

Christ-Siemens Touraine syndrome

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A B S T R A C T

Introduction: The plight of any syndrome is the impact they have on the quality of life of the sufferer. The adverse impact on both the physical and emotion well being cannot be overemphasised. Christ-Siemens Touraine syndrome characterized by a congenital dysplasia of one or more ectodermal structures and their accessory appendages.

Case report: A 2 ½ year old child urged his father to get his teeth made. The child was normal in demeanour and did not show any signs of low I.Q. The child responded to questions and obeyed to verbal commands. His clinical appearance was suggestive of a syndrome and exhibited many of the manifestations of ectodermal dysplasia.

Conclusion: The patient suffering from ectodermal dysplasia has multiple issues due to the syndrome. The facial aesthetics are affected and the functional disability due to absence of teeth is also present. Medical intervention for the various clinical symptoms present and sessions with a child psychologist are also inherent in the treatment regime.

Keywords: Oligodontia, anhidrosis, hyperthermia.

INTRODUCTION

'Ectodermal Dysplasia' comprises of a large, heterogeneous group of inherited disorders that are defined by primary defects in the development of 2 or more embryonic ectodermal tissues. This term was first described by Thurnam in 1848 and it was coined by Weech in 1929.^{1,2} This syndrome is characterized by a congenital dysplasia of one or more ectodermal structures and their accessory appendages. The patient may suffer from dry skin, hyperthermia, and unexplained high fever due to the deficiency of sweat glands. Genetic studies done on more than 300 cases reveal an X-linked mode of inheritance, with its gene locus being Xq11-21.1. It is inherited as a X-linked recessive trait so that the frequency and severity of the condition is more pronounced in males than in females.³⁻⁵ A latest study on four consanguineous Palestinian families with features of ectodermal dysplasia reported that appearance of ectodermal dysplasia was consistent autosomal recessive inheritance. The phenotype is due to homozygosity on chromosome 22. The variation occurs at the domain which has high affinity for receptor that regulates Wnt signalling; which in turn is crucial for the development of ectodermal structures. Mutations and other genes in this pathway lead to tooth agenesis with or without other ectodermal anomalies.⁶ This disorder might occur during the first trimester of pregnancy. If it appears before the sixth week of embryonic life the dentition will be affected. After eighth week, other ectodermal structures may be affected.³ These disorders have been considered to

be relatively rare, with an incidence of 7 cases occurring in every 10,000 births.⁴

CASE REPORT

A male boy aged about 2 ½ years reported to the Dental OPD of HAH Centenary Hospital with chief complaint of uneruption of teeth. The child's extra oral appearance was characteristic of ectodermal dysplasia with scanty hair, sparse eyebrows, frontal bossing, saddle nose, wrinkling of the skin over the bridge of the nose, eye-lids and protuberant lips. (Fig. 1,2,3) Intra oral examination reveal edoligodontia with the presence of two peg shaped incisors in the maxilla. (Fig.4) History revealed that the child birth was a normal delivery and the younger sibling, a sister 9 months old is normal. The father also revealed that the boy has extreme intolerance to heat. Medical history was non-contributory. None of the family member from the paternal or maternal side has suffered from this condition. The child was noncooperative for orthopantomogram hence no radiographs could be taken on record.

The patient is on regular follow-up.

DISCUSSION

Clinically, ectodermal dysplasia may be divided into two broad categories. One is the hypohidrotic form which is X-linked, which is characterized by the classical triad of hypodontia, hypotrichosis and hypohidrosis, which is also termed as Christ-Touraine Syndrome. The other category



Figure-1: Front profile showing the wrinkling of skin, sparse hair; **Figure-2:** Lateral profile showing saddlenose, protuberant lips



Figure-3: Birds eye view showing the scanty hair; **Figure-4:** Intra-oral view showing peg shaped lateral

is the hidrotic form which was described by Clouston, which usually spares the sweat glands and can affect the teeth, hair and nails.⁷ The various syndromes which are associated with ectodermal dysplasia are Rapp-Hodgkin Syndrome, Strandberg- Ronchese's Syndrome, Rosselli-Gulienetti Syndrome and various others.⁸ The clinical presentation of the patient with the characteristic facial features and intra oral findings is similar to observations in previous reports. The wrinkling of the skin, thin scanty hair and the extreme intolerance to heat can be explained by the partial or total absence of the hair follicles, sweat glands and sebaceous glands. The observation of normal form of the digits and nails is in agreement with the observations made by Shaw.^{9,10} In studies that utilized serial cephalometric measurements^{11,12} reported that ingeneral facial growth proportion and pattern of jaw growth appear to be normal in these children despite the absence of tooth development. Therefore, the protuberant lips observed in these children may be attributed to the reduction in the height of the alveolar process due to uneruption of teeth.

CONCLUSION

The patient suffering from ectodermal dysplasia has multiple issues due to the syndrome. The facial aesthetics are affected and the functional disability due to absence of teeth is also present. Besides these extreme intolerance to heat depending on the extent to which the ectodermal derivatives are affected pose a definite compromise on the quality of these individuals. The treatment plan constitutes prosthesis fabrication for the compliant patients to allow

the normal growth of the orofacial musculature. Medical intervention for the various clinical symptoms present and sessions with a child psychologist are also inherent in the treatment regime.

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