Patient story: Abigail

When little Abigail celebrated her first birthday, she was a bubbly girl who had reached all her milestones. But, within months, her parents were worried. Her baby babbling had stopped, she wasn't making eye contact, was having trouble using her hands, and had begun repetitive movements, like pacing.

The family returned home to Australia from overseas to seek medical help and Abigail was initially diagnosed with autism. She began a rigorous routine of therapy.

But her progress was limited and, by her third birthday, Abigail's parents had been given the "devastating" diagnosis that she had Rett syndrome following genetic testing.

Abigail's mother, Mary, says they began a journey from disbelief to acceptance, along with a lot of grief.

Rett syndrome is a rare genetic neurological and developmental disorder that affects the way the brain develops. It causes a progressive loss of motor skills and language and can also cause seizures, severe scoliosis, breathing abnormalities and unusual hand movements.

It impacts every aspect of Abigail's daily life.

"She is dependent on us for everything," Mary explains. "She has no functional hand use. So even for something as simple as having a drink, she relies on someone to remember to offer her one. We are very lucky that Abigail can walk – so many people with Rett syndrome never do."

Abigail is learning to communicate using an eye gaze device. The family were able to purchase the $13,000 piece of technology following a donation from an anonymous benefactor. Abigail uses her eyes to 'point' to symbols on a screen.

"We now some have some ways of communicating," Mary says.

Abigail, now seven, is still a little girl with a big personality. Amid a busy routine of various therapies and going to school, the highlight of her week is her horse riding lessons.

Mary has been working to raise awareness of the condition and to raise funds towards medical research for a cure. This includes launching Speechless, an annual event during Rett syndrome awareness month in October where volunteers are sponsored to spend a day without talking.

She is optimistic about the potential of a breakthrough from medical research like the work of A/Prof Wendy Gold and her team.

> **"I am confident that we will find a cure within Abigail's lifetime. I don't know what that will mean for Abigail. But I am really focused on finding a cure."**
> **– Mary, Abigail's mum**