LETTER TO THE EDITOR
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CHRIST-SIEMENS-TOURAINE SYNDROME : AUTOSOMAL RECESSIVE - ANHIDROTIC ECTODERMAL DYSPLASIA- A VARIANT MERITING MENTION

Dermatology

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KEYWORDS

Sir,

Ectodermal dysplasia(ED) is a group of rare inherited diseases encompassing a primary defect in the development of ectodermal tissues and can be grouped under hidrotic, anhidrotic or hypohidrotic depending on the presence or absence of sweating. XLR inheritance is encountered in 80% cases of anhidrotic ectodermal dysplasia (AED) that classically presents as a triad of hypohidrosis, adontia and atrichia. [1] Autosomal recessive AED (AR-AED) is clinically indistinguishable from the x-linked recessive AED (XLR-AED)which is the most common type of ectodermal dysplasia. We describe here a rare case of Christ-Siemens-Touraine syndrome (AR-AED).

CASE

A 6-year-old developmentally normal male child, full term normal vaginal delivery without any complications, presented with complaints of sparse scalp hair, absent eyebrows ,eyelashes, no body hair and delayed dentition.(Figure1) Parents reported history of unexplained fever, absence of sweating and temperature intolerance since birth. His elder brother has similar complaints along with recurrent pyrexia and dysmorphic facies but the parents are asymptomatic. Cutaneous examination revealed normal palmoplantar skin and dermatoglyphics. Child had a typical old- man appearance, smooth, dry, finely wrinkled periorbital skin, frontal bossing, depressed nasal bridge, facial hyperpigmentation, everted lower-lip and bat ears.(Figure2,3) Oral examination revealed four peg-shaped widely spaced maxillary teeth and a low-arched palate.(Figure4) No nail abnormalities, limb-length discrepancy or lobster-claw deformity were found. Systemic examination and biochemical investigations were unremarkable. Hypothenar skin biopsy revealed absence of sweat glands. Genetic study was not done due to limited resources.

DISCUSSION

Our patient falls under 1-3-4 subgroup as per the Freire-Maia Pinheiro classification(1994) which designated numerals 1-5 representing hair, nail, teeth, sweat-glands and other ectodermal defects respectively. [2] Consanguinous marriage, asymptomatic parents, pedigree analysis, disease expression in >1 sibling and classical clinical picture(anhidrosis, atrichosis, hypodontia)supports the diagnosis of AR-AED. Mutations in the ectodysplasin anhidrotic receptor (EDAR) gene or ectodermal dysplasia autosomal recessive (EDARADD) gene can lead to the pathogenesis of AR-AED. [3] About fifty percentage of affected individuals exhibit mild nail dystrophy. Progressive nail bed injury is more commonly encountered in older patients. [4] Complications like hyperthermia, recurrent chest infections and diarrhea are seen in neonates due to anhidrosis/hypohidrosis and inadequate development of mucous glands in the gastrointestinal tract and respiratory tract.[5] Cool environment, light clothing, repeated water sprays and avoiding exertion are helpful in combating hyperthermic episodes. Timely diagnosis, genetic counselling and prompt treatment of febrile seizures, diarrhea and respiratory infections are vital. Placement of partial or full dentures is commonly recommended by 2 years of age. These must be periodically modified because tooth agenesis and its secondary effects on the growth and development of the jaw is a significant clinical problem.[5] Life expectancy and intelligence of ectodermal dysplasia patients are usually normal but brain damage and early neonatal deaths have been reported due to hyperthermia.[1] AR-AED carriers are phenotypically asymptomatic in contrast to partially symptomatic XLR-AED heterozygotes which becomes an important clinical clue to guess the mode of inheritance in case genetic studies cannot be performed in resource limited settings.

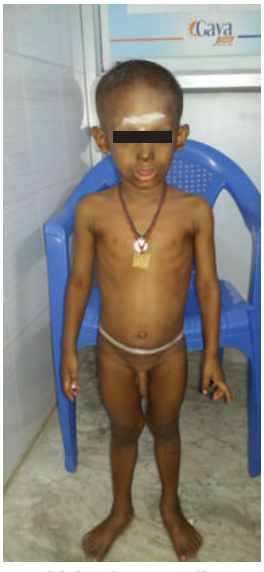
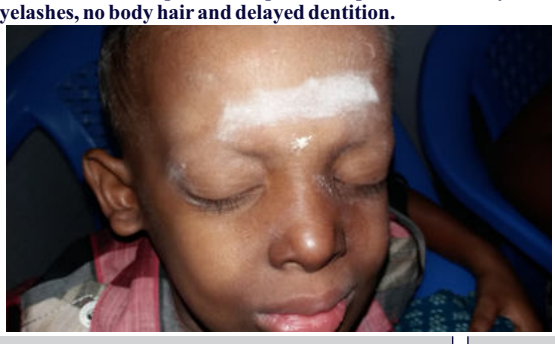


FIGURE 1 : A 6-year-old developmentally normal male child, full term normal vaginal delivery without any complications, presented with complaints of sparse scalp hair, absent eyebrows ,eyelashes, no body hair and delayed dentition.



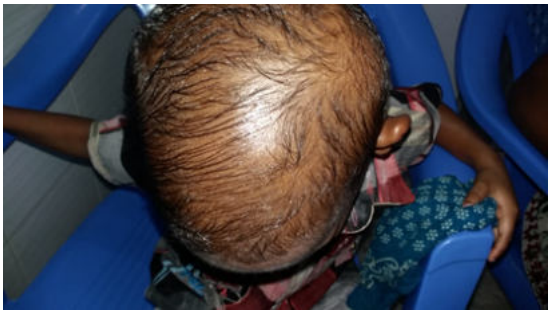


FIGURE 2 AND 3: . Child had a typical old- man appearance, smooth, dry, finely wrinkled periorbital skin, frontal bossing, depressed nasal bridge, facial hyperpigmentation, sparse hair, everted lower-lip and bat ears.

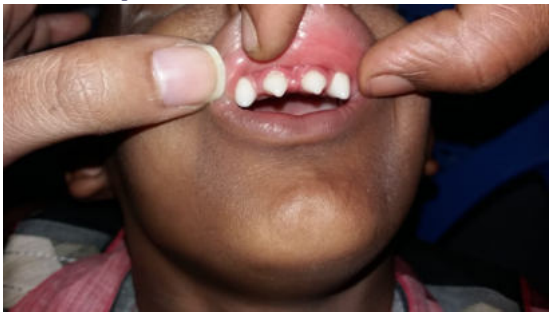


FIGURE 4: Oral examination revealed four peg-shaped widely spaced maxillary teeth and a low-arched palate.

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