Climb™

Supporting those affected by Inherited Metabolic Disorders

Primary Carnitine Deficiency

This condition is grouped with the defects known as fatty acid oxidation disorders. Fat or adipose tissue is the main energy store of our bodies derived not only from the fat that we eat but also made in the body from any excess calories obtained from our food and drink. When fat is broken down it releases fatty acids.

Our bodies rely on fatty acids as a major energy source particularly when we have not eaten for a number of hours. Mitochondria are located in virtually every cell of the body and act as the powerhouse of the cell capturing the energy obtained from the food we eat. This energy is then used to keep our bodies warm, move our muscles and enable us to grow, fuel our brains and to perform all our other bodily functions. Carnitine is a substance that is both acquired from the diet and also made within our liver and kidneys. The role of carnitine is to transport long-chain fatty acids i.e. most of the fat we eat and the fat in our adipose tissue, into the mitochondria within our cells so that these fatty acids can be broken down to release their energy. In this disorder, the protein that transports carnitine into the cells is defective or missing completely. This protein is known as the carnitine transporter protein.

When the carnitine transporter is not working carnitine cannot be transported (absorbed) into our bodies from our food and the carnitine that is made in the body cannot be transported into cells and is lost in the urine. As a result of this, carnitine is not available to transport fatty acids into the mitochondria & the body runs out of energy. The heart, which relies very heavily on fatty acids for its energy, is often affected.

Synonyms

Alternative names for this condition are:

- PCD

Further information about this condition is available from Climb.

Disclaimer

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Updated 10/10/17

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