

Genetic carrier screening should be recommended for all couples planning a family

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Credit: Murdoch Childrens Research Institute

New research by the Murdoch Children's Research Institute (MCRI) has shown that the combined affected pregnancy rate of cystic fibrosis (CF), fragile X syndrome (FXS) and spinal muscular atrophy (SMA) is comparable to the population risk for Down syndrome – highlighting the need to routinely offer carrier screening.

Testing for Down syndrome has been routinely offered for over twenty years, yet most [health care providers](#) aren't offering screening for other

serious inherited conditions to couples planning a family, even though these tests have been available since 2012. This because there is a common misconception that testing is only relevant to those individuals with a known [family history](#).

These three genetic conditions – CF, FXS and SMA - have serious health consequences and are some of the most common inherited conditions in the general population. They are the three conditions that meet key criteria for population screening indicated by the relevant professional bodies.

Senior author, Professor David Amor said that professional bodies recommend that all couples considering or in early pregnancy be informed that testing for these conditions is available.

"In fact, offering screening for these conditions is recommended even when there is no family history."

The research, published in *Genetics in Medicine*, evaluated the 'prepair' genetic screening program and found that 1 in 20 people screened are carriers of at least one of the three conditions.

- 88% of carriers had no family history of the condition highlighting that screening is relevant to everyone
- One in 240 couples had an increased risk of having a child with these [conditions](#) and approx.
- Approximately 1 in 1000 pregnant women screened had an affected pregnancy which is comparable to the estimated incidence of Down syndrome

The test offered by the Victorian Clinical Genetics Services (VCGS) is simple and can be performed on a blood or saliva sample, typically beginning with testing the female partner. It can be performed before

pregnancy or in [early pregnancy](#) (prior to 12 weeks).

Having the test before a woman is pregnant means there is time to consider the options if they are found to be a carrier of CF, FXS or SMA.

Cystic Fibrosis Community Care, the Fragile X Association of Australia and Spinal Muscular Atrophy Australia Inc. support offering carrier screening for CF, FXS and SMA and have endorsed this [carrier screening](#) program.

First author, Dr Alison Archibald, Associate Genetic Counsellor with VCGS said, "It's important that all carriers are given the option of genetic counselling so they can discuss the implications of their results with an expert."

Provided by Murdoch Childrens Research Institute

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