

# TAT Gene

Subjects: Genetics & Heredity

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Tyrosine aminotransferase: The TAT gene provides instructions for making a liver enzyme called tyrosine aminotransferase.

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## 1. 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.

## 2. Health Conditions Related to Genetic Changes

### 2.1. 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.

## 3. Other Names for This Gene

- ATTY\_HUMAN
  - L-tyrosine:2-oxoglutarate aminotransferase
  - tyrosine transaminase
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## References

1. Bouyacoub Y, Zribi H, Azzouz H, Nasrallah F, Abdelaziz RB, Kacem M, Rekaya B, Messaoud O, Romdhane L, Charfedine C, Bouziri M, Bouziri S, Tebib N, Mokni M, Kaabachi N, Boubaker S, Abdelhak S. Novel and recurrent mutations in the *TAT* gene in Tunisian families affected with Richner-Hanhart syndrome. *Gene*. 2013 Oct 15;529(1):45-9. doi: 10.1016/j.gene.2013.07.066.
  2. Mehre P, Han Q, Lemkul JA, Vavricka CJ, Robinson H, Bevan DR, Li J. Tyrosine aminotransferase: biochemical and structural properties and molecular dynamic simulations. *Protein Cell*. 2010 Nov;1(11):1023-32. doi:10.1007/s13238-010-0128-5.
  3. Sivaraman S, Kirsch JF. The narrow substrate specificity of human tyrosine aminotransferase--the enzyme deficient in tyrosinemia type II. *FEBS J*. 2006 May;273(9):1920-9.
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