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.

Other Names for This Gene

- KIAA0196
- MGC111053
- SPG8
- STRUM_HUMAN
- strumpellin

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of WASHC5 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9897[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?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; WASHC5 (https://omim.org/entry/610657)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/9897)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=WASHC5[gene])

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Genomic Location

The *WASHC5* gene is found on chromosome 8 (https://medlineplus.gov/genetics/chromosome/8/).

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