GM3 Synthase Deficiency

Gene involved: *ST3GAL5*

What is GM3 Synthase Deficiency?

GM3 synthase deficiency is an inherited disorder that primarily causes problems with brain development. People with GM3 synthase deficiency have significant delays in reaching their developmental milestones. They frequently have seizures, fussiness, sleep difficulties, feeding and growth problems. Children are born with GM3 Synthase Deficiency and do not grow out of it.

What are the early signs and symptoms?

- Affected infants often appear normal at birth, but within a few months exhibit fussiness or agitation, poor feeding and difficulty gaining weight (failure to thrive)
- Infants have slow head growth and typically have a small head (microcephaly)
- Many babies are floppy, lack muscle tone, and may develop seizures (epilepsy) within the first few months of life

What Happens as Children Get Older?

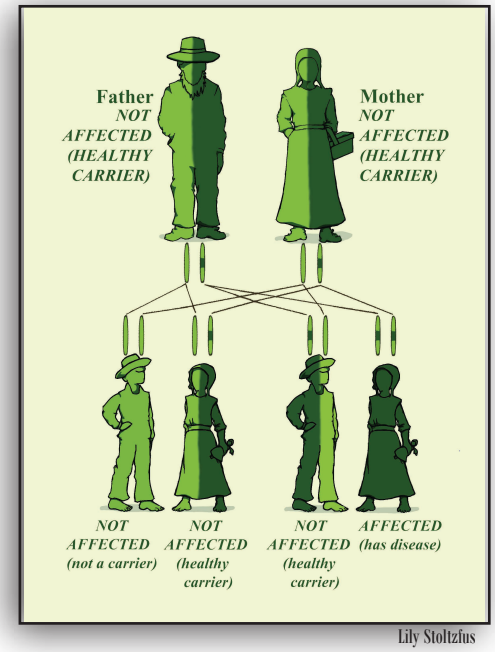
- Affected children have delays in developmental milestones such as sitting, head control, crawling, walking, talking and interacting with their family
- They can develop muscle stiffness or remain floppy
- Most children will have seizures, although they may be difficult to see
- Sleep difficulties and irritability are common
- They can develop non-purposeful movements
- Vision and hearing may be poor
- Digestive problems including acid reflux and constipation are common
- They will need significant support from the family for activities like eating, bathing, and getting dressed

Management

Although there currently is no cure for GM3 synthase deficiency, the various symptoms of the disorder can be managed. There are medications to assist with seizures, irritability, and sleep difficulty. A feeding tube may help with proper nutrition. These and other helpful therapies (e.g. physical therapy to aid movement) should be coordinated through the doctor(s) assisting the care of the affected child.

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 GM3 synthase deficiency have a spelling change in BOTH copies of their *ST3GAL5* genes. This means they have no working copy of the *ST3GAL5* instruction.

People with one normal copy of the *ST3GAL5* 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 GM3 synthase deficiency, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *ST3GAL5* 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)

NEW LEAF CENTER
CLINIC FOR SPECIAL CHILDREN
WINDOWS OF HOPE PROJECT

16014 E. Chestnut St., PO Box 336
Mt. Eaton, OH 44659

330-359-9888
330-359-9890 (F)
info@newleafclinic.org
www.newleafclinic.org

Additional PCHC Members Include:

Center for Special Children (La Farge, WI) **608-625-4039**
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**