What is Sanfilippo (MPS III)?

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

Lysosomal Storage Disorders
- LSD
- 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.

Mucopolysaccharidoses
- MPS
- 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 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.

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.

Early Years (2-6)
- Stage 1: Mild developmental delays
- Stage 2: Extreme activity and difficult behaviour
- Stage 3: Gradual decline with shortened lifespan

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 breakthroughs giving real hope for the future.