RESEARCH ARTICLE

ANHIDROTIC HEREDITARY ECTODERMAL DYSPLASIA IN SIBLINGS - A CASE REPORT.

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Abstract:
A hereditary disease characterized by congenital dysplasia of one or more ectodermal structures and other accessory appendages is medically termed as Ectodermal dysplasia. Hypohidrotic or Anhidrotic forms are the most common among this large group of hereditary disorders. Hypohidrosis, Hypotrichosis and Hypodontia constitute the main symptoms of this syndrome. Here with presenting a case study of Anhidrotic ectodermal dysplasia in siblings of 5 and 7 yr. old.

Key Words: - Ectodermal dysplasia, Anodontia, Hidrotic, Anhidrotic, Hypohidrosis, Hypotrichosis.

Introduction:-
Ectodermal dysplasia (ED) is defined by National foundation for Ectodermal dysplasia as a genetic disorder in which there are congenital birth defects of two or more Ectodermal structures (Hickey, 2001). Thurman published the first report of a patient with ED in 1848, but the term was not coined until 1929 by Weech.¹³ Female carriers outnumber affected men but females show little or no signs of the condition (Pigno et al, 1996) the syndrome involve overlapping features, Lamartine in 2003 has described various well defined Ectodermal dysplasia as
Hypohidrotic (Anhidrotic),
Hidrotic (Clouston ‘syndrome),
Ectrodactyly-ectodermal dysplasia-cleft syndrome (EEC),
Rapp- Hodgkin Syndrome,
Hay-Wells syndrome or ankyloblepharon ectodermal dysplasia.

Usually the Ectodermal dysplasia is divided into two types based on the number and function of sweat glands (viera et al, 2007) as mentioned,

Hypohidrotic (Anhidrotic) Ectodermal Dysplasia (Christ-Siemens- Touraine Syndrome)
This disorder is usually inherited as either autosomal dominant / recessive or X-linked recessive trait and the gene locus is X q13-q21. It is commonly X-linked recessive with full expression in males. Female carriers have a minimal expression. In this form sweat glands are absent or significantly decreased. 60-70% of cases usually show manifestations restricted to minimal Hypodontia, Aplastic or Hypoplastic mammary glands, impaired lacrimal gland function, glaucoma and increased susceptibility to allergic disorders such as asthma or eczema.

Typical general mental development, frontal bossing with characteristics reduction in amount of hair (Hypotrichosis), absence of sweat glands (Anhidrosis) resulting in temperature elevation, absence of sebaceous glands (Asteatosis) resulting in dry skin, depressed nasal bridge, protuberant lips, prominent
supra orbital ridges, sunken cheeks, wrinkled hyper pigmented skin around the eyes and large low set ears (Crawford et al, 1991).

The oral manifestations include conical or peg shaped teeth, Hypodontia (partial absence of teeth) or complete Anodontia (complete absence of teeth) of both the deciduous and the permanent dentition with malformation of any teeth that may be present, generalized spacing, underdeveloped alveolar ridges and delayed eruption of permanent teeth. Even when complete Anodontia exists the growth of the jaw is not impaired. This would imply that the development of the jaws except for the alveolar process is not dependent upon the presence of teeth. However, since the alveolar process does not develop in the absence of teeth, there is a reduction in the normal vertical dimension resulting in the protuberant lips. In addition, in oropharynx the defect may be manifested as a high palatal arch or even a cleft palate. The salivary glands including the intraoral accessory glands are sometimes Hypoplastic in this disease. This result in Xerostomia and the protuberant lips may be dry and cracked.

**Hidrotic Ectodermal Dysplasia (Clouston Syndrome):**
Here the clinical features include nail dystrophy, hair defects and palmo-plantar dyskeratosis. The patients have normal facies, normal sweating and no specific defect is seen.

**Case Report:**
A seven year old female child patient reported to the outpatient department with the chief complaint of absence of teeth since toddler age. The parents of the child gave a history of delay in eruption of deciduous as well as permanent teeth, intolerance to heat, and loss of sweating and excessive urination. They also gave a history that the child use to dampen herself with water to get rid of heat and dryness of skin during hot summer months. There was no history of consanguineous marriage between parents, but the grand fraternal parents are the first cousins. On extra oral examination the patient had dry, scaly and wrinkled skin with areas of hyper pigmentations on face around the nose and sparse hair on body and scalp. Hair present were fine in texture and light brownish in color. Facial profile shows typical features like prominent supra orbital ridges, with loss of eye brows and eye lashes. Frontal bossing, flattened nasal bridge were prominent. Patient had no other general complaints other than excessive micturition reflex, as her parents complain of her going to urination very frequently. She is very attentive and exhibited well-coordinated movements.

Intra oral examination reveals normal soft tissue color and texture; there was evidence of partial loss or sparse salivation. Her occlusion was deprived from absence of teeth both in maxillary and Mandibular arches and there is decrease vertical dimension pertaining to typical ‘Old mans’ face appearance. Only permanent teeth present were tooth no 16, 26, and 36. Similar findings were present in her younger sister who had accompanied her. She was five years old and exhibit all the features mentioned above. Based on these findings a working diagnosis of Hereditary Ectodermal Dysplasia (Anhidrotic type) is given. Further investigations are carried out such as radiographs like OPG, lateral Ceph which revealed multiple missing teeth and evidence of unerrupted permanent tooth buds of 17, 27, and 46. As the patients are from poor economic background their further investigative procedures like gene mapping are put on hold. And they were referred to the prosthodontics rehabilitation with support from Chief Minister’s relief fund.

**Discussion:**
The Ectodermal dysplasia’s are a group of inherited disorder that share in common developmental defects involving at least two of the major structure classically hold to derive from the embryonic ectoderms like hair, teeth, nails, sweat glands [3]. The incidence of Ectodermal dysplasia in males is estimated at 1 in 100,000 births [3]. Thurman first described this entity in 1848[4]. More than 192 distinct disorders have been described till date. Frère Maia and Pinheiro published an exhaustive review and classification system for these disorders using a numeric system of 1 (hair), 2 (teeth), 3 (nail), 4 (sweat glands) for characterization [4]. The complete syndrome does not occur in females but females may show dental
defects, sparse hair, reduced sweating and dermatoglyphic abnormalities \(^2\). The major concern seen in these patients is the lack of teeth and the special appearance, as seen in our case \(^1\). The most characteristic findings are the reduced number and abnormal shape of teeth. The delay in eruption of teeth is often the first step in the diagnosis. The men have an easily recognizable facies, also referred to as an old man facies. Some infants have a premature look with scaling of the skin. This can also form a clue to the diagnosis \(^1\). The extra oral features seen in this disorder are frontal bossing with the forehead appearing square in shape, prominent supra orbital ridge, depressed nasal bridge (saddle nose) as seen in our case. The other features include midface hypoplasia, pointed chin and protuberant and everted lips;\(^5\) however these features were not seen in our case. Abnormalities of skin, nails and teeth are also noted \(^1\), which was prominent in our case. Prenatal diagnosis of Ectodermal dysplasia has occasionally been reported which has been diagnosed by fetal skin biopsy, obtained by fetoscopy by 20 weeks gestation \(^6\). The characteristic facies is pathognomonic but may not be recognized in infancy. In partial forms, the pointed conical teeth provide the most valuable indication and should suggest the need for sweat test and a skin biopsy \(^2\). Both autosomal dominant and recessive mode of inheritance has been described.

ILLUSTRATIONS:

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Another variety of Hidrotic Ectodermal dysplasia called as Clouston syndrome is inherited in an autosomal dominant manner, was described by Clouston in 1929 and Lowery et al in 1966, which is found in Canadian families of French descent. This syndrome usually spares the sweat glands \(^8\). The treatment usually comprises of complete restoration of function and aesthetics normalize the vertical dimension and provide adequate support to the facial soft tissues.

Based on Frère Maia and Pinheiro exhaustive review and classification system for these disorders our case comes under AED1,2,4(Anhidrotic Ectodermal Dysplasia 1,2,4).the differential diagnosis considered were,

1. Pituitary deficiency/craniofacial dysostosis where we can find no eruption of teeth in spite of presence of teeth like structures in both jaws.
2. Ellis-Vancrevald syndrome (Chondroectodermal dysplasia) manifested by, Dwarfism Polydactyl, Hypohidrotic Ectodermal dysplasia affecting Nails, Teeth and Hair, Congenital heart disease.
3. Book’s syndrome as quoted by Pindborg 1970 characterized by absence of premolars (Hypodontia), Hypohidrosis and premature whitening of hair.
4. EEC syndrome (Ectodactly Ectodermal dysplasia-cLEFTING) which is a combination of Lobster claw deformity of the hands and feet(reduction in no of fingers or toes with sundactyly), nasolacrimal duct
obstruction and cleft lip-palate, sparse scalp hair and eye lashes, brittle nails and absence of sebaceous glands associated with Anodontia or severe Oligodontia.

**Conclusion:**

Young patients with ED need to be evaluated early 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. In developed countries, diagnosis of ED pertains to laboratory identification of defective genes and mode of inheritance of mutant gene associated with recessively X chromosome or autosomal dominant or recessive. This may be difficult in developing countries like India where such facilities are insufficient and requires further probing through genetic analysis. In our case, as the patient comes from a very economically poor background, we had applied for the chief minister relief fund for their prosthetic treatment concerning both the children and their genetic profiling had been kept aside. It is to be noted that a positive family history should not be a factor in causing any diagnostic dilemmas with respect to Ectodermal dysplasia, a condition that shows multiple modes of inheritance.

**References:**

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