

Anhidrotic Ectodermal Dysplasia: Report of two Cases and Review of Literature

Abstract

Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures. The X-linked recessive ED (Christ-Siemens-Touraine syndrome) is the most common disorder (80% of EDs); it affects males and is inherited through female carriers. It is characterized by the triad of signs comprising sparse hair (atrachosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis). The lack of teeth and the special appearance were reported to be major concerns. The incidence in male is estimated at 1 in 100,000 births, the carriers-incidence is probably around 17.3 in 100,000 women. Most patients with EDA have a normal life expectancy and normal intelligence. However, the lack of sweat glands may lead to hyperthermia, followed by brain damage or death in early infancy, if unrecognized. Thus an early diagnosis is important. Families with EDA should therefore be offered genetic counselling. Currently the genes and gene products are defined, but the function of the proteins is not fully known. Once the exact function is known, we can understand the embryogenesis and morphogenesis of the ectodermal structures. It is quite possible that these kinds of observations of gene function and interaction may form the basis of new therapeutic methods in the future.

For the patients as well as the dentists tooth agenesis and its secondary effects on growth and development of the jaws is often the most significant clinical and therapeutical problem. The course of the treatment is to restore the function and the aesthetics of the teeth, normalise the vertical dimension and support the facial soft tissues.

We report two cases of ectodermal dysplasia. Both of them are anhidrotic ectodermal dysplasia type.

Key Words

Ectodermal Dysplasia; Tooth Agenesis; Christ-Siemens-Touraine Syndrome, Atrichosis, Hypotrichosis, Anodontia, Hypodontia, Anhidrosis, Hypohidrosis

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INTRODUCTION

Ectodermal dysplasia (ED) is a hereditary disorder that can affect several ectodermal structures. These structures may include: skin, hair, nails, teeth, nerve cells, sweat glands, parts of the eye and ear, and parts of other organs. Thurman published the first report of a patient with ED in 1848, but the term was not coined until 1929 by Weech.^{1,2,3} ED syndromes have been described as a group of disorders of morphogenesis displaying two or more of the following signs and symptoms.^{1,2,3}

1. Trichondysplasia (abnormal hair)
2. Abnormal dentition
3. Onchondysplasia (abnormal nails)
4. Dyshidrosis (abnormal or missing sweat glands)

Lamartine in 2003 has described various well defined ectodermal dysplasia as Hypohidrotic (anhidrotic), Hidrotic (Clouston's syndrome), Ectrodactyly-

ectodermal dysplasia-cleft syndrome (EEC), Rapp-Hodgkin Syndrome, Hay-Wells syndrome or ankyloblepharonectodermal dysplasia. Usually the ectodermal dysplasia is divided into two types based on the number and function of sweat glands as mentioned below⁴:

1. Hypohidrotic (anhidrotic) Ectodermal Dysplasia (Christ-Siemens-Touraine Syndrome)
2. Hidrotic Ectodermal Dysplasia (Clouston Syndrome)

Hypohidrotic (anhidrotic) Ectodermal Dysplasia (Christ-Siemens-Touraine Syndrome)

Hypohidrotic (anhidrotic) Ectodermal Dysplasia (Christ-Siemens-Touraine Syndrome)

The most reported ED syndrome is x-linked hypohidrotic (anhidrotic) ED (Christ-Siemens-Touraine syndrome) which affects

one to seven individuals per 10,000 with males afflicted more frequently than, females^{1,2,5,6-9}.

Patients generally have prominent supraorbital ridges, frontal bossing, and a saddle nose. The maxilla may be under developed and the lips are thick and prominent. The nose may appear pinched and the alar nasi appears hypoplastic. The patient may resemble like an old edentulous person^{8,9}.

The skin is usually dry, scaly, and easily irritated as a result of poorly developed or absent sebaceous glands. Sweat glands may also be absent or few in number or non-functioning which may result in increased body temperature. Scalp hair may be absent, sparse, very fine pigmented, or abnormal in texture. Eyebrows, eye lashes, and other body hair may also be sparse or absent. When hairs are present, they may be fragile, dry, and generally disorderly because of the lack of sebaceous glands. Finger and toe

nails are usually normal⁹. Orofacial characteristics of this syndrome include anodontia or hypodontia, hypoplastic conical teeth, underdevelopment of the alveolar ridges, frontal bossing, a depressed nasal bridge, protuberant lips, and hypotrichosis^{10,11}. Teeth in the permanent dentition are frequently small, conical, tapered (peg shaped), and widely spaced. Lack of alveolar growth may be associated with this condition and frequently results in increased inter-occlusal distance which allows optimum artificial tooth placement¹². Patients may present with a marked mandibular protrusion. Depending on the severity of the condition, various prosthodontic treatments are available to improve appearance, mastication, and speech¹³.

Children usually have a normal mentality and life expectancy, and their facial appearance warrants professional concern for their emotional well being and social progress^{8,14}. Tanner¹⁵ states ectodermal dysplasia with an abnormal appearance may affect normal social and psychological development in young patients. Functional needs also must be considered since the difficulty these children experience in masticating may cause nutritional problems^{8,14}. Therefore, dental care for such patient is important.

Hidrotic Ectodermal Dysplasia (Clouston Syndrome)

Here the clinical features include nail dystrophy, hair defects and palmo-plantar dyskeratoris. The patients have normal facies, normal sweating and no specific defect is seen.

Anhidrotic ectodermal dysplasia (EDA) is a rare disorder commonly transmitted as an X-linked recessive disorder. However, rarely autosomal recessive and autosomal dominant inheritance have also been seen. This syndrome manifests as a triad of defects partial or complete absence of sweat glands anomalous dentition and hypotrichosis.¹⁶ In view of the rarity of this entity, we report two cases of anhidrotic ectodermal dysplasia.

Case reports:

Case 1:

A 12-year old male child reported to our hospital with complaint of lack of esthetics and difficulty in mastication. Patient also complained of fever on and off. Fever used to be high grade and more so during summer and would subside with medication. The patient had history of parchment like skin sheet at birth

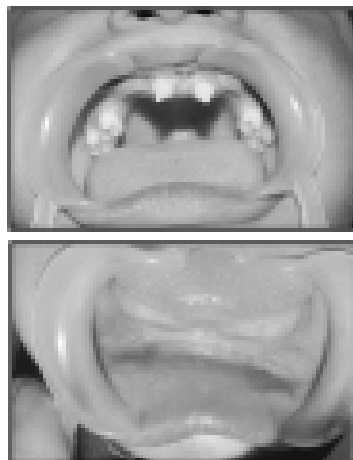
Clinical examination of the patient revealed a fine, sparse scalp hair on scalp while hair were absent on eyebrows, and eyelashes. There was frontal bossing, depressed nasal bridge and malar hypoplasia. Wrinkled hypo-pigmented skin around periorbital region was noted. Skin was rough and dry with no sweat on it (Figure 1).

(Fig1)



The teeth were reduced in number and conical in shape in upper jaw and anodontia in lower jaw (Figure 2), the alveolar ridges were underdeveloped, and the lips were prominent.

Fig 2: Oral examination of Case No. 1, showing teeth were reduced in number and conical in shape in upper jaw and anodontia in lower jaw.



The treatment option preferred was of a removable partial denture for the missing maxillary teeth and complete denture for edentulous mandibular arch. The peg shaped teeth were modified with composite resin which helped in enhanced retention of upper denture. The dentures were delivered and the patient was instructed on the maintenance of oral hygiene and dentures. Recall appointments were done for adjustments. (Figure 3)

Figure 3: Post-operative picture of Case No. 1

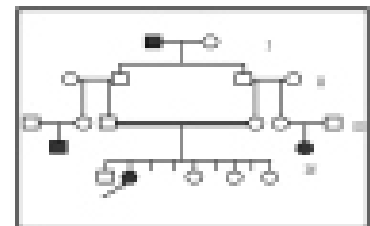


Case 2:

An 11 year old young female, presented to our hospital for evaluation of teeth abnormalities. She had a history of recurrent high fevers during infancy, and the patient's mother reported that her daughter is not able to sweat she had to apply some precautions to protect her from overheating during warm weather and physical exertion.

Other family members (including first degree cousins) were affected by hypohidrosis and dental abnormalities. Her mother's medical history included four death births. Her family pedigree is represented at Figure 4.

Figure 4: Pedigree of Case No.2



Clinical examination of the patient revealed a fine, sparse scalp hair, eyebrows, and eyelashes. The patient's skin was smooth and dry. Mild periorbital and perioral hyperpigmentation were evident. Additional facial features included frontal bossing, prominent supraorbital ridges, and

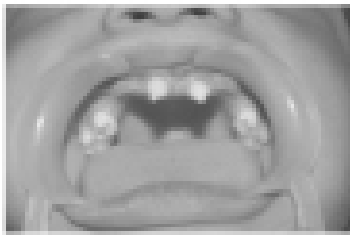
midfacial hypoplasia with a depressed nasal root and bridge. (Figure 5)

Figure 5: Clinical picture of Case No. 2, revealing fine, sparse scalp hair, eyebrows, and eyelashes.



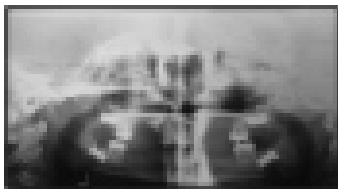
The patient had oligodontia. The teeth were reduced in number and conical in shape. (Figure 6)

Figure 6: Oral examination of Case No. 2, showing oligodontia. The teeth were reduced in number and conical in shape.



OPG revealed complete absence of some permanent tooth germs and underdeveloped maxillary and mandibular alveolar ridges. (Figure 7)

Fig 7: OPG of Case No. 2, showing complete absence of some permanent tooth germs and underdeveloped maxillary and mandibular alveolar ridges.



The treatment option preferred was of a removable partial denture for the missing teeth. The peg shaped teeth were modified with composite resin which helped in enhanced retention of upper denture.

Discussion and review of literature:

Disease name and synonyms

- Ectodermal dysplasia anhidrotic (EDA)
- Anhidrotic ectodermal dysplasia
- Hypohidrotic ectodermal dysplasia (HED)
- Christ-Siemens-Touraine syndrome

Definition

Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures, such as hypodontia or anodontia.^{1,2,3}

More than 190 clinically and genetically distinct hereditary ectodermal dysplasias have been catalogued¹⁷.

Anhidrotic (hypohidrotic) ectodermal dysplasia (EDA) is the most common ED (80%); it is characterized by hypoplasia of hair, teeth and sweat glands. Since there is not a complete lack of sweat glands the term hypohidrotic is more adequate than anhidrotic. The anhidrotic (hypohidrotic) ectodermal dysplasia is often inherited as an X-linked disorder (XLEDA /Christ-Siemens-Touraine syndrome).¹⁸ X-linked anhidrotic ED was first described in 1848 by Thurnam and later in the 19th century by Darwin. Both autosomal dominant and autosomal recessive inheritance of clinically similar conditions have now been demonstrated and the molecular defects defined¹⁹.

Genetics of EDA

All the ectodermal dysplasias appear to be genetic in etiology. Of more than 190 EDs described, the molecular basis has been elucidated for more than 30 of them²⁰. A little less than a hundred years after Darwin's discussion, the gene for EDA was the first X-chromosomal gene whose map position was suggested, based on the occurrence of an X; auto some translocation in a female patient ("Anly") with the disease phenotype. Later linkage studies confirmed the suggested position of the gene at Xq12-q13.1 (XLHED-gene)²¹. The localisation of the gene has permitted more accurate diagnostics of the disorder, both in carrier females and in prenatal screening. The discoveries of disease genes and the identification of mutations in patients represent great progress in biomedical science. In order to help patients suffering from the diseases it is obviously not enough that the gene defects are known. It is necessary to understand the context in which genes function and the details of the biological processes where they are involved.

The recent cloning of the gene has led to

the identification of a novel transmembrane protein "ectodysplasin" (TNF family ligand) and receptor "edar" (TNF receptor). This TNF ligand and receptor have a developmental regulatory role and are tightly associated with epithelial-mesenchymal interactions and signaling pathways that regulate ectodermal appendage formation and organogenesis during the initiation of development²².

The next step in research, the analysis of the function of the genes and the effects of gene mutations will be a challenge for researchers for many decades to come, and this can be expected to result in the development of new therapeutic methods. It is quite possible that these kinds of observations of gene function and interaction may form the basis of new therapeutic methods in the future.

Epidemiology

The prevalence of EDA is unknown; however, the incidence in male is estimated at 1 in 100,000 births although the condition is usually overlooked in infants²³. This X-linked recessive disorder affects males and is inherited through female carriers. This carriers-incidence is probably 17.3 in 100,000 women. The autosomal dominant and autosomal recessive inheritance EDA is an extremely rare condition.²⁴

Clinical Features

EDA is characterized by the triad of signs comprising sparse hair (atrachosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis). The lack of teeth and the special appearance were reported to be major concerns.^{1,2,3}

Most patients with EDA have a normal life expectancy and normal intelligence. However, the lack of sweat glands may lead to hyperthermia, followed by brain damage or death in early infancy, if unrecognized. Thus an early diagnosis is important. Families with EDA should therefore be offered genetic counselling.^{1,2,3}

Craniofacial structures

Considering how much osseous and dental tissue is missing, these patients have surprisingly normal facial structures. Already in 1936, Tannhauser stated that the characteristic deformity of the cranial bones of all affected patients is such that the resemblance among the patients is bigger than when compared with their own unaffected sib²³.

Clinically, the forehead appears square, with frontal bossing, and there is a prominent

supra-orbital ridge. The nose has a depressed nasal bridge and is called a saddle nose. The midface is depressed and hypoplastic, giving it a "dished-in" appearance. The cheekbones are high and broad, although they appear flat and depressed as well. The chin may be pointed and the lips everted and protuberant²⁵.

In non-treated patients with EDA, craniofacial deviations from the norm increased with advancing age. At age 26 there is a tendency toward a Class III pattern with anterior growth rotation²⁷. Cephalometric analysis and anthropometry studies have been performed. The quantitative findings show reduced facial dimensions, decreased lower facial height, variable pattern in facial widths, the maxilla has been relatively more retruded than the mandible, thenasal alar width and mouth width were significantly smaller²⁸.

This remarkable variability in facial dimensions and harmony found in patients with ED probably corresponds to the different kinds of dysplasia, with different expression of the interested genes²⁹.

Oral structures

Dental defects represent a core clinical feature of many EDs: anodontia, polydontia, dysplastic teeth, retained primary teeth, deficient enamel development (amelogenesis imperfecta), dentine deficiency (dentinogenesis imperfecta), and underdevelopment of the alveolar ridge. In some EDs, the number of erupted teeth is reduced, the spacing of the teeth disrupted, and the periodontium affected. Disturbance of the enamel matrix may occur, making the teeth more susceptible to caries and altering the shape of the teeth, leading to a pegged appearance and additional accessory cusps^{30,31}.

Missing teeth or the delay in teething often starts to worry the parents and leads to the diagnosis of EDA in the second year of life. A dentist should not hesitate to radiographically examine a patient whose teeth have not erupted by the appropriate age in order to exclude EDA. The screening limit for the first tooth to erupt is 15 months³². Besides the delay in teething, the teeth appear radiographically abnormal in shape and structure. The enamel layer is thin and the cervical area of the tooth is constricted. Enamel is rarely hypoplastic²³. If at that stage aplasia of several teeth is seen, the patient should be referred to a geneticist in a paediatric unit with a suspicion of EDA diagnosis. Tooth crowns are small and abnormal in shape. Upper incisors and cuspids are always conical or pointed.

Taurodontism, frequently on the second deciduous molars, is a common feature. Not only the shape is abnormal, but also the number. A severe hypodontia is a universal feature amongst affected individuals. All lacked some deciduous teeth and permanent teeth. The number varies from four to twenty. A few patients have congenital anodontia. There are generally more teeth in the maxilla than in the mandible, although both jaws can be toothless. Most often the lower incisors and premolars are missing, followed by the upper premolars and incisors. The upper cuspids and first upper and lower molars are formed.³³

The edentulous EDA patients do not have any alveolar processes either. In those patients with some natural teeth, there is a striking difference in the intra-oral height and breadth of the bone. In areas where no teeth have developed, the alveolar bone is missing and the bone ridge is very thin in contrast to the normal alveolus surrounding an occasional tooth.³⁴

Many patients complain of dry mucous membranes in mouth and nose. Reduced salivary secretion has been spotted in some EDA patients³⁴. The oral mucous glands should be missing in the lips. Autopsy has also shown absence of mucous glands in the pharynx, larynx, trachea and bronchi. This is in agreement with the susceptibility to respiratory infections. The other salivary glands are not described in literature. Analysis of the saliva has revealed a reduced buffer capacity and an increased number of bacterial cultures. Most affected individuals were susceptible to dental caries³³.

Hair, nails, skin and skintags

Abnormalities of hair are present in all affected individuals. Most individuals have sparse, fine, slowly growing scalp hair. Some individuals are completely bald by their middle teens, whereas other individuals have normal amounts of scalp hair, though they may exhibit an abnormal texture. Sparse eyebrows and eyelashes were always found. Most individuals show decreased body hair, pubic hair, and/or axillary hair, but these features are more variable. However, beard and moustache hair are normal. Electron microscopy of hairs from affected and unaffected individuals showed no abnormalities.

About half of the affected individuals exhibit mild fingernail abnormalities and nail dystrophy. Slow nail growth and split nails are most often reported. A few individuals had a longitudinal ridging, thinning and superficial peeling. Nail

problems occur more frequently in older individuals. This suggests that the nail beds are more susceptible to progressive injury with age. Toenails were generally normal.

Most individuals report dry skin. Affected individuals have a smooth, almost velvety skin texture. The skin of patients also seems to be "thinner" than expected for age. Some infants may have a premature look because of the thin skin. Scaling in the neonate may form a clue to diagnosis.

Almost all affected relatives have decreased sweating, and many show heat intolerance. Some individuals only sweat in certain areas on their body. Common sites of sweating include palms, soles and axillae. Because of the reduced number of sweat glands, there is a danger of hyperthermia. In this way EDA has been associated with sudden infant death. The hyperthermia may also lead to brain damage, and is probably the cause of the rare cases of EDA reported with mental retardation. Subcutaneous fat is often diminished and over one third of the boys have abnormalities of the breast, including absent or accessory nipples. Episodes of hyperpyrexia and severe respiratory infections are life-threatening components in EDA. The delay in teething often leads to the diagnosis in childhood. After the first critical years of life the patients experience a general improvement in health. The lack of teeth is the most important factor in determining the quality of life in these patients, particularly in later life. They all suffer greatly from their abnormal facial and dental appearance.³³

Differential Diagnosis

The differential diagnostic problem is the distinction of the autosomal recessive form of HED (AR-HED) from X-linked HED. AR-HED is considerably less common than XLHED. The clinical features are quite similar in both conditions but due to the different mode of inheritance AR-HED affects both males and females and the heterozygotes have no signs at all. For adequate genetic counselling it is thus important to recognize XLHED heterozygotes by dental examination and sweat tests. To distinguish between AR- and XL-forms, the HED diagnosis should be followed by careful family history for ectodermal manifestations both in male and female and by tests for heterozygote identification. The findings of equally affected males and females, as well as the presence of consanguinity, support an autosomal recessive mode of inheritance³⁶.

Genetic Counselling

Because of the importance of an early diagnosis, families with X-linked EDA should be offered genetic counselling. This implies a calculation of the risk of having an affected child. For genetic counselling the diagnosis of female carriers is very important. The advantage of diagnosing female carriers of EDA includes the optimisation of neonatal and paediatric care for affected male infants, who may be at substantial risk of death in infancy. There is substantial mortality and morbidity in male infants, with about 30% dying in the first two years of life, because of fever or a chest infection. So it is important for carrier females to be aware of their 1/4 risk of having an affected child, for the sake of their child's health. For the calculation of the risk for a particular female to be a carrier, both clinical and pedigree information are necessary.²⁶

Phenotypic tests, however, are still of practical importance in genetic counselling. Signs of EDA are found in about 70% of obligate carriers. In most cases, it is difficult to place much weight upon subjective assessments of scalp hair density, heat intolerance, breast feeding difficulties or the appearance of the eyebrows. So the most significant finding is hypodontia, which is easily recognized. There is also a greater tendency for abnormal crown form and smaller tooth size in carrier females. Without accurate information regarding past extractions, it is impossible to be certain which teeth were congenitally absent. Dental radiographs can provide useful additional information and can be a simple screening test for the carrier status. Certainly, in cases of uncertain diagnosis there is a correlation between the hypodontia and the result of the sweat tests.²⁶ Two methods of assessment of sweating have been developed to identify possible female carriers. The first sweat test is performed on the backs of the carrier female and gives a V-shaped pattern of streaks that refers to the lines of Blaschk. The other method of assessing sweat pores in female carriers is to make counts of the sweat pores along ridges on the fingertips or palms, but there are methodological difficulties.²⁶

The size of the patches is variable and to count sweat pores where they are clearly visible will bias the results obtained. Patches of skin where the ridges appear flattened and the pores reduced maybe caused by domestic labour or by pure quality application. Such considerations have led us to favour the first method of assessing sweating, by performing sweat tests on the whole back of the subjects in search of

patchiness that might follow the lines of Blaschk. The test is regarded as positive if several areas of at least 1 cm diameter are clear of active sweat glands, by an asymmetry between the two sides or if a complete absence of sweating is found. If it gives a clearly abnormal result, then one can be fully confident of its accuracy. The results seem clear cut, but unfortunately the interpretation of the sweat test is inherently subjective. The value of the test is limited due to temporary functional differences of sweat glands (a patchy pattern may also be present in normal individuals). Secondly, the density of sweat pores may vary among normal individuals and a low mean number may lead to false results. Combination of both dental examination and sweat testing enhances clearly the chances of making a correct diagnosis, namely of identifying female carriers of XLHED. In a substantial number of carriers no signs of EDA are found. The calculation of the risk of being a carrier may then be based on the family history and pedigree information. The mapping of a gene for X-linked EDA has given new possibilities for the detection of carriers of XLHED by molecular genetics. The gene locus of EDA has been mapped to proximal Xq and close flanking markers are available. This often allows female carriers in a family to be identified with a high degree of accuracy. Even if DNA diagnosis is feasible, it is not yet available as a service in most centres and will continue to be expensive and thus unattractive for the foreseeable future.³⁷

Prenatal Diagnosis

Prenatal diagnosis of EDA has occasionally been reported. The diagnosis has been made on fetal skin biopsy, obtained by fetoscopy by 20 weeks gestation after determination of the sex of the fetus²³. By histological analysis they demonstrate of either complete lack of, or reduction in, the number of pilosebaceous follicles and by the lack of sweat glands primordia in multiple skin biopsies. The interpretation of the biopsy can be difficult if one does not appreciate the normal regional variability of the distribution of skin appendages of fetal skin, and that sweat gland primordia only begin to develop at around ²⁰ week gestation. This procedure is complicated and implies a considerable risk to the pregnancy.³² The use of linked markers on DNA from chorionic villi has greatly improved the safety of prenatal diagnosis of X-linked EDA. This new method of prenatal diagnosis has major advantages as well as disadvantages. It permits the diagnosis to be

made in the first trimester of pregnancy prior to the development of the affected structures, thereby allowing an early determination of an affected pregnancy. It is technically simpler and may present a lower risk to the pregnancy than the fetoscopy and multiple skin biopsies. Disadvantages to a linkage based indirect analysis include the need for the sampling of previously affected individuals. The counseling of families is more complex since one is dealing with the probabilities of an affected fetus, rather than a more definitive diagnosis based on direct observation. However, these statistical concepts are difficult for many families to comprehend fully.

The identification of mutations in the family will further improve the accuracy of prenatal diagnosis. However, EDA is a disorder which in most cases is associated with a normal life expectancy and a normal intelligence. Prenatal diagnosis will therefore probably not be an option in most families with EDA.²¹

Treatment

Children with ectodermal dysplasia present many and different clinical problems from early childhood through adolescence and also present a life-long need for maintenance care and revisions. For the patients as well as the dentists tooth agenesis and its secondary effects on growth and development of the jaws is often the most significant clinical problem. The course of the treatment is to restore the function and the aesthetics of the teeth, normalise the vertical dimension and support the facial soft tissues. As long as there are no physical, psychological or social burdens, no treatment is necessary. Early placement of partial or full dentures is commonly recommended from the age of two or three years onwards. The denture must be periodically modified as alveolar growth, erupting teeth and rotational jaw growth change both the alveolar, occlusal and basal dimensions. In children, breakage and even loss of removable protheses is quite common. They have also a limited retention and stability, a fastened bone destruction of an already hypoplastic alveolar process and the middle of the upper jaw is covered and so it blocks the sutural growth. For this reason, in young children we prefer a treatment with crowns and bridges.

Prior to that it is generally advantageous to modify the crowns of the existing teeth with direct or indirect composite crowns. When conical anterior teeth are crowned the appearance of the child is very much normalised. Restoration of facial height improves both facial aesthetics and

speech.

Impaired salivary secretion rates constitute an increased risk for dental diseases, namely caries. The fragile oral mucosa may affect the clinical situation as well as the possibilities to wear removable prostheses. It is commonly agreed that osseointegrated implants should be not placed before cessation of growth. Even in young adults, alveolar growth can be remarkable. In EDA boys, the situation is different as no alveolar growth takes place in the totally edentulous areas of the mouth. There are several published cases of early implant placement in toothless EDA patients; the success, however, has been variable²³.

Summary:

Ectodermal dysplasias (EDs) are a heterogeneous group of disorders characterized by the triad of signs comprising sparse hair (atrachosis or hypotrachosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis). The lack of teeth and the special appearance were reported to be major concerns.

Most patients with EDA have a normal life expectancy and normal intelligence. However, the lack of sweat glands may lead to hyperthermia, followed by brain damage or death in early infancy, if unrecognized. Thus an early diagnosis is important. Families with EDA should therefore be offered genetic counselling.

Young patients with EDA need to be evaluated early by a dental professional to determine the oral ramifications of the condition. When indicated, appropriate care needs to be rendered throughout the child's growth cycle to maintain oral functions as well as to address the aesthetic needs of the patient.

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