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A rare genetic variant of Hypohidrotic Ectodermal Dysplasia – A case report

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ABSTRACT:

Ectodermal dysplasia (ED) is a rare genetic disorder that is brought on by abnormalities in the development of tissues, organs, and other accessory appendages formed from embryonic ectoderm. Ectodermal dysplasia usually presents with Bilateral congenital tooth absence. Other ectodermal abnormalities such as anhidrosis, asteatosis, hypotrichosis, and salivary gland anomalies may coexist with anodontia or hypodontia. This case report highlights the unique features when compared with a typical Ectodermal Dysplasia

KEYWORDS:

Ectodermal dysplasia, Hypohidrotic ectodermal dysplasia, Fusion, Ectodermal deformity

INTRODUCTION:

One of the uncommon genetic illnesses known as ectodermal dysplasia (ED) has primary developmental abnormalities in two or more ectoderm-derived tissues. The main tissues impacted ofthese are eccrine glands, teeth, hair, nails, skin, and hair **[1,2]**. Around 7 instances of ED occur for every 10,000 live births. **[3].** The prevalence of ectodermal dysplasia is between 1:10,000 and 1:100,000. The predilection of ectodermal dysplasia is towards the male gender and it is usually seen in Caucasians.

Ectodermal dysplasias are currently divided into approximately 150 distinct kinds. **[4]** In order to classify them, various subgroups are developed based on the presence or absence of the four

primary ectodermal dysplasia (ED) defects. ED1 refers to trichodysplasia or hair dysplasia, ED2 refers to Dental dysplasia, ED3 refers to Onchodysplasia or nail dysplasia and ED4 refers to Dyshidrosis or sweat gland dysplasia.

The salient features of Hipohydrotic ectodermal dysplasia are Frontal bossing, sunken cheeks, a saddle nose, thick and everted lips, wrinkled and hyperpigmented skin around the eyes, and big, low-set ears.**[5]**

As several organ-systems are involved in ED, the treating physician faces several treatment challenges.This case study primarily deals with the management of the disease in the pediatric age group, keeping in track with the latest developments in the field.**[5]** This case report is about ectodermal dysplasia with other unique features.

CASE REPORT:

A Girl child aged 3 years and 10 months walked into the department of pediatric and preventive dentistry who was conscious, alert normally built with normal gait; with the chief complaint of missing teeth in the lower front tooth region with difficulty inchewing solid food.

On eliciting the medical history, parents gave a history the of lip polyp and Y shaped tongue tie. During her 6 months of age tongue tie and lip polyp were excised in private hospital with post-surgicalinstructions was given. (the parent was asked not to disturb the area of suture and was asked to properly wipe the mouth after feeding and antibiotics were given)

On Eliciting the family history of the patient, the parents were consanguineously married, and thepatient has an elder brother who is apparently normal.

On enquiring the dental history, this was the patient's first dental visit

On general examination the patient was found to be alert and fully conscious. The patient had normal gait and posture and was well responsive to verbal communication. On extraoral examination, there is a prominentloss of hair seen in the forehead region and also had oddly shaped eyeballs. The head of the patientwas seen to be dolichocephalic and the facial form was euryprosopic and the nasal septum was deviated to the left side. **[Figure 1], [Figure 2]**

On clinical examination the patient had Congenitally missing 71,72,81,83 and Fusion of 82 and 84were seen. Peg shaped 82 was appreciated. **[Figure 3, Figure 4]**

Orthopantomographic examination reveals missing 71,72,81 and 83 were noted. Erupting tooth buds of 33 and 43 are seen with congenitally missing permanent incisors in the lower region. **[Figure 5]** Fusion of 82 and 84 wereseen. At the same time the fusion of 63 and 64 were also seen. Deviated nasal bridge and loss of bone thickness on the right side was appreciated.

Based on the above-mentioned clinical features and the symptoms, we provisionally diagnosed that this child has X- linked autosomal recessive Hypo hidrotic ectodermal dysplasia. This case is especially unique when compared to the other cases of ED as the Fusion of teeth is usually not seen inED, but this child presented with the symptom.

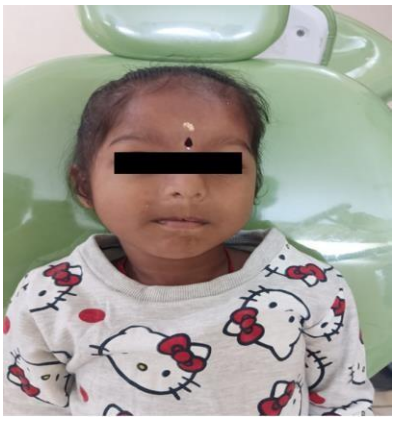

Figure 1. Deviated nasal septum

Figure 2. Lateral view of face

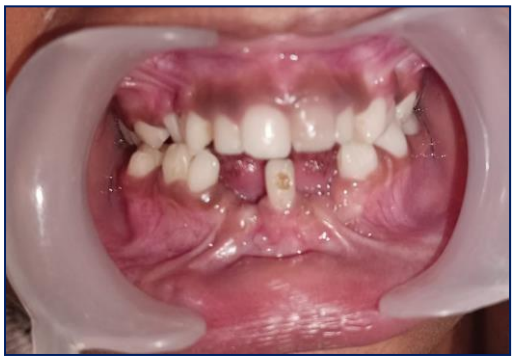

Figure 3. Intraoral frontal view

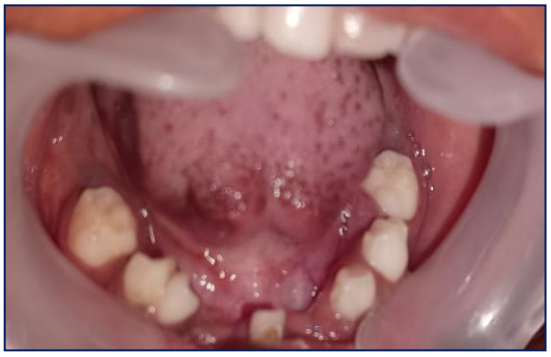

Figure 4. Intraoral occlusal view

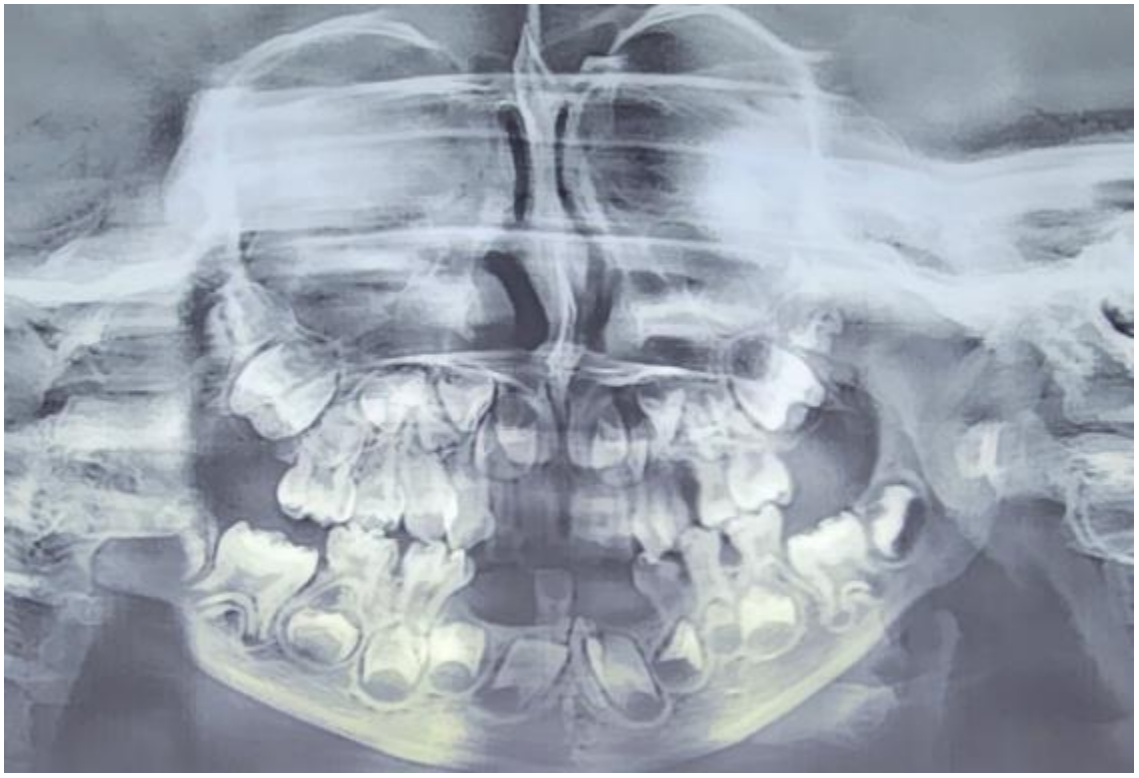

Figure 5. OPG view

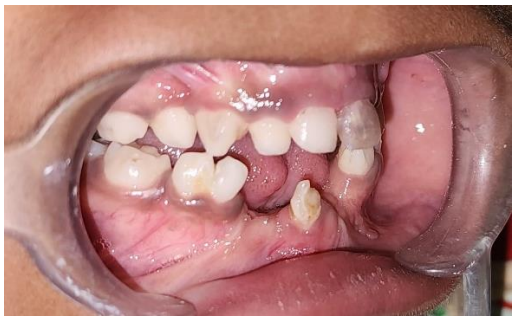

Figure 6. Intraoral lateral view

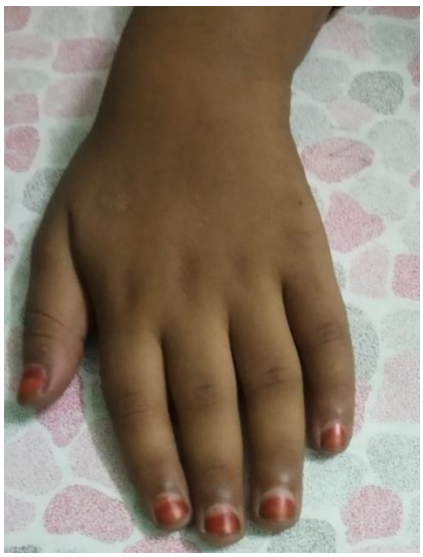

Figure 7. Curving of fingers

After an 18 month observation period, the patient had the same clinical features and dental caries were seen to develop in the proximal regions of the fused teeth and discoloration of 61 was noted due to trauma which later exfoliated. **[Figure 6]**

The absence of mandibular permanent canine crypts are seen in the orthopantomogram.

The fingers of the patients show abnormal curvature of the digits. **[Fig 7]**

DISCUSSION:

Hypohidrotic ectodermal dysplasia (HED) typically inherited as an X-linked recessive disorder, with the gene carried by the female and exhibited in the male.**(6)** The unaffected female has a fifty percent likelihood of passing on this disorder to her male children, and each female offspring has a 50% chance of inheriting the defective gene and thus becoming a carrier.

The onset of ectodermal dysplasia is most likely in the first trimester of pregnancy. If it is severe, itmanifests before the sixth week of embryonic life, which will have an impact on the dentition. The other ectodermal structures will be affected after the eighth week.**[7]**

The subgroup 1-2-3-4 includes the three most well-known ectodermal dysplasia syndromes because they share characteristics with all four of the primary ectodermal dysplasia abnormalities.

The mandibular dentition has a higher incidence of missing teeth than the maxillary dentition. The teeth that are most frequently present in the primary dentition are the mandibular canines, canines, central incisors,and maxillary 2nd molars. **[8,9]** Children with ectodermal dysplasia show maxillary retrusion due to sagittally underdeveloped maxilla, forward and upward displacement of mandible and collapsed anterior facial height. The palatal arch is frequently high and cleft palate may be present**[6,10,11]**

This particular case has all the usual findings of hipohydrotic ectodermal dysplasia with a new finding that was never documented before. This patient presented with the fusion of teeth both in the mandibular and maxillary arch. However, during early infancy diagnosis is difficult because manifestations involving teeth, hair andinability to sweat are hard to detect. **[6]**

The usual pattern of treatment involves dentures to replace the congenitally missing teeth and as the child develops, the dentures can be modified and replaced. During the mixed dentition stage, the dentures have to be modified to accommodate for the loss of primary teeth. During the permanent dentition, the removable prosthesis may be replaced by fixed depending on the number and position of the teeth. Once the Kid reaches adulthood fixed implants can be given to replace the missing teeth. **[9,12,13]**

The treatment plan for this patient involves replacement of congenitally missing teeth with partial dentures and as the child grows to the mixed dentition phase, the existing dentures has to be replaced and altered. In the permanent dentition stage, the tone of the muscles can be toned using

a vestibular screen after which an implant can be given. This condition can be managed through meticulously planned construction of dentures during the primary and mixed dentition phase.

CONCLUSION

Ectodermal Dysplasia is a very rare genetic condition that has many differing and overlapping clinicalfeatures and it is very difficult to categorize and classify them. The various salient features of ED causes the patient affected with ED to develop a social stigma. This disturbs both the normal body functions and oral functions of the patients. The main management of ED is a swift diagnosis and prosthetic rehabilitation through a multidisciplinary approach and therapy. Pedodontists have an important part in maintaining the outward appearance of the child and to maintain the Child's psychology.

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