

## IGFBP7 gene

insulin like growth factor binding protein 7

### Normal Function

The *IGFBP7* gene provides instructions for making a protein called insulin-like growth factor-binding protein 7 (IGFBP7). Insulin-like growth factors (IGFs) are molecules that are involved in promoting cell growth and division and preventing cells from self-destructing (undergoing apoptosis) prematurely. The IGFBP7 protein is one of a group of proteins that help control the availability of IGFs in body fluids and tissues and increase or decrease the attachment (binding) of IGFs to other molecules called receptors. The binding of IGFs and their receptors activates the cell signaling processes in which they are involved.

The IGFBP7 protein is active in the lining of blood vessels (the vascular endothelium). Its interactions with IGFs and their receptors are thought to help stop a pathway called BRAF signaling, which is involved in directing cell growth.

### Health Conditions Related to Genetic Changes

#### Retinal arterial macroaneurysm with supravulvar pulmonic stenosis

A single *IGFBP7* gene mutation has been identified in people with retinal arterial macroaneurysm with supravulvar pulmonic stenosis (RAMSVPS), a disorder that affects blood vessels in the eyes and heart. In affected individuals, the mutation occurs in both copies of the gene in each cell. The mutation, written as 830-1G>A, is called a splice-site mutation, which means that it affects the way the protein is pieced together. The protein produced from the mutated *IGFBP7* gene is abnormally short and does not function properly.

Without normally functioning IGFBP7 protein to control BRAF signaling, this signaling is increased. It is unknown how this increase is related to the specific blood vessel abnormalities that occur in RAMSVPS, or why these abnormalities are confined to the eyes and the pulmonary artery. Researchers suggest that differences in normal levels of IGFBP7 protein in various parts of the body or the presence of other proteins with a similar function in different tissues may account for the specific signs and symptoms of this disorder.

## Other Names for This Gene

- AGM
- angiomodulin
- FSTL2
- IBP-7
- IGF-binding protein 7
- IGFBP-7
- IGFBP-7v
- IGFBP-rP1
- IGFBPRP1
- insulin-like growth factor binding protein 7
- MAC25
- PGI2-stimulating factor
- prostacyclin-stimulating factor
- PSF
- RAMSVPS
- TAF
- tumor-derived adhesion factor

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of IGFBP7 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3490\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=3490[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28IGFBP7%5BTIAB%5D%29+OR+%28insulin-like+growth+factor+binding+protein+7%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+1440+days%22%5Bdp%5D%29%29>)

### Catalog of Genes and Diseases from OMIM

- INSULIN-LIKE GROWTH FACTOR-BINDING PROTEIN 7; IGFBP7 (<https://omim.org/entry/602867>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/3490>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=IGFBP7\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=IGFBP7[gene]))

## References

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## Genomic Location

The *IGFBP7* gene is found on chromosome 4 (<https://medlineplus.gov/genetics/chromosome/4/>).

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