

TS 2000 Study 2023 Annual Newsletter

Hello everyone!

It's been a few years since we were last in touch, so we would like to update you on our progress on the TS2000 study and associated projects. Thank you for your ongoing participation in the study!

Phase 1

- We recruited individuals newly diagnosed between 2001 and 2006 across the UK. The final sample consisted of 125 children.
- All cases underwent comprehensive assessments, to measure medical and biological 'risk factors', which are factors that increase the chance that a different outcome will occur. In Phase 1, we looked at features of TSC that might be associated with cognitive and behavioral outcomes later in life.

Phase 2

- We visited 90 families to gather information on developmental and neuropsychiatric outcomes.
- We found a strong association between epilepsy in the first two years of life and later childhood outcome, in terms of i) intellectual ability and ii) autistic behaviours.
- We also found a developmental pathway linking the type of genetic mutation to the neurological manifestations of TSC through to ADHD symptoms.

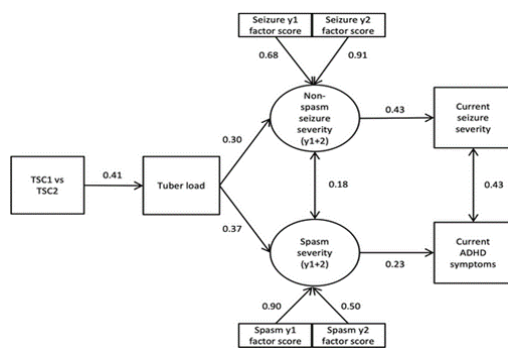
Phase 3

- We invited families to visit our lab in London so that we could examine brain function in TSC. We saw 48 participants with TSC and 20 young people without TSC. Participants completed tasks while their brain activity was recorded with electro-encephalography (EEG). Tasks included viewing pictures of faces, playing computer games, and watching videos.
- We found differences in brain activity in TSC that are associated with the number of tubers, infantile spasms in the first two years of life, and increased symptoms of autism.
- The results show that more severe infantile spasms and a greater number of tubers are associated with atypical neural processing of social information (faces) in young people with TSC, which in turn is associated with increased autistic symptoms.



Recent publications

The TS2000 team recently published a paper that looked ADHD symptoms in children with TSC. The team found relationships between the type of genetic mutation, cortical tuber load, epileptic spasm severity and how severe child ADHD symptoms are.



We need your help!

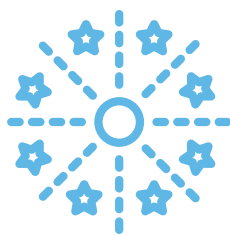
Phase 4 of the TS 2000 study will be starting in this year. We will be starting to look at the everyday experiences of young adults with TSC using smartphone technology. We are hoping to recruit individuals to try out this new smartphone app sometime in winter this year. More information will be provided in autumn this year.

As part of phase 4, we are running a focus group study where we will be asking young adults with TSC about their smartphone use and how we can use smartphones to monitor their everyday experiences. If you are aged between 16-30 with a diagnosis of TSC and are interested in taking part, please head to the TS2000 website www.ts2000study.co.uk for more information!

Our Research Team

Kate Fifield is a PHD research student at Kings College. Kate has joined the team to investigate the everyday experiences of young adults with TSC using smartphone technology.

Charlotte has returned from maternity leave and is helping Kate in starting Phase 4 of the TS2000 study.



Thank you!

Once again, we would like to thank you for all of your participation taking part in the TS2000 study.

Contact details

If any of your contact details have changes, we would appreciate if you could drop us an email on ts2000@kcl.ac.uk to let us know so that we can update our database.

With best wishes,

Dr Charlotte Tye & the TS2000 Study Team

Recent publications:

Tye, C., McEwen, F. S., Liang, H., Woodhouse, E., Underwood, L., Shephard, E., ... & Bolton, P. F. (2023). Epilepsy severity mediates association between mutation type and ADHD symptoms in tuberous sclerosis complex. *Epilepsia*.

