Patient:
DOE, JANE

Accession: NXGMDX
Created: December 16, 2021
Patient Gender: Female
Date Of Birth: August 11, 1995
Specimen Type: Whole blood
Collection Date: December 16, 2021

Receiving Physicians:

Received Date: December 16, 2021

Receiving Facilities:
NxGen MDx 801 Broadway Avenue Northwest Suite 203 Grand Rapids MI 49504

RESULTS SUMMARY

NXGEN MDX PRENATAL SCREEN

NEGATIVE: PREGNANCY AT LOW RISK

Estimated Due Date: 2022-9-24
Gestational Age: 10 weeks 2 days
Maternal Height, Weight, BMI: 5'1",196.8 lbs, 37
Fetus Count: 1

NXGEN INFORMED PRENATAL SCREEN		Fetal cfDNA Percentage: 7.1%
Condition	**Result**	**Risk After Test**
Trisomy 21 (T21)	Low Risk	< 0.01% (1 in 10,000)
Trisomy 18 (T18)	Low Risk	< 0.01% (1 in 10,000)
Trisomy 13 (T13)	Low Risk	< 0.01% (1 in 10,000)
Predicted Fetal Sex	Male	
Sex Chromosomes	Low Risk	< 0.01% (1 in 10,000)

INTERPRETATION
Results consistent with low risk for the above chromosomal disorders.

The risk after test percentage represents the remaining chance that the pregnancy is affected with the indicated chromosome anomaly in view of a negative result. NIPS is a screening test; therefore, false positive and false negative results can occur.

No irreversible clinical decisions should be made based on these screening results alone. Clinical correlation is suggested and a consultation with a genetic counselor is recommended for any questions or concerns. Healthcare providers may recommend other types of genetic testing during or after pregnancy.

Sample Report
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METHODOLOGY

NxGen MDx uses massively parallel shotgun sequencing to analyze cell-free DNA present in maternal plasma. The test is validated for pregnancies with gestational age of at least 10 weeks 0 days, as estimated by last menstrual period, crown rump length, or other appropriate method. These performance metrics are unaffected by fetal fraction. Fetal fraction and positive predictive value are provided in compliance with ACOG and American College of Medical Genetics and Genomics (ACMG) guidelines.

NxGen MDx's non-invasive prenatal screen (NIPS) is designed as a screening tool to identify pregnancies at risk for aneuploidy (abnormal chromosome counts) and deletions involving specific chromosomal regions (1p36.3-p36.2, 4p16.3-p16.2, 5p15.3-p15.1, 15q11.2-q13.1 and 22q11.21). Microdeletions and aneuploidy of chromosomes other than 13, 18, and 21 are reported only when requested. In about 0.5% of cases it is not possible to determine whether a fetus is XY or XX. It is important to note that these cases are distinct from sex chromosomal aneuploidies, and there is not evidence of only one sex chromosome (XO) or three chromosomes (XXY) in these cases. We know that patients value both the aneuploidy risk reduction and the fetal sex information, and we include an expedited NxGen Fetal Gender Test in these rare cases so that fetal sex chromosome detail can be included in the report. Fetal cell-free DNA is isolated from the maternal plasma and sex-specific markers are amplified and analyzed using a multiplex PCR assay. Analysis of the amplification data based on Y-chromosome markers is used to predict the fetal sex of a singleton pregnancy.

LIMITATIONS OF THE TEST

These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal or subchromosomal abnormalities, birth defects, and other conditions. A negative test result does not eliminate the possibility of chromosomal abnormalities. This test is not intended to identify pregnancies at risk for open neural tube defects. The American Congress of Obstetricians and Gynecologists (ACOG) recommends patients who are undergoing cell-free DNA screening be offered maternal serum alpha-fetoprotein screening or ultrasound evaluation for risk assessment.

When an aneuploidy detected or suspected result is reported in a twin pregnancy, the status of each individual fetus cannot be determined. Therefore, diagnostic testing is recommended. Although the presence or absence of Y chromosome material can be reported in a twin pregnancy, the occurrence of sex chromosome aneuploidies such as monosomy X, XXX, XXY, and XYY cannot be evaluated in twin pregnancies.

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) or of the mother (chromosomal mosaicism).

Disclaimer: The use of this information to guide patient care is the responsibility of the health care provider, including advising for the need for genetic counseling or diagnostic testing. Any test should be interpreted in the context of all available clinical findings. This test was developed and its performance characteristics were determined by NxGen MDx's CLIA-accredited laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration.

This test was performed at NxGen MDx, located at 801 Broadway Suite 203, Grand Rapids, Michigan, 49504. CLIA Number: 23D2059943.

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NONINVASIVE PRENATAL SCREENING PERFORMANCE DATA

Chromosome	Sensitivity	Specificity	Sex Chromosome	Sensitivity	Specificity
21	99.7%	99.96%	XX	97.6%	99.2%
18	97.9%	99.96%	XY	99.1%	98.9%
13	99.0%	99.96%	Monosomy X	95.8%	99.86%
XXX/XXY/XYY		Other sex chromosome aneuploidies will be reported if detected. Limited data for these rarer aneuploidies preclude performance calculations.			
Sex chromosome mosaicism cannot be distinguished by this method (occurrence <0.3%). Patient with such mosaicism will have a sex chromosome result reported and will fall into one of six categories: Monosomy X, XXX, XXY, XYY, XX, XY					
When expanded analysis is requested, aneuploidy in other chromosomes and included microdeletions (22q11.2, 15q11.2, 1p36, 4p, and 5p) will be reported if detected. Limited data for these rare subchromosomal anomalies preclude performance calculations.					

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