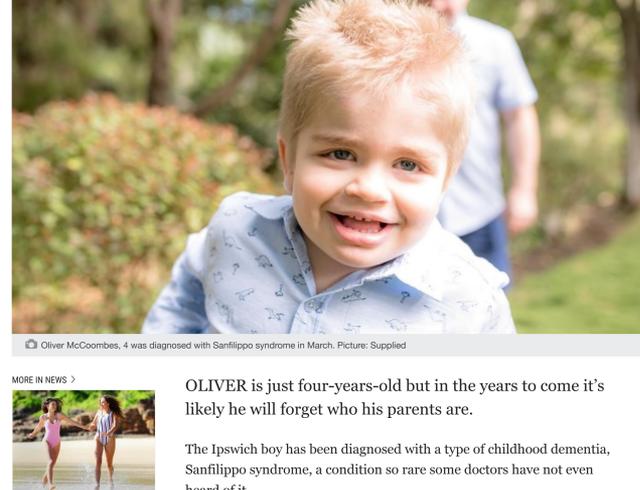


QLD NEWS **McCoombes family's heartbreak and hope for four-year-old Oliver with Sanfilippo syndrome**

Oliver McCoombes will likely very soon forget who his parents are. The four-year-old Queensland boy is living with a type of childhood dementia so rare some doctors have not even heard of it.

Antonia O'Flaherty, The Sunday Mail (Qld) Subscriber only | June 28, 2020 12:00am



Oliver McCoombes, 4, was diagnosed with Sanfilippo syndrome in March. Picture: Supplied

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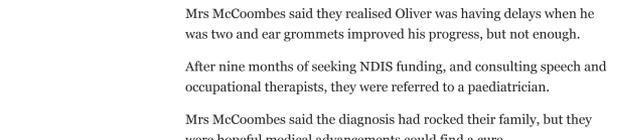
OLIVER is just four-years-old but in the years to come it's likely he will forget who his parents are.

The Ipswich boy has been diagnosed with a type of childhood dementia, Sanfilippo syndrome, a condition so rare some doctors have not even heard of it.

Phillip and Catherine McCoombes received the life-changing diagnosis in March after a nine month journey to investigate developmental delays.

The syndrome causes progressive brain damage, it has no cure and children with the condition have a life expectancy of 12 to 20 years.

Experts believe there are less than 100 cases in Australia.



Phillip and Catherine McCoombes with Oscar, 4, and Maisy, 9 months. Picture: Supplied

Mrs McCoombes said they realised Oliver was having delays when he was two and ear grommets improved his progress, but not enough.

After nine months of seeking NDIS funding, and consulting speech and occupational therapists, they were referred to a paediatrician.

Mrs McCoombes said the diagnosis had rocked their family, but they were hopeful medical advancements could find a cure.

"You go in thinking he just had developmental delays, but then you first get told he has something that's terminal, then you get told there's no cure, it was really devastating," she said.

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"We're both carriers, we don't show any symptoms, so unless we're actually tested we wouldn't know and it is extremely rare, we hadn't heard of it, no one we've told has heard of it."



Oliver McCoombe. Picture: Jamie Hanson

Mrs McCoombes said they had to then test their younger daughter, who did not inherit the condition.

"Obviously we're glad she's okay, she will be okay, she'll get to grow up but that doesn't really help the fact Oliver does have it and there's no cure," she said.

Mrs McCoombes said the diagnosis hit her husband hard the moment they received it, but she was in shock and didn't break down until a few days later, when she told her own father.

She said Oliver's symptoms manifest as hyperactivity and he can be destructive, throwing and chewing on objects.

"As time progresses he will lose more of his abilities, he will lose his speech completely, then eventually lose his mobility, the ability to chew and swallow and be fed through a tube and eventually pass away," she said.

She said Oliver could go to kindy, but would have to be enrolled in a special school when it came time to start prep.

"I'm happy that he is sort of having a bit more normality, going to kindy and I know he does have a lot of fun," she said,

"He's really happy there, but it's hard at the same time because you want to spend as much time as possible with them."



But their story of heartbreak is also one of hope, that a silver lining might deliver a treatment to slow the progression of the syndrome, or a cure.

"It's my hope they will find a cure, and hope that our children will get to grow up and they won't have to suffer," Mrs McCoombes said.

"We're focusing on Oliver's campaign and raising awareness and funds for the foundation and looking forward to spending time together.

"He's very affectionate, loves giving kisses and cuddles, and Maisy absolutely adores Oliver, she's always going up to him and he'll give her little cuddles."

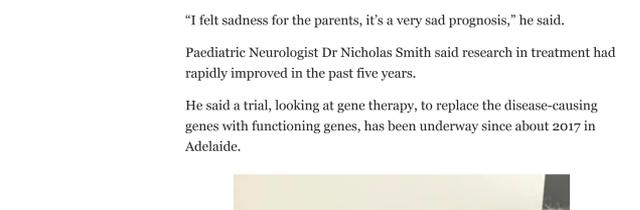
Mrs McCoombes said they would be willing to try any gene therapy trial in Australia or around the world, and would stop at nothing to try and find a treatment or a cure.

For the family's Paediatrician, Dr Paul Koch, it was the first time he diagnosed a child with Sanfilippo syndrome.

"I felt sadness for the parents, it's a very sad prognosis," he said.

Paediatric Neurologist Dr Nicholas Smith said research in treatment had rapidly improved in the past five years.

He said a trial, looking at gene therapy, to replace the disease-causing genes with functioning genes, has been underway since about 2017 in Adelaide.



Dr Smith said the children first show symptoms at about two years of age, experiencing slowed development and at about five years of age they begin to have significant loss of skills, accompanied with behavioural issues, and they lose their language skills.

"Over time they develop motor disabilities, it's a slow thing but they lose their fine motor skills and eventually they lose the ability to walk," he said.

Dr Smith said they can become deaf, non-verbal, and they increasingly lose their ability to interact and can end up in vegetative state before death.

"There is significant hope for these therapies, while they remain experimental there is potential for them to make a profound impact for children and families living with this devastating disease."

Sanfilippo Children's Foundation founder Megan Donnell, mother of Isla and Jude who are both battling the condition, said several programs were being funded in hopes of discovering new treatments or a cure.

The foundation's head of research, Kristina Elvidge, said since 2015 the foundation had committed \$4.7 million to 24 projects in a bid to research and trial effective treatments in search for a cure.

They also advocate for screening programs to identify the condition early in children as newborns, and for parents to determine whether they're carriers.

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Oliver McCoombe's diagnosis has rocked his family. Picture: Jamie Hanson

Oliver McCoombe with his mum Catherine. Picture: Jamie Hanson

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