# UBE3B gene

ubiquitin protein ligase E3B

## Normal Function

The *UBE3B* gene provides instructions for making a protein that plays a role in the ubiquitin-proteasome system, which is the cell machinery that breaks down (degrades) unwanted proteins.

The UBE3B protein is called an E3 ubiquitin ligase. E3 ubiquitin ligases function as part of the ubiquitin-proteasome system by forming part of a protein complex that 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. The specific proteins tagged by complexes involving the UBE3B protein are unknown, but research suggests that the protein functions in the nervous system, digestive tract, respiratory system, and other organs and tissues, from before birth into adulthood.

## Health Conditions Related to Genetic Changes

Kaufman oculocerebrofacial syndrome

At least 15 *UBE3B* gene mutations have been identified in people with Kaufman oculocerebrofacial syndrome, which is a disorder characterized by eye problems (oculo-) , severe intellectual disability (-cerebro-), and a distinctive pattern of facial features (-facial). The mutations associated with this disorder are thought to result in an abnormal UBE3B protein that cannot function properly or that is unstable and is rapidly broken down. Loss of this protein's function likely prevents cells from eliminating certain unnecessary proteins, resulting in problems with development and function of the nervous system and other parts of the body.

## Other Names for This Gene

- BPIDS
- HECT-type ubiquitin transferase E3B
- KOS

- ubiquitin-protein ligase E3B isoform 1
- ubiquitin-protein ligase E3B isoform 3

## Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of UBE3B (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=89910[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28UBE3B%5BTIAB%5D%29+OR+%28ubiquitin+protein+ligase+E3B%5BTIAB%5D%29%29+OR+%28%28HECT-type+ubiquitin+transferase+E3B%5BTIAB%5D%29+OR+%28ubiquitin-protein+ligase+E3B+isoform+1%5BTIAB%5D%29+OR+%28ubiquitin-protein+ligase+E3B+isoform+3%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+1800+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- UBIQUITIN-PROTEIN LIGASE E3B; UBE3B (https://omim.org/entry/608047)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/89910)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=UBE3B[gene])

## References


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## Genomic Location

The *UBE3B* gene is found on chromosome 12 (https://medlineplus.gov/genetics/chromosome/12/).

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