



GRM6 gene

glutamate metabotropic receptor 6

Normal Function

The *GRM6* gene provides instructions for making a protein called metabotropic glutamate receptor 6 (mGluR6). This protein is a glutamate receptor, which is a type of protein that attaches (binds) to the signaling molecule glutamate on the surface of cells. The mGluR6 protein is found within the membrane that surrounds cells called bipolar cells, which are part of the light-sensitive tissue at the back of the eye (retina). Bipolar cells receive visual signals from cells called rods that are used to see in low light. Rod cells release glutamate, which then binds to mGluR6 on bipolar cells. This binding ultimately triggers bipolar cells to transmit the visual signals to other retinal cells and eventually to the brain.

Health Conditions Related to Genetic Changes

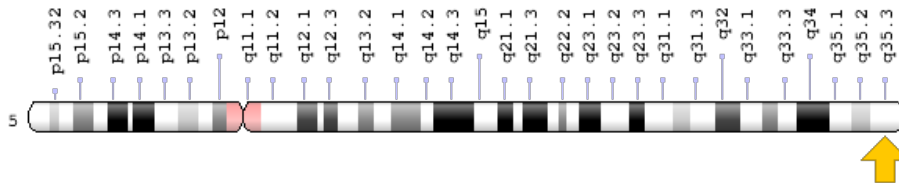
Autosomal recessive congenital stationary night blindness

At least 25 mutations in the *GRM6* gene have been found to cause autosomal recessive congenital stationary night blindness, which is characterized by the inability to see in low light and other vision problems such as nearsightedness (myopia). Most *GRM6* gene mutations impair the function of the mGluR6 protein by changing single protein building blocks (amino acids) in the protein. These mutations prevent the protein from reaching the cell membrane where it is needed to bind to glutamate. Without any mGluR6 protein at the cell surface, the glutamate released from rod cells in low light is not detected by bipolar cells, so visual signals are not transmitted. The brain does not receive the visual information sent by rods, leading to difficulty seeing in low light.

Chromosomal Location

Cytogenetic Location: 5q35.3, which is the long (q) arm of chromosome 5 at position 35.3

Molecular Location: base pairs 178,978,327 to 178,995,123 on chromosome 5 (Homo sapiens Annotation Release 109, GRCh38.p12) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- glutamate receptor, metabotropic 6
- GPRC1F
- GRM6_HUMAN
- metabotropic glutamate receptor 6
- mGlu6
- MGLUR6

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Metabotropic Receptors Modulate Synaptic Transmission
<https://www.ncbi.nlm.nih.gov/books/NBK27951/>
- Webvision: The Organization of the Retina and Visual System: Different Glutamate Receptor Types for ON and OFF Bipolar Cells
https://www.ncbi.nlm.nih.gov/books/NBK11521/#ch19bcchapter.2_Different_glutamate
- Webvision: The Organization of the Retina and Visual System: Metabotropic Glutamate Receptors
https://www.ncbi.nlm.nih.gov/books/NBK11526/#ch18glu.Metabotropic_Glutamate

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GRM6%5BTIAB%5D%29+OR+%28metabotropic+glutamate+receptor+6%5BTIAB%5D%29%29+OR+%28%28mGlu6%5BTIAB%5D%29+OR+%28MGLUR6%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>

Catalog of Genes and Diseases from OMIM

- GLUTAMATE RECEPTOR, METABOTROPIC, 6
<http://omim.org/entry/604096>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=GRM6%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:4598
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:2916>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2916>
- UniProt
<https://www.uniprot.org/uniprot/O15303>

Sources for This Summary

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<https://ghr.nlm.nih.gov/gene/GRM6>

Reviewed: January 2014
Published: June 11, 2019

Lister Hill National Center for Biomedical Communications
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