

## Sample Abstract

### *Anhydrotic Ectodermal Dysplasia in Omani Families*

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*Hypohidrotic (anhidrotic) ectodermal dysplasia (HED), a genetic disorder characterized by defective development of hair, teeth, and eccrine sweat glands, is usually inherited as an X-linked recessive trait mapped to the X-linked ectodermal dysplasia locus, EDA, at Xq12-q13.1. The existence of an autosomal recessive form of the disorder had been proposed but subsequently had been challenged by the hypothesis that the phenotype of severely affected daughters born to unaffected mothers in these rare families may be due to marked skewing of X inactivation. Consanguineous Omani families with possible autosomal recessive HED have been identified on the basis of the presence of severely affected male and female siblings and unaffected parents. Homozygous mutation in the EDAR gene on chromosome 2, and mutation of the EDA gene on the X chromosome were found in Omani families. The recognition of non-allelic genetic heterogeneity and molecular genetic testing are crucial for genetic counseling.*

*Keywords: Hypohidrotic ectodermal dysplasia, X-linked, genetic mapping, consanguinity, Oman.*

*Category: 1*