



# MASD Syndrome

**Other names:** KPTN-associated disorder    **Gene involved:** *KPTN*

## What is MASD Syndrome?

MASD is an inherited condition. It affects the way the brain grows and develops. Letters in the name stand for the syndrome's most common difficulties:

- Macrocephaly: enlarged head circumference
- Anxiety: unusually high level of nervous or repetitive behavior
- Seizures: increased risk for having seizures
- Developmental delay: learning difficulties and slow development

This condition was identified in 2014 within Ohio Anabaptist/Amish communities. Although it is rare, children around the world have MASD. Children are born with MASD syndrome and do not grow out of it.

## What are the early signs and symptoms?

- A large head
- Floppy or weak at birth, compared to healthy siblings
- Anxious or "fussy" babies/toddlers who do not cope well with change in routine
- Some infants have seizures that can be treated with medication
- Slower physical and intellectual development

## What Happens as Children Get Older?

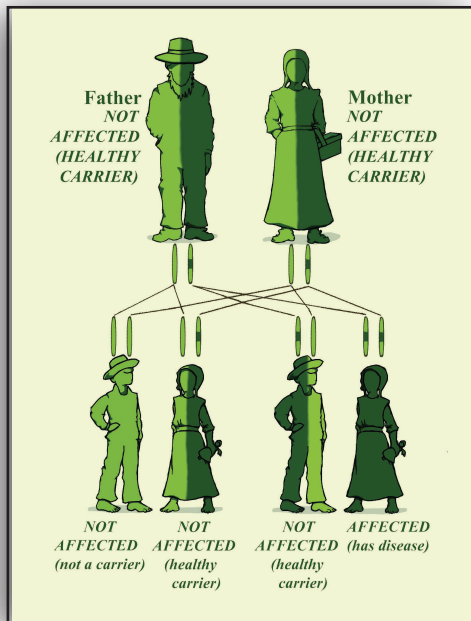
- As they grow, children will make developmental progress. They will walk and communicate. They can go to school, although learning will be at a slower pace than that of their peers. People with MASD will need ongoing support from their families through adulthood.
- When children enter school, they are often anxious and may have trouble adjusting to new routines and being around people they don't know very well.
- Children with MASD do best in "special education" classes.
- Seizures may develop in older children.
- Children with this diagnosis find a controlled work environment helpful. Many older affected individuals enjoy workshops for people with special needs.

## Management

Although MASD is not curable, treatments are available for the problems people face. People with seizures should be under the care of a doctor so the right medication can be given. Children with MASD do well with routine, medications, and patience in stressful situations like school or church. Early intervention for language, physical and social development is also very important. Physical, speech, and occupational therapies help children achieve their full potential.

## Diagnosis

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



Lily Stoltzius

### 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 MASD have a spelling change in BOTH copies of their *KPTN* genes. This means they have no working copy of the *KPTN* instruction.

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



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**