MPS III is a mucopolysaccharide disease and is also known respectively as Sanfilippo syndrome. It takes its name from Dr. Sylvester Sanfilippo who was one of the doctors in the United States who described the condition in 1963.

As yet, there is no cure for individuals affected by Sanfilippo Syndrome, MPS III but Scientists who study this devastating disease continue to look for ways to treat this disorder.

What is Sanfilippo Syndrome?

Mucopolysaccharides are long chains of sugar molecules used in the building of bones, cartilage, skin, tendons and many other tissues in the body. The more modern word for mucopolysaccharides is glycosaminoglycans or GAG, which stands for the sugar-amino-sugar polymer or long repeating sugar chains found in these materials. In the course of normal life, there is a continuous process of building new mucopolysaccharides and breaking down old ones. This continuous process is required to keep your body healthy. The breakdown requires special biochemical tools called enzymes. Individuals with MPS III are missing one of four specific enzymes that are essential in the breakdown of one of the GAG called heparin sulfate. The incompletely broken down heparin sulfate remains stored inside cells in the body and build up causing progressive damage. The GAG itself is not toxic but the amount of it and the effect of storing it in the body and especially in the brain lead to many physical problems such as delayed development and hyperactivity, sleep disorders, loss of speech, dementia and typically death before adulthood. Although babies typically show no signs of the disorder, as GAG accumulates symptoms begin to appear usually anywhere from 2 to 6 years of age.

Different forms of the disorder

There are four different enzyme deficiencies that have been found to cause Sanfilippo Syndrome and they are described as type A, B, C or D. The names of the deficient Sanfilippo enzymes are heparin N-sulfatase (type A), alpha-N-acetylglucosaminidase (type B), acetyl-CoA-glucosaminide acetyltransferase (type C) and N-acetylglucosamine-6-sulfatase (type D). There is little clinical difference among the four types of Sanfilippo Syndrome. All four types accumulate the same GAG, heparin sulfate. All four enzymes are only involved with the breakdown of heparin sulfate.

Heparin sulfate is primarily found in the central nervous system. The accumulation in the brain is the cause for numerous problems that affect individuals with all types of MPS III.

How common is MPS III?

The incidence of MPS III (all four types combined) is estimated to be 1 in 70,000 births. Type A is the most common one in Northwestern Europe, type B in Southeastern Europe, and types C and D are rare everywhere

How is Sanfilippo Syndrome inherited?

As most of MPS disorders, Sanfilippo Syndrome has an autosomal recessive mode of inheritance. This means that the disorder occurs only if both parents carry the abnormal gene. When both parents have such an abnormal gene, there is a one-in-four chance of every child having Sanfilippo Syndrome or 25% in each pregnancy. The unaffected children have in their turn a 2 in 3 chance of being carriers like their parents, and a 1 in 3 chance of being a normal non-carrier.

Most families with an MPS III child do not have a family history of any genetic problem.

How does Sanfilippo progress?

Sanfilippo syndrome affects children differently and progresses at very different rates. Anywhere from age 2 - 6 symptoms typically begin to appear. Change will be gradual and tends to have three main stages.

The child's pre-school years may be a very frustrating stage for the parents. They begin to worry as their child starts to lag behind their friends' children in development, and they may feel they are being blamed for the child's overactive and difficult behavior.

The diagnosis is often made very late as some children do not look abnormal, and their symptoms are among the most common seen in all children. The doctor has to be perceptive enough to recognize that something serious is wrong and ask for urine and blood tests to help reach

a diagnosis. It is not unusual for families to have one or more affected children before the diagnosis is established. The second phase of the disease is characterized by extremely active, restless and often very difficult behavior. Some children sleep very little at night. Many will be into everything. Many like to chew; hands, clothes or anything they can get hold of.

Sadly, language and understanding will gradually be lost and parents may find it hard not being able to have a conversation with their child. Many will find other ways of communicating. Some children never become toilet trained, and those who do will eventually lose the ability.

In the third phase of the disease, children with MPS III syndrome begin to slow down. They become unsteady on their feet, tending to fall frequently as they walk or run. Eventually they lose the ability to walk. Life may be more peaceful in some ways, but parents will need help with the physically tiring task of caring for an immobile child or teenager.