THINK MPS IIIA

Learn about mucopolysaccharidosis type IIIA (MPS IIIA), also known as Sanfilippo syndrome type A

What is MPS IIIA?

MPS IIIA is part of a family of lysosomal storage diseases called mucopolysaccharidoses. These are genetic diseases that affect certain enzymes in the body.

MPS IIIA is specifically caused by a change in a gene called *SGSH*

TYPICAL FORM OF *SGSH*

SGSH provides the instructions for a cell to create an enzyme called sulfamidase

↓

Sulfamidase breaks down a complex sugar in the body called heparan sulfate

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The broken-down parts of heparan sulfate can be recycled or reused

VS

FORM OF *SGSH* IN PEOPLE WITH MPS IIIA

The change in the *SGSH* gene may cause the cell to not make enough functional sulfamidase

↓

Heparan sulfate isn't broken down, so it builds up inside the cell

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Too much heparan sulfate can damage cells, resulting in the signs and symptoms of MPS IIIA

MPS IIIA is rare—it occurs in fewer than

1 in 70,000 births

in the United States.*

*Data from 2017.

How is MPS IIIA inherited?

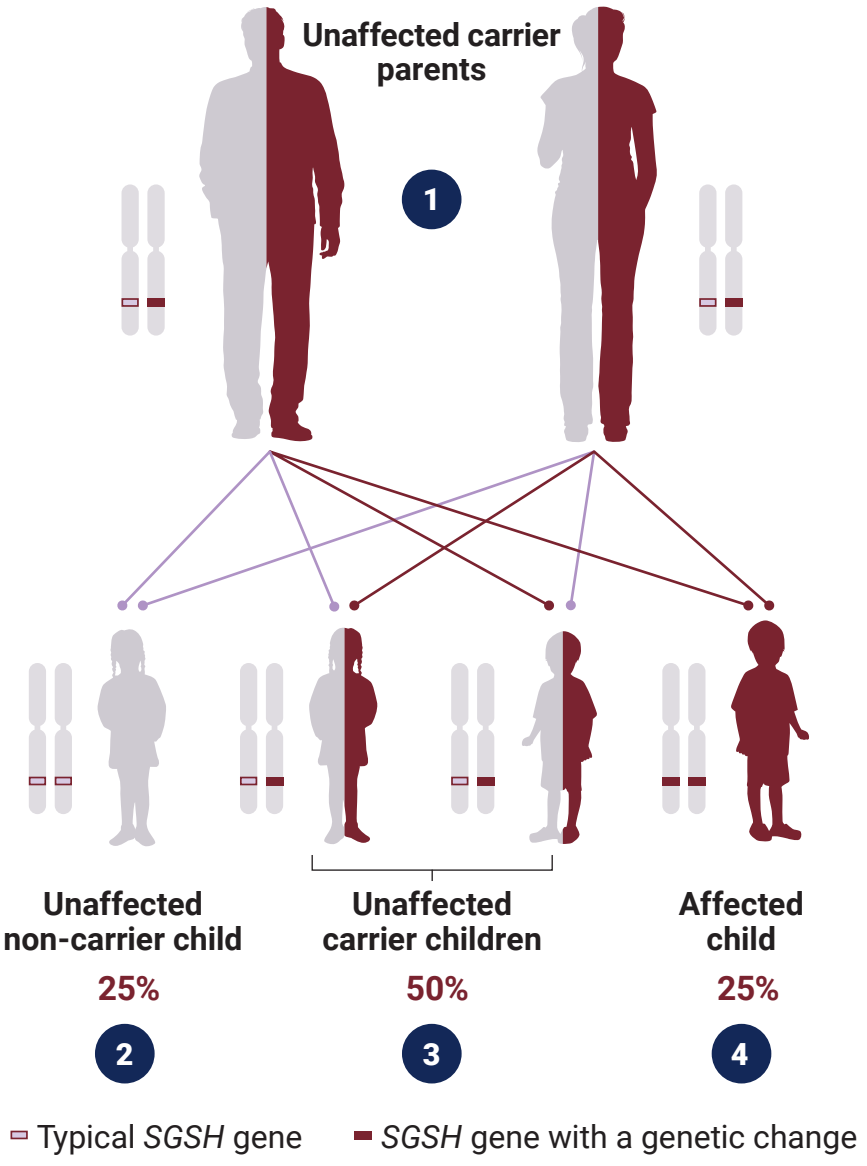
MPS IIIA is inherited in an autosomal recessive manner

1. Each parent carries **1 copy** of the *SGSH* gene with a genetic change but does not have MPS IIIA because 2 copies are needed.

2. Each child has a **1 in 4**, or **25%**, **chance** of not carrying a copy of the *SGSH* gene with a genetic change.

3. Each child also has a **1 in 2**, or **50%**, **chance** of being a carrier, which means they carry 1 copy of the *SGSH* gene with a genetic change but they do not have MPS IIIA.

4. Each child has a **1 in 4**, or **25%**, **chance** of having MPS IIIA, which means they have 2 copies of the *SGSH* gene with a genetic change, inheriting 1 copy from each parent.



What are the signs and symptoms of MPS IIIA?

MPS IIIA can present in many ways, at different ages, and may change over time*

Head and face

- Prominent forehead
- Thick eyebrows
- Full lips and nose
- Enlarged head size

Mouth

- Problems swallowing

Lungs

- Recurring lung infections

Digestive system

- Chronic loose stools/diarrhea
- Pain from problems in stomach and intestines

Muscles and bones

- Pain

Nervous system

- Delay or loss of speech or language
- Intellectual disability
- Seizures
- Challenges walking
- Difficulty sleeping
- Problematic behavior, such as hyperactivity and aggressive, destructive, or impulsive behavior

Ears

- Hearing loss
- Frequent ear infections

Skin

- Thick and tough skin

Other organs

- Enlarged liver or spleen

Because of these effects on brain and body, MPS IIIA is often called "childhood dementia"

*These are not all the possible signs and symptoms of MPS IIIA. Not every child with MPS IIIA will have the same types, severity, and timing of signs and symptoms.

How does MPS IIIA change over time?

Children with MPS IIIA initially develop at a similar rate as their peers but then can lose cognitive abilities and motor skills over time

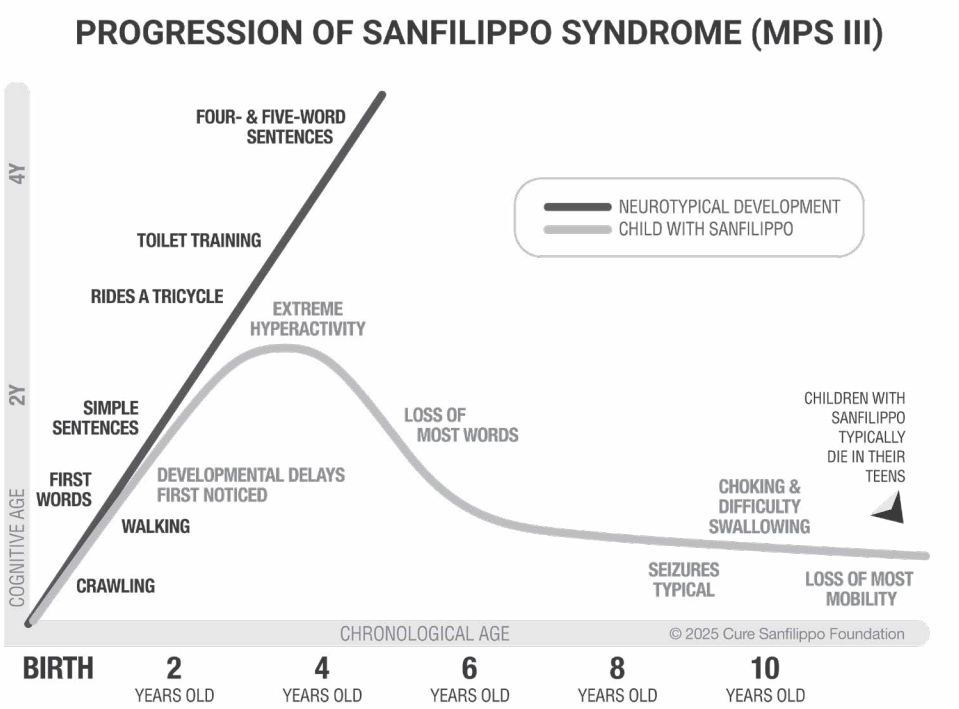


Figure represents studies of disease impact in rapidly-progressing forms of Sanfilippo Syndrome.

MPS IIIA is considered the most severe type of MPS III and the one that progresses most rapidly. How MPS III develops over time can vary significantly from one child to the next.

Figure created by and used with permission of Cure Sanfilippo Foundation.

- Though signs of MPS IIIA may be present at birth, they can go unnoticed
- As early as 2 years of age, development tends to slow down and often stops
- After 3 years of age, any skills learned may be gradually lost, including the abilities to speak, walk, and feed themselves. Behavioral issues and sleeping problems may also start to occur
- People with MPS IIIA have a life expectancy of around 10 to 20 years

How is MPS IIIA diagnosed?

Your child may need specific tests to confirm whether they have MPS IIIA

SCREENING TEST	CONFIRMATORY TESTS		
			
Urine test Detects high levels of heparan sulfate.	**Genetic test** Detects whether any of the changes in the *SGSH* gene that can cause MPS IIIA are present.	**AND/ OR**	**Enzyme test** Measures sulfamidase enzyme activity in a blood or skin sample.

Tests for MPS IIIA are crucial to confirm diagnosis because **the signs and symptoms can be mistaken for other conditions**, such as:

- Autism spectrum disorder because of similar language delay and behavioral issues
- Attention deficit hyperactivity disorder, or ADHD, because of children being hyperactive and impulsive
- General developmental or speech delays

Typically, children with MPS IIIA are diagnosed around 4 to 6 years of age, **and it's not uncommon for it to take 2 years or more to get a confirmed diagnosis.**

What happens after diagnosis of MPS IIIA?

If your child has MPS IIIA, they may receive specialist care for their needs

Depending on their signs and symptoms, your child with MPS IIIA may need:

- Speech, physical, and/or behavioral therapy
- Seizure medications
- Feeding tubes
- Nutritional support
- Sleep medications
- Hearing aids or ear tubes
- Mobility aids
- Modifications to ensure safety at home and in other environments

Your child may need to see different types of specialists, which may include:

- Pediatric neurologists
- Developmental specialists
- Geneticists
- Orthopedic surgeons
- Ear-nose-throat specialists
- Audiologists

THINK MPS IIIA

MPS IIIA is an inherited condition that mainly affects the way the nervous system functions.

MPS IIIA has many signs and symptoms and can slow or stop the development of skills and abilities, which may be lost over time.

Testing for MPS IIIA is an important step toward getting specialist care for the condition.

Learn more about MPS IIIA by scanning the QR code or visiting ThinkMPSIIIA.com

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