

## Condition: Ectodermal dysplasia

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### Inheritance:

Genetically heterogeneous.

### Genetic etiology:

There are multiple genes known to be responsible for various forms of ectodermal dysplasia. The hypohidrotic form is X-linked and due to mutation in the *EDA* gene. A rarer form of hypohidrotic ectodermal dysplasia is due to mutation in the *EDAR* or *EDARDD* genes, and can be inherited both as autosomal recessive and autosomal dominant traits. Hidrotic ectodermal dysplasia type 2 is inherited as an autosomal dominant trait and is due to mutation in the gene *GJB6*, which encodes a gap junction protein. There are many other forms of ectodermal dysplasia, usually associated with additional features, such as ectodermal dysplasia with immune deficiency, due to mutation of the *IKBKG* gene on the X chromosome.

### Frequency:

HED2 has relatively high frequency among French-Canadians due to a founder effect. Hypohidrotic ectodermal dysplasia affects 1/5,000 – 1/10,000 males.

### Clinical features:

Hypohidrotic ectodermal dysplasia is characterized by the triad of hypotrichosis (deficient hair), hypodontia (absent, small, misshapen teeth), and hypohidrosis (deficient sweating). The major features of hidrotic ectodermal dysplasia are deficient hair, hyperkeratosis of the palms and soles, and dystrophic nails.

### Management:

Symptomatic treatment; avoidance of hyperthermia for hypohidrotic forms.

### Genetic counseling:

Depends on mode of genetic transmission (autosomal or sex-linked, dominant or recessive); genetic testing is available for the major forms of the disorder.