

## ADGRG1 gene

adhesion G protein-coupled receptor G1

### Normal Function

The *ADGRG1* gene, formerly known as *GPR56*, provides instructions for making a protein that is critical for normal brain development. Before birth, the ADGRG1 protein appears to be essential for the normal growth and movement (migration) of nerve cells (neurons) in a part of the brain called the cerebral cortex. This outer layer of the brain carries out many important functions, such as sensation, voluntary muscle movement, thought, planning, and memory.

Although the ADGRG1 protein has been studied most extensively in the brain, it is active in many of the body's tissues. This protein interacts with other proteins on the cell surface to trigger a series of chemical signals within the cell. Studies suggest that ADGRG1 signaling may play an important role in attaching cells to one another (cell adhesion).

### Health Conditions Related to Genetic Changes

#### Polymicrogyria

At least eleven mutations in the *ADGRG1* gene have been identified in people with a severe form of polymicrogyria called bilateral frontoparietal polymicrogyria (BFPP). This disorder causes intellectual disability, delayed development, problems with speech and movement, and recurrent seizures (epilepsy). Most of the identified mutations change a single protein building block (amino acid) in the ADGRG1 protein. These mutations interfere with the normal processing of the protein. The abnormal protein becomes trapped within the cell, where it is unable to reach the cell surface to carry out its normal signaling functions.

A loss of ADGRG1 protein function likely disrupts the normal migration of neurons in the developing brain. As a result, certain regions of the cerebral cortex—areas known as the frontal and parietal lobes—develop too many folds (called gyri), and the folds are unusually small. These brain abnormalities cause intellectual disability and the other neurological problems associated with bilateral frontoparietal polymicrogyria.

## Other Names for This Gene

- 7-transmembrane protein with no EGF-like N-terminal domains-1
- DKFZp781L1398
- EGF-TM7-like
- G protein-coupled receptor 56
- GPR56
- GPR56\_HUMAN
- TM7LN4
- TM7XN1

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of ADGRG1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9289\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=9289[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28GPR56%5BTIAB%5D%29+OR+%28G+protein-coupled+receptor+56%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%29%29>)

### Catalog of Genes and Diseases from OMIM

- ADHESION G PROTEIN-COUPLED RECEPTOR G1; ADGRG1 (<https://omim.org/entry/604110>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/9289>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ADGRG1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ADGRG1[gene]))

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## **Genomic Location**

The *ADGRG1* gene is found on chromosome 16 (<https://medlineplus.gov/genetics/chromosome/16/>).

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