Climb™

Supporting those affected by Inherited Metabolic Disorders

Glutaric Aciduria Type 2

Glutaric Aciduria Type 2 is a rare disorder that belongs to a group of disorders known as the Organic Acidaemias. It can be caused by a deficiency in either the electron transfer flavoprotein (ETF) enzyme or the ETF-ubiquinone oxidoreductase (ETF:QO) enzyme. This results in a build-up of organic acids in the blood and urine. There are two forms of Multiple Acyl CoA Dehydrogenase Deficiency. A neonatal form which is a complete deficiency of the enzyme and is often fatal during the newborn period and a late onset form which is less severe and can present at any age.

Symptoms of the neonatal form of this disorder include severely low blood sugar levels, low muscle tone, and respiratory distress. Those affected by this disorder may have characteristic facial features including an enlarged head, ear abnormalities, a high forehead and a flat nasal bridge. Other symptoms include an enlarged liver, kidney abnormalities, genital abnormalities and a distinctive odour. The late onset form has symptoms, which vary greatly and may include low blood sugar and periods of nausea, vomiting and weakness.

Synonyms

Alternative names for this condition are:

- Deficiency of Electron Transfer Flavoprotein
- GA II
- MADD
- Multiple Acyl-CoA Dehydrogenase Deficiency

- Further information about this condition is available from Climb.

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