



## 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 strumpellin 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.

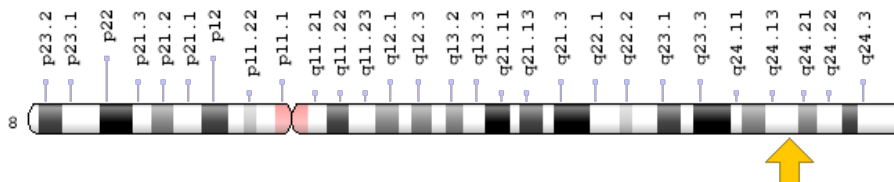
#### Cancers

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 Annotation Release 109, GRCh38.p12) (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](http://atlasgeneticsoncology.org/Genes/GC_WASHC5.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=WASHC5%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:28984](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:28984)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:9897>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/9897>
- UniProt  
<https://www.uniprot.org/uniprot/Q12768>

## **Sources for This Summary**

- Hedera P, Rainier S, Alvarado D, Zhao X, Williamson J, Otterud B, Leppert M, Fink JK. Novel locus for autosomal dominant hereditary spastic paraplegia, on chromosome 8q. *Am J Hum Genet.* 1999 Feb;64(2):563-9.  
*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/>
- Porkka KP, Tammela TL, Vessella RL, Visakorpi T. RAD21 and KIAA0196 at 8q24 are amplified and overexpressed in prostate cancer. *Genes Chromosomes Cancer.* 2004 Jan;39(1):1-10.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/14603436>
- Rocco P, Vainzof M, Froehner SC, Peters MF, Marie SK, Passos-Bueno MR, Zatz M. Brazilian family with pure autosomal dominant spastic paraplegia maps to 8q: analysis of muscle beta 1 syntrophin. *Am J Med Genet.* 2000 May 15;92(2):122-7.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/10797436>
- Valdmanis PN, Meijer IA, Reynolds A, Lei A, MacLeod P, Schlesinger D, Zatz M, Reid E, Dion PA, Drapeau P, Rouleau GA. Mutations in the KIAA0196 gene at the SPG8 locus cause hereditary spastic paraplegia. *Am J Hum Genet.* 2007 Jan;80(1):152-61. Epub 2006 Dec 1.  
*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/>
- Valdmanis PN, Meijer IA, Rouleau GA. Spastic Paraplegia 8. 2008 Aug 13 [updated 2013 Jul 25]. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Ledbetter N, Mefford HC, Smith RJH, Stephens K, editors. *GeneReviews®* [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. Available from <http://www.ncbi.nlm.nih.gov/books/NBK1827/>  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20301727>
- OMIM: WASH COMPLEX, SUBUNIT 5  
<http://omim.org/entry/610657>
- van Duin M, van Marion R, Vissers K, Watson JE, van Weerden WM, Schröder FH, Hop WC, van der Kwast TH, Collins C, van Dekken H. High-resolution array comparative genomic hybridization of chromosome arm 8q: evaluation of genetic progression markers for prostate cancer. *Genes Chromosomes Cancer.* 2005 Dec;44(4):438-49.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16130124>

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