SMARCE1 gene

SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily e, member 1

Normal Function

The *SMARCE1* gene provides instructions for making a protein that forms one piece (subunit) of several different SWI/SNF protein complexes. SWI/SNF complexes regulate gene activity (expression) by a process known as chromatin remodeling. Chromatin is the network of DNA and protein that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression is regulated during development; when DNA is tightly packed, gene expression is lower than when DNA is loosely packed.

Through their ability to regulate gene activity, SWI/SNF complexes are involved in many processes, including repairing damaged DNA; copying (replicating) DNA; and controlling the growth, division, and maturation (differentiation) of cells.

The role of the SMARCE1 protein within the SWI/SNF complex is not completely understood.

Health Conditions Related to Genetic Changes

Coffin-Siris syndrome

At least five variants (also known as mutations) in the *SMARCE1* gene cause Coffin-Siris syndrome, which is characterized by delayed development, abnormalities of the fifth (pinky) fingers or toes, and characteristic facial features that are described as coarse. Most *SMARCE1* gene variants involved in Coffin-Siris syndrome change single protein building blocks (amino acids) in the SMARCE1 protein. Although it is unclear how these changes affect SWI/SNF complexes, researchers suggest that the variants result in abnormal chromatin remodeling. Disturbance of this process alters the activity of many genes and disrupts several cellular processes, which could explain the diverse signs and symptoms of Coffin-Siris syndrome.

Other Names for This Gene

- BAF57
- BRG1-associated factor 57

- chromatin remodeling complex BRG1-associated factor 57
- SMCE1_HUMAN
- SWI/SNF-related matrix-associated actin-dependent regulator of chromatin e1
- SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily E member 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SMARCE1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6605[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28SMARCE1%5BTIAB%5D%29+OR+%28%28BAF57%5BTIAB%5D%29+OR+%28BRG1-associated+factor+57%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+2880+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY E, MEMBER 1; SMARCE1 (https://omim.org/entry/603111)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6605)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=SMARCE1[gene])

References

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- Santen GW, Kriek M, van Attikum H. SWI/SNF complex in disorder: SWItching frommalignancies to intellectual disability. Epigenetics. 2012 Nov;7(11):1219-24.doi: 10.4161/epi.22299. Epub 2012 Sep 25. Citation on PubMed (https://pubmed.ncbi.nl

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- Tsurusaki Y, Okamoto N, Ohashi H, Kosho T, Imai Y, Hibi-Ko Y, Kaname T, Naritomi K, Kawame H, Wakui K, Fukushima Y, Homma T, Kato M, Hiraki Y, YamagataT, Yano S, Mizuno S, Sakazume S, Ishii T, Nagai T, Shiina M, Ogata K, Ohta T,Niikawa N, Miyatake S, Okada I, Mizuguchi T, Doi H, Saitsu H, Miyake N, MatsumotoN. Mutations affecting components of the SWI/SNF complex cause Coffin-Sirissyndrome. Nat Genet. 2012 Mar 18;44(4):376-8. doi: 10.1038/ng.2219. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/22426308)

Genomic Location

The *SMARCE1* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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