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Research study testing new treatment for Sanfilippo disease progressing well

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A study into a new treatment for Sanfilippo disease, a rare and fatal condition which causes progressive dementia in children, is progressing well with results set to be published later this year.

Also known as Mucopolysaccharidosis (MPS) III, there is currently no effective treatment for the condition which affects around 150 children in the UK.

Sanfilippo disease is diagnosed in childhood, with sufferers experiencing deafness, hyperactivity and behavioural problems, progressive developmental delay, and seizures during the later stages of the condition. It is usually fatal in late childhood or early adulthood.

The study aimed to establish whether high doses of the treatment Genistein Aglycone was effective. Patients with Sanfilippo disease have too much of the substance heparan sulphate in their cells, particularly cells in the brain, because they lack the enzyme that usually breaks the heparan sulphate down. It is thought that Genistein Aglycone works by blocking the production of heparan sulphate and associated damage to the cells.

Genistein is a naturally occurring chemical found in soya beans. In the study the researchers used a synthetic version, Genistein Aglycone, to maximise absorption through the gut.

Previous research has shown that low doses of Genistein reduce the

heparan sulphate in the blood and urine, but are not sufficient to be effective in the brain. However, research at The University of Manchester using higher doses of Genistein Alygone in the mouse model of Sanfilippo disease has shown that this is effective in reducing neurodegeneration.

All 22 patients were recruited within the allocated time frame. Monitoring of the final participant to join the trial will be complete in July, after which the results will be published.

The study was funded by charity The Society for Mucopolysaccharide Diseases (The MPS Society) and the GEM Appeal. It was conducted by The University of Manchester and the National Institute for Health Research (NIHR) / Wellcome Trust Manchester Clinical Research Facility at The Royal Manchester Children's Hospital.

Patients received either Genistein Aglycone or placebo (an inactive substance that looks like the treatment) with food, over a period of 12 months. After that, all children received Genistein Aglycone for a further 12 months. Participants attended regular clinic visits to have levels of heparin sulphate in the spinal fluid measured, alongside other assessments.

Dr Simon Jones, Consultant in Paediatric Inherited Metabolic Disease at Saint Mary's Hospital, Manchester and Honorary Senior Lecturer at The University of Manchester, who is leading the study explains: "This was the first study to test the effectiveness of higher doses of Genistein, to assess whether it has an effect on the brain of patients with Sanfilippo disease. Now that we are nearing completion of the trial, we will soon be in a position to publish those findings.

"We know how distressing this condition can be for children and their families. During the later stages of the disease, patients experience

seizures, become wheelchair bound and can have trouble swallowing. Conducting research into rare conditions like Sanfilippo disease is important to help us provide patients with the best possible care."

Dr Brian Bigger, Chief Scientific Investigator for the study from The University of Manchester said: "We would like to thank all of the families who have supported this study whether that has been by raising the vital funds to deliver the study or by deciding to take part in the study. We eagerly anticipate the results of the trial and hope that this may ultimately result in a potential treatment for Sanfilippo disease."

Provided by University of Manchester