New insight into Rett syndrome severity

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A research collaboration between Australia and Israel has identified a genetic variation that influences the severity of symptoms in Rett syndrome.

The finding is published in the latest edition of the international journal *Neurology*.

Dr Helen Leonard, who heads the Australian Rett Syndrome Study at the Telethon Institute for Child Health Research, said the finding was exciting in that it identifies a potential new target for treatment of the debilitating neurological disorder.

"We know that there is a wide range in the onset and severity of symptoms in patients with Rett syndrome but it has been difficult to give families a firm idea of how the disorder would progress," Dr Leonard said.

"This information is potentially helpful in predicting the clinical progression, but importantly, gives us another area to explore for potential therapies."

In the study, clinical information and DNA samples were gathered from 125 patients from the Australian Rett Syndrome Database and an Israeli cohort coordinated by Dr Bruria Ben Zeev at the Safra Pediatric Hospital, Sheba Medical Centre, Sackler School of Medicine, Tel Aviv. The genetic testing was undertaken by Professor John Christodoulou, from the NSW Centre for Rett Syndrome Research at the Children's Hospital at Westmead in Sydney and Dr Eva Gak from the Sagol Neuroscience Center at the Sheba Medical Centre.

Professor Christodoulou said while it has been established that Rett syndrome is caused by mutations in the MECP2 gene, these new findings have established a correlation between the severity of clinical symptoms and a common brain-derived neurotrophic factor (BDNF) polymorphism.

"Those patients with the normal BDNF genetic variant had less severe symptoms, with later onset and frequency of seizures," Dr Christodoulou said.

"We know that BDNF plays a major role in the development, survival and function of brain cells. What we now have to establish is the nature of the interaction between MECP2 and BDNF."

"It may be that if we can stimulate BDNF within patients with Rett syndrome, there is a chance that we can delay the onset of seizures and reduce some of the more debilitating aspects of the disorder."

Source: Research Australia (news : web)