

Genetic Carrier Screening

Factsheet

Genetic carrier screening tests healthy adults to assess their chance of having a child with an inherited single gene condition. This screening test is ideally performed prior to pregnancy so that individuals/couples with an increased chance of having an affected child have access to all of the available reproductive options.

Why is genetic carrier screening important?

Although single gene conditions are individually rare, collectively, around 1 in 200 babies are born with an inherited single gene condition⁽¹⁾. These conditions account for 20% of infant mortality⁽¹⁾.

Most individuals are carriers of 2 or more single gene conditions but are unaware of this because being a carrier most often has no health implications⁽¹⁾.

Who is it for?

The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) recommends that all individuals planning a pregnancy, or in early pregnancy should be offered genetic carrier screening⁽²⁾.

What are the options?

Monash IVF offers two genetic carrier screening panels. Both of our panels can be accessed at home (via saliva sample) or in-clinic (via blood sample):

The **3-gene** panel tests for 3 conditions: cystic fibrosis, fragile X syndrome and spinal muscular atrophy. Around 1 in 240⁽³⁾ couples will have an increased chance of having a child with one of these conditions.

The **expanded** panel tests for 400+ genetic conditions, including the 3 conditions above. It also includes other common conditions such as thalassaemia and Duchenne muscular dystrophy (DMD). Collectively, around 1 in 50⁽⁴⁾ reproductive couples will have an increased chance of having a child with one of these conditions.

Medicare has recently introduced a rebate for three gene genetic carrier screening.

Monash IVF strongly recommends that patients consider expanded carrier screening, as it is a pan-ethnic test which makes it appropriate for individuals of all ethnicities. All the conditions on our expanded panel are just as significant as the first 3-genes and can significantly affect the health of a future child.

A full list of conditions included can be viewed at:

bit.ly/3sLNICZ

Genetic Carrier Screening Options	3-Gene	Expanded
Conditions included in screen	3	400+
Patient out-of-pocket cost (for Medicare eligible individuals)	\$0 (bulk billed)*	\$695/individual \$990/couple^
Turnaround time once sample received	3 weeks	4-6 weeks
At-home saliva or in-clinic blood test options	✓	✓
Support from an in-house, qualified Genetic Counsellor	✓	✓
Support from reproductive and genetic experts in the event of a high chance result	✓	✓
Expected number of couples identified as having a high chance result	1 in 240 ⁽³⁾	1 in 50 ⁽⁴⁾

*A \$25 postage and handling fee applies for at-home 3-gene orders.

^Pricing is correct as at 1 November 2023. Please check the Monash IVF website for up-to-date pricing.

What if the results indicate a high chance?

If the results indicate an increased chance result, our experienced genetic counselling team are available to offer support and consultation to both the referring doctor and the patient(s).

What are the reproductive options for carriers of genetic conditions?

For people who are carriers of genetic conditions wanting to start a family there are options which may include testing in pregnancy, testing after the birth of a child, considering IVF with a gamete donor or considering IVF with preimplantation genetic testing of embryos. The Monash IVF team are equipped to advise on all reproductive options.

A referral to see a fertility specialist is a good first step. Monash IVF has fertility specialists in every mainland state and territory in Australia, for a full list of locations, visit: monashivf.com/locations

Medicare funding may be available for IVF treatment and embryo screening for those identified as having an increased chance of having a child with a single gene condition.

Get more information:

To get more information or order a kit, scan this QR code or visit: monashivf.com/genetic-testing

E geneticsadmin@monashivf.com

T 1800 684 198 (free call)

monashivf.com



(1) The Royal Australian College of General Practitioners (2020) Genetic carrier screening: What GPs need to know, NewsGP, accessed 23 January 2024, <www.racgp.org.au/newsGP/>

(2) The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (2019) Genetic carrier screening (statement), March 2019.

(3) Archibald, A. D., Smith, M. J., Burgess, T., Soarff, K. L., Elliott, J., Hunt, C. E., ... Amor, D. J. (2018). Reproductive genetic carrier screening for cystic fibrosis, fragile X syndrome, and spinal muscular atrophy in Australia: Outcomes of 12,000 tests. *Genetics in Medicine*, 20(5), 513-523. doi:10.1038/gim.2017.134

(4) Berbic M, Hesson L, Clarke J, et al. (2022) Reproductive carrier screening (RCS) to identify Australian couples at risk of having children with autosomal recessive and X-linked conditions, RANZCOG Annual Scientific Meeting, October 2022.

Genetic Carrier Screening Locations

For at-home testing via saliva sample visit: monashivf.com

For in-clinic testing via blood sample, contact your nearest location below:

NSW

Albury The Gardens Medical Centre, Level 2 470 Wodonga Place Albury 2640 (02) 6041 2677	Bondi Junction Westfield Tower 1 Level 26 520 Oxford Street Bondi Junction 2022 (02) 9389 1177	Sydney CBD Level 10 207 Kent Street Sydney 2000 (02) 9154 1130	Parramatta Level 2 1 Fennell Street Parramatta 2151 (02) 9890 9022	Penrith Level 3 38 Somerset Street Kingswood 2747 (02) 9154 1155
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Bella Vista Q Central Building Level 1, Suite 114 10 Norbik Drive Bella Vista, 2153 (02) 9629 2011	Bondi Junction Level 17, Suite 1705 Westfield Tower 1 520 Oxford Street Bondi Junction 2022 (02) 9388 0955	Frenchs Forest Suite 5, Level 6 Northern Beaches Hospital 105 Frenchs Forest Road Frenchs Forest 2086 (02) 9154 1174	Kogarah Suite 1A, Level 1 4 Belgrave Street Kogarah 2217 (02) 9553 9611	Liverpool Level 2, Suite 205 161 Bigge Street Liverpool 2170 (02) 9822 8447
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Newtown RPA Medical Centre Suite 404 100 Carillon Avenue Newtown 2042 (02) 9516 2064	St Leonards Tower A, Level 4 North Shore Health Hub 7 Westbourne Street St Leonards 2065 (02) 9413 9196	Sydney Suite 6.02, Level 6 309 Kent Street Sydney 2000 (02) 9290 2122
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QLD

Brisbane Level 1 Leichhardt Ct 55 Little Edward Street Spring Hill 4000 (07) 3069 9600	Gold Coast Level 1 7 Short Street Southport 4215 (07) 5519 1600
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VIC

Clayton Monash Surgical Private Hospital Suite 1, 252-256 Clayton Road Clayton 3168 (03) 9590 8300	Cremorne Level 1 510 Church Street Cremorne 3121 (03) 9420 8200	Geelong Geelong Private Medical Centre, Level 2 73-77 Little Ryrie Street Geelong 3220 (03) 5222 8599
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Berwick Clinic on Clyde Suite 3 40-42 Clyde Road Berwick 3806 (03) 9707 0887	Box Hill Ekeru Medical Centre Level 2, Suite 2.04 116-118 Thames Street Box Hill 3128 (03) 9890 6177	Clayton Monash Surgical Private Hospital 252-256 Clayton Road Clayton 3168 (03) 9544 6744	Cremorne Level 1 510 Church Street Cremorne 3121 (03) 9427 7610	Mulgrave 441 Police Road Mulgrave 3170 (03) 9790 1766	Sunshine Sunshine Private Floor G, Suite 1 147 Furlong Road (03) 9097 1450
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WA

West Leederville (Previously PIVET Medical Centre) 166-168 Cambridge Street West Leederville 6007 (08) 9422 5400

SA/NT

Dulwich 180 Fullarton Road Dulwich, SA 5065 (08) 8333 8111	Darwin Harry's Place 1 Willeroo Street Tiwi, NT 0810 (08) 8945 4211
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