

## Study: Rett syndrome can strike males

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Australian researchers say they've discovered Rett Syndrome can strike males, as well as females.

Dr. Helen Leonard, who heads the Australian Rett Syndrome Study at the Perth-based Telethon Institute for Child Health Research, says the finding means testing for the genetic disorder should be considered in some baby boys who develop progressive serious neurological problems.

"The common thinking in the past had been that Rett syndrome only affects girls, and that the genetic flaw would be so serious in boys that they would die before birth," Leonard said.

"Worldwide there have only been 11 previously established cases in boys who have presented early in life with a severe clinical picture of progressive neurological decline and breathing abnormalities starting soon after birth," she said. "All but two had a family history of a girl in the family with Rett syndrome. This study has confirmed a further four cases with no family history."

She said it's likely some baby boys with early severe progressive encephalopathy might go undiagnosed. "We encourage pediatricians to think about (Rett Syndrome) as a possible cause of severe neurological abnormalities."

The study appears in the journal *Neurology*.

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