



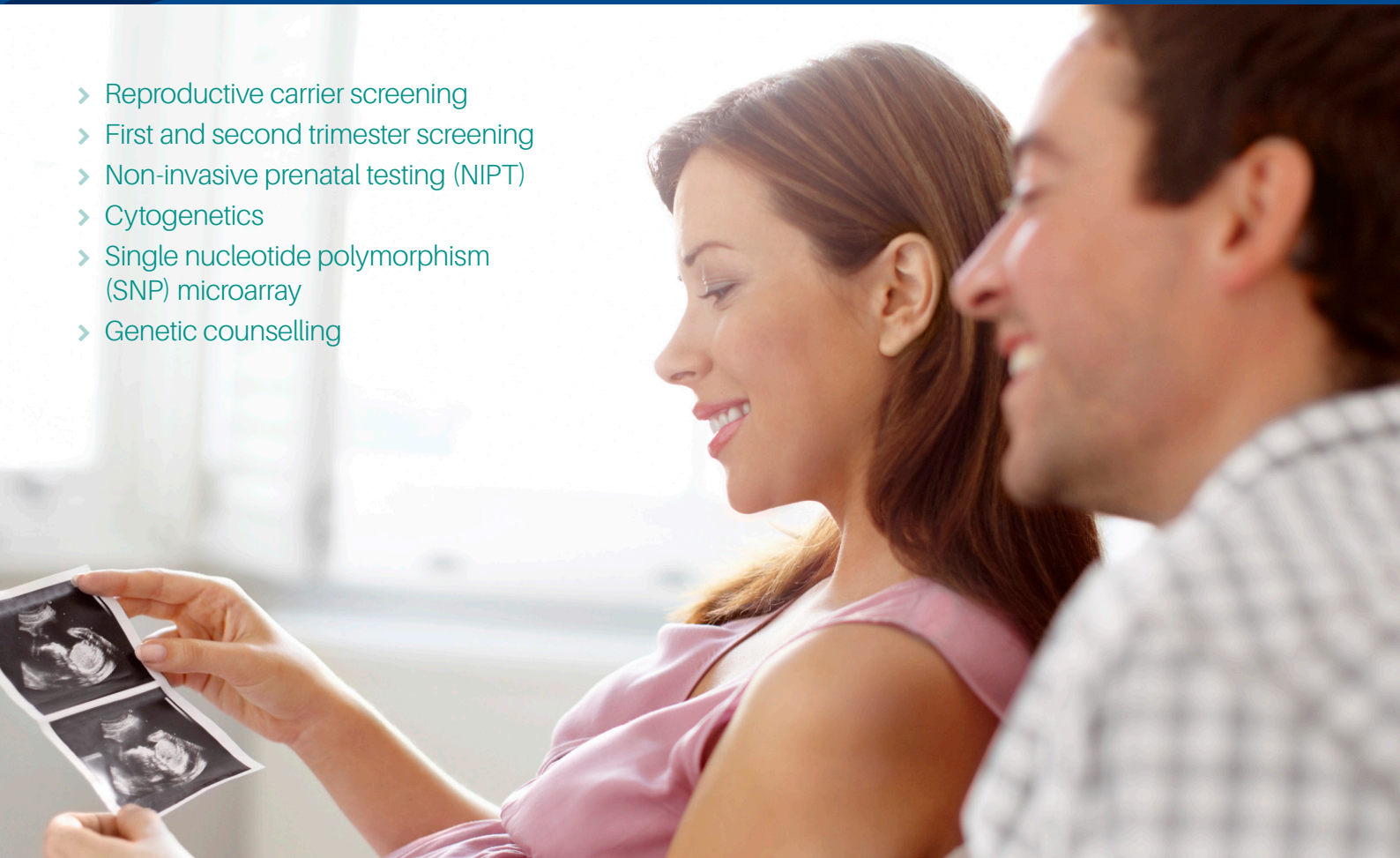
Sonic  
Genetics

Quality is in our DNA

# Genetic testing: Preconception to pregnancy

Information for Doctors

- › Reproductive carrier screening
- › First and second trimester screening
- › Non-invasive prenatal testing (NIPT)
- › Cytogenetics
- › Single nucleotide polymorphism (SNP) microarray
- › Genetic counselling



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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.

## Our Genetic Pathologists



**Professor Graeme Suthers**

BSc (Med), MBBS, PhD, FRACP, FRCPA, GAICD  
*Director, Genetics*  
*Sonic Pathology Australia*



**Dr Melanie Galea**

MBBS (Hons), BMedSc (Hons), FRCPA (Genetics)  
*Director, Genetics*  
*Douglass Hanly Moir Pathology*



**Dr James Harraway**

MB ChB, FRCPA, DPhil  
*Director, Genetics*  
*Sullivan Nicolaides Pathology*



**Dr Eric Lee**

MBBS, FRCPA  
*Genetic Pathologist*  
*Sullivan Nicolaides Pathology and*  
*Douglass Hanly Moir Pathology*

## First and second trimester screening



**Dr Michael Sinosich**

BSc, AIMLS, PhD, FFSRCPPA  
*Scientific Director/Department Manager*  
*Douglass Hanly Moir Pathology*



**Dr David Kanowski**

BSc (Hons), MBBS, FRCPA  
*Chemical Pathologist*  
*Sullivan Nicolaides Pathology*



**Dr Narelle Hadlow**

MBBS, MAACB, FRCPA, GAICD, AFCHSM  
*Chemical Pathologist*  
*Clinipath Pathology*

# Sonic Genetics preconception to pregnancy testing services

## Reproductive carrier screening

RANZCOG recommends that information about reproductive carrier screening for common recessive disorders be offered to every woman either prior to conception (preferred) or in early pregnancy.<sup>1</sup>

Whilst recognising the clinical and medico-legal significance of reviewing family histories, the reality is that most children with a familial genetic disorder do not have a family history of that disorder. The presence of a family history of a disorder is an important clue; the absence of such a history provides little information. 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 6% of people in Australia will be identified as carriers for one or more of these disorders, and one in 160 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 350 autosomal recessive disorders and more than 50 genes for X-linked recessive disorders. Approximately 70% of people in Australia will be identified as carriers for one or more of these disorders, and one in 20 couples will be found to be at high reproductive risk of having an affected child.

## First and second trimester screening

The prenatal biochemical screening service, through Sonic Pathology Australia laboratories, annually tests above one quarter of all pregnant women across the country, analysing more than 75,000 samples per year. Our panel of fetoplacental derived biochemical markers provides insight into fetomaternal wellbeing, beyond the major trisomies.

- Our laboratories use a variety of platforms to provide assays of free  $\beta$ hCG, PAPP-A,  $\alpha$ FP, unconjugated E3 and PLGF. Combinations of these assays and platforms are available through your local Sonic Pathology Australia 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.
- 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.
- Turnaround time is typically one working day after specimen receipt.





## 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), trisomy 13 (Patau syndrome) and optionally for 22q11.2 deletion (DiGeorge syndrome), with greater accuracy than conventional first trimester screening. NIPT can also screen for abnormalities in sex chromosomes and report gender.

NIPT can be incorporated with biochemical screening (sometimes known as contingent screening). Many peer-reviewed studies have documented the clinical utility of the NIPT.<sup>2</sup>

- All testing by Sonic Genetics is performed in Australia and not sent overseas. Our laboratories are NATA-accredited to perform this test.
- 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. 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) which is Medicare rebatable.
- Turnaround time is typically 5–8 days.

## Cytogenetics

Sonic Genetics is Australia's largest provider of cytogenetics testing, with reference laboratories in Melbourne (Melbourne Pathology) and Brisbane (Sullivan Nicolaides Pathology). They provide a comprehensive range of investigations including prenatal diagnosis, analysis of products of conception and preconception screening for couples experiencing recurrent miscarriages.

- The karyotype can be determined by cytogenetic analysis (light microscopy) or microarray. If a woman is at increased risk of having a chromosomally abnormal fetus (for example, fetal malformations on ultrasound), the fetal karyotype can be assessed by microarray (recommended) or cytogenetic analysis of CVS or amniocytes. Following a high-risk NIPT result, we recommend the fetal karyotype be assessed by cytogenetic analysis (microarray could miss the rare instance of trisomy due to a heritable translocation). Rapid testing for common trisomies is also available with a 24-hour turnaround.
- Most miscarriages are due to chromosome abnormalities that arise during the formation of the sperm or egg during early embryogenesis. They are not inherited and hence do not constitute a hazard in subsequent pregnancies. Many clinicians and couples wish to confirm this. The analysis can be done by microarray (recommended) or cytogenetic analysis of the products of conception.
- About 4% of couples with recurrent miscarriages have a balanced chromosome translocation in one or both parents that can cause miscarriages or congenital malformations in future pregnancies. If a potential parent is found to have a balanced translocation, we recommend genetic counselling. Preconception testing for a parental translocation should be done by cytogenetic analysis, not by microarray.



## 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.<sup>1</sup> This test replaces the need for fetal cytogenetics. It is also recommended for the analysis of products of conception, 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. It can identify the trisomy but may not detect the rare instance of trisomy due to a familial translocation.

## Genetic counselling

Sonic Genetics actively encourages pre- and post-test genetic counselling where clinically applicable. To help you support your patients, we provide a link to a national list of general genetics clinics on our website at [www.sonicgenetics.com.au/counsellingservices](http://www.sonicgenetics.com.au/counsellingservices). We can also direct patients to private genetic counselling services in their region. 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).



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 Genetics is part of Sonic Pathology Australia, the largest pathology provider in the country. Our 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  
Clinipath Pathology  
Hobart Pathology  
Launceston Pathology  
North West Pathology  
Southern.IML Pathology

For further information, please refer to our website,  
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