Climb™

Supporting those affected by Inherited Metabolic Disorders

Pyruvate Dehydrogenase Deficiency

This is a disorder of carbohydrate metabolism, the breaking down of glucose (sugar) to provide energy for the body. In this disorder there is a deficiency of the pyruvate dehydrogenase complex which causes difficulty in converting pyruvate into acetyl-CoA, a key step in the use of glucose for energy. The complex is made up of three separate enzymes; E1, E2 and E3. The E1 enzyme contains two different components, designated alpha and beta. The most common form of Pyruvate Dehydrogenase deficiency is caused by a defect in the gene for the E1 alpha subunit. This gene is known as PDHA1. About one quarter of individuals with a condition called Leigh disease have defects in the pyruvate dehydrogenase complex.

The range and severity of symptoms varies greatly in pyruvate dehydrogenase deficiency. This is related to the level of enzyme activity in the patient, with more severe symptoms the lower the amount of functioning enzyme. With very low levels of residual function, patients may present with severe acidosis in the first few days after birth. With a more modest reduction in activity, there may be chronic moderate lactic acidaemia, often with recurrent episodes of muscle incoordination (ataxia).

Synonyms

Alternative names for this condition are:

- ataxia with lactic acidosis
- intermittent ataxia with pyruvate dehydrogenase deficiency
- PDH deficiency
- PDHC deficiency
- pyruvate dehydrogenase complex deficiency

Further information about this condition is available from Climb.

Disclaimer

Please read our disclaimer and information on data protection.

Updated 10/10/17

Telephone: **0845 241 2173** Freephone: **0800 652 3181**
Email: contact@climb.org.uk Website: www.climb.org.uk