GENETICS AND GENODERMATOSES

CHRIST SIEMENS TOURAINE SYNDROME-A RARE CASE PRESENTATION

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Background: Ectodemal dysplasias(ED) comprise rare heterogenous group of disorders involving structures developing from primordial external germ layer like hair, teeth, nail and sweat glands. Hypohidrotic ED is a rare syndrome with incidence of 1:100000. Males are more commonly affected. Triad of signs comprising sparse hair, abnormal or missing teeth and inability to sweat are common. Here we present a case report of a female child with Hypohidrotic ED.

Observation: A 5 year old girl presented with sparse hair since 2 years of age. On examination skin was dry and wrinkled, with sparse thin hair, loss of eye brows and eye lashes, peg shaped tooth, oral mucosa and palate normal. History of reduced sweating and heat intolerance noted. History of consanguinity in parents and no other siblings had similar history. Developmental milestones were normal. Physical, mental development and systemic examination was normal. Diagnosis of Hypohidrotic ED was made on basis of history and clinical features.

Key message: Management of Hypohidrotic ED is a challenge for dermatologists. Stresses on importance of genetic counseling and psychological support.