Non-Invasive Prenatal Testing Consent Form

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

Trisomy 21 (Down syndrome), Trisomy 18 and Trisomy 13, are three of the most common chromosomal abnormalities, and are usually due to the presence of one extra copy of chromosome 21, 18 or 13. The NIPS test is an optional screening test to assess the chance of these conditions being present in the fetus. This test is performed by measuring DNA from the fetus in the mother's blood from the 10th week of gestation. This test will detect 99 out of 100 (99%) pregnancies with trisomy 21, about 96% of trisomy 18 pregnancies and 91% of trisomy 13 pregnancies. The test can be used in singleton and twin pregnancies, pregnancies from IVF or those using donated eggs.

Limitations of the test:

1. Although this test will detect most pregnancies with trisomy 21, trisomy 18 or trisomy 13 it will not detect all cases, therefore a 'low risk' result does not completely rule out these conditions. The test will miss approximately 1 out of 100 fetuses with Down syndrome, 4 out of 100 fetuses with trisomy 18 and 9 out of 100 fetuses with trisomy 13. The test is also only designed to detect these conditions – a "low risk" result does not exclude other chromosomal or genetic abnormalities.
2. This test is very accurate but may indicate a condition is present when it is not (false positive). Typically for every 10 "high risk" results between 3 and 8 pregnancies will have a trisomy depending on the trisomy detected, mother's age, family history and any previous screening / ultrasound scan results. This is not a diagnostic test, "high risk" results require follow up testing in consultation with an obstetrician, usually by amniocentesis.
3. In some instances (<1 tests per 100 requested tests) a result cannot be provided by the laboratory. This may occur because there is insufficient fetal DNA present in the sample. The chance of this happening is higher in twin pregnancies (up to 6 tests per 100 twin pregnancies screened). Maternal chromosomal abnormalities including aneuploidies, mosaicism, microdeletion, and microduplication may compromise the accuracy of the NIPS test. 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.
4. Conditions other than trisomy 21, trisomy 18 or trisomy 13 may occasionally be detected, these include sex chromosome aneuploidy (Turner syndrome (XO), XYY, XXY), microdeletions, microduplications or maternal aneuploidy (either inherited or acquired).

Informed consent of the pregnant woman:

1. I have provided true and reliable personal information, and fully understand the indication, intended purpose and potential risks of this test. The test has been explained to me, and all my questions answered. I fully understand the limitations of this test (as outlined above).
2. I understand the report will be available within 10 days from the time the referral laboratory receives the sample. I understand that a repeat blood sampling (up to 3%) may be required due to insufficient concentration of fetal DNA, damage of the blood sample or technical failure.
3. I understand that the result cannot be used as the sole evidence for a diagnostic conclusion. Results from alternative examinations or tests should also be considered to make a final diagnostic determination.
4. I agree to provide the relevant information of this pregnancy, in particular if my baby is subsequently found with a chromosomal or genetic disease. I understand and agree that my clinician may contact me for such information. I agree to the use of my clinical information by my clinician and/or the laboratory for the purpose of auditing, quality assurance and research provided that I remain anonymous and unidentifiable during data analysis and that all my personal information are removed from any reports or publications.

My signature below acknowledges my voluntary participation in this testing, but in no way releases the laboratory and staff from their professional and ethical responsibility to me.

☐ I **consent** to the use of my DNA sample for research purposes, or

☐ I **do not** consent to the use of my DNA sample for research purposes.

☐ **GENDER IDENTIFICATION** Please tick if you WOULD like to know fetal gender.

Medical Practitioner/Counsellor Statement

I have explained the potential clinical utility for the requested molecular test to this individual. I have determined that this is a screening test for chromosomal aneuploidies. I have discussed genetic testing with them, presented the information outlined above, discussed the options, have addressed the limitations outlined above, and I have answered this individual's questions. Results from these tests may be used to direct medical management.

Signature: …… Print Name: …… Date: ……/……/……

Patient Statement

I agree to the genetic analysis, and I have had the opportunity to ask questions about the testing.

Patient's Name (PRINTED): …… Patient's Date of Birth: ……/……/……

Patient's Signature: …… Date Signed: ……/……/……

This form should be accompanied by a completed laboratory request form.