

It all starts in our DNA....

Errors in any one of 4 genes called SGSH, NAGLU, HGSNAT and GNS cause Sanfilippo (types A, B, C and D respectively).

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

The enzymes recycle "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 two faulty copies of one of these genes, which they inherit from both of their parents.

Because of this the **cells** cannot produce a particular enzyme, or the enzyme that is produced does not work.

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 approaches try to find a solution to the problem

Genetic Counseling

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

Gene Therapy Stem Cell Therapy

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

Enzyme Replacement Therapy

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

Substrate Reduction Therapy

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

Palliative Treatments

... 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