Climb™

Supporting those affected by Inherited Metabolic Disorders

Galactosaemia

Galactose is a sugar which is a constituent of lactose (the form of sugar found in milk, including breast milk, and in the majority of infant milk formulas). The term Galactosaemia (increased galactose in the blood) usually refers to "classic" Galactosaemia. In Classic Galactosaemia the body's ability to convert galactose into glucose is impaired. Galactose-1-phosphate, derived from galactose, builds up in the body and becomes toxic. This disorder is caused by deficiency of a liver enzyme known as galactose-1 -phosphate uridyl transferase (GAL-1-PUT). The incidence of Classic Galactosaemia is estimated to be approximately 1:50,000 with a higher prevalence in Southern Ireland, particularly in travelling communities.

Early symptoms become apparent within a few days of birth. These include poor feeding, vomiting, poor weight gain, jaundice, and an enlarged liver. Older infants who have not been recognised and treated show irritability, listlessness, cataracts, liver failure and kidney damage. Other symptoms may include low blood sugar, and reduced growth of the head. Severe E.Coli infections in untreated infants can be life threatening. Early recognition and treatment permits complete resolution of cataract and liver disease. There is a reduction in the expected IQ, and impairment of verbal and speech skills, which appears to be unaltered with treatment. Puberty may be late or absent in females due to the ovaries not producing enough oestrogen.

Synonyms

Alternative names for this condition are:

- Classic Galactosaemia
- GALT Deficiency
- Galactose-1-Phosphate Uridyl Transferase Deficiency

Further information about this condition is available from Climb.

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