

## **Innovative test could revolutionise Fragile X screening and diagnosis**

Researchers from Murdoch Childrens Research Institute have developed an innovative new test that could revolutionise the way Fragile X syndrome is screened and diagnosed.

The Murdoch Childrens test uses a DNA region that was previously thought to have no function. Researchers say the new technology could be used as an early detection test for both male and females, and could be included in newborn screening tests, leading to improved quality of life for thousands of patients and their families.

Fragile X syndrome is a genetic disorder caused by a faulty switch of an important gene called FMR1 which is located on the X chromosome. Fragile X syndrome is associated with a range of developmental, physical and behavioural problems and is the most common known cause of inherited developmental disability worldwide. It is estimated that 1 in 130 females and 1 in 180 males carry the FXS gene and that 12 FXS carriers and one fully affected FXS child is born in Australia each week.

Early identification and intervention improves outcomes for children with Fragile X syndrome but until now it has been difficult for doctors to diagnose the disorder until the age of three years or older.

In a world-first, the researchers have shown that the new test can detect both the type and severity of symptoms in Fragile X syndrome with unparalleled accuracy in DNA samples from 154 females, with 18 of these having the 'faulty switch' in the FMR1 gene.

Lead researcher, Dr David Godler, from Murdoch Childrens said that in the study the test was shown to be superior to others available in predicting developmental disability particularly in females, and that the discovery could pave the way for a simple, accurate and inexpensive test for Fragile X syndrome that could be used for population screening.

"The test is especially advantageous for diagnosis and screening in females, because it can specifically and accurately identify those individuals who are expected to develop cognitive impairment and can therefore potentially identify those most likely to benefit from early intervention," Dr Godler said.

John Kelleher, President of the Fragile X Association of Australia has welcomed the finding, saying the test has the potential to benefit thousands of Fragile X patients by providing sufferers and their families with earlier detection and intervention, leading to better treatment and improved outcomes.

"This test has the potential to become one of the most powerful tools to be discovered this decade for accurate diagnosis of children with Fragile X syndrome. With it we may be able to test and treat affected individuals earlier on in their lives giving them the best chance to live to their full potential and to save parents the anguish of spending years searching for a diagnosis. The association has spent several years lobbying for the

inclusion of FXS testing within the heel prick test and now Dr Godler and his associates have developed the technology.”

The novel test was developed by Murdoch Childrens researchers Dr David Godler and Dr Howard Slater in collaboration with Dr Danuta Loesch from the School of Psychological Science, La Trobe University. The work was supported by the Thrasher Research Fund and National Health and Medical Research Council Development grant.

Larger studies are now underway internationally to further validate the findings. This work was published online today in the leading clinical laboratory journal *Clinical Chemistry*.

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**Vision and interview opportunities available:**

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Fragile X patients and families in Brisbane, Sydney, Perth and Melbourne