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Missense mutation of the *EDA* gene in a Jordanian family with X-linked hypohidrotic ectodermal dysplasia: phenotypic appearance and speech problems

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ABSTRACT. Mutations in the *EDA* gene are responsible for X-linked hypohidrotic ectodermal dysplasia, the most common form of ectodermal dysplasia. Males show a severe form of this disease, while females often manifest mild to moderate symptoms. We identified a missense mutation (c.463C>T) in the *EDA* gene in a Jordanian family, using direct DNA sequencing. This mutation leads to an amino acid change of arginine to cysteine in the extracellular domain of ectodysplasin-A, a protein encoded by the *EDA* gene. The phenotype of a severely affected 11-year-old boy with this mutation included heat intolerance, sparse hair (hypotrichosis), absence of 17 teeth (oligodontia), speech problems, and damaged eccrine glands, resulting in reduced sweating (anhidrosis). Both the mother (40 years old) and the sister (10 years old)

were carriers with mild to moderate symptoms of this disease, while the father was healthy. This detailed description of the phenotype caused by this missense mutation could be useful for prenatal diagnosis.

Key words: EDA; Mutation; Ectodermal dysplasia; Jordan

