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Genetic analysis reveals range of Rett syndrome

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The first comprehensive analysis of the clinical effects of genetic mutations involved in Rett syndrome will enable affected families to receive a more accurate indication of their child's prognosis.

The research was undertaken by an international collaboration based on information provided by families and clinicians from around the world to the InterRett online database at the Telethon Institute for Child Health Research.

The results have just been published in the March edition of the prestigious international journal *Neurology*.

Report co-author, Dr Helen Leonard, who heads the Australian Rett Syndrome Study, said the findings have revealed a wide variability in the effects of the syndrome.

"This is the first time that we've had quality information about a sufficient number of cases to be able to do a rigorous analysis comparing specific genetic mutations with they way the disorder is manifested in affected girls," Dr Leonard said.

"While Rett syndrome is caused by a mutation on the MECP2 gene on the X chromosome, variations in the mutation determine the severity of symptoms.

"Our analysis of eight common mutations that account for two-thirds of

cases, showed considerable variation in abilities”

Dr Leonard said the information would be of great value to families, clinicians and carers.

“Many parents have found the lack of information about their daughter’s prognosis very distressing and will welcome a clearer indication of what they might expect in the future,” Dr Leonard said.

“Our findings really emphasised how much variability there is in the syndrome and we hope that information will assist with earlier diagnosis.”

Dr Leonard said that girls with the milder form of Rett syndrome may retain some language, hand function and the ability to walk. In contrast, those with the severe form don’t show the usual pattern of regression but are affected from birth.

Source: Research Australia