SMARCAD1 gene

SWI/SNF-related, matrix-associated actin-dependent regulator of chromatin, subfamily a, containing DEAD/H box 1

Normal Function

The *SMARCAD1* gene provides instructions for making two versions (isoforms) of the SMARCAD1 protein: a full-length isoform and a shorter, skin-specific isoform. The full-length isoform is active (expressed) in multiple tissues, where it regulates the activity of a wide variety of genes involved in maintaining the stability of cells' genetic information. The skin-specific isoform is expressed only in skin cells, and little is known about its function. However, it appears to play a critical role in the formation of dermatoglyphs, which are the patterns of skin ridges on the pads of the fingers that form the basis for each person's unique fingerprints. These ridges are also present on the toes, the palms of the hands, and the soles of the feet. Dermatoglyphs develop before birth and remain the same throughout life. The activity of the skin-specific isoform of the SMARCAD1 protein is likely one of several factors that determine each person's unique fingerprint pattern.

Health Conditions Related to Genetic Changes

Adermatoglyphia

At least four mutations in the *SMARCAD1* gene have been found to cause adermatoglyphia, which is the absence of dermatoglyphs on the hands and feet. Because affected individuals do not have skin ridges on the pads of their fingers, they cannot be identified on the basis of their fingerprints. Adermatoglyphia can occur without any related signs and symptoms, or it may be associated with other features, typically affecting the skin.

The mutations that cause adermatoglyphia affect the skin-specific isoform of the SMARCAD1 protein but not the full-length isoform. These genetic changes prevent the production of any functional skin-specific isoform from one copy of the gene, which reduces the total amount of this protein in skin cells. Although it is unclear how these genetic changes cause adermatoglyphia, researchers speculate that a shortage of the skin-specific version of the SMARCAD1 protein impairs signaling pathways needed for normal skin development and function, including the formation of dermatoglyphs.

Other Names for This Gene

- ADERM
- ATP-dependent helicase 1
- DKFZP762K2015
- ETL1
- HEL1
- KIAA1122
- SMRCD_HUMAN

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SMARCAD1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=56916[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SMARCAD1%5BTIAB%5D%29+OR+%28HEL1%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- SWI/SNF-RELATED, MATRIX-ASSOCIATED ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A, DEAD/H BOX-CONTAINING, 1; SMARCAD1 (https://omim.org/entry/612761)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/56916)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SMARCAD1[gene])

References

- Adra CN, Donato JL, Badovinac R, Syed F, Kheraj R, Cai H, Moran C, Kolker MT, Turner H, Weremowicz S, Shirakawa T, Morton CC, Schnipper LE, Drews R. SMARCAD1,a novel human helicase family-defining member associated with geneticinstability: cloning, expression, and mapping to 4q22-q23, a band rich inbreakpoints and deletion mutants involved in several human diseases. Genomics. 2000 Oct 15;69(2):162-73. doi: 10.1006/geno.2000.6281. Citation on PubMed (https:

//pubmed.ncbi.nlm.nih.gov/11031099)

- Burger B, Fuchs D, Sprecher E, Itin P. The immigration delay disease: adermatoglyphia-inherited absence of epidermal ridges. J Am Acad Dermatol. 2011May;64(5):974-80. doi: 10.1016/j.jaad.2009.11.013. Epub 2010 Jul 8. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20619487)
- Costelloe T, Louge R, Tomimatsu N, Mukherjee B, Martini E, Khadaroo B, DuboisK, Wiegant WW, Thierry A, Burma S, van Attikum H, Llorente B. The yeast Fun30 andhuman SMARCAD1 chromatin remodellers promote DNA end resection. Nature. 2012 Sep27;489(7417):581-4. doi: 10.1038/nature11353. Epub 2012 Sep 9. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22960744) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3493121/)
- Nousbeck J, Burger B, Fuchs-Telem D, Pavlovsky M, Fenig S, Sarig O, Itin P, Sprecher E. A mutation in a skin-specific isoform of SMARCAD1 causesautosomal-dominant adermatoglyphia. Am J Hum Genet. 2011 Aug 12;89(2):302-7. doi:10.1016/j.ajhg.2011.07.004. Epub 2011 Aug 4. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21820097) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3155166/)
- Nousbeck J, Sarig O, Magal L, Warshauer E, Burger B, Itin P, Sprecher E. Mutations in SMARCAD1 cause autosomal dominant adermatoglyphia and perturb theexpression of epidermal differentiation-associated genes. Br J Dermatol. 2014Dec;171(6):1521-4. doi: 10.1111/bjd.13176. Epub 2014 Oct 26. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/24909267)
- Okazaki N, Ikeda S, Ohara R, Shimada K, Yanagawa T, Nagase T, Ohara O, Koga H.The novel protein complex with SMARCAD1/KIAA1122 binds to the vicinity of TSS. JMol Biol. 2008 Oct 3;382(2):257-65. doi: 10.1016/j.jmb.2008.07.031. Epub 2008 Jul17. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/18675275)
- Rowbotham SP, Barki L, Neves-Costa A, Santos F, Dean W, Hawkes N, Choudhary P,Will WR, Webster J, Oxley D, Green CM, Varga-Weisz P, Mermoud JE. Maintenance ofsilent chromatin through replication requires SWI/SNF-like chromatin remodelerSMARCAD1. Mol Cell. 2011 May 6;42(3):285-96. doi: 10.1016/j.molcel.2011.02.036. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21549307)

Genomic Location

The *SMARCAD1* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

Last updated April 1, 2015