AUSTRALIAN Clinicallabs
Your experts in antenatal testing

How do Genetic Carrier Screening and NIPT differ?

Confused about the difference between Genetic Carrier Screening and Non-Invasive Prenatal Testing (NIPT)? Read below to learn how these two antenatal blood tests differ and why health experts recommend both during your pregnancy journey.

Genetic Carrier Screening

- *Ideally performed before conception to offer greater reproductive choice*
- *Uses the parent's DNA to screen for parental carrier status*

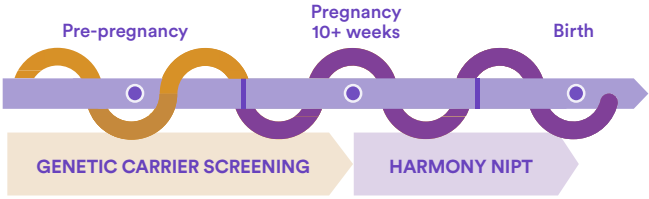
Genetic Carrier Screening analyses DNA in your blood to determine whether you are a genetic carrier for specific conditions. Australian clinical guidelines [1,2] recommend that doctors offer carrier screening to every woman or couple planning or in the first stage of pregnancy for common genetic conditions, such as Cystic Fibrosis (CF), Spinal Muscular Atrophy (SMA), and Fragile X Syndrome (FXS), regardless of their likelihood of having these conditions. 90% of carriers for these conditions have no family history.

- **CF:** A severe genetic condition that causes lung and gastrointestinal problems. ≈ 1 in 25 people are carriers. ≈ 1 in 2,500 people are affected.
- **SMA:** A neuromuscular disease historically associated with high morbidity and mortality. ≈ 1 in 35 people are carriers. ≈ 1 in 6,000 people are affected.
- **FXS:** The most common inherited form of intellectual disability, affecting ≈ 1 in 3,600 men and 1 in 6,000 women.

Carrier screening for CF, SMA, and FXS (Gene Access Carrier Screening) is available at Clinical Labs and is now 100% Bulk-Billed, subject to Medicare eligibility criteria.

1. The Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) Guidelines.
2. The Royal Australian College of General Practitioners (RACGP) Guidelines.

For more information on our Genetic Carrier Screening options, please visit antenatal.clinicallabs.com.au/patient/carrier-screening.



Harmony® NIPT:

- *Analyses fragments of baby's DNA in the mother's blood from 10+ weeks gestation*
- *Assesses the risk of fetal chromosomal abnormalities, such as Down Syndrome (trisomy 21)*
- *Can also determine the baby's gender*

Australian clinical guidelines (RANZCOG)[1] recommend that doctors provide all pregnant women with information about screening for Down syndrome.[1] Chromosomal conditions like Down syndrome, typically do not run in families and can occur in any pregnancy.

Clinical Labs is the exclusive Australian provider of Harmony NIPT, which screens for the following conditions:

- **Down syndrome (trisomy 21):** A common chromosomal condition occurring in ≈ 1 in 800 babies that is associated with mild to severe differences in physical and intellectual development.[2]
- **Edwards syndrome (trisomy 18), Patau syndrome (trisomy 13), and optional sex chromosome aneuploidies**
- **Optional 22q11.2 microdeletion (DiGeorge syndrome).** The most common genetic cause of heart defects and intellectual disability after Down syndrome.[3] Occurring in ≈ 1 in 1,000 pregnancies.[4,5]

Harmony NIPT is not Medicare-rebatable and costs $430. If ordered, 22q11.2 microdeletion costs an additional $70.

1. RANZCOG Guideline (C-Obs 59) July 2018.
2. U.S. National Library of Medicine. Genetics Home Reference. Down Syndrome. https://ghr.nlm.nih.gov/condition/down-syndrome. Accessed September 13, 2019.
3. McDonald-McGinn et al. Gnet Med.2001 Jan-Feb:3(1):23-9.
4. Grati et al. Prenat Diagn 2015;35:801-809.
5. Wapner et al. N Engl J Med 012;367:2175-2184.

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For more information about Harmony NIPT, please visit antenatal.clinicallabs.com.au/patient/harmony.

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