GSD III Glossary

Cori Disease
Another name for GSD III in honour of the 1947 Nobel laureates Carl Cori and Gerty Cori

Cornflour, Cornstarch
A source of slow-release energy used to treat Glycogen storage diseases. The intestines convert it into glucose and pass it directly into the bloodstream.

Debranching Enzyme
A chemical made by the body that helps to breakdown glycogen into glucose (sugar).

Forbes Disease
Another name for GSD III in honour of American clinician Gilbert Burnett Forbes

Glucose
A simple sugar and the human body's key source of energy.

Glycogen
The main storage form of glucose (sugar) in the body.

GSD IIIa affects both the liver and muscles.

GSD IIIb affects only the liver.

Hypoglycaemia often refered to as a "hypo"
An abnormally low level of blood-sugar. Early warning signs are hunger, trembling, and sweating. If severe, there may be confusion and difficulty concentrating. In very severe cases, the person experiencing hypoglycaemia may lose consciousness.

Hepatomegaly
An enlargement of the liver caused in GSD III by excessive amounts of stored glycogen.

Ketones
Ketone bodies are molecules produced by the liver from fatty acids during periods of carbohydrate restriction. The body can produce energy from ketones instead of glucose.

Ketosis
A metabolic state where most of the body's energy supply comes from ketone bodies in the blood. Significant keytosis often accompanies hypoglycaemia in GSD III patients.

Acknowledgements
This leaflet was produced by the AGSD-UK GSD III support team who are grateful for the considerable input from Elaine Salmons, Senior Specialist Nurse, Birmingham Children's Hospital.

Further reading

AGSD-UK literature available online or in print. Please contact the coordinator or office for details of our growing number of publications.

Genetic Alliance www.GeneticAlliance.org.uk
Produce a number of useful patient leaflets including
Health Insurance : Life Insurance : Genetics

GSDNet
An online forum where questions can be answered by people from all over the world relating to GSD III and many other GSD issues. Find the link on the AGSD-UK website under 'Communications'.

GOV.UK www.gov.uk/browse/disabilities
Advice about government services for disabled people. Includes carers, your rights, benefits and the Equality Act.

Citizens Advice Bureau www.citizensadvice.org.uk
For benefits information and advice. Use this link or contact your local bureau to arrange an appointment.

AGSD-UK Support

GSD III Coordinator email: type3@agsd.org.uk

Patients and families often find help from talking with others who have lived through the same experiences. The AGSD-UK can put you in touch with other patients, families or carers.

AGSD-UK Conference
We hold an annual conference where families can meet to share experiences and talk to UK and international GSD III expert healthcare professionals, and researchers.

AGSD-UK office address:
Old Hambledon Racecourse Centre
Sheardley Lane
Droxford
Hampshire SO32 3QY

☎ 0300 123 2790 (UK: National rates on mobiles)
☎ +44 (0) 1489 877 991 (International)

email info@agsd.org.uk

August 2014

Association for Glycogen Storage Disease (UK) Ltd
Company registered in England number 06981121

Registered office:
Crowe Clark Whitehill LLP, Black Country House,
Rounds Green Road, Oldbury, West Midlands, B69 2DG

LOTTERY FUNDED

GSD III
Cori Disease

Also known as

Glycogen Storage Disease Type 3
Cori Disease
Forbes Disease

AGSD

Association for Glycogen Storage Disease (UK)
Registered charity in England number 1132271

☎ **0300 123 2790**

GSD III GSD 3

Before reading too much about GSD III, please be assured that with *good dietary management*, people can lead a relatively normal life, can have unaffected children, and should live to old age.

Symptoms

Initial symptoms can vary, common childhood symptoms are:

- A swollen abdomen (tummy) due to an enlarged liver
- A history of poor growth or short stature
- Frequent feeding - always hungry
- Episodes of low blood sugars (hypoglycaemia) on fasting
- Excessive sweating (particularly in the morning)

Those affected by GSD III may find their symptoms become easier to manage as they become adults. We generally need less energy as we get older because we are not growing.

Potential complications in later life:

- Progressive muscle weakness (GSD IIIa)
- Thickening of the heart muscles (GSD (IIIa)
- Cirrhosis (Scarring) and/or adenomas on the liver

GSD III is caused by a defect in the "*debranching enzyme*".

The defective enzyme prevents glucose from being properly extracted and so glycogen continues to build up in the body. This accounts for the enlarged liver and swollen abdomen and produces the symptoms of low blood-sugar.

www.agsd.org.uk

Treatment

Management of GSD III is aimed at avoiding hypoglycaemia and maximising the child's growth potential. With frequent feeds many of the potential side-effects of GSD III can be avoided.

Children with GSD III cannot go without food for more than a few hours, and are otherwise at risk of dangerously low blood sugars. Regular feeds or meals throughout the day, and overnight feeds are necessary.

After the child is over one year old a slow release carbohydrate can be used such as uncooked cornstarch (cornflour) or a modified cornstartch (e.g. Glycosade®), divided into a few doses during the day, to maintain a steady blood glucose level.

To ensure the child receives a constant supply of energy, 'tube feeding' is usually required overnight. This is because our bodies still need energy while we're asleep and it is easier to use a tube than to be waking up during the night every few hours to have something to eat. It means the child can have the energy they need and get continuous sleep.

A nasogastric tube will be inserted and parents are helped to manage the feeds. When the person with GSD III is older a gastrostomy may be required.

With both of these tubes, your child is still able to eat and drink. They are used to enable your child to have a regular energy supply, and are both reversible so that if your child's fasting tolerance increases they can be removed.

A specialist dietician will give you a regular feeding regime (a plan, for each day) that must be followed. Any problems or difficulties with this regime should be discussed with your specialist metabolic team.

Most children do very well with this management, particularly as symptoms tend to improve as they get older.

Emergency plans for use at home and in hospital will be given to you for your child in case of illness or suspected hypoglycaemia.

Rapid access will be set up for you at your local hospital so that your child can be seen immediately and treatment started without delay.

Each hospital has its own system in relation to emergencies and you will be told who to contact and where to take your child if such a situation should arise.

Parents will quickly learn to assess their child and make decisions on the use of an emergency plan. This is not always easy, and your specialist team will support you in this decision making process.

Glucogel can be used when your child shows signs of severe hypoglycaemia (such as, not responding or fitting), as it is instant sugar that is given and absorbed in the mouth. This should be given in an emergency whilst waiting for an ambulance.

A Glucagon injection is often given for diabetes but should NOT be given when the patient is hypoglycaemic becase it stimulates glycogen release. GSD children have problems releasing glycogen stores, so it will be ineffective.

Why has my child got GSD III?

Glycogen Storage Diseases are genetic. This means that they are not brought about by anything that my have occurred during or after pregnancy. This genetic disorder is inherited from both mum and dad, neither of whom are likely to have the condition themselves.

The instructions to make a human being are contained in our genes. For most of our characteristics we have two copies of a gene; one inherited from mum and one from dad. We each have 6 or 7 mistakes (mutations) in the thousands of our genes, but so long as we have one working copy of a gene it can do the job of both genes.

With this form of inheritance, when you have a baby you and your partner carry the same genetic mistake, there is a 1 in 4 chance with every pregnancy that these two faulty genes will come together, and so the baby has no fully-working copy of the gene. The result is that the enzyme is not made correctly as the instructions are faulty, and the condition therefore develops.

Your hospital team will offer you the opportunity to meet with a geneticist who can explain inheritance and any other concerns you may have for siblings or other relatives.