

## It all starts in our DNA....

Of the 23 pairs of chromosomes we have, number 17 carries 1200 to 1300 genes, including 4 genes called SGSH, NAGLU, HGSNAT and GNS.

These genes' role is to produce specific enzymes which live in the cell's recycling centre (called the lysosome).

Those enzymes' job is to recycle the heparan sulfate, one of many complex sugar molecules (called GAGs), which are used in the building of bones, cartilage, skin, and tissues.

The body continuously produces GAGs and the recycling process is essential to keep the cells clean...

... and help our body and brain develop and stay healthy.

## Sanfilippo (MPSIII) is the result of a genetic condition

Some people are born with a different version of one of those 4 genes (mutated gene) which they **inherit** from both their parents.

Because of this mutation, the **gene** cannot produce enough of a particular enzyme.

Without the **enzyme**, the heparan sulfate doesn't get recycled properly.

The heparan sulfate then accumulates in the recycling centre (the lysosome) and this **surplus storage** affects the functions of the cells, particularly those in the brain...

... setting off very serious **symptoms** that get worse as the waste builds up, eventually causing the brain and the body to stop functioning.

## Different types of approaches for MPSIII...

Genetic Counseling

Gene Therapy Stem Cell Therapy

Enzyme Replacement Therapy

Substrate Reduction Therapy

Palliative Treatments

## ... try to find a solution to the problem by...

...identifying carriers of the mutated gene and managing the risk of having a child with the disease.

... introducing a healthy copy of the gene for the body to start producing the missing enzyme normally.

... replacing the missing enzyme so that the GAGs get recycled correctly.

... reducing the production of GAGs so there is less waste that gets stored in the cell.

... treating the symptoms through therapies and medication to increase the patient's quality of life.

Preventing the disease

Curing the disease

Stopping the disease

Slowing down the disease

Managing the symptoms

Root Cause

Symptoms