Bone health key in aiding Rett's syndrome patients

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Dr Leonard says maintaining regular contact with a large cohort of individuals and their families was invaluable in identifying potential factors that could be adversely affecting bone health. Credit: amanda tipton

An international panel of experts, led by WA researchers, is developing guidelines to help clinicians and families better manage bone health in individuals with Rett's syndrome.

Rett's syndrome is a rare neurological disorder occurring in 1/9000 births which is caused by a mutation in the MECP2 gene on the X chromosome which affects girls almost exclusively.

The infant typically develops normally in their first year of life however it eventually becomes apparent that they have a severe intellectual disability.

The individual also tends to develop other serious medical conditions such as epilepsy and osteoporosis.

UWA researcher Dr Helen Leonard has been at the forefront of bone research into Rett's for more than ten years.

Dr Leonard says her latest research was designed to ascertain what was happening to bone density over time in individuals with the disease.

"Generally we found that they're bone density measurements decreased further over time," she says.

"I mean they weren't that good to begin with; these girls have growth problems so they're small and quite thin.

"If you took into account their poor muscle and lean tissue mass they weren't quite as bad as they would seem, but on average they did definitely deteriorate."

Dr Leonard says measuring bone density was a fairly good indicator of the overall health of each individual.

"In terms of their health, mobility and physical activity…that varies according to the severity of their mutation," she says.

Puberty delay weighs on sufferers

"But those who struggled, or weren't capable, of walking, were malnourished or whose pubertal development was delayed were much more likely to have poorer results."

However, they found significantly less deterioration in individuals who had achieved, or were going through, puberty.

Dr Leonard says maintaining regular contact with a large cohort of individuals and their families was invaluable in identifying potential factors that could be adversely affecting bone health.

"We've been administering a questionnaire to their parents every two years so we have this ongoing register," she says.

"We wanted to be able to look at some of the things that might affect bone density such as their medications, diet and mobility."

Dr Leonard says she hopes recommendations from her own long-term work and other collaborative research can be made available this year.

"The aim is to produce literature that can help to improve, or maintain the quality of life for individuals with Rett's syndrome and their families."

More information: "Longitudinal bone mineral content and density in Rett syndrome and their contributing factors." DOI: dx.doi.org/10.1016/j.bone.2015.01.023

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