Fact Sheet 2

Rett syndrome: What is it?

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Rett syndrome: What is it?

Brief history

The syndrome, itself, was first described in 1966 by an Austrian paediatric neurologist, Dr Andreas Rett. However, it was not until 1983 when an article on the condition appeared in a widely read neurological journal that the condition became internationally recognised. It was then that individuals (almost exclusively female) began to be diagnosed in Australia and elsewhere.

Incidence of Rett syndrome in Australia is 1: 9,000 live female births.

What Rett syndrome looks like.

A severe disorder of the nervous system in which early brain growth is affected. Children with Rett appear to be developing perfectly normally for the first part of their life. They will often crawl, walk, talk and run. Then things start to 'go backwards' or regress. The symptoms usually start to become apparent in the individual's initial 6 to 18 months of life.

There may follow periods of worsening, stabilization or even sometimes, apparent improvement. Many children with Rett are unable to speak, walk or use their hands. Breathing problems, feeding tubes, seizures, anxiety, gastrointestinal and orthopaedic issues are common. However, the degree of severity can vary considerably among affected individuals.

How is it caused?

Rett syndrome results from a chain of events which begin with a genetic mutation at the time of conception. In October 1999, it was discovered that mutations in the MECP2 gene cause the syndrome. To date, mutations in this gene have been found in 95% of individuals who have been diagnosed with Rett syndrome. It is a random event, can affect ANYONE, and in 99.5% of cases, the mutation was not inherited.

What's the good news?

Rett appears to be unique among neurological disorders. According to a scientific paper authored by Sir Adrian Bird, PhD, in 2007, the symptoms of Rett in mice dramatically reversed when treated. This raises the possibility that RETT COULD BE CURED!

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