

## **SNAI2 gene**

snail family transcriptional repressor 2

### **Normal Function**

The *SNAI2* gene (often called *SLUG*) provides the instructions for making a protein called snail 2. Snail 2 belongs to the snail protein family, which plays a role in the formation of tissues during embryonic development. The snail 2 protein is also found in most adult tissues, so it probably helps maintain the normal function of cells after birth. To carry out these roles, snail 2 attaches to critical regions of DNA and helps control the activity of particular genes. On the basis of this action, the protein is called a transcription factor.

Research indicates that the snail 2 protein is required during embryonic growth for the development of cells called neural crest cells. Neural crest cells migrate from the developing spinal cord to specific regions in the embryo and give rise to many tissues and cell types, including some nerve tissue and pigment-producing cells called melanocytes. Melanocytes produce the pigment melanin, which contributes to hair, eye, and skin color. Melanocytes are also found in certain regions of the brain and inner ear. The snail 2 protein probably plays a role in the formation and survival of melanocytes.

### **Health Conditions Related to Genetic Changes**

#### Piebaldism

One copy of the *SNAI2* gene is missing (deleted) in some cases of piebaldism, a condition characterized by white patches of skin and hair caused by a lack of pigmented cells (melanocytes). Loss of one copy of the gene probably reduces the production of the snail 2 protein. Shortage of the snail 2 protein may disrupt the development of melanocytes in certain areas of the skin and hair, causing the patchy loss of pigment.

#### Waardenburg syndrome

In some cases of Waardenburg syndrome type II, both copies of the *SNAI2* gene are missing. With no copies of the gene, the snail 2 protein is absent. Lack of snail 2 may disrupt the development of melanocytes in certain areas of the skin, hair, eyes, and inner ear, leading to hearing loss and the patchy loss of pigmentation that are characteristic features of Waardenburg syndrome type II.

## Other Names for This Gene

- neural crest transcription factor SLUG
- SLUG
- slug homolog, zinc finger protein (chicken)
- SLUG\_HUMAN
- SLUGH1
- snail 2
- snail family zinc finger 2
- snail homolog 2 (Drosophila)
- SNAIL2
- WS2D

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of SNAI2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6591\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6591[geneid]))

## Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SNAI2%5BTIAB%5D%29+OR+%28snail+homolog+2%5BTIAB%5D%29%29+OR+%28snail+2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>)

## Catalog of Genes and Diseases from OMIM

- SNAIL FAMILY TRANSCRIPTIONAL REPRESSOR 2; SNAI2 (<https://omim.org/entry/602150>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6591>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SNAI2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SNAI2[gene]))

## References

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## Genomic Location

The *SNAI2* gene is found on chromosome 8 (<https://medlineplus.gov/genetics/chromosome/8/>).

**Last updated August 16, 2022**