

ARFGEF2 gene

ARF guanine nucleotide exchange factor 2

Normal Function

The *ARFGEF2* gene provides instructions for making a protein that helps with the movement (trafficking) of small sac-like structures (vesicles) within the cell. The ARFGEF2 protein converts a molecule called guanine diphosphate (GDP) to another molecule called guanine triphosphate (GTP). This reaction activates an ADP-ribosylation factor, a molecule that is involved in vesicle trafficking. Vesicles transport many types of molecules from the interior of the cell to its surface, where they may attach and interact with other substances, or be secreted by the cell.

Health Conditions Related to Genetic Changes

Periventricular heterotopia

A few mutations in the *ARFGEF2* gene have been identified in individuals with periventricular heterotopia. These mutations may interfere with vesicle trafficking, which is important in controlling cell migration during the development of the brain. Nerve cells (neurons) that do not migrate properly during development form clumps around fluid-filled cavities (ventricles) near the center of the brain, resulting in the signs and symptoms of periventricular heterotopia.

Mutations in the *ARFGEF2* gene may also result in weakening of the attachments (adhesion) between cells that form the lining of the ventricles, by impairing the trafficking of the molecules needed for this adhesion. A weakened ventricular lining could allow some neurons to form clumps around the ventricles while others migrate normally to the exterior of the brain, as seen in periventricular heterotopia.

Other Names for This Gene

- ADP-ribosylation factor guanine nucleotide-exchange factor 2
- BIG2
- BIG2_HUMAN
- brefeldin A-inhibited guanine nucleotide-exchange protein 2

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of ARFGEF2 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10564\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10564[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ARFGEF2%5BTIAB%5D%29+OR+%28BIG2%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

- ADP-RIBOSYLATION FACTOR GUANINE NUCLEOTIDE EXCHANGE FACTOR 2; ARFGEF2 (<https://omim.org/entry/605371>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10564>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ARFGEF2\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ARFGEF2[gene]))

References

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Genomic Location

The *ARFGEF2* gene is found on chromosome 20 (<https://medlineplus.gov/genetics/chromosome/20/>).

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