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New recommendations ease challenges for Rett sufferers

January 23 2014, by Lizzie Thelwell

"Since publication, we have developed leaflets for clinicians and a booklet for families based on the recommendations, which provide information in a readable and understandable format," Dr Leonard says. Credit: Bradley Gordon

The Telethon Institute for Child Health Research together with international collaborators have created a comprehensive approach to tackle feeding and growth problems in Rett Syndrome sufferers.

A rare neurodevelopmental disorder caused by a mutation in the X-linked MECP2 gene, Rett Syndrome mainly affects girls and is associated with loss of hand and communication skills between the ages of one and two years.

The children, although apparently normal at birth and in the early

months of life, become severely intellectually and physically impaired following this regression.

Impairments include feeding difficulties related to problems with chewing and swallowing, hypersalivation and hyperventilation, which leads to poor growth and weight gain.

Telethon Institute expert and lead author Dr Helen Leonard says the project included a literature review, parental input and contributions from a multidisciplinary international panel of 27 clinicians.

The result is a comprehensive set of management recommendations to do with clinical assessment of factors such as growth, feeding difficulties and management to increase energy intake.

"The project pulled together data from 77 articles and three sets of guidelines, which we used to create 34 open–ended questions to inform areas in which the literature was lacking," Dr Leonard says.

"We then received feedback from a range of specialists, including gastroenterologists, paediatricians, child neurologists, clinical geneticists, dieticians, speech pathologists and nurses.

"Using a two–stage Delphi process, agreement was achieved on 101 of 112 statements, which inform recommendations we hope will have a major impact on managing the gastrointestinal and growth problems in Rett Syndrome.

"Since publication, we have developed leaflets for clinicians and a booklet for families based on the recommendations, which provide information in a readable and understandable format."

Dr Leonard says drawing together experts was difficult at times and

involved contacting practitioners directly by phone and sending encouraging emails at regular intervals to elicit responses.

Dr Leonard established the first Australia–wide register in 1993 to determine the prevalence of Rett Syndrome, and has managed an international register since 2003.

"I first met girls with this condition over 20 years ago as a doctor working at Disability Services Commission in WA, when the cause of the condition was unknown," she says.

"Since 1993, I have published over 70 journal articles and am continuing my research, including developing guidelines for bone health and evaluating the benefits of gastrostomy feeding."

More information: Leaflets and the booklet for families can be accessed here: rett.childhealthresearch.org.a … ports-and-books.aspx

Provided by Science Network WA

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