

SMARCAL1 gene

SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a like 1

Normal Function

The *SMARCAL1* gene provides instructions for producing a protein whose specific function is unknown. The SMARCAL1 protein can attach (bind) to chromatin, which is the complex of DNA and protein that packages DNA into chromosomes. Based on the function of similar proteins, the SMARCAL1 protein is thought to influence the activity (expression) of other genes through a process known as chromatin remodeling. 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.

Health Conditions Related to Genetic Changes

Schimke immuno-osseous dysplasia

More than 40 mutations in the *SMARCAL1* gene have been found to increase the risk of Schimke immuno-osseous dysplasia. The mutations associated with Schimke immuno-osseous dysplasia disrupt the usual functions of the SMARCAL1 protein or prevent the production of any functional protein. People who have mutations that cause a complete lack of functional protein tend to have a more severe form of this disorder than those who have mutations that lead to an active but malfunctioning protein. Mutations in the *SMARCAL1* gene are thought to lead to disease by affecting protein activity, protein stability, or the protein's ability to bind to chromatin. It is not clear how *SMARCAL1* mutations contribute to short stature, kidney disease, and a weakened immune system in people with Schimke immuno-osseous dysplasia. In order for people with *SMARCAL1* gene mutations to develop Schimke immuno-osseous dysplasia, other currently unknown genetic or environmental factors must also be present.

Other Names for This Gene

- HARP
- HepA-related protein
- HHARP

- SMAL1_HUMAN
- SMARCA-like protein 1
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a-like 1
- SWI/SNF-related matrix-associated actin-dependent regulator of chromatin a-like 1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SMARCAL1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=50485\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=50485[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SMARCAL1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A-LIKE PROTEIN 1; SMARCAL1 (<https://omim.org/entry/606622>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/50485>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SMARCAL1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SMARCAL1[gene]))

References

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Genomic Location

The *SMARCAL1* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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