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90% of genetic carriers have no family history

Did you know that 90% of carriers for cystic fibrosis (CF), spinal muscular atrophy (SMA), and fragile X syndrome (FXS) have no family history of these conditions?

This is why routine screening is so important, and the great news is that genetic carrier screening for CF, SMA, and FXS is now bulk-billed* and accessible to all Australians.

Australian clinical guidelines (RANZCOG & RACGP)[1,2] recommend that doctors offer genetic carrier screening for common genetic conditions, such as cystic fibrosis, spinal muscular atrophy, and fragile X syndrome, to every woman or couple who are either planning or in the first stage of pregnancy, regardless of their probability of having these conditions.

As many as 1 in 25 Australians are carriers of cystic fibrosis, 1 in 35 individuals are carriers of spinal muscular atrophy, and approximately 1 in 250 females in the general population carry a genetic change that puts them at risk of having a child affected with fragile X syndrome.

One in 160 Australian couples will be found to be at risk of having a child affected by CF, SMA, or FXS.

Ideally, screening is performed before pregnancy to offer greater reproductive choice, such as pre-implantation genetic diagnosis through IVF, the use of donor eggs (or donor sperm for CF and SMA), donor embryos, or adoption.

To more information about Gene Access, the reproductive genetic carrier screening test for cystic fibrosis, spinal muscular atrophy, and fragile X syndrome offered by Clinical Labs, please visit antenatal.clinicallabs.com.au/patient/carrier-screening

1. The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) Guidelines.
2. The Royal Australian College of General Practitioners (RACGP) Guidelines.
*Subject to Medicare eligibility criteria.

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