DSG4 gene
desmoglein 4

Normal Function

The DSG4 gene provides instructions for making a protein called desmoglein 4 (DSG4). This protein is found in specialized structures called desmosomes that are located in the membrane surrounding certain cells. Desmosomes help attach cells to one another and play a role in communication between cells. The DSG4 protein is found in cells in certain regions of hair follicles, including the inner compartment of the hair strand (shaft) known as the cortex. Hair growth occurs at the hair follicle when cells divide and the hair shaft is pushed upward and extends beyond the skin.

Desmosomes provide strength to the hair and are involved in signaling between neighboring cells within the hair shaft. The DSG4 protein may play a role in communicating the signals for cells to mature (differentiate) and form the hair shaft. In addition, the DSG4 protein is found in the upper layers of the skin where it provides strength and communicates signals for the skin cells to mature.

Health Conditions Related to Genetic Changes

Autosomal recessive hypotrichosis

At least 10 mutations in the DSG4 gene have been found to cause autosomal recessive hypotrichosis, a condition that results in sparse hair growth (hypotrichosis) on the scalp, and less frequently, other parts of the body. A particular mutation that deletes a piece of genetic material in the DSG4 gene (written as Ex5_8) is a common cause of the condition in individuals of Pakistani ancestry. This mutation impairs the protein's ability to help cells attach to one another. Other DSG4 gene mutations result in the production of abnormal DSG4 proteins that cannot communicate signals between cells within hair follicles or skin. As a result, hair follicles are structurally abnormal and often underdeveloped. Irregular hair follicles alter the structure and growth of hair shafts, leading to fragile hair that breaks easily. A lack of normal DSG4 protein function may weaken the skin and contribute to the skin problems sometimes seen in individuals with autosomal recessive hypotrichosis.

Monilethrix

Mutations in the DSG4 gene have been found in people with monilethrix, a hair condition characterized by strands of hair with a beaded appearance. The hair is also short, brittle and breaks easily. The mutations associated with this condition can affect any part of the DSG4 protein, but these changes typically alter the extracellular domain, which is the region of the protein outside the cell that interacts with other cells. In people with monilethrix, the cortex of the affected hair shaft
appears abnormal. However, it is unclear how mutations in the DSG4 gene are related to the abnormality in the cortex or the beaded appearance of the hair.

It is unknown why some individuals with DSG4 gene mutations develop monilethrix and others develop autosomal recessive hypotrichosis (described above). These conditions may represent different forms of the same disorder.

**Chromosomal Location**

Cytogenetic Location: 18q12.1, which is the long (q) arm of chromosome 18 at position 12.1

Molecular Location: base pairs 31,376,777 to 31,415,791 on chromosome 18 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)

**Other Names for This Gene**
- cadherin family member 13
- CDGF13
- CDH family member 13
- CDHF13
- desmoglein-4
- DSG4_HUMAN
- LAH

**Additional Information & Resources**

**Educational Resources**
  https://www.ncbi.nlm.nih.gov/books/NBK26857/#A3488
Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DSG4%5BTIAB%5D%29+OR+%28desmoglein+4%5BTIAB%5D%29+OR+%28desmoglein-4%5BTIAB%5D%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- DESMOGLEIN 4
  http://omim.org/entry/607892

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DSG4%5Bgene%5D

- HGNC Gene Symbol Report

- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:147409

- NCBI Gene

- UniProt
  https://www.uniprot.org/uniprot/Q86SJ6

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16533311

- OMIM: DESMOGLEIN 4
  http://omim.org/entry/607892

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21495994

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16770573


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