Reach Out

May 2013 Issue 97

Australasian
Tuberous Sclerosis
Society

www.atss.org.au

Contents

Australasian Tuberous Sclerosis Society Inc.

Who are we?

The Australasian Tuberous Sclerosis Society is a voluntary organization established in 1981 to connect families affected by Tuberous Sclerosis Complex. There are hundreds of members around Australia, New Zealand and internationally, including families living with TSC and professionals from the medical, caring and education fields.

Our Vision:

That TSC families are empowered through access to information and support.

Our Mission:

As the only Tuberous Sclerosis organization in Australia and New Zealand, we:

- Provide access to current information and resources;
- Help TSC families build relationships and support networks;
- Ensure best practice management and care for TSC affected people in Australia and New Zealand.

Need more information about living with TSC?

More information about Tuberous Sclerosis Complex is available at:
The ATSS website: www.atss.org.au
Tuberous Sclerosis Alliance, USA: www.tsalliance.org
Tuberous Sclerosis Association, UK: www.tuberous-sclerosis.org
Email or call us to get in touch with a volunteer in your area. ATSS has regional contacts throughout Australia and New Zealand that can help you with the challenges of living with TSC and connect you with services and other families in your local area:
1300 733 435 or info@atss.org.au

Not in Australia or New Zealand?

ATSS is a founding member of Tuberous Sclerosis Complex International, a worldwide association of Tuberous Sclerosis Complex organizations.

Tuberous Sclerosis Complex international, www.tscinternational.org

Editorial

David Matheson – Editor

Over many years I have heard and read of lots of stories from people affected by Tuberous Sclerosis Complex. Each story is unique. Sometimes the stories are so vastly different from each other that it is difficult to imagine that the people involved are affected by the same condition. This is the nature of TSC: a wide range of possible symptoms that can vary markedly from one individual to another.

> *"Diagnosis is followed by a steep learning curve of becoming familiar with the symptoms and treatments of TSC.."*

Because of the varied nature of TSC it is difficult to come up with a list of common themes in the stories of those affected. Yet, that is what I am now attempting to do! The experiences listed below are not in every TSC story, but emerge frequently in the stories that are told and written about living with TSC.

- The experience of diagnosis is one of devastation and despair.
- Diagnosis is followed by a steep learning curve of becoming familiar with the symptoms and treatments of TSC, as well as understanding health service systems.
- There is a pattern of frequent visits to doctors for tests and treatment, which can sometimes be reassuring, but at other times be terrifying.
- Life eventually settles into a new routine, different from what went before.
- Individuals and families affected try to live day by day and deal with each new challenge as it arises.
- Achievements, however small, are something to be celebrated.
- Being an advocate for yourself or your child is important to obtain the best care and services.
- Support from family, friends or some people that understand is vital to help cope with the daily life of living with TSC.

This issue of *Reach Out* again presents a range of stories from individuals and families affected by TSC. There are stories that describe the experience of TSC over a number of years. There are also stories about particular events, such as in the Celebrations page, or particular aspects of living with TSC, such as in the new Just Between Us section. Again, while each story is unique, some of the themes listed above seem to be present.

ATSS is always looking for new ways to provide information and support for those affected by TSC. The internet and social media are now often the first places that people turn to.

The ATSS website was redesigned last year and provides a vast amount of information about TSC, details of various services available, stories form people affected, and links to other useful sites. The Facebook pages, 'Australasian Tuberous Sclerosis Society' and 'Discussions of Australasian Tuberous Sclerosis Society', as well as the ATSS Twitter stream, provide information about current events and a way to connect with others affected by TSC.

> *"Meeting others who are affected is reassuring and supportive."*

Although the use of social media has now become the norm in many forms of communication, the value of face to face contact remains. Many TSC families find that meeting others who are affected is reassuring and supportive. It provides the opportunity share stories and to hear the experiences of others. Picnics will be held across Australia and New Zealand on 19 May that will enable TSC families to meet others. I encourage you to make the effort to attend one of these picnics. Details are in this issue of *Reach Out*.

Australasian Tuberous Sclerosis Society

TSC Tuberous Sclerosis Complex International
A Worldwide Organization of Tuberous Sclerosis Complex Associations

Reach Out Official journal of the Australasian Tuberous Sclerosis Society, Inc.
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President's Report

Debbie Crosby - President

It is with great pleasure that a write my first President's Report since becoming President of ATSS last year. Since joining the ATSS Committee a couple years ago I now understand what an amazing job our volunteers and Project Manager do in keeping ATSS running, and supporting individuals and their families affected by Tuberous Sclerosis Complex.

Taking on the role of President has been very rewarding as I get to meet so many people and families, and it is a privilege representing ATSS in the wider community. I look forward to meeting more of our members and families in the future.

It has been a very busy six months since our last issue of *Reach Out*. Part of our strategic plan is to enhance the information provided by ATSS via our website, and provide up to date information to individuals and families. Our focus on online communication has been very successful with the new website, monthly email newsletters and blog posts on our website. The ATSS facebook group has grown; connecting with others and sharing our thoughts and stories on a more regularly basis is an important part of feeling supported and getting the latest information about TSC.

The TREATMENT clinical trial, which is studying the topical use of Rapamycin to treat angiofibromas, has now been running for over 6 months. This trial is being run at Sydney Children's Hospital with approximately 20 participants, including both children and adults, and is part of the international study which includes ten other centres in North America. The trial has been approved for an extension of a further 18 months, collecting even more data to help us understand the long term implications of the use of this medication.

After many generous donations which successfully raised over $200,000 to make this trial possible, it is great to see the trial up and running. We look forward to seeing the results and then working towards making the cream accessible to everyone at an affordable price.

In October 2012 ATSS made a submission on behalf of all families living with TSC to the Pharmaceutical Benefits Advisory Committee (PBAC), which considered listing Everolimus for PBS subsidy. Everolimus can be an effective treatment for Subpendymal Giant Cell Astrocytomas (SEGAs) caused by Tuberous Sclerosis Complex, and having this medication subsidised through PBS will allow greater access to this treatment for everyone. I would like to thank the families who made personal submissions to the PBAC. The PBAC's decision was handed down in January and the panel deferred making a recommendation. However we did have very positive feedback regarding our personal submissions. They were very impressed with the amount of the submissions and the information included. We will keep on working towards gaining approval of Everolimus on PBS in the future.

On 2 & 3 March 2013 ATSS held a conference in Perth for the first time. It was wonderful to meet people from Western Australia and those who travelled from other states as well. It has been a long term goal of ATSS to hold a conference in Perth and the conference was very informative. It was great to see people share their stories, ideas and connect with one another. Clare Stuart, ATSS Project Manager, did a fantastic job organising the conference and I would like to thank everyone from Perth who helped us out in making the conference a success.

Our Annual Seminar Day will be held in Sydney in August 2013, and more information will be available on our website and in our newsletters in the near future.

Another important day approaching is TSC Global Awareness Day on 15 May 2013. There will be many activities and events planned around the world to raise awareness of TSC.

Here in Australia ATSS are planning picnic days around the country and a media campaign; we encourage everyone to get involved. If you would like to plan an event or have any other fundraising ideas please contact ATSS.

ATSS's vision is that TSC families are empowered through access to information and support.

I look forward to the future as President of ATSS and working towards new and innovative ways in supporting individuals and families affected by TSC.

Treasurer's Report

Hayley Hill - Treasurer

We are already closing in on June 2013. The year is flying by.

ATSS held a conference in Western Australia on 2 and 3 March. It was a very productive conference and many people attended from Western Australia, South Australia, New South Wales, Australian Capital Territory and Tasmania. It was great to see so many new faces and catch up with others that have supported ATSS.

The conference dinner provided a chance to talk and swap information and strategies, or get to know someone else who is dealing with TSC in their lives. Many people commented positively on the conference, and especially on how amazing Professor Petrus deVries was. The people who attended the conference loved the fact that so many doctors and professionals gave up their own time to present. Thank you to all the professionals who made this event possible.

The conference could not have happened without the grants from Lotterywest, Novartis, and the Department of Families, Housing, Community Services and Indigenous Affairs. Thank you.

As at February 2013 ATSS has paid $132,247.97 for the Rapamycin Topical Cream Trial. There is still an expected $112,049.50 remaining to be paid over the next year. I am pleased to say that Sydney Children's Hospital is seeing some positive results from the trial. This trial couldn't have taken place without all the generous donations from you, your families and friends.

It is getting close to the end of financial year and Supporter fees will be due on 1 July 2013.

If you would like to take advantage of tax deductibility on all donations over $2, you can do so by renewing your Supporter fees or make a donation before 30 June 2013.

Thank you again for supporting ATSS during 2012-2013. This allows ATSS to provide support services, information and events to families around Australia and New Zealand living with TSC.

Another option is to become an ATSS Champion, setting up a monthly donation to ATSS. This option is on our website in the Ways To Help section. Perhaps you can ask friends or families to donate on a monthly basis. As little as $10 per month makes a real difference to funding our work.

All the best for the rest of the year and I will update you once again in our November issue of *Reach Out*.

CBA Cheque Account	$21,474.40	
CBA Cash Investment Account	$79,503.88	
Macquarie Term Deposit	$31,645.04	
Macquarie Cash Management	$7,807.10	
CBA Term Deposit	$113,025.46	Matures 19th May 2013
CBA Term Deposit Elizabeth Pinkerton	$5,034.03	Matures 19th May 2013
Total	**$258,489.91**	

As at 28 February 2013 ATSS held accounts with Commonwealth Bank & Macquarie Bank.

Membership Report

Hayley Hill - Membership Secretary

ATSS Supporter Renewals are due on 1 July 2013. Become a Bronze, Silver or Gold Supporter for 2013.

If you would like to take advantage of tax deductibility before 30 June, you can make a donation or renew your support of ATSS. Please complete the bottom half of the Supporter Renewal form that is enclosed with your copy of *Reach Out* Or Renew Online – www.atss.org.au

Click on renew your financial support now (right hand side) and complete using your credit card. By renewing online you will receive an automatic receipt from ATSS that you can have ready for tax time.

Currently there is a total of 131 Supporters of ATSS and 22 Honorary Members. Discussions of ATSS (Facebook group) has 234 members – there is always support to all that are posting on the page. Thank you to everyone who contributes their opinions or strategies: it can really put someone at ease knowing they are not alone and that there are others out there dealing with similar situations. If you aren't a member of this discussions page, go to Discussions of Australasian Tuberous Sclerosis Society and click Join Group.

Our wonderful Clare Stuart is always amazing and has been publishing a monthly newsletter that is sent to the email addresses that ATSS has on its database. As *Reach Out* is published twice a year, ATSS decided that a monthly newsletter would be a great way to keep supporters up to date with news and events. ATSS hope that you have all been enjoying this. If you would like to receive the monthly newsletter go to www.atss.org.au and complete your name and email address to be a subscriber.

TSC Global Awareness Day is on 15 May. TSC Awareness Picnic Day will be on Sunday, 19 May. I hope you can take some time and go to one of the picnics being held throughout Australia and New Zealand. More information is available in this issue of *Reach Out* and on the ATSS website.

It has been a pleasure to once again volunteer as Treasurer and Membership Secretary for another year.

Branching Out in Perth

The 2013 ATSS Family Conference was held in Perth on 2 and 3 March. This was the first time ATSS has held a major event in Western Australia. It was a great opportunity for people from Perth and surrounds to meet each other and at the same time learn more about Tuberous Sclerosis Complex. Many families also travelled from other parts of Australia to attend. The TSC families that attended made valuable connections and enjoyed the conference dinner held on Saturday night.

The keynote speaker for the weekend was Professor Petrus de Vries. Petrus is an experienced neuropsychiatrist and a leading researcher into the behavioural, intellectual, learning and mental health challenges experienced by people with Tuberous Sclerosis Complex. In addition to speaking at the Family Conference, Professor de Vries spoke to a professional audience of neurologists and psychiatrists, raising awareness of these aspects of Tuberous Sclerosis Complex, and the developments in research and new treatments for TSC.

The family conference included a variety of talented speakers covering topics such as neurology, genetics, autism, the kidneys and the skin. Several speakers outlined the services available to individuals and families living with TSC offered by organisations such as Kalparrin, the Neurological Council of Western Australia and Epilepsy Action Australia. All speakers donated their weekend time willingly and ATSS is very grateful for their support. The conference was made possible by the hard work of several ATSS volunteers in Perth: Sue Williamson, Nicole Stone, Karen McCagh, Samantha Stone, and Joelle and Peter Neville.

Recordings of some sessions from the weekend will be made available on the ATSS website or you can contact ATSS for a copy on DVD.

Conference attendees

Dr Lakshmi Nagarajan

Professor Petrus de Vries

Nicole Stone sharing her TSC story

Enjoying a coffee break

Conference attendees

Thank you to the following organisations for their support of the 2013 ATSS Family Conference:

The conference was supported by an educational grant from Novartis Pharmaceuticals Australia

Lottery West provided a generous grant to minimise costs to attendees

Funds for Travel Grants were provided byFaHCSIA's National Disability Conference Initiative

The Country Comfort Inter City Perth offered discounted rates for the conference venue

Operation Lend A Hand

In March, Sydney mother of two, Leanne Park, was grateful to receive a helping hand from staff volunteers from NetApp. Leanne's son Luke has Tuberous Sclerosis and was one of the stars in ATSS's 2007 DVD production 'Tuberous Sclerosis: You are not Alone'. Leanne has also previously volunteered as a Committee Member and Vice-President of ATSS.

Using their paid volunteer leave, staff at the company spent five days renovating Leanne's house and garden. The project was the brain child of NetApp staff member Elliott Cunnew, father to Amelia, who has TSC. Elliott pitched the idea to his company and his colleagues got together to not only do the renovations, but also to fund donations and give Leanne and her family a holiday while their house was being worked on.

Operation Lend A Hand is a wonderful project that has come from connections made through ATSS. Thank you to Elliott and the staff at NetApp.

Some of the volunteers who worked on Operation Lend A Hand

Leanne Park returning home at the end of the week

TSC Awareness Picnic Day

North Queensland
Date: Sunday 19 May
Location: Cairns Esplanade (Northern End)
Time : 12.00 p.m.
RSVP: Fiona Hiatt **Telephone:** 0419787521 by 17 May 2013
Please bring own picnic lunch and chairs/blanket.

South-east Queensland
Date: Sunday 19 May
Time: 10.00 a.m.
Location: Capalaba Regional Park, Pittwin Road, North Capalaba
All abilities playground (includes Liberty Swing)
RSVP: Tamara Goong **SMS**: 0412 199302 or
Telephone: (07) 3206 0975
Please bring own picnic lunch and chairs/blanket.
Look for the blue and white balloons!

Victoria
Date: Sunday 19 May
Time: 12.00 p.m.
Location: Princes Park Playground, Cemetery Drive, North Carlton
RSVP: Email: kate.green10@gmail.com by 12 May 2013
Please bring own picnic lunch and chairs/blanket.

New South Wales
Date: Sunday 19 May
Place: Timbrell Park "Livvi Place", Henley Marine Drive, Five Dock
Time: 11.00 a.m.
RSVP: Hayley **Telephone:** (02) 9920 0261 or 0402 074 326
Please bring own picnic lunch and chairs/blanket.

South Australia
Date: Sunday 19 May
Place: Bush Magic Playground, near Adelaide Aquatic Centre, Jeffcott Rd, North Adelaide.
Free parking in Aquatic Centre car park; playground fully fenced; barbecue facilities available.
Time: 11.00 a.m.
RSVP: Tania Colman **Telephone:** 0403 397 003
Please bring own picnic lunch and chairs/blanket.

Tasmania
Date: Sunday 19 May
Place: Punchbowl Reserve, Launceston
Time: 11.30 a.m.
RSVP: Georgine Schilg **Telephone:** 0438 918 165 or
Email: sandgschilg@bigpond.com
Please bring own picnic lunch and chairs/blanket.

Western Australia
Date: Sunday 19 May
Place: Kings Park Lotterywest Family Area (grassed area outside Ivey Watson Playground)
Time: 11.00 a.m.
RSVP: Nicole Stone **Telephone:** (08) 9446 9151
Please bring own picnic lunch and chairs/blanket.

New Zealand
TSC New Zealand Awareness Get Together Day on Sunday, 19 May 2013 in New Zealand
Locations: 3 Regions: Wellington; Waikato/Bay of Plenty; Auckland
In New Zealand there will be get togethers to meet others around the country. If you don't live in the areas above and are interested please make contact as more can be added.
Meet for lunch. Details will be worked out after you express you interest in attending. Call or email now.
RSVP: Sheryll Brasell **Telephone:** 06 3047 998 or email: sheryllbrasell1@xtra.co.nz as soon as possible.

ATSS Seminar Day and Annual General Meeting

The ATSS AGM and Seminar Day will be held in Sydney on 18 August.
Date: Sunday, 18 August 2013
Time: 8.30 a.m. – 4.00 p.m.
Venue: Sydney Childrens Hospital, Lecture Theatre 1, High Street, Randwick
Speakers: To be advised

Elections for positions on the Australasian Tuberous Sclerosis Society Committee will be conducted at the AGM. In accordance with the ATSS Constitution, nominations for positions must be made prior to the Annual General Meeting, must be made in writing, must be signed by two members of ATSS and be accompanied by the written consent of the candidate. More information about the Seminar Day will be available on the ATSS website.

At last, an explanation!!

Rosemary McDonald

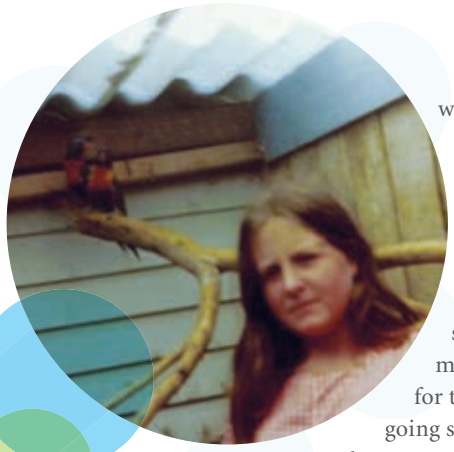
Hi, my name is Rosemary. I was diagnosed in October 2010 at the age of 46, with both Tuberous Sclerosis Complex and a rare lung disease, lymphangioleiomyomatosis (LAM for short). I live on the gorgeous Gold Coast and work for the Gold Coast City Council in their records department. Not having had any children and never marrying, I keep myself busy with my six pet parrots, which are a passion of mine, along with travel and gardening, reading and a few other interests.

About five years ago, I was finding it increasingly harder to exercise or even walk up slight inclines without becoming breathless. I was also having mild panic attacks when carrying moderately heavy shopping bags to the car. In 2010 my GP referred me to a respiratory specialist, where I undertook a lung function test. The results from this and a heart stress test led the specialist to a diagnosis of emphysema, even though I had never smoked before. Upon hearing this diagnosis I was very resentful of my mother, actually very angry to say the least. I had looked after her the year before she died in 2005 and I assumed second hand smoke was the cause of my contracting emphysema (wrongly, as I later learnt). She had smoked around me and my siblings while we were growing up and was diagnosed with emphysema in early 2001; even being told this was killing her, she refused to give up smoking until the day she died.

My GP then sent me for a total blood test and my Alpha-1-Antitrypsin levels were normal as well as Phenotype M, which I understand is not normally the case for those with emphysema and have never smoked. From there I was sent back to the respiratory specialist, had a CT scan of the thorax. It came back showing my lungs had multiple cysts throughout and I also had multicystic kidneys.

I was advised that I had LAM. While I stood there looking at the scans and all the black dots on my lungs I knew this wasn't good and had to sit down. Feeling the blood drain from my face, I asked "How long do I have?" thinking he would say "Oh, you have years and years". He said maybe 10 years or so, depending on how severe the progression of the condition becomes, and that there was no cure. I will likely need oxygen as the disease progresses and a double lung transplant was the only treatment to date. From there, I was advised that there were two other conditions that were associated with LAM, one being Tuberous Sclerosis, and the other being meningioma. The doctor wanted me to have a brain MRI and kidney CT scan.

Rosemary McDonald (left) with her brother and sister



After the consultation I walked out into the beautiful sunny day and wondered how could something so horrible and unbelievable have just occurred on such a perfect day. My world felt like it had spun out of control and my mind was working overtime for the next couple of days, going slightly overboard, thinking about organising a will, prepaying for a funeral. I then "got a grip" on myself and thought "have things really changed since before the diagnosis?" No, so I carried on with my life much the same as I had done before and used the wisdom of Winston Churchill: "Keep calm and carry on"! The brain MRI confirmed that I had Tuberous Sclerosis but luckily no meningioma.

"My world felt like it had spun out of control and my mind was working overtime."

The CT of my kidneys showed multiple cysts also. About two months later, I had my first consultation with my neurologist and I was put through the battery of tests for visual, cranial nerves, reflexes, etc, Luckily, I was advised that they appeared normal and that I was fortunate not to have epilepsy and to be of normal intellect. He advised that I had a hypomelanotic lesion on my hip and adenoma subaceum (facial angiofibromas) on my chin and nose I had seen a dermatologist regarding these spots several years ago and he diagnosed them as being rosacea, and gave me several sessions of dermabrasion, which did little.

It was a real "light bulb" moment, when I was told I had TSC and what that all involved. It just explained so much about my past that I didn't understand. I was born with the umbilical cord around my neck and this in turn caused blood vessels on one side of my face to burst and cause a large portwine birthmark. At around 3 months old my parents were awoken to me having a seizure and they rushed me to hospital. I am not sure what happened after that, whether a diagnosis was given or not and since both parents are now long deceased, I regret not enquiring further into my health records.

My mother always told me that she worried because I was slow in both walking and talking as a toddler. I was incredibly shy and found it difficult to make friends or try new things. My shyness was put down to my having the large scar on my face and being self-conscious (I had a skin graft operation at the age of 3). At primary school, my parents were advised to "hold me back" a year as I was not reaching the normal milestones for my age group, but my parents wouldn't hear of it, so, I had to try and keep up with other children. I absolutely HATED mathematics classes and vividly remember one teacher yelling at me because I could not answer one of the maths problems; it was humiliating and I hated that teacher for the rest of my time at that school. Other than maths, I enjoyed other subjects and went on to get a clerical cadetship at the High Court, where I was promoted up the ranks and stayed there for 15 years before leaving to join the Gold Coast Council and have been here since then.

"It just explained so much about my past that I didn't understand."

It is fantastic that there are now organisations such as the Australian Tuberous Sclerosis Society for individuals and families to turn to. Also, The LAM Foundation and LARA, for those women with LAM, are other support organisations. Let's hope the future is near where a cure or efficient treatment is discovered.

Olivia Amazes Me Every Day

Lana Cole

I love reading the stories of our amazing TSC kids so thought I would share the story of my little girl, Olivia. Olivia is 8 years-old and we live in New Zealand.

I helped to diagnose her at 9 months-old after three unsuccessful visits to our doctor and scouring the internet for information.

Never ever give up! You are your child's best advocate!

Armed with pages of printed out information I took us to Starship Children's Hospital in Auckland. I was seen relatively quickly and was told promptly to stop doing research as it could upset me.

The stubborn streak in me did not listen; I wanted to know everything!

After three days of tests in hospital it was confirmed that my baby girl had Tuberous Sclerosis Complex. As I had already prepared myself for this diagnosis I was not upset and was very relieved that the results were what I had discovered.

> *"Never ever give up! You are your child's best advocate!."*

Livvy was my youngest with three older sisters and an older brother. She is the only one with this condition in our family. All the tubers are contained in her brain: lots and lots of them.

She amazes me every day.

She is in a mainstream school and has a teacher's aide a few hours a day. Her speech is now only slightly delayed after lots of therapy. Her sentences are still very short but she lets everyone know what she wants!

She has a lot of absence seizures and can now recognise and tell others when she starts to have a seizure. She currently takes Epilim chewable tablets (Sodium Valproate). We will be adding a new medication soon to help curb the small seizures she regularly has. This will be a huge effort as she cannot swallow tablets and doesn't like liquid medications. But we will find a way!

Olivia

I'm not sure if anyone else has been paranoid at bedtime, but I've only just been putting her into her own bed in a room with her 10 year-old sister. She has always slept in my bed since she was two as I was scared I would never know if she had a seizure. As I am a single parent there was loads of room!

Every small change in breathing or twitch I would know, and was right there for her. But as she's getting older and I'm getting extremely tired from the wiggling worm, I've decided it's time for my baby to grow up and be a big girl. So with a fancy nightlight and some bribery with a glow stick, she happily became her sister's roommate.

Olivia is a bit of a girly girl. She learnt to ride a two wheeler bike last year and loves to climb! She is very very strong in the arms and upper body. She can hang on the monkey bars at school longer than any kid and is not afraid of heights! Olivia also loves to help mum make salads and bake.

Today she played netball for sports day: something she has never done before. Took her a while to learn how to catch and throw a ball correctly, but the joy on her face after catching one and throwing it to a teammate was priceless.

Thank you to all the parents and caregivers for sharing your stories through ATSS and the Facebook group. I think all the stories and experiences help to guide or relate to another family.

Celebrations

Want to share your family's news, milestones or triumphs?
We'd love to hear your stories and share them with others on this page.
Email info@atss.org.au, write to us or call us on 1300 733 435.

Celebrate…Believe…Achieve

Hi! My name is Lily Kerr. I am almost 10 years-old and I have TSC. I am in Grade 4 at Hampden Specialist School in Terang, Victoria, and I was appointed Campus Captain for 2013. I think the most important things about being a Campus Captain are helping other students, being a good leader, always being kind and caring, and being a good role model. The highlight of my year (so far) was having my photo in the local paper. My mum and dad and all my family are very proud of me and my achievements.

Greg's Graduation

This photo was taken on the night of Greg Silva's Year 12 formal in December 2012. From his mum Josephine: "The most nerve racking, anxious, and proudest night of our lives. Intractable epilepsy will do that to you!"

Greg Silva (middle) with his mother and sister at his graduation ceremony

Ravi's painting

Just wanted to share this painting that Ravi did at Kindy! Ravi is three and a half years old and has Tuberous Sclerosis. I can't express how awesome it is to me, as he's only ever done a couple! Such a proud Mummy I am. Amazing how we all appreciate the littlest things
Hannah, New Zealand

Blake

My son Blake is 17 and was diagnosed with TSC at age three. I don't think I can begin to understand the battles faced by Blake, and am so very proud of how hard he tries at school, and what a beautiful son he is. Blake is a super star in the pool and competed at the 2012 NSW All Schools Carnival (school state titles) in the multi class disability 50m freestyle and the relay and will compete again this year.

I've moved from tears to goose bumps every time I see him go off on his own to the marshalling area, when not that long ago it would have been impossible for him to do that. Good luck beautiful boy!
Suzie, Newcastle

Cooper

Sometimes it's the simple things in life. Like your boogie board blowing in the wind. A happy and relaxed Cooper enjoying the beach. Cooper has Tuberous Sclerosis and lives in the Southern Highlands in NSW.
Cooper's mum, Naomi.

Just Between Us

Sleep Disorders –
From Massachusetts General's 'Living with TSC' resource

Sleep disturbances, such as night waking, waking early, seizure-related sleep problems, and excessive daytime sleepiness, are some of the most common behavioural manifestations of TSC. These problems, which are sometimes caused by night-time seizures, affect approximately 60 percent of people with TSC. Studies that monitored the night-time electrical activity in the brains of people with TSC found that the majority experience many more awakenings and far less efficient sleep than do people without the disorder.

Oh my, where to start! Greg has always had a problem with falling asleep. Except when he was on Tegretol (Carbamazepine) it made him drowsy and practically seizure free for six years which was heaven for all of us. In the past I have tried Melatonin but that only worked for a couple of weeks. Greg can lay awake for hours, be up and down to toilet, with rituals galore every night. Because he sleeps in our bedroom due to seizures, it has greatly affected us and honestly seems to be getting worse as he gets older. If Greg gets eight hours sleep it's a miracle. He will need something for it soon before we go too crazy. Once he's asleep he sleeps like a log and doesn't want to get up in the mornings, for school especially! This can be very very frustrating as a parent.
Josephine, Wollongong

This is a new feature we'll be running in each issue of Reach Out. We'll publish the collective wisdom of TSC families from around Australia and New Zealand on a particular topic. For our first topic, we've asked members of the ATSS Facebook group for their thoughts. All medicines have their chemical name in brackets after the brand name. Remember that, like all information in Reach Out, this page represents the opinions of the author(s) and not official pronouncement of ATSS Inc. This information should not be used without first seeking medical advice.

We had such major sleeping issues, but thankfully have found the answer in Catapres (Clonodine). Hayley was only having 4 hours sleep a night and we could not take it anymore. For a non-verbal child she had plenty to say at 2 a.m.! She is also on Sabril (Vigabatrim), Lamictal (Lamotrigine), Epilim (Sodium valproate) and Frisium (Clobazam) for her seizures.
Linda, Lismore

Jacob had many sleep problems at 2.5 years and that continued to some extent for some years. There was some abnormal brain wave activity that we found when we did a sleep study. When Jacob was around 4 years old we added another anti-convulsant because the Epilum (Sodium valproate) was not enough but Lamictal (Lamotrigine) made the night time issues worse. Weaning the dose down helped. But he would still wake at around 3 a.m. until he was about 7 years. I think then it was about needing to pee. He is 10 now and sleeps well until 5 a.m., sometimes 6 a.m. Other parents I have spoken to use Melatonin (a naturally occurring neuro hormone) for their kids sleep problems which I believe was very helpful. Good luck.
Lisa, Perth

Kristin has her seizures under control now, but one of the things we found when they weren't is that her sleep was very disrupted, for a couple of reasons I think. When she was having seizures very frequently she would always sleep for a long time after the seizure, which would then mean she often wouldn't be tired at night. The other reason I think was that

...Sleep

at night our whole household was a bit disrupted. My husband and I both just had to check on her before either of us went to bed, and if we heard anything in the night we would again get up and check in case of a seizure. We all had our doors open to hear things, so it was no wonder she didn't sleep well. All that was complicated, of course, by the changing of multiple medications and it was always such a challenge to know which one was causing her to be hyper and which one dozy during the day.

Regarding ideas, there was no silver bullet, although we did put a dimmer switch in the hall light so when we did check on her we could just peer in with a dim light for background rather than a normal bright one. We also really watched what food she had for dinner and made sure she had dinner early in case food was hyping her up a bit more. Similarly, the old mantra of a really clear and consistent bedtime ritual - even if it meant we always came home early if we were out or just stayed home. Finally we decided to just de- emphasise it for all of us - we just had to accept tiredness, aim for us to all have some naps if we could on the weekend. We also told Kristin, and ourselves, that you can't make yourself sleep - as long as you stay on bed and rest it still does you good so just make up a dream you enjoy in your head and enjoy it, even if you are still awake.
Michele

Josh has had his moments with sleeping. Sometimes he is absolutely unbelievable and other times he is an angel. I did find that trying to keep him awake or at least letting him sleep for only short times during the day, late dinners or giving him a tub of yoghurt before bed, and doing things during the day making him exercise, was a huge help.
Karyn, Mackay

Our boy Ravi, aged 3 and a half, has always been a good sleeper except for a month in June last year when he was a nightmare waking at 2 a.m., 3 a.m. etc and refusing to go back to sleep. Eventually we changed his Tegretol (Carbamazepine) from 2 daily doses to 3 and his night waking stopped. Since then he has come off Tegretol. In retrospect we think the dose was too high for him.
Hannah, New Zealand

Yes, sleep seems to be a problem with TSC. Rebekah as a child would lie in her bed for ages often singing before going to sleep. She did take Melleril (Thioridazine) for a few years and was on Tegretol (Carbamazepine) and now at 39 sleep is still an issue. Recently she had an overnight sleep study done which showed "mild sleep-apnoea" but I believe they just put that label on being ignorant about TSC. Rebekah has been seizure free for almost 20 years and medication free for 10. Anxiety is definitely an issue and I believe this plays a part in her sleep issues. It used to amuse us how she would stay wide-awake. For example, if we would all come home late at night from somewhere, a typical child would zonk out, but Rebekah wouldn't. I have read one study done on sleep issues but the result seems to be inconclusive. Good luck to everybody, just another one of those issues you have to work out what suits you and your family. If Rebekah gets 4-6 hours solid sleep she thinks she's doing well!
Sue, Perth

Just Between Us: Next Issue

Just Between Us in the November Reach Out will focus on autism. We would welcome your comments and experiences, however short or long. Email reachout@atss.org.au or visit the Discussions of Australasian Tuberous Sclerosis Society Facebook page.

Online seizure diary

Introducing Epilepsy Action Australia's new seizure recording system for the web!

EPILEPSY ACTION AUSTRALIA

A huge challenge of epilepsy is its unpredictably. This is the origin of many impacts on people's wellbeing, both physical and psychological. It becomes especially difficult if seizures are not well controlled. One way to regain some control is keeping a seizure diary. Not only does a diary help people begin to recognise possible triggers or patterns and get clues to improving their situation based on what is really happening. The details can also be shared with significant others and most importantly with medical teams, to help them provide the best and most informed care and advice. But exhaustive tracking can be off-putting. Instead some people rely on their general feelings about how things are going. Others write seizure information on scraps of paper or on a calendar along with other events they need to remember. But these handwritten records are too easily lost or not brought to appointments. Plus, the only way to identify trends would be by a very lengthy process of trying to translate all the notes into some kind of graphic representation.

An exciting new virtual solution

In recognition of both the seizure management challenges facing people with epilepsy and the exciting potential of solutions found in technology, Epilepsy Action Australia has launched a new online seizure diary.

The Epilepsy Action Australia online seizure diary called 'My Epilepsy Diary' is being offered free of charge on our website www.epilepsy.org.au as part of our new integrated set of services available online for people living with epilepsy, which are an Australian first.

The seizure diary was developed by the American company Irody Inc., a leader in online medication management, and is a comprehensive clinical information organiser for people living with epilepsy. It was designed for use by patients or care givers, to gather quality seizure data for better seizure management and control.

Importantly people can choose to give carers direct access to their diary and in turn, to their seizure information, via an easy to use share function.

To ensure privacy and security, the diary runs using Hypertext Transfer Protocol Secure (or https), a connection that provides encrypted communication for better data protection.

Once people have entered profile information such as their medications, medical history and descriptions of seizures, the seizure diary is as easy use as other online tools such as Facebook.

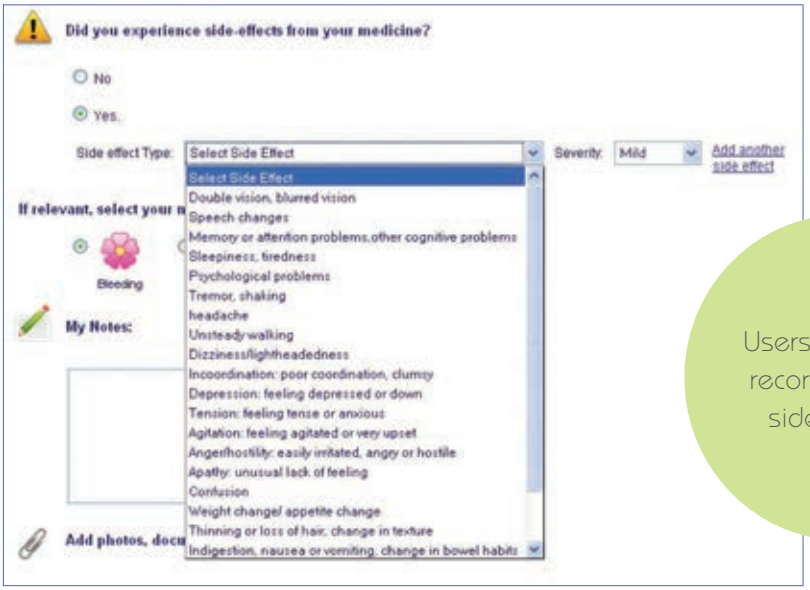
My Epilepsy Diary – Benefits

There are a range of pluses for people, their families and medical professionals. These include:

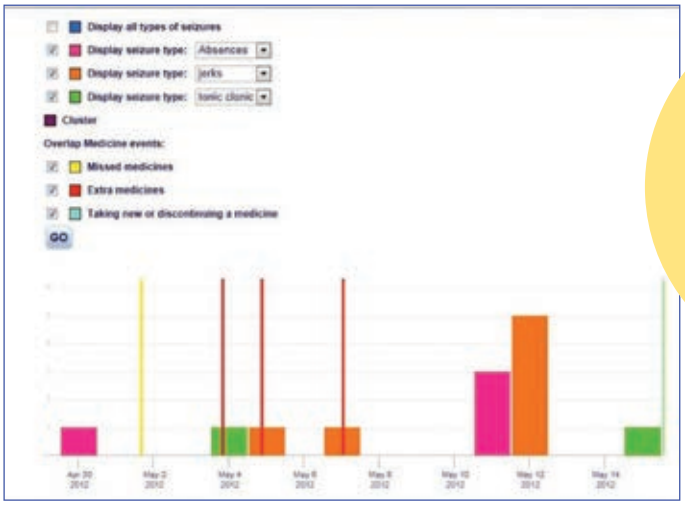
- **A simple but comprehensive system to gather data** using icons to represent seizures activity, side-effects, medications and dosages. So it is quick and straightforward to log important events and also easy for a person and their caregivers – whether loved ones, doctors or nurses – to review later and get an accurate picture of what is happening at a glance.
- **Creation of easy-to-follow reports and graphs** mapping patterns surrounding people's seizures to help them take more control. These can then be taken to medical appointments as evidence of trends over time.
- **Accessibility** through the Epilepsy Action Australia website via an easy secure log-in process. Users can update their diary on a desktop computer or mobile device (Android or Apple iPhone, iPod Touch and iPad) to record or look up details anywhere there is an internet connection.
- **A note-making facility plus the ability to upload other relevant documents and photos** to capture any extra details that may prove valuable to know.
- **Help to comply with medications** through an email reminder system that ensures doses are remembered and taken. This overcomes the difficulty with compliance often reported by people with epilepsy and has an obvious positive flow-on effect to seizure management.
- **Caregivers can log on and add data** in the event a person is unable to do it themselves.

Visit www.epilepsy.org.au today to start creating your new seizure diary! To watch a video on how the diary works, visit http://my.epilepsy.com/node/989665.

Data display is visual, with icons for variables like mood and medication doses.

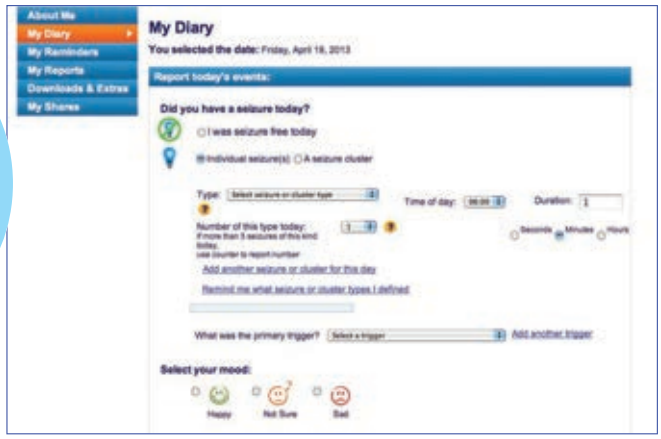


Users can easily record different side effects.



The diary makes noting down details of seizure activity a quicker simpler task.

Graph reports include this one for medicines and seizures.



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TSC Clinical Consensus Conference Leads to New Diagnosis, Surveillance and Management Recommendations

This article was written by Steven L Roberds. Phd, Chief Scientific Officer for the TS Alliance in the United States of America. It has been adapted with permission by Clare Stuart, ATSS Project Manager.

On 14-15 June 2012 more than 60 healthcare professionals, each having expertise treating one or more aspects of Tuberous Sclerosis Complex (TSC), gathered in Washington, DC, to update consensus recommendations for the diagnosis, surveillance (or ongoing monitoring), and management of TSC. We wrote about the consensus conference in the November 2012 issue of Reach Out. This article provides an update on this valuable work.

Including work before the conference, a total of 80 individuals contributed to the recommendations. Prior guidelines were based on a 1998 consensus conference, and the TSC field has made tremendous advancements in the meantime. To ensure the resulting recommendations would benefit from diverse perspectives and be internationally applicable, experts from 14 different countries participated. This included Dr David Mowat, ATSS Medical Advisor, who was selected to participate in the genetics working group. Doctors Hope Northrup and Darcy Krueger co-chaired this important effort that included specialists in:

1. Genetics
2. Brain Structure, Tubers, and Tumors
3. Epilepsy
4. Neurodevelopment and Behaviour
5. Dermatology and Dentistry
6. Nephrology
7. Pulmonology
8. Cardiology
9. Ophthalmology
10. Gastroenterology
11. Endocrinology
12. Coordination of Clinical Care

Diagnostic Criteria

The most significant change to the diagnostic criteria was to enable diagnosis of TSC based on genetics alone if an individual has a mutation in TSC1 or TSC2 that is known to cause TSC in other individuals or that produces a non- functional TSC1 or TSC2 protein.

The value of having a diagnosis based on genetics - even in the absence of sufficient clinical signs - is that the genetically diagnosed individual can have the appropriate monitoring throughout his or her lifetime. For example, even if he/she does not currently meet the clinical criteria for TSC, a newborn diagnosed by genetics can be closely monitored for development of infantile spasms, and a teen or adult diagnosed by genetics can be monitored regularly for the appearance of angiomyolipomas (AML) or lymphangioleiomyomatosis (LAM). Without a formal diagnosis, the monitoring (or surveillance) for these events - which are rare in the general population - would be difficult to justify.

That said, a genetic finding is not required for a clinical diagnosis of TSC. Because some TSC- causing mutations cannot be detected by current genetic tests, a normal genetic testing result or the finding of mutations of unknown significance does not exclude the clinical diagnosis of TSC. However, if the mutation in an affected relative is known, testing for that mutation has very high predictive value for family members. Thus, genetics can make a diagnosis alone, but a genetic finding is not required to make a diagnosis.

The updated clinical diagnostic criteria for TSC now include 11 major features and 6 minor features (see Table 1).

> *"The most significant change to the diagnostic criteria was to enable diagnosis of TSC based on genetics alone if an individual has a mutation in TSC1 or TSC2."*

Additionally, the confidence levels for diagnosis were simplified to "definite" (2 major features or 1 major feature with 2 minor features) or "possible" (either 1 major +/- 1 minor feature, or 2 minor features). The previous diagnostic criteria included a confidence level of "probable" that clinicians now feel adds little to helping physicians monitor or treat the disorder. Small changes in the names and numbers of occurrences of major and minor features were made for clarification. But overall the clinical features important for the diagnosis of TSC remain consistent with those in place for the past 15 years.

Surveillance and Management Recommendations

Because knowledge about TSC and new treatment options have grown so much since 1998, the bulk of the conference's efforts were directed toward updating the recommendations for surveillance and management of TSC.

Months before the conference itself, working groups for each focus area were formed, led by clinicians with advanced expertise in TSC and the relevant specialties. Each working group was charged with formulating key clinical questions to address within their focus area, reviewing relevant medical literature, evaluating the strength of published data, and providing a recommendation based on evaluated literature or - if data were lacking - on expert opinion based on experience or case studies. The conference provided time for face-to-face discussion of the most important or controversial points.

For each feature of TSC, the recommended treatment approach, such as surgery vs. drug therapy, differed depending on the relative

risks, benefits, and state of knowledge. Of course, every individual's specific situation will influence choices of treatment, but the consensus recommendations will help improve the quality of care to everyone affected by TSC by providing state-of- the-art guidance to physicians around the world who are less familiar with TSC. Some of the relatively major additions and changes are described here.

Since 1998, our ability to treat infantile spasms has greatly improved. For infants diagnosed with TSC, parents should be educated to recognize these even if none have occurred at time of first diagnosis. Based on clinical data, the conference participants recommend vigabatrin as first-line therapy for infantile spasms in TSC, with ACTH as a second option.

> *"Every individual's specific situation will influence choices of treatment, but the consensus recommendations will help improve the quality of care to everyone affected by TSC."*

To help raise awareness of the neuropsychiatric aspects of TSC, conference participants recommended adopting a new name for the variety of mood, cognitive, and behavioural problems that affect those with TSC. Collectively, these functional, clinical manifestations will be referred to as TSC-associated neuropsychiatric disorders (TAND). The hope is that giving a single name to this group of symptoms that can occur to varying degrees in individuals with TSC will increase awareness and understanding. A TAND screening checklist for primary clinicians is being developed by the consensus conference's neuropsychiatry working group that will take less than 10 minutes to complete for an initial evaluation. The attendees at the 2013 ATSS Family Conference in Perth were asked to provide feedback on this draft checklist by Professor Petrus de Vries, the chair of the neurodevelopment and behaviour group at the consensus conference.

One of the biggest changes in TSC treatment in the two years leading up to the conference was the approval of everolimus (Afinitor®), previously known as RAD001, for two life-threatening manifestations of TSC. In 2010, everolimus was approved in the USA to treat subependymal giant cell astrocytomas (SEGA) that are not candidates for surgical removal. Approval was given in Australia by the Therapeutic Goods Administration (TGA) and in New Zealand by Medsafe in 2012. The consensus conference recommendations for treatment of SEGA will reflect this, although the choice of surgical removal of SEGA or drug treatment depends on the relative benefits and risks in each individual situation.

In 2012, everolimus was also approved in the USA to treat growing angiomyli pomas (AML) that do not yet require surgery. The medicine is currently being considered by the TGA and Medsafe for approval for treatment of AMLs. In cases that require immediate intervention, for example due to bleeding or pain, embolism or kidney- sparing surgery may be needed. But because kidney surgery can destroy healthy kidney tissue that cannot be replaced, and often more kidney tissue is removed than planned, the conference participants strongly recommended avoiding surgery whenever possible, including partial and complete nephrectomy (kidney removal). In cases that do not require emergency intervention, the use of everolimus treatment or the embolization of large AML are clearly preferred over surgery.

Detailed recommendations for surveillance, such as the frequency of MRI or EEG, are dependent on the history of manifestations affecting each individual with TSC. As we know, the effects of TSC vary widely from person to person. Guidance on surveillance will be provided to clinicians in the medical articles being published as a result of this consensus conference.

Doctors Northrup and Krueger are submitting two overarching articles, one for diagnostic criteria and one for recommendations for surveillance and management of TSC, to an international medical journal. These articles will describe the overall consensus and highlight the importance of comprehensive and co-ordinated care in this disease that involves so many different medical specialists. The individual working groups will also produce discipline-specific manuscripts to provide more detailed guidance in journals that are frequently read by specialists in their areas. The TS Alliance will pay the necessary publication fees to make all of these articles open for access to everyone worldwide free of charge. The TS Alliance and conference organizers are also making plans to update recommendations on a regular, more frequent basis given the rapid advances that are occurring in our understanding of TSC and our ability to treat it better. The 2012 International TSC Clinical Consensus Conference was sponsored and organized by the TS Alliance. The conference was supported by generous sponsors who donated funds without playing a role in the planning or having a presence at the conference, including the Rothberg Institute for Childhood Diseases, Novartis, Sandra and Brian O'Brien, and Questcor.

Table 1:

Genetic Diagnostic Criteria	
The identification of either a TSC1 or TSC2 pathogenic mutation in DNA from normal tissue is sufficient to make a definite diagnosis of TSC. A pathogenic diagnosis is defined as a sequence variant that clearly prevents TSC1 or TSC2 protein production. Additionally, some mutations compatible with protein production (e.g. some missense changes) are well established as disease causing and also as sufficient to make a definite diagnosis of TSC. Other TSC1 and TSC2 variants may be consistent with a clinical diagnosis of TSC but are not considered to be diagnostic.	
Clinical Diagnostic Criteria	
Definite Diagnosis: 2 major features or 1 major feature with 2 minor features Possible Diagnosis: Either 1 major feature, 1 major and 1 minor, or ≥2 minor features	
Major features	**Minor features**
• Hypomelanotic macules (≥3) • Angiofibromas (≥3) or forehead plaque • Ungual fibromas (≥2) • Shagreen patch or multiple collagenomas • Multiple retinal hamartomas • Cortical dysplasias (≥3)* • Subependymal nodules (≥2)	• "Confetti" skin lesions • Dental enamel pits (≥3) • Intraoral fibromas (≥2) • Retinal achromic patch • Multiple renal cysts • Nonrenal hamartomas

National Disability Insurance Scheme

What is a National Disability Insurance Scheme (NDIS)?

An NDIS will be aimed at those who are most in need, providing long term, high quality support for around 410,000 people who have a permanent disability that significantly affects their communication, mobility, self-care or self-management.

It will focus on intensive early intervention, particularly for people where there is good evidence that it will substantially improve functioning or delay or lessen a decline in functioning.

It will also include a comprehensive information and referral service, to help people with a disability who need access to mainstream, disability and community supports.

An NDIS will give all Australians the peace of mind to know that if they have or acquire a disability that leaves them needing daily assistance with everyday life, or if they care for someone who has a disability, that they will be supported.

How will it improve the lives of people with disability, their family and carers?

An NDIS will look beyond immediate need, and will focus on what's required across a person's lifetime. At its core will be:

- A lifetime approach – funding is long-term and sustainable. People with disability and their carers will have peace of mind that the individualised care and support they receive will change as their needs change.
- Choice and control – people choose how they get support and have control over when, where and how they receive it. For some, there may be the potential to manage their own funding.
- Social and economic participation – people with disability will be supported to live a meaningful life in their community to their full potential.
- Focus on early intervention – the system will have enough resources and will be smart enough to invest in remedial and preventative early intervention instead of just providing support when a family is in crisis.

Who will be eligible?

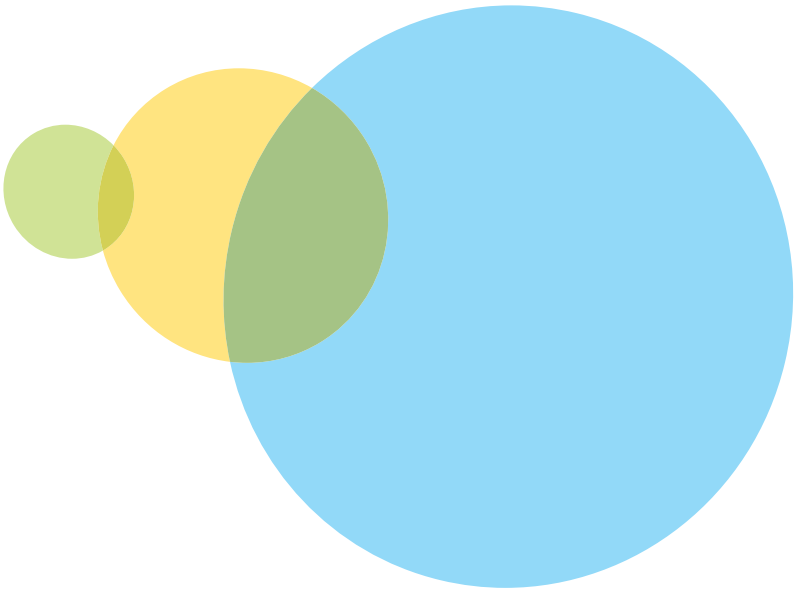
An NDIS will work with people who have a permanent disability that significantly affects their communication, mobility, self-care or self-management to ensure that they get the support that is reasonable and necessary to meet their needs. This could include an individual plan and an individually funded package.

Individual support will also be given to people for whom there is good evidence that early intervention would substantially improve functioning (for example, autism, acquired brain injury, cerebral palsy or sensory impairments), and those for whom early intervention will delay or lessen a decline in functioning (for example, multiple sclerosis and Parkinson's disease).

Others will be able to use the scheme to get information about what supports they might be able to use in the community (for example community groups like sporting clubs) and from other government programs such as health, employment support and education.

For more information go to www.ndis.gov.au

Department of Families, Housing, Community Services & Indigenous Affairs



Research News

Two notable publications on new medicines in Tuberous Sclerosis Complex have been published since our last edition of *Reach Out*. Both were published in *The Lancet*, a high impact medical journal with a large audience. One paper was from the EXIST-1 trial, looking at the use of everolimus for subependymal giant cell astrocytomas (SEGAs) in Tuberous Sclerosis Complex[1].

The second was the first major publication from the EXIST-2 trial into the use of everolimus for angiomyollipomas (AMLs) in the kidneys of people with either TSC or LAM[2].Both studies were funded by Novartis, manufacturers of everolimus.

The Treatment Trial, a randomized trial examining the topical use of rapamycin for the treatment of facial angiofibromas, has recently been approved for an 18 month extension phase. This extension will collect valuable additional data about the longer term use of this experimental treatment. Thank you again to the ATSS donors whose generosity has made this extension phase possible.

Work is progressing on **a clinical trial into a new treatment for epilepsy** in TSC is in the early stages of planning. Centres in Sydney, Brisbane, Melbourne and Perth are all being considered. More information will be provided on the ATSS website and in our e-newsletter when it becomes available.

The Association of Genetics Support of Australasia (AGSA) has received a grant to **identify unmet needs of rare disease community in the Hunter area** of New South Wales. AGSA would welcome contact from interested families in the Hunter region via www.agsa-geneticsupport.org.au or (02) 9211 1462.

Other opportunities for Australian and New Zealand families to participate in research are included in our monthly email newsletters and promoted via our Facebook pages and website. Connect with ATSS on line to be kept up to date. Thank you to the TSC affected individuals and their families that have participated in recent research projects conducted through the University of Newcastle and Epilepsy Foundation of Victoria.

1. *Efficacy and safety of everolimus for subependymal giant cell astrocytomas associated with tuberous sclerosis complex (EXIST-1): a multicentre, randomised, placebo-controlled phase 3 trial. Franz DN, Belousova E, Sparagana S, Bebin EM, Frost M, Kuperman R, Witt O, Kohrman MH, Flamini JR, Wu JY, Curatolo P, de Vries PJ, Whittemore VH, Thiele EA, Ford JP, Shah G, Cauwel H, Lebwohl D,Sahmoud T, Jozwiak S. Lancet. 2013 Jan 12;381(9861): 125-32. doi: 10.1016/S0140-6736(12)61134-9-32*

2. *Everolimus for angiomyolipoma associated with tuberous sclerosis complex or sporadic lymphangioleiomyomatosis (EXIST-2): a multicentre, randomised, double-blind, placebo-controlled trial. Bissler JJ, Kingswood JC, Radzikowska E, Zonnenberg BA, Frost M, Belousova E, Sauter M, Nonomura N, Brakemeier S, de Vries PJ, Whittemore VH, Chen D, Sahmoud T, Shah G, Lincy J, Lebwohl D, Budde K. Lancet. 2013 Jan 10. doi:pii: S0140-6736(12)61767-X. 10.1016/S0140-6736(12)61767-X*

New Fibre Optic Laser Allows Minimally Invasive Treatment of TSC Brain Complications

A new fibre optic laser technology is available for treatment of brain tumours in TSC patients. The laser generates focused heat that can be used to treat growing SEGA or SEGA-causing hydrocephalus, or it can be used for tuber ablation to treat medically resistant epilepsy. Additionally, this method can be used for some forms of epilepsy besides TSC.

Although the optical fibre of the laser apparatus must be inserted into the brain to reach the tumour, this new approach is much less invasive than surgical removal of a tumour. In the laser process, the individual undergoing treatment is put to sleep, and a metal frame is attached to his or her head. The surgeon uses magnetic resonance imaging (MRI) to guide surgical placement of the laser fibre tip inside the tumour. The surgeon then turns the laser on while visualizing the tumour site directly with MRI. The tip of laser gets hot, destroying the surrounding cells with heat. The surgeon watches the process by MRI as the treated area grows, and the laser is turned off once the entire tumour is gone.

Advantages of this process include no shaving of the head, a smaller incision (half-inch scar instead of 5-10 inches), and no need to open the skull bone (just "puncture" it instead of having to remove a "plug" of bone). The result is less pain, lower risk of infection or bleeding, and a shorter hospital stay (1 night instead of 7-10 days) and recovery (2 weeks instead of 6-12 weeks).

The fibre optic laser procedure is best for those cases in which traditional surgery is most difficult. For example, it is excellent for deep lesions because there is much less injury to overlying non-tumour tissue. Also, the laser approach is useful for tumours near areas of critical brain function because it allows patients to be awake, and because the process is monitored by MRI in real time as the area is treated.

The fibre optic procedure is clinically available at Miami Children's Hospital where four epileptologists and two neurosurgeons have treated six patients so far, the first of whom was treated over one year ago and remains seizure free. This procedure promises to significantly change brain tumour treatment options in TSC, and the techniques will only continue to improve over time.

Reprinted with permission from TS Alliance Perspective. Summer 2012.

Fundraising Thank You

Hayley Hill
ATSS Treasurer

Charity Golf Day

ATSS would like to thank Caroline Cox and the Riversdale Golf Club in Victoria who raised over $5,000 to improve support for families living with TSC. Caroline decided to support the Australasian Tuberous Sclerosis Society as her daughter Jess has Tuberous Sclerosis Complex and they wanted to support an organisation that did not receive government funding. This money was raised with three wonderful events organised by Caroline and her friends Sue Godfrey, Jan Hays and Vic Leckey.

Wednesday, 30 January 2013 was the Riversdale Golf Club's annual charity day. The club donated free golf rounds for the entire field and all money raised this year went to ATSS. The success of the day was due to many members donating prizes for a raffle which was then supported by the club members. Our thanks also go to ATSS Supporter Catherine Catlow for her heart felt talk about her family and daughter Amanda, who has Tuberous Sclerosis Complex. Her talk motivated many people attending the day to be extra generous.

In addition to the charity golf day, the ladies' committee had two film days that raised approximately $650. They also held a Girls Calendar, which included some very brave members posing. More than 100 copies were sold, raising over $1,000. The success of all three events was due to the hard working committee, the very generous members of Riversdale Golf Club, and the 80 ladies who attended the charity day from other clubs. Thank you to all.

Carmen Lewicki, Lady President Riversdale Golf Club; Caroline Cox, who presented the cheque on behalf of the Charity Committee; and Cathy Catlow on the right holding the envelope with the cheque.

Glittering Christmas Lights

ATSS would like to thank Chris and Julie Graham of Maitland area, New South Wales, who once again lit their house up with Christmas lights during Christmas 2012. Chris and Julie are good friends of ATSS Committee Member Janiffer Reynolds, whose son Nathan has Tuberous Sclerosis Complex. From the Christmas lights display $316 in donations were given. This effort has a little extra meaning as Julie has been ill recently. ATSS would like to wish Julie all the best in her recovery. Thank you again for thinking of families living with TSC.

Elliott's Amazing Triathlon

ATSS would like to congratulate Elliott Cunnew and his many sponsors who raised over $1100 when Elliott ran in the Forster Sprint Triathlon on Saturday, 6 October 2012. Elliott's daughter Amelia has Tuberous Sclerosis Complex and his wife Sally Nicholson is an ATSS Committee Member.

Elliott woke up at 4.30 a.m. to join 328 other participants at the swim start at Forster Keys. The triathlon included a 2 km swim, 120 km bike ride, and finished off with a 20 km run. Elliott mentioned to ATSS that as he ran up the final ramp to the finish line with some techno music blaring in his ears, he looked up and saw his amazing family wife Sally, daughter Amelia (his biggest fan), and son Lachie cheering him on, which helped him finish hard in 7 hours and 6 minutes. Some of what also got Elliott through was the belief from his family and friends in him completing this triathlon, which was shown by the generous donations ATSS received.

These are just a few of the ways our supporters have contributed to ATSS during the last year. ATSS would also like to thank all of the people who make donations year after year, most of whom are families affected by TSC and their friends. ATSS would not be where it is now without the constant support from you all. On behalf of all the families that we help each year I thank you.

If you would like to get involved in holding an event to raise money for ATSS, here are some ideas: host a morning tea, a mufti-day at school or workplace, or a barbecue at your local Bunnings or shopping centre. I look forward to sharing your fundraising successes on this page in the next edition of Reach Out.

Supporters and Donations

1st October 2012-14th April 2013

Gold Supporters & Donations.	Silver Supporters & Donations	Bronze Supporters & Donations	General Donations
Frederick Alexander	Sarah Brice	Peter Abery	Peter Abery
R Barassi	Ray Bryant	Activ Library	Hilary Blackburn
Mary Benedetti	J Cahalane	Lisa Anfuso	Jenny Boyce
Dawn Bowra	Linda Cameron	Kristine, Barlow-Stewart	Sarah Brice
A CakeShape	Lucy Di Falco	DJ Barron	Kerry Bromhad
Peter Clisdell	Pam George	Leslie Bishop	June & Vic Brown
Michael Cochran	G K Gray	C Boulter	Peter Clisdell
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Joanne Crosby	Val Woodyatt	Tania Colman	Richard Close
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Carly Davis	Margaret Horn	Chris Doyle	Tania Colman
Franca Di Fiori	Patricia LaPouple	R Drury	Dan & Maria Daly
Joy English	James Lowe	Debbie Duffy	Franca Di Fiori
Lloyd English	Frank Martin	Early Childhood Intervention Program	Alison & John Dick,
Epilepsy Action Australia	Sue McCormack	Pam Edgell	Lynne Downes
Shirley Faravoni	Malcolm McLean	Natalie Groom	Debbie Duffy
Lachlan Foster	David Meredith	Marie Hell	Pam Edgell
MaryAnne Hartley	Joelle Neville	Dave Woodyatt	L Edgerton
Olive Hill	Dianne Nitz	Gloria Hoffman	Joy English
David & Hayley Hill	Leanne Park	James Laird	Andrew Goldstiver
Kelven Hill	Shirley Peipman	Valda Lang	G K Gray
Daniel Strosberg & Lisa Liebman	Valleys to Plateau Community-	Barbara Lewis	A Green
Brigitte Mansour	Support Services	Dawn Lonnie	Kelven Hill
Karen McCagh	Mal Whatmore	Bernadette McGlynn	Gloria Hoffman
Sally McKillop	Kay Woodcock	Faye McLean	Lyle & Ethan Holt
Jill Mustard		Kay McMillan	Berice Hopwood
Valerie Nhan		Noah's Ark Centre of Shoalhaven Inc.	Kerrie & Mason Horsley
Sally Nicholson		Beverley O'Reilly	Lynne Johnson
Kevin Niklaus		Sharlene Oxenbridge	Jennie Kidd
Julie Osborne		Hannah Scanlon	Valda Lang
Catherine Panich		Graeme Shaughnessy	Mikey Leung
Sue Pinkerton		D Taylor	Suzanne Lewis
Georgina Schilg		Kaylene Trunk	Faye McLean
David Somerville		Geoff Tugby	Kay McMillan
Samantha Stone		Jay Yardi	David Meredith
Loren Wakeley			Sharolyn Moyle
Robyn Walker			Jill Mustard
Stephen Walker			Jeff Nelthropp
Catherine Wiles			Mark & Oyen Nelthropp
Sue Williamson			Sally & Alan Nelthropp
Alison Zehnwirth			Sally Nicholson
			Gerard O'Donnell
			Tony & Kate O'Muhoney
			Daniel Patterson
			David, Val & Daniel Patterson
			Rob Pinkerton
			Joyce Ryde
			Georgina Schilg
			Graeme Shaughnessy
			Gina Smith
			M Smith
			Nicole Stone
			Samantha Stone
			Clare Stuart
			Yvonne Sutherland
			Riversdale Golf Club Ladies Commit- tee
			Pamela Thomson
			Mal Whatmore
			Jay Yardi

Calendar of events

4 May 2013	ATSS Committee Meeting
15 May 2013	Global TSC Awareness Day
19 May 2013	TSC Awareness Picnic Day
20-23 June	International Research Conference on Tuberous Sclerosis Complex and Related Disorders, Washington DC, USA
3 August 2013	ATSS Committee Meeting
18 August 2013	ATSS Seminar Day and Annual General Meeting
2 November 2013	ATSS Committee Meeting

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