PHKB Gene

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phosphorylase kinase regulatory subunit beta

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1. Introduction

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.

2. Health Conditions Related to Genetic Changes

2.1. 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 sugar (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.

3. 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

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