North Shore Mums
For mums, by mums

My son, Quinn: A mother's story for Jeans for Genes day

By North Shore Mums on July 25, 2018

NORTH SHORE MUM SASHA AND HER SON QUINN WITH THEIR FAMILY

Six-year-old Quinn may not be able to talk, but he gets his message across loud and clear as a star of the new ad campaign to mark the 25th Anniversary of Jeans for Genes. Quinn, from North Wahroonga, has a metabolic disorder known as Mucolipidosis. He is in a wheelchair, doesn't speak and is losing his vision. Here his mum Sasha shares the family's incredible story.

When my son Quinn was around nine months old, I had niggles that something wasn't right. I was watching a little boy who was the same age. I thought to myself, "Quinn doesn't seem as alert or as switched on." By the time he was one, it became clear our life was going on a different path to the one I had envisaged. At that time, I read something about a child with autism, and suddenly I could see it in Quinn. I had noticed that he didn't turn his head, and he was flapping his fingers all the time. The next day, I was at the school where I worked as a teacher, and there was a support teacher who worked with children who have autism, and I just burst into tears and told her.

QUINN (RIGHT) HAS MUCOLIPIDOSIS TYPE IV (ML4)

I went to my GP, and he recommended Quinn do physio and see a pediatrician. He had an MRI, we did some blood tests, and everything came back normal. Then our GP noticed that Quinn held his toys close to face when he was playing and recommended an ophthalmologist. That doctor saw cloudiness in his eyes and said he had a metabolic disorder. **It took six months of testing to find out he had Mucolipidosis Type IV (ML4).**

When I was given the terminology, I realised the full impact of what Quinn's life would be like. His vision was already obscured. He's six now, and by 12 he'll probably be blind. He'll get spasticity in his joints. So far, we can see his arms stiffening. He's in a wheelchair, and he's non-verbal. But having a diagnosis validated everything I'd been feeling and seeing. Finally, I felt like we were on the right path. I'd had the trauma of when he was one and he wasn't progressing. I had reverse feelings when he was diagnosed. I was relieved. For 95% of the time, we've embraced who Quinn is and what he's all about. **It's only 5% of the time that it's really difficult.**

It's amazing what we can get out of him. In some ways, communicating is like having a baby: you learn what their cries and their behaviours are about. I've learnt what he's all about, and he's learnt how I deliver messages to him. Those close to him work out some magical way to speak to him.

I've explained it all to my older son, but my youngest daughter knows no different. The younger one says, "Quinny don't talk, Quinny don't walk". We walk on the street, and she pushes her doll in her pram and always parks it next to Quinn's wheelchair. My older son has this gorgeous thing where he says, "Quinn, I'm going to give you a head rub," and he shakes his head violently against Quinn's and they cackle every time.

We've found things to impress upon the other two about having a brother with a genetic condition. We told them how, when we went to Disneyland, we got the Fast Pass because of their awesome brother in a wheelchair. We invite lots of kids over so they can see this is our normal.

At present, there are no treatments, no cure. There is a lot of testing but no gene therapy yet. We need lots to happen. We live in hope that a cure will be found, or some treatment will be made available- even if it's for future generations. I understand that all rare genetic conditions have overlapping qualities, so research may not find a cure for us but it might help someone else. It's like a massive jigsaw. You start with just one piece and every piece makes a difference to the whole puzzle. It's very common to ask, why us? But I want to know – what are we going to do about it?

On a personal level, if people support research into genetic diseases, they should know that even one dollar could someday improve the quality of life for someone they love.For 95% of the time, we've embraced who Quinn is and what he's all about. It's only 5% of the time that it's really difficult.

'Fight for Me, Fight with Me' is the theme of this year's Jeans for Genes Day campaign, which encourages everyone to sign up now to help raise money or give a donation that will make finding cures for children's genetic diseases possible. Sign up here to raise money and help find cures for children's genetic diseases.