PQBP1 gene

polyglutamine binding protein 1

Normal Function

The *PQBP1* gene provides instructions for making a protein called polyglutamine-binding protein 1. This protein attaches (binds) to stretches of multiple copies of a protein building block (amino acid) called glutamine in certain other proteins.

While the specific function of polyglutamine-binding protein 1 is not well understood, it is believed to play a role in processing and transporting RNA, a chemical cousin of DNA that serves as the genetic blueprint for the production of proteins.

In nerve cells (neurons) such as those in the brain, polyglutamine-binding protein 1 is found in structures called RNA granules. These granules allow the transport and storage of RNA within the cell. The RNA is held within the granules until the genetic information it carries is translated to produce proteins or until cellular signals or environmental factors trigger the RNA to be degraded. Through these mechanisms, polyglutamine-binding protein 1 is thought to help control the way genetic information is used (gene expression) in neurons. This control is important for normal brain development.

Health Conditions Related to Genetic Changes

Renpenning syndrome

At least 14 *PQBP1* gene mutations have been identified in people with Renpenning syndrome, a disorder that occurs almost exclusively in males and causes intellectual disability and characteristic physical features. Most of the *PQBP1* gene mutations that cause Renpenning syndrome result in an abnormally short polyglutamine-binding protein 1. The function of a shortened or otherwise abnormal protein is likely impaired and interferes with normal gene expression in neurons, resulting in abnormal development of the brain and the signs and symptoms of Renpenning syndrome.

Anophthalmia/Microphthalmia

MedlinePlus Genetics provides information about Anophthalmia/Microphthalmia

Coloboma

MedlinePlus Genetics provides information about Coloboma

Other Names for This Gene

- 38 kDa nuclear protein containing a WW domain
- nuclear protein containing WW domain 38 kD
- polyglutamine tract-binding protein 1
- polyglutamine-binding protein 1
- PQBP-1
- PQBP1_HUMAN
- RENS1

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PQBP1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10084[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28PQBP1%5BTIAB%5D%29+OR+%28polyglutamine+binding+protein+1%5BTIAB%5D%29%29+OR+%28%28MRX55%5BTIAB%5D%29+OR+%28MRXS3%5BTIAB%5D%29+OR+%28MRXS8%5BTIAB%5D%29+OR+%28NPW38%5BTIAB%5D%29+OR+%28polyglutamine+tract-binding+protein+1%5BTIAB%5D%29+OR+%28polyglutamine-binding+protein+1%5BTIAB%5D%29+OR+%28PQBP-1%5BTIAB%5D%29+OR+%28RENS1%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+1080+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- POLYGLUTAMINE-BINDING PROTEIN 1; PQBP1 (https://omim.org/entry/300463)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/10084)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PQBP1[gene])

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Genomic Location

The *PQBP1* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

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