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## Australian first: Genetic testing gives families the answers

Australia's first genetic testing for Tuberous Sclerosis Complex (TSC) will help parents take the guesswork out of deciding whether or not to have more children.

Sydney Children's Hospital, Randwick and South Eastern Area Laboratory Services (SEALS) will today sign an agreement to develop and provide gene testing for the disease, making it one of only a few laboratories in the world to do so.

This great step forward has been taken thanks to funding from The Australasian Tuberous Sclerosis Society (ATSS). The funding will enable SEALS and Sydney Children's Hospital to provide both laboratory and genetic counselling staff to develop the service.

TSC is a genetic condition with severe manifestations including seizures, intellectual impairment, tumours, autism and skin lesions.

The disease affects approximately one in 6000 people with different degrees of severity.

"In about 70 per cent of cases tuberous sclerosis has come as a 'bolt from the blue', the result of a new genetic mutation (mistake), and no one else in the family is affected," said Sue Pinkerton, President of ATSS.

"In the remaining 30 per cent of cases, one of the parents and other family members are also found to have TSC."

Some people with the disease are mildly affected and may go through life without the diagnosis ever having been made, she said. Others are only identified after the diagnosis of an affected child.

Dr David Mowat, Clinical Geneticist at Sydney Children's Hospital, said it is important for TSC families to receive accurate genetic counselling as an adult with the TSC gene mutation has a one in two chance of passing it on to any child they might have.

"Previously parents would be screened for the disease by skin examination, brain and kidney scans to see if they were mildly affected or not," said Dr Mowat.

He said this was not an exact science so many chose not to have more children and risk passing on TSC.

media release

“With the availability of gene testing, parents and other relevant family members can make an informed decision about having children,” Dr Mowat said.

“Those testing negative for the TSC gene mutation can be reassured that they do not have TSC nor can they pass it on to future children. Those families testing positive can use the information to monitor their health and family planning.

“The new genetic testing will enable families to make decision about having children with a clear understanding of whether or not the disease can be passed on through the parents,” he said.

SEALS and Sydney Children’s Hospital will begin collecting blood samples in the coming weeks.

**Interviews:** Leanne, mother of 6-year-old Luke with TSC.  
Dr David Mowat, Clinical Geneticist, Sydney Children’s Hospital  
Ms Sue Pinkerton, President of ATSS

***Interviews can be arranged by contacting Kylie Rafferty, (02) 9382 3571 or 0411 730 842.***