Carrier screening is recommended either before pregnancy or early in pregnancy (during the first 12 weeks).

Screening is relevant to everyone - most people who are carriers do not have a known family history of the condition. Making a decision about screening is a choice only you can make.

Recommended carrier screening provider

GENETIC HEALTH QUEENSLAND PRACTICES
Royal Brisbane And Woman’s Hospital
E: ghq@hwath.qld.gov.au
P: +61 7 3646 1680
Queensland Children’s Hospital
P: +61 7 3068 4152
The Townsville Hospital
E: GHQ@health.qld.gov.au
P: +61 7 5202 3425
Toowoomba Base Hospital
P: +61 7 4616 6995
Gold Coast University Hospital
P: +61 7 5687 1515

SOGI
W: www.sogiscan.com.au
P: +61 7 3120 4200
E: counsellor@sogican.com.au

Medical Genetic Australia
W: www.medgen.com.au
P: 1300 651 741
E: andrea@medgen.com.au

Genes Australia
W: www.genesaustralia.com.au
P: 07 3088 6983
E: admin@genesaustralia.com.au

Northern Territory Clinical Genetics Service
P: (08) 8944 8731
E: genetichealth.ths@nt.gov.au

For more information
Cystic Fibrosis Queensland
W: cfqld.org.au
E: services@cfqld.org.au
P: (07) 3359 8000

Fragile X Association of Australia (FXAA)
W: fragilex.org.au
P: 1300 394 636

Spinal Muscular Atrophy Australia Inc. (SMA Australia)
W: smaaustralia.org.au
P: (03) 9796 5744

1 in 20 people carry a gene change for cystic fibrosis, fragile X syndrome, or spinal muscular atrophy, but most people are unaware that they are carriers.

Genetic carrier screening services are available in Australia. This screening helps people find out whether they have a chance of having a child with a genetic condition.
If you decide to have carrier screening, we recommend you follow the steps below:

1. **Visit your healthcare provider**
   Genetic carrier screening is usually requested by general practitioners, obstetricians, fertility specialists, midwives, genetic counsellors or medical geneticists.
   Let your healthcare provider know that you wish to have genetic carrier screening.
   Your healthcare provider will need to complete a test request form that you can take to a local pathology collection centre.
   Alternatively, if you do not live near a pathology collection centre you can order a saliva sample kit that can be mailed to you.
   If you are aware of a family history of an inherited condition, knowing the specific gene change in your family and communicating this to the healthcare provider arranging your testing, will ensure the appropriate test is ordered for you.
   It is recommended that your testing be performed by a specialist genetic testing laboratory.

2. **Have the test**
   It is recommended that the female partner is tested first, as testing may include conditions carried on the X chromosome.
   Take your test request form to an affiliated pathology collection centre and have your blood sample taken.
   If you have a family history of some conditions, such as FXS or CF, Medicare and Private health insurance may subsidise the cost of carrier screening.

3. **Visit your healthcare provider to get your results**
   Typically your results will be communicated to your healthcare provider within 2-4 weeks. They will inform you if you carry a gene change for any of these conditions.

4. **If the test shows you are a carrier**
   It is recommended that you discuss your result with a genetic counsellor.
   Some carrier screening services provide genetic counselling as part of their service.
   Depending on your result, your healthcare provider or a genetic counsellor will discuss carrier testing for your partner.