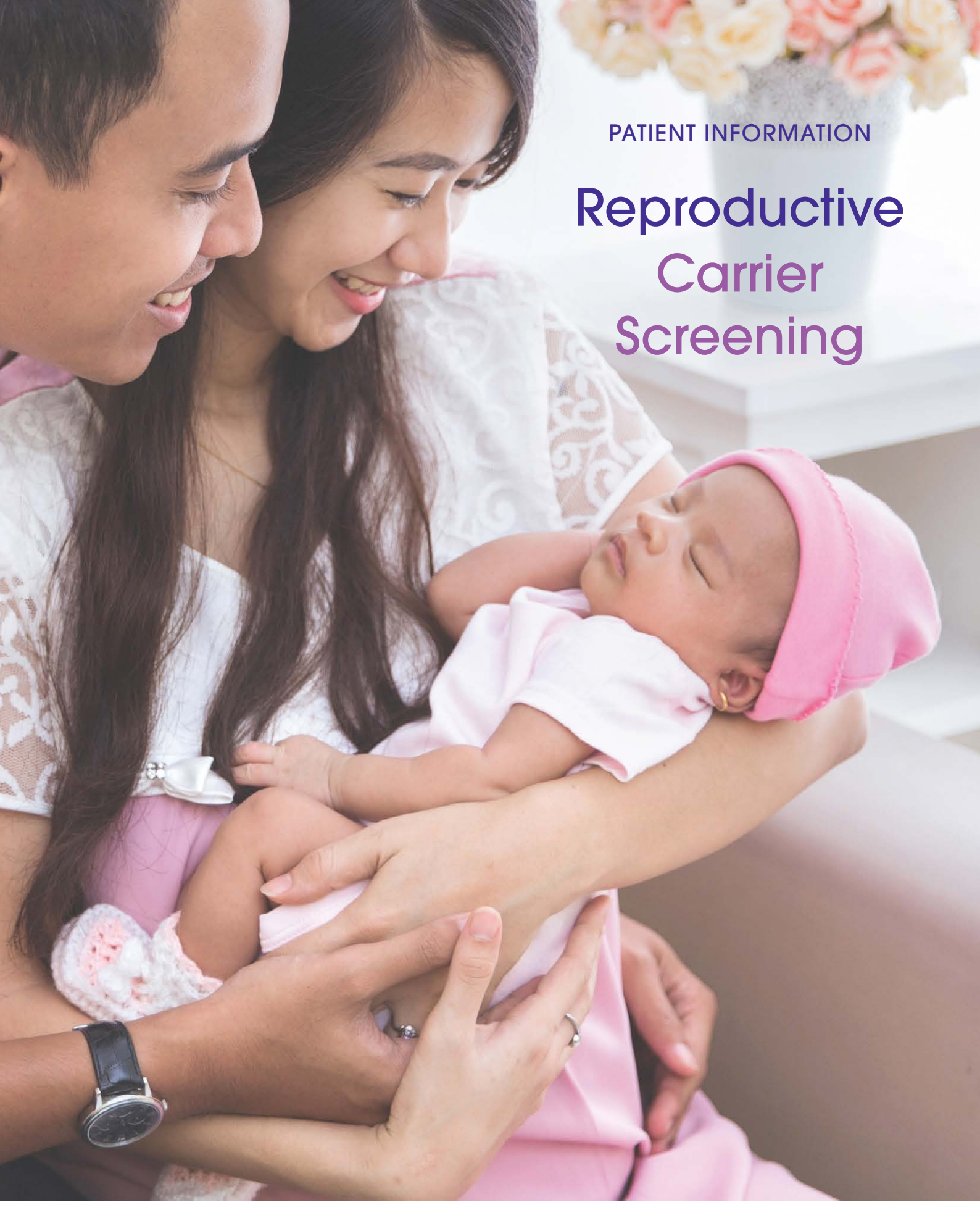

PATIENT INFORMATION

Reproductive Carrier Screening

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What is reproductive carrier screening?

Reproductive carrier screening identifies individuals and couples at risk of passing on certain genetic conditions to their children.

These childhood-onset conditions may lead to a shortened life expectancy, intellectual disability, and/or physical disability. Although most of these conditions are very rare, collectively they occur in more than 1 in 200 live births.

Why should I undergo screening?

Carrier screening results may inform your reproductive decision making. Below are some of the options available to couples and individuals identified as at-risk.

Be aware of risks and conceive naturally

Prenatal testing in early pregnancy

IVF and preimplantation genetic testing

IVF and donor egg, sperm, or embryo

Adoption

Virtus Genetics is a leading provider of preimplantation genetic testing (PGT) in Australia. A Medicare rebate for PGT is now also available for couples at risk of having children affected by a genetic condition.

Who should be screened?

Carrier screening may be suitable regardless of your family history, genetic ancestry, or age.

- Most children born with such conditions will not have a family history of the condition. Nevertheless, if you are aware of a family history of a genetic condition, it is important to discuss this with your doctor.
- For most of the screened conditions, the risk of being a carrier is not limited to those of a specific genetic ancestry.
- Unlike chromosomal conditions (eg. Down syndrome) that occur more commonly with advanced maternal age, the risk of being a carrier of a genetic condition does not vary with age.

When should I be screened?

The ideal time for carrier screening is when planning a pregnancy. Screening before becoming pregnant allows time for all necessary screening to be performed and allows for the widest array of reproductive options.

Carrier screening is also useful when performed in early pregnancy. However, there may be less time for all necessary screening to be performed and there is a narrower range of reproductive options available.

Couples may be screened simultaneously or sequentially (one partner is tested first, followed by the other partner if they are found to be a carrier). In sequential screening, we recommend screening the female partner first, in order to exclude X-linked conditions.

What results should I expect?

There are two potential screen results:

- No genetic variants are identified, or
- You are identified as a carrier of one or more genetic conditions

If no genetic variants are identified, this significantly reduces the chances of you being a carrier for the screened conditions, and of having an affected child. However, it does not completely eliminate these risks.

If you are found to be a carrier, it will be for an 'autosomal recessive' and/or 'X-linked' condition:

- Autosomal recessive conditions: If both you and your reproductive partner are found to be carriers in the same gene, there is a 1 in 4 (25%) chance of having a child affected by that genetic condition. Most of the screened conditions are autosomal recessive.
- X-linked conditions: Female carriers of an X-linked condition have up to a 1 in 2 (50%) chance of having a child affected by that genetic condition, irrespective of the male reproductive partner's carrier status.

In most circumstances, being a carrier will not have health implications for you and will only increase your risk of having an affected child. In rare situations, test results may have health implications for your own health, such as revealing an undiagnosed condition or increased risk of a medical condition. Your results may also have implications for your family members.

What screening options are available?

TEST	Basic carrier screen	Expanded carrier screen
Number of screened genes	3	289 in females 268 in males (excluding X-linked conditions)
Identifies cystic fibrosis (CF) carriers CF is associated with respiratory and digestive problems. Approximately 1 in 25 individuals are carriers.	✓	✓
Identifies spinal muscular atrophy (SMA) carriers SMA is associated with muscle problems throughout the body. Approximately 1 in 40 individuals are carriers.	✓	✓
Identifies fragile X syndrome carriers Fragile X syndrome is an X-linked disorder and the most common known cause of inherited intellectual disability. Approximately 1 in 250 women are carriers.	✓	✓
Test fee	$400	$650
Turnaround time	2-3 weeks	3-4 weeks
Testing laboratory	Virtus Genetics (Australia)	Invitae (U.S.)*
Eligible for Medicare rebate	Medicare rebates may apply to part of the test fee if relevant clinical criteria are met	A Medicare rebate is not available and test will be privately billed

* Your specimens and personal information will be made available to Invitae for the purposes of providing test services and performing related activities, as further described in Invitae's Privacy Policy (www.invitae.com/privacy).

Test limitations

Carrier screening does not exclude the possibility of having a child affected by a genetic condition. There will remain a small residual risk of being a carrier in one or more of the tested genes. Test reports rely on currently available information in the medical literature. A repeat specimen collection may be required in a minority of cases.

How do I arrange to be screened?

Step 1: Discuss carrier screening with your doctor, including any known family history.

Step 2: **Option 1** - Take the request form completed by your doctor to a Virtus Diagnostics collection centre to have a blood specimen collected. Specimen collections through other pathology providers may incur additional transport/handling fees from that provider.

Option 2 (available for expanded carrier screening only) - On request from your doctor, we will send out cheek swab kits to your home. Kits will include everything you need to self-collect a specimen and instructions on how to post the kit back.

Step 3: Results will be securely returned to your doctor in up to 3-4 weeks.

Virtus Genetics is a member of Virtus Health, an Australian owned and publicly listed company and one of the most successful medical collaborations of its kind globally. Our heritage is in assisted reproductive services as a leading fertility treatment provider in Australia, Europe and South East Asia.

Our network of accredited genetic laboratories offers boutique genetic testing services to specialists and general practitioners. We have a dedicated team of experienced scientists and specialist pathologists to provide expert advice and ensure testing is of the highest quality.

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Information herein is correct as of 31.01.2022

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