Ectodermal Dysplasia- 2 rare case reports

[1]Arjit Vihan, [2]Rajendragouda Patil, [3]Ambika Murari

[1,3]Post graduate student, Department of Oral Medicine and Radiology, Kothiwal dental college and Research Centre, Moradabad

[2]Professor and Head, Department of Oral Medicine and Radiology, Kothiwal dental college and Research Centre, Moradabad

E-Mail Id- Arjitvihan@gmail.com

Correspondence Author: Dr. Arjit Vihan, Department of Oral Medicine and Radiology, Kothiwal dental college and Research Centre, Moradabad

Contact No: 8191974974

E-Mail Id: Arjitvihan@gmail.com

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ABSTRACT

Ectodermal dysplasia (ED) is a rare genetic condition affecting multiple ectodermal structures such as the hair, nails, teeth, and sweat glands. Depending on the specific syndrome, ectodermal dysplasia may also impact the skin, the lens or retina of the eye, portions of the inner ear, the formation of fingers and toes, nerves, and other areas of the body. Each syndrome typically presents with a unique combination of symptoms, varying in severity from mild to severe. The syndrome is estimated to occur in approximately 1 case per 100,000 live births. The most prevalent types of ectodermal dysplasias are hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome) and hidrotic ectodermal dysplasia (Clouston syndrome). In this paper we present two case reports of two male patients reported with the features suggestive of this condition. Comprehensive medical care for patients with ED necessitates a multidisciplinary approach involving physicians from various specialties.

KEYWORDS- Ectodermal Dysplasia; Multidisciplinary; Hypohidrosis; Anhidrosis.

INTRODUCTION

Ectodermal dysplasia (ED) is a rare hereditary disorder characterized by primary developmental abnormalities in two or more tissues originating from the ectoderm. These primarily affected tissues include the skin, hair, nails, eccrine glands, and teeth. The disorders are congenital, diffuse, and non-progressive[1,2]. Among these disorders, the most prevalent syndromes are hypohidrotic (anhidrotic) ED and hidrotic ED. Hypohidrotic ED, also referred to as Christ-Siemens-Touraine syndrome, is the more frequent phenotype and typically follows X-linked recessive inheritance. It is marked by various anomalies such as reduced sweating, abnormal dentition, onychodysplasia, and hypotrichosis. Characteristic facial features often include prominent forehead, sunken cheeks, a saddle-shaped nose, thickened and protruding lips, wrinkled and hyperpigmented skin around the eyes, and large, low-set ears. Dental characteristics can involve cone-shaped or peg-shaped teeth, partial or complete absence of teeth (hypodontia or anodontia), and delayed eruption of permanent teeth. Eccrine sweat glands may be absent or underdeveloped, especially in individuals with hypohidrotic ED[3]. In certain instances, mucous glands may be lacking in the upper respiratory tract, bronchi, esophagus, and duodenum. Additional prevalent symptoms include short stature, eye abnormalities, reduced tear production, and sensitivity to light (photophobia)[4].

CASE- 1

A 27 year old male patient reported with the complaint of missing teeth in upper, lower, front and back tooth region. On further questioning the patient revealed that he had this condition since 1 year of age but as his parents were not aware of the condition so did not went for any consultation. Parents of the patient gave history that child had no teeth till 4-5 years of age but when he was around 5-6 years two teeth in front region started erupting followed by few teeth in posterior but they noticed that the shape was abnormal and along with that patient also started difficulty in biting and chewing from those teeth. Apart from this he also gave history of lack of hair in scalp, eyebrows and eyelashes since 1 year of age. On medical history the patient revealed that he suffers repeated episodes of frequent fever since 10 years. Such episode used to aggravates during summer months usually June and July. For this patient used to bath 2-4 times everyday and increase intake of fluids but again there was no relief. The dental history of the patient was non- contributory. The family history of the patient revealed that the parents of the patient had non-consanguineous marriage but there

was no complication during the birth of the patient. The personal history of the patient was non-contributory and the vital signs of the patient were under normal limits.

During the clinical examination, extra-orally, the patient had frontal bossing, there was prominent supraorbital ridge, thick upper and lower lips, sparse hair, scanty eyelashes, depressed nasal bridge, there were absence was secondary sexual characteristics, along with that there was lack of hair in scalp and loss of body hair (figure.1).

Upon intra-oral examination 16 permanent teeth were absent including 12, 14, 15, 22, 24, 25, 32, 31, 33, 34, 35, 41, 42, 43, 44, 45 and he also had cone-shaped teeth (figure.2).

The orthopantomogram (OPG) was performed which showed multiple absent teeth and generalized bone loss interdentally. The shape of the teeth seen in the OPG were showing abnormal presentation (Figure.3).

The mode of inheritance was established through family history. Diagnosis was confirmed via skin biopsy, which showed thinning and flattening of the epidermis, along with a decrease in the number and presence of underdeveloped eccrine sweat glands.

Hence, on the basis of clinical features as well as radiographic features, Ectodermal dysplasia was diagnosed.

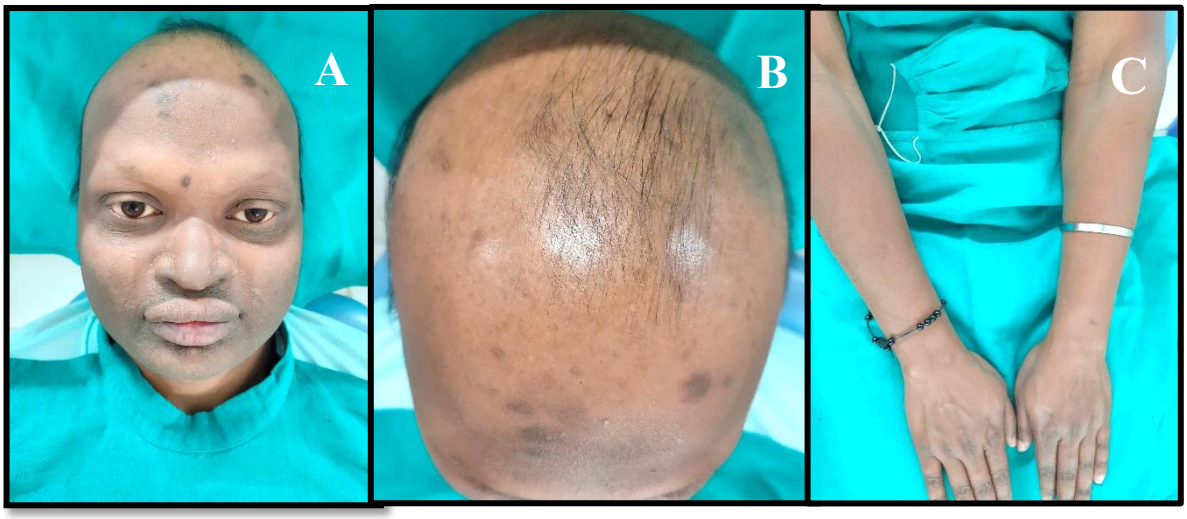

Figure 1. Extra-oral pictures. A) Patient profile, B) Sparse hairs seen on the scalp, C) Lack of body hair on hands of the patient.

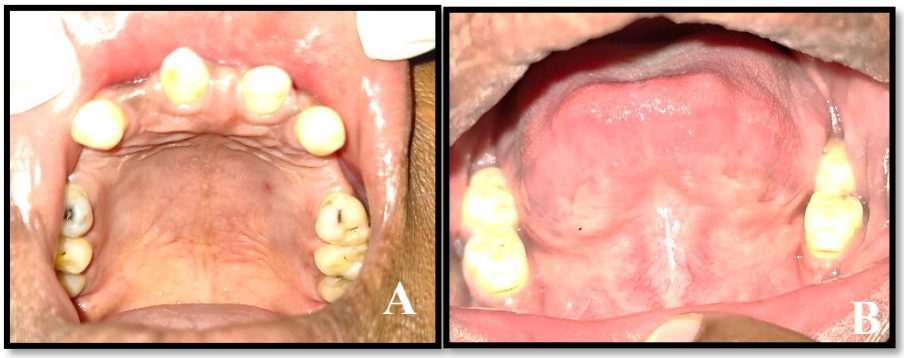

Figure 2. Intra-oral pictures. A) Maxilla with multiple missing teeth and cone shaped teeth, B) Mandible with multiple missing teeth.

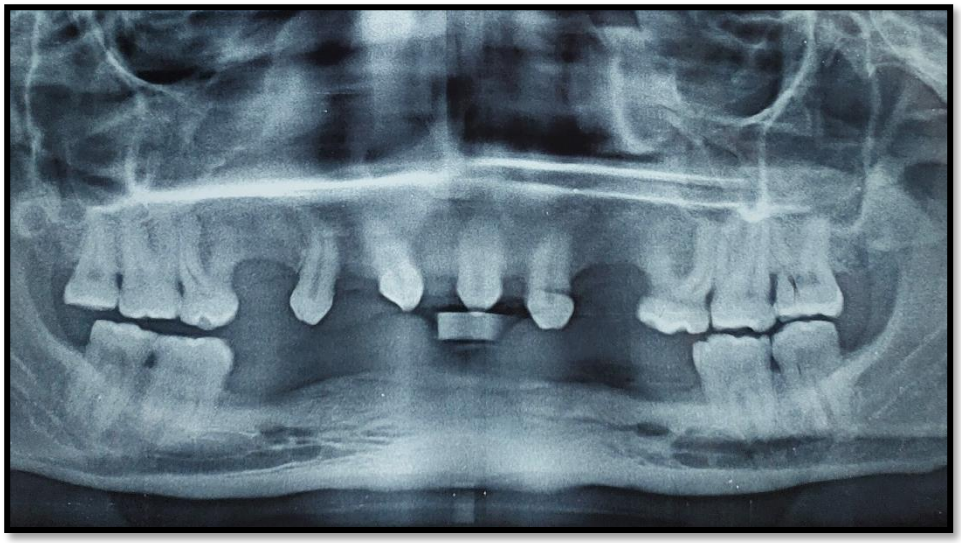

Figure 3. OPG of the patient.

CASE- 2

A 15 year old patient reported with the complaint of missing teeth in upper and lower front tooth region since childhood resulting in difficulty with chewing food. He had heat intolerance and a medical history of recurrent chest infections. According to his family history, his uncle also experienced oligodontia and heat intolerance. The dental history revealed that the patient was a denture wearer since 5 years of age. The personal history of the patient was non-contributory.

On extraoral examination, patient presented with fine, sparse scalp hair, scanty eyebrow and eye lashes, thick upper lip, depressed nasal bridge, pigmented and coarsed skin, there was absence of sweat glands as well, lack of body hair was appreciated as well (figure.4).

On intra-oral examination, it revealed complete anodontia of the deciduous teeth and partial anodontia of the permanent. A total of 14 teeth were missing from the patients mouth and interestingly the teeth present were only in the posterior region, maxillary and mandibular anteriors were missing since birth of the patient only (figure. 5).

The orthopantomogram (OPG) was performed and it revealed multiple retained deciduous in the first and third quadrant of the jaw, along with that multiple missing tooth in the maxillary and mandibular anterior region. Generalized interdental bone loss was evident, Also the shape of the posterior teeth appear abnormal in shape (figure.6).

Hence, on the basis of clinical features as well as radiographic features, Ectodermal dysplasia was diagnosed.

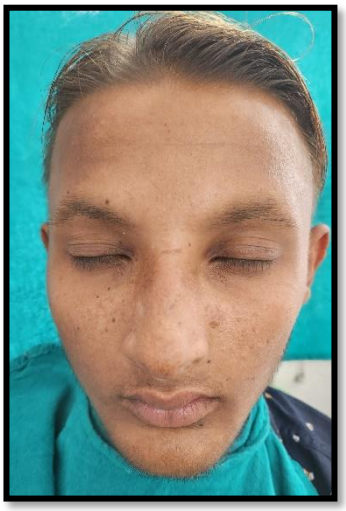

Figure 4. Patient profile.

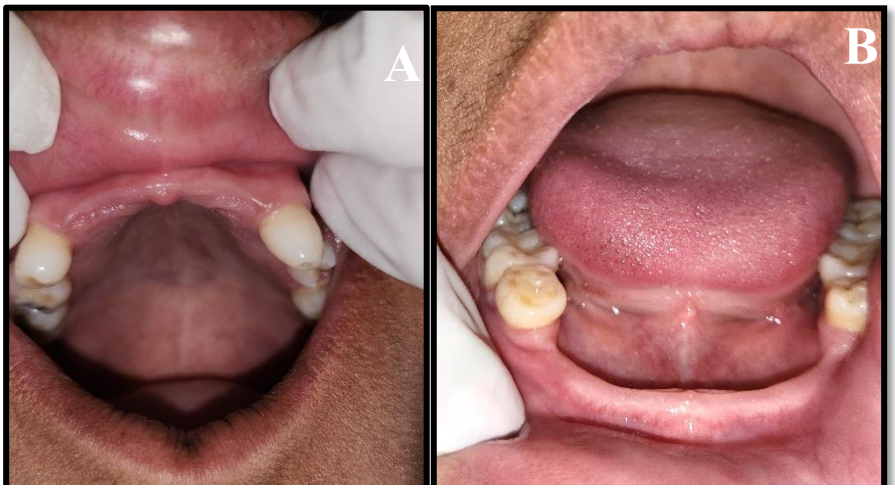

Figure 5. Intra-oral pictures. A) Maxilla with multiple missing teeth and cone shaped teeth, B) Mandible with multiple missing teeth.

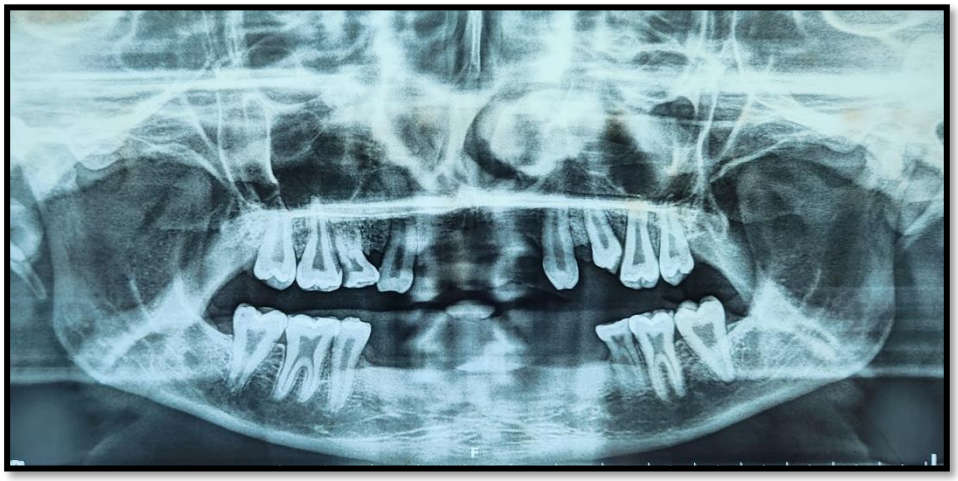

Figure 6. OPG of the patient.

DISCUSSION

Ectodermal dysplasia is an uncommon congenital condition that arises from disruptions in ectodermal development, initially described by Thurman[5]. Ectodermal dysplasias (EDs) are a diverse group of hereditary disorders that occur in approximately one out of every 100,000 births. They arise from primary developmental defects affecting two or more tissues derived from the embryonic ectoderm[6].

Clinical characteristics may encompass nail dystrophy, sparse hair growth (hypotrichosis), reduced skin thickness on the palms and soles (palmoplantar hypokeratosis), tooth abnormalities (odontodysplasia), and dysplasia affecting other ectoderm-derived structures. Hair abnormalities are the most prevalent manifestation (>90% of patients), followed by dental defects (80%), nail issues (75%), and sweat gland abnormalities (42%)[7]. Along with this the individual exhibits dull blonde hair on the scalp, accompanied by widespread skin scaling. Unexplained fever and heat intolerance commonly result from lack of sweating (anhidrosis). Normal intelligence is typically observed[3]. Other notable facial characteristics include a prominent forehead, sunken cheeks, a flattened nasal bridge, thick protruding lips, wrinkled and hyperpigmented skin around the eyes, and large, low-set ears[8].

ED is generally classified into two main categories: the hypohidrotic form (Christ-Siemens-Touraine Syndrome), which is X-linked and marked by the classical triad of hypodontia, hypotrichosis, and hypohidrosis; and the hydrotic form (Clouston syndrome), which affects the teeth, hair, and nails while sparing the sweat glands[3]. The most prevalent oral symptom is the absence of some or all deciduous and permanent teeth, often accompanied by cone-

shaped tooth morphology[9]. Diagnosis relies on a thorough assessment of family history, along with comprehensive clinical and radiographic examinations[10].

The two patients reported to us had similar clinical findings including frontal bossing, thick lips, sparse hair, scanty eyeslashes, absence of sweat glands, anodontia, cone shaped teeth. Along with these findings one of the patient gave a history of recurrent fever and the second patient gave a history of heat intolerance. The clinical and radiological findings helped us in diagnosing both the cases as Ectodermal dysplasia.

CONCLUSION

Ectodermal dysplasias (EDs) are uncommon genetic disorders with numerous overlapping features, making classification challenging. The clinical manifestations of ED can lead to significant social challenges for affected individuals, affecting both oral and overall bodily functions. A thorough evaluation of the patient's family history, coupled with an assessment of the family's socioeconomic status, is crucial for developing an effective treatment plan for ectodermal dysplasia. The cornerstone of successful management for ED lies in prompt diagnosis and prosthetic rehabilitation through a multidisciplinary approach. This approach further contributes to fostering positive self-esteem and significantly improves social integration.

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