

# Ectodermal dysplasia - A case report from Nepal

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## Abstract

Ectodermal dysplasia (ED) is a rare genetic disorder with abnormal development of two or more ectodermal derivatives. These include defects in skin, nails, hair, teeth and sweat glands. A multidisciplinary approach is required for management of such cases. A case of 9 years old boy with ED is reported here.

## Introduction

Ectodermal dysplasia (ED) is a rare heterogenous group of heritable disorder that manifests as developmental defects of two or more tissues of ectodermal origin.<sup>(1)</sup> The skin, hair, nails, eccrine glands, and teeth are the tissues primarily affected<sup>(2)</sup>. Other tissues of ectodermal origin that may be involved are mammary gland, central nervous system, external ear, cornea, conjunctiva, melanocytes, and lacrimal gland and duct<sup>(3)(4)</sup>. All patterns of inheritance like autosomal recessive/dominant, X-linked and mitochondrial have been described. The ED which is congenital, diffuse and nonprogressive, has an estimated incidence of around seven cases per 100,000 live births<sup>(1)</sup>. Traditionally ectodermal dysplasia is classified into two types based on sweat gland function. The first one is X-linked hypohidrotic type (Christ-Siemens-Touraine syndrome) presenting with a classical triad of hypodontia, hypohidrosis (reduced or absent sweating), and hypotrichosis. In contrast to that, the second one is the hydrotic type (Clouston syndrome) characterized by normal sweat gland function with other ectodermal defects and is inherited in autosomal dominant manner<sup>(1)</sup>. But this simple classification has failed to address the anomalies associated with nail, hair and teeth in various forms of ED.

## Case Report

A 9 years old boy presented to our OPD with complaints of dry skin, multiple missing teeth in both upper and lower jaws, absent right middle finger distal phalanx and nail, and sparse fine grey hairs in the scalp.

His vitals and systemic examination were normal. The child had mandibular hypodontia with peg-shaped mandibular canine. He had sparse, hypopigmented hairs in the scalp. His distal phalanx of right middle finger along with its nail plate was missing (Figure 1,2 and 3). Orthopantomograph (OPG) revealed multiple missing permanent tooth buds. His family history was also significant. His siblings i.e, brother and sister were normal. But his father had dry, coarse skin with sparse hair in scalp with multiple missing teeth in upper and lower jaw. However, father had normal fingers and nails. On the basis of history, clinical and radiological examinations, a final diagnosis of hypohidrotic ectodermal dysplasia was made.

## Discussion

The EDs are hereditary genetic disorders characterized by primary developmental defects of two or more ectodermal structures, one of which is either hair, teeth, nail or sweat glands. X-linked hypohidrotic form (Christ Touraine Syndrome) with absent or reduced sweat production is the most common ED.<sup>(5)</sup> However autosomal dominant/recessive cases of hypohidrotic EDs have been reported.<sup>(3)</sup> Clinically they present with hypotrichosis i.e, fine, slow growing scalp and body hair and sparse eyebrows with hypodontia and nail

anomalies.<sup>(6)</sup> Both deciduous and permanent teeth are typically peg-shaped and reduced sweat production results into dry skin and heat intolerance.<sup>(5)(6)</sup>

Hydrotic ectodermal dysplasia (Clouston syndrome) with autosomal dominant inheritance affects the teeth, hair and nails but sweating is normal. The pathophysiology usually involves mutations in connexin gene leading to hypotrichosis (partial or total alopecia), brittle hair, dystrophic nails, clubbing, finger pulp dermal ridges and keratoderma. Eye lashes and eye brows are sparse or absent. Paronychia infections are commonly observed.<sup>(7)</sup> The management involves keratolytics and emollients for palmoplantar keratoderma. Minoxidil has been used to enlarge miniaturized hair follicles while wigs can be used in cases of total alopecia. Nail dystrophy can be addressed with professional pedicures and manicures.<sup>(8)</sup>

The other features commonly observed in EDs are frontal bossing, sunken cheeks, depressed nasal bridge, thick everted protuberant lips, wrinkled hyperpigmented periorbital skin, and a large low set of ears.<sup>(5)</sup> The most common oral sign is hypodontia or anodontia of deciduous and permanent dentition associated with conical teeth. The pathophysiology of ED involves the genetic defect in complicated network of signalling pathways that coordinates the formation and function of ectodermal structures. Some of these important molecular pathways are Hedgehog, Wntless, TNF- $\alpha$ , NF- $\kappa$ B, ED and p63 signalling pathways, Gap junctions-connexin pathway and Axin pathway.<sup>(3)(4)</sup>

The diagnosis of ED can be made based on family history, thorough clinical evaluation, molecular analysis and imaging studies. In our case, permanent central incisor and canine teeth are missing in lower jaw along with dry, rough skin. There was absence of distal phalanx of right middle finger with missing nail plate. Molecular analysis couldn't be done in our resource constrained settings. OPG and hand radiographs were taken to confirm the clinical diagnosis. These clinical features and imaging studies were suggestive of hypohidrotic ED.

## Conclusion

A multidisciplinary team consisting of pediatricians, pedodontists, prosthodontists, orthodontist, dermatologists, otolaryngologists, psychiatrists and speech therapists are required for diagnosis and treatment of child with ED and to address the significant social, physical and psychological problems in affected individuals. Early diagnosis of hypohidrotic ED is of utmost importance to avoid life threatening complications induced by hyperthermia and infections. Early prosthetic treatment in children with ectodermal dysplasia can result in significant improvement in esthetics, masticatory, phonetic function and will be helpful in psychosocial development of child.

## CONFLICT OF INTEREST

The authors have no conflicts of interest.

## AUTHOR CONTRIBUTIONS

KA was involved in manuscript preparation, literature search and she is correspondence author. SK was also involved in literature search. SM was involved in concept, manuscript editing, guidance, and in the final approval of manuscript.

## CONSENT

A written consent was obtained from the patient party for the publication of the case and images.

## DATA AVAILABILITY STATEMENT

We agree to make the manuscript available to general people and are also ready to provide other necessary data regarding the case report when required.

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