REP-2-020 Attachment D- Patient Informed Consent for Y-Chromosome

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Review and Approval	• Reviewers: Jonathan Hayden; Christina Deer; Lisa Schmitt • Approver (Laboratory Director Only): Terrence Hallahan
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Revision Log

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Aug 1, 2018	1	Eurofins D CDS US Quality Management Department; Eurofins D CDS US Reporting Department; Eurofins D CDS US Laboratory Management	New Test Offer

Electronic Signatures

Christina Deer;Review;Jul 2, 2018 4:03 PM EDT
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Lisa Schmitt;Review;Jul 11, 2018 10:40 AM EDT
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Patient Informed Consent for Y Chromosome Assessment

PLEASE NOTE: A signed Physician Acknowledgement of NY Informed Consent for Genetic Testing must be on file at Eurofins NTD, LLC to permit testing and processing.

I, (Patient name)_____, voluntarily request of Eurofins NTD, LLC to perform Y Chromosome Assessment DNA-based testing as ordered by my physician in an attempt to determine fetal sex for the purpose of adjusting my maternal serum screening analytes. My physician has obtained my informed consent for this testing, including the following information, which I have read and understand:

- This analysis tests for the presence of Y chromosome-specific DNA in maternal blood samples. There are two sex chromosomes in humans, the X and Y chromosomes. Females have two X chromosomes while males have one X and one Y chromosome.
- This test has been ordered because fetal sex is a parameter that has been found to alter the analysis results of maternal serum screening markers. The current optimum screening practice for the detection of fetal chromosomal anomalies is the first trimester measurement of maternal serum markers combined with fetal ultrasound markers. Detection of Y chromosome allows for the appropriate corrections to each maternal serum screening analyte in order to calculate more accurate patient-specific risk.
- This analysis can have the following outcomes:
  - **Positive:**
    Y chromosome DNA was detected in the mother's blood sample and therefore a male fetus is present in the current pregnancy.
  - **Negative:**
    Y chromosome DNA was not detected in the mother's blood sample and therefore most likely (98%), a female fetus is present in the current pregnancy and the maternal serum screening test will be interpreted as such.
    Note: 2% of negative results may occur because the amount of fetal DNA in the mother's blood was below the limit of detection of this DNA test.
  - **Inconclusive:**
    The sample failed to generate enough material to provide an accurate result. The mother has the option to resubmit another blood sample.
- Sensitivity and specificity of the detection of Y Chromosome DNA at 10 weeks of gestation for properly collected maternal blood samples is 98.7% and 96.4% respectively. The amount of fetal DNA in the maternal blood is very low. Please note that if the sample is collected after 10 weeks of pregnancy then the technique is usually sensitive enough to detect the small amount of fetal DNA in mother's blood. Approximately 2% of the samples have fetal DNA fraction that is lower than the detection limit.
- Y Chromosome Assessment requires a blood sample, which has risks associated with obtaining the sample. A second blood sample for the analysis of maternal serum markers is also required to complete your prenatal risk assessment. Additional samples may be needed if the samples are damaged in shipment or inaccurately submitted.
- As with any complex genetic test, there is always a small possibility of a failure or error in sample analysis. Extensive measures are taken to try to avoid these errors. The methods are not 100% accurate due to the possibility of rare genetic variations in the DNA of an individual or due to the complexity of the testing itself. Possible diagnostic errors include sample mix-ups, rare genetic variants that interfere with analysis, or other sources of interference. A low error rate, approximately 1 in 1000 samples, is generally estimated to exist in a laboratory.
- It is the responsibility of the referring physician or health care provider to understand the specific use and limitations of the testing ordered, including the above referenced information, and to educate the patient regarding these limitations.
- Due to the complexity of DNA testing and potential implications of test results, results will be reported directly to the patient's ordering provider, who will then review and discuss the test results with me.
- Patient-identifying results and information at Eurofins NTD, LLC will remain confidential and may only be released to other parties with my expressed written consent or as permitted or required by applicable law.
- I understand no tests other than those authorized shall be performed on my sample and that the sample shall be discarded within sixty days after being taken. However Eurofins NTD, LLC performs research and development studies to improve and to validate existing and new tests and to advance biomedical knowledge. I and my heirs will not receive payments, benefits, or

rights to any resulting products or discoveries. Patient permission is requested for the use of patient de-identified sample in research and development studies and is entirely voluntary.

- If I have additional questions, I understand that I may wish to obtain further professional genetic counseling prior to consenting to this testing.

My signature below acknowledges my voluntary participation in the genetic tests ordered by my physician and verifies that I have been appropriately counseled about the testing process and the different possible outcomes.

_____	_____	_____
Patient/Guardian Signature	Printed Name	Date

Physician/Counselor/Clinician Statement:

I have explained Y Chromosome Assessment to the patient/parent/guardian. The consent form and limitations of genetic testing were reviewed with the patient/parent/guardian. I accept responsibility for pre- and post-test genetic counseling. I will use my independent professional judgment and the patient's best interests in advising the patient/parent/guardian regarding DNA test results, the use and limitations of same, and any research study, clinical trial, drug, treatment or device brought to my attention by Eurofins NTD, LLC or others.

_____	_____	_____
Healthcare Provider Signature	Printed Name	Date

A signed physician acknowledgement of NY informed consent for genetic testing must be on file at Eurofins NTD, LLC to permit testing and processing.

This Patient Informed Consent form is to be maintained by the physician in the patients' medical file. DO NOT Return to Eurofins, NTD, LLC.