PDHX Gene

Subjects: Genetics & Heredity

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pyruvate dehydrogenase complex component X

genes

1. Introduction

The *PDHX* gene provides instructions for making a protein called E3 binding protein, which is part of a large group of proteins known as the pyruvate dehydrogenase complex. This complex is made up of several enzymes, including one called E3, and other proteins. E3 binding protein attaches E3 to the complex and provides the correct structure for the complex to perform its function.

The pyruvate dehydrogenase complex plays an important role in the pathways that convert the energy from food into a form that cells can use. This enzyme converts a molecule called pyruvate, which is formed from the breakdown of carbohydrates, into another molecule called acetyl-CoA. This conversion is essential to begin the series of chemical reactions that produces adenosine triphosphate (ATP), the cell's main energy source.

2. Health Conditions Related to Genetic Changes

2.1. Pyruvate dehydrogenase deficiency

Mutations in the *PDHX* gene cause pyruvate dehydrogenase deficiency in a small number of people. This condition is characterized by a potentially life-threatening buildup of a chemical called lactic acid in the body (lactic acidosis), delayed development, and neurological problems. *PDHX* gene mutations associated with pyruvate dehydrogenase deficiency result in the complete absence of E3 binding protein. Loss of this protein impairs the binding of the E3 enzyme to the pyruvate dehydrogenase complex, which leads to a reduction of the complex's activity. With decreased function of this complex, pyruvate builds up and is converted, in another chemical reaction, to lactic acid, causing lactic acidosis. In addition, the production of cellular energy is diminished. The brain, which is especially dependent on this form of energy, is severely affected, resulting in the neurological problems associated with pyruvate dehydrogenase deficiency.

2.2. More About This Health Condition

Leigh syndrome

3. Other Names for This Gene

- dihydrolipoamide dehydrogenase-binding protein of pyruvate dehydrogenase complex
- DLDBP
- E3BP
- lipoyl-containing pyruvate dehydrogenase complex component X
- ODPX_HUMAN
- OPDX
- proX
- pyruvate dehydrogenase complex, component X
- pyruvate dehydrogenase complex, E3-binding protein subunit
- pyruvate dehydrogenase complex, lipoyl-containing component X
- pyruvate dehydrogenase protein X component, mitochondrial

References

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