Australian Paediatric Surveillance Unit (APSU)
Parent/Carer Information Sheet
MECP2 Duplication syndrome

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BACKGROUND

Individuals who have two copies of the *MECP2* gene have common symptoms which are collectively referred to as *MECP2* Duplication syndrome. This syndrome has only recently been described and the number of people who are affected, and the rate at which it occurs in the population, are currently unknown.

Symptoms associated with *MECP2* Duplication syndrome include limited speech development, intellectual impairment, autistic like behaviours, poor muscle tone, epilepsy and recurrent respiratory infections. Some facial features, appear to be common in those affected. Diagnosis is difficult as little is known about the syndrome.

Most of the cases identified to date are male but some females have also been identified with a *MECP2* duplication. However females usually not have any symptoms. The majority of affected males have inherited the duplicated gene from their symptom free mother. In some the duplication occurs spontaneously and is not inherited from their mother.

THIS STUDY

We are conducting a national surveillance study through the Australian Paediatric Surveillance Unit (APSU) to learn more about *MECP2* Duplication syndrome. Over time this surveillance will allow us to determine how frequently the syndrome occurs and how many cases there are in Australia. Through a further study we also hope to contact with the families of those affected to better describe health outcomes. The information we collect will help health professionals gain an understanding of *MECP2* Duplication syndrome, improve diagnose and develop management guidelines.

LINKS

For more information about MECP2 Duplication syndrome, you can visit:
The Van Wright Foundation in Australia: www.vanwrightfoundation.org/
US Family website: mecp2duplication.com/
UK Family website: www.mecp2.co.uk/