

## Case Report

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# Christ Siemens Touraine syndrome: a rare entity

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## ABSTRACT

Ectodermal dysplasia is a rare entity with incidence of 1 in 1,00,000 births with male predominance. Most commonly it presents with appendageal abnormality with facial dysmorphism. The two most common types of ectodermal dysplasias are hypohidrotic ectodermal dysplasia (Christ-Siemens-Touraine syndrome) and hidrotic ectodermal dysplasia (Clouston syndrome). Clinical recognition varies depending on severity of symptoms and associated complications. The prognosis is good after infancy if diagnosed early with appropriate management of complications. Here we present a case of eight-month-old female with hypohidrotic ectodermal dysplasia.

**Keywords:** Ectodermal dysplasia, Genetic, Hypohidrotic, Hypodontia, Skin

## INTRODUCTION

Hereditary ectodermal dysplasia (ED) is characterized by defective formation of one or more structures derived from the ectoderm.<sup>2</sup> It was first described by Thurnam in 1848 and the name was coined by Weech in 1929.<sup>1</sup> Ectodermal dysplasias are congenital, diffuse, and non-progressive. It is of two types: hypohidrotic ED, also known as Christ-Siemens-Touraine syndrome, which presents with a classic triad of hypohidrosis, hypotrichosis, and hypodontia and; hidrotic ED, also known as Clouston syndrome, in which hair and nails are affected with sparing of sweat glands.<sup>3</sup>

Hypohidrotic ED is the most common form of ectodermal dysplasia. It is most commonly X linked recessive condition. Inability to produce sweat, intolerance to heat, presence of dry scaly skin should give rise to suspicion of ED. Genetic testing, family screening should be done. Family education regarding care of the child plays crucial role in the prognosis. History and clinical examination remains gold standard to reach the diagnosis. Genetic testing to confirm the diagnosis, to identify the carrier state

and to offer prenatal diagnosis is essential in this era of molecular diagnosis.

## CASE REPORT

An eight-month-old female child presented with failure to thrive, recurrent lower respiratory tract infections since birth, choking and coughing episodes after taking food. On enquiry there was absence of sweating and high temperatures recordings on and off from birth. There was no significant family history. On examination, she was euthermic with stable vital parameters. Sparse scalp hairs with hardening of skin and dry scaly skin was noted. There was no dentition. Systemic examination were within normal limits.

We had a differential diagnosis of Christ Siemen Touraine syndrome, Rothmund-Thomson syndrome. Skin biopsy was done which was suggestive of hypohidrotic ED. Genetic testing was planned to confirm the diagnosis. EDAR gene was found to be pathogenic which confirmed our diagnosis of hypohidrotic ED.



Figure 1: Clinical photo.



Figure 2: Dry scaly skin with hardening of skin.

DNA TEST REPORT - MEDGENOME LABORATORIES						
Full Name / Ref No:	MARIAM BELDAR		Order ID/Sample ID:	103376/252944		
Gender:	Female		Sample Type:	Blood		
Date of Birth / Age:	8 months		Date of Sample Collection:	14 <sup>th</sup> March 2019		
Referring Clinician:	Dr. Alpana Kondekar, Nair Hospital, Mumbai		Date of Sample Receipt:	15 <sup>th</sup> March 2019		
Test Requested:	Ectodermal dysplasia panel genes		Date of Order Booking:	16 <sup>th</sup> March 2019		
			Date of Report:	17 <sup>th</sup> April 2019		
<b>CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY</b> Baby Mariyam Beldar, born of a consanguineous marriage is suspected to be affected with ectodermal dysplasia and has been evaluated for pathogenic variations in the genes listed in appendix 1.						
<b>RESULTS</b>						
LIKELY PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED						
Gene (Transcript) *	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
<i>EDAR</i> (+) (ENST00000258443.2)	Exon 12	c.1144G>A (p.Gly382Ser)	Homozygous	Hypohidrotic ectodermal dysplasia-108	Autosomal recessive	Likely Pathogenic
*Genetic test results are reported based on the recommendations of American College of Medical Genetics [1].						
ADDITIONAL FINDINGS: NO VARIANT(S) OF UNCERTAIN SIGNIFICANCE (VUS) DETECTED						
No other variant that warrants to be reported was detected. Variations with high minor allele frequencies which are likely to be benign will be given upon request.						

Figure 3: Genetic diagnosis report.

## DISCUSSION

EDs occur approximately 1 in every 100,000 births and are caused by primary defects in the development of two or more tissues derived from the embryonic ectoderm - hair, teeth, nails, sweat glands. There are more than 150 types

of EDs.<sup>4</sup> The mode of inheritance is X-linked with its gene locus being Xq11-21.1. It is carried by the female but manifested only in the male.<sup>5</sup> In recent times a transmembrane protein "ectodysplasin" (TNF family ligand) and a receptor "edar" (TNF receptor) have been identified which regulate the ectodermal appendage formation and organogenesis.<sup>6</sup>

The genes affected are ectodysplasin A (EDA), EDA anhidrotic receptor, EDAR and NEMO. Clinical features include absence of sweat, dry scaly, wrinkled skin with hypo pigmented lesions and intolerance to heat. Missing/pointed conical teeth, frontal bossing, sunken cheeks and saddle nose, thick everted lips, low set ears, prominent supraorbital ridge, midfacial hypoplasia, brittle nails with ridges, sparse scalp hair with normal sexual hair growth are some of the other features. Ophthalmologic and otolaryngic abnormalities are present secondary to decreased saliva and tear production. High incidence of atopic diseases, high incidence of GERD is present in 20% of cases. Infant mortality rate is 30% in these conditions. The presentation of facial deformity, dry skin, and sparse hair in our case report was similar to previous reports. Most incredible oral finding in our patient was absence of primary teeth which is a rare finding in ED.<sup>7</sup> Patients with hypohidrotic ED usually have oligodontia of primary and permanent teeth. In addition, high-arched palate can be more frequent and cleft palate may be present.<sup>8</sup>

Investigations include starch iodine test, palmar and scalp biopsy. For prenatal diagnosis, fetal skin biopsy by fetoscopy is advised at 20 weeks. Genetic diagnosis for confirmation of diagnosis.

Management usually demands a multidisciplinary team approach. It includes protection from exposure to high temperatures, early dental evaluation for adequate nutrition, removable prosthodontics and dental implants, use of lacrimal tears to prevent corneal damage. Family screening and other sibling screening is also advised. The oral rehabilitation of ED patients usually consists of complete or removable prosthesis in the developing years, followed by a definitive prosthesis. Vestibuloplasty and ridge augmentation should be considered prior to prosthetic treatment. Orthodontic treatment needs to be considered for mal-aligned teeth. Dental implant treatment may be considered for anodontia.<sup>9</sup>

## CONCLUSION

Clinical acumen always takes a precedence in early diagnosis and thus early intervention, treatment and rehabilitation.

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