

## RGS9BP gene

regulator of G protein signaling 9 binding protein

### Normal Function

The *RGS9BP* gene (which is also known as *R9AP*) provides instructions for making a protein called RGS9 binding protein. This protein is found in the retina, which is the specialized tissue at the back of the eye that detects light and color. Within the retina, the protein is associated with light-detecting cells called photoreceptors. As its name suggests, RGS9 binding protein interacts with a protein called RGS9 (which is produced from the *RGS9* gene). It anchors the RGS9 protein to photoreceptors and stimulates RGS9's activity.

When light enters the eye, it stimulates specialized pigments in photoreceptor 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 and RGS9 binding protein are involved in a chemical reaction that helps return photoreceptors to their resting state quickly after light exposure.

### Health Conditions Related to Genetic Changes

#### Bradyopsia

At least six mutations in the *RGS9BP* 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 *RGS9BP* gene mutations that cause bradyopsia prevent RGS9 binding protein from anchoring the RGS9 protein to photoreceptor cells. The resulting loss of RGS9 protein 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 *RGS9BP* 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

- FLJ45744
- R9AP
- regulator of G-protein signaling 9 binding protein
- regulator of G-protein signaling 9-binding protein
- RGS9 anchor protein
- RGS9-anchoring protein

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of RGS9BP ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=388531\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=388531[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28RGS9BP%5BTIAB%5D%29+OR+%28R9AP%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

### Catalog of Genes and Diseases from OMIM

- REGULATOR OF G PROTEIN SIGNALING 9-BINDING PROTEIN; RGS9BP (<https://omim.org/entry/607814>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/388531>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=RGS9BP\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=RGS9BP[gene]))

## References

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- Nishiguchi KM, Sandberg MA, Kooijman AC, Martemyanov KA, Pott JW, Hagstrom SA, Arshavsky VY, Berson EL, Dryja TP. Defects in RGS9 or its anchor protein R9AP in patients with slow photoreceptor deactivation. *Nature*. 2004 Jan 1;427(6969):75-8. doi: 10.1038/nature02170. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14702087>)

## Genomic Location

The *RGS9BP* gene is found on chromosome 19 (<https://medlineplus.gov/genetics/chromosome/19/>).

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