Glycogen Synthase Deficiency

Other names that may be used for this condition are:
- Glycogenosis Type 0
- GSD-0
- Glycogen Storage Disease Type 0
- Liver Glycogen Synthase Deficiency

Glycogen Synthase Deficiency is a rare genetic disorder characterized by low blood sugar and an abnormally high level of acidic substances called ketones in the blood and urine. This disorder is caused by a deficiency in the liver glycogen synthetase enzyme which leads to a low glycogen content in the liver. The defective gene which codes the enzyme is known as GYS2 and is located on the long arm of chromosome 12.

Treatment includes consulting a dietician to management of the diet, the patients diet should consist of adequate proteins and calories to aid growth. Those affected should eat frequently and avoid long periods without food. Uncooked cornstarch is recommended as it releases glucose slowly. Studies have shown that dietary treatment over one year, which includes frequent meals that are high in protein and low in carbohydrate improved growth and treated symptoms of low blood sugar. Genetic counselling may be of benefit to 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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Climb National Information Centre for Metabolic Diseases

Climb Building, 176 Nantwich Road, Crewe, Cheshire, CW2 6BG, UK

Freephone: 0800 652 3181
Email: ir.svcs@climb.org.uk

Climb is the only charity in the United Kingdom that provides support on all Metabolic Diseases with links worldwide

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