



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

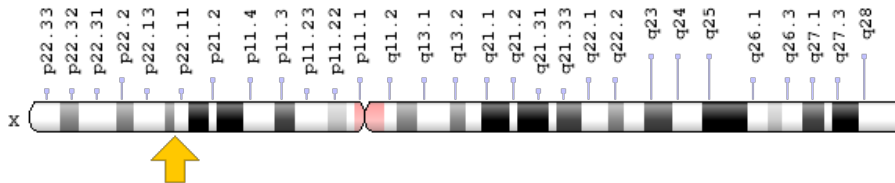
Snyder-Robinson syndrome

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.

Chromosomal Location

Cytogenetic Location: Xp22.11, which is the short (p) arm of the X chromosome at position 22.11

Molecular Location: base pairs 21,940,573 to 21,994,837 on the X chromosome (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- spermidine aminopropyltransferase
- spermine synthase isoform 1
- spermine synthase isoform 2
- SPMSY
- SpS
- SPSY_HUMAN
- SRS

Additional Information & Resources

Educational Resources

- Lund University: The Role of the Polyamines in Cell Cycle Control and Programmed Cell Death
<https://www.biology.lu.se/research/research-groups/animal-physiology/research-projects/cancer-stem-cell-research/cell-proliferation/the-role-of-the-polyamines-in-cell-cycle-control-and-programmed-cell-death>

Clinical Information from GeneReviews

- Snyder-Robinson Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK144284>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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>

Catalog of Genes and Diseases from OMIM

- SPERMINE SYNTHASE
<http://omim.org/entry/300105>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SMS.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SMS%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11123
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6611>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6611>
- UniProt
<https://www.uniprot.org/uniprot/P52788>

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