# EDA Gene

Subjects: Genetics
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## Definition

Ectodysplasin A: The EDA gene provides instructions for making a protein called ectodysplasin A.

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## 1. Normal Function

This protein is part of a signaling pathway that plays an important role in development before birth. Specifically, it is critical for interactions between two embryonic cell layers called the ectoderm and the mesoderm. In the early embryo, these cell layers form the basis for many of the body's organs and tissues. Ectoderm-mesoderm interactions are essential for the formation of several structures that arise from the ectoderm, including the skin, hair, nails, teeth, and sweat glands.

One version of the ectodysplasin A protein, known as ectodysplasin A1, interacts with a protein called the ectodysplasin A receptor (produced from the *EDAR* gene). On the cell surface, ectodysplasin A1 attaches to this receptor like a key in a lock. When these two proteins are connected, they trigger a series of chemical signals that affect cell activities such as division, growth, and maturation. Starting before birth, this signaling pathway controls the formation of ectodermal structures such as hair follicles, sweat glands, and teeth.

## 2. Health Conditions Related to Genetic Changes

### 2.1 Hypohidrotic Ectodermal Dysplasia

More than 300 mutations in the *EDA* gene have been found to cause hypohidrotic ectodermal dysplasia, the most common form of ectodermal dysplasia. Starting before birth, ectodermal dysplasias result in the abnormal development of the skin, hair, nails, teeth, and sweat glands. Hypohidrotic ectodermal dysplasia is characterized by a reduced ability to sweat (hypohidrosis), sparse scalp and body hair (hypotrichosis), and several missing teeth (hypodontia) or teeth that are malformed. *EDA* gene mutations are the most frequent cause of hypohidrotic ectodermal dysplasia, accounting for more than half of all cases.

Some mutations in the *EDA* gene change single DNA building blocks (base pairs), whereas other mutations insert or delete a larger section of DNA. These changes lead to the production of a nonfunctional version of the ectodysplasin A1 protein. A shortage of functional ectodysplasin A1 prevents the protein from interacting effectively with its receptor, which impairs chemical signaling needed for interactions between the ectoderm and the mesoderm in early development. Without these signals, hair follicles, teeth, sweat glands, and other ectodermal structures do not form properly, which leads to the characteristic features of hypohidrotic ectodermal dysplasia.

### 2.2 Other Disorders

*EDA* gene mutations have also been reported in some people with a condition called nonsyndromic tooth agenesis. This condition causes one or more teeth not to form. Although missing teeth is a common feature of ectodermal dysplasias, "nonsyndromic" suggests that in these cases tooth agenesis occurs without the other signs and symptoms of those conditions. It is unclear why the effects of some mutations in this gene appear to be limited to tooth development, while other mutations affect the formation of multiple ectodermal tissues.

## 3. Other Names for This Gene

- Ectodermal dysplasia protein

- ectodysplasin
- ectodysplasin-A
- ED1
- ED1-A1
- EDA-A1
- EDA-A2
- EDA1
- EDA_HUMAN
- HED
- XHED
- XLHED

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## Keywords

genes