



The University of Sydney

the  
children's  
hospital at Westmead

## INFORMATION SHEET

### STK9: A Second Rett Syndrome Gene?

We have recently identified mutations in a gene (*STK9*) which may be associated with a Rett-like phenotype, in association with autistic features, and often with early childhood onset severe seizures. We wish to extend our mutation screening to a larger group of Rett syndrome patients, who have been tested for mutations in the *MECP2* gene but in whom no mutations were found.

Enclosed with this document is a more detailed information sheet, two consent forms (a general consent form and a consent form for collection, storage and testing of human tissue for research). We would appreciate if you could ask your doctor to assist you with completion of the consent forms. The consent forms then need to be returned to me at:

*The Western Sydney Genetics Program,  
Children's Hospital at Westmead,  
Hawkesbury Rd,  
Westmead, NSW, 2145, Australia.*

Alternatively the completed forms may be faxed to me at 612 – 9845 1864 or emailed to me at [johnc@chw.edu.au](mailto:johnc@chw.edu.au)

If you have any questions relating to the project or the consent forms you can contact me (Ph. 612 – 9845 3452).

DNA samples for testing can be sent to me at the above address. Ideally, we would like to receive DNA that has been extracted from at least 5ml of blood which has been collected into an EDTA tube.

In addition, we would appreciate it if you and your doctor could complete the questionnaire over the page and e-mail, fax or post it to me.

Once again, thank you for your help.

With kind regards.

Yours sincerely,

John Christodoulou  
Professor & Director,  
Western Sydney Genetics Program



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Patient name: \_\_\_\_\_

DOB: \_\_\_\_\_  
(DD/MM/YYYY)

Patient's sex: \_\_\_\_\_  
(M/F)

Patient's doctor: \_\_\_\_\_

Doctor's contact address: \_\_\_\_\_

\_\_\_\_\_

Doctor's fax number: \_\_\_\_\_ Doctor's email address: \_\_\_\_\_

Clinical diagnosis: Rett syndrome \_\_\_\_\_  
(yes/no)

If yes, clinical subtype of Rett syndrome: \_\_\_\_\_

If no, clinical diagnosis: \_\_\_\_\_

Does the patient have intellectual disability? \_\_\_\_\_  
(yes/no)

Has a formal IQ assessment been performed? \_\_\_\_\_  
(yes/no)

Result: \_\_\_\_\_

Does the patient have non-febrile seizures? \_\_\_\_\_  
(yes/no)

Seizure type: \_\_\_\_\_

Does the patient have autistic features? \_\_\_\_\_  
(yes/no)

If yes, please provide details: \_\_\_\_\_

\_\_\_\_\_

MECP2 mutation screening performed (please tick the boxes):

Screening of exons 2, 3, and 4  screening of exon 1

Testing for a large MECP2 deletion  If yes, method used: \_\_\_\_\_

Other tests (please specify): \_\_\_\_\_

\_\_\_\_\_



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## PATIENT INFORMATION SHEET

Screening of candidate genes in children and adults with intellectual disability with or without autistic spectrum disorder.

Investigators:

Royal Alexandra Hospital for Children, Westmead

Professor John Christodoulou      Western Sydney Genetics Program – (612) 9845 3452  
Dr Bruce Bennetts                  Western Sydney Genetics Program – (612) 9845 3426  
Dr Carolyn Ellaway                Western Sydney Genetics Program – (612) 9845 0000  
Dr Katrina Williams                Dept of Clinical Epidemiology – (612) 9845 2006

Sydney Children's Hospital, Randwick

Dr David Mowat                      Dept of Clinical Genetics – (612) 9382 1708

University of NSW

Associate Professor Stewart Einfeld      School of Psychiatry – (612) 9350 2447

We are conducting a research study to see if mutations in newly identified genes can be responsible for non-specific intellectual disability in females, with or without autistic features. Your child has been diagnosed as having intellectual disability with or without an associated speech, social interaction or behaviour problem of the type seen in autism. There is a genetic disorder called Rett syndrome, which affects mainly girls, in who these types of behaviours are frequently seen. A gene (called *STK9*) responsible for some cases of Rett syndrome has recently also been found to be associated with autistic spectrum disorder associated with intellectual disability. We wish to explore how common this finding might be by undertaking DNA testing using a blood sample from your child.

As part of our research study, we would like to collect a blood sample for DNA testing from your child. In addition, we wish to set up a special cell bank derived from white blood cells that can be stored along with the DNA sample from your child for future research studies. These studies may include testing other new genes as they are identified in future.

If you agree to your child participating, a blood sample of 5-20ml [1 – 4 teaspoons] (depending on age) will be collected. A local anaesthetic cream can be applied if requested beforehand to minimise pain. Apart from the pain at the time that the blood is taken, a small bruise at the site of blood collection occasionally develops. Genetic testing can have significant consequences, which we will discuss with you. It is unlikely that our research studies will directly benefit your child.

Participation in this project is completely voluntary, and if you decide not to take part or decide to withdraw at any time this will not affect you/your child's care at the hospital in any way. For further information, please contact Professor Christodoulou (Ph 612-9845 3452), Dr Carolyn Ellaway (Ph 612-9845 0000), Dr Katrina Williams (Ph 612-9845 2006) or Dr David Mowat (Ph 02-9382 1708).

A copy of this Information Sheet and signed Consent Form will be given to you. You will be informed of the outcome of these studies.

If you have any concerns about the conduct of this study, please do not hesitate to discuss them with Professor Christodoulou or with Anne O'Neill (telephone: 612 - 9845 1316), the secretary of the Ethics Committee which has approved this project.



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PATIENT CONSENT FORM

Screening of candidate genes in children and adults with intellectual disability with or without autistic spectrum disorder.

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Dr Bruce Bennetts Western Sydney Genetics Program – (612) 9845 3426
Dr Carolyn Ellaway Western Sydney Genetics Program – (612) 9845 0000
Dr Katrina Williams Dept of Clinical Epidemiology – (612) 9845 2006

Sydney Children's Hospital, Randwick

- Dr David Mowat Dept of Clinical Genetics – (612) 9382 1708

University of NSW

- Associate Professor Stewart Einfeld School of Psychiatry – (612) 9350 2447

I have read and understand the Parent Information Sheet, and give my consent for my child to participate in this research study, which has been explained to me by \_\_\_\_\_

I understand that I am free to withdraw from the study at any time and this decision will not otherwise affect my child's care at the Hospital.

Name of Child:.....(Please print)

Name of Parent or Guardian:.....(Please print)

Signature of Parent or Guardian:.....

Name of Witness:.....(Please print)

Signature of Witness:.....

Name of Interpreter:.....(Please print)

Signature of Interpreter:.....

If you have any concerns about the conduct of this study, please do not hesitate to discuss them with Professor John Christodoulou (Ph 612-9845 3452), or with Ms Anne O'Neill (Ph 612-9845 1316), the Secretary of the Ethics Committee which has approved this project.