EDA mutation as a cause of hypohidrotic ectodermal dysplasia: a case report and review of the literature

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ABSTRACT. Ectodermal dysplasia (ED) represents a collection of rare disorders that result from a failure of development of the tissues derived from the embryonic ectoderm. ED is often associated with hair, teeth, and skin abnormalities, which are serious conditions affecting the quality of life of the patient. To date, a large number of genes have been found to be associated with this syndrome. Here, we report a patient with hypohidrotic ED (HED) without family history. We identified that this patient’s disorder arises from an X-linked HED with a mutation in the EDA gene (G299D) found by whole-exome sequencing. In addition, in this paper we summarize the disease-causing mutations based on current literature. Overall, recent clinical and genetic research involving patients with HED have uncovered a large number of pathogenic mutations in EDA, which might contribute to
a full understanding of the function of *EDA* and the underlying mechanisms of HED caused by *EDA* mutations.

**Key words:** Hypohidrotic ectodermal dysplasias; Whole exome sequencing; *EDA*