INFORMED CONSENT

NIFTY™
BGI technology inside

Sample Barcode

NIFTY test for Fetal Trisomy 21, Trisomy 18, and Trisomy 13

Introduction of disease and detection method:

Trisomy 21, Trisomy 18 and Trisomy 13, are three of the most common chromosomal abnormalities, usually 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 materials 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 pregnancy. It is also possible to use this test to discover other chromosomal aneuploidies **in singleton pregnancy** such as XO and other sex chromosomes aneuploidoes, sub-chromosomal deletion and duplication (concluded as **Additional Findings**). 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 the diagnostic procedures 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 pregnancy stage (<10 gestational weeks), uninformative result could be produced because of inadequate amount of fetal chromosomal materials. The following situations may compromise the accuracy of NIFTY test: maternal chromosomal aneuploidies, mosaicism, chromosome microdeletion, microduplication and multiple pregnancies. If the pregnant women have received allogeneic blood transfusion, transplantation or stem cell therapy, there will be a possibility of misleading results because of exogenous DNA.

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. My doctor has explained the test to me, and answered all my questions. I fully understand the limitation of this test, in particular i) 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 aneuploidoes, sub-chromosomal deletion and duplication are available., ii) 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 aneuploidy is undetermined.

2. I understand that the test may also include an estimation of percentage of Y chromosome for 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 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 pregnancy only T21, T18 and T13 risk assessment results are available from 12th week of gestation.

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 2 weeks. I understand that a repeat blood sampling (up to 3%) may be required due to insufficient concentration of fetal DNA, damage of blood samples or abnormal experiment procedures.

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. I agree that BGI will act on my behalf for handling insurance procedures with PICC Health Insurance Company Ltd. Shenzhen Branch. If the test result is “low risk”, and my baby has been diagnosed with Trisomy 21, 18 or 13 by hospital specialists within one year of my test, the insurance company will compensate me with RMB￥200,000 (or equivalent). If my test result is high risk for T21, T18 or T13, PICC will reimburse me for the cost of further confirmatory diagnostic tests including amniocentesis, CVS and chromosome fluorescence in situ hybridization (FISH). The reimbursement will be RMB￥2,500 (or equivalent) maximum.

I agree to take this test for the prenatal detection of fetal Trisomy 21, Trisomy 18, Trisomy13.

Name(In capital) :_____ Signature:_____ Date(DD/MM/YYYY):_____

Witness

Name(In capital): DR. _____ Clinic/Hospital:_____Signature:_____

Date(DD/MM/YYYY):_____

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

I understand there exist certain risk at late pregnancy (>24 weeks) because I miss the ideal time for prenatal diagnosis. I agree to take NIFTY test and I will take responsibility to all the risks due to I cannot take a clinical diagnostic test to confirm the results.

Name(In capital) :_____ Signature:_____Date(DD/MM/YYYY):_____

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