

Tuberous Sclerosis

Complex

It can be devastating, but there is hope for people living with this unusual condition.

Lizzie Pinkerton had a seizure the day she was born. “She twitched and jerked for about 10 seconds,” remembers her mother Sue. “We thought it was just new baby stuff, even though she was our third.”

Seven months later Lizzie’s parents were told she had Tuberous Sclerosis Complex, or TSC. This genetic disorder occurs in around one in 6,000 people, 80% of whom have epilepsy. It causes tuber-like growths to appear which are benign but calcify over time, becoming hard or ‘sclerotic’. These have the potential to affect the function of the brain, skin, kidneys, lungs, eyes, heart and other organs. Often when doctors make a diagnosis of TSC it comes as an enormous shock to a family. But the Pinkertons had struggled with the uncertainty surrounding Lizzie’s different symptoms, which ranged from seizures to being temporarily blind. So for them, discovering the truth “was a relief.”

Later, faced with Lizzie’s many health challenges and the “doom and gloom” of literature about TSC, “it felt like we were in a vacuum,” says Sue. “We didn’t know where to go, who to turn to.” They contacted the Australasian Tuberous Sclerosis Society (ATSS). Then in 1994, when Lizzie was 10 years old, Sue became president of this vibrant support organisation, which today is helping to improve

quality of life for people with TSC in Australia and New Zealand. As an advocate, speaker and fundraiser, Sue is “passionate” about working for change. While “there’s a lot more awareness now,” she says many doctors still haven’t heard of the condition.

Symptoms

Some people may be unaffected and never even find out they have TSC, or experience only minor impacts. For others, symptoms can be wide-reaching. Sue’s daughter Lizzie didn’t walk until she was seven and “anything she achieved she had to be taught,” says Sue. Common signs include:

- 1. Epilepsy** Hard-to-control seizures caused by tubers in the brain are often the first indication of the condition.
- 2. Developmental delays** Half of people with TSC have learning difficulties, from minor to serious.
- 3. Skin signs** These include white patches of de-pigmented skin, a raised rash on the nose and cheeks, or skin nodules around fingers or toenails becoming more obvious in adolescence.
- 4. Behaviour** Autistic and hyperactive tendencies can occur, along with attention problems.

Treatment

TSC is incurable. But medical management can vastly improve a

Living with TSC

Lizzie Pinkerton needs 24-hour care because of her condition. At 23, she is unable to talk, has daily complex partial seizures, regular bouts of non-convulsive status epilepticus and is hearing and vision impaired. But her disability has also given her determination a chance to shine. She is busy and active. When she's not delivering Meals on Wheels and doing work experience at Bunnings as part of her adult community participation program, she enjoys hobbies including swimming, horse riding and going to movies or out for coffee.

Many strategies can help people make the most of life with TSC, regardless of their disability. Sue's suggestions are:

1. Get informed Find out as much as you can, especially if you're the parent of a child with TSC, and pass information on to your child's school. Knowledge gives you confidence, and enables you to become an advocate for your child.

2. Meet others Connect and share knowledge and experiences with other people who are in a similar situation. Ask the ATSS about social opportunities like the National Picnic Day or biannual TSC conference.



Lizzie Pinkerton "needs assistance for nearly all activities," says her mother Sue (together in Sydney at Christmas). Even so, "she has a very fulfilling life."

3. Use early invention services Occupational therapy, physiotherapy and speech therapy can all help to minimise the impacts of TSC on a child's development.

4. Find a family doctor Go to the same GP every time instead of visiting different medical centres as this will help you manage issues better.

5. Use respite services Make the most of opportunities for support and rest.

6. Get gene counselling Learn about the likelihood of other children being affected, as this can help families to make informed decisions for the future.

Most importantly Sue recommends people stay positive about the future. While diagnosis "can seem devastating at first... there is hope," she says. "That's the message I want to get across."

TSC FAST FACTS

- TSC causes tuber-like growths in the organs.
- 1 in 6000 people have TSC.
- 80% have epilepsy due to brain lesions.
- A third of people inherit TSC from a parent.
- 70% of cases occur from a new gene mutation.

person's prospects.

Thanks to technology like CT and MRI brain scans, diagnosis can be made earlier. So treatment can begin sooner, helping to limit the reach of the condition.

Managing seizures early with antiepileptic drugs is particularly important. This is because reducing seizure frequency, especially in a child's crucial first year, can minimise the impact on their learning and development. Newer drugs are also more effective in controlling seizures, with fewer side-effects.

A small number of people may also be suitable for surgery. Although it can be hard to isolate and remove the growths causing the seizures, more than half of people who have surgery become seizure-free. The high-fat low-carbohydrate ketogenic diet and Vagal Nerve Stimulators (devices interrupting seizure activity through an electrical pulse) can also help to control seizures in children.

Other treatments include early intervention for developmental delays such as speech therapy or physiotherapy, and laser treatment for skin rashes and growths if appearance is a concern.

The work of ATSS

The ATSS is a volunteer organisation founded in 1982 by a woman with TSC to raise awareness and support families. Now it's instrumental in bringing about positive change for people in Australia and New Zealand.

Last year alone saw three achievements. The ATSS helped to establish Australia's first multidisciplinary TSC clinic, which runs monthly at Sydney Children's Hospital in Randwick and is a model for other hospitals to copy. It held the first Medical Meeting for international experts to share knowledge and improve the care and management of patients. The Society also funded a new genetic testing laboratory – the first non-profit facility of its kind in the world - in collaboration with the South Eastern Sydney Area Laboratory at the Prince of Wales Hospital in Sydney.

Gene testing can help families living with TSC to make decisions about having children. If parents have either of the two genes causing TSC they will pass it on in one out of two births. For the remaining 70%

of people, the condition results from new gene mutations which rarely recur in other children. This is the case with Lizzie, who is the only one of Sue's five children affected. Previously testing was only possible by sending blood samples to the UK or the US. From mid next year it should be available in Australia for around half the cost thanks to the new service, says Dr David Mowat, a clinical geneticist at Sydney Children's Hospital in Randwick and at the TSC clinic.

Dr Mowat believes there could be international demand for the service. He expects better access to genetic information will "restore options" for parents of a child with TSC as well as reassuring siblings who also worry about their risks. At the same time, gathering information about different mutations will increase understanding and could ultimately help scientists to develop better treatments.

FOR MORE INFORMATION

Contact The Australasian Tuberous Sclerosis Society on 1300 73 34 35 or visit the website www.atss.org.au.

Epilepsy Action congratulates the work of the ATSS. We have a large client group in common and are committed to working closely together.