NTD | Eurofins Clinical Diagnostics

Patient informed consent

INTRODUCTION: This form describes the benefits, risks, and limitations of this screening test. You should seek pre-test counseling by a genetic counselor or other experienced health care provider prior to undergoing this test. Read this form carefully – and ask any questions you may have of your health care provider -- before making your decision about testing.

PURPOSE: The purpose of the Verifi™ Test and the Verifi™ Plus Test is to screen your pregnancy for certain chromosomal abnormalities, also known as "aneuploidies." Both tests give information about whether there may be extra copies (trisomy) of chromosomes 21, 18, and 13, and the option to know if there is an extra copy of a sex chromosome (X or Y), and/or a missing copy of sex chromosome (thMX). Fetal sex may also be reported. The Verifi™ Plus Test has the option to screen for aneuploidies (extra copies) in all chromosomes. In addition, the option to screen for the following microdeletions (small, missing parts of chromosomes) syndromes: 1p36 deletion, 4p- (Wolf-Hirschhorn syndrome), 5p- (cri-du-chat syndrome), 15q11.2 (Prader-Willi syndrome/Angelman syndrome), 22q11.2 deletion (DiGeorge syndrome or velocardiofacial syndrome) is also available. For chromosomes 21, 18, and 13, the Verifi™ Test is validated in singleton and twin pregnancies. In twin pregnancies, sex chromosome testing can only screen for the presence or absence of the Y chromosome, and not for extra or missing sex chromosomes. Both Verifi™ and Verifi™ Plus can be performed as early as 10 weeks 0 days gestational age. Consult your health care provider if you would like more information about this screening test, including risks, limitations, performance data, error rates, descriptions of the conditions being screened, and what these results may mean to your pregnancy.

HOW THIS TEST WORKS: This test screens for specific chromosomal abnormalities by looking at the DNA (genetic material) in your blood. The sample of blood includes a combination of both your DNA and the DNA from the pregnancy. A technology called massively parallel sequencing is used to count the amount of DNA from each test chromosome and/or from specific regions of chromosomes. The laboratory then uses an analysis method to determine if each of the conditions you have elected to test for is likely to be present or absent.

SEX OF PREGNANCY: Depending upon the option you and your health care provider elect, the test results may include the sex of the pregnancy. If you do not wish to know the sex, please tell your health care provider not to disclose this information to you. Depending upon the test ordered, you may not be able to prevent learning the sex of your pregnancy. In rare instances, incorrect sex results can occur.

LIMITATIONS OF THE TEST: These are screening tests that look only for specific chromosomal abnormalities. This means that other chromosomal abnormalities may be present and could affect your pregnancy. A "No Aneuploidy Detected" result does not guarantee a healthy pregnancy or baby and does not eliminate the possibility that your pregnancy may have birth defects, genetic conditions, or other conditions, such as open neural tube defects or autism.

There is a small possibility that the test results might not reflect the chromosomes of the fetus, but may reflect chromosomal changes of the placenta (confined placental mosaicism, CPM) or of you (maternal chromosomal abnormalities). While these tests are not designed to assess your health, in some cases, information about your health may be revealed directly or indirectly (e.g., when combined with other information). Examples include maternal XXX, sex chromosome status or benign or malignant maternal neoplasms. In a twin pregnancy, the status of each individual fetus cannot be determined.

These tests, like many tests, have limitations, including false negative and false positive results. This means that the chromosomal abnormality being tested for may be present even if you receive a negative result (this is called a 'false negative'), or that you may receive a positive result for the chromosomal abnormality being tested for, even though the abnormality is not actually present (this is called a 'false positive').

In the case of a twin pregnancy, the presence or absence of Y chromosome material can be reported. The occurrence of sex chromosome aneuploidies cannot be evaluated in twin pregnancies. In the case of a vanishing twin, the test result may reflect the DNA of the vanishing twin, leading to a higher probability of false positive or false negative results.

No irreversible clinical decisions should be made based on these screening results alone. If definitive diagnosis is desired, chorionic villus sampling or amniocentesis would be necessary. In some cases, other testing may also be necessary. Some rare chromosomal aneuploidies may only occur in mosaic form. Clinical consequences depend on the chromosome involved and cannot be predicted prenatally.

Consult your health care provider for more information about your results and what they may mean for your pregnancy, what options you will have for further testing, and whether additional testing is recommended for you based on your clinical history.

TEST PROCEDURE: A tube of your blood will be drawn and sent to Verinata Health, Inc., a wholly owned subsidiary of Illumina, Inc., which will then analyze your blood.

PHYSICAL RISKS: Side effects of having blood drawn are uncommon, but may include dizziness, fainting, soreness, bleeding, bruising, and, rarely, infection.

DISCRIMINATION RISKS: Genetic information could be used as a basis of discrimination. To address concerns regarding possible health insurance and employment discrimination, some countries, U.S. states, and the U.S. government have enacted laws to prohibit genetic discrimination in those circumstances. The laws may not protect against genetic discrimination in other circumstances, such as when applying for life insurance or long-term disability insurance. Talk to your health care provider or genetic counselor if you have concerns about genetic discrimination prior to testing.

PREGNANCY OUTCOME INFORMATION: Collecting information on your pregnancy after testing is part of a laboratory's standard practice for quality purposes and is required in several states. As such, Illumina or its designee may contact your health care provider to obtain this information. By executing this informed consent, you agree to allow your health care provider to provide this information to Illumina or its designee.

SECONDARY FINDINGS: In the course of performing the analysis for the indicated tests, information regarding other chromosomal alterations, also known as "secondary findings" may become evident. Our policy is to NOT REPORT on any secondary findings that may be noted in the course of analyzing the test data.

PRIVACY: Test results are kept confidential. Your test results will only be released in connection with the testing service, to your health care provider, his or her designee, other health care providers involved in your medical care, or to another health care provider as directed by you (or a person legally authorized to act on your behalf) in writing, or otherwise as required or authorized by applicable law.

CROSS-BORDER DATA TRANSFER: If you are from outside the United States, your specimen and associated health information will be sent to the United States in order for the testing to be completed. As part of the testing, additional health information about you will be created and maintained. Your country may consider the legal privacy protections in the United States to be inadequate.

USE OF INFORMATION AND LEFTOVER SPECIMENS FOR RESEARCH: Eurofins NTD, LLC performs research to help develop and provide safe and effective screening tests and to contribute to advancing biomedical knowledge. Your sample will be discarded within sixty days after taken, unless you specifically give consent. Permission to allow the use of your de-identified sample in research and development studies is entirely voluntary.

TEST RESULTS: Your test results will be sent to the health care provider.

PATIENT CONSENT: By signing this form, I, the patient having this screening performed, acknowledge that: (i) I have been offered the opportunity to ask questions and discuss with my health care provider the benefits, risks, and limitations of the test to be performed; (ii) I have discussed the test limitations (reliability of positive and negative test results; the predictive value of the test results; and that the test is not a diagnostic test, but a screening test and is not definitive) with the health care provider who ordered the test; (iii) I have been informed about the availability and importance of genetic counseling and have been provided with information identifying an appropriate health care provider from whom I might obtain such counseling; (iv) I have received, read, and understood the Patient Informed Consent in its entirety and that I may retain a copy for my records (v) I consent to having this test performed; and (vi) I will discuss the results and appropriate medical management with my health care provider.

Patient Signature: **Date:**