Fact sheet

What is Sanfilippo (MPS III)?

Sanfilippo Children's Foundation

A rare, life-limiting disease that is part of a wider group of disorders

LSD

Lysosomal Storage Disorders
50 main diseases
1 in 5,000 births

The lysosome is a very small unit in the cell and contains enzymes responsible for recycling cellular materials. A LSD is a disease where those materials don't get recycled correctly and accumulate in the cell, causing dysfunctions.

MPS

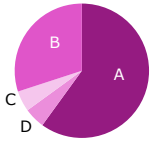
MucoPolySaccharidosis
7 main diseases
1 in 25,000 births

Mucopolysaccharides are complex sugar molecules (also called GAGs) naturally produced by the body and used in the building of bones, cartilage, skin, and tissues. The body continuously produces GAGs which need to be recycled, but in MPS diseases this does not occur and the GAGs accumulate.

MPS III

MPS III or Sanfilippo
4 disease types
1 in 70,000 births

Children with mucopolysaccharidosis type 3 (MPS III) or Sanfilippo lack an enzyme required to recycle heparan sulfate, one of the body's many complex sugar molecules (GAGs), resulting in the storage of these molecules in the cells.



Sanfilippo Types A, B, C, D
A & B most common,
C & D rarer

Each Sanfilippo type corresponds to a particular enzyme that is deficient. There is considerable variation in severity and life expectancy within each type.

An autosomal recessive genetic condition

Genetic

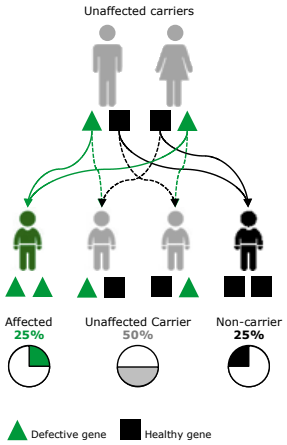
It is inherited: children get one defective gene from both their parents. With both parents carriers, there is 1 chance out of 4 for each child to inherit the disease.

Recessive

Each individual has two copies of each gene. In recessive disorders, affected individuals have two defective genes. Individuals who have one defective gene and one healthy copy are called carriers and are unaffected.

Autosomal

The defective gene is on a chromosome that is not a sex chromosome. Both males and females can be affected in autosomal disorders and males and females can be carriers.



A metabolic form of childhood dementia

Metabolic

Metabolism is the set of life-sustaining chemical transformations within the cells. These transformations are done through the actions of **enzymes** which act as catalysts; the missing enzyme in Sanfilippo results in a metabolic disorder.

Childhood dementia

This disorder affects primarily the cells in the brain and is considered a type of **childhood dementia**. Children experience hyperactivity, sleeplessness, loss of speech and cognitive skills, cardiac issues, seizures, loss of mobility and finally death.

A progressive and fatal disease

Children at birth appear healthy; the first symptoms are often mild developmental delays. The disease progresses very differently from one child to the next, making it very difficult to predict.

Symptoms spectrum

- Brain and eyes
- Ear, nose and throat
- Mouth & teeth
- Speech & swallowing
- Heart and lungs
- Bowel
- Liver & spleen
- Hair & facial features

The disease does not yet have a cure and only limited palliative treatments currently exist; average life expectancy is 12-20 years. However medical research is achieving promising break-throughs giving real hope for the future.

Dec 2019