

News from TSCNZ - Newsletter 14, November 2020



Tuberous Sclerosis Complex New Zealand

Inform | Connect | Support

In this issue: [Progress on Funding of Sirolimus](#), [Research News](#), [Global Awareness Day](#), [Fair for Rare Campaign](#), [New Brochure](#), [Mail Chimp Newsletter](#)

Progress on Funding of Sirolimus

Welcome to this TSCNZ newsletter. Almost time to pop the champagne corks! We are pleased to announce that PHARMAC has released a proposal to fund sirolimus for renal angiomyioma and intractable epilepsy in TSC from 1 February 2021.

Thanks to all the families who provided their personal stories to support our application for widening the funding of mTOR inhibitors and the clinicians who also made applications. This is a significant step forward for the NZ TSC community.

Although we are delighted at the progress, TSCNZ would like even broader access to m-TOR inhibitors and will be making a submission asking for sirolimus to be funded for LAM and facial angiofibromas. We would also like everolimus to be a funded option in the event that sirolimus is not tolerated. We have made a submission to this effect. Email info@tsc.org.nz if you would like a full copy of TSCNZ's submission.

Research News

Topical Rapamycin Trial now open to participants New Zealand wide

We are delighted to let you know that this study of a new cream for the treatment of facial angiofibroma is now open to participants from anywhere in New Zealand. The first two people in New Zealand have completed 6 months of daily use of the cream and three other people in the South Island are currently using the cream. There are 30 people around the world who have used or are currently using the cream as part of the study.

Christchurch Hospital is the only place in NZ where the study is being carried out, therefore costs of travel to Christchurch will be provided as part of being involved. This study is the first opportunity for people with TSC to take part in a clinical trial in NZ and give people with TSC opportunities to access treatments that might otherwise not be available.

The study involves 8 clinic visits over about 8 months including screening and follow up for the study, daily application of the cream and filling in a diary. Out of pocket expenses including travel from anywhere in NZ to Christchurch for clinic visits as part of the study are reimbursed. Anyone aged 6-65 with facial angiofibroma might be suitable for this study, so if you or a family member are interested please contact malina.storer@cdhb.health.nz (or 03 364-1157) or caroline.mahon@cdhb.health.nz for more information.

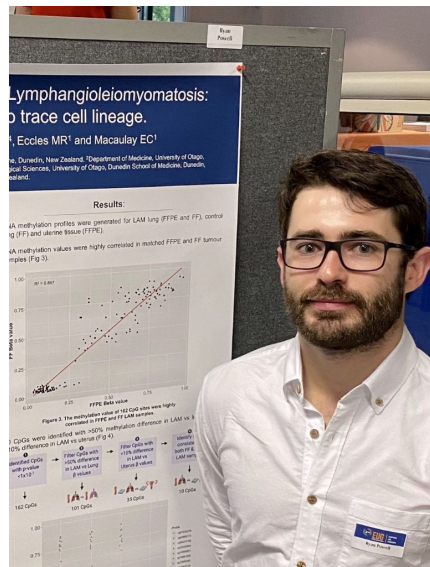
Special thanks to principal investigator Dr Caroline Mahon for her informative talk about the trial and skin manifestations of TSC at our AGM held in July. An audio recording of her talk and her slide presentation is now available to view on our youtube channel https://youtu.be/T5A74h9Qc_U

Introducing Ryan Powell

In other research news, the TSCNZ committee were pleased to be able to award a grant of \$2000 to Ryan Powell to support his PhD research and interest in TSC.

Ryan Powell is currently undertaking a Doctor of Philosophy (PhD) degree at the University of Otago, Dunedin. His primary research project entails a case study on a single patient with Tuberous Sclerosis Complex. In this project, the Dunedin research team is

attempting to understand the genetic relatedness between the TSC-associated manifestations of this patient. In doing so, they hope to unravel the genetic changes required to progress from a benign growth to malignancy. Ryan is also investigating the cell of origin for sporadic-Lymphangioliomyomatosis, a common manifestation also observed in Tuberous Sclerosis Complex. For the final portion of his PhD, Ryan aims to investigate cellular vulnerabilities resulting from TSC mutations as targeted treatment options. Ultimately, Ryan hopes to pursue a varied and exciting career as a research academic. Ryan seeks to one day establish his own research group, investigating how the TSC1 and 2 genes contribute to the 'rewiring' of various genetic and metabolic processes within cells.



Global Awareness Day



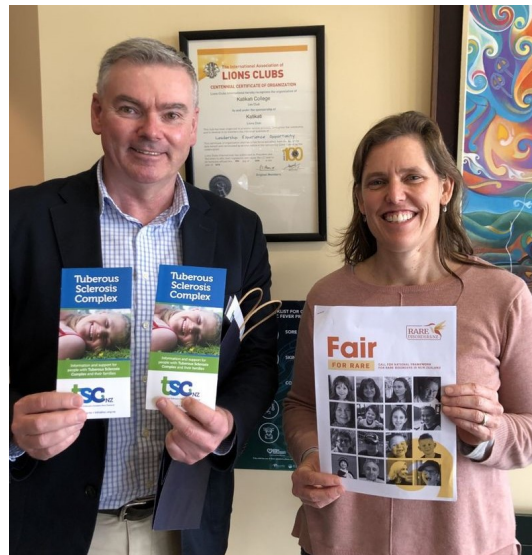
Thanks to all those who took part in our Global Awareness Day activities. We saw a real boost in donations which was very

heartening.

It was wonderful to have the opportunity to talk to Dan Klein from the TS Alliance about Global Awareness Day and New Zealand's response to Covid-19 for the [Global Awareness Day podcast](#) . The collaboration between the global TSC community continues despite closed borders.

Thanks all who have recently become financial members and/or made generous donations. A big thank you to Constellation Brands who matched their employee Tom Maling's donation. What a great idea! Now is the time of year we ask people to renew their \$30 financial memberships if they haven't joined recently. Though we are happy to inform our community even if they are not financial members, we greatly appreciate your regular support. It is now super easy to join TSCNZ through our website www.tsc.org.nz/support-us and click on the **Join Today** button.

Fair for Rare Campaign



MP Meetings

TSCNZ President Helen Willacy met with Labour MP Jan Tinetti and National MP Scott Simpson to discuss the New Zealand National Rare Disorder Framework being promoted in the Rare Disorders NZ Fair For Rare Campaign. There was also some time to tell them about TSC and our campaigns to improve access to medicines and co-ordinated care. They were both good listeners and very interested. It was also good to hear that Rare Disorders NZ has a positive reputation in

Wellington. It was very encouraging how easy it was to set up the meetings and speak directly to those representing us.

Thanks to Leah Bos, TSCNZ committee member for creating a video and sharing it as part of the Fair for Rare campaign. The Fair For Rare campaign will resume after the election. Sue Haldane's petition is now open until March 2021 <https://raredisorders.org.nz/fair-for-rare-nz/parliamentary-petition/>. Details of the campaign can be found at <https://raredisorders.org.nz/fair-for-rare-nz/fair-for-rare/>

Communication Updates

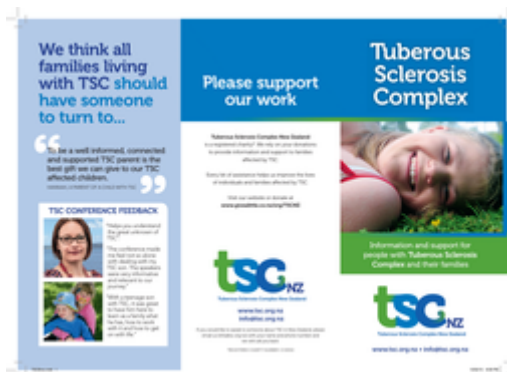
New Brochure Photos Request

One of our projects for 2020 is to update our 3-fold brochure. We would love some recent photos that reflect the diversity of ages and cultures in the TSC community.

If you would like to contribute, please email a high resolution photo of you or your family member with TSC to info@tsc.org.nz

We are delighted to announce that we have received to recent generous donations which we will use to adapt TS Australia's recently published Parachute Pack resources for newly diagnosed individuals and families and information for health professionals to the New Zealand context. The excellent Australian versions are available on their website in the meantime. <https://tsa.org.au/parachute-packs-now-available/>, <https://tsa.org.au/health-professional/>

Please contact TSCNZ at info@tsc.org.nz if you ever require brochures or copies of the Surveillance and Management guidelines.



Conference News

Covid permitting, here are some tentative dates for your diaries.

July 2022: We are hoping to lead a contingent of New Zealanders to attend the next Global Conference in in Dallas.

October 2022: New Zealand TSC Conference

Mail Chimp Newsletter

We hope you are enjoying the new newsletter format. It certainly makes it easier to get out information in a timely fashion. If would also like to receive our communications by email. Please check the settings in your email to check it will not send these emails to your spam folder - you may need to click an option like 'Trust this sender' or it may help to use the 'Add us to your address book' link at the bottom of this email.

You can also help us by updating your contact details if they change - use this link at the bottom of all emails: [update your preferences](#)



[TSCNZ on Facebook](#)



[Website](#)



[YouTube Channel](#)



info@tsc.org.nz

**If you need information or support on any aspect of living with TSC please
email us or call 021 154 7017**

Copyright © 2020 TSCNZ, All rights reserved.