

## ABAT gene

4-aminobutyrate aminotransferase

### Normal Function

The *ABAT* gene provides instructions for making the GABA-transaminase enzyme. This enzyme helps break down a brain chemical (neurotransmitter) called GABA when it is not needed. GABA normally helps slow down (inhibit) brain cell activity when necessary, to prevent the brain from being overloaded with too many signals. For this reason GABA is called an inhibitory neurotransmitter.

### Health Conditions Related to Genetic Changes

#### GABA-transaminase deficiency

At least 10 mutations in the *ABAT* gene have been identified in people with GABA-transaminase deficiency, which is a brain disease (encephalopathy) that begins in infancy. Babies with this disorder have recurrent seizures (epilepsy), movement problems, and profoundly delayed development. They may grow more rapidly in length than usual (accelerated linear growth), even though they have feeding problems and may not gain weight as quickly as expected (failure to thrive). Individuals with this disorder usually do not survive past the first 2 years of life, but some live longer into childhood.

The *ABAT* gene mutations that cause GABA-transaminase deficiency lead to a shortage (deficiency) of functional GABA-transaminase enzyme. As a result, GABA is not properly broken down, so this neurotransmitter and another molecule called beta-alanine accumulate abnormally in brain cells. This accumulation alters the balance of neurotransmitters in the brain, leading to the neurological problems characteristic of GABA-transaminase deficiency. Excess GABA also leads to abnormal release of a protein that is necessary for growth of the body's bones and tissues (growth hormone), resulting in the accelerated linear growth that sometimes occurs in this disorder.

### Other Names for This Gene

- (S)-3-amino-2-methylpropionate transaminase
- 4-aminobutyrate aminotransferase, mitochondrial precursor
- 4-aminobutyrate transaminase

- GABA aminotransferase
- GABA transaminase
- GABA transferase
- GABA-AT
- GABAT
- gamma-amino-N-butyrate transaminase
- NPD009

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of ABAT ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=18\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=18[geneid]))

### Scientific Articles on PubMed

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

### Catalog of Genes and Diseases from OMIM

- 4-AMINO BUTYRATE AMINOTRANSFERASE; ABAT (<https://omim.org/entry/137150>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/18>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ABAT\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ABAT[gene]))

## **References**

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## Genomic Location

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

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