

Reach Out

May 2012 Issue 95



Riding the TSC
Wave P8



Adventures
with Hamish
P15



Launch of
Treatment Trial
P18



www.atss.org.au

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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:

1 300 733 435 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.

Visit www.tscinternational.org to find a TSC organization in your area.

President	Sue Pinkerton
Vice President	Vacant
Treasurer	Hayley Hill
Membership Secretary	Hayley Hill
Minutes Secretary	David Matheson
Committee Members	Debbie Crosby Kate Green Narelle Kerr Sally Nicholson Janiffer Reynolds
Life Members	Lynn Wilson OAM JP Andrew McKinnon JP
Public Officer	David Matheson
Medical Advisory Board	Dr David Mowat, Clinical Geneticist Dr John Lawson, Paediatric Neurologist

Editorial

David Matheson – Editor

Regular readers of *Reach Out* will notice that its appearance has changed with this edition. The new look brings a modern appearance and reflects some of the changes in the Australasian Tuberous Sclerosis Society. ATSS has grown from a small, locally-based organization to one that provides services to families all over Australia and New Zealand.

A major recent activity of ATSS was the ‘Riding the TSC Wave’ conference that occurred in October last year. This conference attracted people from many distant parts of Australia and New Zealand, and featured presentations from a number of local and international experts. Participants heard about advances in treatments for Tuberous Sclerosis Complex and details of current research. Being up to date with the latest TSC information is one way of helping to make living with TSC easier. Personal stories from families affected by TSC were also presented, and these provided a way to connect with the experience of others. Inside this edition of *Reach Out* is a report on the ‘Riding the TSC Wave’ conference. The feedback for the conference was overwhelmingly positive, as can be seen from some of the comments contained in the article. A DVD is available from ATSS that contains presentations from three of the key speakers at the conference.

ATSS has also changed in that it now has a leading role in major initiatives that improve the lives of TSC affected families. Around six years ago ATSS funded the development of a genetic testing laboratory to make genetic testing more readily available for Australian families. In recent years ATSS has raised funds for research into the development of a topical cream for the treatment of facial angiofibromas. A trial at Sydney Children’s Hospital is



now beginning. Information about the trial appears in this edition of *Reach Out*.

The content of *Reach Out* has evolved in recent years and now includes more articles regarding Australian events. Again, this is an indicator of the higher level of activity of ATSS in providing resources, helping families, and ensuring the care of TSC affected families. ATSS now has a paid employee, something that has been a dream for many years. Also, ATSS is soon hosting an information evening in Brisbane and a conference in Melbourne. These are great achievements for a small but dynamic organization.

Although ATSS has grown considerably and will continue to grow in coming years, it remains family-based. All of the members of the ATSS Committee are volunteers who have been affected by Tuberous Sclerosis Complex in some way. ATSS relies on the support and assistance it receives from families and individuals who all have their own TSC story. The activities of ATSS focus on supporting families affected by TSC.

Being up to date with the latest TSC information is one way of helping to make living with TSC easier.

I hope that this edition of *Reach Out* in some way is valuable for you in whatever way you are affected by TSC. The readers of *Reach Out* are diverse, and include families, individuals, professionals working with TSC affected families, and others who have an interest in supporting the society. *Reach Out* will aim to continue meeting the needs of all readers as it presents the ongoing work of the Australasian Tuberous Sclerosis Society.



Reach Out: Official journal of the Australasian Tuberous Sclerosis Society, Inc.

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President's Report



Sue Pinkerton - President

It has been an exciting and busy six months since the last issue of *Reach Out*. ATSS has achieved some remarkable goals, including the very successful 'Riding the TSC Wave' Family and Medical Conference last October; a new look *Reach Out*, with a new designer and printer; and completion of the second three-year Strategic Plan.

Another exciting achievement for ATSS is the employment of a part-time Project Officer, who is undertaking to grow ATSS to better meet the needs of TSC affected families and professionals. I welcome Clare Stuart to this role. Her first major project is to create a new website and resource directory with a financial grant provided by Novartis. The new website will be launched at the Conference 'Making the Puzzle Easier, Together' in Melbourne on 2 June.

For those families living in Queensland or northern New South Wales an evening 'Meet the Expert' meeting is to be held in Brisbane on Thursday, 31 May, with international and local experts in TSC sharing their knowledge and expertise.

The 31st ATSS AGM and annual Seminar Day will be held in Sydney on Sunday, 12 August with three outstanding presenters. This will be an opportunity for New South Wales families to

attend and hear Associate Professor Ernie Somerville from Prince of Wales Hospital's Comprehensive Epilepsy Unit, Dr Vicki Holets-Whittemore, and Dr Vanessa Sarkozy, a developmental paediatrician, present. This meeting will also provide an opportunity for interested members to join the ATSS Committee and contribute to the growth of this wonderful organization.

There is more information in *Reach Out* about all these initiatives. I encourage each of you to respond to some, one or all the invitations to these marvellous events in Victoria, Queensland and New South Wales.

On Monday, 12 March ATSS signed an agreement with Sydney Children's Hospital, Randwick, to fund the Treatment Trials for the study of topical rapamycin, which is about to start. Just over \$200,000 was raised by donations from generous families, relatives and friends of TSC affected families. The generosity of people wanting to improve the quality of life of people living with TSC is extraordinary and greatly appreciated. Special mention of thanks to Deb Duffy, whose initiative in forming the ATSS Fundraising Friends raised over \$1

15,000 towards the research project. Because this trial is part of an international study, it will also enhance future opportunities to participate in research.

During the early part of this year the ATSS Committee reviewed and delivered the second three-year Strategic Plan which can be viewed on the website. This will give direction and purpose to the work of the Committee in meeting the ongoing challenges of supporting TSC affected families.

TSCi (TSC International), of which ATSS is an integral member, after worldwide consultation, has declared 15 May as International TSC Day. Regardless of where you are on that day, know that you are not alone and that thousands of people around the world are thinking of you.

I congratulate David Matheson, Editor of *Reach Out*, who has gathered interesting and relevant articles for another excellent edition of *Reach Out* for your enjoyment.

I will not be standing for re-election at the Annual General Meeting in August. After the death of our daughter Lizzie in 2010 and 19 years as President, it is time I gave the opportunity to someone else to lead and serve ATSS through the coming exciting years.

I encourage you to read *Reach Out*, and if you are able, to attend one of the events being held this year for the families and friends of TSC affected people.

Treasurer's Report



Hayley Hill - Treasurer

Financially, ATSS has had a great start to 2012.

The transition from a single membership fee to three Supporter options, Gold, Silver and Bronze, has resulted in an overall increase in donations. This programme also allows our supporters to claim a tax deduction from their donation, with only \$5 being for a membership fee.

ATSS financial funds could not grow without the continuous generosity of Supporters, family and friends. Thank you to all who have supported ATSS during 2011 and 2012.

One of the CBA Term Deposits that was holding funds donated to the topical Rapamycin project was closed on 30 December 2011 and it was moved to the Cash Investment Account. On 28 February 2012 ATSS paid the first instalment for the Treatment Trial for topical Rapamycin; this was for a total of \$96,676.37. The remainder of the funds will be paid in instalments over the next 12 months as the trial progresses; this will come from the other CBA Term Deposit of \$107,736.46.

It is very exciting for Australia to be a part of this global trial and to be

the only centre outside of the United States of America. I know many of us think it could have come sooner, but all good work takes time to prepare. The last three years of donations has been worth it. Good luck to the team at Sydney Children's Hospital, and thank you to all who donated because this project wouldn't have been possible without your generosity. Building on the success of prior projects, such as the genetic testing laboratory, all supporters of ATSS should be proud of reaching this goal.

As at 29 February 2012 ATSS held accounts with Commonwealth Bank and Macquarie Bank.

CBA Cheque Account	\$62,941.08	
CBA Cash Investment Account	\$74,956.16	
Macquarie Term Deposit	\$29,966.89	
Macquarie Cash Management	\$7,539.69	
CBA Term Deposit	\$107,736.46	Matures 19 May 2012
CBA Term Deposit Elizabeth Pinkerton	\$5,000.00	Matures 19 May 2012
Total	\$288,140.28	

Membership Report

Hayley Hill - Membership Secretary

As at March 2012 the ATSS database held 564 contact details. Of these contacts there were:

121	Financial Supporters
22	Honorary members
96	Medical Professionals / Organizations (13 Financial Supporters)
27	Non-financial Supporters
298	Non-active

ATSS Supporter Renewals are due on 1 July 2012. Payments can be made before then if you would like to make a tax deductible donation before the end of financial year.

Please take the time to update your details, including your email address, on the renewal form. This helps us to inform you of upcoming events and other news in between editions of *Reach Out*. You can also renew your support online at www.atss.org.au.

Other ways to keep up to date are by regularly checking the ATSS website

and by finding us on Facebook. There are 183 members in our Facebook group that discuss various aspects of living with TSC. The ATSS Facebook page is also a way to find out about the latest developments in TSC in Australia, New Zealand and overseas.

It has been a pleasure to be the volunteer Treasurer and Membership Secretary for another year. I hope you all have a wonderful 2012.

ATSS Welcomes Its First Employee

Clare Stuart (nee Pinkerton) started as ATSS' first Project Officer in November 2011. Clare has commenced work on two projects:

Rebuild of the ATSS website, transforming it into an online directory to help TSC families share resources;

The 2012 Family Conference, being held in Melbourne on 2 June 2012.

The employment of a Project Officer has been made possible by a grant from Novartis Pharmaceuticals. The funding from Novartis will not last forever, so another part of Clare's job is to secure other grants and increase donations; this will allow ATSS to grow so that it can empower more TSC families.

ATSS has operated on 100% voluntary labour for the last 30 years. This is an amazing accomplishment that all current

and past volunteers should be proud of. The ATSS Committee has felt for some time that the task of growing ATSS would involve bringing on a paid employee. When the opportunity to hire Clare to deliver these two projects arose, the Committee knew this was the right decision for the future of ATSS.

Many of you will know Clare from her many years as a volunteer and Committee Member. Clare's first project for ATSS was making the first membership database – more than 15 years ago! More recently Clare was the main organizer of the very successful 2011 Family Conference. Clare now has a one year-old son and is working 10 hours per week from home, helping ATSS avoid the cost and commitment of office space and a permanent staff member.

Clare brings passion, experience and a

variety of professional skills to the role of Project Officer. As the big sister to Lizzie, who had TSC, Clare understands the experience of living in a family with TSC. As a management and IT consultant for the last 10 years, Clare has strong skills in building relationships, communication, strategic planning and technology. Clare has also started her Masters in Public Health at Sydney University, building her credentials and network in the health sector. Last, but not least, due to Clare's previous role as a Committee Member, the current ATSS Committee can trust that she will get on with the job with minimal training and supervision.

We wish Clare well in her role and look forward to many more milestones in the growth of ATSS.

The ATSS Committee

The Genetic Support Council of Western Australia

Sue Williamson

The Genetic Support Council of Western Australia Inc. held a lecture series on 22 February 2012 to acknowledge 'Rare Disease Day' in The Niche in Nedlands, Perth. It was an opportunity for support groups to share resources at tables in the foyer and then to hear a variety of speakers.

On the technical side Dr Gareth Baynam from Genetic Services Western Australia spoke on 'New and emerging diagnostic tests for rare disease', particularly on 3D Facial Analysis. Dr John Beilby, Head of Molecular Genetics, gave an interesting insight into diagnostic genetics and using the example of the latest diagnostic testing for Fragile X. Dr. Beilby explained the process of breaking down DNA for research focusing on the role of genes in common diseases.

I spoke on 'Tuberous Sclerosis – living with a rare condition'. As we are 38 years along our journey with TSC I found it interesting to parallel Rebekah's early years with what families experience now as there have been so many advances in

the treatments and management of TSC. However, TSC remains a constant in our lives and there are still hiccups along the way. We have just found out that Rebekah has early stages of LAM, so that will be our next hurdle.

Jemma Neal, a mother of a child living with Velo-cardio-facial Syndrome, told us her personal story. Jemma focused on the difficulty of attaining appropriate services and needs to care for her critically ill son and the daily struggle.

It was timely that the last speaker, Lesley Murphy, who is the Community Support Manager for Muscular Dystrophy (and has a son with Duchenne Muscular Dystrophy), had just returned from Sydney from the meeting to push forward the establishment of the National Rare Diseases Group. It was wonderful to hear of the progress made and Lesley's enthusiasm for what had been achieved already.

A Question and Answer session followed which highlighted the diversity of the participants, but was also encouraging

to realize that all stakeholders are appreciative of each others role in obtaining better understanding and outcomes for people living with rare diseases.



TSC remains a constant in our lives and there are still hiccups along the way.

Annual General Meeting and Seminar Day

Annual General Meeting

You are cordially invited to attend the 31st Annual General Meeting on Sunday, 12 August at 9.30 am at Sydney Children's Hospital, Randwick, to support the work of the volunteer Committee of ATSS. The coming year presents many opportunities to really grow and strengthen the Australasian Tuberous Sclerosis Society, and your attendance will encourage the Committee Members as they continue to improve the outlook for TSC affected families. The Elizabeth Pinkerton Memorial Award will be presented during the AGM to a professional who has made a significant contribution to improving the lives of TSC affected families.

If you are willing and able to make a contribution to the ATSS then please nominate for a position as Committee

Member. Roles and responsibilities vary but a commitment to making a difference is vital. Attendance at meetings held every three months can be in person or by teleconference.

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.

Seminar Day

The annual ATSS Seminar Day will be held on Sunday, 12 August immediately following the Annual General Meeting at Sydney Children's Hospital, Randwick. ATSS is privileged to have three very expe-

rienced professionals providing valuable information. Sessions will be presented by Vicki Holets-Whittemore PhD from USA on 'Worldwide Research Trends in TSC'; Associate Professor Ernie Somerville, neurologist and epilepsy specialist; and Dr Vanessa Sarkozy, a developmental paediatrician on 'Assessment and Beyond'.

All TSC affected families and friends are invited to attend the Seminar Day. Cost is \$30 per person, which will include morning tea and lunch. The Seminar Day is also an opportunity to renew acquaintances and meet others who also travel a similar TSC path.

More information is available on the ATSS website.

Please RSVP to Hayley Hill by Friday, 3 August at: hayley@atss.org.au or call 02 9920 0261.

New Policy on Working with Pharmaceutical Companies

The Australasian Tuberous Sclerosis Society Committee has recently approved a policy that governs the way we work with pharmaceutical companies. The purpose of the policy is to provide

clear principles and guidelines about how ATSS works with pharmaceutical companies to achieve our objectives, without compromising our integrity and our ability to support our members.

The policy is available on our website, or by contacting ATSS by phone or post. If you have any questions about ATSS' relationship with pharmaceutical companies, please don't hesitate to contact ATSS.

Can you help ATSS?

ATSS is primarily run by volunteers and there are many opportunities to get involved. Most work is done from home and meetings are by teleconference, so geography is no barrier. Perhaps you could contribute through one of these volunteering roles:

- Committee Members: the ATSS Committee meets quarterly and directs the activities of ATSS. Nominations are

open for the 2012/2013 Committee to be elected at the Annual General Meeting in August.

- Occasional administrative tasks such as mailouts: computer skills preferred.
- Local activities: organizing events locally to connect with TSC families in your area. Get in touch with the Regional Contact in your state.
- Fundraising: every effort, big or small,

helps. Get in touch to discuss how ATSS can support you through the provision of signs, printed material, receipting for tax purposes, or even for ideas to get you started.

- Your professional expertise: get in touch if you have a skill that we might be able to use.

If you'd like to get involved, email info@atss.org.au.

Riding the TSC wave: The 2011 ATSS Family Conference

Entertaining, Educational, Emotional, Empowering

Clare Stuart

More than 120 people, from every state and territory of Australia, as well as New Zealand, gathered in Sydney in October for the 2011 Family and Medical Conference. Highlights included the high calibre of international and local speakers, the varied and comprehensive programme and the opportunities for all attendees to share experiences.

The combined conference was given the theme of the wave. For the medical programme, this expressed in the wave of progress that has been made by understanding the mTOR protein pathway. This wave impacts TSC, but also Neurofibromatosis 1, Fragile X and some cancers. Session content included understanding of the overlaps between conditions, best practices in epilepsy control and multi-disciplinary management in TSC. Clinicians also heard the latest results and plans for clinical trials in the area and world-leading research being done on the mental health and cognition aspects of TSC.

For the family meeting, the wave metaphor was liberally applied in many ways:

- The wave of research washing up useful treatments onto the shore for TSC families;
- The ups and downs of day to day life with TSC;
- The benefits an early warning system (regular surveillance) to warn of incoming tsunamis;
- The need for a set of floaties (your support network) to help you when it feels like you might be sinking.

The Saturday family programme included a panel discussion, with neurologists focused on the decisions facing doctors, patients and parents in TSC. A review of the genetics of TSC was well received by both new families and those that could always do with some revision. The Parents as Case Managers

workshop was very interactive and focused on goal setting. This workshop ran in parallel to the first ever stream at an ATSS event dedicated to adults mildly affected by TSC.

Sunday was a combined programme that featured an all-star lineup in the TSC world. On top of hearing from Dr David Franz on Saturday morning, Sunday saw Drs Vicky Whittemore and Petrus de Vries share their expertise. Vicky, who herself has TSC, shared the journey so far – how far our understanding of TSC has come since the discovery of the TSC genes in the 1990s. As exciting as this progress has been, the conference also heard of the aspects of TSC that we still do not understand and areas where we are hopeful for more effective treatments in the future.

Dr Petrus de Vries gave an entertaining and insightful talk on the neuropsychiatry of TSC. This aspect is often under recognized and has the potential to be the

most challenging aspect of life with TSC. I thoroughly recommend that those that missed the conference take the opportunity to view the recording of this session.

The Sunday programme also featured local experts. Dr Deborah Yates, recognized as a world leader in research into lymphangioleiomyomatosis (LAM), gave a comprehensive review of management and surveillance of TSC-related pulmonary disease. Dr Sean Kennedy also reviewed the various aspects of kidney involvement

The conference also heard of the aspects of TSC that we still do not understand and areas where we are hopeful for more effective treatments in the future.



in TSC. The day was capped off with a series of small group workshops. These provided participants with a chance to ask questions on the information they had heard. The workshops also provided an update on progress in the area of a topical cream treatment for angiofibromas.

One of the favourites of the conference programme was the personal stories. Four families affected by TSC shared their stories with both the medical meeting on Saturday and the combined meeting on Sunday. Many tears and much laughter were shared as we listened to the various ways TSC affected these families.

Saturday night's conference dinner was a good chance to unwind, appreciating the views over Sydney's iconic Bondi Beach. The celebration of 30 years of ATSS was marked by the cutting of a birthday cake and much dangerous knife-wielding by Sue Pinkerton!

It took more than a year for the ATSS Committee to organise this meeting, but it was a privilege to do so. The closing remarks included copious "thank yous" to the team of people involved in making this life-changing event a reality. We would like to echo some of these messages of thanks:

- Clare Stuart, in a voluntary capacity, who was the main organizer of the family conference;
- The medical team at Sydney Children's, especially Drs David Mowat and John Lawson (ATSS Medical Advisors);
- Sue Pinkerton (ATSS President), who was a fantastic mentor and support;
- Hayley Hill (ATSS Treasurer) for her tireless administrative and logistics support;

As people with TSC or parents of TSC affected children we are constantly educating the many people in our children's lives about TSC. These people include family, friends, paediatricians, GPs, occupational and speech therapists, and teachers and aides at school. One helpful tool for families is the DVD from the 2011 ATSS Family Conference.

The DVD includes presentations from:

Dr Vicky Whittemore: *From Genes to Therapies*, describes the research on TSC completed already and future research directions that will take us closer to a cure.

Dr Sean Kennedy: *Kidney Involvement in TSC*, provides an overview of the renal manifestations of Tuberous Sclerosis Complex as well as recommended surveillance and treatment.

Dr Petrus de Vries: *Behaviour, Learning and Psychological Aspects of TSC*, dissects these various aspects in a very entertaining and insightful lecture.

For only \$20 including postage, the DVD is very informative and provides an easy to understand summary of the latest information, developments in research and treatments for TSC. It also comes with printed versions of the slides for your reference.

To order the DVD please head to the ATSS website (www.atss.org.au) or call 1300 733 435.



“ Many tears and much laughter were shared as we listened to the various ways TSC affected these families.”



“ Clinicians also heard the latest results and plans for clinical trials in the area and world-leading research being done on the mental health and cognition aspects of TSC.”

- All speakers for willingly giving up their weekend time to present at the conference;
- Our personal story speakers for sharing their TSC stories;
- Very importantly, thanks need to go to all of the families of the above for providing the space and time to for your family member to voluntarily contribute to this event.

We would also like to acknowledge our two financial sponsors: Novartis Australia and FaHCSIA's National Disability Conference Initiative provided much needed funds, particularly to allow us to cover

the majority of travel costs for interstate families. In addition, Sydney Children's Hospital provided the venue and co-hosted the event. Murrays bus company provided a generous discount on transport to and from the conference dinner.

The ATSS Committee continues to organise these events as they are always a source of information, inspiration and support. This year's conference was no exception - some of the comments from attendees are included with this article. The message that I took out of the conference was that every case of TSC is unique. Research is providing much needed insight

into the many different aspects of TSC and revealing promising treatments. But for those living with TSC, a team approach is needed to ensure that surveillance plans are made and followed and that each person with TSC has access to the best treatment plans for them.

We appreciate that not everyone was able to make the journey to Sydney. For some, it may be too early in their TSC journey to attend what can be a very emotional weekend. For these reasons, video recordings of selected conference sessions are available via our website or to order by calling ATSS.



Your Comments - things you liked about the 2011 Family Conference

- Family stories were awesome and entertaining, international experts had amazing knowledge shared and gained.
- Fantastic, travel grant made it happen.
- The quality of presenters and the timing and running of the event.
- Networking, information, amazing guest speakers.
- Presenters, opportunity to talk to doctors and other professionals.
- Love meeting families; personal stories were great. I really appreciate all the effort that goes into the conference.
- Very interesting speakers and personal stories. Friendly lovely people.
- Meeting other families. Hearing personal stories, loads of great information.
- Thanks for organising would love to come again.
- Very positive and informative. Meeting others in the same situations. Wonderful food.
- Engaging, informative, latest research. Sincere thanks to Clare and the committee for organising an inspiring and very educational conference. Also to Sue for her tireless efforts and energy in leading ATSS into what is an exciting phase of treatment and management of TSC.
- Brilliant combining medicos and families. Calibre of presentations was outstanding.
- Great casual atmosphere, wonderful speakers and well planned.
- Enjoyed the whole weekend.
- Being able to catch up and feel normal.
- Everything! Especially the audience involvement.
- Felt welcome, Conference info, dinner was lovely. Having doctors and research on hand.
- The conference was a success for me even after 10 years in the "system" I always learn and meet someone new that will help me in my journey.
- Extremely informative, fantastic lectures. My wife and I would fly anywhere to these events if they are run like this one.

Melbourne

TUBEROUS SCLEROSIS
COMPLEX

MAKING THE PUZZLE
EASIER, TOGETHER

A conference for
FAMILIES EDUCATORS
AND HEALTH PROFESSIONALS

Saturday, 2nd June, 2012
The Royal Children's Hospital, Melbourne
www.atss.org.au

Australasian
Tuberous Sclerosis
Society

Brisbane

Brisbane Meet the Expert Evening



In May, leading neurologists will be in Brisbane for the International Child Neurology Congress. Don't miss this opportunity to:

Hear about the latest TSC
treatments

Find out about clinical trials
for new medicines

Meet other families



Key note speaker:

Sergiusz Jóźwiak, MD, PhD
Children's Memorial Health Institute, Poland
World leading researcher into new therapies for TSC

From 6pm, Thursday 31st May 2012

Full programme and registration :

www.atss.org.au

1300 733 435

Brisbane top of mind symposium

ATSS partnered with Epilepsy Action to host a TSC stream within the Top of Mind symposium held in Brisbane in November 2011. A group of wonderful local doctors spoke to a group of TSC families.

- Associate Professor Michael Gabbett explained the genetics of TSC and how to access genetic services in Queensland.
- Dr Stephen Malone promoted the best practice surveillance and monitoring guidelines.
- Dr Kate Riney presented the various treatment options for TSC epilepsy.

One of the highlights from the rest of the day was Associate Professor Ernie Somerville's insights into what neurologists do when their patients are still having seizures. The topic is so relevant to TSC families that ATSS has invited Associate Professor Somerville to speak at the Seminar day to be held in Sydney on 12 August.

Perth Picnic

ATSS members and friends in the Perth region are invited to gather for a picnic in Perth during May.

Date: 13 May 2012

Time: 11.00 am

Venue: King's Park, in the vicinity of Sticky Beaks Cafe

Look for the blue and white balloons for Western Australia to acknowledge Global TSC Day. For further details please contact Sue Williamson: suemika1@aapt.net.au.

29th February A Rare Day for Rare Disease Day

All over the world, rare disease patient organisations, like ATSS, and their partners will joined forces to promote awareness about rare diseases and the millions of people affected by them. This year Tuberous Sclerosis Complex and ATSS were involved in a number of events.

Rare Disease Day at Novartis

Clare Stuart and Hayley Hill spoke to staff members at Novartis Australia and raised awareness of Tuberous Sclerosis Complex and other rare diseases. More than 120 people heard a talk entitled 'A tale of two strong girls: The difference 20 years makes' as Clare told the stories of both Lizzie Pinkerton (Clare's sister) and Alana Crosby (daughter of Deb Crosby from the ATSS Committee). Clare explained the difference that emerging treatments make to the hope that TSC families have for the future. The event was a great opportunity to raise awareness of TSC and collect a few donations. Thank you to the Novartis team for getting involved in Rare Disease Day 2012.

Celebratory Lunch in Manly

Sue Pinkerton and Clare Stuart represented ATSS at Rare Disease Day celebrations for those working in the area. The event was attended by many other patient organisations and it was a

wonderful opportunity to connect with others, highlight the many challenges faced by people living with rare disease, and get an update on the work happening in the area.

APSU Rare Disease Workshop

Clare Stuart represented ATSS at a workshop hosted by the Australian Paediatric Surveillance Unit (APSU). The day included researchers, clinicians and patient organisation representatives discussing successes and challenges in the rare disease area.

Rare Voices Australia working party

Clare Stuart was a part of a working group that has led to the formation of a new organization called Rare Voices Australia. Rare Voices aims to be a united voice for Australians affected by rare diseases in the journey towards a National Plan that will lead to improved health and other services.

Clare explained the difference that emerging treatments make to the hope that TSC families have for the future.

Adventures with Hamish:

A Tuberous Sclerosis Story

Kate Green

Hamish was born at 2.15 pm on a freezing cold August afternoon in 2005. Tiny, but beautiful and perfectly formed. An hour after birth his little heart went into Supraventricular Tachycardia (SVT), which subsequently resolved itself. The paediatrician was paged. Four hours later he was in a New-born Emergency Transport Service ambulance being transferred to the cardiac ward of the Royal Children's Hospital, Melbourne; not before a large BMW which was blocking his ambulance was towed away from the maternity unit car park, much to the delight of the nurses. So began our adventures with Hamish and Tuberous Sclerosis Complex.

That SVT turned out to be a blessing in disguise, as it meant we had a diagnosis of TSC when Hamish was just forty-eight hours old. He had rhabdomyomas on his heart, astrocytomas behind his right eye and a head full of tubers. Masses of them, according to the results of his first MRI. We were taken to the quiet room and spoken to gently by the cardiologist. He talked about seizures, developmental delays and possible heart issues. I heard it all, but could only see an adorable little baby boy snuggled into a tiny ball on my chest. Happily, he is still snuggly.

Hamish was liberated from the Royal Children's Hospital at three days old and set about endearing himself to his big brother Alex, his grandparents, and anyone else who crossed his path. Especially to Alex, who at seven years old had been living in fear of having a baby sister and being surrounded by Barbie dolls! He also got to work his charm on the Mobile Intensive Care Ambulance paramedics who were called when he went into SVT again at ten days old. Fortunately they captured a very useful ECG and Hamish was promptly put on beta blockers. He hasn't had issues with his heart since then and the rhabdomyomas eventually disappeared entirely as his heart grew.

Next on Hamish's Tuberous Sclerosis Complex to do list was epilepsy. He met

his first neurologist at the Royal Children's Hospital when he was three months old. At this point he was diagnosed as having infantile spasms. Fortunately, his paediatrician had described, accompanied by hand and arm actions, what epilepsy with Hamish may look like. Ever helpful, Hamish's seizures started in just that way, so we were on to it pretty quickly. Another drug, Clonazepam, was added to the beta blockers. Unfortunately, his seizures only became worse so Sabril was also added to his daily cocktail. Despite the onset of infantile spasms, Hamish started smiling at six weeks of age and laughing at about twelve weeks. He was interested in the world around him, fed well, slept well and continued to charm everyone around him. A model baby, apart from the whole TSC thing.



Hamish in Royal Children's Hospital

Over the next twelve months Hamish continued to thrive, despite marked developmental delays and increasingly worse seizures. Most importantly, he was happy and healthy. By the time he was twelve months old he was spending four hours a week with the wonderful people at the Malvern Special Needs Playgroup and we were attending a weekly playgroup at Noah's Ark. We met some lovely people and their children through these organizations, which became an essential support and outlet for us. Hamish started crawling at seventeen months and astoundingly, started walking at twenty-one months. Although it wasn't so much walking as a drunken-like stagger. Truly frightening to

watch sometimes. It was during this time that Hamish had a seizure that lasted for over an hour. A trip to the hospital in an ambulance followed and Hamish was discharged with another anti-epileptic drug.

When Hamish was about eighteen months old his original neurologist retired and we were introduced to his infectiously enthusiastic epileptologist, Simon. This was where the fun really started. After several rounds of EEG video monitoring, SPECT scans and MRIs, Hamish was considered a candidate for neurosurgery to hopefully remove the tubers in his brain which were causing his seizures. In July 2007, just weeks before his second birthday, Hamish had his first round of neurosurgery. Two large tubers were removed during ten hours of surgery. It seemed like the



Hamish with his mother Kate

longest of days, but if I had known what was to come later, I think I would have run screaming from the recovery room! Hamish bounced back from his craniotomy in excellent spirits and somehow still regarded "his" neuroscience ward as a place of adventure and interesting people.

Sadly his seizures continued to get worse, so another round of surgery was scheduled. This was to be the incredibly high-tech procedure where probes were placed directly onto his brain during neurosurgery and left there for a week while his seizures and EEG activity were monitored. So, three months after his first round of neurosurgery, Hamish was back for round two. During eleven hours



Hamish

“Hamish was kicked out of ICU the next day and spent the next six days connected to his new best friend, the EEG monitor.”

of surgery, eighty probes were placed on Hamish’s brain, his skull stapled back into place (leaving a little hole for all the wires and a drain), and the probes hooked up to a portable EEG monitor. Hamish came through the surgery with flying colours. Unfortunately, at about 11.30 that night Hamish’s oxygen levels started to drop and his breathing became laboured. The medical emergency team arrived and Hamish was whisked off to Intensive Care with a collapsed lung, which was an unexpected by not unusual consequence of such a long anaesthetic. I was doing the sleepover that night and will never forget how quickly his little room filled with staff, once the red emergency button had been pressed. I knew Hamish was in the best possible hands, but ringing my husband

and mother in tears at midnight to say he had been rushed to ICU wasn’t something I ever want to repeat.

Not to mention the various central lines and IV pole. He still managed a couple of little walks to keep an eye on his ward. As luck would have it, on the day the probes were due to come out and more tubers removed, Hamish developed a nasty case of gastro, which he also managed to pass onto his poor neurosurgeon. Surgery was dramatically shortened and just the probes were removed. Hamish and his neurosurgeon both looked a little pale and queasy afterwards, and his anaesthetist was very stoic about having been put on nappy changing duty during the procedure. After a much needed blood transfusion, which put the pink back in his cheeks, and a bottomless supply of chocolate frogs (just don’t tell the paediatrician), Hamish was successfully discharged again. Still smiling, still bouncing, despite the trauma of having Collingwood football players sing Christmas carols to him while he was in hospital.

Following this, Hamish had two more rounds of neurosurgery to resect tubers. Everything went smoothly and six months after his last surgery, Hamish’s seizures

“Hamish at six is an amazing, happy, gentle and charming little boy with the face of an angel.”

went away. His walking was still alarmingly off-kilter, but once the seizures stopped he found his balance and even started running in an attempt to chase his cat. By this stage Hamish was attending preschool at the Port Philip Specialist School’s Early Education Program two mornings a week, where he is now in grade one at school.

Hamish at six is an amazing, happy, gentle and charming little boy with the face of an angel. Like all six year old boys, he can be evil on the inside! It’s been hard on my older son Alex. He has worried about all of Hamish’s procedures, cried when a child at primary school made a cruel comment about his brother, and celebrated Hamish’s first swear word.

He’s also seen some pretty confronting things while Hamish has been in hospital. I believe Alex will be a more understanding person because of Hamish. He is very protective of his little brother, yet at fourteen, old enough to have his own interests and agenda apart from Hamish. Hamish adores Alex, although I think Hamish’s adoration sometimes has more to do with Alex’s array of computers, iPad and other interesting gadgets with bright lights and tempting buttons.

Tuberous Sclerosis can be a cruel, unpredictable condition and we don’t know what the future will hold for Hamish. He was diagnosed with mild focal seizures a year ago, but his latest drug, Topamax, has almost completely dampened them; it has also had the happy side effect of making him extremely chatty and vocal. He is starting to mimic words, especially a particularly rude one, and walks around giving his opinion on everything, in his own special language, whilst pointing, gesticulating and throwing in the odd expletive. He’s endlessly funny and entertaining and shamelessly plays to an audience, constantly looking at them to make sure they are listening. Best of all (for me) he’s an extremely affectionate mummy’s boy. He still climbs onto my lap and moulds

himself to me like a baby, which is bliss. Hamish's TSC issues will be monitored as closely as modern medicine allows and problems dealt with when and if they arise. He will be loved all the more for it.

Sometimes I wonder what Hamish would have been like without Tuberous Sclerosis Complex, but without TSC he wouldn't be Hamish. We wouldn't have met the amazing friends, doctors, nurses, teachers and others who constitute our TSC world. We wouldn't have learnt to appreciate Hamish's many achievements with such joy or been nearly as tolerant of his attempts to wreak havoc. I personally have discovered depths of patience, understand-

ing and empathy I didn't know I had. Not to mention love. Pure, unadulterated and fiercely protective love for this little creature I've been blessed with.

Finally, without Hamish and Tuberous Sclerosis Complex, I wouldn't have found myself last week gleefully describing, in lurid detail, his every swear word to his neurologist. Hamish was standing on a chair, talking at the top of his voice and our long suffering neurologist was in fits of laughter, both at Hamish and his rather filthy new words. I came away thinking that if a neurological appointment can end up being so funny, then maybe things won't be so bad.



Foot painting

Launch of Treatment Trial

A cream to treat facial angiofibromas



Three years of fundraising has led to Sydney Children's Hospital being funded by ATSS to take part in the first randomized controlled trial for the topical use of Rapamycin for angiofibromas, the skin condition that affects up to 90% of people with Tuberous Sclerosis Complex. Thank you to the hundreds of people that organized fundraising activities and donated. Over \$200,000 was raised; however, the average donation was under \$100.

This is an important study for several reasons. Our goals are to have topical Rapamycin to be approved by the Therapeutic Goods Administration (TGA) and subsequently included in the pharmaceutical benefits scheme (PBS); this is the way that we will ensure this treatment is made accessible and affordable for all TSC affected people that require the cream. The Treatment Trial will provide the clinical trial evidence required for this approval process. It will also help to confirm the strength of the cream and the way it should be used and give doctors and families more evidence of its safety.

The principal investigator for the trial at Sydney Children's Hospital, Dr Orli Worgan, will provide an update on the trial progress in the November edition of *Reach Out*.

During the trial and the lengthy approvals process that might follow, there is still a way for TSC families to access the treatment. You need to discuss this treatment option with a dermatologist who will assess each individual case. This is called using the medicine "off-label" and may also be subject to approval processes. The

team at the Princess Margaret Hospital for Children in Perth have published their experience with the cream and their recipe. You may like to refer your dermatologist to the following scientific journal article:

Foster, R. S., Bint, L. J. and Halbert, A. R. (2012), Topical 0.1% rapamycin for angiofibromas in paediatric patients with Tuberous Sclerosis: A pilot study of four patients. *Australasian Journal of Dermatology*, 53: 52–56

This article suggests an annual cost of approximately \$3000. Because there is no centralized manufacturer and the medicine is not yet on the Pharmaceutical Benefits Scheme, funding arrangements will differ between health services, so you should also discuss this with your doctor. Please contact ATSS if you need any more information.

The Treatment Trial will provide the clinical trial evidence required for this approval process.

First mTOR Inhibitor Approved by TGA for TSC

Everolimus, an mTOR inhibitor medicine, has been approved by the Therapeutic Goods Administration in Australia for subependymal giant cell astrocytomas in Tuberous Sclerosis. The drug is marketed under the brand name 'Afinitor'. This is the first drug to be approved for a Tuberous Sclerosis Complex indication in Australia, in this case specifically for

subependymal giant cell astrocytomas or SEGAs. SEGAs, a benign brain tumour, affect 15-20 % of people with TSC and typically become symptomatic in children and adolescents. If left untreated, SEGAs can lead to a build up of fluid in the brain and even death. Afinitor has been approved for use in patients that are not candidates for surgical resection.

The approval of Everolimus for treatment of SEGAs has been made possible by the TSC families, in Australia and worldwide, who have taken part in clinical trials for this medicine. This is a milestone in the journey towards better treatments and, one day, a cure for TSC.

Are you taking Everolimus for SEGAs? Help us get Everolimus on the PBS

Everolimus will be considered for inclusion on the Pharmaceutical Benefits Scheme over the next few months. Inclusion will minimise the cost of the medicine for families throughout Australia. There is an opportunity for you to make a statement to the advisory committee for them to consider while making this decision.

You can choose to contribute your own statement or be a part of a combined statement from ATSS.

Please email clare@atss.org.au for more information.

At this stage, Everolimus is only being considered for inclusion for the treatment of SEGAs (subependymal giant cell astrocytomas). We expect that future submissions might include other signs and symptoms of TSC. For the current advocacy activity we are particularly looking for families taking Everolimus for SEGAs.

Fundraising Thank You

Hayley Hill

Wedding Gift Donation

The marriage of Craig and Bard Martin was celebrated on 29 October 2011; their wedding gifts included donations to charities. Bruce and Katina Core, long term ATSS supporters, wrote a cheque to ATSS on behalf of Craig & Bard Martin's wedding gift. Thank you Bruce and Katina and to the newlyweds - all the best for your future together.

Christmas Donations from the Heeley Family

Every year for the last 15 years, Donald and Margaret Heeley and family have decorated their house in Mallee Street, Quakers Hill, Sydney. Santa even makes an appearance and for Christmas 2011 he handed out more than 6,000 lollipops!

Not only do they bring Christmas cheer to everyone who visits to see the house, but they also fundraise for charity. For 2011 the Heeley Family chose the Australasian Tuberous Sclerosis Society (ATSS) for their donations. An amazing \$2,768.00 was donated by the people that visited the house to see the lights this year. ATSS would like to sincerely thank the Heeley Family for choosing ATSS and thank everyone who donated.

Christmas Donations from Chris & Julie Graham

Chris and Julie Graham of Maitland area, New South Wales, again adorned their house with a Christmas lights display during

Christmas 2011. Chris and Julie received \$272.35 in donations for ATSS from the many who visited their lights display this year. Thank you to Chris and Julie for thinking of ATSS; once again, it is greatly appreciated.

Christmas Appeal Donations

In the busy weeks leading up to Christmas in 2011, ATSS sent out a Christmas Appeal to some of you. Thank you to all who donated; a total of \$1,155.00 was raised.

Birthday Celebrations

Ian Webb of Queensland, who has a grand-daughter Chelsea with Tuberous Sclerosis, recently celebrated his 60th birthday. As part of his birthday celebrations Ian requested "Donations to ATSS in the quest to find a cure for Tuberous Sclerosis". The total of donations received was an amazing \$3,600.00 from his friends and family. Thank you to Ian, his friends and family for your generous donations. ATSS hopes that you had a wonderful 60th birthday.

I would also like to say thank you to everyone who has donated to ATSS since the November issue of *Reach Out*. A list of all donors is included in this issue. The donations help to keep ATSS working to improve the lives of all TSC families in Australia and New Zealand.

Supporters & Donations For ATSS September 2011 - March 2012

Gold Supporters & Donations. Total Received \$2085.00	Silver Supporters & Donations. Total Received \$505.00	Bronze Supporters & Donations. Total Received \$545.00	General Donations. Total Received \$5,655.35	Topical Rapamycin Donations. Total Received \$3,288.00	Christmas Appeal Donations. Total Received \$1,155.00
Peter Abery	Linda Cameron	Shirley & Michael Aisbett	Peter Abery	Shirley & Michael Aisbett	Lynne Abbott
K & J Bromhead	Tania Colman	D & C Barron	D & C Barron	K & J Bromhead	H Blackburn
Gavin & Suzie Clark	Franca Di Fiori	D & E Bartels	K & J Bromhead	MaryAnne Hartley	Lorraine Booth
Peter & Helen Clisdell	Steve & Natalie Hale	Catherine Catlow	Peter & Helen Clisdell	Lynda Heeley	Megan Bowley
Peta Cope	Ron Heard	Sarah-Jane Deckert	Bruce & Katina Core	Susan & Andrew King	Sandra Capper
Debbie & Anthony Crosby	Malcolm McLean	Sally Dewhurst	Franca Di Fiori	Jill Kowalewski	Warren Cheney
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Lisa & Lachlan Foster	Val Woodyatt	R & J Drury	Irina Giler	Malcolm McLean	Eileen Cooke
MaryAnne Hartley	Dave & Paula Woodyatt	Michael & Tamara Goong	Emma Goodison	Doris Murray	G, S & P Faravoni
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Rose Travers		Edna Kennedy	Mark James		Olive Hill
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Helen & Tam Willacy		Barbara Lewis	Paul LaPouple		Y F James
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		Clare Stuart	Graeme Smith		Guy Romeo
		Josephine Taffs	Rose Travers		Judy & Tony Taylor
		D Taylor	Trish & John Weaver		Sharron & Simon Tolsher
			Ian Webb		
			Chris & Mal Whatmore		
			Kay Woodcock		

Calendar of events

5 May 2012	ATSS Committee Meeting, Sydney
13 May 2012	Family Picnic, Perth
15 May 2012	Worldwide TSC Awareness Day
31 May 2012	'Meet the Expert' Evening for Families, Brisbane
2 June 2012	TSC Conference, Melbourne
28 July 2012	ATSS Committee Meeting, Sydney
12 August 2012	ATSS AGM and Seminar Day, Sydney
15 September 2012	Closing date for November Reach Out
6-9 September 2012	International TSC Research Conference and TSC International Meeting, Naples, Italy
3 November 2012	ATSS Committee Meeting, Sydney

Sponsorship opportunities

Would your business, employer or family like to support ATSS by sponsoring an edition of *Reach Out*?

ATSS relies on generous donations to cover costs of publishing *Reach Out* twice a year.

To discuss sponsorship opportunities, email clare@atss.org.au.