What is Mucopolysaccharidosis type III (Sanfilippo syndrome)?

MPS III, also known as Sanfilippo syndrome, is a progressive and currently fatal lysosomal storage disorder. Primary features and disability are due effects within the central nervous system. There are 4 types of MPS III (A, B, C, D) each caused by a unique genetic defect and enzyme deficiency. However, all types result in the excessive build up of the glycosoaminoglycan (GAG) heparan sulfate.

Sanfilippo syndrome is inherited in an autosomal recessive pattern. Parents who are carriers have a 25% chance in each pregnancy of having an affected child. 1 in 133 adults carry an abnormality in one of the genes causing Sanfilippo.

Children with MPS III generally do not display overt features of their condition at birth. Signs and symptoms of their disease typically begin to be recognized in early childhood.

Early:
- Transient tachypnea of newborn
- Macrocephaly/J-shaped sella on lateral skull x-ray
- Recurrent ear/sinus infections
- Speech/developmental delays
- Autistic symptoms
- Hyperactivity
- Loose stools
- Poor sleep
- Umbilical hernia

Later:
- Progressive intellectual disability with brain atrophy
- Seizures/Dystonia
- Enlarged liver/spleen
- Coarsening facial features (eyebrows)
- Hearing loss (may be early)
- Loss of ambulation
- Loss of oral feeding
- Early death

What are 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 and autism. Sometimes children carry these diagnoses for many years before the underlying cause is discovered.

Please consider ordering a urine MPS screening (non-sterile urine specimen) for patients with developmental delays or a combination of features listed to the left.

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