



**CHRIST-SIEMENS-TOURAINÉ SYNDROME WITH PULMONARY TUBERCULOSIS: A CASE REPORT**

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**ABSTRACT**

Ectodermal dysplasia is a rare disorder with defects in two or more of the following structures: the teeth and the skin and its appendages including hair, nails, eccrine, and sebaceous glands. Anhidrotic ectodermal dysplasia is the most common type of disease. Affecting 1 in 17000 individual. This rare disorder, also known as Christ-Siemens-Touraine syndrome, which is a form of anhidrotic ectodermal dysplasia (ED) characterized by triad of hypodontia, hypotrichosis, and hypohidrosis.

Here we report a case of 2 year old boy who was admitted to the department of paediatrics with complains of fever for one month. There was sparse hair, hypodontia with sharp incisor teeth and abnormal facies and B/L bronchial breath sounds were heard. Chest radiography showed B/L non homogenous opacity. Hence, a diagnosis of Christ-Siemens-Touraine syndrome with Pulmonary Tuberculosis was made.

Here we want to emphasize that there is immunodeficiency in ED which makes the children susceptible to many infections.

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**INTRODUCTION**

Ectodermal dysplasia is divided into two types based on the number and function of sweat glands: hidrotic ectodermal dysplasia (Clouston syndrome) and hypohidrotic (anhidrotic) ectodermal dysplasia (Christ-Siemens-Touraine syndrome) [1]. Christ-Siemens-Touraine syndrome has characteristic triad of reduction in the amount of hair (hypotrichosis), absence of sebaceous glands (asteatosis), and absence of sweat glands (anhidrosis) [2]. It is an X-linked recessive condition seen exclusively in males resulting due to a mutation in transmembrane protein (ectodysplasin A), associated with ectodermal structures. It was first described by the British physician, Dr. J. Thurnam in 1848. [3]. The children may have depressed nasal bridge, protuberant lips, prominent supraorbital ridges, sunken cheeks, and wrinkled hyperpigmented skin around the eyes. The oral manifestations include conical or peg-shaped teeth, hypodontia or complete anodontia in both deciduous and permanent dentition, malformation of present teeth, generalized spacing, delayed eruption of permanent teeth, underdeveloped alveolar ridges, and high palatal arch or a cleft palate. Hypoplasia of salivary glands and the absence of oral accessory glands are also present and result in xerostomia and dry cracked lips [4]

**Case Report**

A 2 year old boy was admitted to emergency department of paediatrics with complains of fever for 1 month. The boy was born out of non consanguineous marriage by normal vaginal delivery and was immunized as per National immunization schedule. The boy's mother had noticed slight sweating and episodes of fever without clinical symptoms, which were more frequent in summer. *There was history of contact with an open case of tuberculosis.* On general examination, he had sparse, thin, light, blond hair over the scalp, scanty eyebrow and eyelashes, depressed nasal bridge, frontal bossing, and prominent supraorbital ridges. Lips were dry, everted, and prominent. (Fig 1). Oral examination revealed hypoplastic conical teeth, and delayed dentition. (Fig 2). On respiratory examination B/L bronchial breath sounds were heard and Chest radiography showed non homogenous opacity more on left side (Fig 3). Following which a diagnosis of Christ-Siemens-Touraine syndrome with Pulmonary Tuberculosis was made. The child was started on Anti Tubercular drugs under RNTCP category 1. The parents were counseled about the disease. They were advised to keep baby away from sunlight, avoid hot and spicy food and to make the child bath twice a day. The fever subsided within 7 days and the baby was discharged successfully with antitubercular drugs.

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Fig 1 showing sparse hair and abnormal face



Fig 2 showing abnormal conical teeth



Fig 3 showing chest radiography with patch

## DISCUSSION

The frequent occurrence of rhinitis and other upper and lower respiratory tract infections like pharyngitis, bronchitis, and pneumonia can be attributed to the scanty mucus production and deficient ciliary action along this span of area<sup>[5]</sup> Moreover, immune deficiency can also be a possible cause for the recurrent infections. Affected patients manifest dysgammaglobulinemia and, despite therapy, have significant morbidity and mortality. The hypohidrotic/anhidrotic form of ED (HED/EDA) has been attributed to at least 4 genes *EDA1* [ectodysplasin]; *EDA3*; *EDAR* [the EDA-A1 isoform receptor]; and *EDARADD* [EDAR-associated death domain]), with at least 3 modes of inheritance: X-linked recessive

(OMIM 305100), autosomal dominant (OMIM 129490), and autosomal recessive (OMIM 224900). X-linked ectodermal dysplasia with immunodeficiency (XL-EDA-ID) is described in patients with hypomorphic mutations in *IKBKKG* (the inhibitory  $\kappa$ B kinase  $\gamma$  gene), which encodes nuclear factor  $\kappa$ B essential modulator (NEMO). Features include hypohidrosis, dental anomalies, alopecia, and immunodeficiency. Boys with NEMO mutations often present with serious infections, but the NEMO mutations are rarely diagnosed early in infancy<sup>[6]</sup>. The overlapping clinical features makes diagnosis difficult with different clinical conditions like limbmammary syndrome, incontinentia pigmenti (IP), De Sanctis-Cacchione syndrome, and Turnpenny type of ED.<sup>[7,8]</sup>

## CONCLUSION

Hypohidrotic ectodermal dysplasia should be included in the differential diagnosis of fever of unknown origin. The diagnosis of ED can be difficult due to a variety of types, range of abnormalities, and severity of defects. However, it is important to identify the components of the disorder so that appropriate treatment can be rendered to ED patients. It is also important to understand the genetic hereditary patterns so that the parents of an affected child can be advised on the possibility of new cases in the family.

## Contributors

Dr.Nasreen Ali-conception, design and drafting Dr.Sunil Kumar Agarwalla-revising it critically for important intellectual content.

## Conflict of Interest

There was no conflict of interest and no funds received.

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