

TS 2000 Study

Annual Newsletter 2008

Hello to All

This newsletter is our way of thanking you for your participation in the TS 2000 study during the past year and to let you know how the research is progressing.

What Have We Learnt So Far?

Now that we have good quality information for most of the TS 2000 children, we have been able to embark on a detailed analysis of these data. The Tuberos Sclerosis Association International Research Conference held at the University of Sussex in September provided the ideal opportunity for us to present our interim results to a large gathering of scientists and doctors with a special interest in tuberous sclerosis. John Yates talked about the different ways that TS can affect children and the challenges of making the diagnosis. First signs of the disease are being picked up at a younger age than in the past, for example during pregnancy when changes in the heart or kidney are detected by routine ultrasound scanning. Establishing the diagnosis can be difficult in pregnancy and the first year of life and the TS 2000 Study confirms the valuable role of genetic testing in the early diagnosis of TS. Early diagnosis before the start of epilepsy also raises challenging questions about how best to monitor these children to detect the start of seizures and when to begin anticonvulsant therapy. This provoked lively debate among the doctors at the conference. The study data also provides support for current management guidelines and highlights the need for increasing awareness amongst doctors looking after TS children to ensure these guidelines are followed. Patrick Bolton addressed the important question of why some children with TS have learning difficulties and other do not. For children in the TS 2000 study we have been collecting detailed information about their seizures, carrying out assessments of their development and IQ, reviewing their MRI scans to assess the severity of the changes in the brain and obtaining the results of genetic testing. We still have to chase up these data for some children, but Patrick talked about his analysis of the information collected so far. Needless to say, the relationships are complicated and more work needs to be done to sort out the detailed picture, but the preliminary analysis fits in with what we had suspected, that learning difficulties are more likely in children with extensive brain changes and more severe epilepsy. Ayla Humphrey in her presentation to the Conference focused on children who were recruited to the study before they started having seizures. In this group we have been able to carry out repeated assessments of their development and IQ before and after the onset of their epilepsy. Again, the picture is complicated but shows that in some children epilepsy can have a damaging effect on their development.

Completing the Jigsaw

Over the past year we have been working hard checking all the data from the study and chasing us missing information. This has gone well, and we now have good quality data on most of the children. However, there are still a few gaps. Because many of the children attend more than one hospital and have often seen several specialists, it can sometimes be difficult for us to track down the results of investigations, such as brain and kidney scans, or in some cases even to establish what investigations have been done. This is why we circulated a questionnaire earlier in the year asking you to confirm what investigations had been carried out. It has been very useful having this information and we are most grateful to everyone who returned questionnaires to us. For

those of you who haven't yet found time to return your questionnaire, it's not too late and we would still find it very helpful to have this information.

Future Plans

John Yates retired in April 2008 but is continuing to work on the study helping with the analysis and publication of the study data. Cathy MacLean has moved to a new post at Cambridge University, but in her spare time is also helping out chasing up missing data for the study. Having worked for so many years on the TS 2000 Study the whole study team are committed to completing the first phase and publishing the results. Patrick Bolton based at the Institute of Psychiatry and the Maudsley Hospital in London is now leading the TS 2000 Study and is busy planning the next stage of the research. Phase two of the study will involve further follow up of the TS 2000 children to chart their progress. All the children in the study are now old enough to allow detailed assessment of their development and behaviour, and adding these results to all the data we now have on the children should give 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.

Happy Holidays!

We would like to take this opportunity to wish you all a happy and healthy holiday season. And 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,



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