

## BACKGROUND

Rett syndrome was first described in 1983<sup>1</sup> and is now known to be a genetic disorder. Classical Rett syndrome only affects girls, who initially develop normally then regress, with loss of communication and hand skills. Ultimately, most become severely intellectually and physically handicapped. Rett syndrome may be associated with scoliosis, breathing abnormalities, growth retardation, gastrointestinal problems, poor mobility, sleeping problems and epilepsy. In 1999, mutations in the MECP2 gene were identified in a proportion of cases of Rett syndrome<sup>2</sup> and this may be useful for early diagnosis and prenatal testing in some cases.

The Australian Rett Syndrome Register used the APSU as one source of case ascertainment between 1993-5. The prevalence of Rett syndrome in Australia was estimated at 0.72 per 10<sup>4</sup> females and the cumulative incidence at 0.96 per 10<sup>4</sup> females <12 years<sup>3</sup>. This cohort was subsequently used for several clinical studies.<sup>4-8</sup> Few studies have examined the impact of the disorder on the child or family, or its management. The current study will establish a cohort of newly identified cases and follow them over time to provide data on the functional and medical aspects of Rett syndrome. Clinicians notifying cases will also be invited to participate in a study to identify the prevalence of the molecular defect in children with Rett syndrome.

## Objectives:

1. To identify newly diagnosed cases of Rett syndrome in Australia
2. To obtain information about the progression of the disorder
3. To describe the epidemiology (including survival status) of Rett syndrome
4. To define the phenotype-genotype correlation

## Case Definition

*A child < 16 years age with newly diagnosed or possible Rett syndrome according to the clinical criteria below or genetic testing.*

### Diagnostic Criteria for Rett syndrome

- Apparently normal prenatal and perinatal period
- Apparently normal psychomotor development through the first 6 months of life
- Normal head circumference at birth
- Deceleration of head growth between 5 months to 4 years
- Loss of acquired purposeful hand skills between ages 6 and 30 months temporally associated with communication dysfunction and social withdrawal
- Development of severely impaired expressive and receptive language and presence of apparent severe psychomotor retardation
- Stereotypic hand movements such as hand wringing/squeezing, clapping/tapping, mouthing and "washing/rubbing" automatisms appearing after purposeful hand skills are lost
- Appearance of gait apraxia and truncal apraxia/apraxia between ages 1-4
- Diagnosis tentative until 2-5 years

## REPORTING INSTRUCTIONS

Please report any child fulfilling the case definition, whom you have seen in the last month and whom you have not already reported to APSU

*Note:* Not all children with Rett syndrome (particularly Atypical cases) fulfil all diagnostic criteria, so all *suspected* cases should be reported.

### **Follow-up of notifications**

A questionnaire requesting further details will be forwarded to practitioners who report a case Rett syndrome.  
**A copy of the questionnaire is enclosed for your information.**

Clinicians will also be invited to pass on information to the child's family regarding optional participation in follow-up studies. A similar procedure was used previously without problems.

***Any doctor wanting information on the procedures for the research related to genetic testing should contact Dr Helen Leonard (08 9489 7789) or Clinical Associate Professor John Christodoulou (02 9845 3452).***

### **Investigators**

Dr Helen Leonard, Associate Professor Carol Bower, Dr Lakshmi Nagarajan, Dr Jon Silberstein, Dr Sue Fyfe, Associate Professor John Christodoulou, Dr Carolyn Ellaway, Dr Elizabeth Thompson, Dr Jim McGill, Dr Martin Delatycki, Dr MK Thong

### **Any questions should be directed to:**

Dr Helen Leonard, Medical Coordinator, Rett Syndrome Research Program, TVW Telethon Institute for Child Health Research, PO Box 855 West Perth WA 6871.

Tel: 08 9489 7789

Fax: 08 9489 7704

Email: rett@ichr.uwa.edu.au

### **References**

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