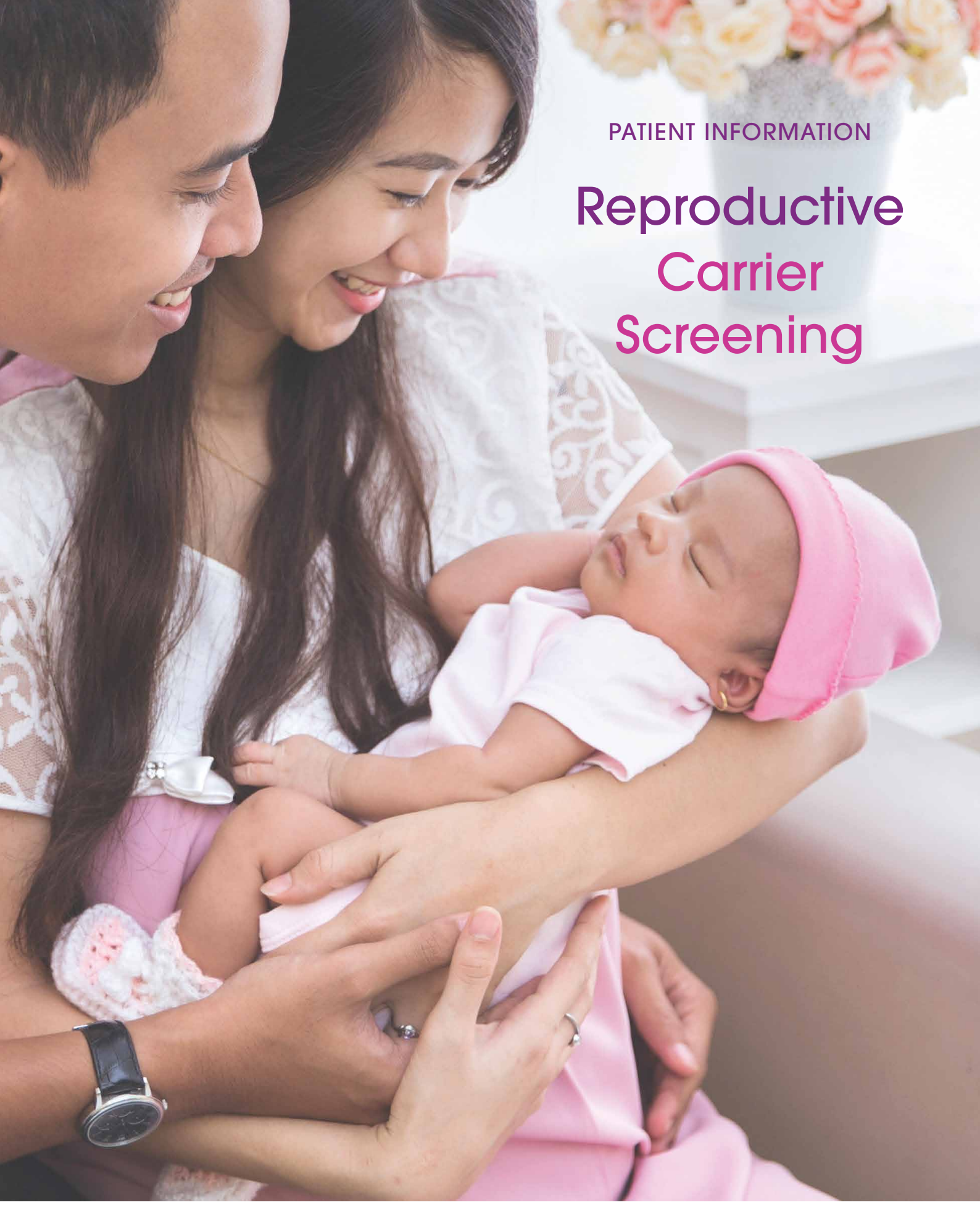

PATIENT INFORMATION

Reproductive Carrier Screening

www.virtusgenetics.com.au

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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?

	virtus core carrier screen	virtus core+ carrier screen	virtus duo carrier screen	Individual Expanded Carrier Screen (Invitae)
Genes	**3 genes** Cystic Fibrosis (CFTR) Fragile X Syndrome (FMR1) Spinal Muscular Atrophy (SMN1)	**6 genes** Cystic Fibrosis (CFTR) Fragile X Syndrome (FMR1) Spinal Muscular Atrophy (SMN1) Alpha and Beta thalassemia (HBA1/HBA2 and HBB)	**Up to 390 genes** • Couple specific - repeat carrier screening required if reproductive partner changes • Individual testing not available • Cannot be used with donated eggs or sperm	**Option 1** Legacy panel - up to 289 genes **Option 2** New panel – up to 556 genes
Testing location	**Australia** (Virtus Genetics)	**Australia** (Virtus Genetics)	**Australia** (Virtus Genetics)	**United States** (Invitae)
Report inclusions	**Individual carrier status for all tested conditions**	**Individual carrier status for all tested conditions**	**Individual carrier status reported for all 6 genes in Core+ Carrier Screen** **AND** Carrier status for other tested conditions are reported only if: • There is a risk to offspring - both partners are carriers or female partner is an X-linked condition carrier • There are personal health implications for carriers themselves	**Individual carrier status for all tested conditions.** Recommended for patients using egg or sperm donors or patients wanting to know their individual carrier status
How to Request	**Standard pathology request form** Requests for "Core Carrier Screen" "Virtus Genetic Screen" or "VGS" will undergo the 3 gene Core carrier screen.	**Standard pathology request form** Requests that specify "Core+" or Core and Alpha and Beta thalassemia (HBA1/HBA2 and HBB) will receive the Core+ 6 gene screen.	**A dedicated Duo Carrier Screen request form must be used** This form ensures that both partners provide the necessary consent and are unambiguously identified	**Standard pathology request form** The default test performed will be the Legacy Panel (up to 289 genes). Please clearly indicate on the request form if the New Panel is being requested (eg."556 genes" or "New Invitae panel").
Sample type	**Blood or cheek swab** To have cheek swab test kits sent to their home, patients can call Virtus Genetics (1800 837 284) or email a copy of their request form to info@virtusgenetics.com.au. Kits will be available at Virtus collection sites.	**Blood or cheek swab** To have cheek swab test kits sent to their home, patients can call Virtus Genetics (1800 837 284) or email a copy of their request form to info@virtusgenetics.com.au. Kits will be available at Virtus collection sites.	**Blood:** MUST be collected together by a Virtus collector **Cheek swab:** Kits may either be handed out in the clinic or requested from Virtus Genetics at info@virtusgenetics.com.au or call 1800 837 284.	**Blood or cheek swab** To have cheek swab test kits sent to their home, patients can call Virtus Genetics (1800 837 284) or email a copy of their request form to info@virtusgenetics.com.au. Kits will be available at Virtus collection sites.
Price	**Bulk Billed** **Private Fee: $340** for non Medicare Eligible Patients	**Approx. $150 out of pocket** **$490 per person** Medicare rebate available for eligible patients.	**Approx. $510 out of pocket** **$850 per COUPLE** Medicare rebate available for eligible patients.	**$720 per person**
Genetic counselling	Pre and Post-test genetic counselling can be arranged through the Virtus Diagnostics Genetics team at no cost to the patient. Clinicians and patients can organise a session via the website at www.virtusgenetics.com.au/book-an-appointment/ or by calling our team on 1800 837 284			

* 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).

Private billing charges will apply in the case of Medicare rejections.

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 - Request that a cheek swab kit is sent to your home, or pick up a kit from the clinic. 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 3-6 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.

General enquiries: 1800 837 284 **Email:** info@virtusgenetics.com.au

Information herein is correct as of 01.11.2023