

OTULIN gene

OTU deubiquitinase with linear linkage specificity

Normal Function

The *OTULIN* gene provides instructions for making a protein that helps regulate inflammation, which is part of the body's early immune response to foreign invaders. Inflammation occurs when the immune system sends signaling molecules and white blood cells to a site of injury or disease to fight the invaders and facilitate tissue repair. Inflammation can be turned on by a cellular process called ubiquitination, in which molecules called ubiquitin are attached to certain proteins. When foreign invaders are recognized, chains of ubiquitin molecules linked end-to-end, called linear ubiquitin chains, are attached to particular proteins. The addition of these chains stimulates signaling pathways that result in inflammation. Once the infection is under control, the body stops the inflammatory response to prevent damage to its own cells and tissues. The OTULIN protein helps control inflammation by removing linear ubiquitin chains.

In addition to inflammation, the OTULIN protein is thought to be involved in regulating development before birth and controlling cell death. Researchers are working to understand the protein's role in these processes.

Health Conditions Related to Genetic Changes

Otulipenia

At least three mutations in the *OTULIN* gene have been found to cause otulipenia, a condition that causes abnormal inflammation in the body, beginning in infancy. Affected babies have recurrent episodes of fever, diarrhea, painful joints, and skin rashes. The gene mutations involved in otulipenia reduce the function of the OTULIN protein. As a result, removal of linear ubiquitin chains is impaired, and signaling pathways that cause inflammation are abnormally active. The excessive inflammation that results causes the signs and symptoms of otulipenia and damages organs in the body; it can be life-threatening if not treated.

Other Names for This Gene

- AIPDS
- deubiquitinating enzyme otulin
- FAM105B

- family with sequence similarity 105, member B
- FLJ34884
- GUM
- OTU domain-containing deubiquitinase with linear linkage specificity
- ubiquitin thioesterase Gumby
- ubiquitin thioesterase otulin

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of OTULIN ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=90268\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=90268[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28OTULIN%5BTIAB%5D%29+OR+%28OTU+deubiquitinase+with+linear+linkage+specificity%5BTIAB%5D%29+OR+%28OTU+domain-containing+deubiquitinase+with+linear+linkage+specificity%5BTIAB%5D%29+OR+%28deubiquitinating+enzyme+otulin%5BTIAB%5D%29+OR+%28ubiquitin+thioesterase+Gumby%5BTIAB%5D%29+OR+%28ubiquitin+thioesterase+otulin%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%29>)

Catalog of Genes and Diseases from OMIM

- OTU DEUBIQUITINASE WITH LINEAR LINKAGE SPECIFICITY; OTULIN (<https://omim.org/entry/615712>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/90268>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=OTULIN\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=OTULIN[gene]))

References

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Genomic Location

The *OTULIN* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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