TAT gene

tyrosine aminotransferase

Normal Function

The *TAT* gene provides instructions for making a liver enzyme called tyrosine aminotransferase. This enzyme is the first in a series of five enzymes that work to break down the amino acid tyrosine, a protein building block found in many foods. Specifically, tyrosine aminotransferase converts tyrosine into a byproduct called 4-hydroxyphenylpyruvate. Continuing the process, 4-hydroxyphenylpyruvate is further broken down and ultimately smaller molecules are produced that are either excreted by the kidneys or used to produce energy or make other substances in the body.

Health Conditions Related to Genetic Changes

Tyrosinemia

At least 22 *TAT* gene mutations have been found to cause tyrosinemia type II. This condition often affects the eyes, skin, and mental development. Most of these mutations change single DNA building blocks (base pairs) within the *TAT* gene. Research suggests that the altered *TAT* gene produces a tyrosine aminotransferase enzyme with reduced activity. Other mutations delete all or part of the *TAT* gene, eliminating enzyme activity. As a result of these mutations, tyrosine is not properly broken down. Tyrosine levels are elevated and some tyrosine is converted into other molecules that may be toxic to cells. It is unclear how impaired break down of tyrosine leads to the skin, eye, and intellectual problems that characterize tyrosinemia type II.

Other Names for This Gene

- ATTY_HUMAN
- L-tyrosine:2-oxoglutarate aminotransferase
- tyrosine transaminase

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of TAT (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6898[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28tyrosine+aminotransferase%5BTIAB%5D%29+NOT+%28glucocorticoid%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- TYROSINE AMINOTRANSFERASE; TAT (https://omim.org/entry/613018)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/6898)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=TAT[gene])

References

- Bouyacoub Y, Zribi H, Azzouz H, Nasrallah F, Abdelaziz RB, Kacem M, Rekaya B, Messaoud O, Romdhane L, Charfeddine C, Bouziri M, Bouziri S, Tebib N, Mokni M, Kaabachi N, Boubaker S, Abdelhak S. Novel and recurrent mutations in the TAT genein Tunisian families affected with Richner-Hanhart syndrome. Gene. 2013 Oct15;529(1):45-9. doi: 10.1016/j.gene.2013.07.066. Epub 2013 Aug 13. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/23954227)
- Mehere P, Han Q, Lemkul JA, Vavricka CJ, Robinson H, Bevan DR, Li J. Tyrosineaminotransferase: biochemical and structural properties and molecular dynamicssimulations. Protein Cell. 2010 Nov;1(11):1023-32. doi:10.1007/s13238-010-0128-5. Epub 2010 Dec 10. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/21153519) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3023147/)
- Sivaraman S, Kirsch JF. The narrow substrate specificity of human tyrosineaminotransferase--the enzyme deficient in tyrosinemia type II. FEBS J. 2006May;273(9):1920-9. doi: 10.1111/j.1742-4658.2006.05202.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16640556)

Genomic Location

The *TAT* gene is found on chromosome 16 (https://medlineplus.gov/genetics/chromosome/16/).

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