Monilethrix: a review and case report

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Monilethrix, a term of Greek and Latin derivation meaning necklace hair, is a rare ectodermal condition inherited as an autosomal dominant trait with high penetrance and variable expressivity.[1,2] The latter characteristic may be partially due to hormonal fluctuations, including puberty and pregnancy, and to environmental factors such as seasonal changes.[2] The degree of expressivity is difficult to quantitate.[2] Some patients are so mildly affected that their hair disorder is overlooked.[3] However, very mild and quite severe cases of monilethrix have been found within the same sibship.[2] Both males and females are equally affected, and expressivity seems to be equally variable within families. Individuals affected with rare autosomal disorders have one affected parent and the condition is transmitted for generations. Fifty percent of the children will have the condition as a result of one affected parent and one unaffected parent.[4] Not every case has affected parents; new mutations are possible. The incidence is unknown. Most known cases are of European origin, however, Indian and Arab pedigrees have been described.[5] Extensively affected kindreds with the pattern of dominant inheritance have been reported.[5] Solomon and Schnyder in McKusick[5] suggested that one of five families might have a recessive form of the disorder.

This condition is characterized by a variation in the hair shaft thickness, with small node-like deformities that produce a beaded appearance, internodal fragility, breakage, and partial alopecia.[6] Alopecia may be the presenting manifestation.[5] The degree of alopecia varies from patient to patient and from time to time in the same individual.[5] In this disorder, normal neonatal lanugo hairs are shed during the first week of life, and subsequent hair growth — generally at about the second month of life — becomes dry, lusterless, and brittle, failing to grow to any appreciable length.[6] Affected hairs are short, usually < 1.9 cm in length, but some patients have longer hair (reaching almost 7 cm).[1] Hairs that have the typical monilethrix morphology can show a spectrum of severity,[1] however, both the normal and altered hairs are short.[1] The normal hairs appear to suffer the same process of breakage that affects the other hairs.[1] In severe cases the infant may remain bald, or the scalp hair may be sparse, easily fractured, and stubble-like.[6] Generally, a disorder of scalp hair, eyebrows, eyelashes, body hair, and pubic hair may also be affected.[6,7] Monilethrix appears as an inborn error of metabolism, due to an enzymatic defect, similar to that found in other enzymatic diseases.[8] In monilethrix, the genetic effect appears to be an arginosuccinase deficiency when arginosuccinic acid is found in the urine.[8] Monilethrix also has been described as a syndrome involving the triad of moniliform hairs, follicular keratosis, and koilonychia (dystrophic fingernails).[3]

The etiology of monilethrix is unknown.[6,7] It is not characteristic of any systemic disease or metabolic defect, but may be seen in association with keratosis pilaris, brittle nails, cataracts, or dental abnormalities.[6] There is no relationship between the beading and the keratosis, which is even absent at times.[3] A tendency to spontaneous improvement or remission may occur at puberty or during pregnancy and, in some cases, may continue during adult life.[6,7] Solomon and Green in Andrew's Diseases of the Skin[9] noted that improvement of the hair may occur during pregnancy, but after delivery the hair returns to its original state. They also state a seasonal improvement during the summer, and a favorable response is also attained with the use of systemic corticosteroids.[9] In some individuals, the disease may persist unchanged throughout adulthood.[6] Prognosis is variable and there is no effective therapy.[6] There are scattered reports of associations among monilethrix, CNS abnormalities, and problems with the dentition, but the relationship has not been defined.[4] The purpose of this article is to present the dental findings of monilethrix that are not well known and to differentiate the disease from ectodermal dysplasia.

Case report

JS, a 7-year-old Caucasian male, was referred to the pediatric dental clinic with a periapical abscess in one of his primary molars. The initial evaluation revealed a very friendly and spontaneous child, well-nourished

with normal physical development. The patient displayed the classical features of ectodermal dysplasia, which included sparse hair, very fine eyebrows, thick

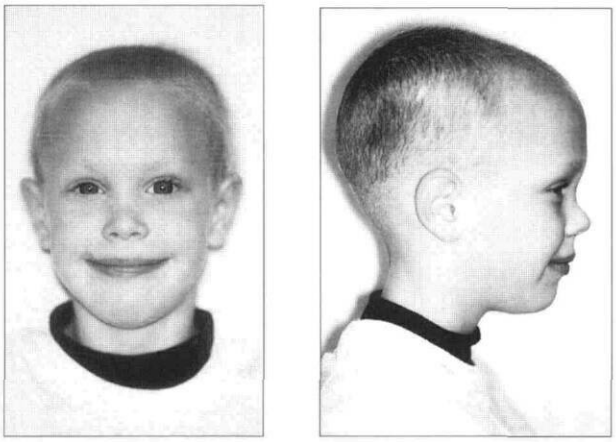

Fig 1. Frontal and lateral views of a 7-year-old with monilethrix. Notice the sparse hair, fine eyebrows, protrusive lips, and mild saddle nose.

protrusive lips and a mild saddle nose (Fig 1). No other physical feature related to ectodermal dysplasia was found. The father was questioned about the patient's health history and no abnormal findings were highlighted in the health history questionnaire. However, the father did mention, after being asked, that his son did not have ectodermal dysplasia but a condition known as monilethrix. The parents noticed in both siblings short hair at birth, and it became noticeable that it was abnormally short within the first 2 years of life. Their scalps were dry and the hair was brittle, with circumscribe keratosis pilaris. As they grew older, the keratosis pilaris became more obvious with a noticeable predilection for the back of the neck. JS's condition was clear, given the family history, but no treatment was implemented because of the family's previous experience of spontaneous improvement at a later age in both the the patient's father and oldest brother.

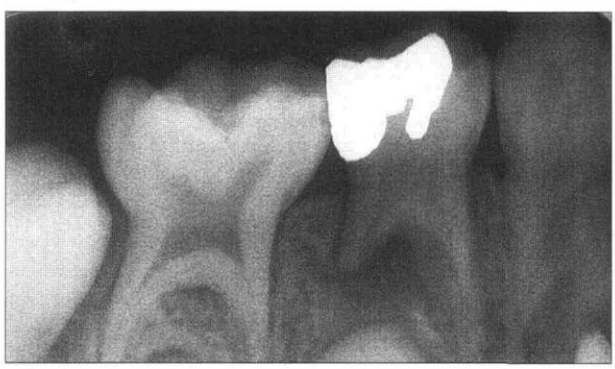

Fig 2. Periapical radiolucency clearly seen in the mandibular right first primary molar. The mandibular right second primary molar reflects the presence of taurodontia.

Oral examination revealed an early mixed dentition with the only permanent teeth erupted being the mandibular central incisors and the six-year molars. Restorations were found in all posterior quadrants. Oral hygiene was fair and the patient had a bilateral mesial step molar relationship and a bilateral class I canine relationship. Saliva outflow and consistency was normal. The father reported a swelling around his son's mandibular right primary first molar for the last 24 hr, increasing in size with no apparent discomfort or pain. Soft tissue examination revealed a 5x4-mm soft, red exophytic lesion with a broad base on the attached gingiva at the distofacial aspect of the mandibular right first primary molar. The attached gingiva around the maxillary left primary first molar appeared red, and a white exudate typical of a periapical abscess was present. Panoramic, periapical, (Fig 2) and bite-wing

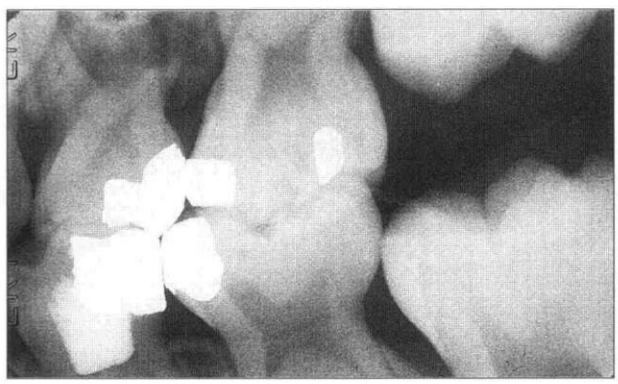

Fig 3. Radiolucency involving the furcae of the maxillary left primary first molar. Large pulp chamber clearly seen in the mandibular left primary first molar.

(Fig 3) radiographs of the areas involved were exposed to rule out any other abnormalities. The radiographs showed a radiolucency in the area of the furcae of both teeth in question. The panoramic view and individual radiographs revealed a localized distinctive taurodontia in the mandibular second primary molars (Fig 4) — a characteristic commonly seen in ectodermal dysplasia. The father consented to the extractions of the abscessed teeth and the placement of space maintainers both in maxilla and mandible.

Discussion

This case emphasizes the importance of a differential diagnosis and also shows variations and correlations between two different ectodermal conditions. The patient's physical features prompted the idea that a relationship with ectodermal dysplasia existed, and it was important to find the link between both conditions. The patient presents the typical physical features of a patient with ectodermal dysplasia, however, no absence of sweat glands, sebaceous glands or dry skin was present. According to the father's narrative, both JS and his 13-year-old brother inherited this rare ectodermal condition from the paternal side of the family.

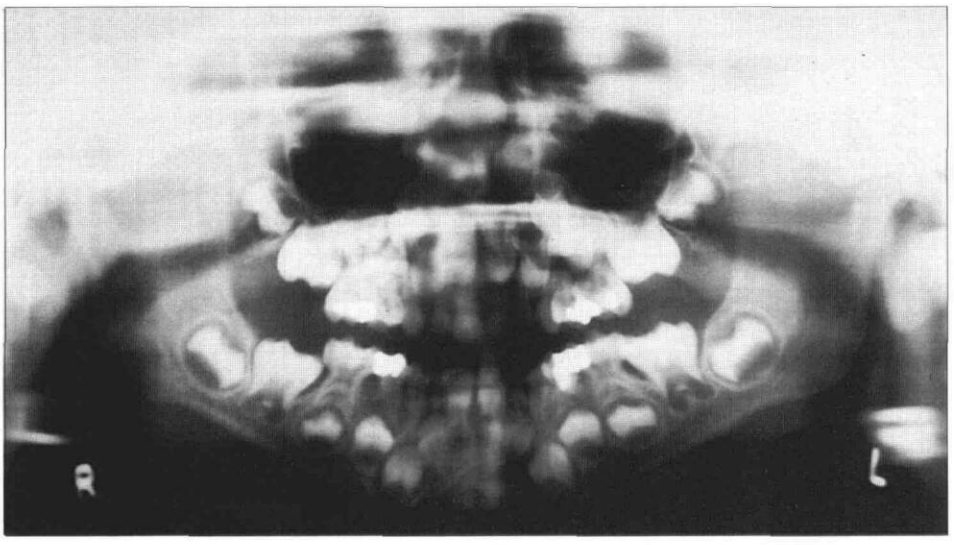

Fig 4. Panoramic radiograph at age 7 showing the presence of taurodontia in the mandibular primary second molars.

No health or developmental problems have been reported in this family except for some common dental problems during childhood. The father had suffered from monilethrix during his childhood but did not mention the presence of monilethrix in previous generations. The father had a normal physical appearance and normal scalp hair.

JS had keratosis pilaris, which is a common feature of this condition. His hair was brittle and broke easily. Improvement has been seen during the summer months in both JS and his brother. A more significant improvement is taking place as JS's oldest brother reaches puberty and has developed very healthy scalp hair.

Recently, a clinical breakthrough utilizing topical minoxidil has been reported to be effective in treating alopecia in monilethrix.[3] This potent vasodilator seems to act on vellus hair follicles.[3] Speculation exists that minoxidil synchronizes the development of anagen hairs and helps prolong the growing phase.[3] The absence of koilonychia in JS separates this particular case from the previous described monilethrix syndrome.

The literature does not provide extensive information or specifics regarding monilethrix and its dental implications. Dental problems are reported in cases where the individual has oculodentodigital syndrome and monilethrix also was present.[3, 10] In this particular case, the only dental abnormality that resembles those who suffer from ectodermal dysplasia is the presence of taurodontia in the molar area. It is important to emphasize that in JS's situation, the presence of taurodontia played a significant role in the patient's dental problem. Typically in cases like this, it is presumed the dental problems are caused by poor oral hygiene indoctrination and, as a result, severe problems are inevitable. A complete health history and radiographic survey in cases such as this, can help us to better understand certain conditions, leading to a better treatment plan and better results.

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