

ZFYVE26 gene

zinc finger FYVE-type containing 26

Normal Function

The *ZFYVE26* gene provides instructions for making a protein called spastizin, which is found in most tissues. Spastizin is important in a process called autophagy, in which worn-out cell parts and unneeded proteins are recycled within cells. Specifically, spastizin is involved in the formation and maturation of sacs called autophagosomes (or autophagic vacuoles). Autophagosomes surround materials that need to be recycled. The autophagosome then attaches (fuses) to a cell structure called a lysosome, which breaks down and recycles the materials.

In addition to being involved in clearing cells of unneeded materials, spastizin also plays a role in ensuring the proper division of cells. During the final stages of cell division, spastizin participates in the process by which the dividing cells separate from one another (cytokinesis).

Health Conditions Related to Genetic Changes

Spastic paraplegia type 15

More than 30 mutations in the *ZFYVE26* gene have been found to cause spastic paraplegia type 15. This condition is characterized by progressive movement problems, intellectual disability, and vision problems. Many of the *ZFYVE26* gene mutations that cause this condition result in a shortened spastizin protein that is quickly broken down. As a result, functional autophagosomes are not produced, autophagy cannot occur, and recycling of materials within cells is decreased. An inability to break down unneeded materials, and the subsequent accumulation of these materials in cells, leads to cell dysfunction and often cell death. The loss of cells in the brain and other parts of the body is responsible for many of the features of spastic paraplegia type 15.

It is unclear whether a lack of spastizin protein interferes with normal cytokinesis and whether impaired cell division contributes to the signs and symptoms of spastic paraplegia type 15.

Other Names for This Gene

- FYVE domain-containing centrosomal protein

- FYVE-CENT
- KIAA0321
- spastizin
- SPG15
- zinc finger FYVE domain-containing protein 26
- zinc finger, FYVE domain containing 26

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ZFYVE26 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23503\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23503[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ZFYVE26%5BTIAB%5D%29+OR+%28%28SPG15%5BTIAB%5D%29+OR+%28spastizin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>)

Catalog of Genes and Diseases from OMIM

- ZINC FINGER FYVE DOMAIN-CONTAINING PROTEIN 26; ZFYVE26 (<https://omim.org/entry/612012>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/23503>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ZFYVE26\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ZFYVE26[gene]))

References

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- Vantaggiato C, Clementi E, Bassi MT. ZFYVE26/SPASTIZIN: a close link

between complicated hereditary spastic paraparesis and autophagy. *Autophagy*. 2014 Feb;10(2):374-5. doi: 10.4161/auto.27173. Epub 2013 Nov 26. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24284334>)

- Vantaggiato C, Crimella C, Airoidi G, Polishchuk R, Bonato S, Brighina E, Scarlato M, Musumeci O, Toscano A, Martinuzzi A, Santorelli FM, Ballabio A, Bresolin N, Clementi E, Bassi MT. Defective autophagy in spastizin mutated patients with hereditary spastic paraparesis type 15. *Brain*. 2013 Oct;136(Pt10):3119-39. doi: 10.1093/brain/awt227. Epub 2013 Sep 11. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/24030950>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3784282/>)

Genomic Location

The *ZFYVE26* gene is found on chromosome 14 (<https://medlineplus.gov/genetics/chromosome/14/>).

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