

## ATG16L1 gene

autophagy related 16 like 1

### Normal Function

The *ATG16L1* gene provides instructions for making a protein that is required for a process called autophagy. Cells use this process to recycle worn-out cell parts and break down certain proteins when they are no longer needed. Autophagy also plays an important role in controlled cell death (apoptosis). Additionally, autophagy is involved in the body's inflammatory response and helps the immune system destroy some types of harmful bacteria and viruses.

### Health Conditions Related to Genetic Changes

#### Crohn's disease

At least one variation in the *ATG16L1* gene is associated with an increased risk of Crohn's disease, particularly a form of the disorder that affects the lower part of the small intestine (the ileum) and the colon. This increased risk has been found primarily in people of northern European ancestry. The identified *ATG16L1* variation changes a single protein building block (amino acid) in a critical region of the ATG16L1 protein. Specifically, it replaces the amino acid threonine with the amino acid alanine at protein position 300 (written as Thr300Ala or T300A).

This change in the *ATG16L1* gene impairs the autophagy process, allowing worn-out cell parts and harmful bacteria to persist when they would otherwise be destroyed. These cell components and bacteria may trigger an inappropriate immune system response, leading to chronic inflammation in the intestinal walls and the digestive problems characteristic of Crohn's disease. Researchers continue to study the relationship between changes in the *ATG16L1* gene and a person's risk of developing this disorder.

### Other Names for This Gene

- APG16 autophagy 16-like
- APG16L
- ATG16 autophagy related 16-like 1 (*S. cerevisiae*)
- ATG16 autophagy related 16-like protein 1

- ATG16A
- ATG16L
- Autophagy 16-like 1
- autophagy related 16-like 1
- autophagy related 16-like 1 (*S. cerevisiae*)
- WD repeat domain 30
- WDR30

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of ATG16L1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55054\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55054[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ATG16L1%5BTI%5D%29+OR+%28APG16L%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%22%5D>)

### Catalog of Genes and Diseases from OMIM

- AUTOPHAGY 16-LIKE 1; ATG16L1 (<https://omim.org/entry/610767>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/55054>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ATG16L1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ATG16L1[gene]))

## **References**

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## Genomic Location

The *ATG16L1* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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