Sanfilippo Syndrome

- **What is Sanfilippo Syndrome?**
  - Sanfilippo (san-fuh-lee-po) syndrome, also known as mucopolysaccharidosis type III (MPS III), is a rare genetic disorder that primarily affects the central nervous system. There are four types. Approximately 1 in 70,000 children are born with the condition, but the symptoms do not typically appear until the third year of life or beyond. At this time there is no cure yet for Sanfilippo syndrome.
- **What are the signs/symptoms?**
  - Between ages 2 and 6 years old, kids with Sanfilippo syndrome will begin to show developmental delays and behavioral issues.
  - Early signs of Sanfilippo syndrome include:
    - Speech and other developmental delays
    - Getting many ear infections or sinus infections
    - Large head size
    - Respiratory infections
    - Ongoing diarrhea
    - Headaches
    - Sleep problems
    - Behavior issues can look like those caused by autism.
    - Compulsive behaviors and hyperactivity,
- **What is Sanfilippo Syndrome a result of?**
  - A child born with Sanfilippo syndrome has a defect in one of the genes that make enzymes needed to break down a type of sugar molecule called heparan sulfate. Without the gene, the used heparan sulfate builds up in the cells and keeps them from working properly. This can damage organs, affecting growth, mental development and behavior. Because there are four enzymes involved in breaking down heparan sulfate there are four types of Sanfilippo syndrome (A,B,C, and D), depending on which enzyme is affected. Type A is the most common and most severe form.
- **What are the complications of Sanfilippo Syndrome?**
  - Sanfilippo syndrome affects life expectancy. Most children reach their teenage years, some can live longer.
  - Over time the early signs of Sanfilippo syndrome will lead to:
    - Seizures
    - Severe cognitive problems
    - Progressive loss of motor skills (walking, speaking, feeding, etc)

References: *https://kidshealth.org/en/parents/sanfilippo-syndrome.html*