EDAR Gene

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Ectodysplasin A receptor: The EDAR gene provides instructions for making a protein called the ectodysplasin A receptor.

Keywords: genes

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.

The ectodysplasin A receptor interacts with a protein called ectodysplasin A1 (produced from the *EDA* 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.

Studies suggest that common variations (polymorphisms) in the *EDAR* gene are associated with the thickness and straightness of scalp hair, particularly in East Asian populations. *EDAR* appears to be one of many genes that influence these hair traits.

2. Health Conditions Related to Genetic Changes

2.1 Hypohidrotic Ectodermal Dysplasia

More than 50 mutations in the *EDAR* 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. *EDAR* gene mutations account for about 10 percent of all cases of hypohidrotic ectodermal dysplasia.

Most of the *EDAR* gene mutations associated with hypohidrotic ectodermal dysplasia change a single protein building block (amino acid) in the receptor protein. Some of the mutations that cause this condition lead to the production of an abnormal version of the ectodysplasin A receptor. Other mutations prevent cells from producing any functional receptor. All of these genetic changes prevent the receptor from interacting with ectodysplasin A1, 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

EDAR 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

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- ectodysplasin 1, anhidrotic receptor
- ectodysplasin A1 isoform receptor
- ectodysplasin receptor
- ED1R
- ED3
- ED5
- EDA-A1R
- EDA1R
- EDA3
- EDAR_HUMAN

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