NIPD GENETICS

VERAgene® comprehensive NIPT

PATIENT INFORMATION		REFERRAL INFORMATION	
NAME		CLINIC NAME **Center of Medcine**	
ID NUMBER		CLINIC ID **104641**	
DATE OF BIRTH (DD/MM/YYYY)	GESTATIONAL AGE **Week: Day:**	REFERRING CLINICIAN **Ekaterina Venskevich**	
IVF STATUS **No**	NUMBER OF FETUSES **One**	CLINIC FAX	
SAMPLE INFORMATION			
ORDER NUMBER	LAB NUMBER	DATE OF COLLECTION (DD/MM/YYYY) **28/06/2022**	DATE RECEIVED (DD/MM/YYYY) **04/07/2022**

VERAgene PRENATAL SCREENING TEST RESULTS

VERY LOW RISK NIPT Results

FETAL FRACTION

8.7%

CONDITION	REMARK
Trisomy 21	The results show very low risk for trisomy 21
Trisomy 18	The results show very low risk for trisomy 18
Trisomy 13	The results show very low risk for trisomy 13
Trisomy X	The results show very low risk for trisomy X
Monosomy X	The results show very low risk for monosomy X
XXY Constitution	The results show very low risk for XXY constitution
XYY Constitution	The results show very low risk for XYY constitution
XXYY Constitution	The results show very low risk for XXYY constitution
Microdeletions: (DiGeorge,1p36 deletion syndrome,Smith-Magenis, Wolf Hirschhorn)	The results show very low risk for microdeletions (DiGeorge (22q11.2), 1p36 deletion syndrome, Smith-Magenis (17p11.2), Wolf Hirschhorn (4p16.3))
Panel of 100 single gene diseases	The results show very low risk for the panel of 100 monogenic diseases
Presence/Absence of Y Chromosome	The results show the presence of Y chromosome

INTERPRETATION

The results show very low risk for all tested conditions. The fetal fraction is 8.7%, which is sufficient for analysis. Please consult Test Method and Test Description sections below for information on the method, performance and limitations of the test. The results should be communicated by the referring clinician with appropriate counselling.

TEST METHOD

VERAgene is a Laboratory Developed Test (LDT) from NIPD Genetics Public Company Ltd for prenatal screening that analyses cell-free DNA (cfDNA) from maternal plasma and sample from biological father. Multiplexed parallel analysis of specific regions of interest was applied for the copy number determination of chromosomes 21,18,13 aneuploidies of X, Y chromosomes, select microdeletions including, DiGeorge (22q11.2 deletion), 1p36 deletion syndrome, Smith-Magenis (17p11.2 deletion), Wolf Hirschhorn (4p16.3 deletion), Y chromosome detection and mutation detection for 100 single gene diseases. A complete list of 100 monogenic diseases and tested mutations is available at www.nipd.com/veragene/report/monogenic_diseases.

TEST DESCRIPTION

Test performance is valid only for full chromosomal aneuploidies for chromosomes 21, 18, and 13, aneuploidies of X and Y chromosomes, select microdeletions, Y chromosome detection and a select number of pathogenic and likely pathogenic mutations associated with monogenic diseases listed in www.nipd.com/veragene/report/monogenic_diseases . It does not exclude other chromosomal abnormalities, birth defects or other complications. VERAgene is available for singleton, twin and vanished twin pregnancies, including in-vitro fertilization (IVF) pregnancies of at least 10 weeks of gestation. Sex chromosome aneuploidies are not reportable for twin and vanished twin gestations. The VERAgene test cannot be performed on pregnancies achieved with egg/sperm donation or surrogacy. Patients with malignancy or history of malignancy, patients with bone marrow, organ transplant or recent transfusion are not eligible for the test. Furthermore, samples from both biological parents are required for the test to be performed. The test result is valid only if the samples are collected from the biological parents. In a small number of cases the amount of fetal DNA present in maternal blood (fetal fraction), is not sufficient for analysis and a redraw maybe requested.

Validation studies are carried out by NIPD Genetics Public Company Ltd. The test is not intended and not validated for mosaicism, triploidy, partial trisomy or translocations. This test has been validated on full region deletions and maybe unable to detect deletion of smaller regions. The test will not identify all deletions associated with each microdeletion syndrome. Furthermore, the test is not intended and not validated for a number of mutations which are associated with the monogenic diseases listed in www.nipd.com/veragene/report/monogenic_diseases , but are not tested. Therefore, a very low risk result reduces but does not eliminate the possibility of the fetus to be affected or carry the mutation. A very high risk result for twin pregnancies indicates high risk for the presence of at least one affected fetus. In twin pregnancies, detection of Y indicates the presence of at least one Y chromosome. Although this test is highly accurate, there is still a small possibility for false positive or false negative results. This may be caused by technical and/or biological limitations, including but not limited to confined placental mosaicism (CPM) or other types of mosaicism, maternal constitutional or somatic chromosomal abnormalities, residual cfDNA from a vanished twin or other rare molecular events. The VERAgene test is not diagnostic but a screening test and results should be considered in the context of other clinical criteria. Clinical correlation with ultrasound findings, and other clinical data and tests is recommended. If diagnosis is desired, amniocentesis is necessary. The referral clinician is responsible for counselling before and after the test including the provision of advice regarding the need for additional invasive genetic testing.

The VERAgene non-invasive prenatal test development and performance evaluation was carried out by NIPD Genetics Public Company Ltd, which is regulated under the Clinical Laboratory Improvement Act of 1998 (CLIA) as qualified to perform high-complexity testing. VERAgene is intended for clinical purposes and should not be regarded as investigational or for research. The test has not been cleared or approved by the U.S. Food and Drug Administration (FDA), which does not require this test to go through premarket FDA review.

Approved by: Elena Kypri, Ph.D, ASCP

Approved by: Philippos Patsalis, Ph.D, HCLD, Laboratory Director

DATE OF REPORT (DD/MM/YYYY): **11/07/2022**

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