

## SPRED1 gene

sprouty related EVH1 domain containing 1

### Normal Function

The *SPRED1* gene provides instructions for making the Spred-1 protein, which helps control (regulate) the Ras/MAPK signaling pathway. The Ras/MAPK pathway is involved in the growth and division of cells (proliferation), the process by which cells mature to carry out specific functions (differentiation), cell movement, and the self-destruction of cells (apoptosis).

The Spred-1 protein attaches (binds) to a protein called Raf, which is part of the Ras/MAPK pathway. The binding of the Spred-1 protein blocks the activation of Raf, stopping the signaling through the remainder of the Ras/MAPK pathway.

### Health Conditions Related to Genetic Changes

#### Legius syndrome

Mutations in the *SPRED1* gene cause Legius syndrome, a condition characterized by multiple café-au-lait spots, which are flat patches on the skin that are darker than the surrounding area.

Several mutations have been identified throughout the *SPRED1* gene. These mutations lead to a nonfunctional Spred-1 protein, usually because the mutated gene provides instructions for a shortened protein. The nonfunctional Spred-1 protein is unable to bind and block the activation of the Raf protein, which means the Ras/MAPK pathway is continuously active. It is unclear how mutations in the *SPRED1* gene cause the signs and symptoms of Legius syndrome.

### Other Names for This Gene

- EVH1/Sprouty domain containing protein
- FLJ33903
- hSpred1
- NFLS
- PPP1R147
- SPRE1\_HUMAN

- spread-1
- sprouty related, EVH1 domain containing 1
- sprouty-related, EVH1 domain containing 1
- sprouty-related, EVH1 domain-containing protein 1
- suppressor of Ras/MAPK activation

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of SPRED1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=161742\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=161742[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28SPRED1%5BTIAB%5D%29+OR+%28spread-1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- SPROUTY-RELATED EVH1 DOMAIN-CONTAINING PROTEIN 1; SPRED1 (<https://omim.org/entry/609291>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/161742>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SPRED1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SPRED1[gene]))

## References

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## Genomic Location

The *SPRED1* gene is found on chromosome 15 (<https://medlineplus.gov/genetics/chromosome/15/>).

**Last updated February 1, 2011**