

A Case of Ectodermal Dysplasia: How Can a Dentist Play Role in Treating it?

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Abstract

Ectodermal dysplasia is a group of systemic conditions that are congenital and are all caused by errors of/in ectoderm and the tissues arising of it. It constitutes of large and complex group of diseases characterized by triad of sparse hairs, abnormal or missing teeth and inability to sweat. Out of 170 ectodermal dysplasia described, <30 have been explained at molecular level with identification of the causative gene. Many cases are associated with anomalies in other organs and in few cases it can lead to mental retardation. Here we present a case of 31-years-old male who reported to us with discomfort in his existing denture and chewing problem.

Keywords: Anodontia, Atrichosis, Ectodermal dysplasia, Hypotrichosis, Tooth agenesis

INTRODUCTION

Ectodermal dysplasia is a heterogenous group of disorders characterized by developmental dystrophies of structures arising from ectoderm.^{1,2} Tissues which primarily affected are teeth, skin, hairs, nails, etc.³⁻⁵ It is characterized by triad of signs comprising sparse hairs (atrichosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohydrosis) the incidence in males estimated at 1 in 100,000 births. Most patients have normal expectancy and normal intelligence.^{6,7} However, lack of sweat glands may lead to hyperthermia, followed by brain damage or death in early infancy if unrecognized.

For dentists and patients both, tooth agenesis and its impact growth and development of jaws are the major concern. The course of the treatment is to restore the function and the esthetics of the teeth, normalize the vertical dimensions and support the facial soft tissues.⁸⁻¹⁰

CASE REPORT

A male patient 31 years of age reported to the clinic with the chief complaint of difficulty in chewing and frequent loosening of existing denture along with sensitivity and occasional pain. On examination following features were found.

On extra oral examination, everted lips, saddle nose, concave facial profile with mild frontal bossing was observed (Figures 1 and 2).

On intraoral examination, multiple teeth were missing, normal alveolar ridge, existing teeth were malformed. No other significant findings were seen (Figures 3 and 4).

On general examination, slurring of speech since childhood as patient informed us was observed.

Patient complained about intolerance to heat because of which he was unable to work in summer days. He used to have frequent baths to keep his skin cool due lack of perspiration (hypohydrosis). Hair follicles were completely absent on his arms, no sweating on hands. He had sparse hairs (hypotrichosis), thin eyebrows, nails were thin. Patient also complained of reduced salivation that led to halitosis.

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Medical and Dental History

Patient had not very significant medical history except frequent episodes of cough and cold. Patient had

undergone multiple teeth extractions in the past. Presently he is wearing removable partial denture. Patient informed that his several teeth had never erupted (hypodontia).

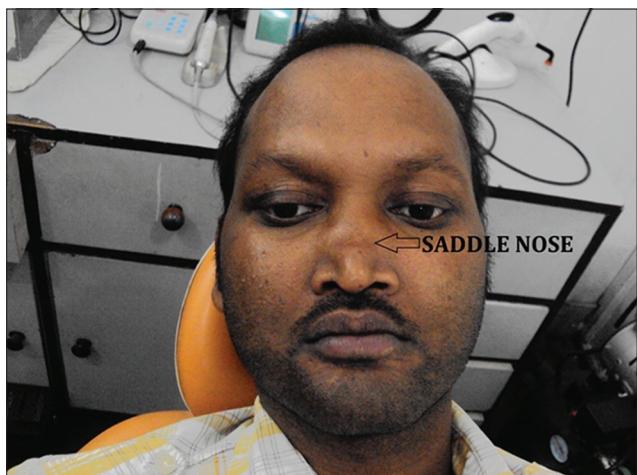


Figure 1: Front view of patient profile along with saddle nose



Figure 2: Brittle nails



Figure 3: Upper arch showing less number of teeth

Radiological Findings

Orthopantomogram was taken, and it showed only four teeth in the upper arch and five teeth in the lower arch were present. Density of bone was poor in upper anterior region and normal in the lower arch (Figure 5).

Lateral cephalogram revealed maxillary teeth deficiency, slight prognathic mandible and frontal bossing which exhibited a concave profile (Figure 6).

Hence considering above all clinical, radiological findings, and clinical examination, final diagnosis of ectodermal dysplasia was made.

DISCUSSION

Ectodermal dysplasias are a large group of heritable conditions characterized by congenital defects of one or more ectodermal structures. Ectodermal dysplasias, as a rule, are not pure “one-layer diseases.” Mesodermal and, rarely, endodermal dysplasias coexist.^{11,12}



Figure 4: Lower arch showing less number of teeth



Figure 5: Orthopantomogram of patient



Figure 6: Lateral cephalogram of patient

Hypodontia, enamel hypocalcification suggested differential diagnosis of Down's syndrome, amelogenesis imperfecta, and ectodermal dysplasias. In Down's syndrome intelligence is poor. In both Down's syndrome and amelogenesis imperfecta other ectoderm derived structures are not affected.¹³⁻¹⁵

Everted lips, frontal bossing, hypotrichosis, hypohydrosis and hypodontia suggests the feature of ectodermal dysplasias, in which structures originated from ectoderm are mostly affected, the absence of tooth buds, hair follicles, and sweat glands and other exocrine glands which develop from ectoderm is primarily affected, whereas structures developing from other germ layers are mostly normal.¹⁶⁻¹⁸

Treatment

Patient with ectodermal dysplasias present many and different clinical problems from early childhood through adolescence and also present a lifelong need for maintenance, care and revision. The course of the treatment is to restore all functions and esthetics of the teeth, normalize the vertical dimension and support the facial soft tissues. Multidisciplinary approach is needed to handle such cases. A team from several specialties will play a major role. Plastic surgeon would need to be consulted to address saddle nose and periorbital wrinkled skin. Dermatologist would be needed to consult check with hypohydrosis and skin infections due to lack glandular secretions. Dental professional team would be needed correct conical teeth and enamel hypoplasia by extraction, crowning or utilization as abutments or support for a prosthesis. A prosthodontist would be needed for fabrication of implant supported denture and orthodontist to address maligned teeth if needed.

CONCLUSION

Ectodermal dysplasias are a heterogeneous group of disorders that affects multiple ectodermal structures, which needs early detection and rehabilitation with multidisciplinary approach. Genetic analysis is critical for early diagnosis and appropriate treatment.

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