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Chondroectodermal dysplasia (Ellis–van Creveld syndrome): a case report

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SUMMARY A case of chondroectodermal dysplasia (Ellis–van Creveld syndrome) with a remarkable number of the classic oral and dental changes is described. This syndrome involves all embryonic tissue layers and is polysymptomatic; yet some oral and dental manifestations are pathognomonic and must be considered in primary diagnostic criteria. However, in some patients, these oral and dental manifestations are not clinically evident leading to misdiagnosis.

Introduction

Chondroectodermal dysplasia is a complex genetic syndrome first described in 1940 by Ellis and van Creveld (1940) which is also called mesoectodermal dysplasia, Ellis–van Creveld syndrome or chondrodystrophy syndrome. It is a rare autosomal recessive disorder whose minimal diagnostic criteria include postaxial polydactyly of the hands, short limb dwarfism and dysplastic fingernails and teeth. A case is described with some remarkable general and oral manifestations which required orthodontic treatment.

Case report

The subject, a girl, was the first child of non-consanguineous and normally developed parents. She has a sister with no congenital abnormalities. Pregnancy and birth were uneventful. The newborn showed bilateral postaxial polidactilia (hexadactilia) of both hands (Fig. 1) and the left foot. A functional systolic cardiac murmur, and a relatively short length with tendency to generalized hyperlaxity were detected. No natal teeth or genital anomalies were found, and the diagnosis of chondrodysplasia ectodermica was not made at that examination. Her psychomotor and mental development were within normal limits. Diagnosis of Ellis–van Creveld syndrome was retrospectively established based on failure of growth with a pattern of acromelic shortening of the extremities. Radiographic examination at that time confirmed shortening of the limb bones with expanded and abnormally shaped and aligned metaphyses. At the age of 11, when first seen at the Unit of Orthodontics, Madrid, Spain, her height was 131 cm (percentile 3). Her head morphology and facial appearance were normal, as was the quantity and quality of the hair (Fig. 2). Her nails were moderately dysplastic. She showed an Angle Class II, division 1 malocclusion with moderate overbite. Cephalometric parameters were normal. Anodontia of the upper right and both lower lateral incisors and severe rotation of the lower central incisors were observed. Hyperplastic fraenula and some morphological abnormalities of the teeth were apparent, particularly the upper left lateral incisor which was peg-shaped, and both upper central incisors which were bell-shaped (Fig. 3).

Some slight serrations of the alveolar ridge were observed distally to the upper central incisors and next to the rotated lower central incisors. Another identation of the gingiva was evident between these teeth when they were orthodontically derotated (Fig. 4). Radiographic examination revealed retarded formation of second molar buds (particularly the bud of the 67 was rudimentary) but no presence of incomplete mandibular or maxillary clefts.

The examination of the hands revealed the scars of the surgical excision of the extra fingers before the first year of life. The radiograph of the wrist showed fusion of the hamate and

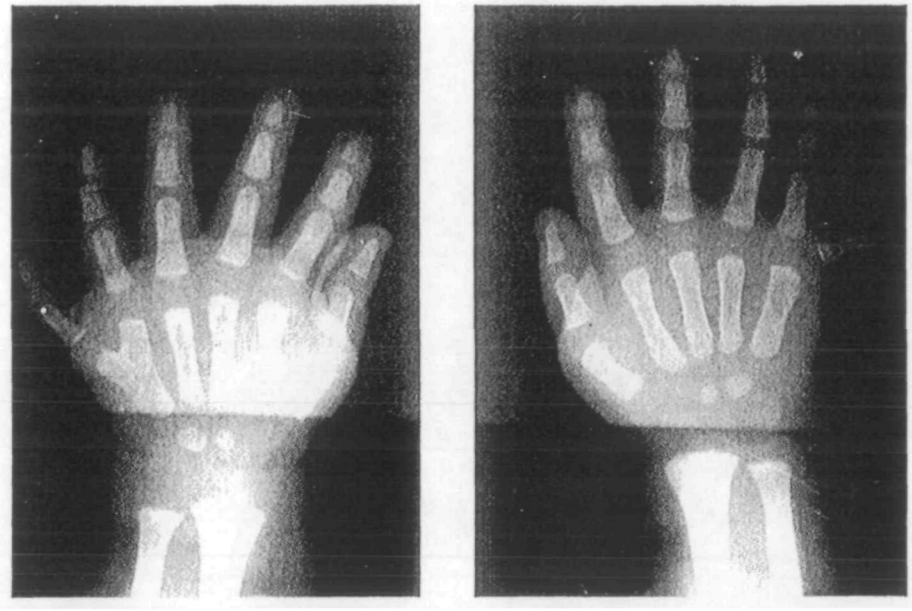

Figure 1 Radiograph of the hands of the newborn patient showing hexadactilia with fusion of 5th and 6th metacarpals of the left hand.

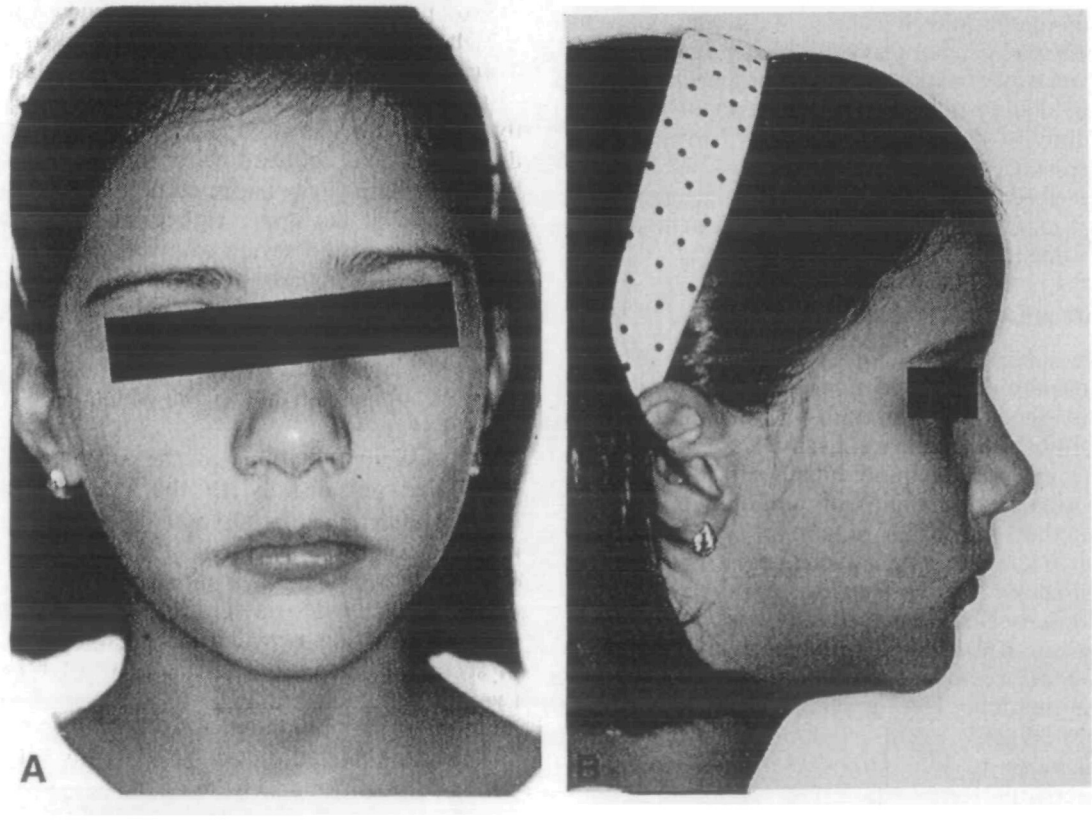

Figure 2 Normal head and facial appearance.

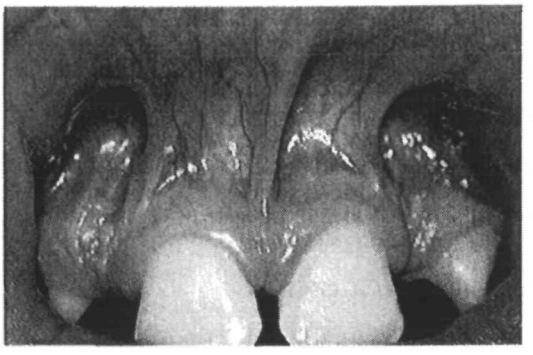

Figure 3 Hyperplastic fraenula, bell-shaped upper central incisors and anodontia of lateral left incisor.

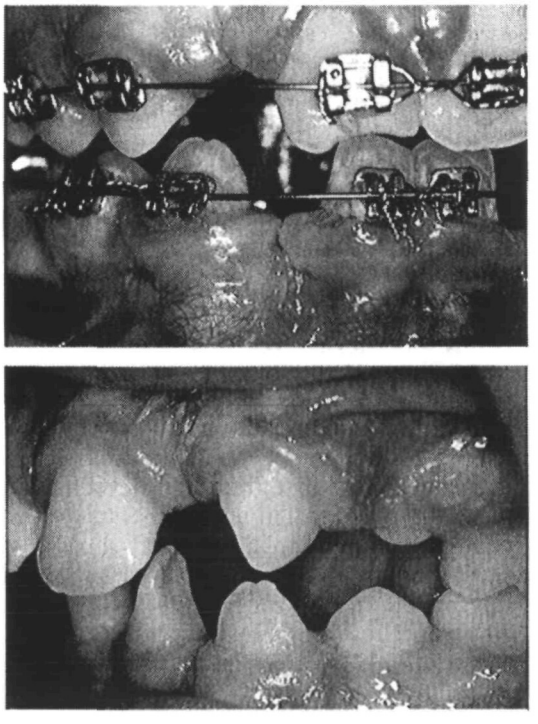

Figure 4 Serration of the alveolar ridges.

capitate bones, but the relative size of the phalanges was normal (Fig. 5).

Orthodontic treatment was started with a cervical headgear and a full fixed appliance to correct the Angle Class II, division 1 malocclusion and to prepare the spaces for implants in the place of the anodontic incisors. The most prominent fraenula were eliminated and both bell-shaped central incisors were recontoured.

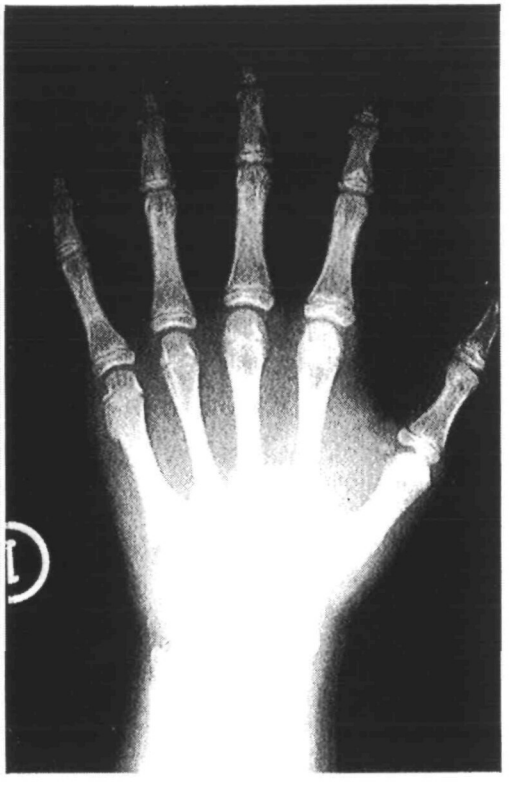

Figure 5 Radiograph of the hand at 11 years of age showing fusion of the hamate and capitate bones of the wrist.

Prevalence and genetics

Ellis–van Creveld syndrome is an autosomal recessive disorder. Sex ratio is M1:F1. It is normally very rare, although with the advent of ultrasonography an increasing number of affected foetuses are being diagnosed (Mahoney and Hobbins, 1977). It does not show racial predilection, although is very common in one Amish group (McKusick *et al.*, 1964) and has been detected in other ethnic groups in which endogamy and consanguinity are common (Oliveira *et al.*, 1980; Goldblatt *et al.*, 1992). The risk of recurrence for siblings is 1 in 4 (25 per cent) for each offspring. For children of an affected person the risk is not increased, unless the spouse is also a carrier, in which event the risk is 50 per cent.

Findings

Table 1 records the general clinical findings and Table 2 the oral and dental findings commonly

Table 1 General features of Ellis–van Creveld syndrome in our patient in comparison with the literature

Clinical features	References	Patient study
Normal intelligence rate	Very frequent: McKusick *et al.* (1964), Ellis and van Creveld (1940)	+
Short stature	Very frequent: McKusick *et al.* (1964)	+
Oral malformations	Constant	+
Short extremities with normal trunk	Very frequent: Oliveira *et al.* (1980), Douglas *et al.* (1959), Ellis and van Creveld (1940), McKusick *et al.* (1964)	+
Pelvic dysplasia	Very frequent: McKusick *et al.* (1964)	+
Polydactyly:		
hands	Constant: Oliveira *et al.* (1980), Ellis and van Creveld (1940)	+
feet	15%: Oliveira *et al.* (1980)	+(left)
Fusion of hamate and capitate wrist bones	Frequent: Oliveira *et al.* (1980), McKusick *et al.* (1964), Taylor *et al.* (1984)	+
Fusion of 5th and 6th metacarpals	Frequent	+
Dysplastic nails (slight)	Very frequent	+
Congenital heart disease (atrial septal defects)	50–60%: Blackburn and Belliveau (1971), McKusick *et al.* (1964), Ellis and van Creveld (1940), Sanchez-Cascos (1974), Oliveira *et al.* (1980)	+
Genital anomalies	Rare: McKusick *et al.* (1964)	–
Genu valgum	Frequent: Oliveira *et al.* (1980), McKusick *et al.* (1964)	+
Normal skin and hair	Constant: McKusick *et al.* (1964)	+
Retinitis pigmentosa	Very rare: Brueton and Dillon (1990)	–
Renal, hepatic dysfunction	Very rare: Blackburn and Belliveau (1971), Brueton and Dillon (1990)	–
Neural malformations	Very rare: Blackburn and Belliveau (1971)	–

Constant = 100%; Very frequent = > 60%; Frequent = > 20%; Rare = < 20%; Very rare = occasional cases.

present in this syndrome and those observed in our patient.

Discussion

All embryonic layers appear involved in Ellis–van Creveld syndrome. The signs of ectodermal dysplasia are usually limited to nails, teeth and gums, although some cases with eye (Salvador *et al.*, 1990) and neural involvement (Nabrady, 1961; Blackburn and Belliveau, 1971; Rosemberg *et al.*, 1983) have been described. Abnormalities of the bones (Ellis and van Creveld, 1940; McKusick *et al.*, 1964; Oliveira *et al.*, 1980; Taylor *et al.*, 1984) and the heart (Ellis and van Creveld, 1940; McKusick *et al.*, 1964; Sanchez Cascos, 1974; Oliveira *et al.*, 1980) and in some patients of the kidneys (Rosemberg *et al.*, 1983) indicate the mesodermal involvement. Endodermal involvement is not very common, but some patients with lung and liver abnormalities have been reported (Brueton and Dillon, 1990).

The partial harelip and maxillary alveolar clefts and continuum fraenum could be related to a partial failure in the normal development of the embryonic median nasal process; yet this mechanism can not explain the partial clefts of the mandibular alveolar process as suggested by Biggerstaff and Mazaheri (1968).

Ellis–van Creveld syndrome presents phenotypic variations. Isolated findings in near relatives, such as polydactyly, short stature or abnormalities of wrist bones without other stigmata of the syndrome, have been frequently described. A maternal aunt of our patient showed a short stature (130 cm). It has been suggested that some oral manifestations, mainly the combination of partial clefts of the alveolar processes, partial harelip and continuous fraenum, are pathognomonic and should be used in primary diagnosis as other criteria are inconclusive (Biggerstaff and Mazaheri, 1968). Based on these signs, the differential diagnosis with other syndromes that include short stature, polydactilia and orofacial abnormalities (I and

Table 2 Oral malformations of Ellis–van Creveld syndrome in our patient in comparison with the literature.

Oral malformation	References	Patient study
Natal teeth	Douglas *et al.* (1959), Ellis and Andrew (1962), McKusick *et al.* (1964)	–
Malocclusion	Biggerstaff and Mazaheri (1968), Oliveira *et al.* (1980), Gorlin and Pindborg (1964), Prahu *et al.* (1978), Winter and Geddes (1967)	+
Partial anodontia (deciduous and permanent dentition)	Biggerstaff and Mazaheri (1968), Prahu *et al.* (1978),	+
	Sarnat *et al.* (1980), Winter and Geddes (1967)	(?, +)
Retarded eruption	Prabhu *et al.* (1968)	–
Enamel dysplasia and hypocalcification	Biggerstaff and Mazaheri(1968), Oliveira *et al.* (1980), Ellis and Andrew (1962), Winter and Geddes (1967)	–
Malformed teeth (peg-, bell-, shovel-shaped...)	Biggerstaff and Mazaheri (1968), Oliveira *et al.* (1980), Ellis and Andrew (1962), Prahu *et al.* (1978), Sarnat *et al.* (1980), Winter and Geddes (1967)	+
Fusion, gemination, supernumerary	Ellis and Andrew (1962), McKusick *et al.* (1964), Prahu *et al.* (1978), Sarnat *et al.* (1980)	+
Transposition	Oliveira *et al.* (1980)	–
Torsion	Oliveira *et al.* (1980)	+
Lack of root development	Ellis and Andrew (1962)	–
Labiogingival adherences	Oliveira *et al.* (1980), Ellis and Andrew (1962), McKusick *et al.* (1964), Prahu *et al.* (1978), Sarnat *et al.* (1980), Winter and Geddes (1967)	–
Fraenula hypertrophy	McKusick *et al.* (1964), Biggerstaff and Mazaheri (1968), Oliveira *et al.* (1980), Sarnat *et al.* (1980)	+
Accessory fraenula	Prahu *et al.* (1978), Sarnat *et al.* (1980), Winter and Geddes (1967)	+
Bilateral incomplete clefts of both alveolar processes	McKusick *et al.* (1964), Biggerstaff and Mazaheri (1968), Ellis and Andrew (1962), Prahu *et al.* (1978), Winter and Geddes (1967)	–
Serrated gingiva	Oliveira *et al.* (1980), Ellis and Andrew (1962), Sarnat *et al.* (1980)	+
Midline puckering of the upper lip (partial harelip)	McKusick *et al.* (1964), Bigerstaff and Mazaheri (1968)	–

II Orofaciodigital Syndromes) (Gorlin and Pindborg, 1964) and asphyxiating thoracic dystrophy (Brueton and Dillon, 1990) can be established.

The clinical variability of the oral abnormalities in Ellis–van Creveld syndrome could be due to the fact that its genetic effect on the teeth and other oral structure development occurs during a relatively long period, and could be the result of other genetic and environmental phenotype modifying factors.

Our patient showed many of the characteristic changes described in the permanent dentition. The morphology of her primary teeth is unknown since when first seen, all her deciduous dentition, except the second molars, had exfoliated.

Malocclusions are common in Ellis–van Creveld syndrome but are not of any specific type. On the other hand, many patients, as reported here, show normal cephalometric parameters. Hypoplasia of the anterior maxilla was a part of the syndrome described in the Amish community (McKusick *et al.*, 1964) and was responsible for the prognathism in other patients (Prahu *et al.*, 1978). A wide gonial angle causing an increased height of the lower third of the face has also been shown in this syndrome (Prahu *et al.*, 1978).

In its full-blown clinical form, this syndrome

is easily diagnosed but, in the absence of some of the most relevant general manifestations, some of the oral and dental signs are so peculiar that they constitute a fundamental cue for the differential diagnosis and genetic counselling. However, in some patients the oral and dental signs are also minor and the orthodontist must interprete them adequately.

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