



# TS 2000 Study

## Annual Newsletter 2013-14

### Hello to All

It's been about a year since we were last in touch, so we've written this newsletter in time for Christmas. We want to let you know how the research is progressing and tell you about some exciting new developments in the study, including our progress in Phase 2 of the study, thanks to your ongoing participation.

### Phase 2

Last year, we started on Phase 2 of the TS 2000 study. Phase 2 aims to follow-up all the children in TS 2000 to see how they have been progressing since they were originally assessed. We hope to learn more about the types of problems that develop as children grow up. We are very keen to find out about the development of each and every child in the TS 2000 study, because we want to get as accurate a picture as possible about the 'natural history' of Tuberous Sclerosis.

In particular, TS increases the risk for developmental disorders, including autism and attention deficit hyperactivity disorder (ADHD). In Phase 2, we are trying to find out whether these disorders are the same in TS compared to 'typical' cases, and also which specific aspects of development in TS increase the risk for these disorders. We are also interested in whether individuals with TS are receiving appropriate and timely assessments of behavioural problems.

We started visiting families at their home, school or local clinic early this summer to carry out in-depth assessments of behaviour and cognition. We asked parents to complete an interview about how their child's behaviour, communication and play has developed, and complete some questionnaires that ask about specific questions of communication, social interaction 'adaptive' abilities for day-to-day life, activity levels and attention.

We have also asked parents to complete an online questionnaire on their child's development and behaviour. We left some families with home saliva kits and a questionnaire about access to services you receive for the various medical problems associated with TS. Please **contact us** if you are having any problems completing the saliva kit (or need a new kit), online or paper-based questionnaires, or would like to go through the questions in-person over the phone. **We're very keen to get all of the information back as soon as possible!**

We have also been asking children or young people with TS to take part in a range of assessments, tasks and games. These are designed to assess intellectual ability, social understanding, language, planning, memory, attention and thinking style. This involves a mix of conversation, paper and pencil tasks, simple social games, and computer-based tasks (such as listening to different stories and answering questions about them). Some of these assessments are used in clinics that specialise in autism and related disorders and ADHD. If children are found to be experiencing these types of difficulties then we offer to send a report to children's doctors. Families who take part in these assessments are being given gift vouchers as a token of our appreciation.

We have now seen almost 60 of the original TS 2000 participants – we are very grateful for your time and all of your hard work! We still have lots of families to see and will be in contact in the

coming weeks and months. If you haven't received information from us about Phase 2, or if you did but have lost it, please **contact us** and we will send it out to you.

### **Sibling assessments**

We are now offering certain assessments to some siblings who are *unaffected* by TS. It is important to have a comparison group who are closely matched to children with TS in terms of family and social factors and siblings provide an ideal group. We will ask the parent to complete online and paper-based questionnaires, and the sibling to complete some tasks to measure intellectual ability. Please **contact us** if you'd like to take part in this assessment, which will involve a short (1 hour) visit to your home, school or to our research centre, with travel costs reimbursed and a voucher for your time.

### **Research Update**

There have been a number of findings from Phase 1 of the TS 2000 Study. Firstly, we have identified a possible 'causal pathway' from genetic abnormality to tuber count to epilepsy severity to intellectual outcome in TS. This shows a strong relationship between epilepsy and intellectual ability. Likewise, in a detailed study of a few families in which TS was identified before seizures began, children who develop a form of epilepsy called infantile spasms are more likely to have problems in intellectual development, particularly if infantile spasms are not treated quickly and effectively. Taking this further, we are now exploring the distribution of intellectual ability in TS. To date our findings suggest that changes in intellectual ability might be due to a lack of development of IQ in a subset of TS cases that is associated with a set of specific risk factors (aspects that increase the chance of difficulties), now under investigation in Phase 2.

We have also investigated the impact and burden of TS on TS 2000 parents, showing that behavioural problems and difficulties (like attention problems and anxiety) account for most of parents' perceived impact of TS.

The added information we are gathering during Phase 2 is providing us with a much better understanding of the factors that lead to some children with TS having developmental and behavioural problems whilst others do not. We look forward to telling you more about the outcomes of the ongoing assessments of behaviour and cognition in next year's newsletter.

### **New members of the Research Team**

**Dr Holan Liang** is a consultant child psychiatrist who will be part of the Phase 2 team. Holan studied experimental psychology and medicine at Cambridge University. She trained in psychiatry and child psychiatry at the South London & Maudsley NHS Foundation Trust. She was awarded lecturer status by King's College London in 2005. She has been a Consultant Child Psychiatrist specialising in neurodevelopmental disorders in the NHS in London and Hertfordshire. Her recent publications and research interests are in the field of neuropsychiatry. She joins the TS2000 team as the Tuberous Sclerosis Association Research Fellow.

**Dr Lisa Underwood** is a postdoctoral researcher who will be part of the Phase 2 team. She has a degree in Psychology from the University of Staffordshire. Having previously worked as a researcher for the Royal College of Psychiatrists' Research Unit and Institute of Education, Lisa has been based at the Institute of Psychiatry since 2006. Her PhD in Health Service Research explored the mental health and service use of adults with learning disability and autism spectrum disorder.

### **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. The data we collect from this study is a very important way to learn more about the natural history of Tuberous Sclerosis.

With Best Wishes,

A handwritten signature in black ink that reads "P.F. Bolton". The signature is written in a cursive style and is followed by a long, sweeping horizontal line that extends to the right.

**Professor Patrick Bolton**                      & the TS 2000 Study Team  
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