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CASE REPORT

Hay–Wells syndrome (AEC): a case report

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We would like to present a case of the rare genetic skin disorder catalogued as AEC syndrome. This rare disorder was described in 1976 by Hay and Wells in seven individuals from four families, and it entails a complex polymalformative syndrome with an autosomal-dominant inheritance pattern and variable penetration. Descriptive explanation and facial and intraoral images of this rare disorder constituted the study design. The neonatal report outlines dysplastic phenotype, micrognathia, hypoplasia of the hard and soft palate, cleft palate, small nose, mammary hypoplasia with ectopic mammary nodules, hypoplastic external genitalia with clitoral hypertrophy, hypoplasia of the nails, a tendency towards dorsiflexion of the big toe on both feet, ankyloblepharon filiforme, low positioning of the auricles and faulty development of the left auricle, scaly exanthema with eritrodermatitis and hyperkeratosis, good lung ventilation, normal heart rhythm and normal neurological examination. Although only a few cases published are available, clinical variability is one of the hallmarks of AEC syndrome. The majority of authors consider ankyloblepharon, ectodermal dysplasia and orofacial clefting as cardinal signs. They are all are present in the case reported.
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Clinical case

The patient came to our orthodontic office at the age of 8 years and 9 months. On facial examination she displayed (Figure 1) maxillary hypoplasia, sparse and thin hair, cup-shaped auricles with hypoplasia of the scafoid earhole of the left auricle, absence of eyelashes, diminished eyebrows, commissural keilosis and keilitis. Extremities were palmoplantar keratodermy, dystrophic and hypoplastic nails, slight syndactyly between third and fourth toes. Lateral skull radiograph revealed maxillary hypoplasia with anterior crossbite and decreased lower anterior face height with severe destruction and absence of dental structures (Figure 2). Orthopantomography (Figure 3) revealed right upper molar with gemini appearance and right lower molar with supernumerary roots and a pearly enamel.

Occlusal photography (Figure 4) revealed oligodontia and pronounced dental anomalies in both shape and size with severe dentine loss and amelogenesis imperfecta.

To carry out the differential diagnosis, we have to consider the clinical sets of symptoms related to ectodermal dysplasia and ankyloblepharon filiforme adnatum and also, although to a lesser extent, all signs caused by oral and/or facial clefting. Special emphasis must be placed on the signs shared by both conditions (Hay and Wells, 1976). The phenotypic variability makes it difficult to establish a correct diagnosis. There are more than a hundred clinical conditions known to be related to ectodermal dysplasia. We can identify at least four types of syndrome involving ankyloblepharon, and finally, there is a multitude of symptoms related to facial clefting. In clinical practice, the most similar syndromes are Bowen Armstrong syndrome (EEC syndrome), ectodermal dysplasia, ectrodactyly and cleft/lip palate, Rapp-Hodkin syndrome, acro-dermato-ungual-lacrimal-tooth (ADULT) syndrome. Their similarities have been covered in depth by some authors (Cambiaghi *et al*, 1994). All these syndromes show an autosomal-dominant inheritance. Clinical distinction among these syndromes is based on the degree of expressivity of each disorder and the occurrence of unique characteristics.

Comments

Hay–Well's syndrome is a type of exceptional autosomal-dominant, ectodermal dysplasia caused by heterozygous missense mutations in the carboxyl terminal region with a sterile alpha motif (SAM) domain of the p63 gene (McGrath, 2001). P63, a P53 family gene member, is required for craniofacial and limb development as well as for proper skin differentiation. Clinical variability is one of the hallmarks of AEC syndrome (Spiegel and Colton, 1985; Greene *et al*, 1987), although the majority of authors consider ankyloblepharon, ectodermal dysplasia and orofacial clefting as cardinal signs (Mancini and Palleras, 1997).

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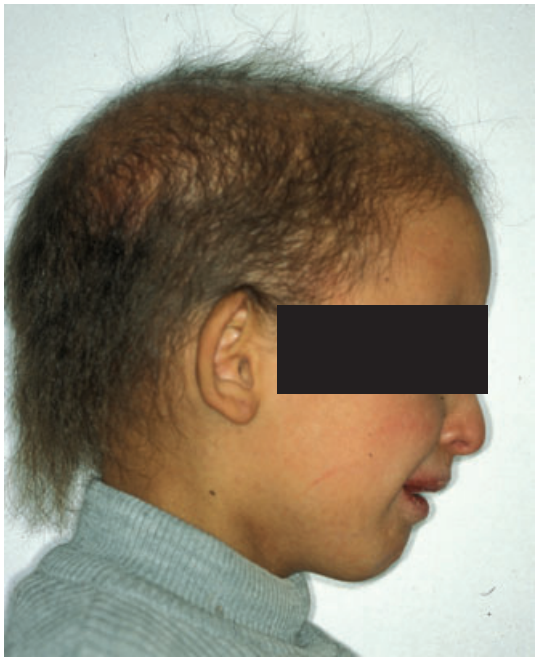

Figure 1 Facial lateral view

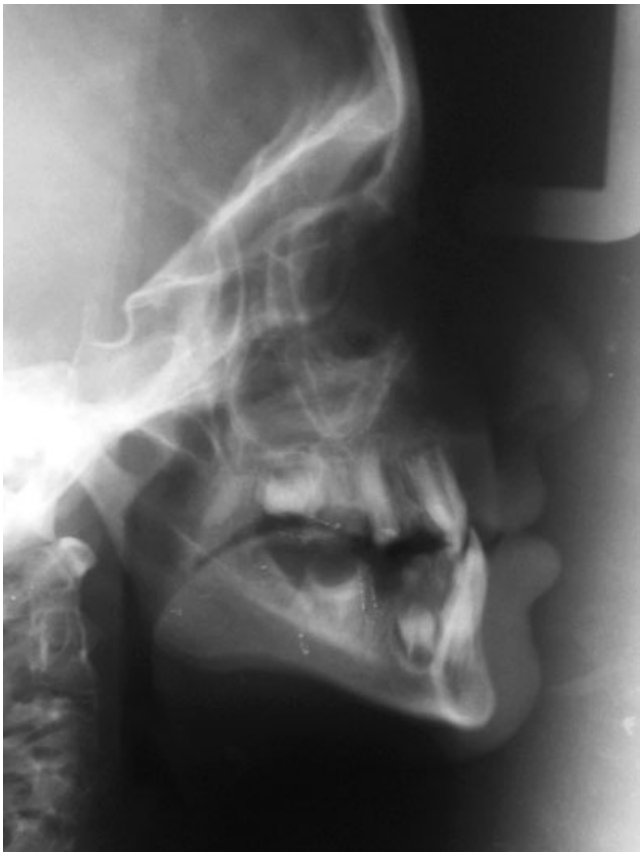

Figure 2 Lateral skull radiograph

In our patient, the parents were not affected, although the father was 47 years old, which might be the cause for the autosomal-dominant mutation that caused the syndrome.

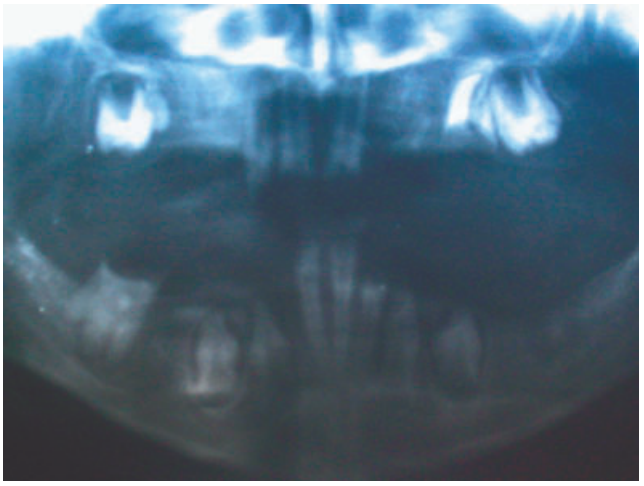

Figure 3 Orthopantomography

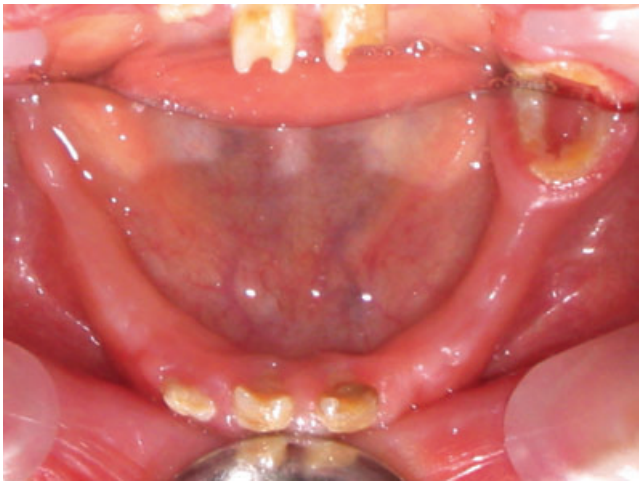

Figure 4 Lower occlusal view

These disorders are normally classified and treated first in different hospital departments (dermatology, ophthalmology, etc.). They arrive late at the dental office due to the severity of systemic complications (Vanderhooft *et al*, 1993; Drut *et al*, 2002) which may arise, and the large number of necessary consultations between different medical specialists (Saxena and Kaur, 1965; Ehlers and Jensen, 1970; Lodha and Ng, 2004) it is difficult to have a good team approach to the patient.

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