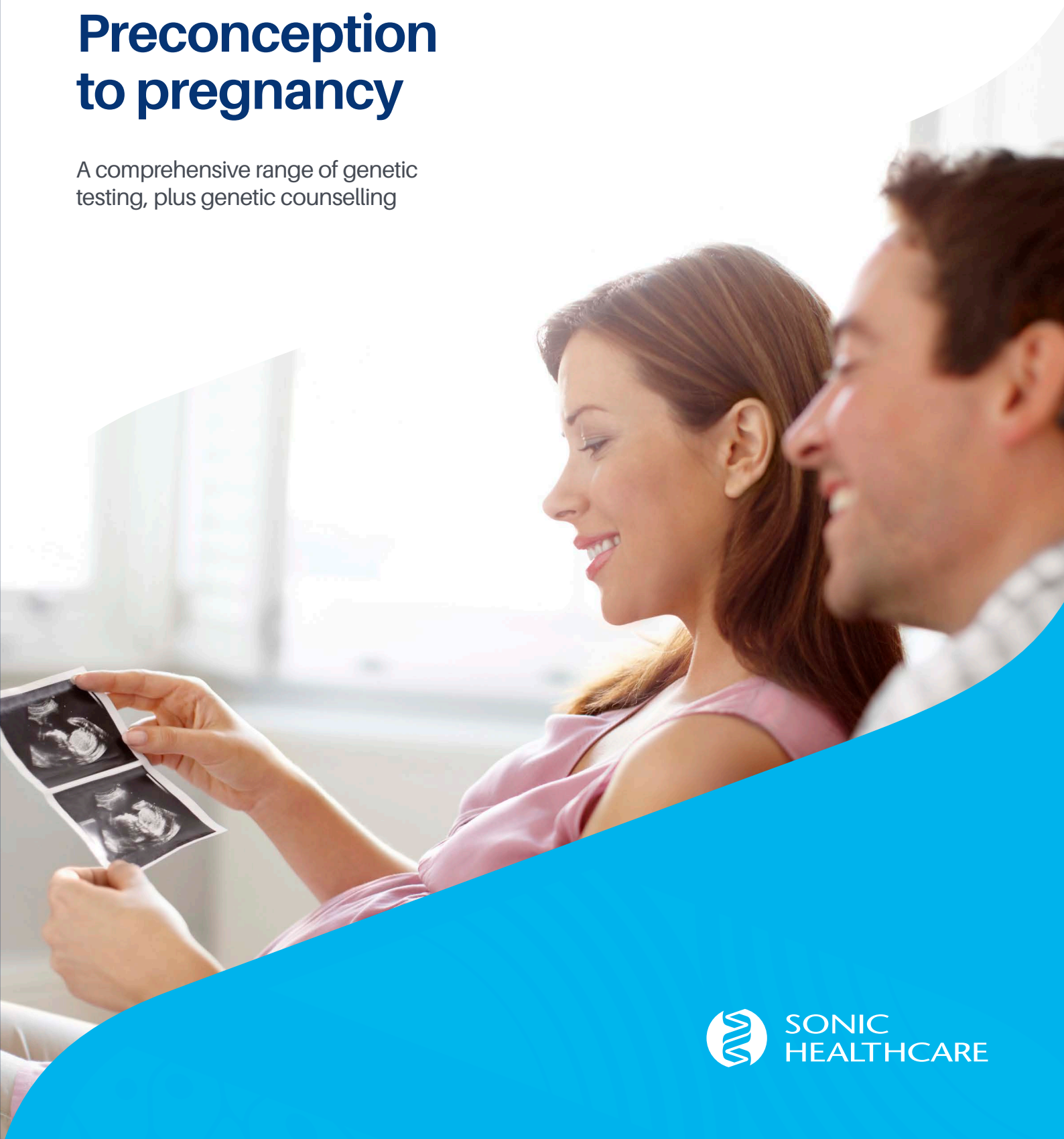
Sonic Genetics

Information for Doctors

Preconception to pregnancy

A comprehensive range of genetic testing, plus genetic counselling



SONIC HEALTHCARE

Preconception to pregnancy testing services

Sonic Genetics provides a comprehensive range of preconception and pregnancy genetic testing services throughout Australia. Our genetic pathologists and medical scientists have established national and international reputations in genomic testing.

Pre-pregnancy

Reproductive carrier screening

RANZCOG recommends that information about carrier screening for common recessive disorders be offered to every woman prior to conception (preferred) or in early pregnancy.[1]

Most children with a familial genetic disorder do not have a family history of that disorder. A test to screen for genetic carriers can provide information about the risk of a familial disorder that is not evident from reviewing the family history.

There are two types of reproductive carrier screening provided by Sonic Genetics:

- A three-gene panel that looks for mutations responsible for three common genetic disorders: cystic fibrosis, spinal muscular atrophy and fragile X syndrome. Approximately 1 in 20 people in Australia will be identified as carriers for one or more of these disorders, and 1 in 240 couples will be found to be at risk of having an affected child.
- The Beacon expanded carrier screen is a screening test that looks for mutations in genes for more than 400 disorders. Approximately 3 in 4 people will be identified as carriers for one or more disorders, and 1 in 20 couples will be shown to be at high reproductive risk of having an affected child.

An MBS rebate for 3-gene screening is being considered by Medicare.

First trimester

Non-invasive prenatal testing

Non-invasive prenatal testing (NIPT) is a cell-free DNA-based blood test that screens for trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome), and trisomy 13 (Patau syndrome) with greater accuracy than first trimester screening by ultrasound and maternal serum screening.[2] NIPT can also optionally screen for abnormalities in sex chromosomes, report fetal sex and screen for less common deletions and duplications of genetic material.

NIPT can be performed in conjunction with or after first trimester screening. Given the better performance of NIPT for detecting trisomies, we recommend that first trimester screening not be performed after NIPT.

- We report a result only if there is sufficient fetal DNA to be confident of accuracy. For patients with low fetal DNA, alternative test methods, such as serum screening or invasive diagnostic testing, may be more appropriate.
- The turnaround time is typically 3–8 days.
- For women who have a high-risk NIPT result, we provide follow-up cytogenetic testing on CVS or amniocentesis (rapid FISH test plus full karyotype) at no cost to the patient (Medicare rebated).

NIPT is not covered by Medicare. Please refer to our website for current pricing and options.

First & second trimester

Prenatal biochemical screening

The prenatal biochemical screening service provided through Sonic's Australian pathology laboratories tests more than a quarter of all pregnant women across the country, analysing more than 75,000 samples per year.

Our panels of feto-placental derived biochemical markers provide insight into feto-maternal wellbeing, beyond the major trisomies.

Our laboratories use a variety of platforms to provide assays of free βhCG, PAPP-A, αFP, unconjugated E3 and PLGF. Combinations of these assays and platforms are available through your local laboratory.

- Analytical results are provided as either lot-specific MoMs (multiples of the median) or concentration units for incorporation into combined risk assessment by interpretive software.
- The turnaround time is typically one business day after specimen receipt.
- Reports are available electronically by Sonic Dx, and/or by fax. Amendments and updates, especially crucial for MoM calculations, are available electronically or in real time.

These tests are rebated by Medicare and may incur an additional gap fee. Please refer to to your local Sonic pathology practice for current pricing and options.

Cytogenetics

Sonic Genetics is Australia's largest provider of cytogenetics testing, with reference laboratories in Melbourne (Melbourne Pathology) and Brisbane (Sullivan Nicolaides Pathology).

Our laboratories provide a comprehensive range of chromosome investigations for reproductive purposes utilising cytogenetic analysis (light microscopy) or microarray; this includes prenatal diagnosis, analysis of products of conception (POC) and preconception screening for couples experiencing recurrent miscarriages.

Cytogenetics is suitable for:

- Couples with recurrent miscarriages. Approximately 1 in 25 such couples has a balanced chromosome translocation in one or both parents. Balanced translocations are not detected by microarray.
- Invasive confirmation of a high-risk NIPT result for whole chromosome aneuploidy. The fetal karyotype should be assessed by cytogenetic analysis rather than microarray; microarray will not detect that a trisomy is due to a heritable translocation. Rapid testing for common trisomies is also available with a 24-hour turnaround.

Microarray is preferred for investigations of CVS or amniocytes in fetuses with structural abnormalities or after a miscarriage.

These tests are usually rebated by Medicare. Please refer to our website for current options.

Single nucleotide polymorphism (SNP) microarray

Chromosomal microarray is a whole-genome screening test that can detect losses and gains of chromosomal material. With its high resolution, it has a better diagnostic yield than conventional cytogenetic testing in the prenatal setting. For example, where a fetal abnormality has been detected by ultrasonography, microarrays detect an abnormality in an additional 5–7% of cases.

- Microarray testing is recommended for use in prenatal diagnosis in fetuses with one or more structural abnormalities identified on ultrasound.[1] This test replaces the need for fetal cytogenetics. It is also recommended for the analysis of POC, as it does not depend on the presence of viable cells.
- Microarray testing is not recommended when testing for a parental translocation, as it cannot detect a balanced rearrangement, that is, where there is no net gain or loss of chromosomal material. Similarly, it is not recommended when testing CVS or amniocytes for autosomal trisomy detected by NIPT. It can identify the trisomy but may not detect the rare instance of trisomy due to a heritable translocation.

This test is usually rebated by Medicare. Please refer to our website for current pricing and turnaround times.

Genetic counselling

Sonic Genetics encourages pre- and post-test counselling regarding the implications of a genetic test. This counselling may be provided by the doctor requesting the test or by a certified genetic counsellor.

We provide a link to a national list of genetic counselling services in the private and public sectors on our website (sonicgenetics.com.au/counsellingservices).

Sonic Genetics also provides free genetic counselling by telephone for women with a high-risk NIPT result and for couples identified as being at high reproductive risk by reproductive carrier screening (conditions apply).

This is managed as a medical referral from the patient's doctor; details of the process are provided in the reports of eligible patients.

1. Genetic carrier screening (C-Obs 63), March 2019, RANZCOG College Statements and Guidelines
2. Prenatal screening and diagnosis of chromosomal and genetic conditions in the fetus in pregnancy (C-Obs 59), July 2018, RANZCOG College Statements and Guidelines

SONIC HEALTHCARE

Sonic Genetics is Sonic Healthcare's centre of excellence in genetic pathology testing. As the largest pathology provider in the country, Sonic Healthcare's state-of-the-art laboratories and extensive network of collection centres serve each state and territory capital, as well as regional and rural Australia.

Douglass Hanly Moir Pathology
Sullivan Nicolaides Pathology
Melbourne Pathology
Barratt & Smith Pathology
Capital Pathology
Clinipath Pathology
Bunbury Pathology
Clinpath Pathology
Hobart Pathology
Launceston Pathology
North West Pathology
Southern.IML Pathology

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SHG-MKT-0008-00.7
Printed Oct 2021