

## SMS gene

spermine synthase

### Normal Function

The *SMS* gene provides instructions for making an enzyme called spermine synthase. This enzyme is involved in the production of spermine, which is a type of small molecule called a polyamine. Specifically, the enzyme carries out the reaction that creates spermine from a related polyamine, spermidine.

Polyamines have many critical functions within cells. Studies suggest that these molecules play roles in cell growth and division, the production of new proteins, the repair of damaged tissues, and the controlled self-destruction of cells (apoptosis). Polyamines also appear to be necessary for normal development of the brain and other parts of the body.

### Health Conditions Related to Genetic Changes

#### Snyder-Robinson syndrome

At least four mutations in the *SMS* gene have been found to cause Snyder-Robinson syndrome, a condition characterized by intellectual disability and bone and muscle abnormalities. The condition only occurs in males. Most of the identified mutations change a single protein building block (amino acid) in the spermine synthase enzyme. All of the known mutations greatly reduce or eliminate the activity of spermine synthase, which decreases the amount of spermine in cells. A shortage of this polyamine clearly impacts normal development, including the development of the brain, muscles, and bones, but it is unknown how it leads to the specific signs and symptoms of Snyder-Robinson syndrome.

### Other Names for This Gene

- spermidine aminopropyltransferase
- spermine synthase isoform 1
- spermine synthase isoform 2
- SPMSY
- SpS
- SPSY\_HUMAN

- SRS

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of SMS ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6611\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6611[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28spermine+synthase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29>)

### Catalog of Genes and Diseases from OMIM

- SPERMINE SYNTHASE; SMS (<https://omim.org/entry/300105>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6611>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SMS\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SMS[gene]))

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## Genomic Location

The *SMS* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

**Last updated August 1, 2013**