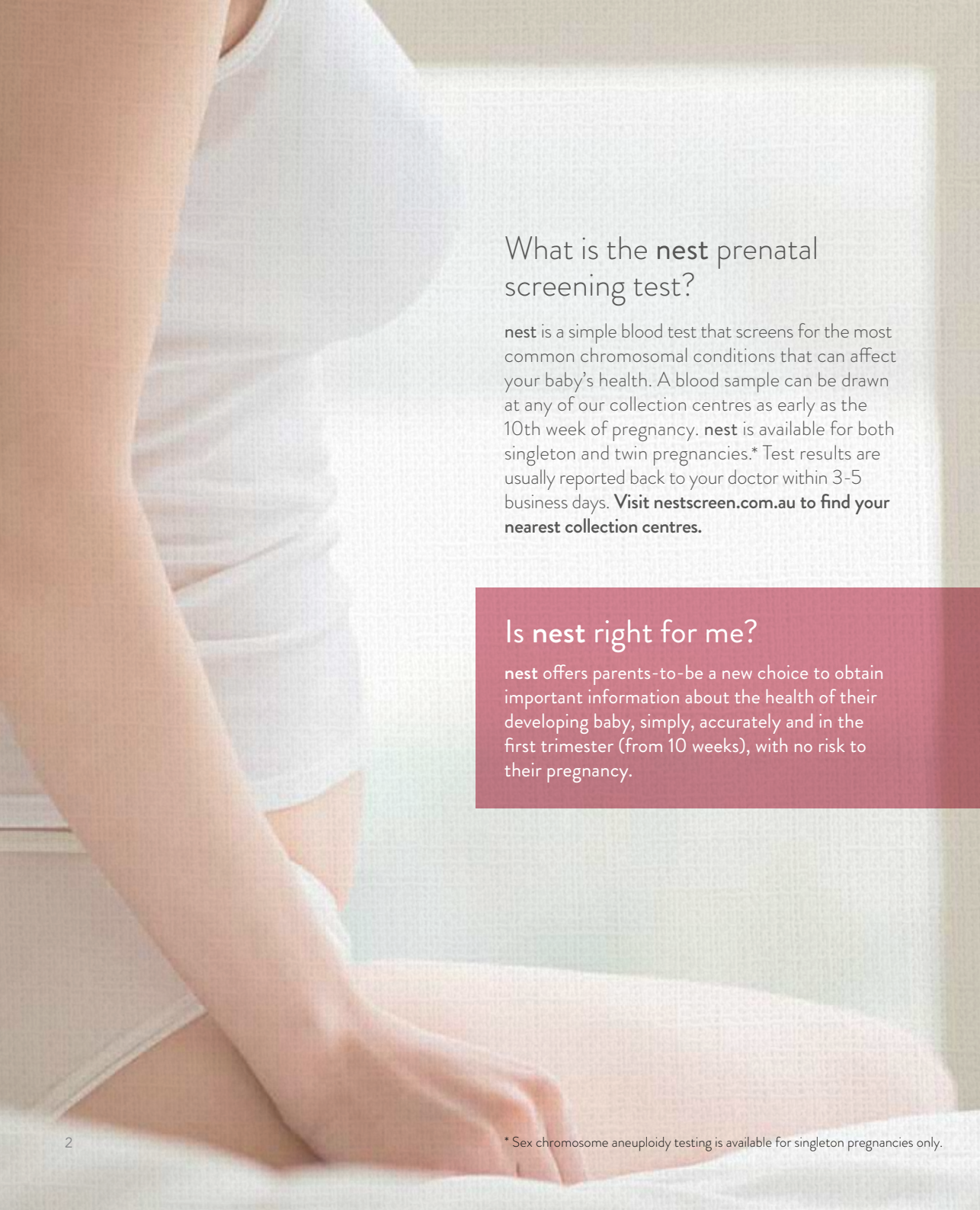
a guide for parents-to-be

nest

non-invasive early screening test



What is the **nest** prenatal screening test?

nest is a simple blood test that screens for the most common chromosomal conditions that can affect your baby's health. A blood sample can be drawn at any of our collection centres as early as the 10th week of pregnancy. **nest** is available for both singleton and twin pregnancies.* Test results are usually reported back to your doctor within 3-5 business days. **Visit nestscreen.com.au to find your nearest collection centres.**

Is **nest** right for me?

nest offers parents-to-be a new choice to obtain important information about the health of their developing baby, simply, accurately and in the first trimester (from 10 weeks), with no risk to their pregnancy.

* Sex chromosome aneuploidy testing is available for singleton pregnancies only.

How does the test work?

A sample of your blood is drawn and the genetic material (DNA) from you and your baby is tested. The **nest** screening is a more precise method, using an advanced technology called 'massively parallel sequencing' to analyse millions of DNA fragments per sample and accurately count the number of chromosomes present. It then uses a special SAFeR™ calculation method to determine if your baby has too many or too few copies of these chromosomes.

What specific conditions can **nest** screen for?

Chromosomes normally come in pairs. Most individuals have 23 pairs of chromosomes. **nest** looks for too few or too many copies of chromosomes.

Missing or extra copies of chromosomes can be associated with intellectual or physical disabilities, with differing levels of severity. The most commonly seen chromosomal conditions include trisomy 21 (Down syndrome), trisomy 18 (Edward syndrome) and trisomy 13 (Patau syndrome) all of which can be accurately detected with **nest**.

If your doctor chooses, **nest** can also be used to screen for sex chromosome conditions such as Turner syndrome (only one X chromosome in a female) or Klinefelter syndrome (an extra X chromosome in a male).

Other possible sex chromosome conditions are Triple X syndrome (an extra X chromosome in a female) and Jacob syndrome (an extra Y chromosome in a male). Men usually have an XY pair of sex chromosomes. Women usually have an XX pair of sex chromosomes. You can also elect to have gender reported to you.

What are my current testing options?

Today there are a number of genetic testing options available for expectant women and their healthcare providers. Some tests are called 'screening' tests and others are called 'diagnostic' tests.

Traditional screening tests are used to predict the chance of a pregnancy having certain chromosomal conditions. Results from screening tests usually provide a 'risk score' (such as 1 in 500 or 1 in 50) that describes the chance of a baby having a certain chromosome condition. They do not provide definitive answers and can have a relatively high rate of 'false negative' or 'false positive' results. But not all screening tests are the same in terms of accuracy and convenience.

Diagnostic tests, such as amniocentesis or chorionic villus sampling (CVS), can accurately determine whether a pregnancy has trisomy 21 (Down syndrome) or other chromosomal conditions. However, these procedures are invasive and carry a risk of miscarriage.

Why should I choose **nest** over other tests?

The **nest** screening test sheds much needed light on the chromosomal health of your unborn baby—providing the reassurance of reliable answers no other screening test can match. Compared to similar options, **nest** offers a highly precise chromosome screening, rather than calculating chances or risk scores. And, it does not carry the risk of miscarriage that an invasive procedure can.

SIMPLE

- **nest** uses a simple, single blood draw from your arm – just 1 tube of blood is all that is needed

CONVENIENT

- **nest** can be conducted conveniently at any of our collection centres – as early as during your 10th week of pregnancy

RELIABLE

- **nest** provides reliable answers about the most common chromosomal abnormalities

FLEXIBLE

- **nest** can screen for sex chromosome conditions if ordered by your doctor (not available for twin pregnancies). The gender of your baby can also be disclosed at your request.

ACCURATE

- **nest** has the lowest test failure in its class (0.1%)[1] - which means there is no need for additional blood tests or a delay in getting your results
- **nest** is able to detect fetal fraction which is the amount of cell-free DNA in the maternal blood that is of fetal origin, it is essential for accurate test results

FAST RESULTS

- **nest** delivers results fast - reports are usually sent to your doctor within 3-5 business days from receipt in our laboratory

What do my **nest** results mean?

Your results will tell your doctor whether or not trisomies 21, 18, 13 or sex chromosome conditions (if ordered) are likely to be present in your pregnancy. In the case of a high probability result, your doctor and genetic counsellor will discuss what the results mean to your pregnancy as well as further testing options to consider.

Your test report will include one of two possible results for chromosomes 21, 18, and 13:

- Low probability—means the expected number of chromosomes were found.
- High probability—means too many or too few copies of one of the chromosomes have been identified. This can indicate a chromosome condition. Diagnostic testing is recommended for confirmation.

If the sex chromosome option is ordered, results will be reported as either low probability or high probability. If gender is requested this will be labelled male or female.

nest genetic counselling

Highly trained **nest** genetic counsellors are available if requested by your doctor to explain this technology and how the results will be reported. They may also deliver your results if requested. Having a deep understanding of this technology gives them the skills to interpret these results and assist in explaining them. If the results do come back showing a high probability, the genetic counsellor can assist in explaining the next steps.

Do **nest** results mean that my baby will be perfectly healthy?

nest is a highly accurate advanced screening test that is non-invasive. No test, however, can guarantee a baby will not have any medical issues. **nest** only looks at chromosomes 21, 18, 13, and sex chromosomes*, if ordered. It does not test for or report all genetic and non-genetic problems that may be present in a baby. If the test result is low probability, it does not completely rule out all potential problems associated with chromosomes 21, 18 and 13, or all sex chromosome conditions in your baby.

Genetic counselling is available to all patients on request to assist in understanding what to expect from their results. Women who receive high probability results should be offered invasive prenatal procedures for confirmation.

A low probability result does not ensure an unaffected pregnancy. CVS and amniocentesis provide definitive diagnostic information, but the invasive nature of these procedures means that they are generally only offered to those who have been identified as a high risk pregnancy.

How do I know I can trust **nest** to be effective?

The performance of **nest** was evaluated in a major scientific study in which more than 60 leading US medical research and teaching institutions participated. The study findings were reviewed and published in the leading journal read by obstetricians and gynaecologists.[2] A second study, published subsequently, presented the test's performance under regular clinical conditions and found similar results.[3]

Fees information

The **nest** non-invasive prenatal screening is excellent value for patients. It is currently not covered by Medicare.

Visit nestscreen.com.au for current fee information.

* Sex chromosome aneuploidy testing is available for singleton pregnancies only.

To learn more about **nest** please visit **nestscreen.com.au** or phone **1800 874 971**

MONASH IVF GROUP

References
1. Bhatt S, Parsa S, Snyder H, Taneja P, Halks-Miller M, Seltzer W, DeFeo E. Clinical Laboratory Experience with Noninvasive Prenatal Testing: Update on Clinically Relevant Metrics. ISPD 2014 poster.
2. Bianchi DW, Platt LD, Goldberg JD, et al. Genome-wide fetal aneuploidy detection by maternal plasma DNA sequencing. Obstet Gynecol. 2012;119:890–901.
3. Futch T, Spinosa J, Bhatt S, de Feo E, Rava RP, Sehnert AJ. Initial clinical laboratory experience in noninvasive prenatal testing for fetal aneuploidy from maternal plasma DNA samples. Prenat Diagn. 2013;33:569-574.