CASE REPORT

An unusual case of ectodermal dysplasia: combating senile features at an early age

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SUMMARY

Ectodermal dysplasia (ED) refers to a group of inherited diseases that have developmental defects in at least two major structures derived from the ectoderm, that is, hair, teeth, nails and sweat glands. Although more than 192 distinct disorders have been described, the most common is X-linked recessive hypohidrotic ED (Christ-Siemens-Touraine syndrome). Since such patients usually presents with missing teeth, dentists are usually the first person to diagnose such cases. Diagnosis of such cases is important because absence of sweat glands can lead to hyperthermia which can be life-threatening if proper care is not taken. Through this manuscript, we report a case of anhidrotic ED affecting deciduous and permanent dentition, which is rare.

BACKGROUND

The ectodermal dysplasia (ED) encompasses a colossal, heterogeneous group of hereditary disorders that are defined primarily by defects in the development of two or more tissues derived from embryonic ectoderm. The most common type of ED is Christ-Siemens-Touraine syndrome, which usually manifest itself as a triad affecting hairs (atrachosis or hypotrichosis), teeth (anodontia or oligodontia) and sweat glands (anhidrotic or hypohidrotic).¹ Though ED affects various tissues, oligodontia is one of the principal features. But involvement of deciduous and permanent dentition is atypical. We document here an infrequent case of anhidrotic ED concerning primary and permanent dentition with senile facial features.

CASE PRESENTATION

A male patient aged 7 years (figure 1A,B) reported with missing teeth in his upper and lower jaws since childhood. The patient also had difficulty in eating and chewing food. The patient also reported with frequent urination and absence of sweat production. There were no other associated symptoms. On extraoral examination, hypotrichosis (sparse hair) was present (figure 1C), eyebrows and eyelashes were rudimentary. Skin appeared to be dry (figure 1D), rough and scaly, and skin over the nose appeared to be shiny. Nails were normal (figure 1E). Lips were everted and ears appeared to be obliquely placed making them prominent. On intraoral examination, peg-shaped deciduous teeth i.r.t 53 and 63 were present with absence of all other primary and permanent teeth (figure 1F). Atrophy of alveolar ridge was evident. On the basis of clinical examination and history given by the

patient, a provisional diagnosis of anhidrotic ED was given.

INVESTIGATIONS

Panoramic radiograph

Panoramic radiograph revealed apical root resorption of deciduous canines i.r.t 53 and 63, developing permanent tooth buds i.r.t 13 and 23 with open apex, missing other primary and permanent teeth (figure 1G).

Skin biopsy

The patient was also referred to dermatology department in a hospital where he was subjected to skin biopsy which on histopathological examination revealed absence of sweat glands.

DIFFERENTIAL DIAGNOSIS

Non-specific partial anodontia, syndromic partial anodontia and trichodonto-osseous syndrome.

TREATMENT

The patient's parents were educated about the condition and were motivated for removable prosthetic treatment for missing teeth which have to be changed continuously until patient's growth ceases.

They were also educated about the absence of the sweat glands and its related hyperthermia, and thus were warned against playing of child in sunlight and other activities that could increase heat production and were educated about keeping cooler environment.

OUTCOME AND FOLLOW-UP

The patient was kept under follow-up for 2 months, the period which the patient had yet not started with prosthetic treatment due to time constraints associated with examination at school.

DISCUSSION

It was in the year 1929 that the term 'ectodermal dysplasia' was coined by Weech, although first published case was reported almost as early as in 1848 by Thurnam.^{1 2} The incidence of ED is approximated to be 0.7 to 1/100 000 births. Freire-Maia and Pinheiro provided an extensive review and thus proposed a provisional classification based on which ectodermal derivatives are affected. According to their classification, '1' indicates hair dysplasia, '2' dental dysplasia, '3' nail dysplasia and '4' sweat gland dysplasia. Recently, many other classifications have been provided; in 2001, Priolo and Laganà reclassified the EDs into two main functional groups: (1) defects in developmental regulation/epithelial-mesenchymal

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Figure 1 (A) Extraoral front view of patient. (B) Extraoral profile view of patient. (C) Sparse hair over scalp. (D) Dry and rough skin. (E) Normal nails. (F) Intraoral photograph depicting missing teeth. (G) Panoramic radiograph depicting missing teeth and tooth buds.

interaction and (2) defects in cytoskeleton maintenance and cell stability. In 2003, Lamartine reclassified the EDs into four functional groups based on the underlying pathophysiological defect. The various genetic forms of ED can be X-linked recessive, autosomal recessive or autosomal dominant. The aetiology is related to either mutation or deletion of certain genes located on different chromosomes. The most commonly implicated genes are EDA (ectodysplasin), EDAR (ectodysplasin receptor) and EDARADD (ectodysplasin receptor associated death domain).³ Since in our case the patient was not subjected to genetic testing, we could not point out the exact gene.

Persons afflicted with ED manifest with deformities in various ectodermal structures. Although noticeable manifestations of the disorder are not clinically evident in most of the newborns, they usually become apparent during infancy or childhood. A family history of similar clinical features is helpful, thus emphasising the importance of case history recording. The facial features of patients with ED are usually referred to as 'old man facies', thus features of senility are obvious at an early age as seen in our case also. The archetypal triad of ED involves teeth, sweat glands and hairs; the same was seen in our case also. Though oligodontia is one of the primary features and it can affect deciduous and permanent dentition,⁴ but still involvement of both the dentitions is rare.³⁻⁵ The presence of canines even in absence of other teeth is an important feature. In our case also, the primary and permanent dentition was affected, and there is presence of canines in both the dentitions.

The various investigative procedures, which aid in diagnosis are radiographs, sweat pore counts that can be easily performed using starch iodine test, pilocarpine iontophoresis, skin biopsy and genetic testing.⁶ In our case, radiographs and skin biopsy were performed.

On the basis of history given by the patient, clinical features, radiographic features and histopathological examination, we came to final diagnosis of anhidrotic ED.

Regarding treatment, multidisciplinary approach is required consisting of pedodontist, prosthodontist, orthodontist, oral surgeon, speech therapist and psychiatrist.⁷⁻⁹ Since absence of teeth leads to atrophy of alveolar ridge and reduced vertical dimension which greatly affects the appearance of face, prosthetic rehabilitation is of utmost importance. And due to continuous growth of child, prosthesis should be changed at required intervals.

Owing to involvement of salivary gland, there could be varying degrees of xerostomia. There are various treatment modalities available for treating it such as replacement therapy, for example, artificial saliva; stimulation therapy, for example, pilocarpine; and disease-modifying drugs, for example, rituximab and epratuzumab.

For counteracting reduced sweating, the patient should be encouraged to maintain cooler environment around him. A similar protocol was followed in our case.

Owing to early senile facial features, patients with such disorders and their parents have to be psychologically motivated at regular intervals to overcome any social stigma associated with ED.

The most important thing is prevention of such cases which can be achieved through genetic counselling and thus consanguineous marriages should be avoided.

Thus, we can see that dentist play an important role in the diagnosis of ED, and are crucial members of the multidisciplinary treatment team.

Learning points

- ▶ Genetic counselling is necessary for preventing such X-linked hereditary disorders.
- ▶ Dentists play a vital role in diagnosing such cases, and thus preventing complications.
- ▶ People should be made aware of such ill effects of consanguineous marriages, and thus such practices should be avoided.
- ▶ Treatment of ectodermal dysplasia involves multidisciplinary approach which involves specialists from different specialties.
- ▶ Psychological motivation is required for patients to combat social stigma associated with early senility.

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