DSC2 gene

desmocollin 2

Normal Function

The *DSC2* gene provides instructions for making a protein called desmocollin-2. This protein is found in many tissues, although it appears to be particularly important in the heart muscle and skin. Desmocollin-2 is a major component of specialized structures called desmosomes. These structures help hold neighboring cells together, which provides strength and stability to tissues. Desmosomes may also be involved in other critical cell functions, including chemical signaling pathways, the process by which cells mature to perform specific functions (differentiation), and the self-destruction of cells (apoptosis).

Health Conditions Related to Genetic Changes

Keratoderma with woolly hair

At least one mutation in the *DSC2* gene has been found to cause a form of keratoderma with woolly hair classified as type III. It is characterized by thick, calloused skin on the palms of the hands and soles of the feet (palmoplantar keratoderma); coarse, dry, fine, and tightly curled hair; and a potentially life-threatening form of heart disease called arrhythmogenic right ventricular cardiomyopathy (ARVC).

The known mutation, which is written as 1841delG, deletes one DNA building block (base pair) from the *DSC2* gene. The resulting abnormal version of the desmocollin-2 protein alters the structure of desmosomes, preventing cells from attaching to one another effectively. Researchers suspect that the impaired connections between cells make the skin, hair, and heart muscle more fragile. Over time, as these tissues are exposed to mechanical stress (for example, friction on the surface of the skin or the constant contraction and relaxation of the heart muscle), they become damaged and can no longer function normally. This mechanism probably underlies the skin, hair, and heart problems that occur in keratoderma with woolly hair type III. Studies suggest that abnormal cell signaling may also contribute to cardiomyopathy in people with this condition.

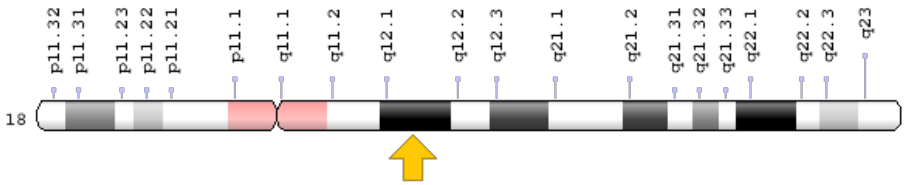
Several other mutations in the *DSC2* gene cause ARVC without palmoplantar keratoderma or woolly hair. It is unclear why some mutations in this gene affect the skin and hair, while others do not.

Arrhythmogenic right ventricular cardiomyopathy

Chromosomal Location

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

Molecular Location: base pairs 31,065,974 to 31,102,432 on chromosome 18 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARVD11
- cadherin family member 2
- CDHF2
- desmocollin-2 isoform Dsc2a preproprotein
- desmocollin-2 isoform Dsc2b preproprotein
- desmosomal glycoprotein II/III
- DG2
- DGII/III
- DSC3

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Desmosomes Connect Intermediate Filaments from Cell to Cell
  https://www.ncbi.nlm.nih.gov/books/NBK26857/#A3488
- Molecular Cell Biology (first edition, 2000): Desmosomes
  https://www.ncbi.nlm.nih.gov/books/NBK21599/#A6512

Clinical Information from GeneReviews

- Arrhythmogenic Right Ventricular Cardiomyopathy
  https://www.ncbi.nlm.nih.gov/books/NBK1131

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DSC2%5BTI%5D%29+OR+%28desmocollin+2%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, 11
  http://omim.org/entry/610476
- DESMOCOLLIN 2
  http://omim.org/entry/125645

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DSC2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DSC2%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:3036
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:1824
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1824
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