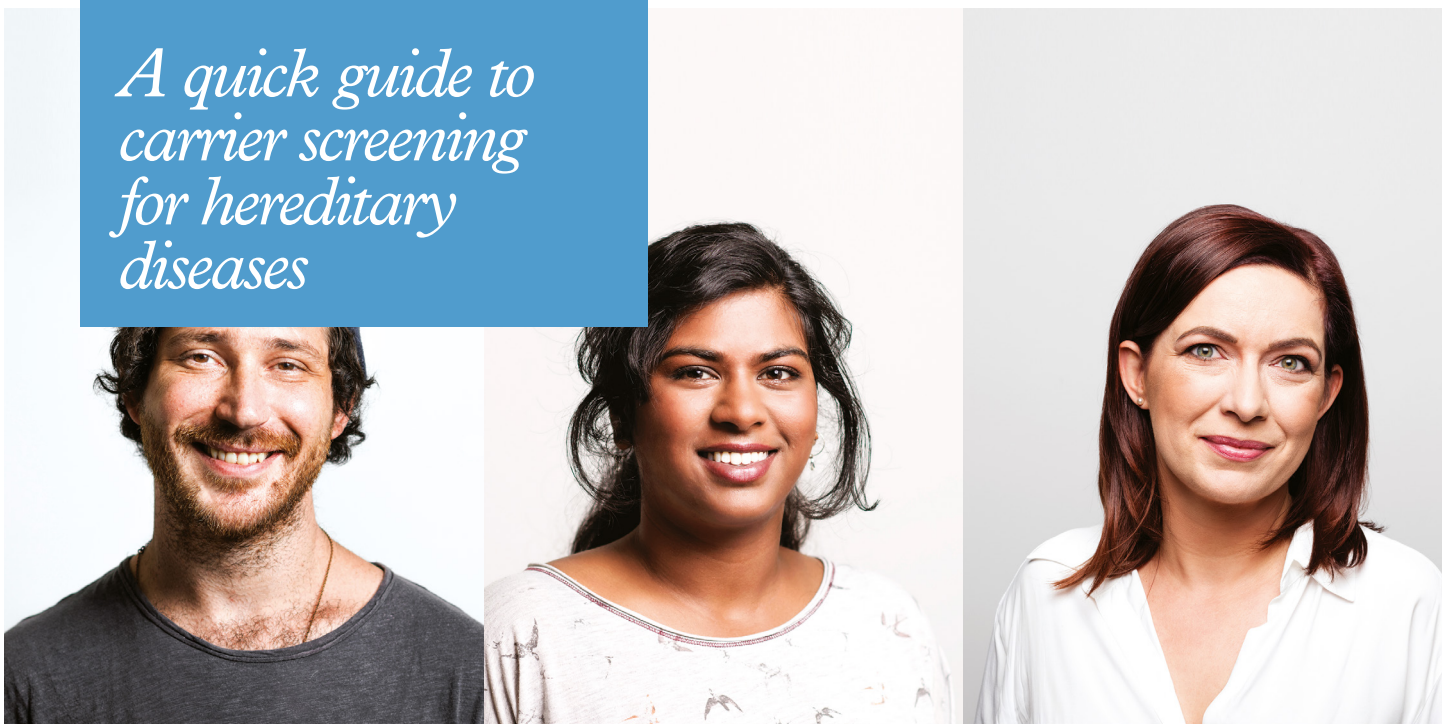
A quick guide to carrier screening for hereditary diseases



Carrier screening is a form of genetic testing that detects whether an individual or couple are carriers of an autosomal recessive and/or X-linked genetic condition.[1] Preconception and early pregnancy genetic screening allows women and couples to understand their risk of passing an inherited condition on to their children and make informed reproductive choices in line with their personal wishes and values.

The stats on inherited diseases

~1–2%
of non-consanguineous couples have a
1 in 4 chance
of having a child with an autosomal recessive or X-linked recessive condition[2]

~1 in 200
babies are born with an autosomal recessive or X-linked recessive condition
THAT'S **x4 higher**
than Down syndrome[2,3]

20%
of all infant deaths
AND **10%**
of paediatric hospital admissions are associated with inherited disorders[4]

What screening options are currently available?

Single-condition screening	Three-condition screening	Expanded carrier screening
Screens for one specific inherited disorder (eg Tay–Sachs disease or haemoglobinopathies)[2]	Screens for three of the most common inherited rare diseases (cystic fibrosis, spinal muscular atrophy, fragile X syndrome)[5]	Screens for hundreds of different inherited disorders regardless of ethnic background or family history[2,6]

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Who should be offered carrier screening?

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists guidelines recommend that clinicians:[7]

- offer basic thalassaemia screening to **all pregnant women** (via full blood examination)
- offer information on carrier screening (both three-condition and expanded panel) to **all women** planning a pregnancy or in the first trimester of pregnancy
- offer additional screening to individuals of Eastern European (Ashkenazi) Jewish descent, due to a higher incidence of conditions such as Tay–Sachs disease in this population
- offer a more detailed discussion about carrier screening with an informed clinician
- obtain informed consent for screening – this should include any out-of-pocket expenses that are required for the chosen test
- refer all carrier couples, and women who are carriers of an X-linked recessive disorder, for genetic counselling.

It is now recommended that carrier screening be offered to all women planning a pregnancy or in the first trimester of pregnancy, regardless of family history or geographic origin.[7]

Does carrier screening replace NIPT and combined first trimester testing?

Carrier screening **does not** replace non-invasive prenatal testing (NIPT) and combined first trimester screening (CFTS). These tests should still be offered to all pregnant women to screen for an increased risk of having a child with Down syndrome or other chromosomal disorder, as carrier screening **does not** detect chromosomal anomalies and NIPT and CFTS **do not** identify carrier status of single-gene disorders.[1,8]

What does carrier screening cost?

Medicare provides rebates for some genetic tests.*
Patients can expect to pay fees in the range of:†

- single-condition screening: $100–$200
- three-condition screening: $350–$400
- expanded carrier screening: $580–$900.

Resources for healthcare professionals

Visit **bewaretherare.com.au** for useful clinical resources, including an RACGP-accredited CPD (Cat. 2) Activity suitable for general practitioners, practice nurses, obstetricians, fertility specialists, general paediatricians and maternal child health nurses.

References **1.** The Royal Australian College of General Practitioners. Genomics in general practice. East Melbourne, Vic: RACGP, 2019. Available at www.racgp.org.au/clinical-resources/clinical-guidelines/key-racgp-guidelines/view-all-racgp-guidelines/genomics-in-general-practice [Accessed 14 January 2020]. **2.** Delatycki MB, Laing NG, Moore SJ, et al. Preconception and antenatal carrier screening for genetic conditions: The critical role of general practitioners. Aust J Gen Pract 2019;48(3):106–10. doi: 10.31128/AJGP-10-18-4725. **3.** Lalani SR. Current genetic testing tools in neonatal medicine. Pediatr Neonatol 2017;58(2):111–21. **4.** Bell CJ, Dinwiddie DL, Miller NA, et al. Carrier testing for severe childhood recessive diseases by next-generation sequencing. Sci Transl Med 2011;3(65):65ra4. doi: 10.1126/scitranslmed.3001756. **5.** Archibald AD, Smith MJ, Burgess T, et al. Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: Outcomes of 12,000 tests. Genet Med 2018;20(5):513–23. **6.** Henneman L, Borry P, Chokoshvili D, et al. Responsible implementation of expanded carrier screening. Eur J Hum Genet 2016;24(6):e1–e12. doi: 10.1038/ejhg.2015.271. **7.** The Royal Australian and New Zealand College of Obstetricians and Gynaecologists. Genetic carrier screening. East Melbourne, Vic: RANZCOG, 2019. Available at https://ranzcog.edu.au/wp-content/uploads/2022/05/Genetic-carrier-screeningC-Obs-63New-March-2019_1.pdf [Accessed 22 June 2022]. **8.** The American College of Obstetricians and Gynecologists. Prenatal genetic diagnostic tests. Washington, DC: ACOG 2019. Available at https://acog.org/Patients/FAQs/Prenatal-Genetic-Diagnostic-Tests [Accessed 17 January 2020]. **9.** Australian Government. Budget 2022–23. Budget Paper No.1: Budget Strategy and Outlook. Canberra: Australian Government, 2022.

*The cost of screening for haemoglobinopathies is generally covered by state/territory government funding. Relatives of affected individuals/carriers **may** be eligible to access funded testing via Medicare or state/territory government funding. Medicare rebates will be introduced in 2022–2023 to support access to carrier screening for cystic fibrosis, spinal muscular atrophy and fragile X syndrome.[9]
†Fee estimates are per person. Actual fees vary by provider.

We acknowledge the Traditional Custodians of the lands and seas on which we work and live, and pay our respects to Elders, past, present and future.

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