

PHKB gene

phosphorylase kinase regulatory subunit beta

Normal Function

The *PHKB* gene provides instructions for making one piece, the beta subunit, of the phosphorylase b kinase enzyme. This enzyme is made up of 16 subunits, four each of the alpha, beta, gamma, and delta subunits. (Each subunit is produced from a different gene.) The beta subunit helps regulate the activity of phosphorylase b kinase. This enzyme is found in various tissues, although it is most abundant in the liver and muscles. One version of the enzyme is found in liver cells and another in muscle cells. The beta subunit produced from the *PHKB* gene is part of the enzyme found both in the liver and in muscle.

Phosphorylase b kinase plays an important role in providing energy for cells. The main source of cellular energy is a simple sugar called glucose. Glucose is stored in muscle and liver cells in a form called glycogen. Glycogen can be broken down rapidly when glucose is needed, for instance during exercise. Phosphorylase b kinase turns on (activates) another enzyme called glycogen phosphorylase b by converting it to the more active form, glycogen phosphorylase a. When active, this enzyme breaks down glycogen.

Health Conditions Related to Genetic Changes

Glycogen storage disease type IX

At least 21 mutations in the *PHKB* gene are known to cause a form of glycogen storage disease type IX (GSD IX) called GSD IXb. This form of the disorder affects the liver and the muscles. The liver problems caused by this disorder include an enlarged liver (hepatomegaly), slow growth, and periods of low blood glucose (hypoglycemia). These features usually improve over time. The condition can also cause mild muscle weakness, although some affected individuals have no muscle problems.

Mutations in the *PHKB* gene reduce the activity of phosphorylase b kinase in liver and muscle cells, although the mechanism is unknown. Reduction of this enzyme's function impairs glycogen breakdown. As a result, glycogen builds up in cells, and glucose is not available for energy. The inability to break down glycogen in the liver and reduced energy production in muscle cells lead to the features of GSD IXb.

Other Names for This Gene

- KPBB_HUMAN
- phosphorylase b kinase regulatory subunit beta
- phosphorylase kinase beta subunit
- phosphorylase kinase beta-subunit
- phosphorylase kinase subunit beta
- phosphorylase kinase, beta

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PHKB ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5257\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5257[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28PHKB%5BTIAB%5D%29+OR+%28phosphorylase+kinase+beta-subunit%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+3600+days%22%5Bdp%5D%29>)

Catalog of Genes and Diseases from OMIM

- PHOSPHORYLASE KINASE, BETA SUBUNIT; PHKB (<https://omim.org/entry/172490>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/5257>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PHKB\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PHKB[gene]))

References

- Brushia RJ, Walsh DA. Phosphorylase kinase: the complexity of its regulation is reflected in the complexity of its structure. *Front Biosci.* 1999 Sep15;4:D618-41. doi: 10.2741/brushia. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/10487978>)
- Burwinkel B, Maichele AJ, Aagenaes O, Bakker HD, Lerner A, Shin YS, Strachan JA, Kilimann MW. Autosomal glycogenosis of liver and muscle due to phosphorylase kinase deficiency is caused by mutations in the phosphorylase kinase beta subunit (PHKB). *Hum Mol Genet.* 1997 Jul;6(7):1109-15. doi: 10.1093/hmg/6.7.

1109. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9215682>)

- van den Berg IE, van Beurden EA, de Klerk JB, van Diggelen OP, Malingre HE, Boer MM, Berger R. Autosomal recessive phosphorylase kinase deficiency in liver, caused by mutations in the gene encoding the beta subunit (PHKB). *Am J Hum Genet.* 1997 Sep;61(3):539-46. doi: 10.1086/515502. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9326319>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1715950/>)

Genomic Location

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

Last updated August 1, 2015