



# MPS III (Sanfilippo Syndrome)

## A Guide For Physicians On Screening

### THE SCREENING RECOMMENDATIONS

The American Academy of Pediatrics suggests considering evaluation for inborn errors of metabolism (including MPS disorders) in children with neuromotor and global developmental delays.

MPS III children have significant behavioral features also seen in ADHD, oppositional behavior, speech/developmental delay, or autism. Children often carry these diagnoses for many years before the underlying diagnosis of Sanfilippo Syndrome is discovered.

Please consider ordering a **urine MPS screening (non-sterile urine specimen)** for your patients with developmental delay or a combination with any features listed to the right.

With the initiation of treatment trials, early diagnosis is key to giving children a lifeline to participate. We need your help identifying patients as early as possible.

Reference: Mol Genet Metab. 2014 Sept-Oct; 113(1-2): 34-41. doi: 10.1016/j.ymgme.2014.07.013. Epub 2014 Jul 16.

### WHAT IS MUCOPOLYSACCHARIDOSIS TYPE III (Sanfilippo Syndrome)?

MPS III, also known as Sanfilippo Syndrome, is a progressive lysosomal storage disorder whose primary features and disability are due to central nervous system effects.

There are four types of MPS III (A, B, C, and D), each caused by a unique autosomal recessive genetic defect and enzyme deficiency. All types results in excessive build up of the glycosaminoglycan (GAG) heparan sulfate. Each subtype has similar signs and symptoms, which progress at varying speeds. However, features of classic MPS IIIA typically appear earlier in life and often progress more rapidly. Most children with subtype A, the most common of the four, do not live past their mid-teens.

Children with MPS III generally do not display overt features of the condition at birth. Signs and symptoms of the disease typically begin to be recognized in early childhood, between 2- to 6-years-old, when the child starts missing developmental milestones.

#### EARLY FEATURES:

- Transient Tachypnea Of Newborn
- Macrocephaly/J-Shaped Sella on Lateral Skull X-Ray
- Recurrent Ear/Sinus Infections
- Speech/Developmental Delays
- Significant Hyperactivity
- Autistic Symptoms
- Diarrhea/Chronic Loose Stool
- Poor Sleep
- Umbilical Hernia

#### LATER FEATURES:

- Progressive Intellectual Disability With Brain Atrophy
- Seizures/Dystonia
- Enlarged Liver/Spleen
- Coarsening Facial Features (prominent eyebrows)
- Hearing Loss (May Be Early)
- Loss Of Ambulation
- Loss Of Oral Feeding
- Early Death

#### PROGNOSIS:

At present, MPS III is terminal. At this time, only palliative care is available. Life expectancy is typically around 15 years.

Medical research has recently achieved promising break-throughs in gene therapy and enzyme-replacement therapy with hopes for the future. Clinical trials are happening or expected around the world. Information about clinical trials and referrals are available through the Foundation.

#### HOW COMMON IS MPS III?

MPS III is the most common type of Mucopolysaccharidosis; the estimated incidence of all four subtypes combined is 1 in 70,000 newborns.

#### FOR MORE INFORMATION ABOUT MPS III

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