Need to talk? Call us 0845 2412173 Monday to Friday 9am to 5pm

Climb™

Supporting those affected by Inherited Metabolic Disorders

X-Linked Creatine Deficiency

Creatine is a product of protein metabolism. It is naturally produced in the liver and also in the kidney and pancreas. It travels in the blood using the Creatine Transporter to different muscles where it is later transformed into phosphocreatine and later into adenosine triphosphate (ATP) which is one of the main sources of energy in the body. Creatine is essential for energy storage and transfer. In X-Linked Creatine Transporter Deficiency Creatine is able to be produced by the body but not transported in the blood to the brain. Creatine Deficiency is caused by a defect in one of two genes that have been identified. The first is a defect in the SLC6A8 gene, also called CT1 or CRTR gene which is predominantly found in the skeletal muscle and the kidneys and is also seen in the colon, brain, heart, testes and prostate. The second gene identified is called the SLC6A10 gene, also known as CT2, which is expressed in the testes only.

X-Linked Creatine Deficiency is a new disorder which was first reported in 2001. However, it may be more common than originally presumed with an increasing number of diagnoses. Symptoms of this disorder are predominantly neurological problems. It is characterised by learning difficulties, developmental delay, severe language delay and autistic-like behaviour.

Synonyms

Alternative names for this condition are:

- Creatine Transporter Deficiency/Defect
- SLC6A8 Deficiency
- SLC6A8-related Creatine Transporter Deficiency
- X-linked Creatine Deficiency Syndrome

Further information about this condition is available from Climb.

Disclaimer

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