Mast Syndrome

Other names: Hereditary Spastic Paraplegia, SPG21 **Gene involved:** *SPG21*

What is Mast Syndrome?

Mast syndrome is in the family of disorders called hereditary spastic paraplegias. It is an inherited disorder that slowly affects the brain and nerves which can cause problems with walking, leg stiffness, and thinking. Children are born with Mast syndrome and do not grow out of it.

What are the early signs and symptoms?

Most families will report that their loved one had issues with slurred speech in early childhood and walking is often delayed.

What Happens as People Get Older?

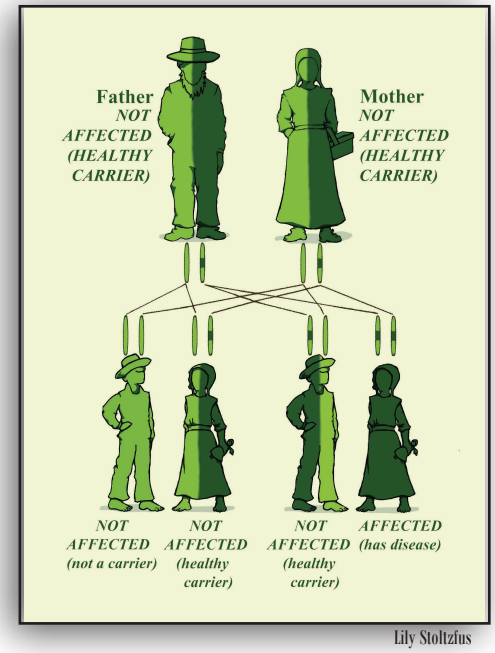
- In young adulthood walking can become clumsy and early signs of difficulties with problem solving and memory can develop
- Families often seek care when their loved one is in their 20s, when problems with walking become more obvious. As their legs become stiffer their walking becomes more off-balance
- People often start to develop non-purposeful movements with their hands and fingers which can limit activities like getting dressed and feeding themselves
- Family members may notice they seem to show less emotion and may appear “flat”
- They might start to struggle with their mood such as depression and anxiety
- They can have a difficult time starting conversation and speech becomes very limited
- They continue to have worsening problems with their memory and problem solving into their 30s
- By their 40s to 50s most people will be completely dependent on the care of their loved ones for daily activities

Management

There is no cure for Mast syndrome, but symptoms can be managed to give people more comfortable and meaningful lives. Prescription medication can be used to help with mood problems, physical therapy can help muscle stiffness and weakness, assistive walking devices and braces can help with mobility, and speech therapy can help with communication.

Diagnosis

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



Lily Stoltzfus

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. People with Mast syndrome have a spelling change in BOTH copies of their *SPG21* genes. This means they have no working copy of the *SPG21* instruction.

People with one normal copy of the *SAMHD1* 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 Mast Syndrome, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *SPG21* 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**