



## PTS gene

6-pyruvoyltetrahydropterin synthase

### Normal Function

The *PTS* gene provides instructions for making an enzyme called 6-pyruvoyltetrahydropterin synthase. This enzyme is involved in the second of three steps in the production of a molecule called tetrahydrobiopterin (BH4). Other enzymes help carry out the first and third steps in this process.

Tetrahydrobiopterin plays a critical role in processing several protein building blocks (amino acids) in the body. For example, it works with the enzyme phenylalanine hydroxylase to convert an amino acid called phenylalanine into another amino acid, tyrosine. Tetrahydrobiopterin is also involved in reactions that produce chemicals called neurotransmitters, which transmit signals between nerve cells in the brain. Because it helps enzymes carry out chemical reactions, tetrahydrobiopterin is known as a cofactor.

### Health Conditions Related to Genetic Changes

#### Tetrahydrobiopterin deficiency

More than 45 mutations in the *PTS* gene have been found to cause tetrahydrobiopterin deficiency. When this condition is caused by *PTS* gene mutations, it is known as 6-pyruvoyltetrahydropterin synthase (PTS) deficiency. PTS deficiency accounts for more than half of all cases of tetrahydrobiopterin deficiency.

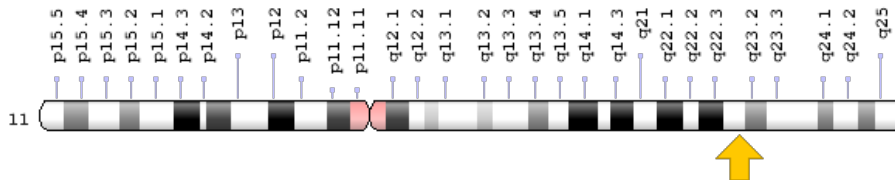
Most *PTS* gene mutations change single amino acids in 6-pyruvoyltetrahydropterin synthase, although some mutations insert or delete small amounts of DNA in the *PTS* gene or disrupt the way the gene's instructions are used to make the enzyme. Changes in 6-pyruvoyltetrahydropterin synthase greatly reduce or eliminate the enzyme's activity. Without enough of this enzyme, little or no tetrahydrobiopterin is produced. As a result, this cofactor is not available to participate in chemical reactions such as the conversion of phenylalanine to tyrosine. If phenylalanine is not converted to tyrosine, it can build up to toxic levels in the blood and other tissues. Nerve cells in the brain are particularly sensitive to phenylalanine levels, which is why excessive amounts of this substance can cause brain damage.

Additionally, a reduction in 6-pyruvoyltetrahydropterin synthase activity disrupts the production of certain neurotransmitters in the brain. Because neurotransmitters are necessary for normal brain function, changes in the levels of these chemicals contribute to intellectual disability in people with PTS deficiency.

## Chromosomal Location

Cytogenetic Location: 11q23.1, which is the long (q) arm of chromosome 11 at position 23.1

Molecular Location: base pairs 112,226,428 to 112,233,973 on chromosome 11 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- 6-pyruvoyl-H4-pterin synthase
- 6-pyruvoyl-tetrahydropterin synthase
- PTP synthase
- PTPS
- PTPS\_HUMAN
- sepiapterin synthase A
- sepiapterin synthesizing enzyme 1

## Additional Information & Resources

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%286-pyruvoyltetrahydropterin+synthase%5BTIAB%5D%29+OR+%28PTS+gene%5BTIAB%5D%29+OR+%28sepiapterin+synthase+A%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

### Catalog of Genes and Diseases from OMIM

- 6-PYRUVOYL-TETRAHYDROPTERIN SYNTHASE  
<http://omim.org/entry/612719>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=PTS%5Bgene%5D>
- HGNC Gene Symbol Report  
[https://www.genenames.org/data/gene-symbol-report#!/hgnc\\_id/HGNC:9689](https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:9689)
- Monarch Initiative  
<https://monarchinitiative.org/gene/NCBIGene:5805>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/5805>
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