

## Case Report

### **Ectodermal Dysplasia, Dental Features and Its Managements: A Report of Two Rare clinical Cases**

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**Abstract:** Ectodermal dysplasia(ED) is defined as a disorder involving structures developing from the ectoderm skin, hair, teeth and sweat glands as well as structures that interact with the ectoderm. These types of ectodermal dysplasias are heritable or genetic disorders. In few cases, it is possible for a child to be the first person in his or her family to be affected by an ectodermal dysplasia. The *EDA*, *EDAR*, and *EDARADD* genes are responsible for cause hypohidrotic ectodermal dysplasia due to mutation in genes mentioned above. The genes *EDA*, *EDAR*, and *EDARADD* provide instructions for making proteins that work together during embryonic development. These proteins form part of a signaling pathway that is critical for the interaction between two germ cell layers, the ectoderm and the mesoderm. In the early embryo, these cell layers form the basis for many of the body's organs and tissues. The interactions between ectoderm-mesoderm are essential for the formation of several structures that arise from the ectoderm. A male patient aged 15 yrs. old (with dental opd registration no.4634/16) reported to the dental outdoor of IGIMS, Patna with complains of yellowish teeth and difficulty in chewing. His mother gave the history that she had twin baby one died at the time of birth and the second one has this problems. On examination his all teeth were erupted (except third molars) with malformed teeth structures and yellowish in colors. There are no cures for ectodermal dysplasias, but many treatments are available to address the symptoms. Second case of a young male child patient aged 05 yrs. old (with dental opd registration no.44460/16) reported to the dental outdoor of IGIMS, Patna, Bihar with complains of teeth are not erupting. The cases are treated symptomatically and long term Prosthodontic treatment plan was advised in case of for second case.

**Keywords:** ectodermal dysplasia, yellowish teeth and cone- occlusion and oligodontia

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#### **INTRODUCTION**

Ectodermal dysplasia(ED) is defined as a disorder involving structures developing from the ectoderm skin, hair, teeth and sweat glands as well as structures that interact with the ectoderm [1]. The affected organs/structures are hair, nails, sweat glands and the teeth mainly. The conditions are a remarkably diverse group of disorders which may also affect other parts of the body [2]. The ectoderm contributes to the formation of the lens of the eye, parts of the inner ear, the fingers and toes, and nerves as well. Therefore, ectodermal dysplasia may cause these parts of the body to develop abnormally. There are more than 150 different types of ectodermal dysplasias [2]. In few rare cases of ectodermal dysplasia affect lifespan and learning disorders. The ectodermal dysplasias divided into two types based on the number and function of sweat glands 1. Hypohidrotic ectodermal dysplasia / Christ-Siemens-Tourine-Syndrome; where sweat glands are absent or markedly reduce in number. 2. Hidrotic Ectodermal Dysplasia / Clouston Syndrome; sweat glands are normal in number. The dentition and hair are

affected similarly in both types, but the hereditary patterns and nail and sweat gland manifestations tend to differ [6].

#### **CAUSES OF ECTODERMAL DYSPLASIAS**

These types of ectodermal dysplasias are heritable or genetic disorders. In few cases, it is possible for a child to be the first person in his or her family to be affected by an ectodermal dysplasia. The *eda*, *edar*, and *edaradd* genes are responsible for cause hypohidrotic ectodermal dysplasia due to mutation in genes mentioned above. The genes *eda*, *edar*, and *edaradd* provide instructions for making proteins that work together during embryonic development. These proteins form part of a signaling pathway that is critical for the interaction between two germ cell layers, the ectoderm and the mesoderm. In the early embryo, these cell layers form the basis for many of the body's organs and tissues. The interactions between ectoderm-mesoderm are essential for the formation of several structures that arise from the ectoderm. The structures of the ectodermal in origin are

skin, hair, nails, teeth, and sweat glands. Mutations in the *eda*, *edar*, or *edaradd* gene prevent normal interactions between the ectoderm and the mesoderm and impair the normal development of the ectodermal organs. In the 1990 edition of the "birth defects encyclopedia", it is stated that as many as seven of every 10,000 babies are born affected by an ectodermal dysplasia. The prevalence of ectodermal dysplasias is almost equal in both sexes and affects all races and ethnic groups [3, 4].

## DIAGNOSIS

In few cases, ectodermal dysplasia is apparent at the time of birth. In other cases, a parent or doctor may only begin to suspect that a problem exists when teeth fail to develop normally. The two cardinal signs of developmental defects first malformed teeth and second is sparse hair confirms the diagnosis of ed. Specific genetics tests to diagnose ectodermal dysplasia are available for only a limited number of ectodermal dysplasias. Features in derivatives of the ectoderm structures: Teeth affected are in shape and in size, may be missing, pointed, widely spaced, or prone to cavities because of defective enamel. Dental treatment is almost always necessary and children may need dentures as early as two years of age. Multiple denture replacements are needed as the child grows, and dental implants may be an option in adolescence. Orthodontic treatment may also be necessary but usually not performed. Scalp and body hair may be absent, sparse, thin, very light in color, excessively brittle, curly, or even twisted. Fingernails and toe-nails may be thick or thin, abnormally shaped, discolored, ridged, slow-growing, or brittle. Many individuals affected by ectodermal dysplasia cannot perspire. Their sweat glands may function abnormally or may not have developed at all this may result in abnormal sweat production. The body temperature rises in the summer. Skin may be thin and pale, dry, scaly and easily irritated, prone to rashes, infections and sunburn, or thick over the palms and sole. Skin erosions or open areas of the skin on the scalp, hands and feet are possible. The other features in some cases are dryness of the eye, sensitivity to sunlight, cataracts, vision defects, hearing problems, respiratory infections, cleft lip and/or palate, or missing fingers and toes [1, 3, 5].

### Case No. 01

A male patient aged 15 years old (with dental opd registration no.4634/16) reported to the dental outdoor of IGIMS, Patna, Bihar with complaints of yellowish teeth and difficulty in chewing. His mother gave the history that she had twin baby one died at the time of birth and the second one has these problems. On intra-oral examination his all teeth were erupted (except third molars) with malformed teeth structures and yellowish in color (fig.no.1 frontal view of occlusion). Enamel was absent in almost all teeth, and dentin was visible, this is the reason behind the yellowish coloration of the teeth. The teeth present are

arranged (occludes) such in way i; e cone to cone arrangement of the teeth rather than cusp-to fossa arrangement (normal arrangement of teeth) which resulted in the difficulty in chewing (fig.no.2 and 03 left and right view of occlusion). On general physical examination, sparse hair on his scalp with dark encircle over the eyes (fig.no.04 facial view). He also complains of hyperthermia (mainly in the face and head part) in the summer seasons this may be due to fewer sweat glands. The few scar marks are seen on the forehead due to thin skin (fig.no.5). The nose is saddled (broad) in appearance. (Fig.no.05, head view). The patient appears normal and understands the commands and talks normal but restless. He was also sent to the department of psychiatric for personality evaluation and development.



Fig-1: Frontal view of occlusion



Fig-2: Left view of occlusion



Fig-3: Right view of occlusion



Fig-4: Facial view of a patient



Fig-5: Head view of patient with sparse hair

#### Case No.02

A male child patient aged 05 yrs. old (with dental opd registration no.44460/16) reported to the dental outdoor of IGIMS, Patna, Bihar with complains of teeth are not erupting. On taking family history his parents revealed that he has two sons of whom second son is completely normal and three daughters, whom all the daughters are also completely normal. On intra oral examinations reveals only both side deciduous upper first molars are erupted. On general physical examination child was with sparse hairs on the scalp. Patient also complains of more dry mouth and warm body temperature during summer season. On panoramic radiographic examination reveals only second deciduous molars are present in the bony arch. All other dentitions are missing including permanent dentition.



Fig-6: Radiographic view (in second case) with oligodontia

#### TREATMENT

There are no cures for ectodermal dysplasias, but many treatments are available to address the symptoms. Patients is advised, not to go for scaling and

polishing as it remove the enamel and expose the dentin, may results in sensitivity of the teeth. Medicated tooth paste is recommended of cleaning the teeth. The soft bristles brush issued for cleaning the teeth. In the case of second patients his Prosthodontics rehabilitations of the whole mouth are advised. The most common treatment plan is removable prosthesis [6]. Implant-supported denture is also suggested as the ideal reconstruction modality for adolescents over 12 years. In case of dry mouth, a mouth wash is recommended [3, 4].

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