



TS 2000 Study

Annual Newsletter 2015

Hello everyone!

It's been a year since we were last in touch, so we want to update you about our progress in Phase 2 of the study as well as tell you about our exciting plans for future research. Thank you for your ongoing participation.

What we have been doing in Phase 2

Phase 2 of the study has now come to an end. Phase 2 aimed to follow-up all the children in TS 2000 to see how they have been progressing since they were originally assessed. We were keen to find out about the development of each and every child in the TS 2000 study, because we wanted to get as accurate a picture as possible about the 'natural history' of Tuberous Sclerosis, and in particular whether children were at increased risk of neurodevelopmental disorders such as Attention Deficit Hyperactivity Disorder (ADHD) and autism.

Between 2013 and 2015 we carried out in-depth assessments of behaviour and cognition. We asked parents to complete an interview about their child's behaviour, communication and play, and complete questionnaires about aspects of communication, social interaction, 'adaptive' abilities in day-to-day life, activity levels and attention. From 2014 we have also investigated obstetric risks, early infant environment and sleep problems.

We also asked children or young people with TS to take part in a range of assessments, tasks and games. These were designed to assess intellectual ability, social understanding, language, planning, memory, attention and thinking style. Over summer 2014 we saw a sample of families to assess siblings who are *unaffected* by TS. They will serve as a comparison group of children closely matched to children with TS in terms of family and social factors.

As well as questionnaire and interview data we have been working on collecting genetic and clinical data. We left/ sent out to some families home saliva kits and/ or cheek swab kits. We have also contacted doctors to obtain recent clinic reports, scan reports and digital images of brain and kidney scans.

Our progress in phase 2

We saw around 90 of the original 125 TS 2000 families, assessed 30 siblings and had contact with over 90 doctors, accessing around 70 digital brain scans. We are very grateful for your time and all of your hard work! Loss to follow-up is inevitable in a long-term follow-up study but every family that we see makes a really valuable contribution to the quality of the research that we are able to produce. In rare disease research we rely on small sample sizes, meaning that every family lost to follow-up and piece of missing data really counts. If you have not been seen by our team, but are now happy to participate, please get in touch! We will be starting a new phase of data collection in the next months. If you have any outstanding questionnaires or genetic samples to send us, please complete them and send them back to us.

Recently we have had a last push to collect outstanding data on the families that we have seen. This includes paper questionnaires, telephone interviews, computer based questionnaires and genetic samples. **If you have a spare moment over the Christmas holidays to complete these and send them back to us, this would be a great Christmas present to us!**

We also presented some of the findings from Phase 2 at the 2015 International Research Conference on Tuberous Sclerosis Complex in Windsor. Colleagues from the international TS community, as well as families affected by TS, were excited to hear about more results from the TS 2000 Study. We hope to publish these results in scientific journals soon and will keep you up to date with results as they are published.

Phase 3 is beginning!

We have recently received funding from Action Medical Research to gather more in-depth information on brain processes using EEG and how they relate to intellectual and developmental outcome in TS. EEG is a non-invasive technique using a special form of hairnet that contains lots of small sensors. The hairnet can measure the naturally occurring 'brain waves' of people from all ages from infancy to adulthood, in the

same way a thermometer measures temperature. The sensor hairnet is connected to a computer that records the activity of the brain while you or your child complete tasks on the computer. The tasks are very simple and are often used with infants and children to see what our brains are doing whilst they are at work.

Beginning in 2016 we will invite TS 2000 participants to visit our research centre in London to take part in this project. We will reimburse participants and their families for travel, meals and other expenses during the trip, and can offer accommodation for long-distance travel. We are interested to see whether different brain processes are associated with different outcomes in TS. Identifying these brain processes will help us to develop tests to predict outcome from an early age. If you'd like more information on this project or to register your interest, please contact Dr Lizzie Shephard (elizabeth.1.shephard@kcl.ac.uk).

MRI brain scans

We are also planning to invite some participants to London to have a state-of-the-art MRI brain scan. We are hoping to use new techniques to study how different parts of the brain are connected to each other, as well as visualizing cortical tubers, SEGAs, and other lesions. We will be able to share these scans with your doctor, so this scan could be carried out in place of a routine clinical scan. We will also pay all expenses for the London visit. Our team might have already been in touch to find out if you are interested in a MRI scan, but you can also contact Dr Lizzie Shephard for more information (elizabeth.1.shephard@kcl.ac.uk).

Our Research Team

We are welcoming new members to the TS 2000 and saying goodbye to others. The study coordinator, Dr Fiona McEwen, will be leaving in the New Year for a new position at Queen Mary University of London, where she will be setting up a new project studying Syrian refugee children. Dr Holan Liang left during 2015 to return to clinical work as a consultant child and adolescent psychiatrist in London. Dr Charlotte Tye, while remaining at King's College London, will be running a new study of infants with TS. She aims to look at how brain activity, measured using EEG, can help predict which infants will develop problems with social interaction, communication, and attention. Drs McEwen, Liang and Tye will remain TS 2000 and will be helping to analyse data and publish results.

Dr Lizzie Shepard has recently joined the team and will be running the Phase 3 EEG study. She has previously worked with infants at risk for autism spectrum disorder and has expertise in using EEG techniques to study brain activity. Students currently on placement with the team – Isabelle Mortl, Juul Steenbruggen, and

Abigail Runicles – are currently providing invaluable support with data collection and administration.

Tea and Scones for TSC

The TS 2000 team took part in Tea and Scones for TSC during 2015, running a bake stall at the Institute of Psychiatry, Psychology & Neuroscience. We sold scones, Victoria Sponge, and mini Pavlovas, raising over £100 for the Tuberous Sclerosis Association.



Tea & Scones for TSC: The team doing their best impression of the Women's Institute!

Contact details

If any of your contact details have changed we'd really appreciate it if you could let us know.

Thank You!

We would like to take this opportunity to thank you again for participating in this study. Please **contact us** if you are having any problems completing the saliva kit (or need a new kit), cheek swabs, online or paper-based questionnaires, or would like to go through the questions in-person over the phone.

With Best Wishes,

Professor Patrick Bolton & the TS 2000 Study Team

Professor of Child & Adolescent Psychiatry and Honorary Consultant, Institute of Psychiatry, London

If you need to Contact us:

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