MID1 gene

midline 1

Normal Function

The *MID1* gene is part of a group of genes called the tripartite motif (TRIM) family. Proteins produced from this large family of genes are involved in many cellular activities. Primarily, TRIM proteins play a role in the cell machinery that recycles unwanted proteins by tagging them with a protein called ubiquitin. Ubiquitin serves as a signal to move these unwanted proteins into specialized structures known as proteasomes, where the proteins are recycled.

The *MID1* gene provides instructions for making a protein called midline-1. This protein attaches (binds) to microtubules, which are rigid, hollow fibers that make up the cell's structural framework (the cytoskeleton). Microtubules help cells maintain their shape, assist in the process of cell division, and are essential for the movement of cells (cell migration). Midline-1 is responsible for recycling certain proteins, including protein phosphatase 2A (PP2A), integrin alpha-4 (ITGA4), and serine/threonine-protein kinase 36 (STK36). The recycling of these three proteins so they can be reused instead of broken down is essential because they are necessary for normal cellular functioning.

Health Conditions Related to Genetic Changes

Opitz G/BBB syndrome

About 90 mutations in the *MID1* gene have been found to cause Opitz G/BBB syndrome. This condition causes several abnormalities along the midline of the body, including widely spaced eyes (ocular hypertelorism), difficulty breathing or swallowing, brain malformations, distinct facial features, and genital abnormalities in males. The majority of the *MID1* gene mutations change a single protein building block (amino acid) in the midline-1 protein. Other mutations delete multiple amino acids and can result in the production of an abnormally short protein. These mutations lead to a decrease in midline-1 function, which prevents protein recycling. As a result, certain proteins are not recycled, and they accumulate in cells. This buildup impairs microtubule function, resulting in problems with cell division and migration. Researchers speculate that the altered midline-1 protein affects how the cells divide and migrate along the midline of the body during development, resulting in the features of Opitz G/BBB syndrome.

Other Names for This Gene

- BBBG1
- FXY
- GBBB1
- midline 1 (Opitz/BBB syndrome)
- midline 1 ring finger
- midline-1
- OGS1
- OS
- OSX
- RNF59
- TRI18_HUMAN
- TRIM18
- XPRF
- zinc finger X and Y

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MID1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4281[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MID1%5BTI%5D%29+OR+%28midline+1%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MIDLINE 1; MID1 (https://omim.org/entry/300552)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4281)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MID1[gene])

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Genomic Location

The *MID1* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

Last updated January 1, 2015