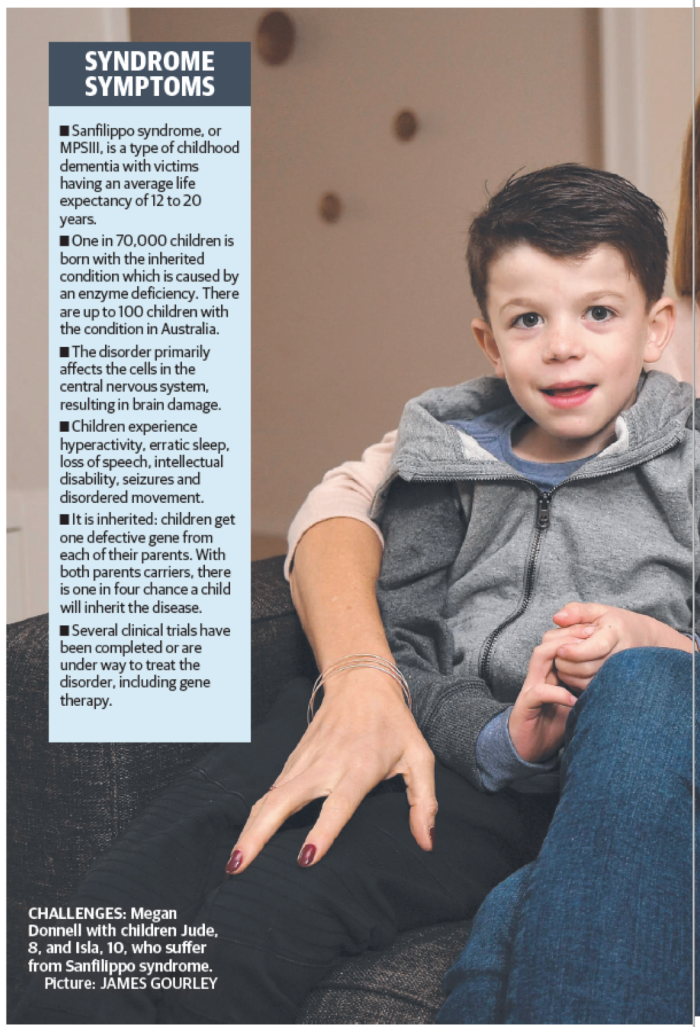

SYNDROME SYMPTOMS

- Sanfilippo syndrome, or MPSIII, is a type of childhood dementia with victims having an average life expectancy of 12 to 20 years.
- One in 70,000 children is born with the inherited condition which is caused by an enzyme deficiency. There are up to 100 children with the condition in Australia.
- The disorder primarily affects the cells in the central nervous system, resulting in brain damage.
- Children experience hyperactivity, erratic sleep, loss of speech, intellectual disability, seizures and disordered movement.
- It is inherited: children get one defective gene from each of their parents. With both parents carriers, there is one in four chance a child will inherit the disease.
- Several clinical trials have been completed or are under way to treat the disorder, including gene therapy.

CHALLENGES: Megan Donnell with children Jude, 8, and Isla, 10, who suffer from Sanfilippo syndrome. Picture: JAMES GOURLEY

Isla has forgotten joy

REBECCA DIGIROLAMO

ISLA Donnell is 10 and she can't remember how to open a present.

It's a simple childhood pleasure that eight months ago brought her great joy under the Christmas tree.

"She would unwrap her presents with such zest, but now she can't even begin to open them," said her mum, Megan Donnell.

Isla and brother Jude, 8, suffer from a rare and fatal childhood form of dementia called Sanfilippo syndrome that is progressively destroying their brains.

Both are living with intellectual disability, behavioural problems, including severe hyperactivity, and sleep disorders. Their cognitive function and motor skills will continue to deteriorate.

The life expectancy for children with Sanfilippo is 12 to 20 years. There is no cure.

"This year, we are seeing a significant progression of the disorder in Isla," Ms Donnell said. The window for a treatment is narrowing: Isla turns 11 next April.

"This is an aggressive condition," she said.

"This is not just about Isla and Jude – it's about all kids with Sanfilippo."

"Every day that passes, we lose a part of these children forever. It is really critical that we find something to help them really quickly."

Ms Donnell established the Sanfilippo Children's Foundation in September 2013 - four months after Isla was diagnosed. She was four.

One month later, two-year-old Jude received the same diagnosis.

The siblings carry a genetic mutation inherited from both their parents – a fact unknown to them until after their children's diagnosis.

The Sanfilippo Children's Foundation has committed more than $6.3 million to 18 research projects across the country and the world.

FOR MORE INFORMATION VISIT SANFILIPPO.ORG.AU

BRAIN IN A DISH

Adelaide scientists' landmark study to cure child dementia

EXCLUSIVE
REBECCA DIGIROLAMO

SOUTH Australian researchers will grow brain cells in a dish to fast-track testing of possibly thousands of drugs in the race for a cure for an aggressive, terminal form of childhood dementia.

The two-year study on Sanfilippo syndrome is being led by scientists from the South Australian Health and Medical Research Institute, Adelaide's Women's and Children's Hospital and the University of Adelaide.

The $2.5 million "Brain in Dish" study will be funded by the Sanfilippo Children's Foundation after securing a $2 million Federal Government Medical Research Futures Fund grant.

Researchers hope that development of this cutting-edge technology will not only revolutionise approaches to personalised medical treatment of Sanfilippo patients, but also pave the way for its application in many more common neurological disorders such as Alzheimer's disease and Parkinson's disease.

Over the next six months researchers will collect skin cell samples from Sanfilippo patients around the country. Once collected, the cells will be reverse-engineered into functioning brain cells.

These cells, representing the brains of individual patients, will be tested against potentially thousands of approved, new and experimental drugs for neurological disorders in the search for a disease cure.

CUTTING EDGE: Dr Nicholas Smith and Associate Professor Kim Hemsley.

Sanfilippo syndrome is a rare genetic condition causing progressive fatal brain damage and currently affects up to 100 Australian children. The neurodegenerative disease does not yet have a cure and only limited palliative treatment exists. Life expectancy for Sanfilippo syndrome is between 12 to 20 years.

Chief researcher Associate Professor Kim Hemsley, from SAHMRI, said the study's use of patients' own cells to rapidly trial multiple drug combinations, without risk to the children themselves, and personalised down to a molecular level, was groundbreaking.

"This study will hopefully deliver improved therapies and a disease cure in an accelerated time-frame compared to traditional pathways of drug discovery," said chief researcher Dr Nicholas Smith, from the University of Adelaide and the Women's and Children's Hospital.

"Waiting up to 15 years for a drug trial for these kids can be too late," Dr Smith said.

The research will involve two other key researchers – SAHMRI's Dr Cedric Bardy and Professor Mark Hutchinson, of the University of Adelaide. A panel of Australian and international experts will also help steer the project.

Sanfilippo Children's Foundation executive director Megan Donnell said the method of personalised drug-screening could not only lead to treatment but potentially spare children suffering from Sanfilippo invasive treatments and side-effects.

"We are thrilled to be partnering with the Government and world-leading researchers in Adelaide to accelerate research towards effective treatments for this devastating condition," she said.

Her own children – Isla, 10, and Jude, 8 – were diagnosed with Sanfilippo syndrome in 2013 (see story at left).

"Five children are born every year in Australia with Sanfilippo syndrome and there is currently no treatment or cure available. We have made it our mission to change this," Ms Donnell said.