WASHC5 gene
WASH complex subunit 5

Normal Function
The WASHC5 gene provides instructions for making a protein called strumpellin. Strumpellin is active (expressed) throughout the body, although its exact function is unknown. The protein's structure suggests that stumpellin may interact with the structural framework inside cells (the cytoskeleton) and may attach (bind) to other proteins.

Health Conditions Related to Genetic Changes

Spastic paraplegia type 8
At least three mutations in the WASHC5 gene have been found to cause spastic paraplegia type 8. These mutations change single building blocks (amino acids) in the strumpellin protein. One mutation that has been seen in multiple families replaces the amino acid valine with the amino acid phenylalanine at position 626 in strumpellin (written Val626Phe or V626F). WASHC5 gene mutations are thought to change the structure of the strumpellin protein. It is unknown how the altered strumpellin protein causes muscle weakness, muscle stiffness, and other features of spastic paraplegia type 8.

Cancers
Research has shown that the WASHC5 gene is abnormally active (overexpressed) in certain types of prostate cancer. Scientists do not know what causes this abnormal expression and have not determined whether the WASHC5 gene plays a role in the development of prostate cancer.
Chromosomal Location

Cytogenetic Location: 8q24.13, which is the long (q) arm of chromosome 8 at position 24.13

Molecular Location: base pairs 125,024,260 to 125,091,819 on chromosome 8 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)

Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA0196
- MGC111053
- SPG8
- STRUM_HUMAN
- strumpellin

Additional Information & Resources

Clinical Information from GeneReviews

- Spastic Paraplegia 8
  https://www.ncbi.nlm.nih.gov/books/NBK1827

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28KIAA0196%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+3600+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- WASH COMPLEX, SUBUNIT 5
  http://omim.org/entry/610657
Research Resources

• Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_WASHC5.html

• ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=WASHC5%5Bgene%5D

• HGNC Gene Symbol Report

• Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:9897

• NCBI Gene

• UniProt
  https://www.uniprot.org/uniprot/Q12768

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9973294
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1377766/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14603436

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10797436

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17160902
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1785307/

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16130124