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

Climb™

Supporting those affected by Inherited Metabolic Disorders

Galactosialidosis

Galactosialidosis is a metabolic disorder which belongs to a group known as the lysosomal storage diseases. It is caused by a defect in the CTSA gene which encodes for the enzyme protective protein/cathepsin A (PPCA); this leads to secondary deficiencies of the lysosomal enzymes β-galactosidase and neuraminidase. Galactosialidosis has symptoms which affect many areas of the body. There are three forms that have been identified, which are distinguished by the age at which they present; these are the Early Infantile form, the Late Infantile form and the Juvenile/Adult form. The Juvenile/Adult form is the most prevalent.

Synonyms

Alternative names for this condition are:

- Deficiency of Cathepsin A
- Goldberg Syndrome
- Lysosomal Protective Protein Deficiency
- Neuraminidase Deficiency with beta-Galactosidase Deficiency
- PPCA Deficiency

Further information about this condition is available from Climb.

Disclaimer

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