GFAP gene

glial fibrillary acidic protein

Normal Function

The *GFAP* gene provides instructions for making a protein called glial fibrillary acidic protein. This protein is a member of the intermediate filament family of proteins. Intermediate filaments form networks that provide support and strength to cells. Several molecules of glial fibrillary acidic protein bind together to form the type of intermediate filament found in astroglial cells. Astroglial cells support and nourish cells in the brain and spinal cord. If brain or spinal cord cells are injured through trauma or disease, astroglial cells react by rapidly producing more glial fibrillary acidic protein.

Although its function is not fully understood, glial fibrillary acidic protein is probably involved in controlling the shape, movement, and function of astroglial cells. Some researchers have suggested that astroglial cells play an important role in the functioning of other cells, including specialized cells that surround nerves (oligodendrocytes) and are involved in the production and long-term maintenance of myelin. Myelin is the fatty substance that forms a protective coating around certain nerve cells and ensures the rapid transmission of nerve impulses. Additionally, astroglial cells may assist in maintaining the protective barrier that allows only certain substances to pass between blood vessels and the brain (the blood-brain barrier).

Health Conditions Related to Genetic Changes

Alexander disease

Researchers have identified more than 50 *GFAP* mutations that cause Alexander disease. Most of these mutations change one of the building blocks (amino acids) used to make glial fibrillary acidic protein. A few mutations add or remove two amino acids in the protein. All of these changes alter the structure of glial fibrillary acidic protein. The altered protein probably disturbs the formation of normal intermediate filaments. As a result, the abnormal glial fibrillary acidic protein may accumulate in astroglial cells, contributing to the formation of Rosenthal fibers, which impair cell function. It is not well understood how impaired astroglial cells contribute to the abnormal maintenance of myelin, causing the signs and symptoms of Alexander disease.

Other Names for This Gene

- FLJ45472
- GFAP_HUMAN
- Glial Intermediate Filament Protein

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of GFAP (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=2670[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GFAP%5BTIAB%5D%29+OR+%28glial+fibrillary+acidic+protein%5BTIAB%5D%29%29+AND+%28glial+fibrillary+acidic+protein%5BMAJR%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- GLIAL FIBRILLARY ACIDIC PROTEIN; GFAP (https://omim.org/entry/137780)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/2670)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=GFAP[gene])

References

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Genomic Location

The *GFAP* gene is found on chromosome 17 (https://medlineplus.gov/genetics/chromosome/17/).

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