PHKA1 gene

phosphorylase kinase regulatory subunit alpha 1

Normal Function

The *PHKA1* gene provides instructions for making one piece, the alpha 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 alpha 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 alpha-1 subunit produced from the *PHKA1* gene is part of the enzyme found in muscle cells.

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 seven mutations in the *PHKA1* gene are known to cause a form of glycogen storage disease type IX (GSD IX) called GSD IXd or X-linked muscle glycogenosis. This form of the disorder is rare and not well understood. It affects muscles and can cause muscle weakness, pain, and cramping, particularly during exercise, although some affected individuals have no signs or symptoms of the condition. Mutations in the *PHKA1* gene reduce the activity of phosphorylase b kinase in 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. Reduced energy production in muscle cells leads to the features of GSD IXd.

Other Names for This Gene

- KPB1_HUMAN

- PHKA
- phosphorylase b kinase regulatory subunit alpha, skeletal muscle isoform
- phosphorylase kinase alpha M subunit
- phosphorylase kinase, alpha 1 (muscle)
- phosphorylase kinase, alpha 1 (muscle), muscle glycogenosis

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of PHKA1 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=5255[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28PHKA1%5BTIAB%5D%29+OR+%28PHKA%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)

Catalog of Genes and Diseases from OMIM

- PHOSPHORYLASE KINASE, MUSCLE, ALPHA-1 SUBUNIT; PHKA1 (https://omim.org/entry/311870)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/5255)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=PHKA1[gene])

References

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Genomic Location

The *PHKA1* gene is found on the X chromosome (https://medlineplus.gov/genetics/chromosome/x/).

Last updated August 1, 2015