EDA gene
ectodysplasin A

Normal Function

The EDA gene provides instructions for making a protein called ectodysplasin A. 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 EDA gene provides instructions for producing many slightly different versions of ectodysplasin A. One version, 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. Before birth, this signaling pathway controls the formation of ectodermal structures such as hair follicles, sweat glands, and teeth.

Health Conditions Related to Genetic Changes

Hypohidrotic ectodermal dysplasia

More than 80 different mutations in the EDA gene have been identified in people with hypohidrotic ectodermal dysplasia. These mutations cause the X-linked form of the disorder, which accounts for 95 percent of all cases of hypohidrotic ectodermal dysplasia. (X-linked disorders are caused by mutations in genes on the X chromosome, one of the two sex chromosomes.)

Some mutations in the EDA gene change single DNA building blocks (base pairs), whereas other mutations insert or delete genetic material in the gene. These changes lead to the production of a nonfunctional version of the ectodysplasin A protein. This abnormal protein cannot trigger chemical signals needed for normal interactions between the ectoderm and the mesoderm. Without these signals, hair follicles, teeth, sweat glands, and other ectodermal structures do not form properly, leading to the characteristic features of hypohidrotic ectodermal dysplasia.
**Chromosomal Location**

Cytogenetic Location: Xq13.1, which is the long (q) arm of the X chromosome at position 13.1

Molecular Location: base pairs 69,616,067 to 70,039,472 on the X chromosome (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

**Other Names for This Gene**

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

**Additional Information & Resources**

**Educational Resources**

- Madame Curie Bioscience Database: Eda/Edar Signaling
  https://www.ncbi.nlm.nih.gov/books/NBK6103/#A21779

**Clinical Information from GeneReviews**

- Hypohidrotic Ectodermal Dysplasia
  https://www.ncbi.nlm.nih.gov/books/NBK1112
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28EDA%5BTIAB%5D%29+OR+%28ectodysplasin+A%5BTIAB%5D%29%29+OR+%28%28ectodysplasin-A%5BTIAB%5D%29+OR+%28ED1%5BTIAB%5D%29%29+OR+%28EDA1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ECTODYSPLASIN A
  http://omim.org/entry/300451
- TOOTH AGENESIS, SELECTIVE, X-LINKED, 1
  http://omim.org/entry/313500

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_EDA.html
- ClinVar
- HGNC Gene Family: Tumor necrosis factor superfamily
  https://www.genenames.org/cgi-bin/genefamilies/set/781
- HGNC Gene Symbol Report
  https://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgncc_data.php&hgnc_id=3157
- Monarch Initiative
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/Q92838
Sources for This Summary


