PATERNITY For Life

NIFTY®

Accredited for compliance with NPAAC Standards and ISO 15189.

NON-INVASIVE PRENATAL GENETIC SCREENING FOR FETAL CHROMOSOMAL ANEUPLOIDIES

PATIENT INFORMATION

Surname:		DOB: ____ /_____ /_____
First Name:		Weight (Kg): Height (cm):
Address:	Phone:	Email:

REQUESTED SCREEN

NIFTY™ Plus

- T21, T18, T13 & specific sex chromosome abnormalities plus additional testing for other trisomies and micro-deletion syndromes (please see website for full list)

GENDER IDENTIFICATION

☐ Please tick if you WOULD like to know fetal gender

REFERRING DOCTOR INFORMATION

Name:

Practice:

Phone number:

Practice Address:

GP Email (for results):

MEDICAL PRACTITIONER STATEMENT

By signing this request form, I confirm that I have patient consent for the NIFTY™ Plus screen to be performed and confirm that the patient understands the purpose, scope and performance of the screen explained by either myself, available patient literature or the Paternity For Life website. I confirm that the patient understand that NIFTY™ Plus is intended to screen for fetal trisomy 21, 18 and 13 for both singleton and twin pregnancy; that it is also possible to use NIFTY™ Plus to screen for specific sex chromosome aneuploidies in singleton pregnancy and identify fetal gender; that NIFTY™ Plus may be used to screen for additional chromosomal aneuploidies such as micro-deletion syndromes; that the results of the screen will be sent to and reviewed by a nominated doctor; that a "high risk" result should be confirmed by a definitive diagnostic test (e.g. amniocentesis or chorionic villus sampling); that repeat blood sampling may be required; and that a small percentage of screens do not yield a result due to biological factors. The patient understands that they can request further information or genetic counselling via Paternity For Life or an alternate health practitioner. The patient understands that their clinical information may be used for the purpose of auditing, quality assurance and research on the basis that they remain anonymous and unidentifiable during data analysis. The patient understands that none of their personal information will be used in any reports or publications.

Name: _____

Signature: _____

Date: ___ / ___ / _____

CLINICAL INFORMATION *Please complete all fields

Gestational Age: _____ weeks _____ days As At date: ____ / ____ / _____

Estimated Due Date: ____ / ____ / _____

IVF: ☐ No ☐ Yes - Implantation Date: ____ / ____ / _____

No. of Fetus: ☐ Singleton ☐ Twin

If Twin: ☐ DCDA ☐ MCDA ☐ MCMA ☐ Unsure

- Please note that some additional analyses (such as sex chromosome abnormalities, Trisomies other than T21, T18 and T13, and micro-deletion syndromes) may not be available for twin pregnancies

PRIOR DOWN SYNDROME SCREENING TEST

☐ No

☐ Yes – estimated risk of T21: 1/ _____, T18: 1/_____, T13: 1/_____

FAMILY HISTORY OF GENETIC DISEASES

☐ No

☐ Yes – Please specify: _____

_____

_____

PLEASE TURN OVER FOR ADDITIONAL INFORMATION

Collection Staff Only		Laboratory Use Only		
Date of Collection:	Collector's signature:	Date of Receival:	Collection tube type:	Signature
Time Of Collection		Time of Receival:	Gestational Age Check:	Comments:

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PATIENT INFORMED CONSENT STATEMENT

Trisomy 21, Trisomy 18 and Trisomy 13, are three of the most common chromosomal abnormalities in newborns and is due to the presence of one extra copy of chromosome 21, 18 or 13. The NIFTY™ test assesses the risk of fetal "Chromosomal Aneuploidies", by detecting fetal chromosomal material with the new generation of high-throughput sequencing technology coupled with advanced bioinformatics analysis. This test is non-invasive, with no risk of causing miscarriage and intrauterine infection, and is highly sensitive, with an accuracy rate of over 99%.

Limitations of the test:

1. This test is intended to detect fetal Trisomy 21, 18 and 13 for both singleton and twin pregnancies. It is also possible to use this test to discover other chromosomal aneuploidies in singleton pregnancies such as XO and other sex chromosomes aneuploidies. This test is highly accurate, with a detection rate over 99% and a false positive rate of less than 1% for fetal Trisomy 21, 18, and 13, however, this test is not a diagnostic test, and the positive results should be confirmed by a diagnostic procedure such as karyotyping. Due to the limitation of the current technology, a negative result cannot totally exclude the possibility of fetal trisomy.
2. If the test is performed at very early stage of pregnancy stage (<10 gestational weeks), an uninformative result could be produced because of an inadequate amount of fetal chromosomal material. The following situations may compromise the accuracy of NIFTY™ test: maternal chromosomal aneuploidies, mosaicism, chromosome microdeletion, microduplication and multiple pregnancies. If the pregnant women has received an allogeneic blood transfusion, transplantation or stem cell therapy, there will be a possibility of misleading results because of exogenous DNA. Results should be interpreted in the context of other clinical and family information.

Supplementary Information about NIFTY™Plus:

1. NIFTY™ Plus will test for additional autosomal trisomies (Trisomy 9, Trisomy 16 and Trisomy 22) and the >10Mb chromosomal deletions which may cause Cri-du-chat (cat's cry) syndrome, also known as 5p- (5p minus) syndrome, 1p36 deletion syndrome, 2q33.1 deletion syndrome, DiGeorge Syndrome 2 (deletion of chromosome 10p14-p13), 16p12.2 deletion syndrome, Jacobsen Syndrome, Van der Woude Syndrome 1, and Prader-Willi and Angelman syndromes. Our data showed that >95% of >10Mb deletion findings are subsequently confirmed. However, the detection rate for chromosomal deletions is unknown at present. Hence, if there is strong clinical suspicion of chromosomal deletions, a conventional diagnostic test should be used instead of the NIFTY™ Plus test.

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. I have received information and had the opportunity to ask questions. I fully understand the limitations of this test, in particular
    a. this test is intended for the detection of Trisomy 21, 18 and 13, detection of other chromosomal aneuploidies in singleton pregnancy such as XO and other sex chromosomes aneuploidies, microdeletion and duplication testing is available as NIFTY™ Pus,
    b. the detection rate for Trisomy 21, 18 and 13 is close to but is not 100%, the detection rate for Monosomy X is over 90%, the detection rate for other sex chromosomal aneuploidies is undetermined.
2. I understand that the test may also include an estimation of the percentage of Y chromosome for a singleton pregnancy. Such information will be used for research and cannot be used to diagnose the sex-linked diseases.
3. I understand that the high sensitivity and specificity of the NIFTY™ test are based on studies in singleton pregnancies. Performance assessment in twin pregnancies by NIFTY™ is still in progress and similar clinical data on twin pregnancies are limited. Based on reported studies so far, and on theoretical grounds, the performance of NIFTY™ in twin pregnancies is similar to that in singleton pregnancies. However, this requires further confirmation by larger studies. For twin pregnancies only T21, T18 and T13 risk assessment results are available from the 12th week of gestation. If the result is "High Risk", it is suggested that at least one of the twin fetuses is at high risk, further diagnostic tests for both fetuses are needed. If the result is "Low Risk", there is a 99% possibility that both fetuses are at low risk for T21, T18, and T13.
4. I understand that the report will be available within 3 weeks from the time the laboratory receives the sample, but in 90% of cases the report will be available within 1 week. 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.
5. 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.
6. 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.
7. In some situations, a sample may produce no result. **No refund will be provided in the case of a NIL result.** Reasons for NIL results include:
    a. The test will not produce any result in approximately 2% of patients. This is more commonly seen in obese patients (>110 kg), older patients and twin pregnancies
    b. Repeating the test may provide a result in up to 50% of cases, but repeat test failure is common in these patients.
    c. A NIL result may be associated with an increased risk a fetal aneuploidy, and an alternative screening test or diagnostic test is recommended for all patients with NIL results.

Supplemental terms for women at late pregnancy (>24 weeks):

1. I understand there exist certain risks in late pregnancy (>24 weeks) because the ideal time for prenatal diagnosis has been missed. I agree to take the NIFTY™ test and I will take responsibility for any risks due to the fact that I cannot take a clinical diagnostic test to confirm the results. I agree to take this test for the prenatal detection of fetal Trisomy 21, Trisomy 18 and Trisomy13.