

OAT gene

ornithine aminotransferase

Normal Function

The *OAT* gene provides instructions for making the enzyme ornithine aminotransferase. This enzyme is active in the energy-producing centers of cells (mitochondria), where it helps break down a molecule called ornithine. Ornithine is involved in the urea cycle, which processes excess nitrogen (in the form of ammonia) that is generated when protein is broken down by the body.

In addition to its role in the urea cycle, ornithine participates in several reactions that help ensure the proper balance of protein building blocks (amino acids) in the body. This balance is important because a specific sequence of amino acids is required to build each of the many different proteins needed for the body's functions. The ornithine aminotransferase enzyme helps convert ornithine into another molecule called pyrroline-5-carboxylate (P5C). P5C can be converted into the amino acids glutamate and proline.

Health Conditions Related to Genetic Changes

Gyrate atrophy of the choroid and retina

More than 60 *OAT* gene mutations have been found to cause gyrate atrophy of the choroid and retina (often shortened to gyrate atrophy). These mutations result in a reduced amount of functional ornithine aminotransferase enzyme. A shortage of this enzyme impedes the conversion of ornithine into P5C. As a result, excess ornithine accumulates in the blood (hyperornithinemia), and less P5C than normal is produced. It is not clear how these changes result in progressive vision loss and the other features sometimes associated with gyrate atrophy. Researchers have suggested that a deficiency of P5C may interfere with the function of the retina, the specialized light-sensitive tissue that lines the back of the eye. It has also been proposed that excess ornithine may suppress the production of a molecule called creatine. Creatine is needed for many tissues in the body to store and use energy properly. It is involved in providing energy for muscle contraction, and it is also important in nervous system functioning.

Other Names for This Gene

- DKFZp781A11155
- HOGA

- OAT_HUMAN
- ornithine aminotransferase (gyrate atrophy)
- ornithine aminotransferase precursor

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of OAT ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4942\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4942[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ornithine+aminotransferase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- ORNITHINE AMINOTRANSFERASE; OAT (<https://omim.org/entry/613349>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/4942>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=OAT\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=OAT[gene]))

References

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Genomic Location

The *OAT* gene is found on chromosome 10 (<https://medlineplus.gov/genetics/chromosome/10/>).

Last updated August 1, 2009