Non-Invasive Prenatal Genetic screening (NIPS)

Canterbury Health Laboratories
www.chl.co.nz | 0800THELAB

Screening for genetic conditions is offered to all pregnant women because all pregnancies have a small chance of a genetic condition regardless of maternal age, family history or personal health. This is an optional screening test for trisomy 21 (Down syndrome), trisomy 18 and trisomy 13. You may wish to discuss your thoughts and how the results of prenatal screening options may affect your pregnancy with your maternity care provider.

What is Non-Invasive Prenatal Screening (NIPS)?

Non-Invasive Prenatal Screening (NIPS) is a screening test for trisomy 21 (Down syndrome), trisomy 18 and trisomy 13 performed on a blood sample taken from the mother and therefore this test poses no risk to the health of the mother or to the pregnancy. DNA from the fetus, circulating in the mother's blood, is measured to estimate the chance of the pregnancy having one of the conditions listed above. Whilst this test provides a better estimate than current, publicly funded first or second trimester screening tests it is still not 100% accurate. Like other screening tests, NIPS can miss a condition when it is present (false negative result) or can incorrectly show there is a high risk for a condition when it is not present (false positive result).

NIPS will detect 99 out of 100 (99%) pregnancies with trisomy 21, and therefore, a "Low risk" result means there is a very low chance of the pregnancy having the specified trisomy. A "Low risk" result does not, however, exclude the possibility the pregnancy has another genetic condition.

Whilst the false positive rate for NIPS is much lower than current, publicly funded screening tests, there are still times when a "High risk" result is given when these conditions are not present. The frequency of false positive results depends on the mother's age, previous history and/or results of any publicly funded screening test and ultrasound scan. For every 10 "High Risk" results, between 3 and 8 pregnancies will have a trisomy.

Who is suitable for NIPS?

NIPS can be performed from the 10th week of pregnancy. The gestational age must be confirmed by ultrasound scan. NIPS can be performed in twin pregnancies but the likelihood of not getting a result (a "No Call") is increased in this group (see below). Pregnancies using donated eggs or IVF can also be screened by NIPS. Previous transplant surgery, immunotherapy, stem cell therapy or recent (within 2 weeks) blood transfusion can affect the accuracy of the result and if you have had any of these treatments NIPS may not be suitable.

How long before I receive my NIPS results?

NIPS results are usually available within 10 days. Canterbury Health Laboratories will send your result to your maternity care provider.

How do I interpret my results?

A "**Low Risk**" result means the pregnancy is unlikely to have trisomy 21, 18 or 13, however it does not completely exclude the chance, this test will miss 1 out of every 100 pregnancies with trisomy 21.

A "**High Risk**" result indicates there is a high chance the pregnancy has a trisomy. The actual chance after the positive test depends on the mother's age, family history and any previous screening/ultrasound scan results. Typically for every 10 "high risk" results between 3 and 8 pregnancies will have a trisomy.

"**Resampling required**" - In a small number of cases (about 3%) not enough fetal DNA is present in the sample to provide a result, in this case a new blood sample is required to complete the screening.

"**No Call**" - In a smaller number of cases (<1%) the laboratory is unable to provide a result.

In the case of a "High Risk" or "No Call" result further diagnostic testing, such as by amniocentesis, is recommended.

How can I get more information and support?

Whilst this screening test may provide some reassurance about your pregnancy, it can also lead to questions such as: what does the result or diagnosis mean? How do I determine if the baby has this condition? What quality of life does a person with this diagnosis have? Where can I find reliable information about this condition? If you have questions of this nature, they should be discussed with your maternity care provider, or you may ask to be referred to a genetic counsellor.

Additional information

Twin pregnancy - twin pregnancies are classified as monochorionic (identical twins) or dichorionic (non-identical twins). In both identical and non-identical twin pregnancies a single risk estimate will be given for the pregnancy, this result must be interpreted in conjunction with ultrasound and other clinical information. In dichorionic twin pregnancies, because each twin contributes a lower percentage of DNA to the mother's blood the chance of a "No Call" is higher and may be up to 6%.

Further information about this test is available at

www.labnet.health.nz/testmanager

Follow the link and click on Non-invasive prenatal genetic testing