Unusual presentation of more common disease/injury

Prosthodontic management of patients with Christ–Siemens–Touraine syndrome

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Christ–Siemens–Touraine syndrome also known as anhidrotic ectodermal dysplasia. Ectodermal dysplasia is a hereditary disorder characterised by developmental dystrophies of ectodermal derivatives. It is characterised by triad of signs comprising sparse hair, abnormal or missing teeth and inability to sweat. Anodontia or hypodontia is the most striking dental manifestation. In severe hypodontia, there is lack of alveolar development with consequent protrusion and eversion of the lips. The case of three children with anhidrotic ectodermal dysplasia with partial anodontia is presented. Dental, oral and physical features were taken into consideration in diagnosis and different treatment modalities for this patient. Clinical management consisted of removable partial prosthesis in maxillary arch and complete denture prosthesis in mandibular arch. The main aim of the treatment was to improve psychological development and to promote better functioning of the stomatognathic system.

BACKGROUND

‘Every child comes with a message that God is not yet discouraged of man’ – Tagore. The most beautiful creation is the innocent smile blossoming on the face of a young child. Eric Erickson has coined a wonderful term in child psychology to illustrate the child’s behaviour ‘d’terrible two’s’ – Little devil and little angel. Although one side of coin of God’s creation can be written with exclamatory mark, the other side, we have to look for, which make us to raise queries? One such query is a condition thrown with poignant smile is ectodermal dysplasia (ED).¹

ED is a congenital, diffuse and non-progressive disease, first described by Thurman, is a hereditary disorder occurring as a consequence of disturbances in the ectoderm of the developing embryo.² More than 60 mutations have been identified in EDA, including nucleotide substitutions (mis sense, nonsense and splicing), small deletions and insertions, gross deletions mainly EDAR and EDARADD genes cause hypohydrotic ectodermal dysplasia. The triad of nail dystrophy (onychodysplasia), alopecia or hypotrichosis (scanty, fine light hair on the scalp and eyebrows) and palmoplantar hyperkeratosis is usually accompanied by a lack of sweat glands (hypohidrosis) and a partial or complete absence of primary and/or permanent dentition. Their incidence is estimated to be 1/100000 births affects male. Differential diagnostic problem is the distinction of autosomal recessive form of HED from X linked HED. AR–HED is less common than X linked. X linked recessive disorders affect males and is inherited through female carriers. This carrier incidence is 17.3 in 100 000 women.³ In children with ED, the appearance of the teeth is extremely important because it can affect the patients self esteem which create challenges for the pediatric dentist. A complete oral rehabilitation of these patients is of great value from the functional stand point as well as for physiologic and psychosocial reasons.^{4 5} The present reports describes the complete rehabilitation of a young child with a full blown condition of ED.

The most frequent prosthetic treatment for the dental management of ectodermal dysplasia is removable

Figure 1 Clinical examination reveals prominent forehead, saddle nose, diminished lower facial height, sparse scalp hair, missing eyelashes and eyebrows and protuberant lips.

Figure 2 Prominent forehead, diminished lower facial height, and protuberant lips.

prosthodontics. Since alveolar bone development is dependent on the presence of teeth, children with ectodermal dysplasia have little or no bone ridge upon which to construct dentures; therefore, restoring function and appearance is more challenging than usual. Follow-up by a multi-disciplinary team involving paediatric dentistry, orthodontics, prosthodontics and oral-maxillofacial surgery specialists is advocated to be the most appropriate approach in such cases.⁶ In these case reports; it is aimed to describe the prosthetic rehabilitation of three young boys with anhidrotic ectodermal dysplasia associated with anodontia.

CASE PRESENTATION

Case report 1

A 10-year-old boy reported to the Department of Paediatric and Preventive Dentistry, with the complaint of absence of teeth, inability to eat and difficulty in speech. The general medical history and family history was non-contributory. The child exhibited the classical features of ectodermal dysplasia: anodontia, hypohidrosis, hypotrichosis, prominent forehead, saddle nose, diminished lower facial height, sparse scalp hair, missing eyelashes and eyebrows, characteristic feature of pinna and protuberant lips (figures 1 and 2).

The intraoral examination revealed absence of multiple primary teeth, as a result of which the edentulous alveolar ridges were deficient in both height and width. The mandibular ridge was of the knife edge variety. The

Figure 3 Intraoral examination reveals tapered (peg-shaped) and widely spaced mandibular canines and root stumps with deciduous incisors along with maxillary permanent first molars.

Figure 4 Intraoral view showing extracted root stumps.

oral mucosa had a slight dry appearance and the tongue seemed relatively large. A clinical examination revealed few permanent teeth present in the maxilla and mandible. The existing teeth were extremely tapered (peg-shaped) and widely spaced mandibular canines and root stumps with deciduous incisors along with maxillary permanent first molars (figure 3).

During the physical examination he showed no signs of mental problems and his vital signs were normal. First, extraction of root stumps (figure 4) was done followed by removable upper and lower partial denture was planned for the patient, considering the patient's need for mastication, esthetics, speech and overall psychological development. Preliminary impressions were made with irreversible hydrocolloid, and then custom trays were prepared for functional impression. Acrylic bases with wax rims were made on the master casts in order to establish maxilla mandibular relations. After making the maxilla mandibular records, the casts were mounted in an articulator (figure 5). Rather than primary tooth forms, permanent tooth forms were selected in order to provide better static and dynamic occlusion. Primary tooth forms were not adequate to fulfill the ideal vertical dimensions because the patient is in the transition period from mixed to permanent dentition. After the final insertion, routine hygiene instructions for the dentures were given to both the child and his parents (figures 6

Figure 5 Maxillary and mandibular records on articulator.

Figure 6 Removable partial dentures.

and 7). The patient was advised to maintain a soft diet for the first few days, and to remove the dentures at night to promote healing of the oral tissue. Continued follow-up is suggested for modification or replacement of the dentures to fit the patient's developing maxilla and mandible.

Case report 2

A 9-year-old boy presented with missing teeth to the Department of Pedodontics and Preventive Dentistry. He came with his father who gave history of uneruption of deciduous teeth except four teeth in the upper jaw and four in lower jaw. On general examination, his hair was fine, sparsely distributed, thinly covering the scalp and the skin was found to be fine smooth and dry, and nails revealed no abnormality. He had characteristic facies with prominence of frontal and temporal regions of the scalp, saddle nose, large ears (megalo – pinna), protuberant lips and depressed central portion of face (figure 8).

Cutaneous examination revealed thin sparsely distributed hair over the scalp and eyebrows. There was hyper pigmentation on the nose and around the eyes. There was no history of consanguinity among parents and none of the family members had history of missing teeth. The parents gave a medical history of repeated respiratory tract

Figure 7 Patient with dentures.

infections and also about heat intolerance at elevated temperatures.

Intraoral examination revealed bone atrophy of maxillary and mandibular alveolar ridges. Maxillary as well as mandibular arch had two short, conical, pointed right and left primary canines and first molars (figures 9 and 10).

In order to improve the appearance, mastication, and speech, the child was provided with removable partial dentures in upper arch and complete dentures in lower arch. Cast partial dentures were avoided because of the changes in oral structures due to growth of child. So, it was decided to change the maxillary and mandibular partial dentures every 6 months serving as interim prosthesis. Other objectives of treatment regarding the dental condition were the preservation of bone, early development of normal mechanisms of chewing, speaking and swallowing, the establishment of normal facial characteristics and smile; the development of normal emotional and psychological profile and the fitting of a functional prosthesis with adequate retention, stability, and support.

Dentures were fabricated following the same procedures described for the first case (figure 11). Primary tooth forms were used and the occlusal vertical dimension was increased by 1 mm in order to improve the balance of both the dentures and facial profile. After the initial insertion, oral hygiene instructions for the dentures were given to the parents. Initially, the patient had some difficulty in accepting the dentures and was unable to keep them in his mouth due to his young age. After a few months, he was fully adapted to using the dentures, and his parents reported that he was able to eat; in addition, his speech improved and he was quite happy with the dentures. Further follow-up have taken place every 3 months. Further adjustments

Figure 8 Characteristic facies with prominence of frontal and temporal regions of the scalp, saddle nose, large ears (megalo-pinna), protuberant lips and depressed central portion of face.

Figure 9 Maxillary arch showing two short, conical, pointed right and left primary canines and first molars.

were made to eliminate interferences at recall appointments; future treatment will include relining, rebasing, or remaking the dentures in order to accommodate growth and development.

Case report 3

A 9-year-old boy reported with the complaint of lack of teeth. He exhibited the classical feature of ED: hypodontia, hypohidrosis, hypotrichosis, saddle nose and everted lips. Intraoral examination reveals dry and sticky mucosa, conical shaped central, lateral incisors and Mesio dens (figures 12 and 13). Conventional over dentures were treatment of choice. Mesiodens were extracted under local anaesthesia and apexification was done for all permanent maxillary teeth as apices were open.

Figure 10 Maxillary as well as mandibular arch had two short, conical, pointed right and left primary canines and first molars.

Figure 11 Patient with dentures.

After 11/2 years, when open apex had closed completely, the conventional root canal treatment carried out. Maxillary teeth were reduced to use bare root abutment for retention of overdenture. Similarly mandibular teeth were reduced close to gingival to use as bare root abutments. Root preparation was done by removing Gutta percha from cervical third of canal. Wax impression for post was made for maxillary teeth and rubber base impression was taken. Wax pattern was tried on the cast. All coping patterns were invested, casted, polished and cemented on root abutments of maxillary teeth (figure 14).

In mandibular arch, an overdenture with intracoronal attachment like zest attachment was planned in which the female component was processed in denture base providing the retention when overdenture is inserted (figure 15). Root preparation seating of male attachment was done, then the female part was snapped in position over male attachment. Wax rims were prepared for maxillary and mandibular denture base, proper vertical height was established. A trial denture base was made for auto polymerising acrylic resin and selected denture teeth were adjusted. After trying the denture base was processed in heat cure acrylic resin (figure 16). After placement of denture, oral hygiene instructions were given to patient.⁴

INVESTIGATIONS

In all the three cases, sweat pore count test is done in which starch-iodine powder applied to palmar or dorsal skin. In unaffected persons, sweating turns the powder to

Figure 12 Intraoral view of conical shaped central, lateral incisors and lower incisor.

Figure 14 Coping on maxillary teeth.

Figure 13 OPG of patient.

Figure 15 ZEST attachments on lower teeth.

deep purple, allowing visualisation of sweat pores. Sweat pores are poorly visualised in affected children.

Blaschko lines- streaky areas of hypohidrosis that follow blaschko lines are observed upon starch-iodine staining, which demonstrates a mosaic pattern of areas of normal numbers of sweat pores alternating with areas of absent pores.

Histologic findings showed reduction in the number of sweat glands, hair follicles and sebaceous glands associated with the different EDs. In EDA, the epidermis is thin and flattened. Sweat glands are few or poorly developed or are very rudimentary.²

DISCUSSION

Ectodermal dysplasias are a group of hereditary disorders involving an absence or deficiency of tissues and structures derived from the embryonic ectoderm. Clinical signs include trichodysplasia (abnormal hair) in 91% of cases, tooth agenesis in 80%, onychodysplasia (abnormal nails) in 75% and dyshidrosis (abnormal sweat glands) in 42%. EDs are rare and incidence has been estimated at 1 in 10 000 and 1 in 100 000 births. ED follows all possible modes of Mendelian inheritance, that is, autosomal dominant or recessive or X linked dominant or recessive and there have also been sporadic cases reported. Now

there are over 170 different, clinically distinct subtypes. Pathogenesis is thought to involve an altered epithelium-mesenchymal interaction. About 30 causative genes have been isolated. These are involved in intracellular communication and signalling, cell development, cell survival and differentiation among other things.³ Recent cloning of genes has led to identification of a novel trans membrane protein – Ectodysplasin and receptor ‘Edar’. This TNF ligand and receptor have a developmental regulatory role and are associated with epithelial mesenchymal interactions and signalling pathways that regulate ectodermal appendages formation. Ectodysplasin is first recognised by Laurikkala.⁷

The anhidrotic form is commonly diagnosed in childhood, subsequent to a feverish episode of unknown origin. Palms and soles are hyperkeratotic; pseudorhages are present around the eyes. Atopic eczema is common, especially in flexures during early childhood. In a newborn, a ‘plastic wrap’ appearance of the skin is there. Partial or complete absence of sweat glands is responsible for the inability to perspire and accounts for intolerance to heat and hyperthermia. Sparse blonde hair including reduced density of eyebrow and eyelash hair is common.

In the present cases, anhidrotic ectodermal dysplasia was suspected due to history of frequent episodes of high fever and senile appearance of the child. There is no

Figure 16 Postoperative intraoral photograph.

specific treatment to minimize the relative consequences of the anomalies of fingernails, skin, and hair. Regarding the dental treatment instituted, it was possible to improve the physical condition of the child, as well as the esthetics, chewing, and phonetic functions. Treatment generally includes a removable or fixed partial denture, a complete overdenture prosthesis (overlying affected teeth when the vertical dimensions of occlusion permits), and an implant retained prosthesis when indicated. These treatment approaches can be used individually or in combination to provide an optimal result.⁶

Early implant placement in a growing child may cause cosmetic problems because the implants act like ankylosed teeth. With the vertical development of the jaws, implant over-structures may not meet with the teeth of the opposite jaw, and may result in prosthetic infraocclusion. Therefore, the use of implants in young children should be considered carefully. In both of the above cases, implant therapy was not the treatment choice due to ongoing growth and development and insufficient alveolar bone support.

Guckes, McCarthy⁸ claimed that implants placed in ED patients younger than 18 years of age have a high risk of failure. Kearns and Sharma⁹ reported submergence of implants placed in two young patients (5 and 7 years) who later required placement of longer abutments and prosthesis revisions to accommodate the change in implant positions due to growth. The disadvantage of changing abutment configurations and replacement of prosthesis should be weighed against the psychological benefits of the patient.

Therefore, the use of implants in young children should be considered carefully, taking into account the above-mentioned issues, especially dental and skeleton maturation as compared to the chronologic age of the patient. In all of the above cases, implant therapy was not the treatment of choice due to ongoing growth and development and insufficient alveolar bone support.^{10 11}

Numerous clinical reports have demonstrated the importance of prosthetic dental treatment in ED patients for physiologic and psychosocial reasons. Serial sets of dentures may be required as a child matures. Shaw describes the need for these to restore vertical dimension and prevent them undesirable protruding lips secondary to over closure, thereby improving the profile. Ideally, a restored dentition should be

in place before the child attends primary school as oral rehabilitation has a major psychological impact on the patient's self esteem and facilitating social acceptance.^{12 13}

In third case, overdentures are used as when there are teeth present for support, over dentures are a desirable treatment options for these patients. Cram provided an excellent overview of the advantages of conventional overdenture. it preserves alveolar bone. If the overdenture is fabricated, retention can be augmented by various attachments anchored to all teeth.¹⁴ Preservation of alveolar bone is imperative in individuals with ED because they must depend on alveolar ridges for prostheses support from an early age.

Dental prostheses may also improve the tone of the muscles of mastication and may compensate for the reduced vertical dimension. Difficulty with mastication has been referred to as a major problem arising from loss of teeth. As seen in all the cases, the facial profile and expression improved significantly with complete dentures; in addition, mastication and dietary patterns also improved.¹⁴

All the cases are male patients as the X linked recessive form of HED is more common which affects males and females are only carriers. These case reports highlight the importance of accurate treatment planning as well as the influence of anodontia on the diagnosis of ectodermal dysplasia. Since the oral rehabilitation of these cases is often difficult, particularly in pediatric patients, treatment should be administered by a multidisciplinary team involving pediatric dentistry, orthodontics, prosthodontics, and oral-maxillofacial surgery.¹⁵

Learning points

- ▶ The HEDs is a genetic disease. Genetic testing is available. HED is diagnosed based upon physical features but genetic testing is used to confirm the diagnosis. Mainly caused by genetic defects in ectodysplasin signal transduction pathways.
- ▶ The diagnosis is usually made with the identification of hypotrichosis, characteristic facial features, hypohidrosis (and more rarely anhidrosis), and teeth abnormalities.
- ▶ Ideally, a restored dentition should be in place before the child attends primary school as oral rehabilitation has a major psychological impact on the patient's self esteem and facilitating social acceptance.
- ▶ The treatment guidelines of the National Foundation for ED state that implants are only recommended for the anterior portion of the mandible in children older than school age (7 years or older). The disadvantage of changing abutment configurations and replacement of prosthesis should be weighed against the psychological benefits of the patient.

Competing interests None.

Patient consent Obtained.

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