.*

Vermilion, Upper Lip, Tented

Definition: Triangular appearance of the oral aperture with the apex in the midpoint of the upper vermilion and the lower vermilion forming the base (Fig. 15). *subjective*

Comment: This finding is distinguished from an ***Exaggerated Cupid's bow*** by the alteration of the shape of the oral aperture.

Replaces: Tented mouth

FIG. 15. *Tented vermilion of the upper lip.*

Vermilion, Upper Lip, Thick

Definition: Height of the vermilion of the upper lip in the midline more than 2 SD above the mean (Fig. 16). *objective* OR

apparently increased height of the vermilion of the upper lip in the frontal view. *subjective*

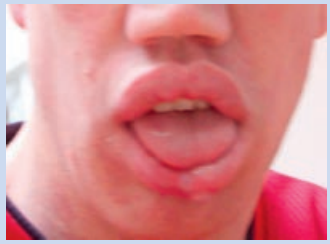

FIG. 16. *Thick vermilion of the upper lip*. This is the preferred designation rather than coarse or full lip since the described feature is the vermilion of the lip, not the lip itself.

FIG. 17. This figure displays the commonly used Likert scale, 1–5, that ranges from the *Thick vermilion of the upper lip* (1) to *Thin vermilion*; in a Caucasian (A) and an African-American (B) (see text). Note as well the philtral scale from 1 to 5 [see Hennekam et al., 2009].

Comments: Normal values for the height of the vermilion are available [Farkas, 1981], but measurements are not commonly used. Most clinicians determine this feature subjectively or utilize the Likert scale of Astley and Clarren [2000] (Fig. 17). The vermilion of the upper lip varies considerably among ethnic groups, and the vermilion should be compared to a population of same ethnic background. The thickness of the upper lip vermilion is sensitive to the facial expression. On profile view, a thick vermilion is more convex than usual.

Replaces: Coarse lip; Thick lip; Full lip

Vermilion, Upper Lip, Thin

Definition: Height of the vermilion of the upper lip in the midline more than 2 SD below the mean (Fig. 18). *objective*

OR apparently reduced height of the vermilion of the upper lip in the frontal view. *subjective*

Comments: Normal values for the height of the vermilion are available [Farkas, 1981], but measurements are not commonly used. Most clinicians determine this feature subjectively or use the Likert scale for Caucasians and African Americans [Astley and Clarren, 2000] (see Fig. 17). The height of the vermilion of the upper lip varies among ethnic groups, and the vermilion should be compared to a population of same ethnic background. The thinness of the upper lip vermilion is sensitive to facial expression (see Anatomy Section). On profile view, a thin vermilion is less convex than usual. A thin upper lip vermilion may be associated with a smooth philtrum and an absence of the Cupid's bow, but these should be assessed separately.

Replaces: Thin vermilion border; Thin upper lip

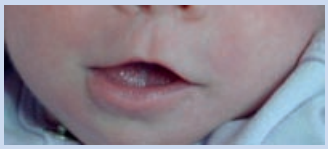

FIG. 18. *Thin vermilion of the upper lip*. Note the *absent Cupid's bow* as well.

MOUTH: DEFINITIONS

Alveolar ridge fusion: see ***Fibrous syngnathia***

Fibrous Syngnathia

Definition: Complete or nearly complete soft tissue fusion of the alveolar ridges (Fig. 19). *subjective*

Comments: This finding is associated with severely reduced mobility, or lack of mobility, between the upper and lower jaws. This finding is the severe end of a spectrum that includes ***Oral synechiae***.

Synonym: Fusion of the alveolar ridges

FIG. 19. *Fibrous syngnathia*. See *Oral synechiae* as well.

Hyperpigmentation, Intra-Oral

Definition: Increased pigmentation, either focal or generalized, of the oral mucosa (Fig. 20). *subjective*

Comment: Pigmentation of alveolar ridges is common in people with dark skin pigmentation. This term encompasses a range of pigmentary findings, from freckles to generalized hyperpigmentation.

Macrostomia: see ***Mouth, wide***

Microstomia: see ***Mouth, narrow***

Mouth, carp: see ***Mouth, downturned corners of***

FIG. 20. *Intra-oral hyperpigmentation.*

Mouth, Downturned Corners of

Definition: Oral commissures positioned inferior to the midline labial fissure (Fig. 21). *subjective*

Comment: This finding should be assessed with the mouth closed, the lips in relaxed contact, and the face relaxed. The finding may be difficult to assess if the lower lip is enlarged.

Replaces: Carp mouth; Fish mouth (pejorative terms)

Mouth, fish: see ***Mouth, downturned corners of***

FIG. 21. *Downturned corners of the mouth*. Here the oral commissures are positioned inferior to the midline labial fissure.

Mouth, Narrow

Definition: Distance between the commissures more than 2 SD below the mean (Fig. 22). *objective*

or apparently decreased width of the oral aperture. *subjective*

Comment: The width of the mouth varies with facial movement and must be assessed when the subject has a relaxed (neutral) face. This term replaces microstomia, small oral aperture, and small mouth because the reduced opening of the mouth is secondary to reduced width.

Replaces: Microstomia; Small oral aperture; Small mouth

FIG. 22. *Narrow mouth.*

Mouth, Upturned Corners of

Definition: Oral commissures positioned superior to the midline labial fissure (Fig. 23). *subjective*

Comment: This finding should be assessed with the mouth closed, the lips in relaxed contact, and the face relaxed. The finding may be difficult to assess if the upper lip is enlarged.

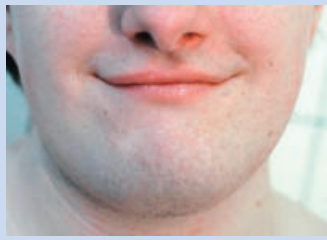

FIG. 23. *Upturned corners of the mouth.*

Mouth, Wide

Definition: Distance between the oral commissures more than 2 SD above the mean. *objective* OR

apparently increased width of the oral aperture (Fig. 24). *subjective*

Comment: The width of the mouth varies with facial movement and must be assessed when the subject has a relaxed (neutral) face. This term replaces macrostomia, large mouth, and large oral aperture because these terms imply a wide and open mouth. The term should not be used to describe a patient with a lateral oral cleft.

Replaces: Macrostomia; Large mouth; Large oral aperture

FIG. 24. *Wide mouth*. This width of the oral aperture can be easily measured.

Oral aperture, small: see ***Mouth, narrow***

Oral Frenulum, Accessory

Definition: Extra fold of tissue extending from the alveolar ridge to the inner surface of the upper or lower lip (Fig. 25). *objective*

Comment: This finding is assessed by gently retracting the oral mucosa from the alveolar ridge. Typically there is a single maxillary and a single mandibular frenulum located in the midline between the two central incisors. Abnormalities of the alveolar ridges may

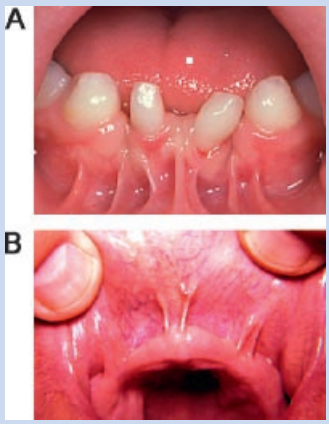

FIG. 25. *Accessory oral frenulum* of the lower (A) and upper lips (B) in different patients.

accompany accessory frenula, but these should be assessed separately.

Synonyms: Supernumerary oral frenulum; Extra oral frenulum

Oral frenulum, extra: see ***Oral frenulum, accessory***

Oral frenulum, supernumerary: see ***Oral frenulum, accessory***

Oral Synechia

Definition: Fibrous band between the mucosal surfaces of the upper and lower alveolar ridges (Fig. 26). *objective*

Comment: These bands must be distinguished from synechiae between the tongue and palate (glossopalatal ankylosis) and from synechiae arising from the floor of the mouth (as in the subglossopalatal membrane), oropharyngeal isthmus (as in persistent buccopharyngeal membrane) or from the lower lip [Gorlin et al., 2001]. If there is a complete soft tissue contiguity between the upper and lower alveolar ridges, the term, ***Fibrous syngnathia*** should be used instead.

FIG. 26. *Oral synechiae*. Note the obvious fibrous bands.

Vermilion, Upper Lip, U-Shaped

Definition: Gentle upward curve of the upper lip vermilion such that the center is placed well superior to the commissures (Fig. 27). *subjective*

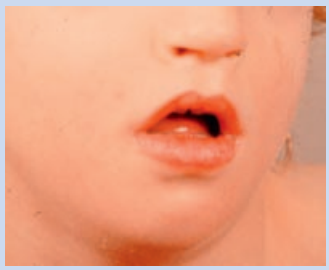

FIG. 27. *U-shaped vermilion of the upper lip*. Note the contour of the vermilion and the shape of the oral aperture.

Comment: The U-shaped upper vermilion is a more rounded version of the ***Tented upper lip vermilion***. In U-shaped upper vermilion there is loss of the central groove of the Cupid's bow.

Replaces: Carp mouth; Fish mouth (pejorative terms); U-shaped mouth

ORAL CAVITY: DEFINITIONS

Aglossia: see ***Tongue, small***

Alveolar ridge hypertrophy: see ***Alveolar ridge overgrowth***

Alveolar Ridge Overgrowth

Definition: Increased width of the alveolar ridges (Fig. 28). *subjective*

Comments: This finding may or may not be accompanied by increased height of the alveolar ridge. This is not to be confused with ***Prominent palatal ridges*** or ***Gingival overgrowth***. This distinction of gingival from alveolar ridge overgrowth may be difficult, especially in milder degrees of the finding.

Replaces: Alveolar ridge hypertrophy

FIG. 28. *Alveolar ridge overgrowth*. Note the increased width of the alveolar ridge.

Ankyloglossia

Definition: Short or anteriorly attached lingual frenulum associated with limited mobility of the tongue (Fig. 29). *subjective*

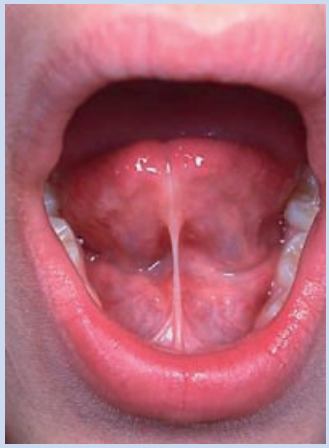

FIG. 29. *Ankyloglossia*. Note the accompanying short lingual frenulum and mild indentation of the tongue tip.

Comment: The anterior third of the tongue is usually free or is partially attached to the floor of the mouth by the lingual frenulum. There is a spectrum ranging from fusion of the tongue to the floor of the mouth ("ankyloglossia inferiorum") to a lingual frenulum that is short or anchored toward the tip of the tongue ("tongue tie"). Ankyloglossia may be associated with a mild indentation of the tip of the tongue, which should not be coded as a ***Bifid tongue***.

Replaces: Ankyloglossia inferiorum; tongue tie

Anodontia: see ***Oligodontia***

Central Incisor, Single Maxillary

Definition: Presence of one maxillary central incisor positioned in the midline (Fig. 30). *objective*

Comment: If a single maxillary central incisor is present but not positioned in the midline, this could be hypodontia (see ***Ologodontia***), but this cannot be evaluated without a radiograph.

FIG. 30. A *single central maxillary incisor*. The lateral incisors are normal making the recognition of this feature difficult on first glance.

Dental Crowding

Definition: Overlapping teeth within an alveolar ridge (Fig. 31). *subjective*

Comment: This is a bundled term. There is a discrepancy in the size or number of the teeth compared to the size of the alveolar ridges.

FIG. 31. *Dental crowding*.

Diastema

Definition: Increased space between two adjacent teeth in the same dental arch (Fig. 32). *subjective*

Comments: Usually there is contact between the lateral aspects of the permanent teeth, at their broadest point. Diastema can apply to any pair of teeth and the term should be modified by a descriptor of the involved teeth. This descriptor must be distinguished from ***Widely spaced teeth***.

Diastemata, multiple: see ***Teeth, widely spaced***

FIG. 32. *Diastema*. See *widely spaced teeth* as well.

Double tooth: see ***Teeth, fused***

Eruption, Advanced

Definition: Tooth eruption more than 2 SD earlier than the mean eruption age. *objective*

Comment: There are established norms for the timing of eruption in both deciduous and permanent teeth [Garn and Rohmann, 1966; Lunt and Law, 1974; McDonald et al., 2004]. Eruption is defined by the appearance of a tooth that has pierced the gum.

Eruption, Delayed

Definition: Tooth eruption more than 2 SD beyond the mean eruption age. *objective*

Comment: This term should not be used in a patient with ***Gingival overgrowth***. There are established norms for the timing of eruption in both deciduous and permanent teeth [Garn and Rohmann, 1966; Lunt and Law, 1974; McDonald et al., 2004]. Eruption is defined by the appearance of a tooth that has pierced the gum.

Gingival hyperplasia: see ***Gingival overgrowth***

Gingival hypertrophy: see ***Gingival overgrowth***

Gingival Overgrowth

Definition: Thickening of the soft tissue overlying the alveolar ridge (Fig. 33). *subjective*

Comments: The degree of thickening ranges from involvement of the interdental papillae alone to gingival overgrowth covering the entire tooth crown.

Replaces: Gingival hypertrophy, Gingival hyperplasia

FIG. 33. ***Gingival overgrowth***. **Note the difference between this finding and overgrowth of the alveolar ridge.**

Glossoptosis

Definition: Posterior displacement of the tongue into the pharynx (Fig. 34). *subjective*

FIG. 34. ***Glossoptosis***. **Note the tongue's posterior placement in the oral cavity and the presence of the formula. [Figure courtesy of Bryan Hall.]**

Comment: Presumably, use of the suffix "ptosis" refers to the situation where the patient is supine, and the displacement is downward. Strictly speaking, the term glossoptosis indicates "falling" of the tongue and thus can also be forward displacement; however by convention it is only used for backward displacement. Glossoptosis may cause obstruction of the airway.

Hypodontia: see ***Oligodontia***

Hypoglossia: see ***Tongue, small***

Macrodontia

Definition: Mesiodistal tooth diameter (width) more than 2 SD above mean for age (Fig. 35). *objective* OR

apparently increased maximum width of the tooth. *subjective*

Comment: The standard reference has means and standard deviations by gender [Moyers et al., 1976].

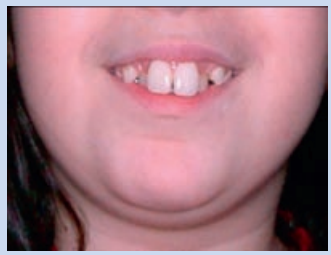

FIG. 35. Macrodontia. The tooth width is easily measured.

Macroglossia: see ***Tongue, large***

Microdontia

Definition: Mesiodistal tooth diameter (width) more than 2 SD below mean (Fig. 36). *objective*

OR apparently decreased maximum width of tooth. *subjective*

Comment: Standard reference has means and standard deviations by gender [Moyers et al., 1976]. In microdontia, the gaps between the teeth, particularly the anterior upper and lower teeth, are increased, creating ***Diastemata***. This should be assessed and coded separately.

Microglossia: see ***Tongue, small***

FIG. 36. *Microdontia*.

Oligodontia

Comment: The term is not defined here since the finding requires a radiograph, as is true for anodontia and for the other designation of tooth agenesis, hypodontia. The terms hypodontia and oligodontia are sometimes used interchangeably in the literature while on other occasions hypodontia is used for selective agenesis of six or less missing teeth while oligodontia is applied when there are more than six missing teeth. Tooth agenesis or oligodontia/hypodontia can be mistaken for delayed eruption and again a radiograph is needed for diagnosis. Absence of teeth may be congenital (tooth agenesis) or acquired. The incidence of congenital absence of teeth is different depending on the type and position of the tooth [Gorlin et al., 2001].

Open Bite

Definition: Visible space between the dental arches in occlusion (Fig. 37). *objective*

Comments: An open bite produces an absence of vertical overlap of the two dental arches. It may be associated with malocclusion, but this should be coded separately. Open bite can be accompanied by malocclusion, which is a complex bundled term. The Angle classification of malocclusion (Classes I–III) is widely used in the orthodontics community [Moyers, 1973] for the characterization of malocclusion.

FIG. 37. *Open bite*. Note the space between the dental arches. (Figure courtesy of Duane Yamashiro.)

Palate, Hard, Short

Definition: Distance between the labial point of the incisive papilla to the midline junction of the hard and soft palate more than 2 SD below the mean (Fig. 38). *objective*

or apparently decreased length of the hard palate. *subjective*

Comment: Objective measurement of the hard palate requires special instrumentation [Hall et al., 2006]. A short hard palate may be associated with velopharyngeal incompetence.

Replaces: Short palate; hypoplastic palate

Palate, short: see ***Palate, hard, short***

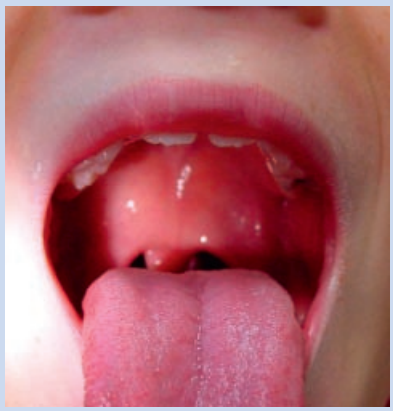

FIG. 38. *Short hard palate*. (Figure courtesy of Alan Rope.)

Palate, High

Definition: Height of the palate more than 2 SD above the mean. *objective* OR

Palatal height at the level of the first permanent molar more than twice the height of the teeth (Fig. 39). *subjective*

Comments: The measuring device for this assessment is described in Hall et al. [2006]. A high palate is often associated with a narrow palate. However, a narrow palate can easily give a false appearance of a high palate. Height and width of the palate should be assessed and coded separately. We do not recommend the subjective determination because this term can be overused and applied inaccurately.

Synonym: High, arched palate

Palate, high arched: see ***Palate, high***

Palate, hypoplastic: see ***Palate, hard, short***

FIG. 39. *High palate*. Note a Narrow palate is a different feature and can produce the false appearance of a high palate.

Palate, Narrow

Definition: Width of the palate more than 2 SD below the mean. *objective*

OR apparently decreased palatal width (Fig. 40). *subjective*

FIG. 40. Narrow palate. Note *Prominent palatine ridges* as well.

Comments: Palatal width is measured as the distance between the maxillary first permanent molar on the right and left sides, at the lingual cervical line, using a specific device. Palate width is typically assessed subjectively in routine clinical practice. Narrowing is often associated with a ***High palate***, but this should be assessed and coded separately. ***Gingival overgrowth*** can give the impression of a narrow palate but should be distinguished and coded separately. The term "gothic palate" is used to indicate that the roof of the palate is not round but rather has an inverted V-shape, and therefore, only the upper part of the palate is narrow.

Palate, Submucous Cleft

Definition: Soft palatal defect with intact overlying mucosa comprising two of the following three findings: (1) notching of the posterior border of the hard palate, (2) bifid uvula, or (3) muscular diastasis leading to a midline translucent zone or furrow in the soft palate (Fig. 41). *objective*

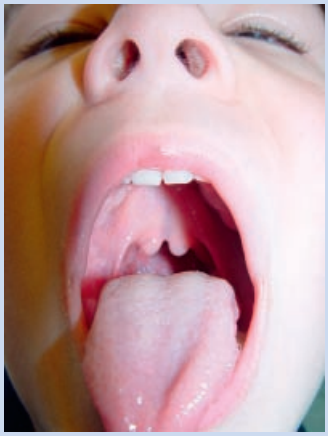

FIG. 41. *Submucous cleft palate*. Note the *Cleft uvula* and the blue indentation of the soft palate. (Figure courtesy of Robert Shprintzen.)

Comment: The notch of the posterior hard palate can sometimes be palpated [Aase, 1990]. Submucous cleft palate is a bundled term but because of its common usage is included here.

Palatine Ridges, Prominent

Definition: Increased size and/or number of soft tissue folds on the palatal side of the maxillary alveolar ridge (Fig. 42). *subjective*

Comments: Soft tissue folds are typically present on the lateral sides of the palate, especially anteriorly.

Synonym: Prominent lateral palatal ridges; Prominent palatine folds

FIG. 42. Prominent palatine ridges. Note the more obvious soft tissue ridges and folds.

Teeth, Fused

Comments: Dental fusion or double tooth (the joining of teeth with separate roots) can only be distinguished from gemination (bifid crown, where only one pulp chamber or root canal is present), with radiographic evidence [Cameron and Widmer, 2003]. Therefore this term is not defined here.

Teeth, Widely Spaced

Definition: Increased spaces (diastemata) between most of the teeth in the same dental arch (Fig. 43). *subjective*

FIG. 43. *Widely spaced teeth*.

Comments: Wide spacing can be secondary to increased room by an unusually large dental arch, microdontia or mixed primary and secondary dentition. It should be carefully noted that slight spacing between the primary teeth is normal, so experience in evaluation is important in determining this feature. This descriptor must be distinguished from ***Diastema***.

Synonym: Multiple diastemata

Tongue, Bifid

Definition: Tongue with a median apical indentation or fork (Fig. 44). *objective*

Comments: Bifid tongue can be associated with ***Ankyloglossia***, but this should be assessed and coded separately. Small indentations of the tip of the tongue should not be coded as a bifid tongue.

FIG. 44. *Bifid tongue*.

Tongue, Furrowed

Definition: Accentuation of the grooves on the dorsal surface of the tongue (Fig. 45). *subjective*

Comments: Usually there is a midline groove with smaller radiating grooves. The deep furrows may extend to the lateral borders. They may follow a regular geometric pattern or be irregular. A furrowed tongue occurs in 10–25% of individuals but is rare in children.

Synonyms: Prominent tongue grooves

Replaces: Scrotal tongue (pejorative)

FIG. 45. *Furrowed tongue*.

Tongue grooves, prominent see ***Tongue furrowed***

Tongue, hyperplasia: see ***Tongue, large***

Tongue, hypertrophy: see ***Tongue, large***

Tongue, hypoplastic: see ***Tongue, small***

Tongue, Large

Definition: Increased length and width of the tongue (Fig. 46). *subjective*

Comments: Normal standards do not exist. Large size usually leads to protrusion of the tongue. This is an acknowledged bundled term, but due to its frequent usage and relative paucity of situations that would call for separate individual assessments of tongue dimensions, the bundled term is retained. ***Micrognathia*** may give the false appearance of a large tongue.

Synonyms: Macroglossia; hyperplasia of the tongue; hypertrophy of the tongue

FIG. 46. *Large tongue*.

Tongue, Lobulated

Definition: Multiple indentations and/or elevations on the edge and/or surface of the tongue producing an irregular surface contour (Fig. 47). *subjective*

Comment: Lobulated tongue can bilobed, trilobed, or show multiple lobes.

FIG. 47. *Lobulated tongue*.

Tongue, Protruding

Definition: Tongue extending beyond the alveolar ridges or teeth at rest (Fig. 48). *subjective*

Comments: ***Protruding tongue*** may or may not be associated with a ***Large tongue***, and that finding should be assessed and coded separately.

FIG. 48. ***Protruding tongue*****. Note the position of the tongue.**

Tongue, rudimentary: see ***Tongue, small***

Tongue, scrotal: see ***Tongue, furrowed***

Tongue, Small

Definition: Decreased length and width of the tongue (Fig. 49). *subjective*

Comment: Normal standards do not exist. The term "aglossia" is often used for extremely small tongue, but a nubbin of tongue tissue is almost always present and aglossia *in sensu strictu* is extremely rare. This is an acknowledged bundled term, but due to its frequent usage and relative paucity of situations that would call for separate individual assessments of tongue dimensions, the bundled term is retained.

Synonyms: Microglossia; hypoglossia; rudimentary tongue

Replaces term: Aglossia; hypoplastic tongue; hypotrophic tongue

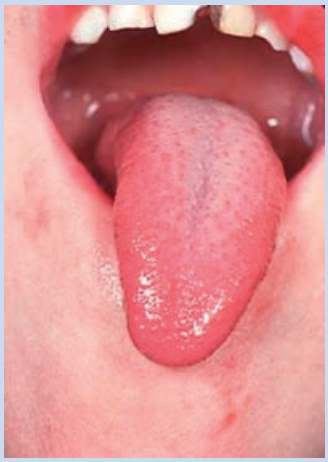

FIG. 49. ***Small tongue*****.**

Tongue, Smooth

Definition: Glossy appearance of the entire tongue surface (Fig. 50). *subjective*

Comment: This is due to reduction in number and/or size of the filiform papillae. A geographic tongue has localized areas of smoothening, but not sufficient to warrant use of the term ***Smooth tongue***.

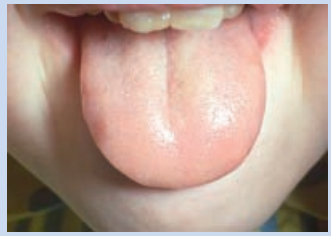

FIG. 50. ***Smooth tongue*****. Note the surface of the tongue in this patient.**

Tooth agenesis: see ***Oligodontia***

Tooth, Natal

Definition: Erupted tooth or teeth at birth (Fig. 51). *objective*

Comment: This is not to be confused with apical dental cysts. Natal teeth most often involve lower central incisors, less often upper central incisors, and rarely first primary molars. Natal teeth occur about once in 3,000 births and are particularly common among some native (First Nation) groups of North America [Mok and Suina, 1986].

FIG. 51. ***Natal tooth*****. Note the eruption of this tooth in this newborn infant.**

Tooth, Premature Loss

Definition: Exfoliation of a primary tooth or teeth earlier than the normal range. *objective*

Comments: See ranges in Kleigman et al. [2007] and Gorlin et al. [2001]. The range of ages in years for normal exfoliation of primary teeth usually precedes the mean age of eruption of each tooth by a year or less.

Tooth, Supernumerary

Definition: Extra tooth or teeth (Fig. 52). *objective*

Comment: The most frequent supernumerary tooth is a mesiodens, which occurs between the two maxillary central incisors. Often it fails to erupt, but creates a large anterior diastema, and would not be detected on physical examination (requires X-ray evaluation). This designation excludes coexistence of primary and permanent dentition due to delayed loss of the former.

FIG. 52. ***Supernumerary tooth*****. Note the extra central incisor between the other incisors. Also there are some** ***widely spaced teeth*** **in the lower arch. (Figure courtesy of Duane Yamashiro.)**

Uvula, Absent

Definition: Lack of the uvula (Fig. 53). *objective*

Comment: Sometimes accompanies a ***Submucous cleft palate***, but this should be coded separately.

FIG. 53. ***Absent uvula*****. (Figure courtesy of Robert Shprintzen.)**

Uvula, Broad

Definition: Increased width of the uvula (Fig. 54). *subjective*

Comments: This finding often accompanies a ***Submucous cleft palate***, but this should be coded separately. A longitudinal groove indicating incomplete fusion of the two parts of the uvula may be present with a broadened uvula and has sometimes been called abortive cleft uvula.

FIG. 54. ***Broad uvula*****. (Figure courtesy of Robert Shprintzen.)**

Uvula, Cleft

Definition: Uvula separated into two parts most easily seen at the tip (Fig. 55). *objective*

Comments: ***Submucous cleft palate*** is a distinct entity.

Synonym: Bifid uvula

Uvula, bifid: see ***Uvula, cleft***

FIG. 55. ***Cleft uvula*****. Often called a bifid uvula, this feature is a component of a** ***submucous palate*** **but is common as an isolated feature.**

Uvula, Long

Definition: Increased length of the uvula (Fig. 56). *subjective*

Comments: In clinical practice, the size of the uvula cannot be easily measured and is not static, since it depends on the position of

FIG. 56. ***Long uvula*****. Note the uvula is also cleft. (Figure courtesy of Robert Shprintzen.)**

the soft palate, the base of the tongue, and the head. Therefore, judgment of change in length of the uvula depends heavily on the experience of the observer.

Uvula, Narrow

Definition: Decreased width of the uvula (Fig. 57). *subjective*

FIG. 57. ***Narrow uvula.*** **(Figure courtesy of Robert Shprintzen.)**

Uvula, Short

Definition: Decreased length of the uvula (Fig. 58). *subjective*

Comments: Objective measurement of the length of the uvula can be determined on a lateral cephalograms. However, in this series we are not relying on radiographs for assessment of findings. In clinical practice, the size of the uvula cannot be easily measured and is not static, since it depends on the position of the soft palate, the base of the tongue, and the head. Therefore, judgment of change in length of the uvula depends heavily on the experience of the observer.

Replaces term: Hypoplastic uvula

Uvula, hypoplastic: see ***Uvula, short***

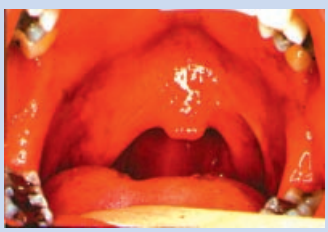

FIG. 58. ***Short uvula.*** **(Figure courtesy of Robert Shprintzen.)**

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Defining Morphology: Hands and Feet

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Abstract

An international group of clinicians working in the field of Dysmorphology has initiated the (re-) definition of all terms used to describe the external human phenotype. The goal is that through standardization of all terms and consensus regarding their definitions the reliability of description of features in humans will increase, comparisons of findings between patients will become more reliable, and discussions with other workers in the field such as developmental biologists and molecular geneticists will become more accurate. Here we report on the (re-) definition of terms needed to describe the major characteristics of the hands and feet. We provide a limited description of the anatomy of this region, limited background anthropometry, and an illustrated list of definitions.

Keywords

nomenclature; definitions; feet; hands; limbs; malformations; anomalies; multiple anomalies; anatomy

INTRODUCTION

This paper is part of a series of six papers defining the morphology of regions of the human body [Allanson et al, 2009a;Carey et al., 2009;Hall et al., 2009;Hennekam et al., 2009; Hunter et al., 2009]. The series is accompanied by an introductory article describing general aspects of this study [Allanson et al., 2009b]. The reader is encouraged to consult the introduction when using the definitions.

An early classification of limb anomalies is that of Isidore Geoffrey Saint-Hilaire (secondary citation, not reviewed). This classification divided anomalies into three categories (phocomèle, hemimèle, and ectromèle). A more recent classification is that by Frantz and O'Rahilly [Frantz and O'Rahilly, 1961]. Modifications of this scheme were implemented by

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others [Swanson et al., 1968; Swanson, 1976]. These schemes are hierarchical classifications and most do not include definitions of the terms.

The similar, but distinct, system of Temtamy and McKusick was developed [Temtamy and McKusick, 1969]. Most of the seven categories that they defined were subdivided into syndromic and non-syndromic forms. While this approach is clearly useful when the goal is to define distinct diagnostic entities, it is not appropriate for a purely descriptive endeavor. The Temtamy and McKusick system focuses on terms for commonly observed patterns of anomalies, not individual structures. Findings that do not conform to commonly recognized patterns cannot be described by their system.

Poznanski lists a glossary of terms used to describe abnormalities and variants of the hands (his text does not address the foot) [Poznanski, 1984]. These definitions are insufficient for a terminological effort such as is undertaken here. However, some are a starting point and others are listed below as being replaced by a more precise term. Finally, the textbook by Dr. Jon Aase includes a number of definitions that were used as starting points [Aase, 1990].

General Descriptors and Background for the Terms

1. The use of the word “digit” versus “finger” and “toe” is problematic. “Finger” is usually specific to digits 2–5 of the hand and “toe” to digits 2–5 of the foot whereas “digit” is a more general term that can be used to describe any finger or toe. We generally use the word “finger” to be distinct from “thumb” (pollex, although this latter term is used rarely in English) and “toe” is distinct from “hallux” (which, oddly enough, is more commonly used in English than is great toe). It is important for the user to read the definition to understand the context in which the term “finger” or “toe” is used. We recognize that some languages do not use a term equivalent to “finger” and “toe” and instead use a modifier of digit (e.g., dita della mano). The term “ray” is a yet more general term that can refer to a finger, toe, thumb, or hallux, but it also includes metacarpals/tarsals, whereas “digit”, “thumb”, “hallux”, “finger”, and “toe” only refers to the phalangeal segments of the ray. The term “ray” is little used in this terminology.
2. The proximo-distal axis of the digits is specified according to the name of the underlying bones e.g., the portion of the finger overlying the distal phalangeal bone is the distal phalanx of the finger.
3. The digits are numbered 1 to 5 (or 6 or more in patients with polydactyly) from the radial/tibial side of the limb, starting with thumb/pollex (great toe/hallux) to little finger/toe; “F” denotes that they are fingers, “T” denotes toes. It is acknowledged that identifying the thumb as “F1” contradicts the general notion that this digit is designated as “thumb” instead of “first finger”, but for simplicity this has been used. In the case of polydactyly, if one can determine the origin of the duplicated digit (e.g., for a partially duplicated thumb where a relatively small digit emanates from the distal first metacarpal, and it is clearly a partially duplicated thumb and not an index finger, the duplicated digit is sub-labeled A and B, with A used for the more anterior (radial/tibial) sub-digit. In this case, the six digits are numbered F1A, F1B, F2, F3, F4, and F5. If the origin of the supernumerary hand digit cannot be clearly ascertained, the digits are labeled F1, F2… recognize that the latter scheme leads to issues comparing pre- and postoperative digit identifiers.
4. Joint specification: interphalangeal joints = IPJ (which is a general term for fingers and toes but is a specific term for the normal thumb and hallux, as they have only a single IPJ) with use of the more specific terms distal IPJ (DIPJ) for the joint between the middle and distal phalanges and proximal IPJ (PIPJ) for the joint

between the middle and proximal phalanges of any triphalangeal digit. The metacarpal-phalangeal joint is abbreviated as the MCPJ and MTPJ is used for the metatarsal-phalangeal joint.

5. Fingertip. The distal segment of the digit overlying the distal phalanx of the finger and including the nail, and dorsal and ventral surfaces.

6. Descriptions should specify bilateral vs. right or left for laterally paired structures. We do not specify terms for findings of asymmetry. Instead, we endorse the approach of defining a body part as small or large, which is less ambiguous and reduces the number of terms that must be defined.

7. The axes of the limb are specified as per embryological terminology. Anterior-Posterior (A/P) (from the thumb/great toe to the little finger/small toe), Dorsal-Ventral (D/V) (from the back of the hand/top of the foot to the palm/sole), and Proximo-Distal (P/D) (from the shoulder/hip to the fingers/toes). Note that common medical usage often substitutes the terms “preaxial” for anterior and “postaxial” for posterior.

8. There are some definitions where one term applies only to the feet and a similar second term applies to the hands. For others, a single term for all limbs may be used with a modifier for “feet” or “hands”, as appropriate. The terms were set out this way primarily for convenience, as some of the terms, definitions, and comments are cumbersome or awkward if one tries to define them in terms of both hand and foot features. In some cases, the definitions were substantially different owing to distinct anatomic features of the hand or foot.

9. For a few of the features, the position of the patient is important (e.g., pes planus). In those cases, the positioning of the patient is discussed in the comments for the term.

10. Note that radiographs are not used in this terminology. See Allanson et al [2009a] for a discussion of this issue.

11. The terms are organized alphabetically in three sections (“Hands and Feet”, “Creases”, and “Nails”). We use the singular form as the default for all terms. For some terms, the singular form cannot exist (e.g., ***Cutaneous syndactyly of the toes***). In such cases, the singular and plural forms of a word are mixed (e.g., “Toe” and “Toes”), alphabetized by the next word of the term. No attempt was made to hierarchically organize the terms. The primary or preferred form of each term appears in the paper in bold and italic font, except within the comments section for that term.

DEFINITIONS

Acheiria: See ***Hand, absent***

Acromicria: See ***Hand, small***

Adactyly

Definition: The absence of all phalanges of all digits of a limb and the associated soft tissues (Fig. 1). *objective*

Comment: This descriptor does *not* require absence of the metacarpal or metatarsal bones. A qualifying phrase is added to specify which limb has the attribute of adactyly.

Replaces: Aphalangy

Amniotic band: See ***Digital constriction ring***

Aphalangy: See ***Adactyly***

Apodia: See ***Foot, absent***

Arachnodactyly: See ***Finger, long; Toe, long; Finger, slender; Toe, slender***

Arch, dropped or fallen: See ***Pes planus***

Arch, high: See ***Pes cavus***

Brachytelephalangy: See ***Finger, short distal phalanx of; Toe, short distal phalanx of***

Camptodactyly

Definition: The DIP and/or PIP joints of the fingers cannot be extended to 180 degrees by either active or passive extension (Fig. 2). *subjective*

Comment: Note that some restrict the use of the term to inability to extend the PIP joint of the fifth finger. We do not restrict the term in this way. This term should not be used if the patient has ***Clenched hand***. A similar effect can be created by radial angulation within the distal phalanx with thickening of the epiphysis, which is called Kirner deformity or dystelephalangy [Ozonoff, 1992]. The affected digits should be specified.

Replaces: Streblodactyly

Clinodactyly

Definition: A digit that is laterally curved in the plane of the palm (Fig. 3). *subjective*

Comment: The curvature in this term is restricted to the phalanges and does not refer to deviation at the MCPJ/MTPJ (see ***Radial deviation of the fingers***). Typically involves an abnormally shaped middle phalanx, but this is not obligate. The identity of the finger/toe should be specified and the directionality of the deviation. This finding is most common in F5, and is almost always radial, but can be seen in any digit and can be in either direction. A similar effect can be created by radial angulation within the distal phalanx with epiphyseal thickening, which is called Kirner deformity or dystelephalangy [Ozonoff, 1992].

Replaces: Incurved finger

Clubbing

Definition: Broadening of the soft tissues (non-edematous swelling of soft tissues) of the digital tips in all dimensions associated with an increased longitudinal and lateral curvature of the nails (Fig. 4). *subjective*

Comment: This definition does not use specification of nail plate angles for several reasons. First, there is no agreement on which angle should be measured. Second, there are no known data to establish the range of normality. Third, clinicians rarely perform such measurements. Clubbed fingers are often described as resembling the end of drumsticks. The affected digits should be specified as described in the introductory comments. If no subtype identifier is specified, it is assumed that all digits are involved. Although increased soft tissue is the sole or major component of clubbing, some cases may be associated with bone spurs or modest overgrowth of the distal phalanx. As this is not relevant to the clinical (non-radiographic) assessment of clubbing, it is not specified in the definition of this term.

Clubhand: See ***Hand, radial deviation of; Hand, ulnar deviation of***

Digit: See various terms under “finger” or “toe” instead of digit

Digit pad, prominent

Definition: A soft tissue prominence of the ventral aspects of the fingertips or toe tips (Fig. 5). *subjective*

Comment: The prominence of the finger or toe pads varies throughout life, being greater in neonates and dependent on the state of hydration. This term should not be used if the digit has clubbing. Note that the synonym “fetal fingertip pads” should not be interpreted to mean that the prominent pad necessarily was present in this individual since fetal life. We do not endorse the use of the term “Persistent fetal pads” as this term implies knowledge of the natural history of this finding, which may or may not be known.

Synonym: Fetal finger/toe tip pads; Fetal finger/toe tip pads

Replaces: Persistent fetal pads

Digital constriction ring

Definition: A narrow segment of significantly reduced circumference of a digit (Fig. 6). *subjective*

Comment: The description should specify the hand and digit that is affected and the approximate location of the band relative to the phalanges. It may be described as partial, if it does not involve the entire circumference of the digit.

Replaces: Amniotic bands

Ectrodactyly: See ***Foot, split; Hand, split***

Fetal fingertip pads: See ***Digit pad, prominent***

Finger, absent

Definition: The absence of all phalanges of a digit of the hand and the associated soft tissues (Fig. 7). *objective*

Comment: This term does not require absence of the metacarpal. The affected digits should be specified. This definition excludes ***Partial absence of the finger***. Note that it can be challenging to number the missing digit if it is a central digit that is missing. In these cases, it is appropriate to modify the term by adding the phrase “non-thumb”. In contrast, it will be obvious that the thumb is present or absent. It may also be difficult to distinguish an absent finger from two digits with an extreme degree of osseous and cutaneous syndactyly. If a patient is missing all digits of a limb, the term ***Adactyly*** should be used. The term Oligodactyly may be used as a synonym, but only in situations where the missing digit is not specified. For example, to state that a patient has Oligodactyly F5 is awkward because the oligodactyly refers to the digits that are present and the F5 refers to the digit that is absent, which is awkward and could become confusing with higher order deficiencies. If a digit that cannot be identified is missing, the term can be modified with a cardinal number, e.g., “Absent fingers, two”.

Replaces: Hypodactyly

Synonym: Oligodactyly

Finger, broad

Definition: Increased width of a non-thumb digit of the hand (Fig. 8). *subjective*

Comment: Note that the girth may be increased in a broad finger, but this must be distinguished from ***Macrodactyly***, because there the length is also increased. This distinction can be subtle. This term should not be used when the increased width is limited to the distal phalanges, instead use ***Broad fingertips***. The affected digit should be specified by the numbering scheme in the introduction. This term is not used for the first digit, see ***Broad thumbs***. When a thumb and one or more fingers are affected, it may be more economical to specify “Broad fingers, F1-5” instead of separately specifying “Broad thumb” and “Broad fingers F2-5”..

Synonym: Wide fingers

Replaces: Pachydactyly; Thick fingers

Fingers, cutaneous syndactyly of

Definition: A soft tissue continuity in the A/P axis between two fingers that extends distally to at least the level of the PIPJ (Fig. 9). *objective OR*

A soft tissue continuity in the A/P axis between two fingers that lies significantly distal to the flexion crease that overlies the metacarpophalangeal joint of the adjacent fingers. *subjective*

Comment: We have set an arguably arbitrary threshold to distinguish the object form the subjective finding. While severe degrees of cutaneous syndactyly are clearly objective, more subtle degrees are subjective. We set this threshold to distinguish these two situations. The digits (or parts of) are joined together by tissue that is not normally present between the digits at that point in the P/D axis. A modifier of “complete” may be used if the cutaneous syndactyly extends to the distal end of the nail bed of the digits. The affected digits should be specified. Note that the unqualified term “syndactyly” is no longer allowed as it is unclear whether this refers to bony or cutaneous syndactyly.

Replaces: Syndactyly (without adjective); Zygodactyly

Finger, hypoplastic: See ***Finger, small***

Finger, incurved: See ***Clinodactyly***

Finger, long

Definition: The middle finger is more than 2 SD above the mean for newborns 27 to 41 weeks EGA or above the 97th centile for children from birth to 16 years of age AND the five digits retain their normal length proportions relative to each other (i.e., it is not the case that the middle finger is the only lengthened digit) (Fig. 10). *subjective* OR

Fingers that appear disproportionately long compared to the palm of the hand. *subjective*

Comment: The first definition is recognized to be problematic, because it implies that the other fingers are all as relatively long as is the middle finger. As the determination of the proportionality of the other four digits is clearly subjective, the term must be regarded as subjective. The term arachnodactyly has been abandoned as it is bundled (narrow and long

digits) and is sometimes used for only narrow or only long digits. The term “long hand” should not be used, as it is a bundled definition of two readily separable terms, ***Long fingers***, and ***Long palm***. If only a subset of the digits of a limb is lengthened, the affected digits should be specified.

Replaces: Arachnodactyly

Finger, narrow: See ***Finger, slender***

Fingers, overlapping

Definition: A finger resting on the dorsal surface of an adjacent digit when the hand is at rest (Fig. 11). *objective*

Comment: This descriptor is ordered depending on which digits are involved (see figure legend for examples). The affected digits should be specified. The ordering of the numbers specifies which digit is dorsal, i.e., with dorsum of the hand facing upward the digit on top is/are recorded first separated by a comma from the digit that is/are overlapped. Fingers that are laterally deviated, but do not rest on top of adjacent fingers should be coded as ***Clinodactyly***.

Finger, partial absence of

Definition: The absence of a phalangeal segment of a finger (Fig. 12). *objective*

Comment: The part that is absent may be specified. The “distal” modifier specifies the loss of the distal phalanx; clinically this is defined by the absence of the nail. The “proximal” modifier specifies the loss of the proximal or middle phalanx with the nail still present and/or the radiographic finding of a remaining phalanx that is similar to a distal phalanx. It may be difficult to determine which phalanx is absent without x-rays and even then, there are circumstances where the missing bone may not be exactly identified (note that no attempt is made to distinguish missing middle from proximal phalanges). In this situation the location adjective will have to be removed. This finding is distinct from ***Short fingers***.

Replaces: Hypophalangy

Finger, radial deviation of

Definition: Angulation of a digit toward the anterior axis (radial side) of the limb (Fig. 13). *subjective*

Comment: The deviation is at the MCPJ. The affected digits should be specified. This finding is distinct from ***Clinodactyly***.

Finger, short

Definition: The middle finger is more than 2 SD below the mean for newborns 27 to 41 weeks EGA or below the 3rd centile for children from birth to 16 years of age AND the five digits retain their normal length proportions relative to each (i.e., it is not the case that the middle finger is the only shortened digit) (Fig. 14). *subjective* OR

Fingers that appear disproportionately short compared to the hand. *Subjective*

Comment: This is an acknowledged bundled term as the definition in most anthropometric sources assumes that the other fingers are all as relatively short as is the middle finger. As the determination of the proportionality of the other four digits is clearly subjective, the term must be regarded as subjective. The Bell classification of brachydactyly (summarized in

[Temtamy and McKusick, 1969]) is a complex assessment of hand patterning – that usage of the word "brachydactyly" is independent of the use here. When "Brachydactyly type *x*" is used, this refers to the Bell classification patterns. When ***Brachydactyly of the hand*** is used, it solely refers to reduced length of the specified digits. "Short hand" should not be used as it is a bundled definition of ***Short fingers*** and ***Short palm***. If the distal phalanges are judged to be proportionately shortened, the additional term of ***Short distal phalanges of the fingers*** should not be used. If the digit is short overall and the distal phalanx is disproportionately shorter than are the finger overall, then both terms should be used.

Synonym: Brachydactyly of the hand

Finger, short distal phalanx of

Definition: Short distance from the end of the finger to the most distal interphalangeal crease or DIPJ flexion point (Fig. 15). *subjective*

Comment: This term differs from ***Partial absence of the finger*** because in that term, the phalanx must be missing, whereas in this term it may be small, but present. Distal phalangeal lengths can be assessed subjectively by comparing that digit segment to the rest of the digit, to other normal digits in that patient, or to typical patients of that age or build. Regarding the subjective definition, for individuals who do not have flexion creases, one may determine this by flexing the DIP joint and estimating the length of the terminal segment of the digit. Alternatively, one may be able to palpate the joint.

Synonym: Brachytelephalangy

Replaces: Short terminal phalanges; Short terminal finger

Finger, slender

Definition: Digits are disproportionately narrow (reduced girth) for the hand/foot size or build of the individual (Fig. 16). *subjective*

Comment: The affected digits should be specified as described. The assessment of this finding is difficult when the digits are long. The bundled and pejorative term "arachnodactyly" is replaced by the separate descriptors ***Long finger*** and ***Slender finger***.

Synonym: Narrow digits; Narrow fingers

Replaces: Thin digits; Arachnodactyly

Finger, small

Definition: Significant reduction in both length and girth of the finger compared to the contralateral finger, or alternatively, compared to a typical finger size for an age-matched individual (Fig. 17). *subjective*

Comment: This is a bundled term, comprising ***Short finger*** and ***Slender digit***, but it is so widely used that it is included. This term is only used if the finger has the normal number of phalangeal segments. An appropriate alternative term for the first digit is ***Small thumb***, when it is the only digit affected. When a thumb and one or more fingers are affected, it may be more economical to specify "Small fingers, F1-5" instead of separately specifying "Small thumb" and "Small fingers F2-5".

Replaces: Underdeveloped finger; Hypoplastic finger

Fingers, splayed

Definition: Divergence of digits along the A/P axis (in the plane of the palm) (Fig. 18). *subjective*

Comment: This may be associated with ***Macrodactyly***, but this should be assessed and coded separately. The affected digits should be specified.

Synonym: Spreading of the fingers

Fingers, spreading of: See ***Fingers, splayed***

Finger, tapered

Definition: The gradual reduction in girth of the digit from proximal to distal (Fig. 19). *subjective*

Comment: If the digits are not uniformly affected, the affected fingers should be specified. If not specified, it refers to all the digits of the hand.

Finger, thick: See ***Finger, broad***

Finger, thin: See ***Finger, slender***

Finger, ulnar deviation of

Definition: Angulation of the digit towards the posterior or postaxial axis (ulnar) of the limb (Fig. 20). *subjective*

Comment: The deviation is at the MCPJ. The affected digits should be specified. This finding is independent of ***Clinodactyly***.

Finger, underdeveloped: See ***Finger, small***

Finger, wide: See ***Finger, broad***

Finger-like thumb: See ***Thumb, triphalangeal***

Fingertip, broad

Definition: Increased width of the distal segment of a finger (Fig. 21). *subjective*

Comment: This term should be reserved for use when the distal digit is significantly broader than the middle part. It should not be used if the digit has ***Clubbing*** or ***Macrodactyly*** or if the entire finger is broad; instead use ***Finger, broad***.

Synonym: Wide fingertips

Replaces: Spatulate fingertips

Fingertip, spatulate: See ***Fingertip, broad***

Fingertip, wide: See ***Fingertip, broad***

Fingertip pads, fetal: See ***Digit, prominent pad***

Foot, absent

Definition: The total absence of the foot, with no bony elements distal to the tibia or fibula (Fig. 22). *objective*

Comment: See ***Foot, partial absence of,*** for a related definition.

Synonym: Apodia

Foot, brachydactyly of: See ***Toe, short***

Foot, broad

Definition: A foot for which the measured width is above the 95th centile for age (Fig. 23). *objective*

OR A foot that appears disproportionately wide for its length. *subjective*

Comment: Note that the normative data [Malina et al., 1973; Hall et al., 2007] only include 4 – 16 year olds. Since foot length is still increasing at the 16 year old age point on the graphs, extrapolation is not recommended. Therefore, persons more than 16 years of age or less than 4 years of age can only be assessed subjectively (until appropriate norms are identified or generated). The Hall et al [2007] handbook (pp. 238-9) states that foot width should be measured from the medial aspect of the first MTPJ to the lateral aspect of the fifth MTPJ. The subjective definition should be used with caution in patients with ***Short feet***. In persons with polydactyly that includes a supernumerary metatarsal, that should be separately coded and the measurement definition from Hall et al, [2007] would need to be modified to account for the supernumerary digit (i.e., if the patient had postaxial polydactyly, measure from the sixth MTPJ). It may be argued that this is double counting polydactyly as a wide foot, as the norms are derived from those with pentadactylous limbs.

Synonym: Wide foot

Foot, cleft: See ***Foot, split***

Foot, long

Definition: Foot length more than 2 SD above the mean for a newborn of 27 – 41 weeks gestation (Fig. 24). *objective*

Foot length above the 97th centile for individuals from birth to 16 years of age *objective* OR A foot that appears longer than expected for age. *subjective*

Comment: Note that for females over the age of 16 years, the curves are essentially flat by 15 years of age, so the 16-year-old norms and distribution can be used for older individuals [Anderson et al., 1956; Hall et al., 2007]. However, the male norms still have significant growth at 16 years, where the graph ends. Therefore, only subjective assessments can be made for males over the age of 16 years. For infants, the norms are specified in SD, per the primary source [Merlob et al., 1984]. Because of the wide range of normal for foot measurements, it may be hard to discriminate a long foot of normal width from that of a narrow foot of normal length, thus the subjective definition is substantially inferior. If the toes are also long, that should be coded separately.

Foot, narrow

Definition: A foot for which the measured width is below the 5th centile for age (Fig. 25). *objective OR*

A foot that appears disproportionately narrow for its length. *subjective*

Comment: The normative data [Hall et al., 2007] only include 4 – 16 year olds. Since the foot width is still increasing at the 16 year old age point on the graphs, extrapolation is not recommended. Therefore, persons more than 16 years or less than 4 years of age can only be assessed subjectively (until appropriate norms are identified or generated). Foot width should be measured from the medial aspect of the first MTP joint to the lateral aspect of the fifth MTP joint [Hall et al., 2007]. It does not seem sensible to apply this term to individuals with polydactyly that includes a duplicated metatarsal.

Foot, osseous syndactyly of

Definition: Lateral (A/P) fusion of the digits (phalanges and/or metatarsals) by hard tissue (cartilage and/or bone) (Fig. 26). *objective*

Comment: Metatarsal syndactyly can be demonstrated by the inability to independently manipulate the two metatarsals (see ***Polydactyly, mesoaxial*** for a description of this maneuver) or the continuity of two metatarsals is demonstrated [Biesecker, 2007]. The description should include the bones that are fused. Osseous syndactyly is distinct from ***Symphalangism***, where the phalanges are fused at the joint longitudinally (P/D axis). The exact nature of the bony anomaly may not be definable without x-ray evidence. Note that the unqualified term “syndactyly” is no longer allowed as it is unclear whether this refers to bony or cutaneous syndactyly.

Replaces: Syndactyly (without adjective)

Foot, partial absence of

Definition: An incomplete absence of the foot, with no bony elements distal to the tarsals, but with preservation of some or all of the tarsals (Fig. 27). *objective*

Comment: See ***Foot, absent*** for a related term.

Foot, postaxial polydactyly of

Definition: Presence of a supernumerary digit that is not a hallux (Fig. 28). *objective*

Comment: Although it is appealing to believe in many cases that the supernumerary (non-hallux) digit is the most fibular, there may be no evidence for this. When the digit is *de minimus*, this seems reasonable by parsimony. When it is fully formed, associated with a supernumerary metatarsal, and fully functional, it may be impossible to determine which toe is supernumerary. Nevertheless, the designation as postaxial is reasonable given the tradition of this designation. Postaxial polydactyly has been divided into two types: A (a fully formed digit) and B (*digitus minimus*, or a pedunculated, non-articulating, non-functional appendage). We recognize these subtypes but note that post-axial polydactyly actually represents a spectrum from type A to type B. When the type is indeterminate, no subtype is specified. The term uses the word “postaxial” instead of the embryologic “posterior” because the former is established in clinical medicine.

Synonym: Posterior polydactyly

Replaces: Posterior duplication of the limb/foot; fibular polydactyly

Foot, preaxial polydactyly of

Definition: Duplication of all or part of the first ray (Fig. 29). *objective*

Comment: There is a wide spectrum of this malformation. The mild end of the spectrum is a bifid (not cleft) nail or a distal phalanx of the hallux with a central lacuna or bifid tip. Broadened halluces without a recognizable anterior/posterior cleft should be coded as ***Broad hallux***. Note that toe number may be affected by the finding of preaxial polydactyly. See the introductory comments for guidance on digit numbering. It is optional to modify the term by adding "partial" to a duplicated hallux that involves less than the entire ray (metatarsal and both phalanges) and "complete" for a digit that has complete duplication of all three bones.

Synonym: Anterior polydactyly

Replaces: Anterior duplication of the limb; Tibial polydactyly

Foot, rocker bottom

Definition: The presence of both a 'prominent heel' and a 'convex contour of the sole' (Fig. 30). *subjective*

Comment: The term is a bundled definition but is in common usage. For that reason, it was included by consensus of the group.

Foot, short

Definition: A measured foot length that is more than 2 SD below the mean for a newborn of 27 – 41 weeks gestation (Fig. 31). *objective OR*

A foot that is less than the 3rd centile for individuals from birth to 16 years of age. *objective OR* A foot that appears disproportionately short. *subjective*

Comment: Note that for females over the age of 16 years, the curves are essentially flat by 15 years of age, so the 16-year-old norms and distribution can be used for older individuals [Anderson et al., 1956; Hall et al., 2007]. However, the male norms still show significant growth at 16 years, where the graph ends. Therefore, only subjective assessments can be made for older males. For infants, the norms are specified in SD, per the primary source [Merlob et al., 1984]. Foot length includes the relatively long calcaneus and tarsal bones as well as the metatarsals and first toe, and so the hand and foot measurements may reflect different growth abnormalities. Foot length should be interpreted with caution if the individual has either ***Pes cavus*** or ***Pes planus***.

Foot, split

Definition: Longitudinal deficiency of a digital ray of the foot except rays 1 or 5 (Fig. 32). *subjective*

Comment: The threshold for this deficiency should be that all of the phalanges in the ray are absent and at least part of the metatarsal. If just the phalanges that are missing, this should be coded as ***Absent toe***. The absence of a central ray or rays frequently yields a cleft appearance of the foot. This term is preferred to "ectrodactyly of the foot" as this has been used (more appropriately) for various forms of phalangeal hypoplasia or amputation. "Lobster claw deformity" is neither descriptive nor acceptable. There is wide variation in the affected feet, but the common factor is the absent central ray, justifying ***Split foot*** as a clinical designation.

Synonym: Cleft foot

Replaces: Ectrodactyly; Lobster claw deformity

Foot, wide: See ***Foot, broad***

Hallux, absent

Definition: The absence of both phalanges of a hallux and the associated soft tissues (Fig. 33). *objective*

Comment: This descriptor does NOT require absence of the metatarsal. The definition excludes partial absent hallux. Oligodactyly and hypodactyly are replaced with more specific terms to allow the distinction of loss of a preaxial digit (thumb or great toe) from loss of digits 2–5. When a great toe and one or more other toes are absent, it may be more economical to specify “Absent toes, T1-3” instead of separately specifying “Absent hallux” and “Absent toes T2-3”.

Synonym: Absent great toe

Replaces: Hypodactyly; Oligodactyly

Hallux, broad

Definition: Visible increase in width of the hallux without an increase in the dorso-ventral dimension (Fig. 34). *subjective*

Comment: Note that girth may be increased in a broad hallux, but this must be distinguished from ***Macrodactyly*** because there the length is also increased. This assessment may be difficult when the hallux is short. Note the separate definition for ***Toes, broad***. When a great toes and one or more other toes are affected, it may be more economical to specify “Broad toes, T1-5” instead of separately specifying “Broad hallux” and “Broad toes T2-5”.

Synonym: Broad great toe

Hammertoe

Definition: Hyperextension of the MTP joint with hyperflexion of the proximal interphalangeal (PIP) joint (Fig. 35). *subjective*

Comment: This term replaces the phrase “dorsiflexed toe” as that term is an incomplete description of the abnormality [Young et al., 2005]. Note that this is not a synonym of mallet toe.

Replaces: Dorsiflexed toe

Hand, absent

Definition: The total absence of the hand, with no bony elements distal to the radius or ulna (Fig. 36). *objective*

Comment: See ***Hand, partial absence*** for a related term.

Synonym: Acheiria

Hand, brachydactyly of: See ***Finger, short***

Hand, cleft: See ***Hand, split***

Hand, clenched

Definition: All digits held completely flexed at the metacarpophalangeal and interphalangeal joints (Fig. 37). *subjective*

Comment: Is distinguished from ***Camptodactyly,*** as that term may describe fewer than five digits of a eudactylous hand and does not involve the MCPJ. The digits may overlap when they lie flexed in the palm. It is not necessary to specify the overlapping fingers finding separately.

Hand, long: See ***Palm, long***

Hand, narrow: See ***Palm, narrow***

Hand, osseous syndactyly of

Definition: Lateral (A/P) fusion of the digits (phalanges and/or metacarpals) by hard tissue (cartilage and/or bone) (Fig. 38). *objective*

Comment: Metacarpal syndactyly can be demonstrated by either a maneuver whereby the two metacarpals cannot be independently manipulated by the examiner (see ***Polydactyly, mesoaxial*** for a description of this maneuver). Phalangeal syndactyly (typically seen in what used to be called a “mitten hand”) may be difficult to demonstrate without X-rays. The description may include the bones that are fused. Osseous syndactyly is distinct from ***Symphalangy*** where the phalanges are fused at the joint longitudinally (P/D axis). See ***Polydactyly, mesoaxial*** for discussion of the use of this term with polydactyly.

Replaces: Syndactyly (without adjective)

Hand, postaxial polydactyly of

Definition: Presence of a supernumerary digit that is not a thumb (Fig. 39). *objective*

Comment: Although it is appealing to believe in many cases that the supernumerary (non-thumb) digit is the most ulnar, there may be no evidence for this. When the digit is *de minimus*, this seems reasonable by parsimony. When it is fully formed with a supernumerary metacarpal and functional, it may be impossible to determine which of the fingers is supernumerary. Nevertheless, the designation as postaxial is reasonable given the tradition of this designation. Postaxial polydactyly has been divided into two types: A (a fully formed digit) and B (*digitus minimus*, or a pedunculated, non-articulating, non-functional appendage). We recognize these subtypes but note that post-axial polydactyly actually represents a spectrum from type A to type B. When the type is indeterminate, no subtype is specified.

Synonym: Posterior polydactyly

Replaces: Ulnar polydactyly; Posterior duplication of the limb/hand

Hand, preaxial polydactyly of

Definition: Duplication of all or part of the first ray (Fig. 40). *objective*

Comment: There is a wide spectrum of this malformation. The mild end of the spectrum is a bifid (not cleft) nail or a distal phalanx of the thumb with a central lacuna or bifid tip. Broadened thumbs without a recognizable line of anterior/posterior clefting should be coded as ***Thumb, broad***. Note that finger numbering may be affected by the finding of preaxial polydactyly. See the introductory comments for guidance on digit numbering. It is optional

to modify the term by adding “partial” to a duplicated thumb that involves less than the entire ray (metacarpal and both phalanges) and “complete” for a digit that has complete duplication of all three bones.

Synonym: Anterior polydactyly

Replaces: Anterior duplication of the limb/hand; Radial polydactyly

Hand, radial deviation of

Definition: Divergence of the longitudinal axis of the hand at the wrist in a anterior (radial) direction (Fig. 41). *subjective*

Comment: This finding may be associated with a short or absent radius, but that is not required and should be coded separately. The term “clubhand” is replaced because it was felt to be pejorative.

Replaces: Radial clubhand; Clubhand

Hand, short: See ***Palm, short; Finger, short***

Hand, small

Definition: A normally proportioned hand (i.e., the various elements of the hand are in proportion to each other) that is overall small for age or overall body size (Fig. 42). *subjective*

Comment: This is acknowledged to be a bundled term, but it is sufficiently common that it was felt to be too cumbersome to describe the component findings. It is arguably superior to code a patient as having objectively determined **Short fingers**, **Short palms**, and **Narrow palms** instead of this bundled term. Note that there are no objective standards that can be used for this assessment, only for middle finger and palm length and palm width. The term acromicria is discouraged because in some definitions, this term also includes small head or facial features; in others it only includes the fingers and toes.

Replaces: Acromicria

Hand, split

Definition: Longitudinal deficiency of a digital ray of the hand except rays 1 or 5 (Fig. 43). *subjective*

Comment: The threshold for this deficiency should be that all of the phalanges of a central ray (not F1 or F5) are absent and at least part of the associated metacarpal (if it is just the phalanges that are missing, this should be coded as ***Absent finger***). The absence of a central ray or rays frequently yields a cleft appearance of the hand. This term is preferred to “ectrodactyly of the hand” as this has been used (more appropriately) for forms of phalangeal hypoplasia or amputation but it is acknowledged that this is a commonly used term. “Lobster claw deformity” is neither descriptive nor acceptable due to its pejorative nature. There is wide variation in the hands of affected individuals, but the common factor seems to be the absence of the central ray, justifying the use of ***Split hand*** as a clinical designation.

Synonym: Cleft hand

Replaces: Ectrodactyly; Lobster claw deformity

Hand, trident

Definition: Splaying of F2-4 along the A/P axis (in the plane of the palm) with relatively normal positioning of F1 and F5 (Fig. 44). *subjective*

Comment: This term is a subtype of splaying of the digits, but its diagnostic utility warrants a separate definition. Note is made that there were a number of different definitions of this term among the terminology group members. However, this definition was the consensus.

Hand, ulnar deviation of

Definition: Divergence of the longitudinal axis of the hand at the wrist in a posterior (ulnar) direction (Fig. 45). *subjective*

Comment: May be associated with a short or absent ulna, but that is not required.

Replaces: Ulnar clubhand; Clubhand

Hand, wide: See ***Hand, broad***

Heel, prominent

Definition: Exaggerated or marked projection of the posterior pole of the heel (Fig. 46). *subjective*

Comment: No sensible thresholds or landmarks could be identified for this finding so the examiners must rely on their experience.

Hypodactyly: See ***Finger, absent; Hallux, absent; Thumb, absent; Toe, absent***

Hypophalangy: See ***Finger, partial absence of; Toe, partial absence of***

Hypothenar eminence, small

Definition: Reduced muscle mass on the ulnar side of the palm (Fig. 47). *subjective*

Comment: The reduced soft tissue is typically abductor digiti minimi and flexor digiti minimi brevis. This may be even more difficult to discern than is ***Small thenar eminence***, but the same considerations outlined in that definition apply.

Limb, anterior duplication of: See ***Foot, preaxial polydactyly of; Hand, preaxial polydactyly of***

Lobster claw deformity: See ***Foot, split; Hand, split***

Macrodactyly

Definition: Significant increase in the length and girth of most or all of a digit compared to its contralateral digit (if unaffected) or compared to what would be expected for age/body build. The increased girth is accompanied by an increase in the dorso-ventral dimension AND the lateral dimension of the digit (Fig. 48). *subjective*

Comment: There are no recognized standards for the girth of digits and this finding can vary substantially in the population. This assessment relies on the judgment of the examiner to recognize when the difference between the expected and the observed is significant. The affected digits should be specified. The definition does not mandate the component of the increased size (bone, connective tissue, etc.), but should exclude edema. The requirement that the girth is most, or all, of the digit is intended to distinguish this from broad fingertips.

This should be distinguished from ***Broad fingers*** or ***Broad fingertips*** as the girth is circumferential in macrodactyly, which is not the case for broad fingers or broad fingertips, which are increased only (or predominantly) in their lateral (A/P) width.

Replaces: Megalodactyly; Pachydactyly

Megalodactyly: See ***Macrodactyly***

Metacarpal, short

Definition: Diminished length of one or more metacarpal bones in relation to the others of the same hand or to the contralateral metacarpal (Fig. 49). *subjective*

Comment: Short metacarpals can involve any of the metacarpal bones, and the affected ray should be specified. The assessment of isolated short metacarpal can be made by viewing the dorsum of the hand when clenched. Note that if metacarpals F2-5 are affected, the correct term is ***Short palm***. The affected digits should be specified.

Metatarsal, short

Definition: Diminished length of a metatarsal, with resultant proximal displacement of the associated toe (Fig. 50). *subjective*

Comment: This is a subjective assessment and one generally compares the position of the MTP joint to that of the contralateral digit or the putatively shortened ray in proportion to the other rays. The affected digits should be specified.

Metatarsus adductus

Definition: The metatarsals are deviated medially (tibially) (Fig. 51). *subjective*

Comment: This is considered subjective, as there is no objective threshold for the finding.

Oligodactyly: ***See Finger, absent; Toe, absent***

Pachydactyly: See ***Finger, broad; Macrodactyly***

Palm, broad

Definition: For children from birth to 4 years of age the palm width is more than 2 SD above the mean; for children from 4 to 16 years of age the palm width is above the 95th centile (Fig. 52). *objective OR*

The width of the palm appears disproportionately wide for the length. *subjective*

Comment: Hand width is measured across the palm at the level of the MCPJ (radial aspect of the second MCPJ to the ulnar aspect of the fifth MCPJ) [Hall et al., 2007]. Caution is advised with the subjective assessment as short metacarpals can mimic a broad palm. In persons with polydactyly that includes a supernumerary metacarpal, that should be separately coded and the measurement technique from Hall et al, [2007] would need to be modified to account for the supernumerary digit (i.e., with postaxial polydactyly, measure to the sixth MCPJ).

Synonym: Wide palm

Palm, long

Definition: For children from birth to 16 years of age the length of the palm is more than the 97th centile (Fig. 53). *objective OR*

The length of the palm appears relatively long compared to the finger length or the limb length. *subjective*

Comment: Palm length standards are found in [Feingold and Bossert, 1974; Hall et al., 2007]. The palm is measured from the distal wrist flexion crease to the flexion crease of the F3 MCPJ. The term “long hand” should not be used as it is a bundled definition of two readily separable terms, ***Fingers, long*** (which, as noted above, can itself be a bundled term) and ***Palm, long***.

Replaces: Long hand

Palm, narrow

Definition: For children from birth to 4 years of age, the palm width is more than 2 SD below the mean; for children from 4 to 16 years of age the palm width is below the 5th centile (Fig. 54). *objective OR*

The width of the palm appears disproportionately narrow for its length. *subjective*

Comment: Palm width is measured across the palm at the level of the MCPJ (radial aspect of the F2 MCPJ to the ulnar aspect of the F5 MCPJ). Norms are specified in [Hall et al., 2007]. Caution is advised for the subjective assessment as the breadth may be in the normal range with disproportionately increased length, which appears narrow. This finding may be associated with elongated/slender limbs in general, but that finding does not bear on the coding of this feature. Proximal narrowing may indicate small thenar or hypothenar eminences. This term replaces “narrow hands” as that term may leave the impression that it includes the thumb, which it does not.

Replaces: Narrow hand

Palm, short

Definition: For children from birth to 16 years of age the length of the palm is less than the 3rd centile (Fig. 55). *objective OR*

The length of the palm appears relatively small compared to the finger length or the limb length. *subjective*

Comment: Palm length is measured as described in [Feingold and Bossert, 1974; Hall et al., 2007]. This term is reserved for individuals with shortening of all four metacarpals 2–5. Individuals with fewer than four shortened metacarpals (in a eudactylous hand, the metacarpals of F2-5) should be coded as ***Metacarpal, short***. See the entry for ***Hand, small*** for a discussion of this finding. “Short hand” should not be used as it is a bundle of two readily separable terms, ***Fingers, short*** (which, as noted above, can itself be a bundled term) and ***Palm, short***.

Replaces: Short hand

Palm, wide: See ***Palm, broad***

Pes cavus

Definition: The presence of an unusually high plantar arch (Fig. 56). *subjective*

Comment: This is a subjective definition for which we could identify no thresholds or standards. Examiners must rely on their experience to make this assessment.

Replaces: High arch

Pes planus

Definition: A foot where the arch is in contact with the ground or floor when the individual is standing (Fig. 57). *objective OR*

In a patient lying supine, a foot where the arch is in contact with the surface of a flat board pressed against the sole of the foot by the examiner with a pressure similar to that expected from weight bearing. *objective OR*

The height of the arch is reduced. *subjective*

Comment: The second definition was conceived by this working group based on a commonsense analysis of how one could generalize the first definition to a non-ambulatory individual. There are no data on this assessment and we invite feedback on its utility.

Replaces: Dropped arches; Fallen arches

Pollex: See various terms under the heading of “Thumb”

Polydactyly, anterior: See ***Foot, preaxial polydactyly of; Hand, preaxial polydactyly of***

Polydactyly, central: See ***Polydactyly, mesoaxial***

Polydactyly, insertional: See ***Polydactyly, mesoaxial***

Polydactyly, intercalary: See ***Polydactyly, mesoaxial***

Polydactyly, mesoaxial

Definition: The presence of a supernumerary finger or toe (not a thumb or hallux) involving the third or fourth metacarpal/tarsal with associated osseous syndactyly (Fig. 58). *objective*

Comment: To determine this finding in the hand without using a radiograph (see [Allanson et al, 2008b] this issue, for a discussion of the use of radiographs), the examiner grasps two adjacent metacarpals, each with a thumb and index finger on the dorsum and palmar aspect of the hand and attempts to independently maneuver the metacarpals. If they cannot be independently manipulated, osseous syndactyly is present [Biesecker, 2007]. Consensus could not be reached on the utility of the bundled term of ***Mesoaxial polydactyly***, so it is specified in the terminology for the time being. It is arguably superior to describe the two separate terms (e.g., Osseous syndactyly of F3,4 (metacarpal) and Postaxial polydactyly of the hand, type A). The term “insertional polydactyly” may be less desirable because the word “insertion” implies mechanism, which is unlikely to be an insertion. The utility of this term should be reviewed over time.

Synonym: Central polydactyly; Intercalary polydactyly

Replaces: Insertional polydactyly

Polydactyly, mirror image

Definition: A hand or foot with more than five digits that has a recognizable A/P axis of symmetry. The axis can lie within a normally formed or partially duplicated digit resembling a middle finger, index finger, thumb, toe, or hallux. Alternatively, the axis can be in an interdigital space with a flanking pair of digits that resemble a middle finger, index finger, thumb, toe or hallux. The most lateral digits on each side of the hand typically resemble fifth fingers/toes (Fig. 59). *subjective*

Comment: This is considered to be a subjective assessment. Although polydactyly is obviously objective, the assessment of the A/P symmetry component is arguably subjective. It may be associated with a forearm that has two ulnae, but this is not required for the finding to be made.

Polydactyly, fibular: See ***Foot, postaxial polydactyly of***

Polydactyly, posterior: See ***Foot, postaxial polydactyly of; Hand, postaxial polydactyly of***

Polydactyly, radial: See ***Hand, preaxial polydactyly of***

Posterior duplication: See ***Foot, postaxial polydactyly of; Hand, postaxial polydactyly of***

Radial clubhand: See ***Hand, radial deviation of***

Ray, absent

Definition: The absence of all phalanges of a digit and the associated metacarpal/metatarsal (Fig. 60). *objective*

Comment: This descriptor requires, in addition to the absence of the phalanges, absence of the metacarpal or metatarsal. Compare this to ***Thumb, absent***, ***Hallux, absent***, ***Toes, absent***, and ***Fingers, absent***. In most cases, the absent metacarpal or metatarsal can be assessed by palpation, but in some cases radiographs may be useful. This definition excludes ***Hand, split*** and ***Foot, split***. If a patient meets the definition of either of those terms, they should be used and ***Ray, absent*** should not.

Sandal gap

Definition: A widely spaced gap between the first toe (the great toe) and the second toe (Fig. 61). *subjective*

Comment: The term is a subjective one but should be used when the gap between the toes is as wide as the second toe is broad.

Sole, convex contour of

Definition: The contour of the foot in lateral profile has a convex shape (Fig. 62). *subjective*

Comment: This term was established as the convex contour may occur without the prominent heel, which together comprise the bundled term ***Foot, rocker bottom***.

Streblodactyly: See ***Camptodactyly***

Syndactyly; See ***Fingers, cutaneous syndactyly of; Toes, cutaneous syndactyly of; Foot, osseous syndactyly of; Hand, osseous syndactyly of***;

Thenar eminence, small

Definition: Reduced palmar soft tissue mass surrounding the base of the thumb (Fig. 63). *subjective*

Comment: The reduced soft tissue is typically abductor pollicis brevis and flexor pollicis brevis muscle bulk. Detection of this abnormality entails clinical judgment, especially in mild cases. The bulk of the muscle mass around the base of the thumb is diminished, and there may be a mild concavity over the volar aspect of the first metacarpal. When the deficiency is unilateral, comparison between the two hands will point up the often-subtle change in contour of the thenar muscles. If the degree of involvement is severe, the palm may taper in width proximally.

Replaces: Thenar hypoplasia

Terminal phalanx, short: See ***Finger, short distal phalanx of; Hallux, short distal phalanx of; Thumb, short distal phalanx of; Toe, short distal phalanx of***

Thenar hypoplasia: See ***Thenar eminence, small***

Thumb abducted: See ***Thumb, hitchhiker***

Thumb, absent

Definition: The absence of both phalanges of a thumb and the associated soft tissues (Fig. 64). *objective*

Comment: This descriptor does not require absence of the metacarpal. This definition excludes ***Partial absence of the thumb***. Oligodactyly and hypodactyly have been replaced with more specific terms to allow the distinction of loss of a thumb from loss of F2-5. The term “oligodactyly” is replaced because oligodactyly refers to the digits that remain, and so “oligodactyly of the thumb” or “oligodactyly F1” is nonsensical.

Replaces: Hypodactyly; Oligodactyly

Thumb, adducted

Definition: In the resting position, the tip of the thumb is on, or near, the palm, close to the base of the fourth or fifth finger (Fig. 65). *subjective*

Comment: The thumb is both flexed and adducted. Lesser degrees of adduction than that specified here may warrant the use of this term, for example, when the tip of the thumb lies near the base of F2 or F3.

Thumb, broad

Definition: Increased thumb width without increased dorso-ventral dimension (Fig. 66). *subjective*

Comment: There is substantial variability thumb width and it may be difficult to determine the threshold for this finding. As it is commonly bilateral, comparing to the contralateral digit will not be helpful. Some have suggested using a definition of thumb width measured at the IP joint of >1.5X the width of the index finger measured at the DIP joint, but no data to support this are available. Note that this term should not be used for thumbs that meet the definition for ***Macrodactyly***. There is a standard for thumb width at birth of 9.8 +/− 0.5 mm (1 SD) (see [Saul et al., 1998] p 45), but this is not widely used and does not take into

account gestational age, or babies who are small for gestational age. The assessment is difficult when the thumb is short.

Synonym: Broad pollex; Wide thumb

Thumb, digitalized: See ***Thumb, triphalangeal***

Thumb, hitchhiker

Definition: With the hand relaxed and the thumb in the plane of the palm, the axis of the thumb forms an angle ≥ 90 degrees with the long axis of the hand (Fig. 67). *objective*

Replaces: Abducted thumb

Thumb, partial absence of

Definition: The absence of a phalangeal segment of a thumb (Fig. 68). *objective*

Comment: The part that is absent may be specified. The “distal” modifier specifies the loss of the distal phalanx; clinically this is defined by the absence of the nail. The “proximal” modifier specifies the loss of the proximal phalanx. This finding is distinct from ***Short thumb***.

Replaces: Hypophalangy

Thumb, proximal placement of

Definition: Thumb placement index >0.55 (Fig. 69). *Objective* OR

The base of the thumb appears closer to the wrist than is typical. *Subjective*.

Comment: The technique for the thumb placement index is described in detail [Malina et al., 1973; Hall et al., 2007]. Briefly, the thumb placement index is the distance from the proximal crease of the index finger to the angle of the first interdigital space divided by the distance from the proximal crease of the index finger to the wrist flexion crease at the base of the thumb. This term should not be used with ***Preaxial polydactyly***.

Thumb, triphalangeal

Definition: A thumb with three phalanges in a single, proximo-distal axis (Fig. 70). *objective*

Comment: The requirement for a single PD axis relates to the issue that partial forms of ***Preaxial polydactyly*** may comprise a partially duplicated thumb with two distal phalanges and a single proximal phalanx. That finding is instead coded as a mild form of thumb polydactyly. Note that this finding can be readily assessed by examination and/or physical manipulation of the thumb.

Replaces: Digitalized thumb; Finger-like thumb

Tibial polydactyly: See ***Foot, preaxial polydactyly of***

Toe, absent

Definition: The absence of all phalanges of a non-hallux digit of the foot and the associated soft tissues (Fig. 71). *objective*

Comment: Note that this descriptor does NOT require absence of the metatarsal. The affected digits should be specified, although this may be difficult. In the latter case, the specified term should be “Absent toe, right foot” (with the identity of the missing toe not specified). This definition excludes ***Partial absence of the toe***. This ***absent toe*** definition excludes ***Absent hallux*** because it is usually obvious if the missing digit is a hallux. It may be difficult to distinguish an absent toe from two digits with an extreme degree of osseous and cutaneous syndactyly. If all digits are missing, the term ***Adactyly*** should be used. The term Oligodactyly may be used as a synonym, but only in situations where the missing digit is not specified. For example, to state that a patient has Oligodactyly F5 is awkward because the oligodactyly refers to the digits that are present and the F5 refers to the digit that is absent, which is awkward and could become confusing with higher order deficiencies.

Replaces: Hypodactyly

Synonym: Oligodactyly

Toe, broad

Definition: Visible increase in width of the non-hallux digit without an increase in the dorso-ventral dimension (Fig. 72). *subjective*

Comment: Note that the girth may be increased in a broad toe, but this must be distinguished from ***Macrodactyly*** because in ***Macrodactyly*** the length is increased as well. The affected digit should be specified. Note that this assessment may be difficult when the toes are short. This term is not used for the first digit, see ***Broad hallux.*** If all five digits are broad, both terms should be used for that patient.

Toes, cutaneous syndactyly of

Definition: A soft tissue continuity in the A/P axis between adjacent foot digits that involves at least half of the P/D length of one of the two involved digits (Fig. 73). *objective*
OR

A soft tissue continuity in the A/P axis between two digits of the foot that does not meet the prior objective criteria. *subjective*

Comment: The digits (or part of) are joined together by soft tissue that is not normally present between the digits at that point in the P/D axis. The definition of toe syndactyly is subtly different from that of the hand. The definition used for the hands was thought to be difficult with toes, as the phalangeal lengths are small and an assessment of the degree of syndactyly is impractical. A modifier of “complete” may be used if the syndactyly extends to the distal end of the nail bed. The affected digits should be specified.

Replaces: Syndactyly (without specification); Zygodactyly

Toe, great; all terms referring to the first toe are listed under ***Hallux*** or ***Toe***

Toe, hypoplastic: See ***Toe, small***

Toe, long

Definition: Digits that appear disproportionately long compared to the foot (Fig. 74). *subjective*

Comment: This finding must be distinguished from digits that are thin but of normal length and that of a short mid and hind foot with normal digit lengths. The affected digits should be

specified. The term arachnodactyly should be discontinued as it may be considered disparaging to liken a patient's digits to that of a spider. If only a subset of the digits of a limb is lengthened, the affected digits should be specified.

Replaces: Arachnodactyly

Toe, narrow: See ***Toe, slender***

Toes, overlapping

Definition: Describes a foot digit resting on the dorsal surface of an adjacent digit when the foot is at rest (Fig. 75). *objective*

Comment: This descriptor is ordered depending on which toes are involved. The overriding toe is labeled, as specified in the introduction (item 3): e.g., T3,4. The ordering of the numbers specifies which toe is dorsal, i.e., with dorsum of the foot facing upward the toe on top is/are recorded first separated by a comma from the digit that is/are overlapped. Toes that are laterally deviated, but do not rest on top of adjacent toes should be coded as ***Clinodactyly***.

Toe, partial absence of

Definition: The absence of a phalangeal segment of a toe or hallux (Fig. 76). *objective*

Comment: The part that is absent may be specified. The "distal" modifier specifies the loss of the distal phalanx; clinically this is defined by the absence of the nail. The "proximal" modifier specifies the loss of the proximal or middle phalanx with the nail still present. It may be difficult to know which phalanx is absent without X-rays and even then, the missing bone may not be identified (note no attempt is made to distinguish missing middle from proximal phalanges). In this situation the location adjective should be removed. Note that this finding is distinct from ***Short toes,*** (which are reduced in length but have the normal number of phalangeal segments). ***Partial absence of the hallux*** is an alternative term that may be used for the first toe.

Replaces: Hypophalangy

Toe, short

Definition: Digits that appear disproportionately short compared to the foot (Fig. 77). *subjective*

Comment: This finding must be distinguished from digits that are of increased girth but of normal length and that of a long mid- and hind foot with normal digit lengths. The affected digits should be specified as described in the introductory comments. Note that we designate brachydactyly as a synonym, but this use of the term is distinct from the use of the same word in Bell's classification (see ***Short fingers***).

Synonym: Brachydactyly of the foot

Toe, short distal phalanx of

Definition: Short distance from the end of the toe to the most distal interphalangeal crease or DIPJ flexion point (Fig. 78). *subjective*

Comment: This term differs from ***Partial absence of the toe*** because in that term the phalanx must be missing, whereas here it may be small, but present. Relative shortening of the distal phalanges of the toes can be harder to assess than in the fingers, as they are

normally quite short. Distal phalangeal lengths can be assessed subjectively by comparing that digit segment to the rest of the digit, to other normal digits in that patient, or to typical patients of that age or build. Although flexion creases can be useful in the hand (see ***Short distal phalanx of the finger,***), this may not be practical in the foot. Alternatively, one may be able to palpate the joint.

Synonym: Brachytelephalangy

Replaces: Short terminal phalanx of the hallux

Toe, slender

Definition: Digits are disproportionately narrow (reduced girth) for the hand/foot size or build of the individual (Fig. 79). *subjective*

Comment: The affected digits should be specified. The assessment of this finding is difficult when the digits are long. The bundled and pejorative term “arachnodactyly” is replaced by the separate descriptors ***Long toe*** and ***Slender toe***.

Synonym: Narrow digit; Narrow toe

Replaces: Thin digits; Arachnodactyly

Toe, small

Definition: Significant reduction in both length and girth of the toe compared to the contralateral toe, or alternatively, compared to a typical toe size for an age-matched individual (Fig. 80). *subjective*

Comment: This is an acknowledged bundled term. There are no standards for this finding, clinical judgment must be used. The affected toes should be numbered.

Toes, splayed

Definition: Divergence of digits along the A/P axis (in the plane of the sole) (Fig. 81). *subjective*

Comment: This may be associated with ***Macrodactyly*** but this should be recorded separately. The affected digits should be specified.

Synonym: Spreading of the digits

Toe, tapered

Definition: The gradual reduction in girth of the digit from proximal to distal (Fig. 82). *subjective*

Comment: If the digits are not uniformly affected, they should be specified. If not specified, it refers to all the digits of the foot.

Toe, thick: See ***Toe, broad***

Toe, thin: See ***Toe, slender***

Toe, wide: See ***Toe, broad***

Toes, widely spaced

Definition: An overall widening of the spaces between the digits (Fig. 83). *subjective*

Comment: This description is based on the width of the gap between the toes. It is usually used when the width of the toes remains normal rather than to describe a situation where the toes are thin or narrow. This term should not be used for the situation where the finding is limited to a gap between T1,2 (see ***Sandal gap***).

Ulnar clubhand: See ***Hand, ulnar deviation of***

Ulnar polydactyly: See ***Hand, postaxial polydactyly of***

Zygodactyly: See ***Fingers, cutaneous syndactyly of; Toes, cutaneous syndactyly of***

CREASES

See Fig. 84 for a diagram of the three major palmar creases. One end of the distal transverse crease is on the radial side of the hand proximal to the base of the index finger or the second interdigital space and extends toward the ulnar side of the palm. One end of the proximal transverse crease begins on the radial (anterior) side of the palm in the first interdigital space and extends across the palm towards, but does not typically reach, the ulnar side of the palm. One end of the thenar crease is typically coincident with the radial part of the proximal transverse crease and extends proximally toward the wrist.

Crease, simian: See ***Palmar crease, single transverse***

Palmar crease, absent

Definition: The absence of a major crease of the palm (distal transverse crease, proximal transverse crease, or thenar crease) (Fig. 85). *subjective*

Comment: This term is not used to describe the “missing” crease in the case of a patient with a single transverse palmar crease.

Palmar crease, bridged

Definition: A crease that connects the proximal and distal transverse palmar creases (Fig. 86). *objective*

Comment: The crease that connects the two transverse creases should itself be more in the transverse (antero-posterior) than longitudinal (proximo-distal) orientation.

Synonym: Transitional palmar crease

Palmar creases, decreased

Definition: Poorly defined or shallow palmar creases (Fig. 87). *subjective*

Comment: This is a completely subjective term, requiring an experienced observer to distinguish this finding from common variation in creases. It refers to an overall diminution of the creases, not to diminution of a subset of the creases.

Replaces: Smooth palms

Palmar crease, deep

Definition: Excessively deep creases of the palm (Fig. 88). *subjective*

Comment: Like the term ***Decreased palmar creases***, this assessment requires an experienced observer to distinguish this from common crease variation. One view is that deep creases are those in which lint could still get stuck, even if the palm is fully opened.

Palmar crease, single transverse

Definition: The distal and proximal transverse palmar creases are merged into a single transverse palmar crease (Fig. 89). *objective*

Comment: The term “five finger crease” for the proximal transverse crease is sourced from [Hall et al., 2007]. The subtypes of single transverse palmar crease are not recognized here [Hook et al., 1974]. Replaces the term “Simian crease” as that term is disparaging and less descriptive.

Replaces: Simian crease

Palmar crease, transitional: See ***Palmar crease, bridged***

Palms, smooth: See ***Palmar creases, decreased***

Plantar crease, deep longitudinal

Definition: Narrow, paramedian longitudinal depressions in the plantar skin of the forefoot (Fig. 90). *subjective*

Comment: As described for a number of other terms, there are no thresholds or guides for the clinician to assess this finding. Instead, the clinician must rely on experience.

Sydney crease

Definition: Extension of the proximal transverse crease (five finger crease) to the ulnar edge of the palm (Fig. 91). *objective*

Comment: The proximal transverse (five finger) crease starts on the radial side of the hand near the base of the index finger and extends toward the ulnar side of the palm, but does not reach the ulnar side. In this finding, the crease extends completely to the ulnar margin of the palm.

NAILS

Brachyonychia: See ***Short nail***

Koilonychia: See ***Nail, concave***

Micronychia: ***See Nail, small***

Nail, bifid

Definition: A digit with two nails, with at least some soft tissue between them (Fig. 92). *subjective*

Comment: See the ***Preaxial polydactyly*** definition for a related point on bifid nails. The affected digits should be specified. If the patient has a partially duplicated digit with two completely separate nails, this term should not be used.

Nail, concave

Definition: The natural longitudinal (P/D) convex arch is not present or is inverted (Fig. 93). *subjective*

Comment: This often results in a saucer- or spoon-shaped nail and the free edge of the nail is typically everted. The affected digits should be specified. Note that the bundled term koilonychia is an abnormal shape of the fingernail where the nail has raised ridges and is thin and concave.

Replaces: Koilonychia; Spoon shaped nails

Nails, dystrophic: The term “dystrophic” is no longer accepted as a descriptor for nails with an unusual morphology. The specific characteristic should be described.

Nails, fused

Definition: A nail plate that has a longitudinal separation with partially separated nails, each with a separate lateral radius of curvature (Fig. 94). *subjective*

Comment: The use of the word “fused” is not meant to imply that pathogenetically these nails were separate and merged. This is distinct from a split or cleaved nail, where the two parts of the nails share the same radius of curvature. The involved digits should be specified. It may be associated with underlying syndactylous digits, but these are coded separately.

Nail, hypoplastic: See ***Nail, small***

Nail, hyperconvex

Definition: When viewed on end (with the digit tip pointing toward the examiner’s eye) the curve of the nail forms a tighter curve of convexity (Fig. 95). *subjective*

Comment: No objective standards were identified for this finding. Another way to describe this finding is to say that the observed curve has a smaller radius than does the typical nail. The affected digits should be specified.

Nail narrow

Definition: Decreased width of nail (Fig. 96). *subjective*

Comment: Standards for newborns were identified [Seaborg and Bodurtha, 1989] but none were identified for older persons. Therefore, for newborns, this could be objective, but this is rarely measured. The affected digits should be specified.

Nail pits

Definition: Small (typically about 1 mm or less in size) depressions on the dorsal nail surface (Fig. 97). *subjective*

Comment: The affected digits should be specified.

Nail, ridged

Definition: Longitudinal, linear prominences in the nail plate (Fig. 98). *subjective*

Comment: There may be only one, or several ridges. The affected digits should be specified.

Nail, short

Definition: Decreased length of nail (Fig. 99). *subjective*

Comment: Use this designation when the length is reduced but the width is normal. Standards for newborns were identified [Seaborg and Bodurtha, 1989] but none were identified for older persons. Therefore, for newborns, this could be objective, but this is rarely measured. The affected digits should be specified.

Synonym: Brachyonychia

Nail, small

Definition: A nail that is diminished in length and width (Fig. 100). *subjective*

Comment: This is a bundled term but is retained because of its common usage. This term may be used when the width and length are reduced, although it may be preferable to code the patient as having both ***Short nail*** and ***Narrow nail***. Standards for newborns were identified [Seaborg and Bodurtha, 1989] but none were identified for older persons. Therefore, for newborns, this could be objective, but this is rarely measured. The affected digits should be specified.

Replaces: Hypoplastic nails

Synonym: Micronychia

Nail, split

Definition: A nail plate that has a longitudinal separation and the two sections of the nail share the same lateral radius of curvature (Fig. 101). *subjective*

Comment: This is distinct from ***Fused nail***, where the two parts of the nail have a separate radius of curvature. The affected digits should be specified as described in the introductory comments.

Nails, spoon shaped: See ***Nail, concave***

Nail, thick

Definition: Nail that appears thick when viewed on end (Fig. 102). *subjective*

Comment: No objective standard for nail thickness could be identified although an unsupported claim suggests that nails are 0.5 mm in females and 0.6 mm in males (http://www.emedicine.com/orthoped/topic421.htm). There is a build up of keratin causing the nail plate to lift away from the nail bed. The thickened nail plate is usually very hard.

Replaces: Onychogryposis; Pachyonychia

Nail, thin

Definition: Nail that appears thin when viewed on end (Fig. 103). *subjective*

Comment: No objective standard for nail thickness could be identified. An unsupported claim suggests that nails are 0.5 mm in females and 0.6 mm in males (http://www.emedicine.com/orthoped/topic421.htm). Thin nails are usually brittle, may easily fray, or break at the free edge. Thin nails usually grow slowly but this definition does not require slow growth of the nail. Note that the term koilonychia is an abnormal shape of the fingernail where the nail has raised ridges and is thin and concave. Since it indicates also

other characteristics than thin nails, it should not be used to indicate this. The affected digits should be specified.

Onychogryposis: See ***Nails, thick***

Pachyonychia: See ***Nails, thick***

Acknowledgments

This paper was reviewed and edited by the co-Chairs of the International Dysmorphologic Terminology working group (Judith Allanson, Leslie G. Biesecker, John Carey, and Raoul C. M. Hennekam). This review and editing was necessary to increase the consistency of formatting and content among the six manuscripts [Allanson et al., 2009b; Carey et al., 2009; Hall et al., 2009; Hennekam et al., 2009; and Hunter et al., 2009] and this paper. While the authors of the papers are responsible for the original definitions and drafting of the papers, final responsibility for the content of each paper is shared by the authors and the four co-Chairs. Images were provided by several of the authors, Raoul C. M. Hennekam, Helen Hughes, and Julia Fekecs. The efforts of LGB were supported by the Intramural Research Program of the National Human Genome Research Institute of the National Institutes of Health.

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Fig. 1.
Adactyly of the feet, bilateral.

Fig. 2.
Camptodactyly of F45, bilateral.

Fig. 3. Clinodactyly, radial, F5, bilateral

This person also has ***Short finger, F5, bilateral, subjective***. See also Figs. 13, 20.

Fig. 4. Clubbing

Note that the clubbing is best viewed laterally (e.g., the thumb) whereas it is difficult to appreciate in the other digits viewed dorsally. See also Figs. 5 and 63.

Fig. 5. Prominent digit pads, right hand, F3,4

Note that this patient also has ***Clubbing*** and a ***Single transverse palmar crease***.

Fig. 6.
Digital constriction rings, left hand, F1-5, near MCPJ.

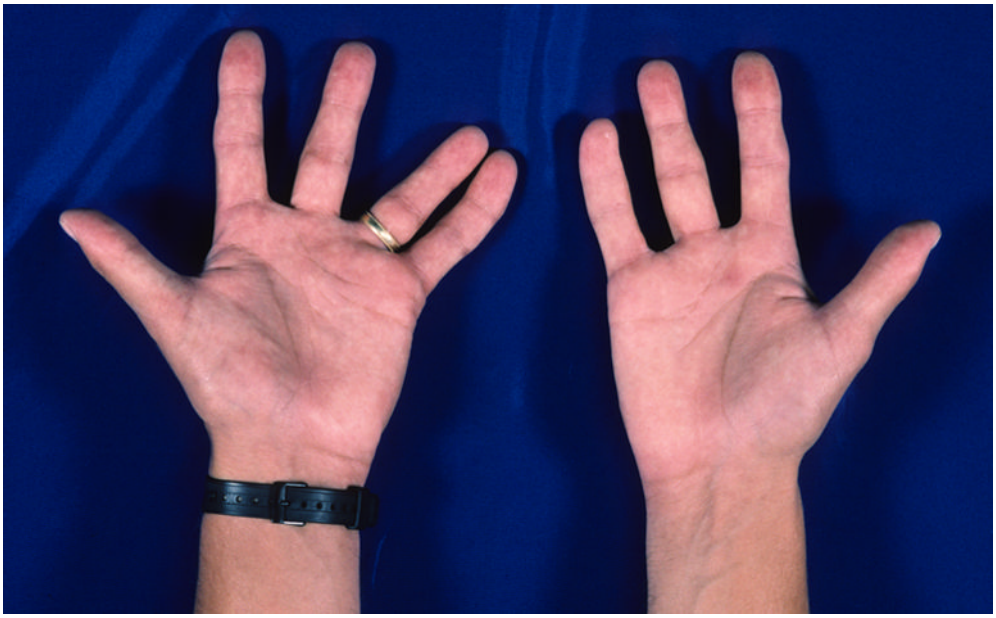

Fig. 7. Finger, absent, right hand

Note here that the identity of the missing digit is not specified, as there are no clinical data to allow this to be determined. See also Fig. 68.

Fig. 8. Broad fingers, right hand

Appreciate that the dorsal-ventral dimension of these digits is not increased, whereas the lateral (proximo-distal) dimension is increased. See also Fig. 99.

Fig. 9.

A. ***Cutaneous syndactyly of F2-5, left hand, complete, objective.*** Note that this patient also has a ***Broad thumb***, and ***Postaxial polydactyly of the hand, type B*** and ***Fused nails F2- 5***. B. This patient has ***Cutaneous syndactyly of F2-4, partial, subjective***. See also Fig. 14.

Fig. 10.
Long fingers, right hand, subjective.

Fig. 11.

A. ***Overlapping fingers, right hand, F54, F56.*** Note that this patient also has ***Mesoaxial polydactyly*** and ***Postaxial polydactyly, type B***. B. ***Overlapping fingers, right hand, F23, F54***. See also Figs. 37, 45, and 58A.

Fig. 12. Partial absence of fingers, left hand, F2-5, proximal
Note that this patient also has ***Small nails, F1-5***.

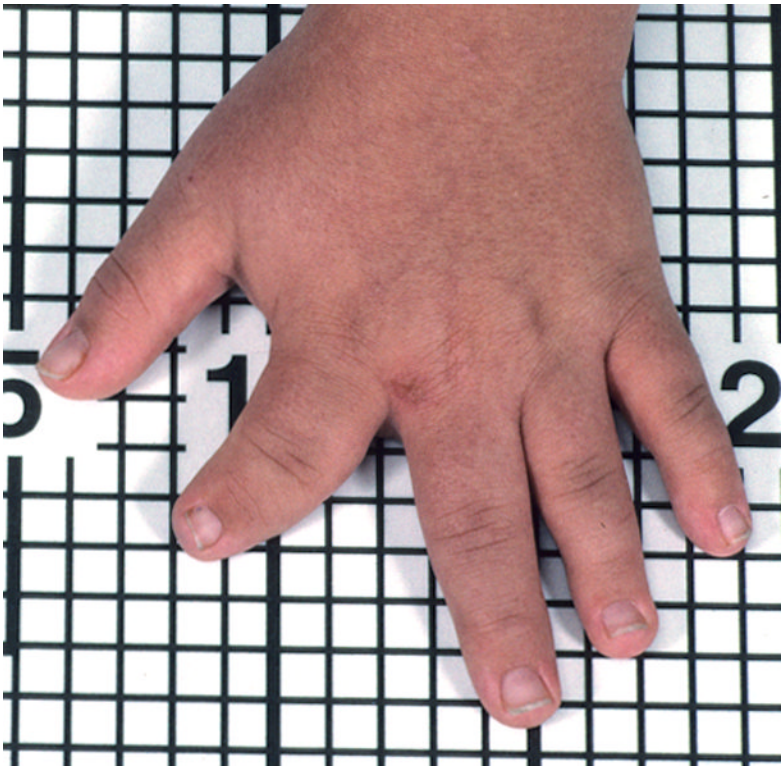

Fig. 13. Radial deviation of the finger, left hand, F2

Note that this patient also has ***Clinodactyly, F2, radial*** and that these two findings are distinct. See also Fig. 47.

Fig. 14. Short fingers, subjective

See also Figs. 3, 47, 69, and 99. Note that this patient also has ***Short palms, subjective*** and ***cutaneous syndactyly of F4,5, right hand, objective***.

Fig. 15. Short distal phalanx of the finger, left hand, F1
Note that this patient also has a ***Short nail, F1***. See also Fig. 60A.

Fig. 16. Slender fingers, right hand, F2-5
Note that only some of the digits are clearly shown in this figure.

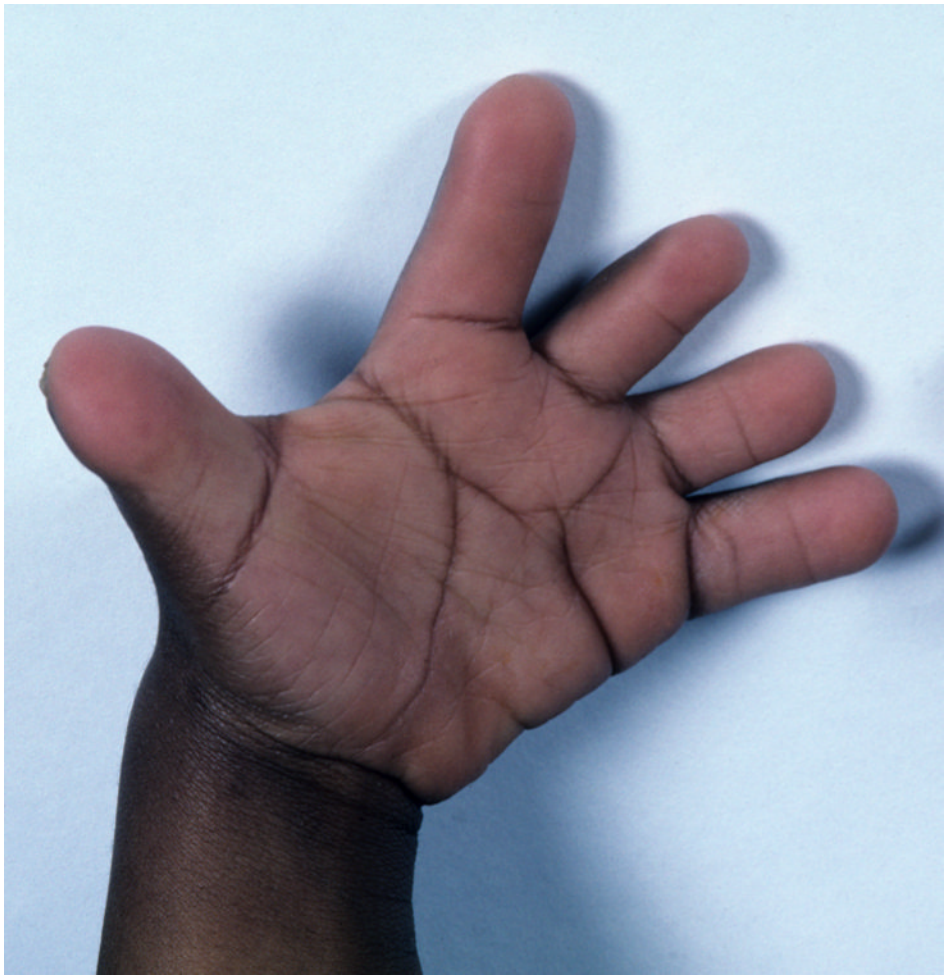

Fig. 17. Fingers, small, left hand, F3,4
This patient also has ***Deep palmar creases***.

Fig. 18. Splayed fingers, left hand, F23, F34
Note that this patient also has ***Macrodactyly F2-4***

Fig. 19.

A. ***Tapered finger, left hand, F4.*** B. ***Tapered fingers, right hand, F2-5.*** See also Figs. 42, 44, and 93.

Fig. 20. Ulnar deviation of finger, right hand, F4

Note that this patient also has ***Clinodactyly of F4, ulnar***. Note that his middle finger manifests ***Clinodactyly radial, F3***, but that this finger is not deviated, demonstrating the distinction of these two features. See also Fig. 45.

Fig. 21.

A. ***Broad fingertip, right hand, F1.*** Note how the digit widens at the IPJ. B. ***Broad fingertips, left hand, F3,4***. See also Fig. 93.

Fig. 22. Absent foot, right

Note that this is the same patient as is shown in Fig. 27. One limb has partial absence of the foot and the other complete.

Fig. 23. Broad foot, left

In this example, the forefoot appears more broad than does the midfoot, but this distinction is not noted in the nomenclature.

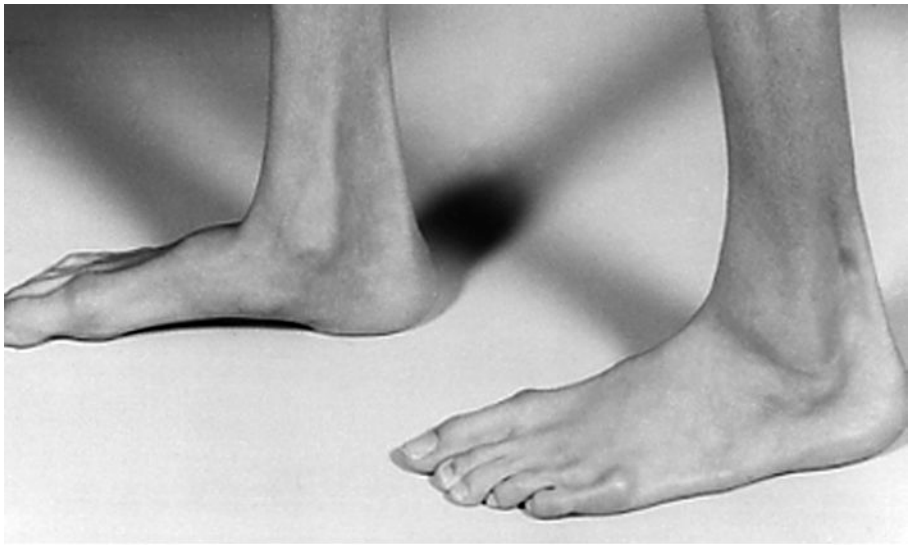

Fig. 24. Long feet, subjective
Note that this patient also has ***Long toes***.

Fig. 25. Narrow foot, left, subjective

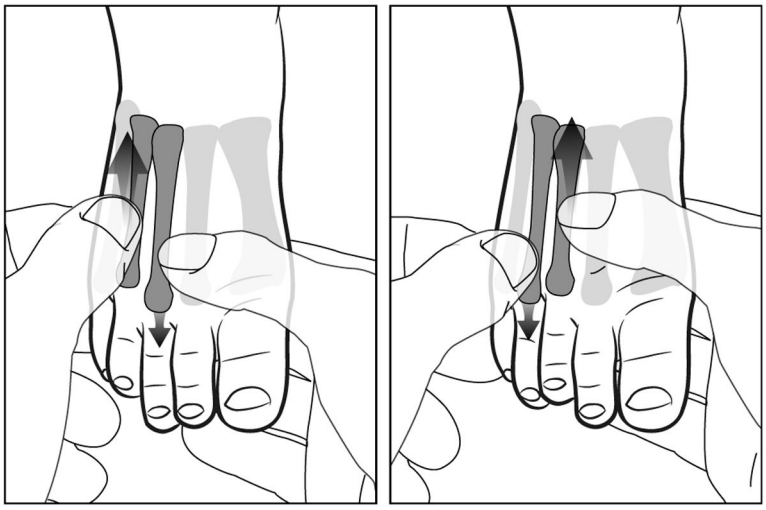

Fig. 26. Foot, osseous syndactyly of

Fig. 27. Foot, partial absence of, left

Note that this is the same patient as is shown in Fig. 22. One limb has complete absence of the foot and the other partial.

Fig. 28. Postaxial polydactyly of the left foot
Note that this patient also has ***Small nails***.

Fig. 29.

A. ***Preaxial polydactyly of the right foot.*** Note that this duplication is nearly complete. B. ***Preaxial polydactyly of the right foot.*** This patient has the same finding, but the duplicated digits are more separated than in Fig. 29A.

Fig. 30.
Rocker bottom foot, left.

Fig. 31.
Short feet, subjective.

Fig. 32.

A. ***Split foot, left.*** **B.** ***Split feet.*** Note that this patient has a more severe, deeper notch in the feet than does the patient in A. The shape and numbers of other affected digits is highly variable.

Fig. 33.
Absent hallux, right.

Fig. 34. Broad hallux, right
See also Fig. 60B.

Fig. 35. Hammertoe, T3, left
See also Fig. 57.

Fig. 36. Absent hand, left

Note that this patient also has a shortened radius and ulna, although that finding is not required for this finding.

Fig. 37. Clenched hand, right

Note that the left hand does not warrant the term because not all of the digits are completely flexed at the MCPJ and IPJ. The left hand would warrant the finding of ***Overlapping fingers F23, F54, left.***

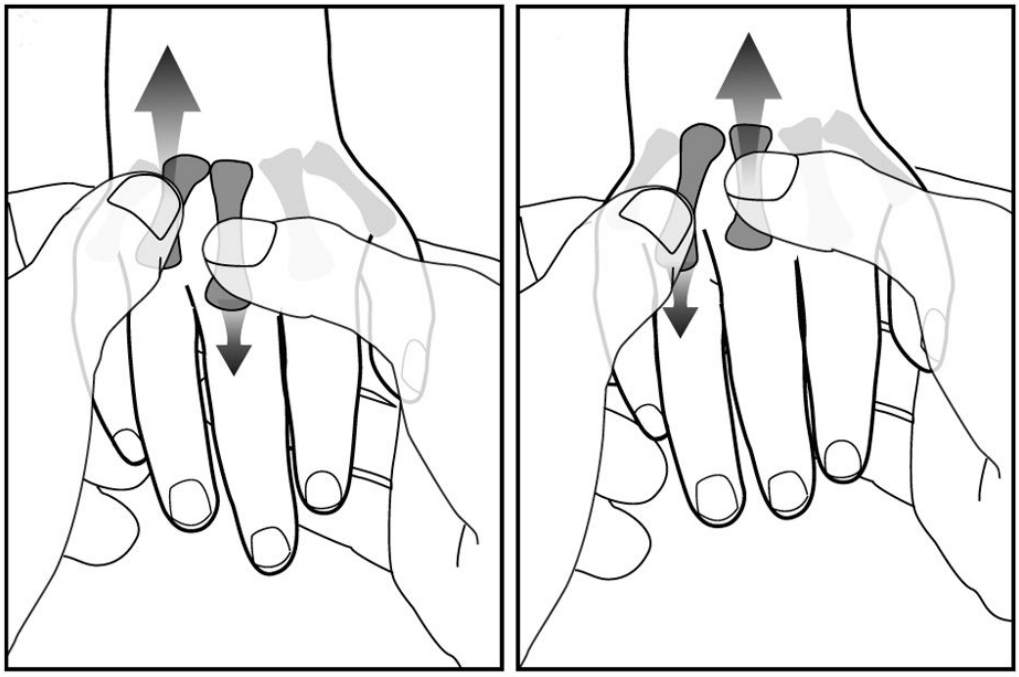

Fig. 38. Osseous syndactyly of the hand

This figure shows the maneuver used to detect this finding. The abnormal finding is not shown. The examiner grasps two adjacent metacarpals and alternately moves them to determine if they are fused or independent.

Fig. 39.

A. ***Postaxial polydactyly of the right hand, type A.*** B. ***Postaxial polydactyly of the right hand.*** Note that this patient has a digit that is intermediate between type A and type B, so

that is not specified. See also Figs. 49A, 58A, and 73A. See Figs. 9A and 11A for examples of ***Postaxial polydactyly, type B***.

Fig. 40.
A Preaxial polydactyly of the left hand, partial. B. Preaxial polydactyly of the right hand

Fig. 41.
Radial deviation of the hand, right.

Fig. 42. Small hands
Note that this patient also has ***Tapered fingers***.

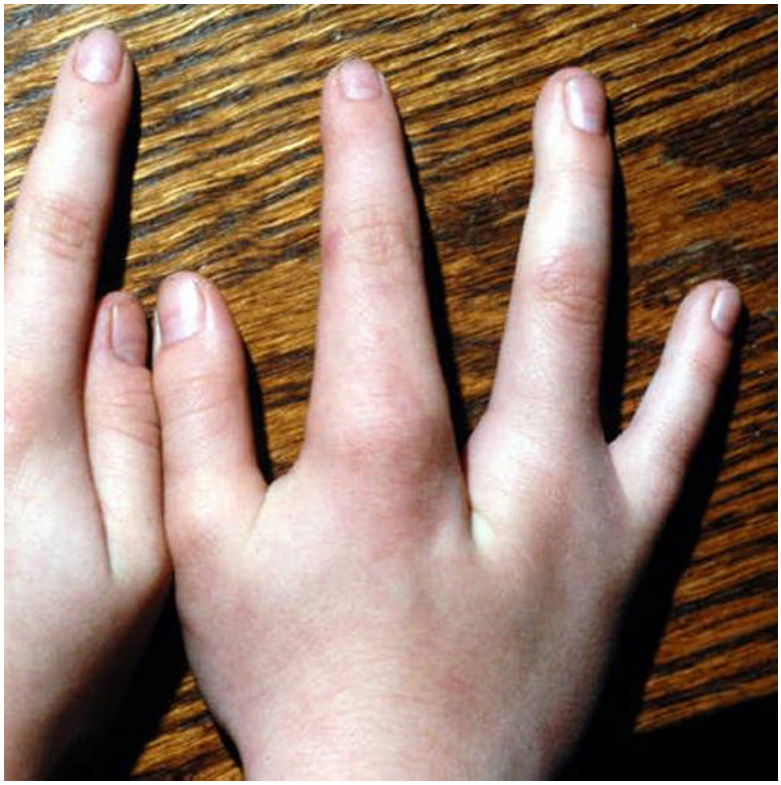

Fig. 43. Split hand, right

Fig. 44. Trident hand, left

Note that this patient also has ***Tapered fingers***, but that is not required for the finding.

Fig. 45. Ulnar deviation of the right hand

Note that this patient also has ***Ulnar deviation of the fingers, F2-3*** and ***Overlapping fingers F45.***

Fig. 46.
Prominent heels.

Fig. 47. Hypothenar hypoplasia, left hand

Note that this patient also has ***Short fingers F2-5*** and ***Radial deviation of fingers F2-3***.

Fig. 48.

A. ***Macrodactyly of F2-3, left hand.*** Note that this person also has Clinodactyly, but that finding should be coded separately. B. ***Macrodactyly of T1-2, right foot.*** This patient also has a ***Sandal gap***. See also Figs. 18, 81, and 83.

Fig. 49.

A. ***Short metacarpal, F5, left hand.*** Note that this patient also has a ***Postaxial polydactyly, partial***. B. ***Short metacarpals, F34, left hand F4, right hand***. Note that this patient's hands are shown in dorsal view, with the fingers flexed, which can facilitate the recognition of this finding.

Fig. 50.
Short metatarsals, T3,4, bilateral.

Fig. 51. Metatarsus adductus, left
Note that this patient also has a ***Sandal gap***.

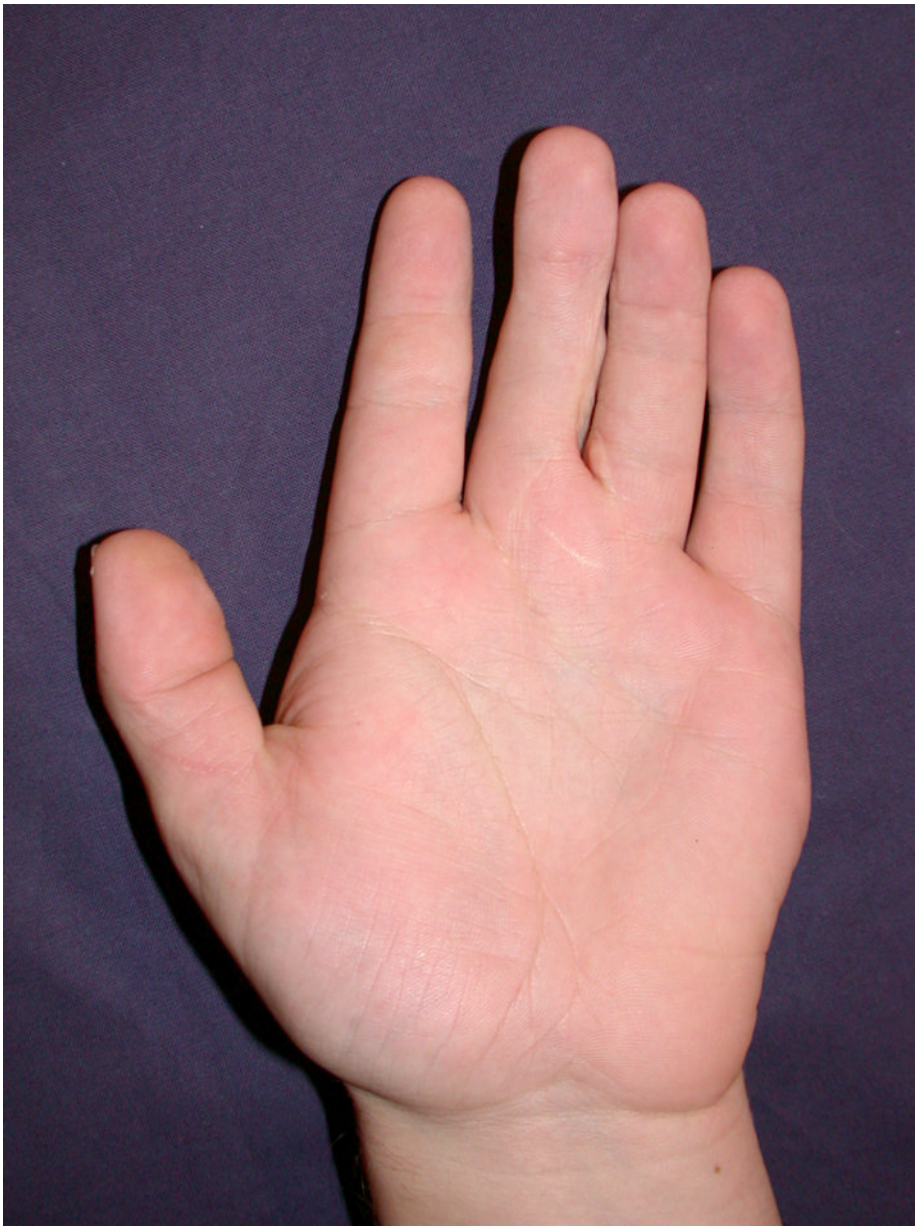

Fig. 52. Broad palm, left, subjective

Note that this patient had normal palm length, so that this is not an example of a short palm with only an apparently broad palm.

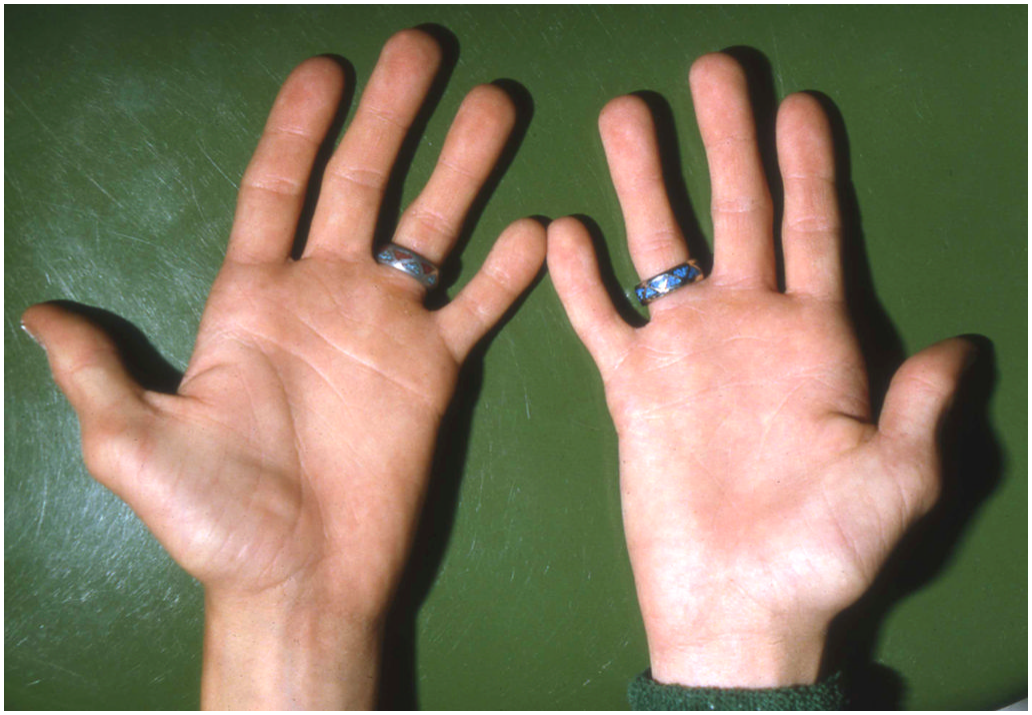

Fig. 53. Long palms

Note that this patient had normal palm width, so that this is not an example of a narrow palm with only an apparently broad palm. Note that this image illustrates a number of other features, which are not delineated. See also Figs. 63 and 85.

Fig. 54. Narrow palms, subjective

Note that this patient had normal palm width, so that this is not an example of a long palms with only apparently narrow palms.

Fig. 55. Short palms, subjective

Note that this patient also has ***Short fingers***. This patient does not warrant a finding of ***Small hands***, because the fingers are of normal caliber. See also Fig. 14.

Fig. 56. Pes cavus

Note that this patient could also be said to have ***pes equinus***, but that term is not included in the current terminology set.

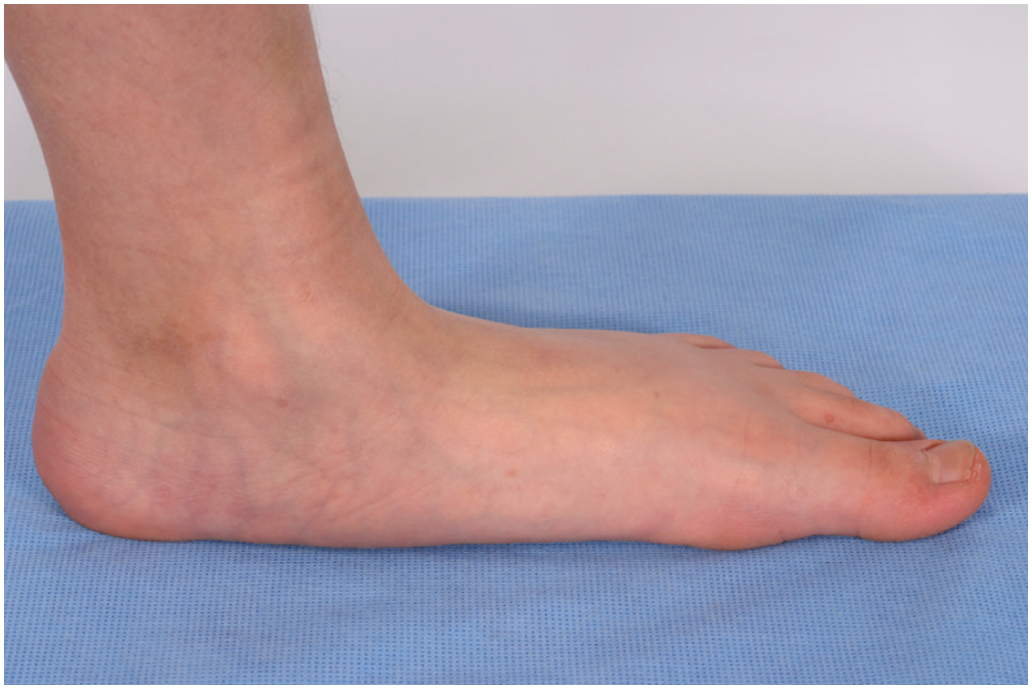

Fig. 57. Pes planus, left foot, objective
Note that this patient also has ***Hammertoes bilateral T2***.

Fig. 58.

A. ***Mesoaxial polydactyly of the hand, right.*** Note that this patient has heptadactyly, manifesting as well ***Postaxial polydactyly, type B***. The figure also shows ***Small nail, F6*** and ***Overlapping fingers F56.*** See also Fig. 11A. B. ***Mesoaxial polydactyly of the right foot.*** This patient also has ***Cutaneous syndactyly of the toes, partial, T23***.

Fig. 59. Mirror image polydactyly, right foot

Note that this patient also has ***Cutaneous syndactyly*** of numerous digits, notable on the right foot.

Fig. 60.

A. ***Absent ray, right hand.*** Note that this patient also has ***Short distal phalanges of the fingers*** and ***Short nails.*** B. ***Absent ray, left foot.*** Note that this patient also manifests a ***Broad hallux***, which should be coded separately.

Fig. 61.
A. ***Sandal gap, right foot, subjective.*** B. ***Sandal gaps, objective.*** See also Figs. 48 and 51.

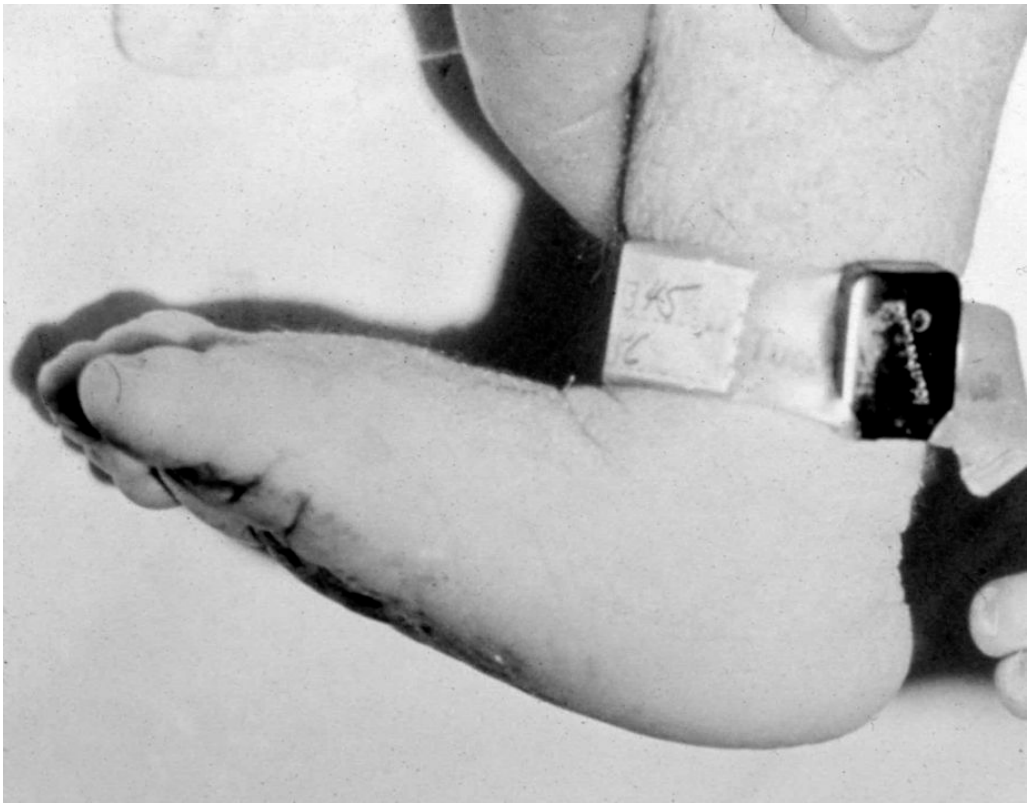

Fig. 62. Convex contour of the sole right foot

Note the heel in this patient is not sufficiently prominent to warrant the descriptor of ***Rocker bottom foot.***

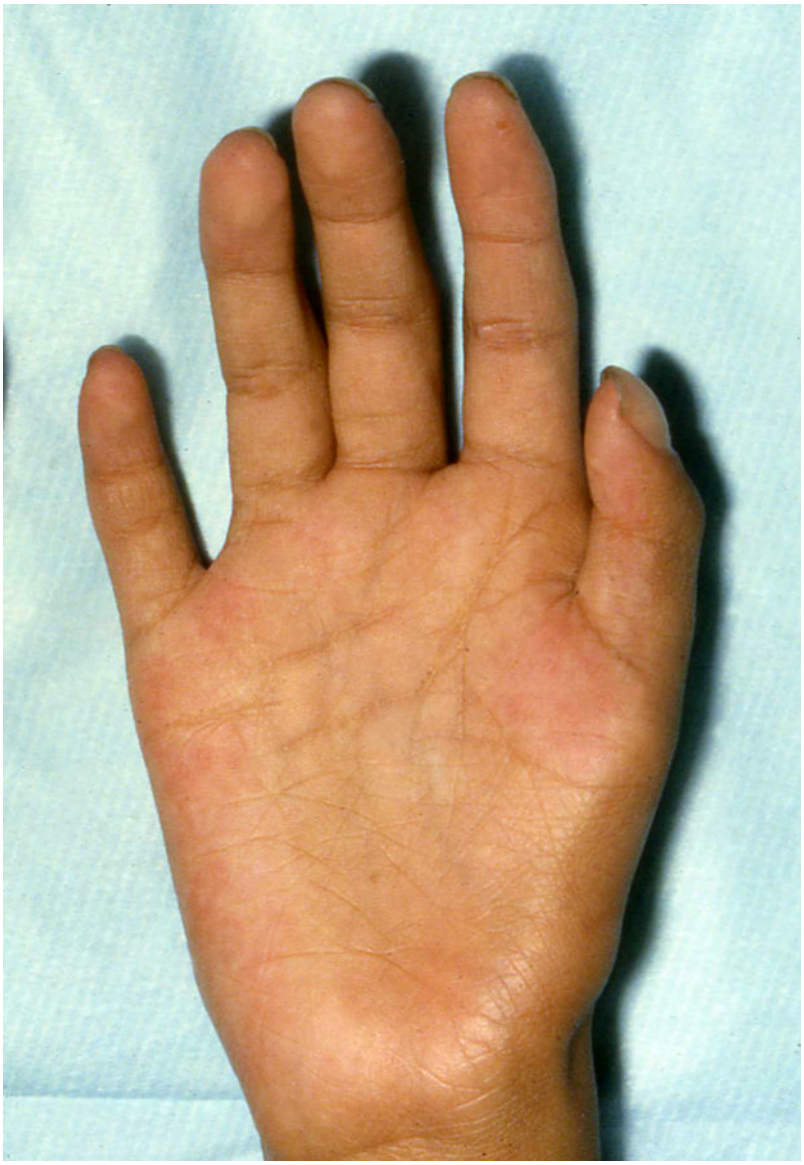

Fig. 63. Thenar hypoplasia right hand

Note that this person also has ***Clubbing*** (visible in the thumb only). This patient also has ***Long palm, right hand, subjective.***

Fig. 64.
Absent thumb, left.

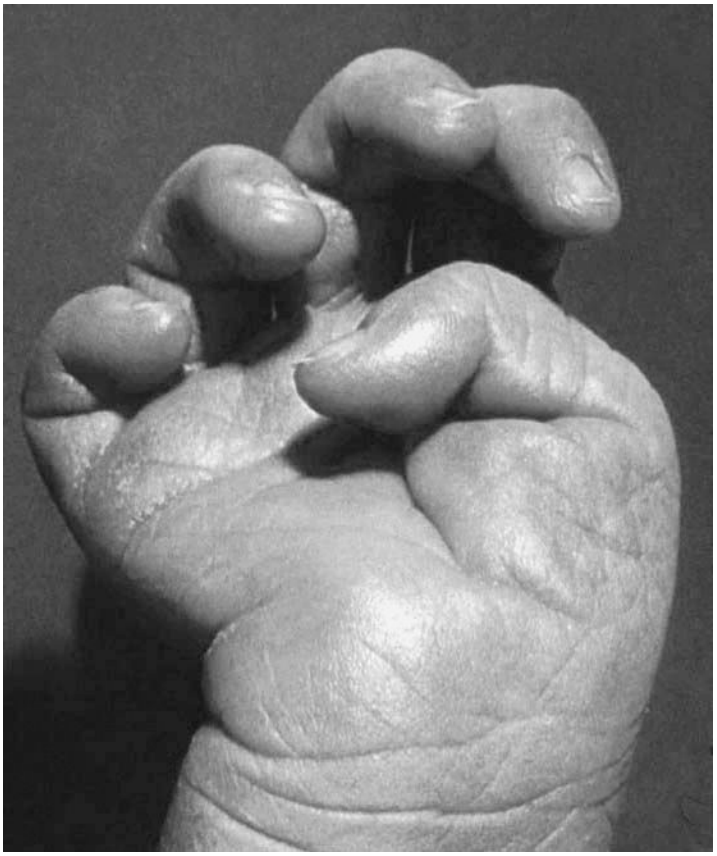

Fig. 65.
Thumb, adducted, right.

Fig. 66. Broad thumbs
See also Fig. 9A.

Fig. 67.
Hitchhiker thumb, right.

Fig. 68. Thumb, partial absence of, right

Note that this patient also has ***Absent fingers, F2-4***, and ***Absent finger F5, partial***.

Fig. 69. Thumb, proximal placement of, right hand, subjective
Note that this patient also has ***Short finger, F2***.

Fig. 70.
Triphalangeal thumb, right.

Fig. 71.
Absent toe, right.

Fig. 72. Toe, broad, left, T1
See also Figs 92, 94, and 101.

Fig. 73.

A. ***Cutaneous syndactyly of the toes, complete, TT1-6, left, objective.*** See also Figs. 58B, 59, 78, 92, 94, and 101. Note that his patient also has ***Preaxial polydactyly of the foot,***

partial, ***left***. B. ***Cutaneous syndactyly of the toes, partial, T2-4, bilateral, objective.*** As specified in the definitions, this finding is objective if the syndactyly extends more than half the proximo-distal length of the digits.

Fig. 74. Long toes, left foot
See also Figs. 24 and 79.

Fig. 75.
Overlapping toes T45, bilateral.

Fig. 76. Toes, partial absence of, right foot, T2,4
Note that this is the same image as in Fig. 82, ***Toe, tapered.***

Fig. 77. Short toes, T1-5, right foot

Note that this patient also has ***Short distal phalanges of toes T2,3***.

Fig. 78. Short distal phalanges of toes T2-4, right foot

Note that this patient also has ***Cutaneous syndactyly of toes T23, subjective.*** See also Fig. 77.

Fig. 79. Slender toes, T2-5, right foot
Note that this patient also has ***Long toes T1-5***.

Fig. 80. Small toe, T1, left foot
See also Fig. 81.

Fig. 81. Splayed toes, T23 right foot, T12, left foot

Note that this finding differs from Toes, widely spaced, because in splayed toes the toes have a divergent axis of orientation. This patient also has ***Small toe, T1, right*** and ***Macrodactyly of T2,3, right, and T1,2, left***.

Fig. 82. Toe, tapered, T5

Note that this is the same image as in Fig. 76, ***Toe, partial absence***.

Fig. 83. Toes, widely spaced, T1,2, T4,5
Note that this patient also has ***Macrodactyly of T1,2***.

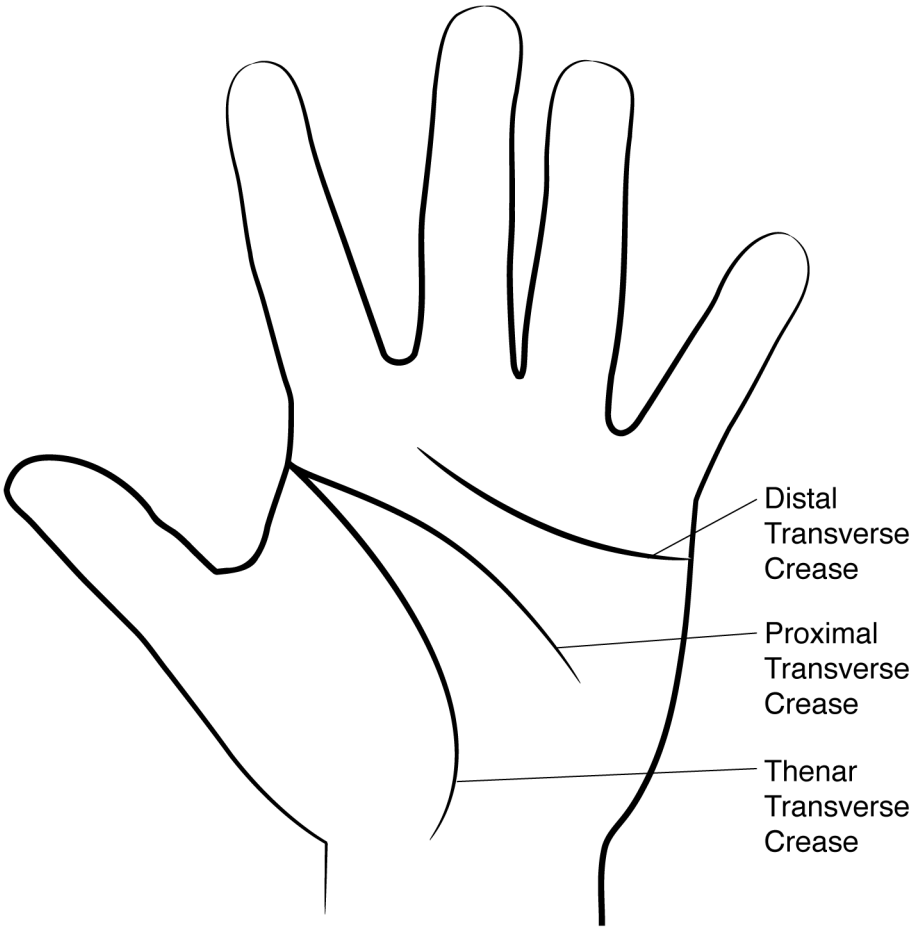

Fig. 84.
Normal palmar creases. See text for explanatory material.

Fig. 85. Palmar creases, absent, left hand

Note that this patient also has ***Long palm, subjective, left*** and ***Single transverse crease, right hand***.

Fig. 86.
Bridged palmar crease, right hand.

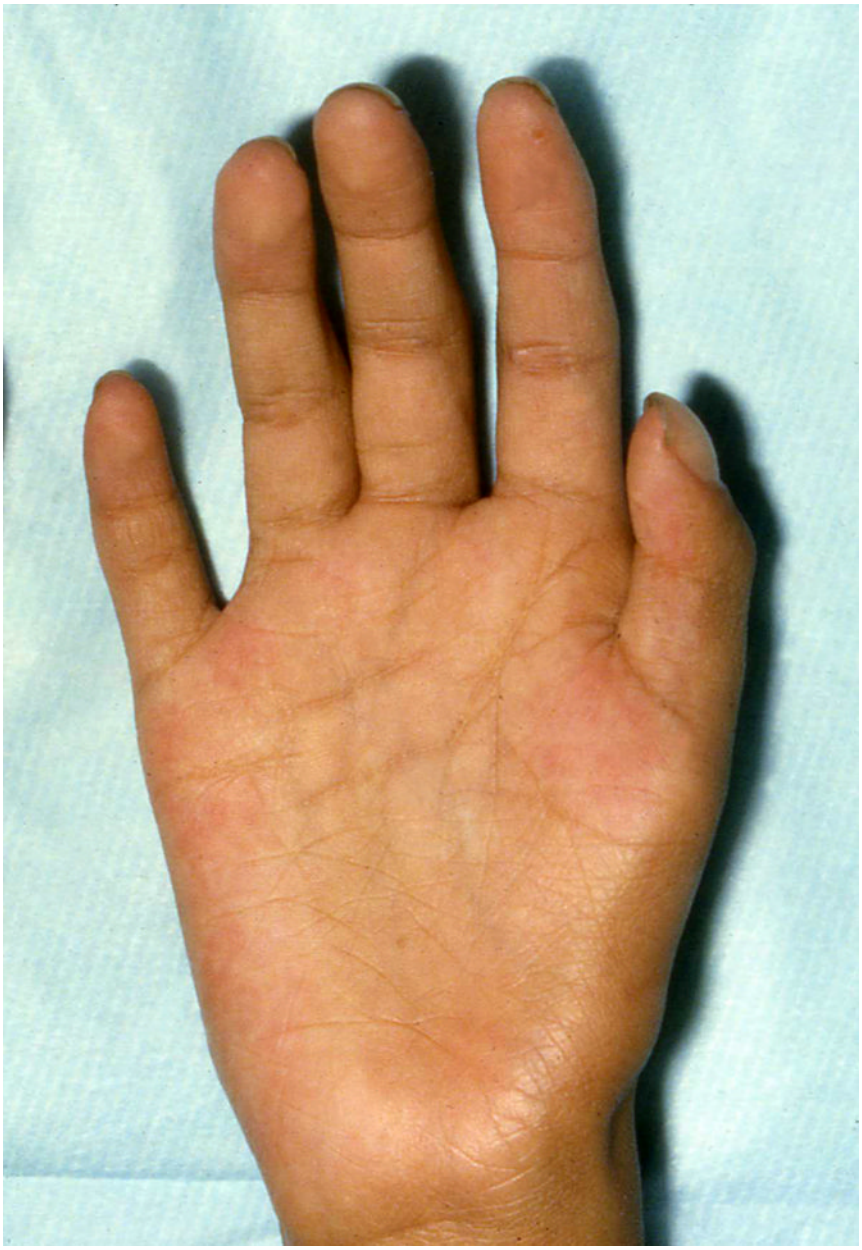

Fig. 87.
Palmar creases, decreased, right hand.

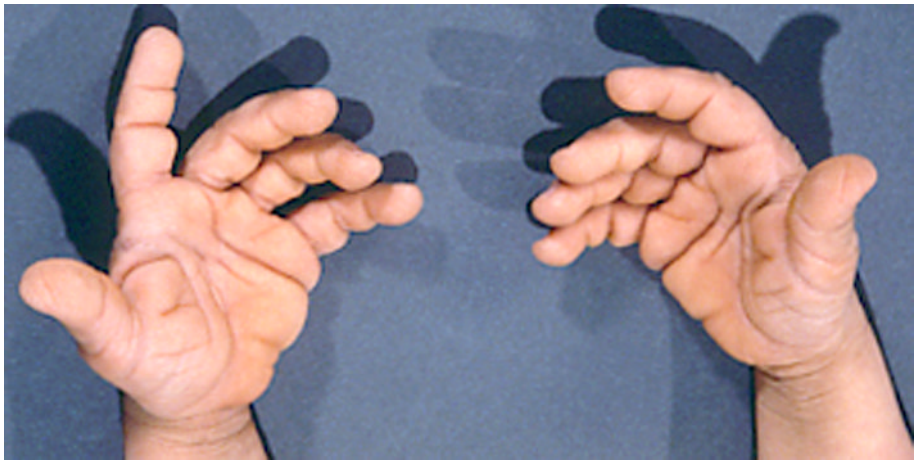

Fig. 88. Palmar creases, deep
See also Fig. 17.

Fig. 89. Single transverse palmar crease, right hand
See also Figs. 5 and 85.

Fig. 90.
Deep longitudinal plantar crease, right foot.

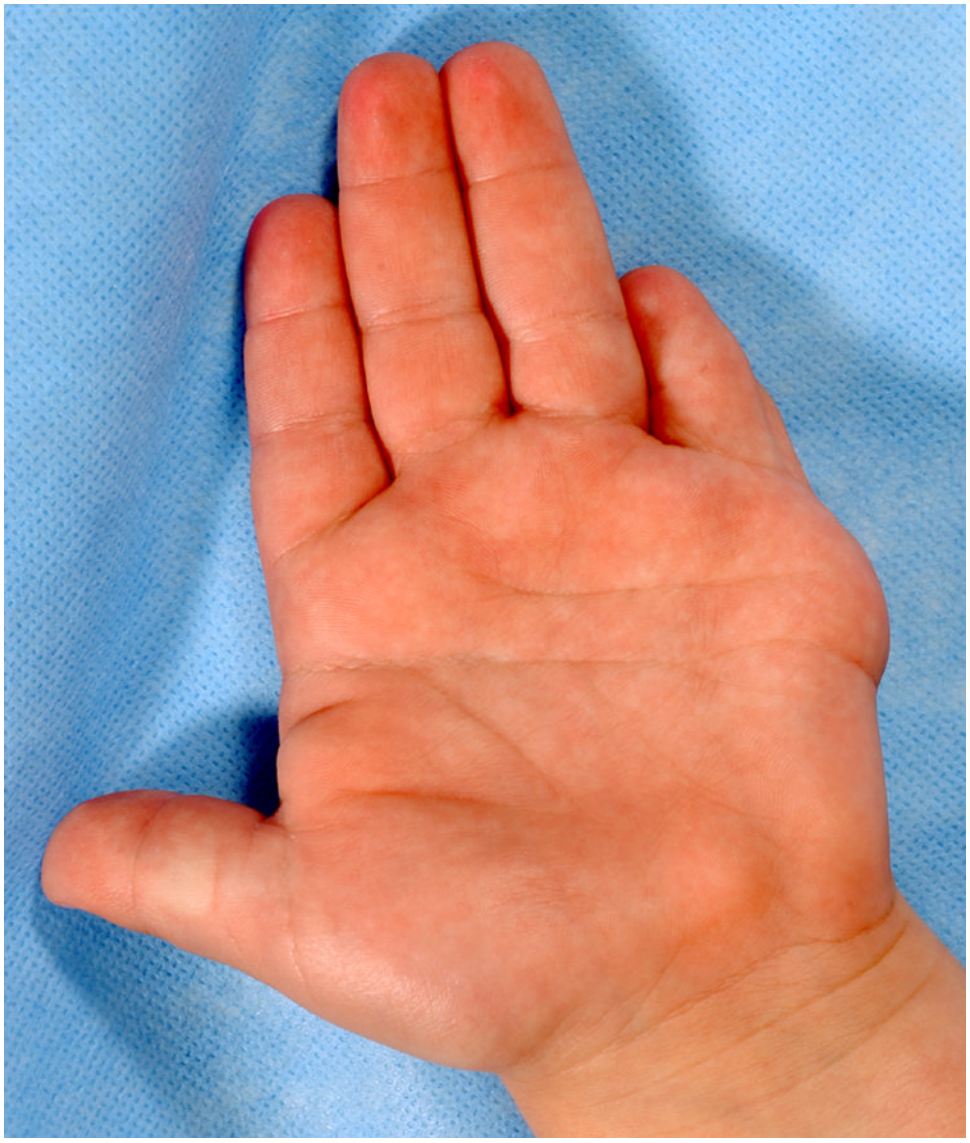

Fig. 91.
Sydney crease, left hand.

Fig. 92. Nail, bifid, T1, left foot

See also Fig. 99. Note that this patient also has a ***Broad toe, T1*** and ***Cutaneous syndactyly, partial, T23***.

Fig. 93. Nail, concave, left hand, F4

See also Fig. 103. Note that this image also shows ***Tapered fingers, left, F23***, and ***Broad fingertip, left, F4***.

Fig. 94. Nail, fused, T1, right foot

See also Fig. 9A. This patient also has a ***Broad toe, left, T1*** and ***Cutaneous syndactyly, partial, left, T23.***

Fig. 95. Nails, Hyperconvex, left, F3,4
See also Fig. 102.

Fig. 96. Nail, narrow, T2, left foot

Note that the length of this nail is normal, so it is coded as narrow, not small.

Fig. 97.
Nails, pitted, F2-4, right hand.

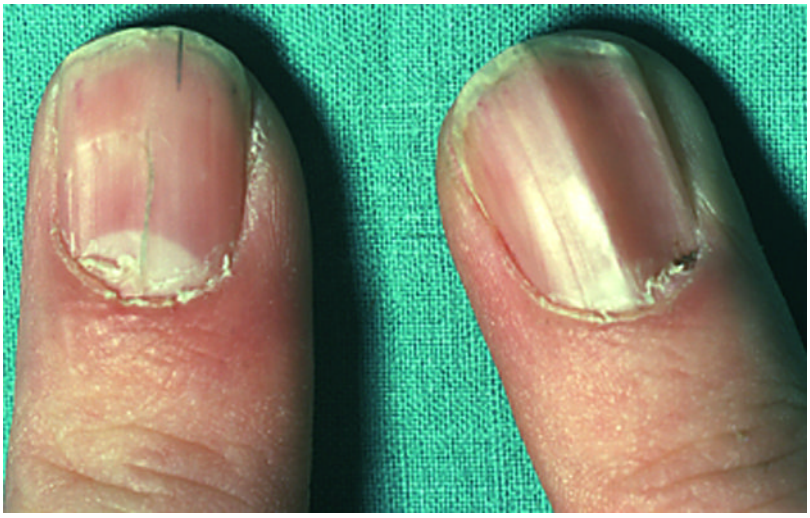

Fig. 98.
Nails, ridged, F1, bilateral.

Fig. 99. Nails, short, F1,5, left hand

See also Fig. 15 and 60A. Note that this patient also has ***Bifid nail, F5 left hand; Broad finger, F5, left hand***; and ***Short finger, left hand, F1***.

Fig. 100.
Nails, small, left foot. See also Figs. 12, 28, and 58A.

Fig. 101.
Nail, split, T1, left foot. Note that this image also shows a ***Broad toe, T1, left foot*** and ***Cutaneous syndactyly, partial, T23, left foot***.

Fig. 102.
Nails, thick, right hand, F1-5. Note that this image also shows ***Hyperconvex nails***.

Fig. 103.
Nails, thin, F2,5, right hand. This image also shows ***Concave nail, F2, left hand.***