

## Rett syndrome research reveals high fracture risk

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Researchers at Perth's Telethon Institute for Child Health Research have found that girls and young women with Rett syndrome are nearly four times more likely to suffer a fracture.

The findings have just been published in the latest edition of the prestigious international journal *Paediatrics*.

The head of the Australian Rett Syndrome Study based at the Institute, Dr Helen Leonard, said the results had important implications for the care of girls with the syndrome.

"Many of the girls had fractures of the femur which are particularly disabling given the limited mobility of many of these children," Dr Leonard said.

"This information will be important for doctors and for families in both identifying fractures and endeavouring to find ways to prevent them where possible."

Dr Leonard said the study had found that girls with epilepsy and more severe forms of Rett syndrome were more likely to suffer fractures.

"Our next step will be to try to identify the mechanism that is making the girls more susceptible to fractures which will include looking at the effect of the specific gene that's responsible for the syndrome and also the impact of the drug therapies used for epilepsy," Dr Leonard said.



"The high incidence of fracture impacts on the quality of life, care needs and outcomes for this group and their families. We have shown previously that having a child with a fracture impacts in a negative way on the mother's mental health status."

Source: Research Australia

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