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Hereditary Ectodermal Dysplasia – A Rare Entity

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A 5-year-old boy presented with complaints of sparse hair on the scalp. The parents gave a history of heat intolerance and inability to sweat noticed in the child. Child was born to a second gravida mother with uneventful antenatal and birth history. The developmental milestones were normal. On close questioning the younger male sibling 1-year-old also had sparse scalp hair and absent teeth. The maternal uncles also had absent scalp hair, premature loss of teeth, and inability to sweat. In addition, he had wrinkles on the face, loose skin folds and absent eyebrows. The picture of the child is shown in Figs. 1, 2, 3 and 4.

Intraoral examination revealed partial anodontia of the maxillary arch with the presence of four teeth suggestive of maxillary central incisors and canines on either side of the midline. The mandibular arch was completely edentulous with poorly developed alveolus. Moderate dryness of the mouth, with inflamed mucosa was evident. Skin below his eyes showed hyperpigmentation as seen with aging.

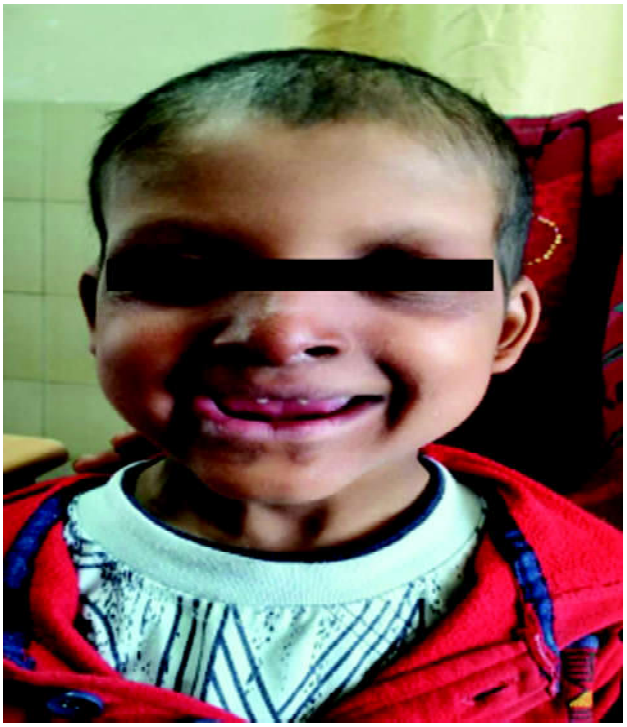

Fig. 1: *Full frontal face showing pigmantation, facial wrinkles, loss of eyebrows.*

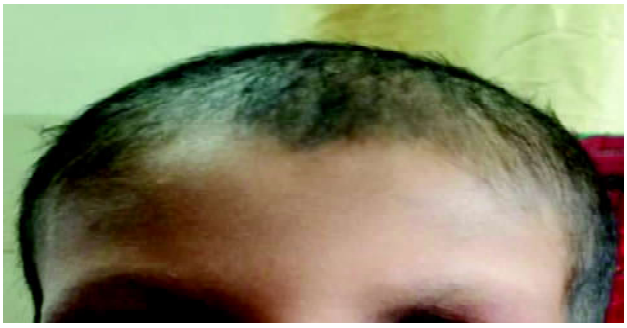

Fig. 2: *Scalp showing hypotrichosis.*

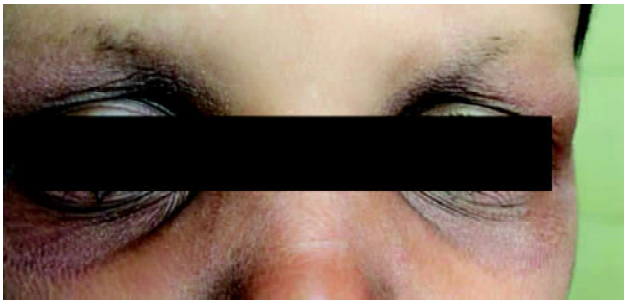

Fig. 3: *Face showing wrinkles and loss of eyebrows.*

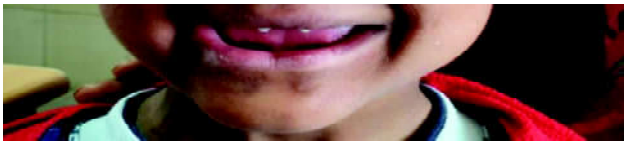

Fig. 4: *Teeth showing edentulous arches.*

Discussion

Hereditary ectodermal dysplasia (HED) is a rare genetic disorder chiefly affecting ectodermally derived structures including hair, nails, sweat glands, etc., with pathognomic manifestations such as hypotrichosis, hypohidrosis, and hypodontia. Hypohidrotic ectodermal dysplasia, is the most frequently encountered subtype and HED, being the rare subtype. HED is primarily transmitted through X-linked recessive trait in which the gene is carried by the female and manifested in male[1]. Although rare, this disorder may be seen affecting lot of members of the same family as seen in this case. In a publication by Gupta *et al*, a series of four cases

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with common classical manifestations accompanied by spoon shaped nails, hyperpigmentation, oligodontia and hypotrichosis have been described. Indian literature on this rare entity is limited. In another publication by Puttaraju *et al* ectodermal dysplasia has been described in identical siblings[2]. These publications focussed on adult patients.

Three disease-causing genes have been hitherto identified, namely, (1) EDA1 accounting for X-linked forms, (2) EDAR, and (3) EDARADD, causing both autosomal dominant and recessive forms. Recently, WNT10A gene was identified as responsible for various autosomal recessive forms of ectodermal dysplasias, including onycho-odonto-dermal dysplasia (OODD) and Schöpf-Schulz-Passarge syndrome[3]. The EDA1 gene was the most common disease-causing gene (58% of cases), and WNT10A and EDAR were each responsible for 16% of cases. Moreover, a novel disease locus for dominant HED/EDA mapped to chromosome 14q12 - q13.1. Although no clinical differences between patients carrying EDA1, EDAR, or EDARADD mutations could be identified, patients harboring WNT10A mutations displayed distinctive clinical features (marked dental phenotype, no facial dysmorphism).

The youngest case report published is an eight-year-old with ectodermal dysplasia[4]. The current report on a 5-year-old is perhaps the youngest paediatric case report.

Early prosthetic treatment leads to significant improvements in appearance, speech and masticatory function. Young patients should be recalled periodically for the prosthetic modification that is required due to continuing growth and development. The transitional prosthesis should be replaced by more definitive prosthesis once the skeletal growth is completed. Other management include protection from heat, wearing clothes that keep body cool and prevention of hyperthermia.

References

1. Gupta AA, Gotmare SS, Jain M. Hypohydrotic Ectodermal Dysplasia in an Indian Family. *J Coll Physicians Surg Pak* 2019; 29 (4): 381-3.
2. Puttaraju GH, Visveswariah PM. Ectodermal dysplasia in identical twins. *J Pharm Bioallied Sci* 2013; 5 (Suppl 2): S150-3.
3. Cluzeau C, Hadj-Rabia S, Jambou M. Only four genes (EDA1, EDAR, EDARADD, and WNT10A) account for 90% of hypohidrotic/ anhidrotic ectodermal dysplasia cases. *Hum Mutat* 2011; 32 (1): 70-2.
4. Ladda R, Gangadhar S, Kasat V, Bhandari A. Prosthodontic management of hypohidrotic ectodermal dysplasia with anodontia: a case report in pediatric patient and review of literature. *Ann Med Health Sci Res* 2013; 3 (2): 277-81.