

## ABCD1 gene

ATP binding cassette subfamily D member 1

### Normal Function

The *ABCD1* gene provides instructions for producing the adrenoleukodystrophy protein (ALDP). ALDP is located in the membranes of cell structures called peroxisomes. Peroxisomes are small sacs within cells that process many types of molecules. ALDP brings a group of fats called very long-chain fatty acids (VLCFAs) into peroxisomes, where they are broken down.

### Health Conditions Related to Genetic Changes

#### X-linked adrenoleukodystrophy

More than 650 mutations in the *ABCD1* gene have been found to cause X-linked adrenoleukodystrophy. This condition is characterized by varying degrees of cognitive and movement problems as well as hormone imbalances. The mutations that cause X-linked adrenoleukodystrophy prevent the production of any ALDP in about 75 percent of people with this disorder. Other people with X-linked adrenoleukodystrophy can produce ALDP, but the protein is not able to perform its normal function. With little or no functional ALDP, VLCFAs are not broken down, and they build up in the body. The accumulation of these fats may be toxic to the adrenal glands (small glands on top of each kidney) and to the fatty layer of insulation (myelin) that surrounds many nerves in the body. Research suggests that the accumulation of VLCFAs triggers an inflammatory response in the brain, which could lead to the breakdown of myelin. The destruction of these tissues leads to the signs and symptoms of X-linked adrenoleukodystrophy.

### Other Names for This Gene

- ABCD1\_HUMAN
- ALD
- ALDP
- AMN
- ATP-binding cassette, sub-family D (ALD), member 1

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of ABCD1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=215\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=215[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ABCD1%5BTIAB%5D%29+OR+%28adrenoleukodystrophy+protein%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- ATP-BINDING CASSETTE, SUBFAMILY D, MEMBER 1; ABCD1 (<https://omim.org/entry/300371>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/215>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ABCD1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ABCD1[gene]))

## References

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## Genomic Location

The *ABCD1* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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