Fumarase Deficiency

Other names that may be used for this condition are:
- Fumaric Aciduria
- Fumarate Hydratase Deficiency

Fumarase Deficiency is a rare disorder of the Krebs cycle (citric acid cycle); this is a metabolic pathway that is central to the breakdown of carbohydrates, fats and proteins into carbon dioxide and water in order to generate energy. In this cycle, Fumarase helps to convert fumarate to malate. Those affected by this disorder show postnatal neurological problems. Patients who suffer with a more severe form of Fumarase Deficiency usually develop respiratory difficulties and this results in life expectancy being reduced in early childhood, whereas patients less severely affected develop non progressive brain problems and survive into adolescence and adulthood.

Treatment depends on the severity of the condition and is mainly symptomatic and supportive. In some cases, nutritional intervention may be appropriate and physiotherapy or use of wheelchairs may be recommended. Removal of certain amino acids that are indicators of fumarate may be beneficial and daily intravenous injections are also sometimes included in treatment. Genetic counselling is recommended for those affected by this condition.

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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