Climb National Information Centre for Metabolic Diseases

Children Living with Inherited Metabolic Diseases

X-Linked Creatine Transporter Deficiency

Other names that may be used for this condition are:

- Mental Retardation, X-Linked, with Seizures, Short Stature, and Mid-Face Hypoplasia
- Mental Retardation, X-Linked, with Creatine Transport 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 located on the long arm of the X chromosome (Xq28). The SLC6A8 gene 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 has been located on the short arm of chromosome 16 (16p11) and is expressed in the testes only.

Treatment is symptomatic and supportive. High doses of creatine supplementation are unsuccessful and no clinical improvement has been seen with this treatment attempt. However, females with symptoms may respond slightly better. Genetic counselling is recommended for those affected by this disorder and their families.

This information is fully sourced and referenced, for more detailed information and references please contact CLIMB by email, letter or telephone.

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