OBSL1 gene

obscurin like cytoskeletal adaptor 1

Normal Function

The *OBSL1* gene provides instructions for making a protein that is thought to help maintain normal levels of another protein called cullin-7, which is produced from the *CUL7* gene. The cullin-7 protein plays a role in the ubiquitin-proteasome system, which is the cell machinery that breaks down (degrades) unwanted proteins.

Cullin-7 helps assemble a complex known as an E3 ubiquitin ligase. This complex tags damaged and excess proteins with molecules called ubiquitin. Ubiquitin serves as a signal to specialized cell structures known as proteasomes, which attach (bind) to the tagged proteins and degrade them. The ubiquitin-proteasome system acts as the cell's quality control system by disposing of damaged, misshapen, and excess proteins. This system also regulates the level of proteins involved in several critical cell activities such as the timing of cell division and growth. In particular, the OBSL1 protein and cullin-7 are thought to help regulate proteins involved in the body's response to growth hormones, although their specific role in this process is unknown.

Health Conditions Related to Genetic Changes

3-M syndrome

At least 29 mutations in the *OBSL1* gene have been identified in people with 3-M syndrome, a disorder that causes skeletal abnormalities including short stature (dwarfism) and unusual facial features. Most of these mutations substitute one protein building block (amino acid) for another amino acid in the OBSL1 protein or result in an OBSL1 protein that is abnormally short and nonfunctional.

Mutations in the *OBSL1* gene likely lead to reduced cullin-7 protein levels, preventing cullin-7 from bringing together the components of the E3 ubiquitin ligase complex and interfering with the process of tagging unneeded proteins for degradation. The body's response to growth hormones may be impaired as a result. However, the specific relationship between *OBSL1* gene mutations and the signs and symptoms of 3-M syndrome are unknown.

Other Names for This Gene

- KIAA0657
- obscurin-like protein 1 isoform 1 precursor
- obscurin-like protein 1 isoform 2 precursor
- obscurin-like protein 1 isoform 3 precursor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of OBSL1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=23363[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28OBSL1%5BTIAB%5D%29+OR+%28obscurin+like+1%5BTIAB%5D%29%29+OR+%28%28KIAA0657%5BTIAB%5D%29+OR+%28obscurin-like+protein+1+isoform+1+precursor%5BTIAB%5D%29+OR+%28obscurin-like+protein+1+isoform+2+precursor%5BTIAB%5D%29+OR+%28obscurin-like+protein+1+isoform+3+precursor%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+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- OBSCURIN-LIKE 1; OBSL1 (https://omim.org/entry/610991)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/23363)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=OBSL1[gene])

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Genomic Location

The *OBSL1* gene is found on chromosome 2 (https://medlineplus.gov/genetics/chromosome/2/).

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