Congenital Skeletal Limb Deficiencies*

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Until the present time, child amputees have been classified as having either non-congenital or congenital amputations. Within the group of congenital amputations, both the so-called true amputation (limb-bud arrest) and the gross limb abnormality are encountered. In the fields of embryology and teratology, limb deficiencies are described with a terminology in which the terms themselves are descriptive as are the implications of these terms. Writers in the field of orthopaedic surgery have failed to adopt a relatively consistent and accurate nomenclature—one which is clear and concise, as compared with the common vague phrase *congenital amputation of the . . . .*

The classification to be presented has been evolved in an endeavor to standardize nomenclature. It does not cover *reduplication* of limbs.

In the past fifteen years, a growing interest in the care of the child amputee has been manifested by the increasing number of amputee clinics that devote their efforts exclusively to the child. These centers have progressively increased the number of patients cared for with single or multiple limb deficiencies or abnormalities. Many of these conditions are bizarre and present problems in prosthetic prescription that cannot be solved by standard prostheses.

Since World War II, the science of prosthetics has advanced phenomenally. Stumps at all levels of amputation can be fitted successfully because of the many mechanical components now available to the prosthetist. Congenital anomalies, however, are a challenge to the orthopaedic surgeon and the prosthetist in that they may be entirely unsuitable for standard prostheses because of gross variations in limb contour, substandard muscle power, and underlying skeletal deficiencies.

The various abnormalities present a frustrating problem to the surgeon who attempts to categorize them, for example, the so-called lobster-claw deformity has no less than ten different synonyms: ectrodactylism, adactyly, oligodactyly, perodactyly, monodactyly, hypodactyly, pincers, claw, crayfish-claw, and crab-claw. All these terms are used to describe partial adactylia.

The term *congenital amputation* is sometimes still used for malformation of a limb formerly thought to be the result of constriction by an amniotic band. However, "most malformations are unaccompanied by amniotic adhesions; and even when these are present, they seldom provide a plausible explanation."[18] Intra-uterine amputations in the late months of pregnancy occur and have been documented.

The relevant literature relating to anomalies has been cited in a previous publication.[13] Certain more recent works are given in the present list of references.

The early development of the limbs has been investigated in detail in the human embryo during the past fifteen years.[14, 15, 16] The upper and lower limbs

appear first as small buds of tissue on the lateral body wall at four postovulatory weeks.[14] These buds grow and differentiate rapidly within the ensuing three weeks, and the various regions of the limbs develop in proximodistal sequence. The arm and forearm, for example, appear before the hand, and the thigh and leg before the foot.

The skeletal elements of the limbs are found first as condensations of mesenchyme within the limb buds. These condensations soon chondrify in a definite order and ossification follows. Initial bone formation is found in the clavicle at five postovulatory weeks, in the humerus, radius, ulna, distal phalanges of the hand, femur, and tibia at seven postovulatory weeks, and in the scapula and fibula at seven weeks.

Endochondral ossification in the shaft of a bone occurs generally from one to five weeks after initial bone-collar formation has taken place. By seven weeks, all the skeletal elements of the limbs (with the exception of the clavicle, which has a different mode of development) are present individually as cartilaginous models, some of which have collars of bone in their shafts. The precise time of appearance of the individual mesenchymal condensation, the area of chondrification, and the site of initial ossification in each of the skeletal elements of the limbs have been calculated and tabulated.[15, 16] By seven weeks, the joints are also well advanced and the larger synovial articulations, such as the shoulder and the hip, have begun to show cavitation. By this time, the skeleton is, in general, a replica in miniature of that of postnatal life.

It is clear that anomalies in which the number of skeletal elements is increased (for example, polydactylia and ulnar dimelia) must arise during the first seven weeks of intra-uterine life. At first sight it may seem that, due to failure of a skeletal element to persist in development, a decrease in the number of skeletal parts (for example, radial hemimelia) may arise after, as well as during, this seven-week period. Both descriptive and experimental embryological investigations, however, suggest that the various types of hemimelia arise at a very early embryonic phase.

It is to be noted that the word *postovulatory* refers to the length of time after the last ovulation, that is, the particular ovulation which resulted in the subsequently fertilized oocyte. The first seven postovulatory weeks are known as the embryonic period and the remainder of intra-uterine life as the fetal period. The distinction between an embryo and a fetus is based on the shift of emphasis in development in general from differentiation to growth. Because this distinction is too general to be of practical use, it is necessary to have available an arbitrary indicator. The level of development in the humerus is a convenient gauge of development. The onset of marrow formation in the humerus occurs when the human organism is about thirty millimeters, crown-to-rump length, at seven postovulatory weeks.

Seven descriptive terms are employed in the classification of the anomalies under discussion: *amelia*, *hemimelia*, *phocomelia*, *acheiria*, *apodia*, *adactylia*, and *aphalangia*. The first three terms are derived from the Greek *melos*, a limb: amelia means absence of a limb, and hemimelia means absence of a large part (*hemi*, a half) of a limb. Phocomelia, based on the Greek *phoke*, a seal, refers to a flipper-like limb, that is, a hand or foot which is attached more or less directly to the trunk. Acheiria and apodia are from the Greek *cheir*, a hand, and *pous*, *podos*, a foot. (These roots are found in such familiar words as chiropody and chirurgia; the latter is the Latin form from which the word *surgery*, literally meaning handwork, is derived.) Hence, acheiria and apodia mean absence of a hand and of a foot, respectively. Adactylia means absence of a digit (Greek *daktylos*, a digit) and is here reserved for those cases in which there is absence of the associated metacarpal or metatarsal as well as of the digit. Aphalangia naturally means absence of one or more phalanges.

TABLE I

CLASSIFICATION OF CONGENITAL SKELETAL LIMB DEFICIENCIES*

TERMINAL (T)

TRANSVERSE (-)

1. *Amelia* (absence of limb)
2. *Hemimelia* (absence of forearm and hand or leg and foot)
3. *Partial hemimelia* (part of forearm or leg is present)
4. *Acheiria or apodia* (absence of hand or foot)
5. *Complete adactylia* (absence of all 5 digits and their metacarpals or metatarsals)
6. *Complete aphalangia* (absence of one or more phalanges from all 5 digits)

LONGITUDINAL (/)

1. *Complete paraxial hemimelia* (complete absence of one of the forearm or leg elements and of the corresponding portion of the hand or foot) - R, U, TI, or FI†
2. *Incomplete paraxial hemimelia* (similar to above but part of defective element is present) - r, u, ti, or fi†
3. *Partial adactylia* (absence of one to four digits and their metacarpals or metatarsals) : 1, 2, 3, 4, or 5
4. *Partial aphalangia* (absence of one or more phalanges from one to four digits) : 1, 2, 3, 4, or 5

INTERCALARY (I)

TRANSVERSE (-)

1. *Complete phocomelia* (hand or foot attached directly to trunk)
2. *Proximal phocomelia* (hand and forearm, or foot and leg, attached directly to trunk)
3. *Distal phocomelia* (hand or foot attached directly to arm or thigh)

LONGITUDINAL (/)

1. *Complete paraxial hemimelia* (similar to corresponding terminal defect but hand or foot is more or less complete) - R, U, TI, or FI†
2. *Incomplete paraxial hemimelia* (similar to corresponding terminal defect but hand or foot is more or less complete) - r, u, ti, or fi†
3. *Partial adactylia* (absence of all or part of a metacarpal or metatarsal) : 1 or 5
4. *Partial aphalangia* (absence of proximal or middle phalanx or both from one or more digits) : 1, 2, 3, 4, or 5

* *List of symbols used:*

- transverse
/ longitudinal
: 1, 2, 3, 4, or 5 denote digital ray involved
FI or fi ---------- fibular

I ---------- intercalary
R or r ---------- radial
T ---------- terminal
TI or ti ---------- tibial
U or u ---------- ulnar

A line below a numeral denotes upper-limb involvement, for example, T-$\underline{2}$ represents terminal transverse hemimelia of the upper limb.

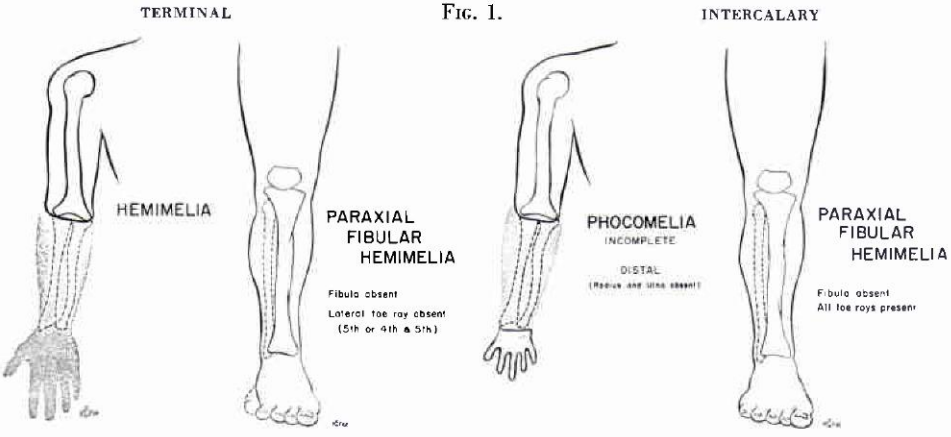
A line above a numeral denotes lower-limb involvement, for example, I-$\overline{1}$ represents intercalary transverse complete phocomelia of the lower limb.

† In capital letters when the paraxial hemimelia is complete, in small letters when the defect is incomplete.

Hemimelia may be complete (when the entire distal half of a limb is absent) or partial (when the greater portion of the distal half is absent). A third category is paraxial hemimelia. The word *paraxial*, meaning beside the axis, has been used in limb deficiencies [13] to indicate that either the preaxial or the postaxial portion of the distal half of the limb is involved. The anatomical term *preaxial*, it may be recalled, refers to the border of a limb on which either the thumb or the big toe is situated, and the term postaxial refers to the opposite border; these two terms are based on the arrangement of the limbs in the embryo at five and one-half postovulatory weeks, when the thumbs and the big toes are both on the cephalic borders of the limbs. Paraxial hemimelia may be radial or tibial (both of which are preaxial) or ulnar or fibular (both of which are postaxial). It is important to note that, by convention, the various subtypes of paraxial hemimelia are named after the absent portion. Thus, radial hemimelia refers to a deficiency of the radial portion of the forearm or of the forearm and hand.

A plan of the proposed classification is presented in Table I. The principles on which this classification is based are as follows. The defects being considered are either *terminal* (T), where there are no unaffected parts distal to, and in line with, the deficient portion, or *intercalary* (I), where the middle portion of a proximodistal series of limb components is deficient but the proximal and distal portions are present. Each of these two main groups may be either *transverse* (denoted by a hyphen), where the defect extends transversely across the entire width of the limb, or *longitudinal* (denoted by a vertical line /), where only the preaxial or postaxial portion is absent and hence the deficiency is longitudinal.

FIG. 1.



Numerals (see Table I) identify subgroups within each of these four categories. A horizontal line (representing the trunk) below a symbol denotes upper-limb involvement; a line above a symbol denotes lower-limb involvement.

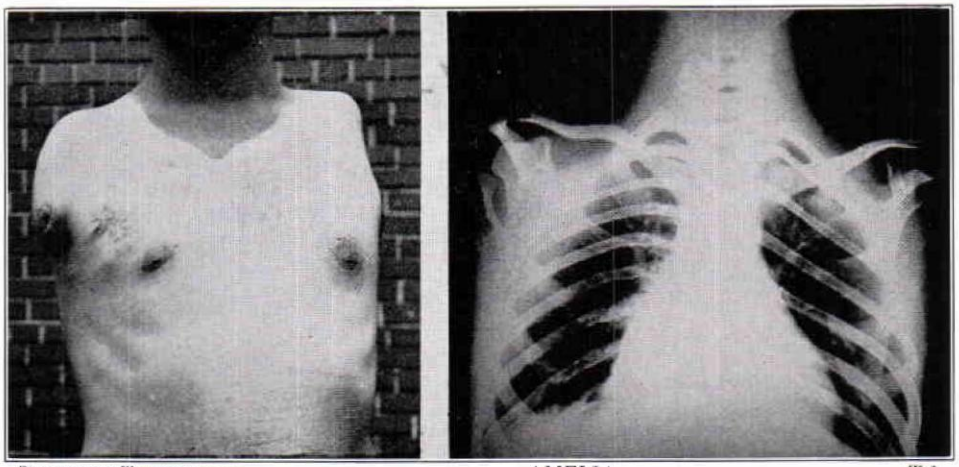
In paraxial hemimelia, the four subtypes (radial, ulnar, tibial, and fibular) are designated by abbreviations. When a capital letter (R, U, TI, FI) are used, the condition is complete, that is, the defective element is completely absent on roentgenographic examination. When lower-case letters (r, u, ti, fi) are used, the condition is incomplete, that is, a portion of the defective element (for example, the radius) is present.

Additional numerals, placed after a colon, denote the digital ray or rays involved. The term *ray* is a convenient designation for a digit, including its metacarpal (or metatarsal) and in some cases, the associated carpals (or tarsals) as well. The thumb, for example, is associated anatomically and developmentally with the trapezium and the navicular. In the case of most of the other digits, however, the relationship to the carpals (or tarsals) is not as clear-cut. It should be stressed that the term *ray* is used here in a purely descriptive sense, without any phylogenetic, or even ontogenetic, implications.

An endeavor has been made to use the internationally accepted *Nomina Anatomica* of 1955 throughout. It may be pointed out that the word *limb* now replaces the pedantic and inaccurately used *extremity* (the upper extremity of the body, as a momentary reflection on the anatomical position will show, is the top of the head).

Use of this classification will allow orthopaedists to designate the various deficiencies accurately. With it we can all speak the same language.

To avoid confusion with deficiencies involving the hand, it is well to keep in mind the *entire* limb: "Think of the arm—not the hand."



TERMINAL TRANSVERSE AMELIA T-1

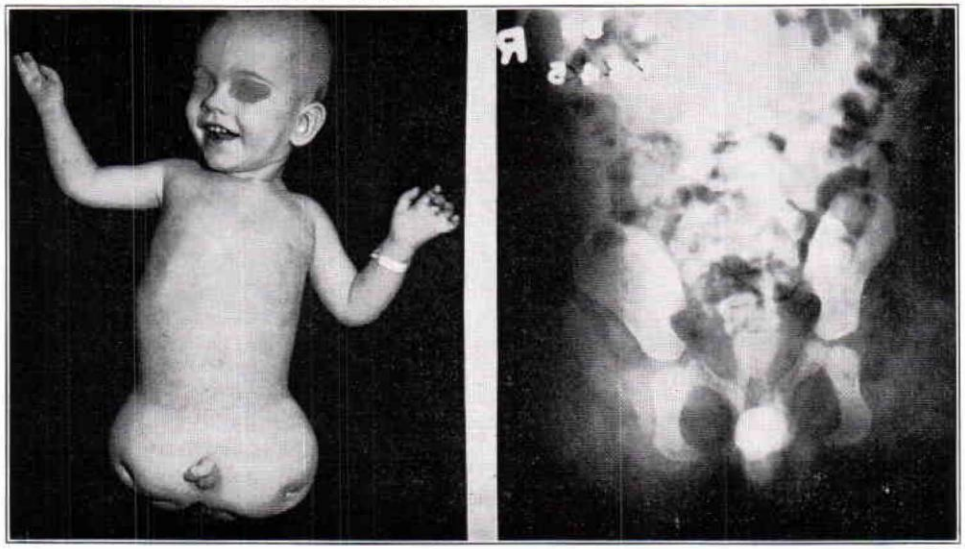
Incidence: Area Child Amputee Center: thirteen cases (nine male, four female) in 300. Seven (54 per cent) bilateral. Birch-Jensen [4] reports 1 in 270,000 births.

Genetics: sporadic abnormality, non-hereditary.

Associated defects: club foot; hare lip; cleft palate; scoliosis; complete phocomelia; distal phocomelia; lower amelia; complete paraxial hemimelia, radial

Note: Area Child Amputee Center has one case of quadrimembral amelia.

Complete absence of the upper limb. There may be a small lobule of fat or dimple on the lateral thoracic wall. Clavicle is present. Shoulder girdle is usually very mobile. There may be accentuation of the shoulder girdle contour from excessive fat. Some cases will present a prominent acromioclavicular joint with little or no subcutaneous fat. Thoracic scoliosis often accompanies this anomaly, especially in bilateral cases.[3, 4, 6]



TERMINAL TRANSVERSE AMELIA T-1

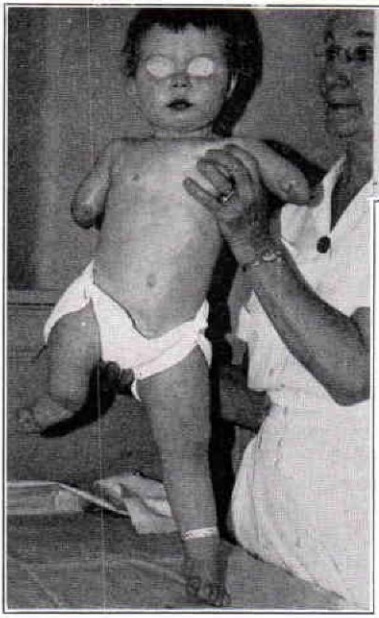
Incidence: Area Child Amputee Center: seven cases in 300. Predominantly male, 5:2. Three (43 per cent) bilateral.

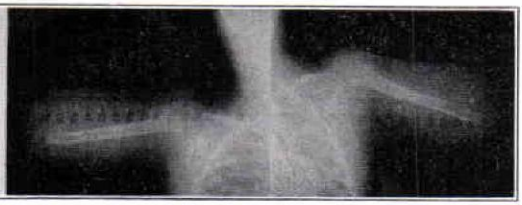
Genetics: sporadic; non-hereditary.

Associated defects: proximal femoral focal deficiency; upper amelia; hemimelia; partial adactylia; complete paraxial hemimelia, ulnar.

Note: Area Child Amputee Center has one case of quadrimembral amelia.

Complete absence of the lower limb. There may be a lobule of fat or there may be a depressed area (pit) on the lateral aspect of the pelvis. The pelvic contour is wide due to accumulation of fat over hip bones.[4, 6]





HEMIMELIA T-2

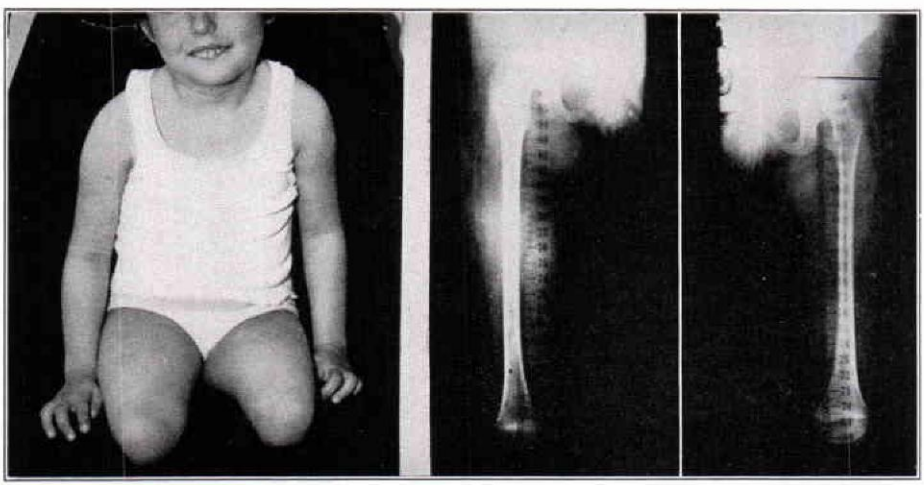
Incidence: Area Child Amputee Center: twenty cases (eleven male, nine female) in 300. Three cases bilateral. There is slight predominance of left unilateral deformities. Birch-Jensen [4] reports nine cases in population of 4,024,000.

Genetics: sporadic; non-hereditary (limb-bud arrest).

Associated defects: amelia; lower hemimelia; partial hemimelia; partial adactylia; complete paraxial hemimelia, fibular or ulnar; proximal femoral focal deficiency; hare lip; cleft palate; heart defect; agnathia.

TERMINAL TRANSVERSE

Absence of forearm and hand—anatomically, an elbow disarticulation with the distal epiphyseal plate of the humerus present. Growth may be deficient in preadolescent age due to the altered functional demand on the humerus. Distal stump end is smooth or dimpled or it presents a small lobule of fat in front of the limb axis. [3, 4, 7, 13]



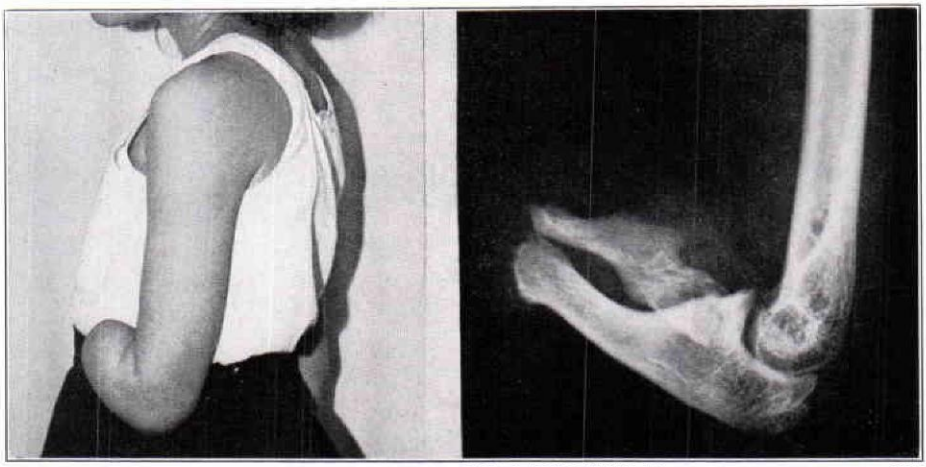
TERMINAL TRANSVERSE HEMIMELIA T-2

Incidence: Area Child Amputee Center: twelve cases (seven male, five female) in 300. Five (41 per cent) bilateral.

Genetics: sporadic.

Associated defects: upper hemimelia; partial hemimelia; acheiria or apodia; partial adactylia; complete aphalangia; amelia.

Absence of leg and foot—anatomically a knee disarticulation with the distal femoral epiphysis present. Stumps are conical and capable of end-bearing. There is usually coxa valga. Distally, there may be fat lobules behind the limb axis (popliteal area). In some cases the patella is present and voluntarily movable. The distal femoral epiphysis is apparently more "active" during growth than is the upper limb counterpart, the distal humeral epiphysis.[1, 3, 7, 13]



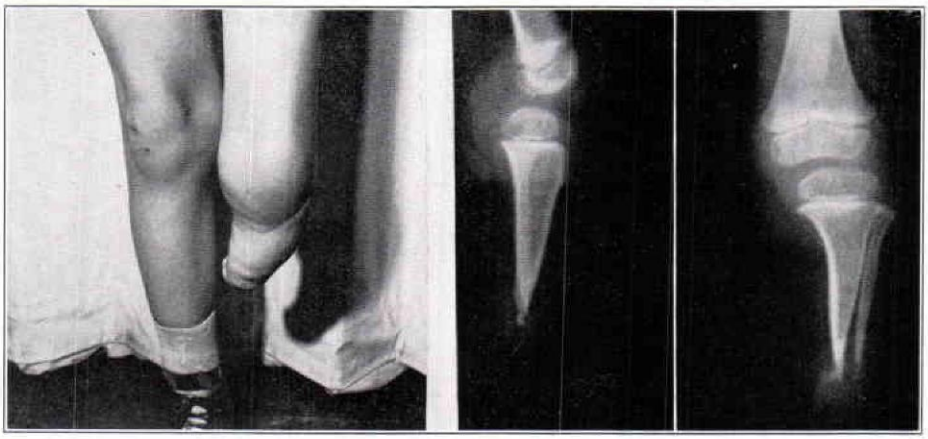
TERMINAL TRANSVERSE PARTIAL HEMIMELIA T·3

Incidence: Area Child Amputee Center: 112 cases in 300. Predominantly female and left-sided; only three bilateral. Birch-Jensen [4] reports 161 cases in population of 4,024,000; female, 3:2; unilateral left, 2:1.

Genetics: sporadic; non-hereditary.

Associated defects: unusual—only ten cases in 112. Syndactylism; partial adactylia; hemimelia; acheiria or apodia; partial aphalangia; club foot; heart defect; hip dysgenesis; agnathia.

Absence of a part of the distal portion of an upper limb. Forearm segment is very short; bone-stump length is less than eleven centimeters. Ulna, olecranon, and trochlea are well developed. Radius is likely to be slightly longer than ulna. Biceps tuberosity may be large and simulate radial bowing. Radial-head development is variable; it may articulate with capitulum or grow latero-proximally beyond capitulum. Skin on stump end may demonstrate small transverse ridge, depressed lobules, or ridge with five or fewer nubbins, some of which may have small finger nails. Usually a strong biceps with excellent elbow flexion power is present. Laterally the elbow is stable but hyperextensible.[2, 3, 4, 7, 12, 13]



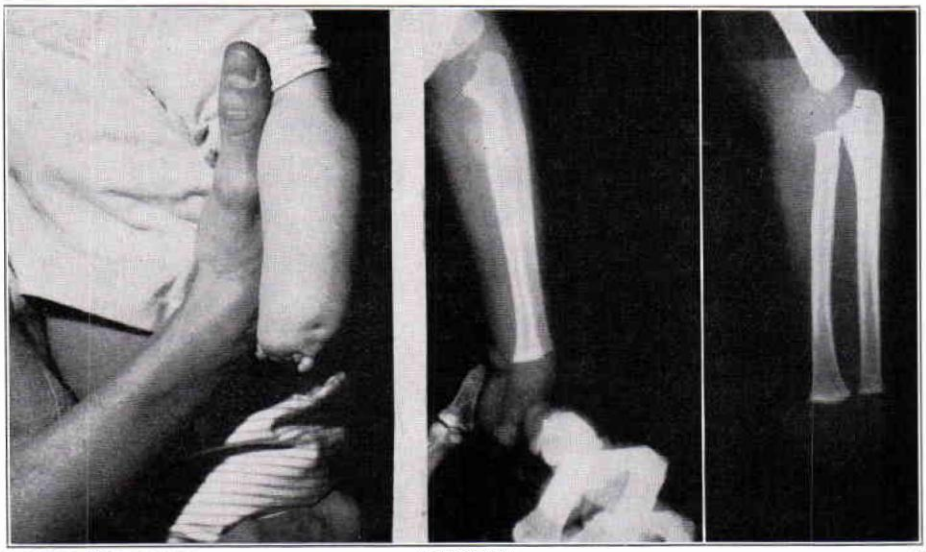
TERMINAL TRANSVERSE PARTIAL HEMIMELIA T·3

Incidence: Area Child Amputee Center: twelve cases in 300. Predominantly male, 9:3; one bilateral.

Genetics: apparently non-hereditary; sporadic.

Associated defects: hemimelia; partial aphalangia or adactylia; complete paraxial hemimelia, fibular; agnathia.

Absence of a part of the distal portion of a lower limb—anatomically a below-the-knee stump. Proximal tibial epiphysis is present with normal contour. Distal end of tibia may form a spike. There is no distal tibial epiphysis. Contour of stump may be symmetrical or deviated slightly in varus. Circular skin depressions are sometimes seen distally (ectodermal deficiency). Fibula may be absent (see T/1 FI).[3, 7, 13]



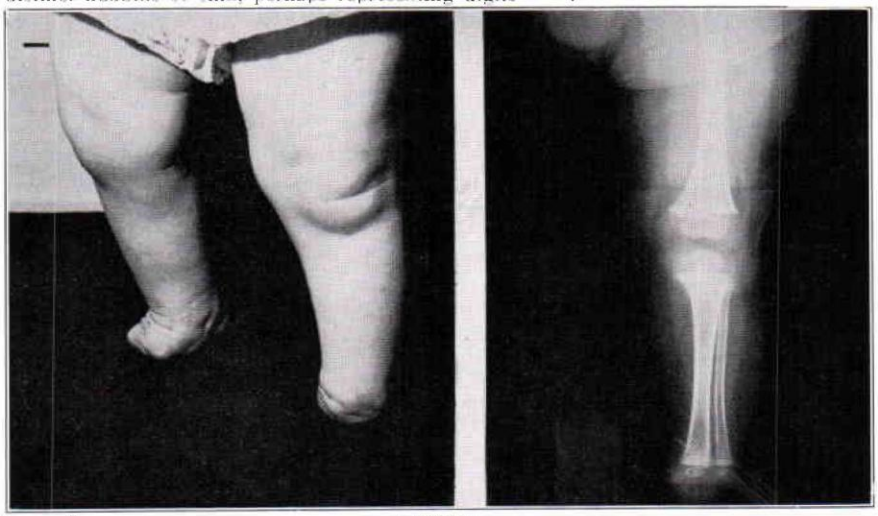
TERMINAL TRANSVERSE ACHEIRIA T-4

Incidence: Area Child Amputee Center: twenty-four (ten males, fourteen females) in 300 cases. Three bilateral. Predominantly left unilateral (62.5 per cent). Birch-Jensen [4] reports fifty-four cases in population of 4,024,000; 60 per cent left unilateral.

Genetics: great majority sporadic; possibly a recessive lesion.

Associated defects: apodia; complete aphalangia; hemimelia; partial hemimelia; radiohumeral synostosis; metatarsus varus; agnathia; hare lip; cleft palate; complete paraxial hemimelia, radial.

Absence of the hand—anatomically a wrist disarticulation with the distal epiphyses of the radius and ulna present. Pronation and supination are usually present; occasionally there is a distal radio-ulnar cartilaginous bar. Skin on stump end may demonstrate depressed transverse dimple or less distinct nubbins of skin, perhaps representing digits [5, 4, 13].



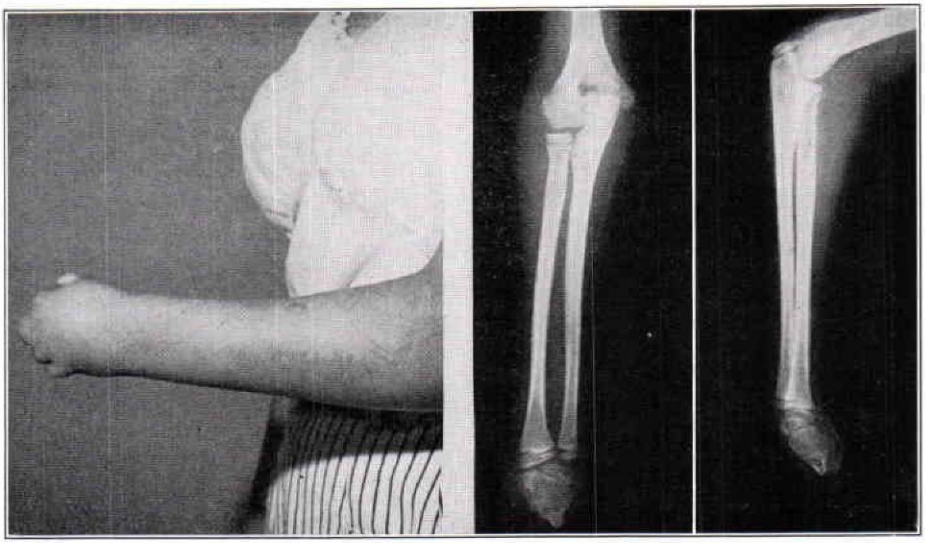
TERMINAL TRANSVERSE APODIA T-4

Incidence: Area Child Amputee Center: six cases in 300. Equal sex distribution.

Genetics: apparently sporadic; non-hereditary.

Associated defects: acheiria; partial hemimelia; hemimelia; complete adactylia; complete paraxial hemimelia; fibular; hare lip; cleft palate.

Absence of foot—anatomically an ankle disarticulation with the distal epiphyses of tibia and fibula present. Stump is fully end-bearing. Skin on plantar surface is cornified as on the normal heel. There is an anterior skin crease on the stump end. Some degree of abnormal tibial torsion may be present [5, 7, 14].



TERMINAL TRANSVERSE COMPLETE ADACTYLIA T-5

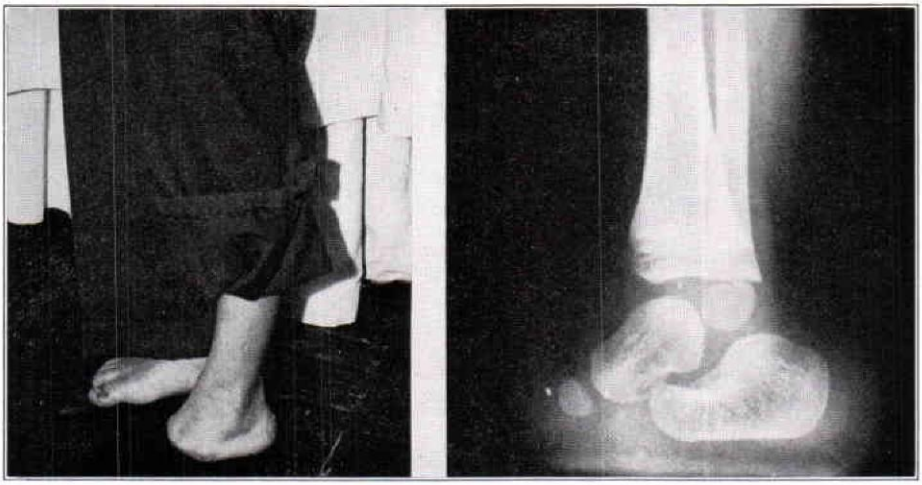
Incidence: Area Child Amputee Center: seventeen cases in 300. Predominantly male, 12:5, no bilateral.

Genetics: great majority sporadic; possibly an inherited recessive lesion.

Associated defects: rare. Complete paraxial hemimelia, fibular.

Synonym: mitten-hand.

Absence of the digits including absence of the metacarpals and phalanges. The carpals may vary in number and configuration; they may be fused *en masse*. Flexion at the wrist is possible. The hand segment may be an oval flipper or demonstrate rudimentary representations of digits which appear as a ridge or as individual small digits.[3, 4, 13]



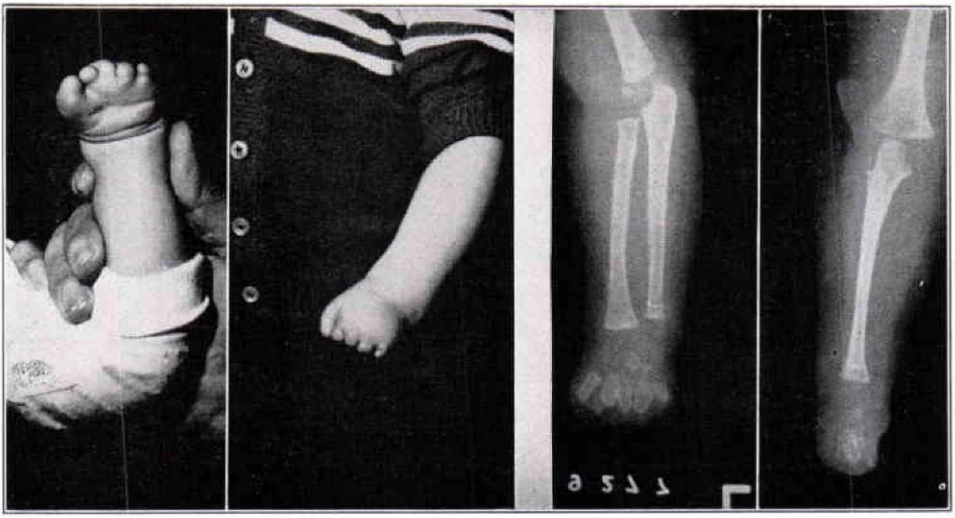
TERMINAL TRANSVERSE COMPLETE ADACTYLIA T-5

Incidence: Area Child Amputee Center: four cases in 300. Predominantly male, 3:1.

Genetics: possibly an inherited recessive lesion.

Associated defects: acheiria; hare lip; cleft palate.

Absence of the metatarsals and phalanges. This may be likened to the Lisfranc amputation. Tibialis anterior is present, preventing progressive equinus of the hind part of the foot. Anteriorly, there may be a raised ridge of skin on the stump end of varying size and shape, sometimes including recognizable digits. Patient has some atrophy of triceps surae and has tendency to knee hyperextension[3].



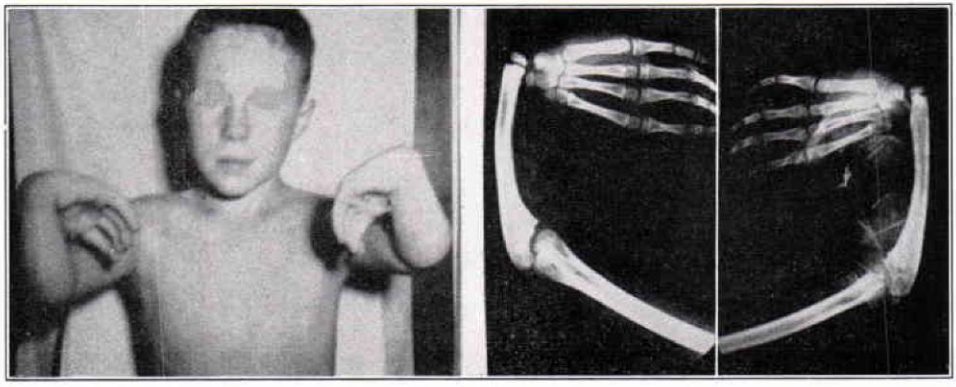
TERMINAL TRANSVERSE — COMPLETE APHALANGIA — T-6

Incidence: Area Child Amputee Center: eight cases in 300. Predominantly male, 5:3. Equally divided right and left unilateral; no bilateral cases. Area Child Amputee Center reports no lower-limb counterparts in this series of patients.

Genetics: probably sporadic; non-hereditary—may be recessive.

Associated defects: radio-ulnar synostosis.

Absence of the phalanges. Metacarpals are present, but they may be attenuated. Hand appears relatively broad due to deficient phalanges. Rudimentary digits represented by skin nubbins with or without rudimentary finger nails are present[3, 4, 7].



TERMINAL LONGITUDINAL — COMPLETE PARAXIAL HEMIMELIA: RADIAL — T/1 R

Incidence: Area Child Amputee Center: six cases in 300. Predominantly male, 4:2. Three (50 percent) bilateral. Birch-Jensen [4] reports seventy-three cases in population of 4,024,000, with associated defects in 40 per cent of the cases. O'Rahilly found numerous cases reported by nine authors through 1951[13].

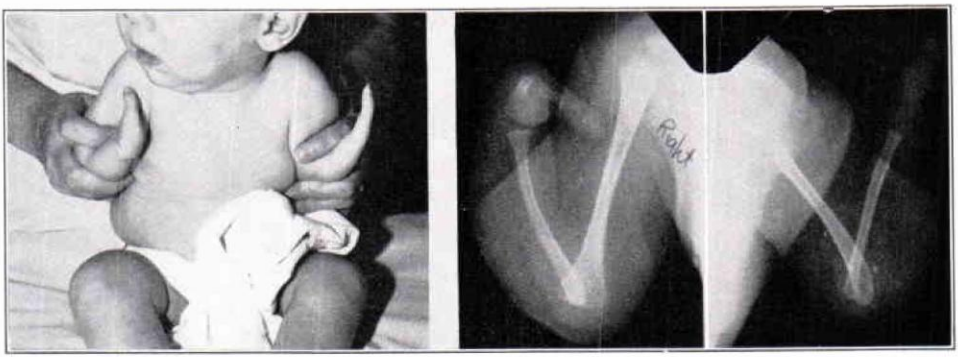
Genetics: usually sporadic; sometimes hereditary—dominant or recessive.

Associated defects: acheiria; partial aphalangia; complete paraxial hemimelia; radial or tibial; proximal femoral focal deficiency; amelia; radiohumeral synostosis.

Synonyms: radial defect; arrest of development; absence of radius; aplasia of radius; hypoplasia of radius; clubbed hand.

Note: This lesion is more prevalent than the Center's statistics reflect; this anomaly is not seen in amputee clinics, but rather in general orthopaedic clinics.

Absence of the radius and radial ray in the hand. There is a short, strong ulna, which may be bowed. The hand is radially deviated. The thumb (first ray), scaphoid, and trapezium are absent [4, 6, 13].



TERMINAL LONGITUDINAL COMPLETE PARAXIAL HEMIMELIA: ULNAR T/1 U

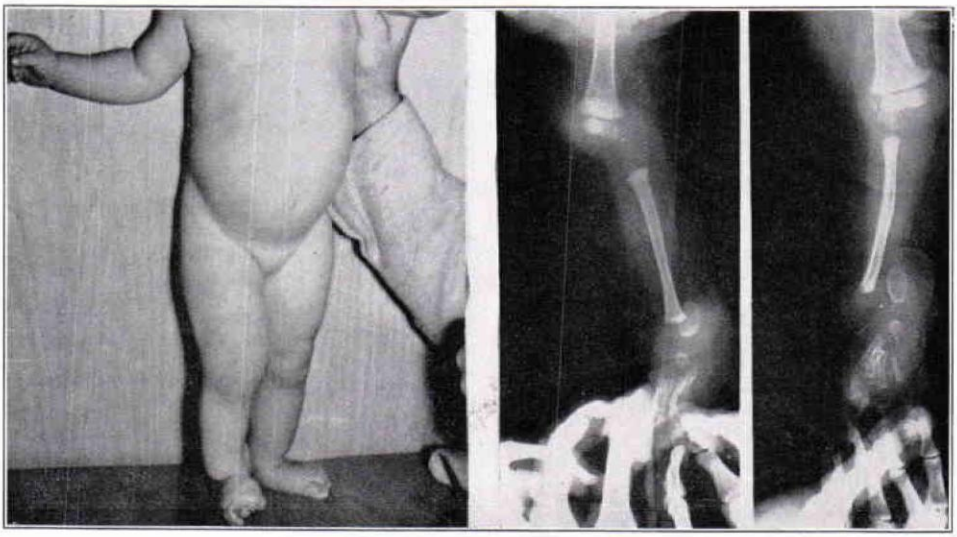
Incidence: Area Child Amputee Center: six cases in 300. Predominantly male, 4:2. One bilateral. Birch-Jensen[4] reports nineteen cases in population of 4,024,000, most often unilateral. O'Rahilly found numerous cases reported by five authors through 1932.

Genetics: apparently not inherited; may appear in families who have an accumulation of joint lesions.

Associated defects: hemimelia: partial adactylia; complete paraxial hemimelia, ulnar (Intercalary Longitudinal).

Synonyms: monodigital arm; monodigital hand; absence of the ulna.

Absence of the ulna. The radius usually articulates with the capitulum. The distal radial epiphysis is present. One digit is present which is unstable in the metacarpophalangeal joint but has flexion power. The pisiform is always present. In the majority of cases, the hamate, triquetrum, and capitate are absent. The forerm cannot be extended beyond 90 degrees of elbow flexion. Further flexion of the elbow is complete but with deficient power. [3, 4, 13, 14]



TERMINAL LONGITUDINAL COMPLETE PARAXIAL HEMIMELIA: TIBIAL T/1 TI

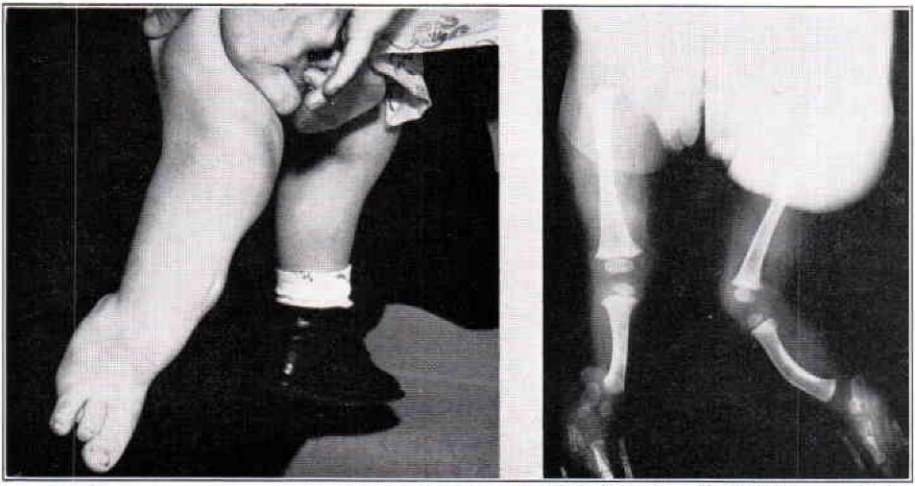
Incidence: Area Child Amputee Center: three cases (two males, one female) in 300. All bilateral.

Genetics: ? sporadic.

Associated defects: partial aphalangia.

Note: the patient illustrated has right-sided tibial hemimelia; the plantar surface of the foot is in inversion.

Complete absence of tibia. The below-the-knee segment of the limb is short. There is a flexion deformity of the knee. The foot is in varus and rotated to place the plantar surface medially. The fibula may be luxated dorsally and proximally toward the popliteal fossa. The calcaneus and talus may be fused to each other and sometimes to the navicular as well. The great toe may be absent or rudimentary. Metatarsals 1, 2, and 3 may be absent. Note that the patient illustrated has no bone elements for the first ray[1, 3, 13, 14].



TERMINAL LONGITUDINAL COMPLETE PARAXIAL HEMIMELIA: FIBULAR T/1 FI

Incidence: Area Child Amputee Center: twenty-six cases (nineteen males, seven females) in 300. Twelve (46 per cent) right-sided unilateral. Coventry and Johnson[5] report twenty-nine cases. Thompson and associates[17] report thirty-one cases. Kruger and Talbott[11] report sixty-one complete fibular absences in forty-seven patients.

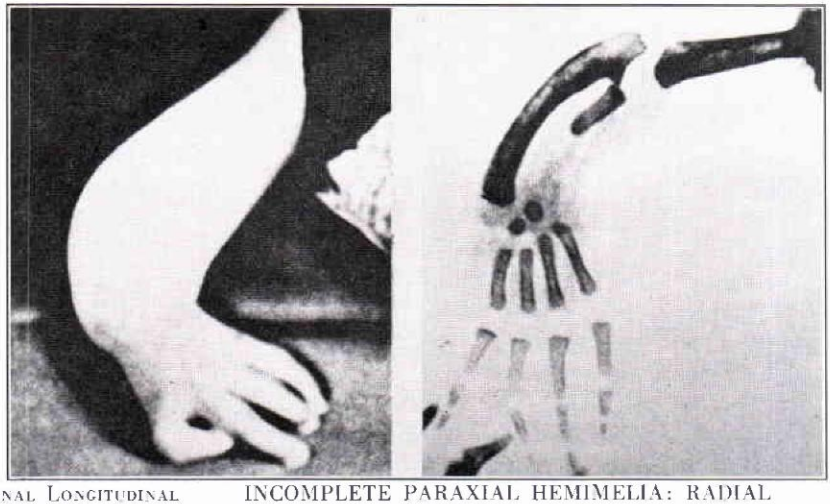
Genetics: recessive, possibly sporadic.

Associated defects: proximal femoral focal deficiency; partial adactylia; complete adactylia; hemimelia; apodia; incomplete paraxial hemimelia, radial; complete paraxial hemimelia, ulnar; partial hemimelia.

Synonyms: kyphosis of tibia; absence of fibula.

Note: proximal femoral focal deficiency, a major lesion, is often accompanied by this abnormality (Amputee Center statistics show 67 per cent).

Complete absence of fibula. The below-the-knee segment is short. Anterior bowing of tibia is present with a skin dimple at the apex of the tibial bow. The distal tibial epiphyseal plate is deficient. There is an equinovalgus deformity of the foot with one or two lateral toe rays absent. The calcaneus may be posterior and above the distal tibial epiphysis. Tarsal fusion may occur. Talus and cuboid may be absent. A fibrous band representing the fibula may be encountered at operation[1, 3, 5, 9, 11, 13].



TERMINAL LONGITUDINAL INCOMPLETE PARAXIAL HEMIMELIA: RADIAL T/2 r

From Birch-Jensen[4]: *Congenital Deformities of the Upper Extremities.*

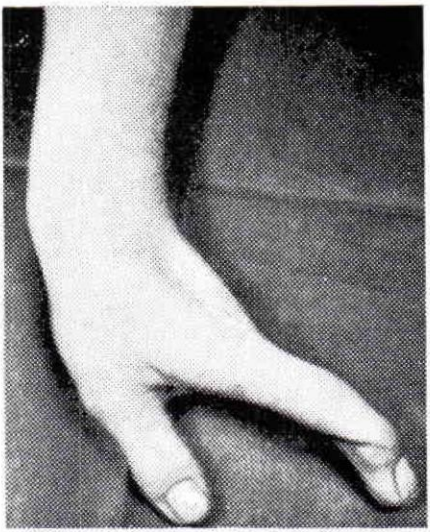
Incidence: Area Child Amputee Center reports two cases (both male, bilateral) in 300. Birch-Jensen [4] reports twenty-four cases, predominantly male, with incidence of one in 55,000.

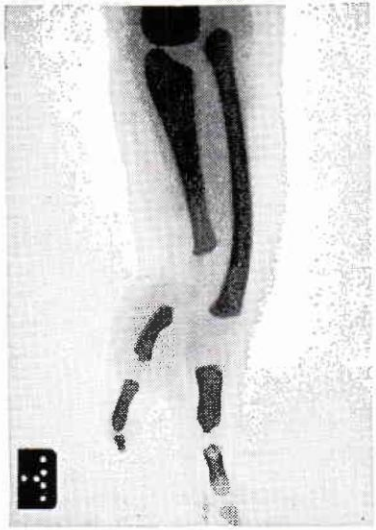
Genetics: very small hereditary incidence; great majority sporadic.

Associated defects: many and varied. Most of Birch-Jensen's cases with accompanying defects were concentrated in a small group of patients. Sixty per cent of his patients with this deficiency had no accompanying anomalies. Aplastic anemia may be encountered with this defect.

(Legend continued from previous page)

This resembles paraxial hemimelia, radial (complete), T/1 R, except that a portion of the radius exists, usually proximally. Note that in this deficiency the thumb (first ray) is absent. Clinically this deficiency may be similar to the complete condition (T/1 R), but roentgenographic examination differentiates them, revealing presence or absence of a part of the radius[3, 4, 13, 14].





TERMINAL LONGITUDINAL INCOMPLETE PARAXIAL HEMIMELIA: ULNAR T/2 u

From Birch-Jensen[4]: *Congenital Deformities of the Upper Extremities.*

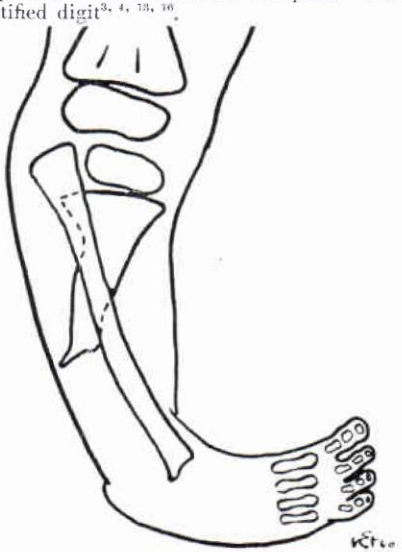
Incidence: Area Child Amputee Center reports no cases. Birch-Jensen[4] reports fourteen cases (eight males and six females), one per 100,000 births and one per 200,000 of population.

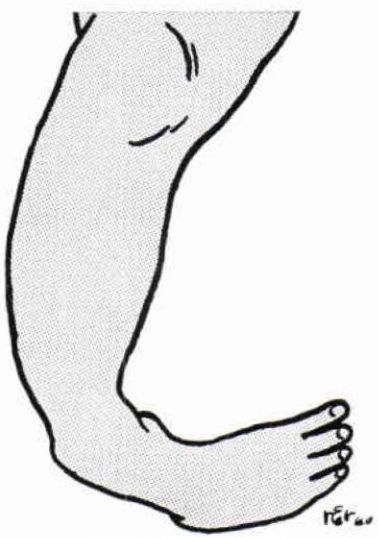
Genetics: insufficient statistics. Most often unilateral. Does not seem to be inherited.

Associated defects: there may be associated defects. Adactylia, partial (split-hand) or lower limb defects, or both, may accompany this deficiency.

Synonyms: hypoplasia of ulna; congenital ulnar defect.

This resembles complete paraxial hemimelia, ulnar (Terminal Longitudinal, T/1 U), except that a portion of the ulna exists. The patient illustrated has two digits, the thumb (first ray) and an unidentified digit[3, 4, 13, 16].

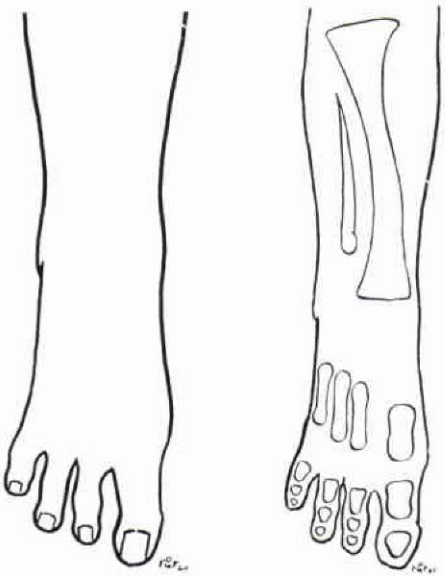




TERMINAL LONGITUDINAL INCOMPLETE PARAXIAL HEMIMELIA: TIBIAL T/2 ti

Incidence: no clear-cut cases reported in the literature.

This deficiency resembles complete paraxial hemimelia (Terminal Longitudinal, T/1 TI) except that a portion of the tibia is present. The foot is inverted and the great toe (first ray) is absent[1, 3, 13, 14, 16].



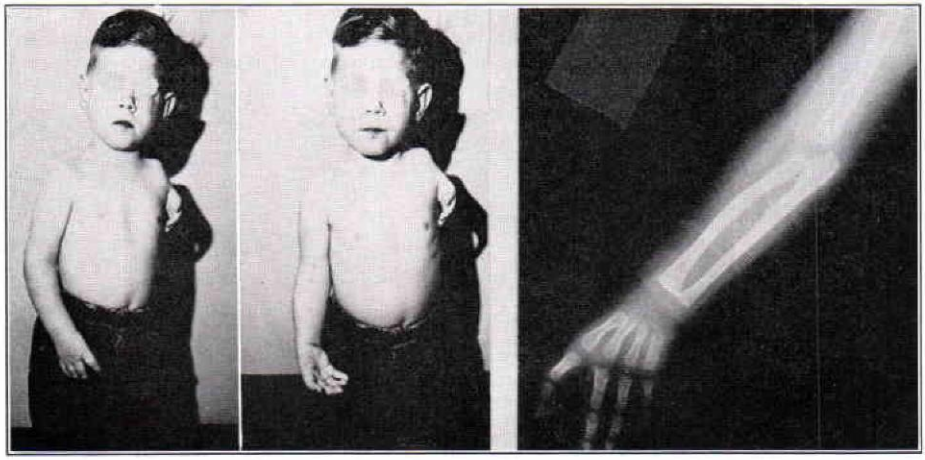
TERMINAL LONGITUDINAL INCOMPLETE PARAXIAL HEMIMELIA: FIBULAR T/2 fi

Incidence: no clear-cut cases reported in the literature.

Genetics: probably parallels paraxial hemimelia, fibular (complete, Terminal Longitudinal, T/1 Fl), which is recessive and possibly sporadic.

Synonyms: incomplete ossification of fibula; kyphosis of tibia.

Clinically this resembles complete paraxial hemimelia, fibular (T/1 Fl). Roentgenographic examination differentiates the two conditions, revealing a portion of the fibula to be present or absent. Note that there are four digits; the fifth is absent. If all toes were present this would be incomplete paraxial hemimelia, fibular (Intercalary Longitudinal, 1/2 fi) [1, 3, 5, 9, 11, 13]



TERMINAL LONGITUDINAL PARTIAL ADACTYLIA T/3

Incidence: Upper Limb: Area Child Amputee Center: seventeen cases (involving divers rays) in 300. Predominantly male 11:6. Usually unilateral. Birch-Jensen[4] reports eighty-five cases.

Lower Limb: Area Child Amputee Center: six cases (four males, two females) in 300.[3, 4, 13, 15]

Genetics: generally sporadic; a few cases may be recessive (inherited).

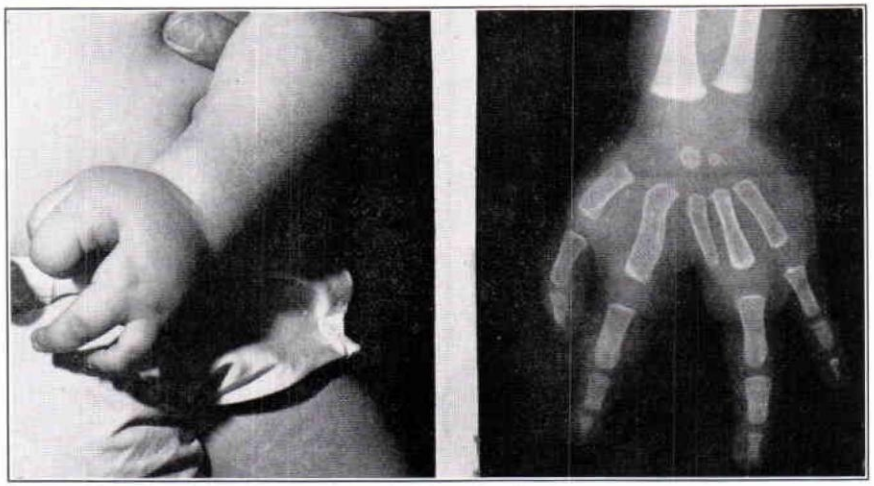
Synonym: synbrachydactylism.

Associated defects: proximal femoral focal deficiency; amelia; hemimelia; complete phocomelia; distal phocomelia; complete paraxial hemimelia, fibular or ulnar; partial hemimelia; hare lip; cleft palate.

(Legend continued from previous page)

Note: patient illustrated presents T/3:1. The left upper limb is intercalary transverse complete phocomelia (I-1).

Absence of one to four digits in the hand or foot along with their metacarpals or metatarsals.



TERMINAL LONGITUDINAL PARTIAL APHALANGIA T/4

Incidence: Upper Limb: Area Child Amputee Center: ten cases (six males, four females) in 300; divers rays involved. Usually bilateral. Birch-Jensen[4] reports fifty-six cases (involving the third ray) in population of 4,024,000.

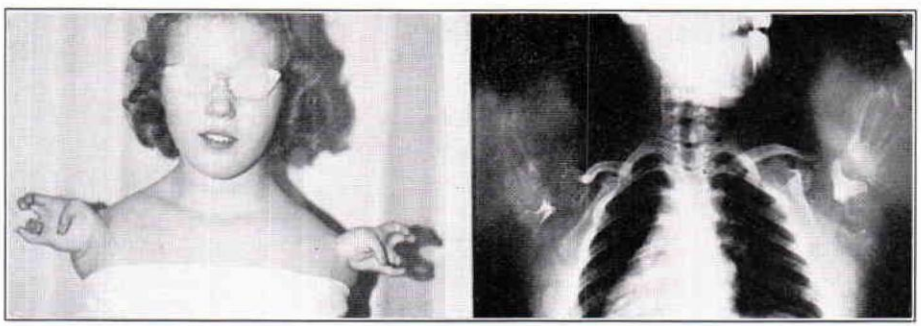
Lower Limb: Area Child Amputee Center: three cases (all female; two bilateral) in 300, involving divers rays.[4, 13, 15]

Genetics: most cases apparently spontaneous. When inherited, condition is *dominant.*

Associated defects: proximal femoral focal deficiency; paraxial hemimelia, fibular or radial; partial or complete aphalangia; partial hemimelia; apodia; dysplastic hip.

Synonyms: ectrodactylism; adactylia; oligodactyly; perodactyly; monodactyly; hypodactyly; pincers; claw; crayfish claw; crab claw; lobster claw.

Absence of one or more phalanges from one to four digits. In the hand the rays may deviate and form a cone-shaped cleft, thus dividing the hand into two parts; there may be syndactylism of fingers on either side of cleft.



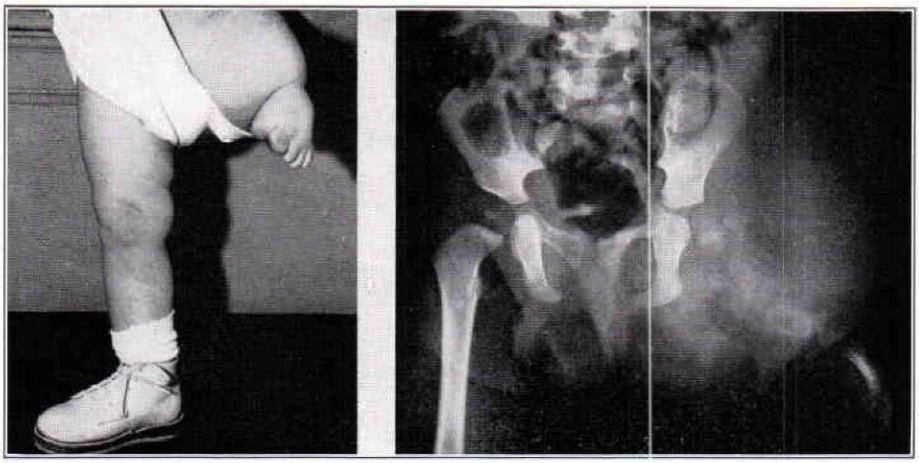
INTERCALARY TRANSVERSE COMPLETE PHOCOMELIA I-1

Incidence: Area Child Amputee Center: six cases in 300. Equal sex distribution. Two bilateral. Birch-Jensen[4] reports five cases (not regionally classified as to complete, proximal, or distal) in population of 4,024,000.

Genetics: insufficient statistics.

Associated defects: amelia; incomplete adactylia; distal phocomelia; arthrogryposis. Note: skeletal deficiencies usually are not symmetrical when bilateral.

Absence of the proximal portions of the upper limb. The hand is attached to the trunk and rests in the coronal plane. There may be deficient rays of the index to little finger inclusive. Digits are in flexion. Flexion power may be sufficient to be functional. Extension power of digits is very poor. Syndactylism may be present; by roentgenographic examination small, undifferentiated bone segments may be recognized[3, 4, 6, 7, 13].



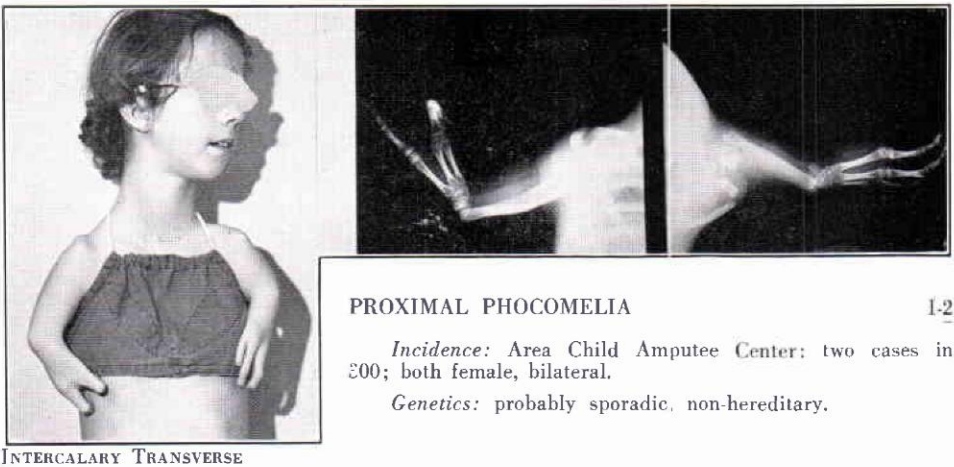
INTERCALARY TRANSVERSE — COMPLETE PHOCOMELIA — I-1

Incidence: Area Child Amputee Center: five cases in 300. All male; one bilateral.

Genetics: insufficient statistics.

Associated defects: partial adactylia; hip dysplasia; proximal femoral focal deficiency; complete paraxial hemimelia, fibular.

Absence of the proximal portions of the lower limb. Foot is attached to trunk wall. Toe rays may be absent. Flexion of the toes is possible but not extension. Roentgenographic examination shows that tarsals may be absent or present in varying degrees. Acetabulum may be nonexistent; its roof index may measure 15 degrees[3, 7, 13].



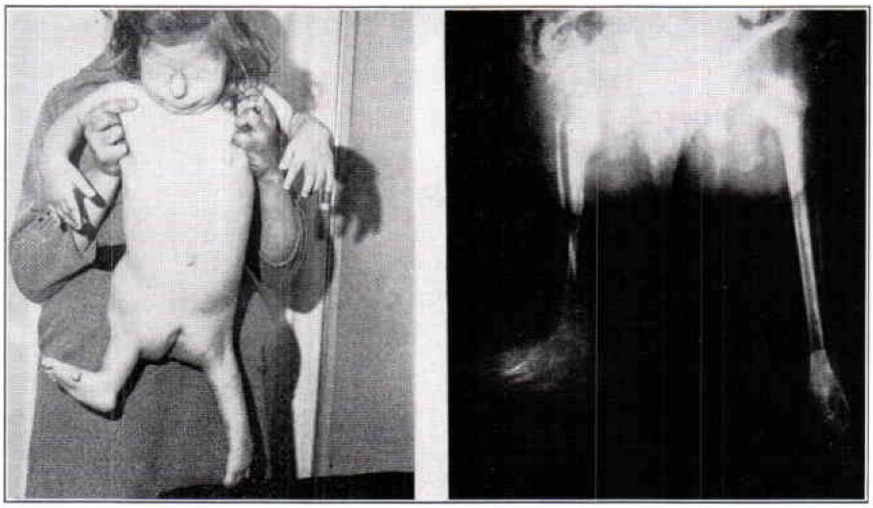
PROXIMAL PHOCOMELIA — I-2

Incidence: Area Child Amputee Center: two cases in 300; both female, bilateral.

Genetics: probably sporadic, non-hereditary.

INTERCALARY TRANSVERSE

Absence of the humerus (the proximal segment of the arm). The patient illustrated demonstrates absence of the radius. Roentgenographically, the forearm here has an ulna. Shoulder girdles are atrophic. All joints are unstable. Hands flex on the arm with fair power, but are deficient in extension. Intrinsic muscle function in the fingers is deficient; they flex but lack extension. Finger rays vary in number.[3, 4, 6, 7, 13]



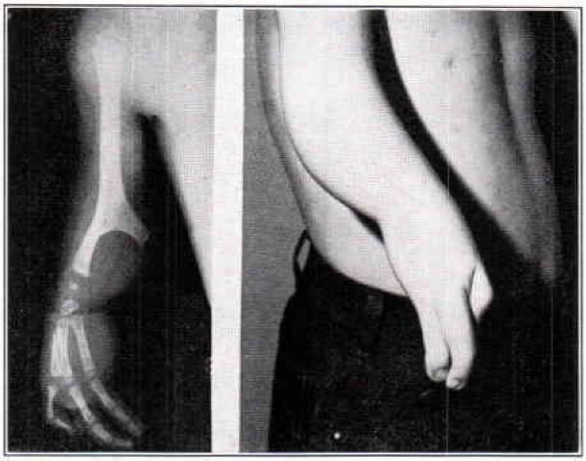
INTERCALARY TRANSVERSE PROXIMAL PHOCOMELIA I-$\bar{2}$

Incidence: Area Child Amputee Center: two cases in 300. Both bilateral.

Genetics: probably sporadic, non-hereditary.

Associated defects: radiohumeral synostosis; cleft palate; complete paraxial hemimelia, fibular or ulnar.

Absence of the femur. The proximal ends of the tibia and fibula rest close to the external surface of the ilium. There is no acetabulum. The junction of the tibia and ilium is very unstable and incapable of weight-bearing. Ligaments if they exist are relaxed. The leg can be rotated much like the hands of a clock[3, 10, 13].



INTERCALARY TRANSVERSE DISTAL PHOCOMELIA I-$\bar{3}$

Incidence: Area Child Amputee Center: ten cases in 300. Predominantly male, 7:3. Three bilateral.

Genetics: sporadic; non-hereditary—may be recessive.

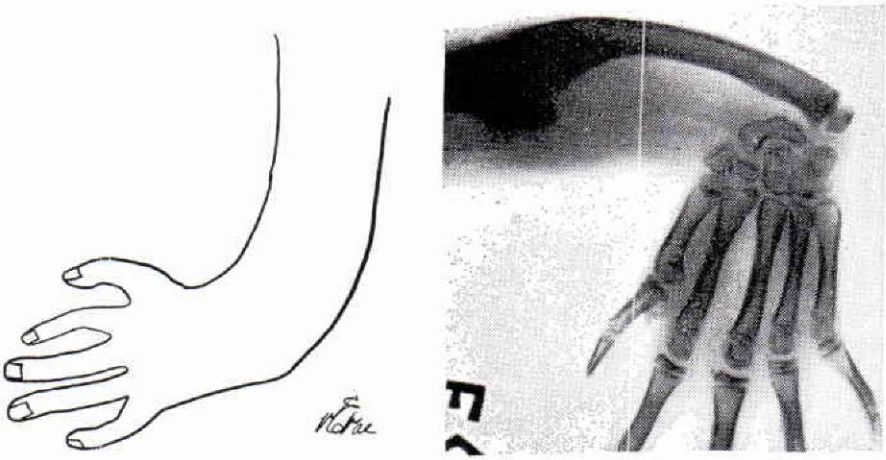
Associated defects: partial adactylia; amelia.

Absence of radius and ulna. The arm is short. Finger rays vary in number. There is good flexion power of the wrist and fingers. Carpohumeral joint is quite stable. Shoulder abduction is limited. Roentgenographic examination usually reveals that the distal end of the humerus is forked. This may represent distorted condylar development.

Lower Limb Counterpart (I-$\bar{3}$):

Incidence: Area Child Amputee Center: one case, male, bilateral.

Associated defects: complete phocomelia; amelia[4, 7, 13].

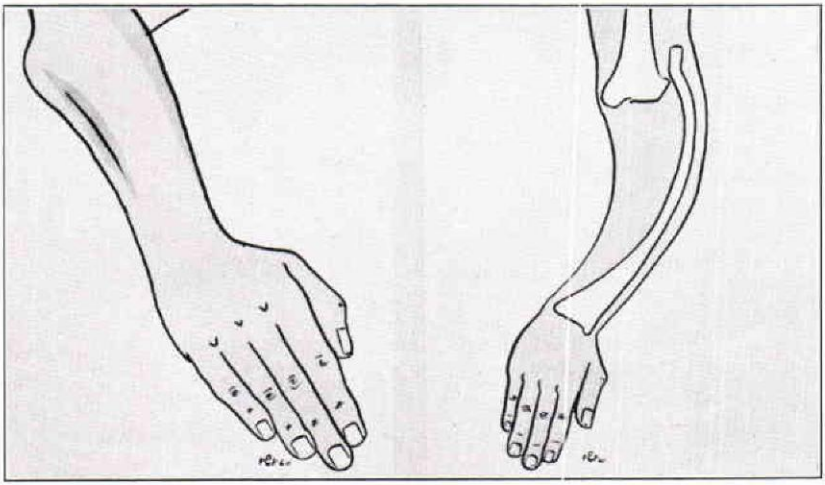


INTERCALARY LONGITUDINAL COMPLETE PARAXIAL HEMIMELIA: RADIAL I/1R

From Birch-Jensen[4]: *Congenital Deformities of the Upper Extremities.*

Incidence: Area Child Amputee Center reports one case in 300. Female, unilateral. Birch-Jensen[4] includes this deficiency in his presentation of radial defects and aplasias.

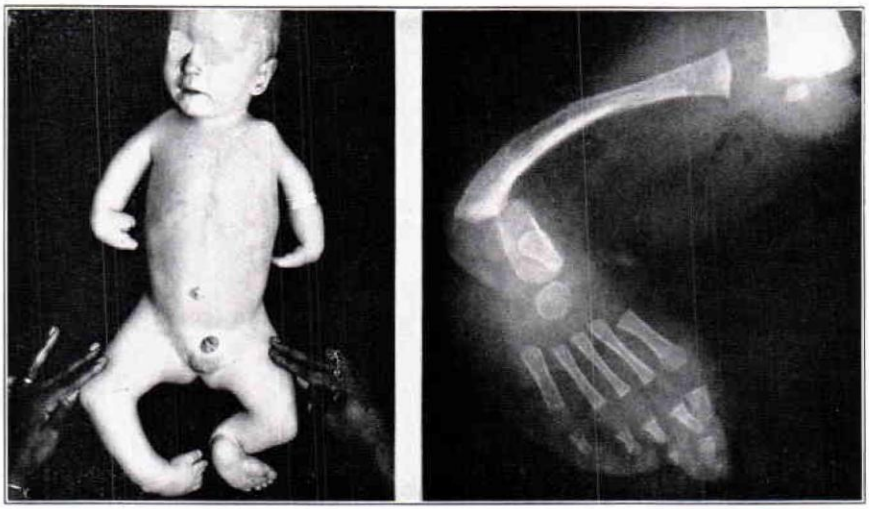
This deficiency resembles complete paraxial hemimelia, radial (Terminal Longitudinal, T/1 R), but demonstrates a complete hand with five digits[4,6,13].



INTERCALARY LONGITUDINAL COMPLETE PARAXIAL HEMIMELIA: ULNAR I/1 U

Incidence: Area Child Amputee Center: one case (male, unilateral) in 300.

This deficiency resembles complete paraxial hemimelia, ulnar (Terminal Longitudinal, T/1 U), but the hand is complete with five digits[3,4,13,14].



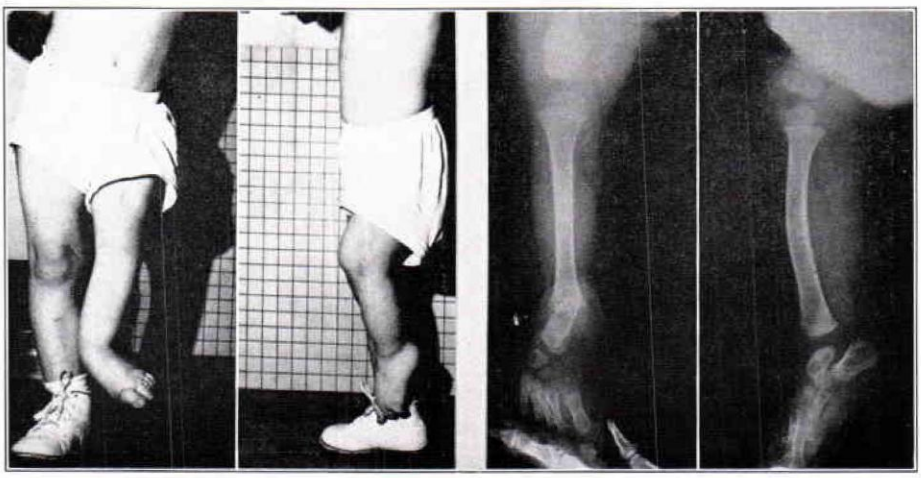
INTERCALARY LONGITUDINAL COMPLETE PARAXIAL HEMIMELIA: TIBIAL I/1 TI

Incidence: Area Child Amputee Center: three cases in 300 (two males, one female; no bilateral).

Genetics: sporadic; possibly hereditary; non-recessive.

Associated defects: complete paraxial hemimelia, radial; microtia.

The tibia is absent. There are five toe rays, indicating that the foot is not involved. Otherwise this anomaly skeletally parallels complete paraxial hemimelia, tibial (Terminal Longitudinal, T/1 TI)[3,7,13].

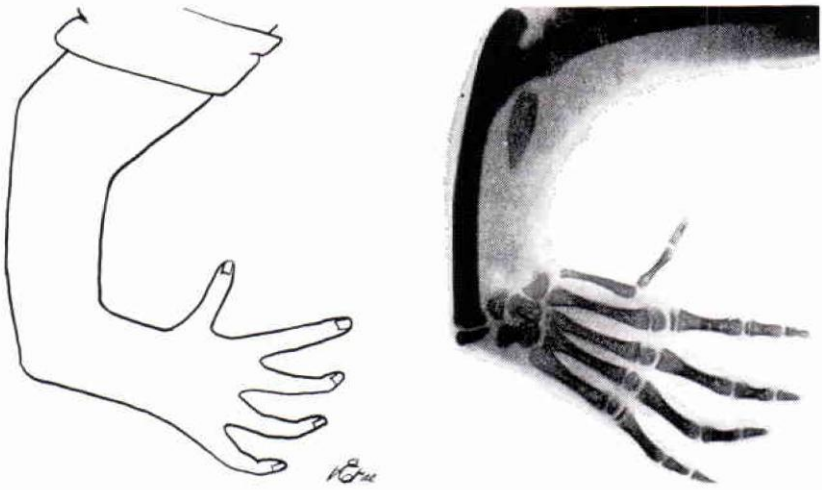


INTERCALARY LONGITUDINAL COMPLETE PARAXIAL HEMIMELIA: FIBULAR I/1 FI

Incidence: Area Child Amputee Center: ten cases in 300. One bilateral; six males, four females.

Genetics and associated defects: see Terminal Longitudinal, Complete Paraxial Hemimelia, Fibular (T/1 FI).

The fibula is absent. This deficiency differs from Terminal Longitudinal Complete Paraxial Hemimelia, Fibular (T/1 FI), in that there are five digits, indicating normal skeletal components in the foot. The foot sometimes rests in valgus[1,3,5,9,11,13].

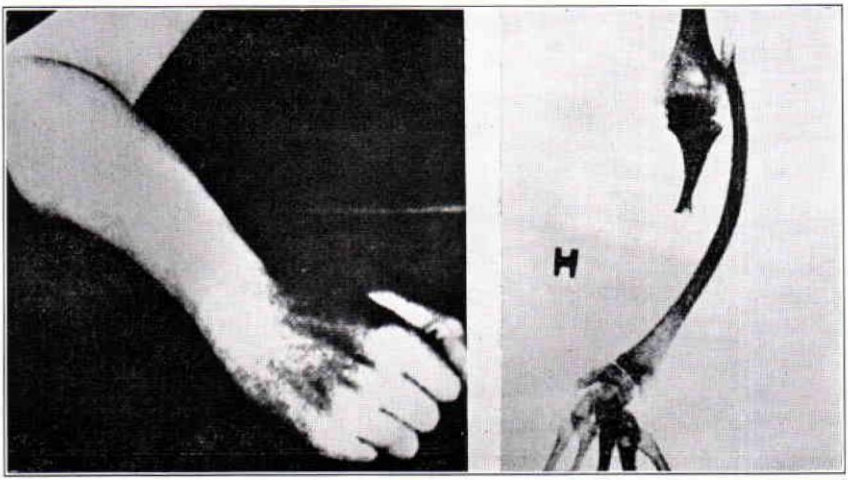


INTERCALARY LONGITUDINAL INCOMPLETE PARAXIAL HEMIMELIA: RADIAL 1/2 r

From Birch-Jensen[4]: *Congenital Deformities of the Upper Extremities.*

Incidence: Area Child Amputee Center reports no cases. Birch-Jensen[4] includes this deficiency in his presentation of radial defects.

This deficiency differs from Incomplete Paraxial Hemimelia, Radial (Terminal Longitudinal, T/2 r) in that there are five digits[3,4,13,14].

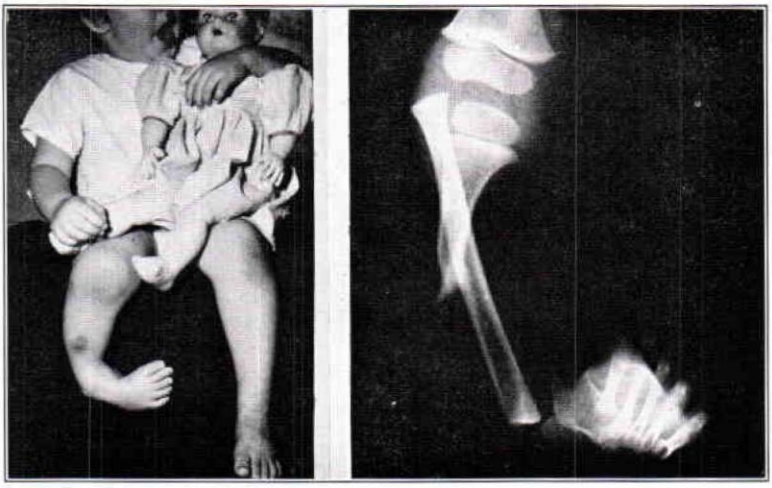


INTERCALARY LONGITUDINAL INCOMPLETE PARAXIAL HEMIMELIA: ULNAR 1/2 u

From Birch-Jensen[4]: *Congenital Deformities of the Upper Extremities.*

Incidence: Area Child Amputee Center reports no cases. Birch-Jensen[4] classifies this among deficiencies with ulnar defects.

Clinically this appears to be a ventrally bowed forearm with apparent luxation of the elbow and a defect on the ulnar side of the forearm. Five digits are present. Roentgenographic examination discloses a portion of the ulna to be present (see Incomplete Paraxial Hemimelia, Ulnar; Terminal Longitudinal, T/2 u)[3,4,13,15].



INTERCALARY LONGITUDINAL INCOMPLETE PARAXIAL HEMIMELIA: TIBIAL I/2 ti

Courtesy of Chestley L. Yelton, M.D., Lloyd Noland Hospital, Fairfield, Alabama

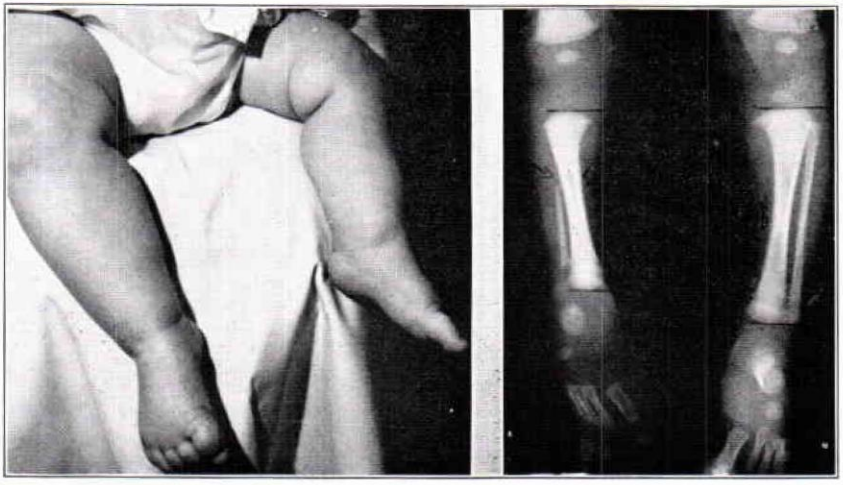
Incidence: Area Child Amputee Center: three cases (one male, two females) in 300.

Genetics: insufficient knowledge.

Associated defects: proximal femoral focal deficiency; complete paraxial hemimelia; radial; partial adactylia; partial aphalangia.

Synonym: delayed ossification of the tibia.

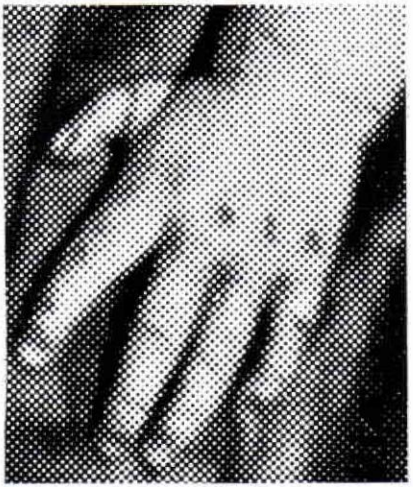
This differs from Complete Paraxial Hemimelia, Tibial, in that a portion of the tibia (usually proximal) is present, with a complete or incomplete proximal tibial epiphyseal plate. If there were fewer than five digits, this would be a terminal longitudinal defect.[1, 3, 13, 14, 19]



INTERCALARY LONGITUDINAL INCOMPLETE PARAXIAL HEMIMELIA: FIBULAR I/2 fi

Incidence: Area Child Amputee Center: one case (female) in 300.

This differs from Complete Paraxial Hemimelia, Fibular (I/1 FI), in that roentgenographic examination reveals a portion of the fibula to be present, usually distally. If there were fewer than five toes, this would be a terminal longitudinal defect[1,3,5,9,11,13].



INTERCALARY LONGITUDINAL — PARTIAL ADACTYLIA — I/3

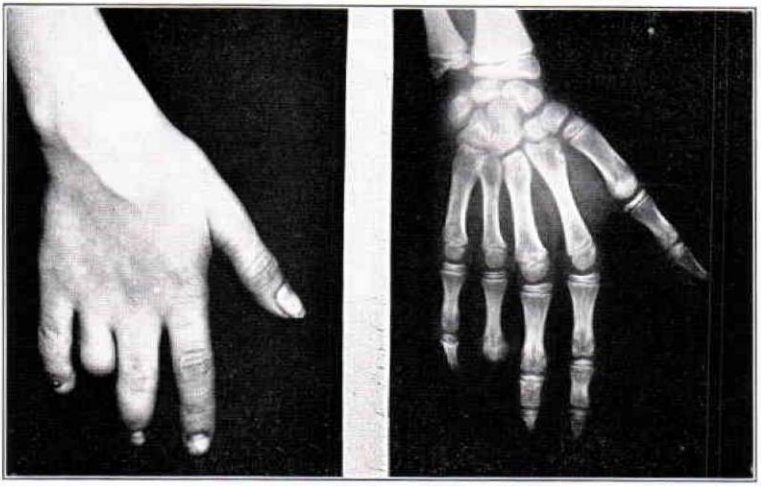
From Birch-Jensen[4]: *Congenital Deformities of the Upper Extremities.*

Incidence: Area Child Amputee Center reports no cases. Birch-Jensen[4] classifies this deficiency with radial defects and aplasias (nineteen cases: reported population incidence is one in 55,000).

Genetics: may be inherited and dominant, but the great majority is not inherited.

Synonym: floating thumb.

Absence of all or part of a metacarpal or metatarsal. The styloid process of the radius, the scaphoid, and the trapezium are usually either absent or small. The hand illustrated shows this deficiency in the thumb ray, the so-called floating thumb. This type of deficiency is not seen in amputee clinics[4,13].



INTERCALARY LONGITUDINAL — PARTIAL APHALANGIA — I/4

Incidence: Area Child Amputee Center: two cases in 300. Birch-Jensen[4] reports forty-one cases in population of 4,024,000.

Genetics: sporadic in majority of cases; when inherited it is dominant.

Synonyms: ectrodactylism; hypophalangia.

Associated defects: lower hemimelia; partial hemimelia.

Absence of the proximal phalanx or middle phalanx, or both, from one or more digits. In the hand illustrated only the little-finger ray is intercalary; note that the distal phalanx is present as is the finger nail (the ring-finger ray is T/4:4—the distal phalanx is absent)[4,13,15].

REFERENCES

1. AITKEN, G. T.: Amputation as a Treatment for Certain Lower Extremity Congenital Abnormalities. J. Bone and Joint Surg., **41-A:** 1267-1285, Oct. 1959.

2. AITKEN, G. T., and FRANTZ, C. H.: Congenital Amputation of the Forearm. Ann. Surg., **141:** 519-522, 1955.

3. AITKEN, G. T., and FRANTZ, C. H.: Management of the Child Amputee. *In* Instructional Course Lectures, The American Academy of Orthopaedic Surgeons. Vol. 17, pp. 246-298. St. Louis, The C. V. Mosby Co., 1960.

4. BIRCH-JENSEN, ARNE: Congenital Deformities of the Upper Extremities. Copenhagen, Ejnar Munksgaard, 1950.

5. COVENTRY, M. B., and JOHNSON, E. W., JR.: Congenital Absence of the Fibula. J. Bone and Joint Surg., **43-A:** 941-955, Oct. 1952.

6. FREIRE-MAIA, N.; QUELCE-SALGADO, A.; and KOEHLER, R. A.; Hereditary Bone Aplasias and Hypoplasias of the Upper Extremities. Acta Genet., Basel, **9:** 33-40, 1959.

7. GILLIS, LEON: Amputations in Children. Ann. Roy. Coll. Surg., **19:** 335-360, 1956.

8. HALL, M. C.: The Trabecular Patterns of the Normal Foot. Clin. Orthop., **16:** 15-20, 1960.

9. HARMON, P. H., and FAHEY, J. J.: Syndrome of Congenital Absence of the Fibula, Report of Three Cases with Special Reference to Pathogenesis and Treatment. Surg., Gynec., and Obstet., **64:** 876-887, 1937.

10. HEPP, OSKER (Munster, Germany): Personal communication.

11. KRUGER, L. M., and TALBOTT, R. D.: Amputation and Prosthesis as Definitive Treatment in Congenital Absence of the Fibula. J. Bone and Joint Surg., **43-A:** 625-642, July 1961

12. MCKENZIE, D. S.: The Prosthetic Management of Congenital Deformities of the Extremities. J. Bone and Joint Surg., **39-B:** 233-247, May 1957.

13. O'RAHILLY, RONAN: Morphological Patterns in Limb Deficiences and Duplications. Am. J. Anat., **89:** 135-193, 1951.

14. O'RAHILLY, RONAN: The Development and the Developmental Disturbances of the Limbs. Irish J. Med. Sciences, No. 397, pp. 30-33, January 1959.

15. O'RAHILLY, RONAN; GARDNER, ERNEST; and GRAY, D. J.: The Skeletal Development of the Hand. Clin. Orthop., **13:** 42-51, 1959.

16. O'RAHILLY, RONAN; GARDNER, ERNEST; and GRAY, D. J.: The Skeletal Development of the Foot. Clin. Orthop., **16:** 7-14, 1960.

17. THOMPSON, T. C.; STRAUB, L. R.; and ARNOLD, W. D.: Congenital Absence of the Fibula. J. Bone and Joint Surg., **39-A:** 1229-1237, Dec. 1957.

18. WILLIS, R. A.: The Borderland of Embryology and Pathology. London, Butterworth and Co., 1958.

19. YELTON, C. L. (Fairfield, Alabama): Personal communication.

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