

## GRIP1 gene

glutamate receptor interacting protein 1

### Normal Function

The *GRIP1* gene provides instructions for making a protein that is able to attach (bind) to other proteins and is important for moving (targeting) proteins to the correct location in cells. For example, the GRIP1 protein targets two proteins called FRAS1 and FREM2 to the correct region of the cell so that they can form a group of proteins known as the FRAS/FREM complex. This complex is found in the thin, sheet-like structures (basement membranes) that separate and support the cells of many tissues. The complex is particularly important during development before birth. One of its roles is to anchor the top layer of skin by connecting the basement membrane of the top layer to the layer of skin below. The FRAS/FREM complex is also involved in the proper development of certain other organs and tissues, including the kidneys, although the mechanism is unclear.

In addition, the GRIP1 protein targets necessary proteins to the junctions (synapses) between nerve cells (neurons) in the brain where cell-to-cell communication occurs. GRIP1 may also be involved in the development of neurons.

### Health Conditions Related to Genetic Changes

#### Fraser syndrome

At least two *GRIP1* gene mutations have been found to cause Fraser syndrome; these mutations are involved in a small percentage of cases of this condition. Fraser syndrome affects development before birth and is characterized by eyes that are completely covered by skin (cryptophthalmos), fusion of the skin between the fingers and toes (cutaneous syndactyly), and abnormalities of the kidneys and other organs and tissues.

*GRIP1* gene mutations alter the genetic blueprint that carries the instructions for making GRIP1 protein. If any GRIP1 protein is made, it is likely unable to function. Without GRIP1, FRAS1 and FREM2 do not get to the correct location to form the FRAS/FREM complex. Lack of the FRAS/FREM complex in the basement membrane of skin leads to detachment of the top layer of skin, causing blisters to form during development. These blisters likely prevent the proper formation of certain structures before birth, leading to cryptophthalmos and cutaneous syndactyly. It is unknown how lack of the FRAS/FREM

complex leads to kidney abnormalities and other problems in Fraser syndrome.

### Coloboma

MedlinePlus Genetics provides information about Coloboma

### Congenital anomalies of kidney and urinary tract

MedlinePlus Genetics provides information about Congenital anomalies of kidney and urinary tract

## **Other Names for This Gene**

- glutamate receptor-interacting protein 1
- GRIP

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of GRIP1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23426\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23426[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GRIP1%5BTIAB%5D%29+OR+%28glutamate+receptor+interacting+protein+1%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+1800+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- GLUTAMATE RECEPTOR-INTERACTING PROTEIN 1; GRIP1 (<https://omim.org/entry/604597>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/23426>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=GRIP1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=GRIP1[gene]))

## **References**

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## Genomic Location

The *GRIP1* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

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