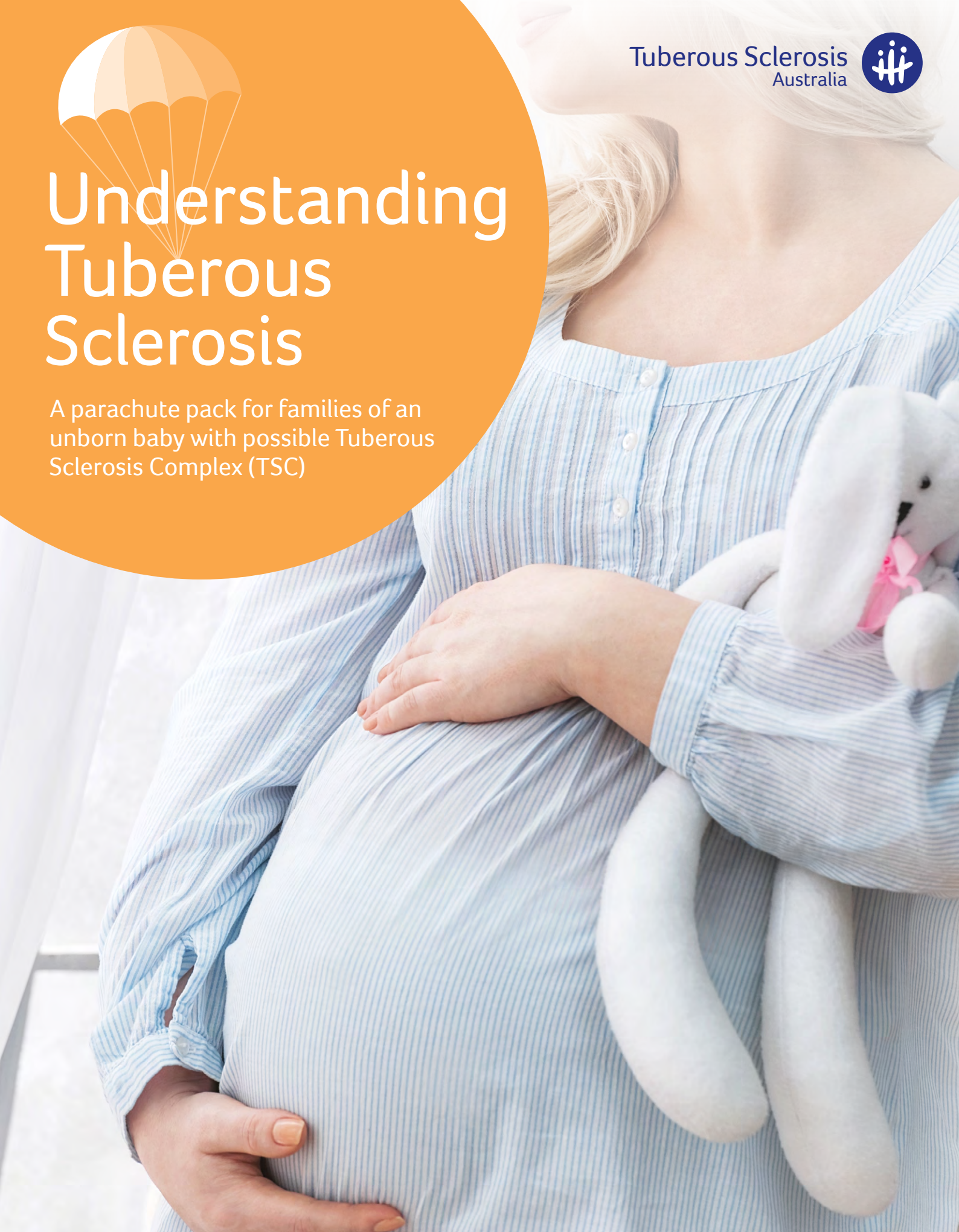
Tuberous Sclerosis Australia

Understanding Tuberous Sclerosis

A parachute pack for families of an unborn baby with possible Tuberous Sclerosis Complex (TSC)



While there is no crystal ball, there is hope

You have been given this booklet because your unborn baby may have tuberous sclerosis complex (TSC), possibly after abnormal clusters of cells have been seen in their heart on an ultrasound. Like most people, you may have never heard of TSC before. TSC is a genetic condition which causes different signs and symptoms in each person who is affected.

Being told your unborn baby may have TSC is a shock. Many families are overwhelmed by their feelings, including being confused by information about TSC, and being afraid of what the future might hold – and all of this before you have even met your baby. You may be feeling something similar.

Some people describe this experience as being in 'free fall'. This is the reason we have called this resource a Parachute Pack. We hope it will help you through this difficult time.

There is no correct way to feel or act when you find out your unborn baby may have TSC. Some people want to learn everything they can about TSC and will spend hours reading about new research or stories of people living with TSC. Other people find they are not ready to learn more about TSC for many weeks, months or even years.

It is possible that receiving a prenatal diagnosis of TSC may have presented you with a heartbreaking choice of whether to terminate your pregnancy. You may benefit from the support that a team of professionals can provide. This may include your doctor, a genetic counsellor and/or a social worker.

It is possible for TSC to cause growths in many organs of the body, including the brain, skin, heart, lungs and kidneys. TSC can also cause epilepsy, developmental delay and autism. However, TSC affects each person differently.

Even people who share the same TSC gene change can have different signs and symptoms of TSC. There is no reliable way to predict how TSC will affect your baby.

In the last 10 years research has changed the way we understand, manage and treat this complicated disease. This means if your baby has TSC, they can benefit from new treatments which can reduce the impact TSC will have on their life.

Progress started in the 1990s when the genes that cause TSC were discovered. This led to the first medicines specifically for TSC being approved in Australia in 2013. Research has also refined the use of other treatment options for TSC. The medical approach to TSC is changing – from a focus on treating symptoms as they arise to a focus on active monitoring and trying to prevent signs and symptoms from occurring.

We hope that this Parachute Pack will help you to understand TSC and help your family during this difficult time.

"I can remember hearing the words 'tuberous sclerosis' for the first time when Michelle was pregnant with Oliver. We were confused and afraid of what our child's future would be. If I could go back in time, I would tell myself that, despite the challenges of the future, you will still enjoy the journey. You have a lot to learn but your child is really not any different to any other child. They will have their own personality and they will have so much joy in their life."

Jeremy, NSW

If your child has already been born, we recommend our Parachute Pack for families of a child newly diagnosed with TSC. You can download this from parachute.tsa.org.au

When TSC information and stories are overwhelming

It is easy to be overwhelmed by what you read about TSC. This is the reason many doctors will still tell parents: "Do not google TSC". There are many reasons why the information you read may cause you distress.

1 Many of the stories you read online about people living with TSC will be from people where TSC has had a significant impact on their life. When TSC has caused severe symptoms such as difficult to control epilepsy and disability, people are more likely to share their stories and get involved in organisations like Tuberous Sclerosis Australia. It may help you to remember that many people with TSC are mildly affected and simply living their lives and not discussing their TSC in public forums.

2 Even factual information can give the impression that TSC always causes a lot of different medical conditions. Remember, most people with TSC will not experience all signs and symptoms.

3 Some information is out of date. The outlook for a child born with TSC today is different to a child born a generation or even ten years ago. This is because we have a better understanding of TSC. Now we are able to look for signs and symptoms of TSC before they cause problems and minimise their impact with several different treatment options.

"When my son Koby was born, the information I found online was confronting and scary, and it seemed to focus only on the worst case scenario."

Fiona, QLD

You may find it helpful to read a variety of stories, including from people for whom TSC has not caused major disability or health complications. We have collected stories from a variety people living with TSC at parachute.tsa.org.au

How might TSC affect your baby?

This page highlights the signs and symptoms of TSC that are most common in early childhood. It will take you some time to become familiar with medical terminology related to TSC. You may find it helpful to concentrate on just the terms on this page to avoid becoming overwhelmed.

TSC may cause changes in your baby's heart

TSC may have caused changes to the cells in your baby's heart. These clusters of abnormal cells are called cardiac rhabdomyomas. These can often be seen on ultrasounds from as early as 16 weeks gestation, and may have been the first sign that your baby could have TSC.

Cardiac rhabdomyomas grow while a baby is growing in the womb but will stop growing after a baby is born. In most cases, they start to shrink after birth. In most babies with TSC, cardiac rhabdomyomas cause no problems. In some babies, these growths affect how blood flows through the heart. Medication to help the heart or shrink the rhabdomyomas quickly, can assist. It is rare that surgery is required.

"We feel confident the early diagnosis and intervention Oskar received has helped him have the very best start to life with TSC."

Miia, NSW

TSC may cause changes in your baby's brain

If your baby has TSC, they may have one or more different types of changes in their brain. Some of these cause problems and some do not. The types of changes are called: cortical tubers, subependymal nodules (SENs) and subependymal giant cells astrocytomas (SEGAs).

TSC may cause seizures

If your baby has TSC, they will be at risk of having seizures and being diagnosed with epilepsy, particularly in the first two years of their life. A seizure can occur when a cortical tuber produces an abnormal electrical discharge in the brain. This discharge can spread to other parts of the brain and cause their normal functions to be abnormally activated, such as brain cells that cause arms and legs to move. There are many different types of seizures. Some of the seizures that young children with TSC experience can go undetected.

Frequent seizures in babies and very young children can interfere with normal brain development or learning. Seizures can also lead to accidents when a child cannot control their body. This is why it is very important to prevent or control seizures. One way this is done is by regular EEG tests to look for early signs of seizures in babies and very young children with TSC.

TSC may cause challenges with learning

Many, but not all, children with TSC will require additional support to learn. If your baby has TSC, they will be more likely than children without TSC to have delays to their development, specific learning difficulties and autism. Monitoring your baby's development and intervening early with therapy and seizure control, can all reduce the chance of them having learning challenges.

TSC can cause other changes

If your baby has TSC, they may also have changes to cells in other parts of their body. These include the eyes, skin, kidney and lungs. Many of these will not cause problems. For those that do, there are treatments available.

These changes may happen as your child gets older. This is why there is a list of recommended tests that will be repeated regularly to look for these changes and help decide if any treatment is required.

"I can clearly remember the day we started on this roller coaster with Oliver's TSC. It was a Friday. In the middle of our 28-week scan the sonographer stopped and consulted a large book. We did not know what was going on."

Jeremy, NSW

What has caused TSC in your baby?

When TSC is detected in an unborn baby, it is often because cardiac rhabdomyomas have been seen on a prenatal ultrasound. When this happens, more tests are required to find out if the baby has TSC. This is particularly important if more than one cardiac rhabdomyoma is found, because it is then likely the baby has TSC.

There are several options for diagnosis that your family can consider. Your health care team will be able to discuss these options in more detail with you.

1. Clinical diagnosis: Diagnosing TSC by signs and symptoms

In an unborn baby, this usually involves an MRI of your baby to look for signs and symptoms of TSC in their brain.

Even if this test does not find any additional signs or symptoms of TSC, it is possible your baby may still have TSC and further tests will be done after your baby is born.

2. Genetic diagnosis: Diagnosing TSC by a gene change

TSC can be diagnosed if a specific change in either the *TSC1* or *TSC2* genes is found. In an unborn baby, this is done through a test called amniocentesis. A sample of fluid is taken from the womb and tested for the gene changes known to cause TSC.

Even if this test does not find a TSC gene change, it is possible your baby may still have TSC and further tests will be done after your baby is born.

3. Waiting until the baby is born to do further tests

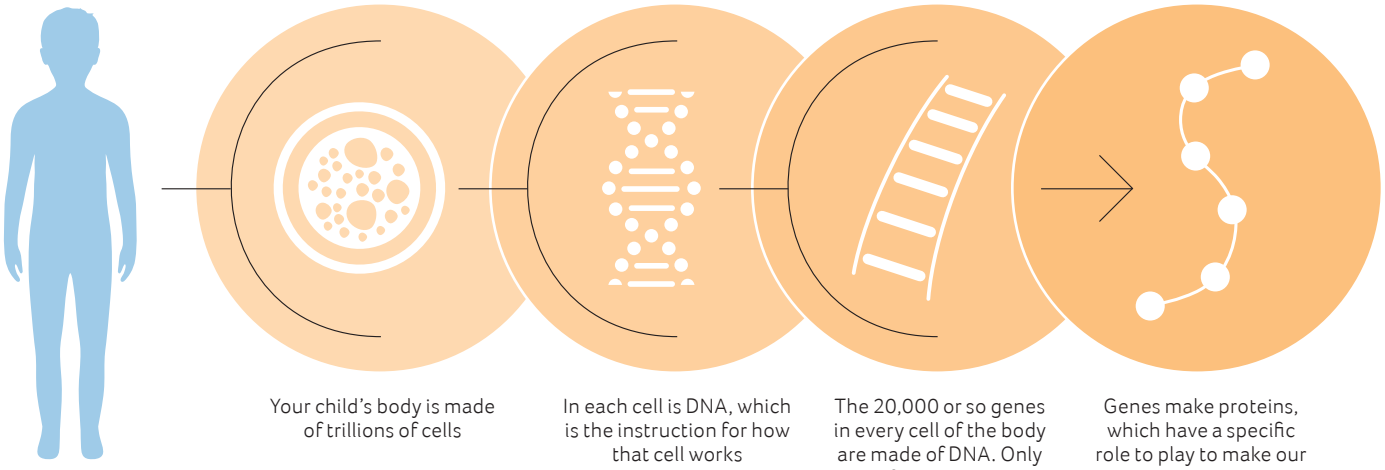
Some families decide to wait until after their baby is born to do either clinical or genetic testing to find out if their baby has TSC. These tests should be arranged immediately after the birth of your baby. If this is what you decide, we suggest that you still prepare for your baby having TSC, including by connecting with your health care team and discussing early monitoring for seizures.

You can visit parachute.tsa.org.au for more information about how TSC is diagnosed.

First, a gene change

If your baby has TSC, they have a change in either the *TSC1* or *TSC2* gene. These changes are sometimes called 'mutations'. All of our bodies contain many gene changes, but only some of them are known to cause genetic diseases.

Genes Make Proteins



There are two possibilities for how a gene change occurred:
1. The gene change was passed onto your baby, from either their mother or father who also has TSC
OR
2. The gene change occurred around the time your baby was created from an egg and a sperm. This is often called a 'spontaneous mutation'. In this case, your baby is the first person in the family to have TSC.

Around two-thirds of children born with TSC have a spontaneous mutation. It may not be clear in your family if your child is a spontaneous mutation. A genetics service can help you understand how your child's gene change occurred.

Second, the gene change causes cells to grow differently

The TSC gene change stops the gene from producing the protein that the body needs to control cell growth. This is why TSC often causes clusters of cells to grow more than they should. The lumps that TSC causes are the result of too many cells growing or cells growing more than usual.

The proteins made by the *TSC1* and genes work together to control cell growth. This is why a gene change in either of these two genes can cause a person to have TSC.

Why is TSC called TSC?

The name tuberous sclerosis comes from how one cluster of abnormal brain cells was first described by doctors. They described these abnormal cells as looking similar to a potato – a 'tuber'. The medical word for 'hard' is 'sclerosis', derived from a Greek word. This is how the name 'tuberous sclerosis' came into being.

Medical research revealed that people with TSC had many different signs and symptoms, not just tubers in their brain. The term 'tuberous sclerosis complex' was adopted to reflect the fact that a number of different organs may be involved in TSC. The acronym TSC is also helpful to distinguish from other 'TS' conditions such as Tourette's syndrome and Turner's syndrome. It is common for doctors and people living with TSC to refer to the disease as simply tuberous sclerosis.

"Because there are so many areas of the body that could be affected by TSC, I found not knowing how our little girl would be hard to deal with."

Linda, NSW

Your health care team

Because TSC can affect many different parts of the body and many different aspects of your child's life, a number of different health professionals are involved in their care. The team will change depending on:

- The age of your child
- Which signs and symptoms of TSC affect your child
- How health care is organised in your local area.

Before your child is born, your team will usually include:

- Maternal and Fetal Medicine specialist – a doctor with special training in looking after women and babies with health conditions or risks
- Cardiologist - a doctor who has special training in the heart
- Geneticist and/or genetic counsellor – health professionals with special training in genes and genetic diseases.

"We have a little book for Caleb, where we write down all his appointments, everything that happens to Caleb in hospital, all the information we have learnt from talking to TSA. I feel really nerdy about it, but it is a great support for us to have this. Our brains are just so full of everything, it is hard to remember. When a nurse commented: "Miriam is really organised, she won't forget this stuff", this was the first time I felt that I wasn't a pain, that I am actually doing something."

Miriam, TAS

You may also start to connect with other professionals who will form part of your child's team once they are born:

- Paediatrician – a doctor with special training in the health of children
- Neurologist – a doctor with special training in the brain and nervous system.

These professionals can answer your questions about TSC and help you to plan for the arrival of your baby, including early monitoring for seizures.

The importance of expert centres

TSC is a relatively rare condition, so not all health professionals have accurate and up to date knowledge about it. TSA maintains a list of Australian health professionals called the TSC Health Professionals Network who we know have up to date knowledge about the disease. These health professionals like to see families with a possible or confirmed diagnosis of TSC as soon as possible so that they can plan tests and ongoing monitoring.

If you are unsure about whether your doctor has up to date knowledge of TSC, you can:

- Provide them with information from TSA
- Talk to TSA's Nurse to check against the latest research and clinical guidelines
- Ask your doctor to refer you to a member of the TSC Health Professionals Network.

Living outside a capital city

Many children with TSC visit specialists in a capital city. Each state and territory has a scheme that provides financial assistance for families living in regional areas who need to travel for their health care. For more information, talk to your child's doctor or visit parachute.tsa.org.au

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TSC is a relatively rare condition, so not all health professionals have accurate and up to date knowledge.

Tests and treatments to be aware of if your baby has TSC

If your child is diagnosed with TSC they will have a series of tests to help understand how TSC is currently affecting them. These are known as 'baseline tests'. The results of these important tests will help your child's doctors identify any treatments that are required. They will also be used in the future to compare results to see what has changed.

Early tests for seizures

An EEG test looks for early signs of seizures. An EEG measures the electrical activity within the brain using small metal discs stuck onto the outside of a child's head. An EEG may show abnormal electrical charges in the brain before your child has their first seizure. Treatment at this stage may reduce the chance of your child developing troublesome seizures.

Vigabatrin (also called Sabril)

Vigabatrin is a medication that is very effective for controlling seizures in young children with TSC, including infantile spasms. Because stopping a young child's seizures and the effects seizures have on the developing brain is important, this medication is recommended as the first choice for treating seizures in TSC, including infantile spasms. Your doctors will talk to you about weighing up the risks of this medication with its benefits.

Oskar having an EEG

"I believe the early intervention helped Koby enormously with seizure control. It has helped him be the best version of himself that he can be."

Fiona, QLD

Epilepsy surgery

In some children with TSC there is a single tuber starting most seizures. Surgery to remove this tuber may be the best way to control seizures. Your child's neurologist will be able to talk to you about whether surgery is an option for your child.

mTOR inhibitors (also called Afinitor, Everolimus, Sirolimus and Rapamycin)

These are relatively new medicines for TSC that can shrink some clusters of abnormal cells and treat some types of epilepsy caused by TSC. Ongoing research projects are helping to understand when a person with TSC can benefit most from these medicines.

An mTOR inhibitor skin cream can be used to treat bumps, called angiofibromas, which can grow on the face of a child with TSC. This skin cream can be used as soon as these small bumps appear and prevent the angiofibromas from forming.

You can discuss these treatments and others with your health care team or contact TSA for more information.

Your family's journey with TSC and how TSA can help

TSC is complicated. From now on, you will start to learn a lot about TSC.

Right now, you may need time and support to adjust to the news that your unborn baby may have TSC. You may have hopes and dreams for your child that may feel out of reach now. You may be feeling alone and scared about what the future will hold. Emotional support may come from your friends, your family and from professionals.

Telling friends and family

It is sometimes difficult to know when and how to share the news that your baby may have TSC. Some parents decide to share this news during their pregnancy, others wait until their baby is born.

You may find that some friends and family cope better with this news than others. You may find that through this experience you connect with new people and find support in unexpected places.

It is likely that your friends and family do not know about TSC. You may find it helpful to share this information with them, or refer them to parachute.tsa.org.au

TSA is here to help you and your family

Tuberous Sclerosis Australia (TSA can provide you with information and support in various forms. We encourage you to contact our team to learn more about TSC, the work of TSA and to connect with other organisations, TSC families and/or health professionals that can help you and your child.

Find out more about TSA's services and other organisations that can support you and your family at parachute.tsa.org.au

TSA's nurse provides telehealth information and support to families affected by TSC

"Right from that first phone call, TSA has been essential to our life with TSC. At no time has TSA told me what to do, but I have been able to come back and ask them what they think about the options we have. They have helped our family make life changing decisions and be confident in these decisions."

Dawn, ACT

"If I could travel back in time to the day of that first ultrasound of Greta I would tell myself it is all going to be OK. You're stronger than you ever knew you could be. I'd also remind myself that my husband is an amazing man and this challenge will make our relationship stronger and teach us truly how to not sweat the small stuff. Yes, there are bad days, but life is pretty good."

Bridie, VIC

Questions for your health care team

Tuberous Sclerosis Australia

This is a publication of Tuberous Sclerosis Australia (TSA)

TSA is the only charitable organisation focused on improving the lives of Australians with tuberous sclerosis complex (TSC). We work to empower people and families living with TSC through access to the best treatment options, up to date information and support.

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nib foundation

Disclaimer

Please note that the information provided in this publication is not medical advice. You should consult your doctor(s) before taking any action based on any information published by TSA.