

EPG5 gene

ectopic P-granules 5 autophagy tethering factor

Normal Function

The *EPG5* gene provides instructions for making a protein that is involved in a cellular process called autophagy. This process recycles worn-out or unnecessary cell parts and breaks down certain proteins when they are no longer needed. Autophagy also helps cells use materials most efficiently when energy demands are high. During autophagy, materials to be recycled or removed are isolated in compartments called autophagosomes. The autophagosomes are then transported to cell structures called lysosomes that break down the materials. The EPG5 protein is important for the interaction between autophagosomes and lysosomes that allows the transfer of materials.

In addition to its role in autophagy, the EPG5 protein aids in the cell's ability to recognize infection from foreign invaders such as bacteria and viruses. The protein transports molecules from these invaders within cells so they can interact with immune system proteins that trigger reactions to fight the infection.

Health Conditions Related to Genetic Changes

Vici syndrome

At least 60 mutations in the *EPG5* gene have been found to cause Vici syndrome, a severe condition that primarily affects the brain, immune system, heart, skin, and eyes. Most of these mutations lead to an abnormally short EPG5 protein that does not function. Without EPG5 protein activity, foreign invaders cannot trigger immune reactions, which leads to recurrent infections. In addition, autophagy is impaired. Researchers speculate that problems with autophagy disrupt the normal development and survival of cells in the brain and other organs and tissues that require large amounts of energy; however, they do not fully understand how the impairment leads to signs and symptoms of Vici syndrome.

Other Names for This Gene

- ectopic P granules protein 5 homolog
- HEEW1
- hEPG5

- KIAA1632
- VICIS

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of EPG5 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57724\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=57724[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28EPG5%5BTIAB%5D%29+OR+%28ectopic+P-granules+autophagy+protein+5+homolog%5BTIAB%5D%29%29+OR+%28%28KIAA1632%5BTIAB%5D%29+OR+%28VICIS%5BTIAB%5D%29+OR+%28ectopic+P-granules+protein+5+homolog%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+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- ECTOPIC P-GRANULES AUTOPHAGY PROTEIN 5 HOMOLOG; EPG5 (<https://omim.org/entry/615068>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/57724>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=EPG5\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=EPG5[gene]))

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Genomic Location

The *EPG5* gene is found on chromosome 18 (<https://medlineplus.gov/genetics/chromosome/18/>).

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