FOR IMMEDIATE RELEASE

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Reproductive carrier testing for three common inherited conditions receives full public funding in Australia

From today, reproductive carrier testing for cystic fibrosis, fragile X syndrome and spinal muscular atrophy is now available for every eligible Australian, thanks to a new Medicare item number, allowing people to make more informed choices.

The cystic fibrosis (CF), fragile X syndrome (FXS) and spinal muscular atrophy (SMA) communities in Australia have worked together for the last decade for equal access for all Australians to genetic carrier testing for these conditions.

In 2020, the Medical Services Advisory Committee recommended the three-condition genetic carrier test be listed on the Medicare Benefits Schedule. Until now, the test cost Australians upwards of $385 per person, limiting testing to only those who could afford it.

Today marks the first day the carrier test will be made freely available to eligible Australians under this item. This pivotal development signifies a substantial leap forward in making reproductive healthcare more accessible, an achievement celebrated by the three communities.

Approximately 1 in 20 Australians are carriers for one or more of these conditions, though many remain unaware of their carrier status. Importantly, children with these conditions are often born to families with no prior history due to the rare nature of these conditions and the patterns of inheritance involved.

“This testing provides couples with important knowledge to inform their reproductive choices,” says Wendy Bruce, Executive Director, Fragile X Australia.

Reproductive carrier testing plays a critical role in identifying couples with an increased chance of having children with CF, FXS or SMA. For autosomal* recessive conditions such as CF and SMA, if both parents are found to be carriers through this testing, the risk of having child with the condition is 25%. For carriers of FXS, which is an X-linked condition, the chance of having children with FXS can be up to 50%.

“Increasing accessibility of carrier testing is an incredibly important step forward in providing people with information to make informed decisions about their family planning and opens reproductive options that they can discuss with their healthcare professional”, says Stephanie Quattromani, Head of Programs and Services at Cystic Fibrosis Community Care.

“This has been a long time in the planning, and we are super excited that all Australian couples now have access to free reproductive carrier testing. All people, not just to those that can afford the test, will now be able to make informed choices about their reproductive plans prior to deciding to start a family”, says Julie Cini, spokesperson for SMA Australia.

Cystic Fibrosis Community Care, Fragile X Association of Australia, and Spinal Muscular Atrophy Australia join forces in their unwavering commitment to enhancing the lives of individuals and families impacted by these genetic conditions. This collaborative effort signifies a significant milestone in ensuring that every Australian family has access to the knowledge they need to make informed choices about their family's health.

For more information about this initiative or to schedule interviews with representatives from the collaborating organisations, please contact:

Available for interview:

- Stephanie Quattromani, Head of Programs & Services at CFCC. squattromani@cfcc.org.au
- Julie Cini, spokesperson for SMA Australia. reception@smaaustralia.org.au
- Wendy Bruce, Executive Director, Fragile X Association of Australia. wendy@fragilex.org.au

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* Autosomal conditions are those where both biological parents are carriers for a change in the same gene.

About cystic fibrosis (CF)

CF is the most common, life-limiting genetic condition affecting Australians, with 1 in 25 people in Australia being carriers. CF causes an abnormal build-up of thick and sticky mucus in the lungs, airways and digestive system. There is no cure but advances in treatment and care are helping people to better manage their CF, which often involves daily physiotherapy, medications and frequent hospitalisations.

About fragile X syndrome (FXS)

Fragile X-associated conditions are a family of inherited conditions caused by alterations in the fragile X gene, located on the X chromosome. Fragile X syndrome is the leading cause of inherited intellectual disability. Approximately, 1 in 250 women and 1 in 800 men are premutation carriers for fragile X.

About spinal muscular atrophy (SMA)

SMA is a condition affecting the muscles involved in movement, which progressively weaken and become wasted (atrophy) over time. This includes the muscles involved in general movement, swallowing and breathing. It is caused by the loss of specialised nerve cells called “motor neurons”, which are controlled by the brain and allow for muscle movement. Approximately 1 in 35 people are carriers for SMA in Australia.

About Cystic Fibrosis Community Care:

Cystic Fibrosis Community Care (CFCC) is a leading organisation dedicated to improving the lives of individuals and families affected by cystic fibrosis across New South Wales and Victoria. With a focus on research, support, and advocacy, CFCC are committed to enhancing the quality of life for those with CF.
www.cfcc.org.au

About Fragile X Association of Australia:

Fragile X Association of Australia is a non-profit organisation devoted to supporting individuals and families impacted by fragile X. Our mission is to support people living with fragile X to live their best possible lives through connection, education, and advocacy.
www.fragilex.org.au

About Spinal Muscular Atrophy Australia:

Spinal Muscular Atrophy Australia is the peak body for SMA in Australia, dedicated to improving the lives of individuals and their families affected by SMA. We work tirelessly to provide support to the community on best practice information care options, resources and treatment choices, raise awareness, and advocate for timely access to treatments.
www.smaaustralia.org.au