GM2 Synthase Deficiency

Gene involved: *BG4GALT1*

What is GM2 Synthase Deficiency?

GM2 synthase deficiency is an inherited condition that affects the brain. It causes difficulties with walking, speech, self-care, and mood. Children are born with GM2 synthase deficiency and do not grow out of it.

What are the early signs and symptoms?

- Babies with GM2 synthase deficiency typically appear normal at birth
- The first concerns parents often report are developmental delays in walking and speech
- Children will have difficulty with balance and are often described as “clumsy”
- They usually do not talk as early as their healthy siblings and their speech can sound slurred

What Happens as Children Get Older?

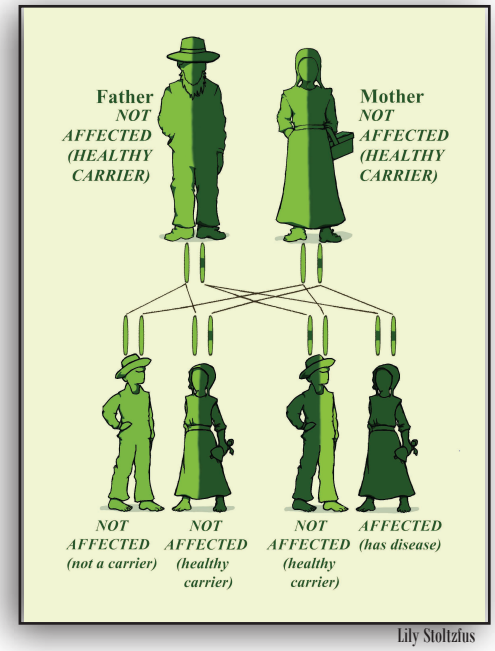
- People with GM2 synthase deficiency often start to have trouble with learning and memory in their late teenage years to early 20s
- In young adulthood their legs start to become stiffer, which makes walking difficult
- They can have difficulty using their hands purposefully
- Mood issues like depression and anxiety are common
- In early adulthood, most families will notice that their loved one talks less and may become less socially engaged
- People with GM2 synthase deficiency will need help from their family for basic needs throughout their life and are not expected to be able to live on their own

Management

Although there is no cure for GM2 synthase deficiency, treatments are available to help children and adults lead comfortable lives. People with this condition benefit from early physical, occupational, and speech therapy to strengthen muscles and encourage hand use. Assistive devices like wheelchairs, and prescription medication for mood problems can be helpful.

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 GM2 synthase deficiency have a spelling change in BOTH copies of their *BG4GALT1* genes. This means they have no working copy of the *BG4GALT1* instruction.

People with one normal copy of the *BG4GALT1* 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 GM2 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 *BG4GALT1* 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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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**