Glycogen Storage Disease

Other names: von Gierke disease, glucose-6-phosphatase deficiency, glycogen storage disease type 1a
Gene Involved: *G6PC*

What is Glycogen Storage Disease?

Glycogen storage disease (GSD) is an inherited disorder of sugar use. Children are born with GSD and do not grow out of it. All types of glycogen storage disease come from problems in how the liver and kidneys use an energy-storing chemical called glycogen. Children around the world have GSD. Sometimes a family will have more than one child with GSD.

What are the early signs and symptoms?

As babies with GSD get older, it becomes easier to recognize they are struggling.

- Babies with GSD look like their siblings
- They can be fussy or sweaty with low blood sugar and may struggle to gain weight
- The body works hard to balance energy, so it may be difficult to notice something is wrong. Babies may still smile and coo but may not have energy to learn to roll over, sit up or use their muscles at the expected ages.

What Happens as Children Get Older?

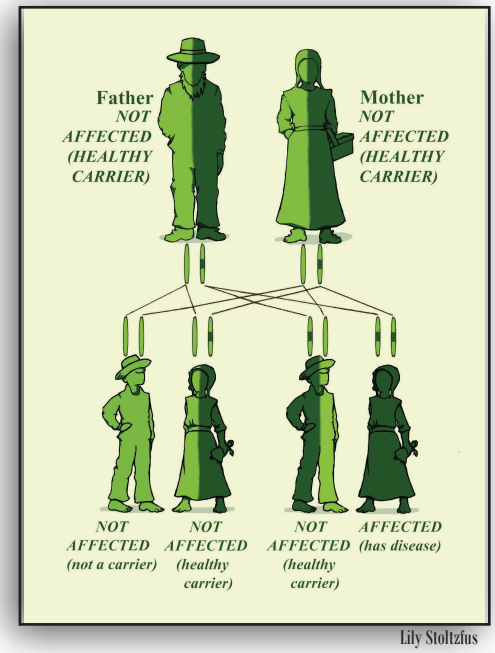
- Children's bellies may look bigger as the liver stores more glycogen and becomes larger
- They may not be as tall as their siblings because the body may not have as much energy to give toward growth
- Children may show signs of low blood sugar (sweating, fussiness, fatigue), or their bodies may not show any signs
- Energy problems become easier to see when the body is stressed during flus, stomach bugs, or fasting

Management

- Patients with GSD avoid problems in glycogen use by limiting how much sugar they eat. It is important to give them the right baby formula and foods.
- Children with GSD can eat many foods, but they must limit foods containing fruit, milk, and table sugars
- GSD patients keep blood sugar levels steady between meals by taking cornstarch
- GSD patients require IV fluids during extra stress with flus or fasting
- Regular bloodwork is required to watch for long term liver and kidney damage, as well as yearly pictures (ultrasound or MRI) of the liver and kidneys

Diagnosis

A gene test is required to make the diagnosis. To arrange this, please see the contact information below.



Autosomal Recessive Inheritance

Genes are instructions for growth and development. We have two copies of each gene. One copy is inherited from our mother, and one copy is inherited from our father. Children with GSD have a spelling change in BOTH copies of their *G6PC* genes. This means they have no working copy of the *G6PC* instruction.

People with one normal copy of the *G6PC* gene and one misspelled copy are called "healthy carriers".

When two healthy carrier parents have a child, there is a:

- 1 in 4 chance the child will have GSD, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *G6PC* gene and be neither affected nor be a carrier

This informational pamphlet is a project of New Leaf Center - a member of the Plain Community Health Consortium (PCHC)

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Central Pennsylvania Clinic (Belleville, PA) **717-935-2065**
Clinic for Special Children (Strasburg, PA) **717-687-9407**
DDC Clinic (Middlefield, OH) **440-632-1668**
Midwest Genetics Network (Okemos, MI) **517-324-8300**
The Community Health Clinic (Topeka, IN) **260-593-0108**
UPMC Children's Hospital of Pittsburgh (Pittsburgh, PA) **412-692-5070**
WeCare Clinic - Medical Care for Special Needs (Pembroke, KY) **270-962-7383**