MEDIA RELEASE **20 OCTOBER 2020**

Reproductive screening for fragile X syndrome, spinal muscular atrophy and cystic fibrosis recommended for funding by MSAC

Following an application by the Royal College of Pathologists of Australasia (RCPA), the Medical Services Advisory Committee (MSAC) has recommended to the Health Minister, Greg Hunt, that reproductive carrier screening for fragile X syndrome (FXS), spinal muscular atrophy (SMA) and cystic fibrosis (CF) be listed on the Medicare Benefits Schedule (MBS).

Dr Melody Caramins, clinical lead for the application and former Chair of the RCPA Genetics Advisory Committee said,

"This is an extremely promising step forward and we are closer than ever to having these tests listed on the MBS. The addition of these items to the MBS would mean that asymptomatic couples with no family history of CF, SMA or FXS, who are planning or in the early stages of pregnancy, would be able to access carrier screening at no personal cost. These disorders are recognised as having either a significant disability associated with them, or are life threatening or life shortening, and together occur at rates comparable to chromosomal disorders such as Down Syndrome, that we already screen for prenatally.

"There is strong support in the medical community to make this testing available. The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) recommends that every woman is offered information about carrier screening for CF, SMA or FXS when they are considering pregnancy. Reproductive carrier screening identifies couples at approximately 25% risk of having a child affected with one of these disorders. There are then more reproductive options open to them in order to avoid or better manage having a child affected with the disorder," said Dr Caramins.

CF, SMA and FXS are three of the most commonly inherited conditions in the general population and carriers are common in Australia. Approximately 1 in 20 people are carriers of one or more of these conditions[1], however most are usually unaware. Many children affected by these conditions are born to families with no history of disease, due to the rare nature of the conditions and patterns of inheritance.

FXS is the most common inherited cause of intellectual disability worldwide whilst SMA is one of the most common genetic causes of infant mortality. Although the average life expectancy for individuals with CF has increased with improved treatment regimens, there is currently no cure. It is estimated that around 1 in 250 women and about 1 in 800 men are carriers of the Fragile X premutation, one in 35 Australians carry the SMA gene, [2]and 1 in 25 Australians carry the gene changes that can cause CF.[3]

"Reproductive carrier screening is already being performed to a significant extent in Australia, but most patients are having to pay out-of-pocket, and this is a significant health equity issue in Australia, as research has demonstrated. The ability to test for genetic diseases such as this enables individuals to make informed reproductive choices and there are some potential treatments available which depend on knowing which mutations you have.

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[1] https://www.nature.com/articles/gim2017134

[2] https://smaaustralia.org.au

[3]https://www.cysticfibrosis.org.au/get-involved/support-and-services/support-and-services-information/cf-carrier-screening

"The addition of these tests to the MBS would have such an impact on the future of many women's decisions and their reproductive options," said Dr Caramins.

Recent research found a strong socioeconomic gradient in the uptake of reproductive carrier screening, with women living in the most advantaged postcodes across Australia significantly being more likely to have reproductive carrier screening than those living in the most disadvantaged areas. This research highlighted the need to minimise social and financial barriers limiting access to testing.[4]

"Testing can be performed on blood or saliva samples and the result will reveal if an individual is a carrier of CF, FXS or SMA. It is recommended that patients who undergo this type of testing, speak to a genetic counsellor who will be able discuss the results and co-ordinate further testing if necessary," said Dr Caramins.

For further information on the RCPA, please visit www.rcpa.edu.au or see updates on Facebook - @PathologyRCPA, Twitter - @RCPAPresident, @PathologyRCPA, or Instagram - @the_rcpa #RCPA #pathology #MedicineIsPathology.

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About the Royal College of Pathologists of Australasia:

The RCPA is the leading professional organisation representing pathologists, medical specialists and scientists who provide pathology testing in Australasia. Its mission is to train and support pathologists and to improve the use of pathology testing to achieve better healthcare.

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[4] https://pubmed.ncbi.nlm.nih.gov/32748403/