

## RGS9 gene

regulator of G protein signaling 9

### Normal Function

The *RGS9* gene provides instructions for making two versions (isoforms) of the RGS9 protein, known as RGS9-1 and RGS9-2. They are found in different parts of the nervous system and have very different functions.

RGS9-1 is produced in the retina, which is the specialized tissue at the back of the eye that detects light and color. Within the retina, RGS9-1 is associated with light-detecting cells called photoreceptors. When light enters the eye, it stimulates specialized pigments in these cells. This stimulation triggers a series of chemical reactions that produce an electrical signal, which is interpreted by the brain as vision. (This process is known as phototransduction.) Once photoreceptors have been stimulated by light, they must return to their resting state before they can be stimulated again. RGS9-1 is involved in a chemical reaction that helps return photoreceptors to their resting state quickly after light exposure.

RGS9-2 is found primarily in an area deep within the brain called the striatum. Although its exact role is unknown, RGS9-2 appears to be part of signaling pathways involving a chemical messenger (neurotransmitter) called dopamine. These pathways are important for planning and coordinating movement. Studies suggest that RGS9-2 also plays a role in the brain's response to opioid drugs, such as morphine and cocaine.

### Health Conditions Related to Genetic Changes

#### Bradyopsia

At least two mutations in the *RGS9* gene have been found to cause bradyopsia, a rare condition that affects vision. In people with bradyopsia, the eyes adapt more slowly than usual to changing light conditions (for example, walking out of a darkened movie theater into daylight or driving into a dark tunnel on a sunny day). Some affected individuals also have difficulty seeing small moving objects, such as a tennis ball.

The *RGS9* gene mutations that cause bradyopsia greatly reduce or eliminate the function of RGS9-1 in photoreceptors. (These mutations do not appear to affect the function of RGS9-2 in the brain.) A loss of RGS9-1 function prevents photoreceptors from recovering quickly after responding to light. Normally they return to their resting

state in a fraction of a second, but in people with *RGS9* gene mutations, it can take ten seconds or longer. During that time, the photoreceptors cannot respond to light. This delay causes temporary blindness in response to changing light conditions and may interfere with seeing small objects when they are in motion.

## Other Names for This Gene

- MGC111763
- MGC26458
- regulator of G-protein signaling 9
- regulator of G-protein signalling 9
- RGS9L

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of RGS9 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8787\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=8787[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28RGS9%5BTIAB%5D%29+OR+%28regulator+of+G-protein+signaling+9%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- REGULATOR OF G PROTEIN SIGNALING 9; RGS9 (<https://omim.org/entry/604067>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/8787>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RGS9\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RGS9[gene]))

## References

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## Genomic Location

The *RGS9* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

**Last updated November 1, 2014**