JUNO diagnostics

PATIENT DEMOGRAPHICS

Patient Name:	Niran Test	LMP:	2022-09-26
Date of birth:	1995-01-01	Sample collection date:	2022-11-22
Email:	nirantestjuno+401@gmail.com	GA at sample collection:	8 weeks/1 days
Report date:	2022-11-24		
Study performed:	Hazel™ NIPS Plus		

About Juno Hazel™ Plus Non-Invasive Prenatal Screening:

Juno Diagnostics' Hazel™ laboratory-developed test (LDT) is a screening evaluation which analyzes circulating cell-free DNA from a maternal blood sample for chromosomal and subchromosomal representations of the fetus and gestational carrier. The screen is indicated for use in human pregnancies for the screening of fetal chromosomal aberrations. Validation data on multiple pregnancies, such as twins, is limited and the ability of this screen to detect aneuploidy in a triplet pregnancy has not yet been validated.

FINAL RESULTS SUMMARY

Result

Increased Risk: Monosomy X

Fetal Sex

Male

Fetal Fraction

14.50%

POSITIVE PREDICTIVE VALUE

Age: 27 years
GA: 8 weeks/1 days

People with this result have an increased chance to have a baby with Monosomy X.

CONDITIONS EVALUATED

	Post-test risk	Interpretation	
Trisomy 21	Approximately 1 in 10,000	Aneuploidy Not Detected	21
Trisomy 18	Fewer than 1 in 5,000	Aneuploidy Not Detected	18
Trisomy 13	Fewer than 1 in 10,000	Aneuploidy Not Detected	13
Sex chromosome aneuploidy	Increased	Aneuploidy Detected	X

WHAT DOES THIS RESULT MEAN?

This result is consistent with a male fetus at increased risk for Monosomy X, but a low risk for Down syndrome and Edwards syndrome and Patau syndrome or other sex chromosome aneuploidies.. Genetic counseling and prenatal diagnosis are recommended.

LAB DIRECTOR COMMENTS

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Juan-Sebastian Saldivar

2022-11-29

Date

SCREENING METHODS

Circulating cell-free DNA (ccfDNA) is purified from the plasma component of maternal blood. The extracted DNA is then converted into a whole genome DNA library for sequencing-based analysis of chromosomes 21, 18, and 13.

SCREENING PERFORMANCE

Juno's Hazel™ laboratory developed test has been evaluated for clinical performance in multiple studies, inclusive of >1600 total venous and capillary samples. Based on this data, expected performance with at-home self-collection is as follows:

	SENSITIVITY (95% CI)	SPECIFICITY (95% CI)
Trisomy 21	>99% (95%-100%)	>99.9% (99%-100%)
Trisomy 18	>99% (94%-100%)	>99.9% (99%-100%)
Trisomy 13	>99% (79%-100%)	>99.9% (99%-100%)
Sex chromosome aneuploidy	Performance Data TBD	Performance Data TBD

SCREENING LIMITATIONS

This screen is for screening purposes only, and is not diagnostic. While the results of these screens are highly accurate, discordant results, including inaccurate fetal sex prediction, may occur due to placental, maternal, or fetal mosaicism or neoplasm; vanishing twin; prior maternal organ transplant; or other causes. Sex chromosomal aneuploidies are not reportable for known multiple gestations.

The screen does not replace the accuracy and precision of prenatal diagnosis with CVS or amniocentesis. A patient with a positive screening result should receive genetic counseling and be offered invasive prenatal diagnosis for confirmation of test results. A negative result does not ensure an unaffected pregnancy nor does it exclude the possibility of other chromosomal abnormalities or birth defects which are not a part of this screening evaluation. An uninformative result may be reported, the causes of which may include but are not limited to insufficient sequencing coverage, noise or artifacts in the region, amplification or sequencing bias, or insufficient fetal representation. The ability to report results may be impacted by maternal BMI, maternal weight and maternal autoimmune disorders.

Screening for whole chromosome abnormalities (including sex chromosomes) and for sub-chromosomal abnormalities could lead to the potential discovery of both fetal and maternal genomic abnormalities that could have major, minor, or no, clinical significance. Evaluating the significance of a positive or a non-reportable result may involve both diagnostic testing and additional studies on the pregnant person. Such investigations may lead to a diagnosis of maternal chromosomal or sub-chromosomal abnormalities, which on occasion may be associated with benign or malignant maternal neoplasms.

This screen may not accurately identify fetal triploidy, balanced rearrangements, or the precise location of sub-chromosomal duplications or deletions; these may be detected by prenatal diagnosis with karyotype and SNP-microarray. Cell-free DNA screening is not intended to identify pregnancies at risk for neural tube defects or ventral wall defects; these may be detected by prenatal ultrasound evaluation.

The results of this screening, including the benefits and limitations, should be discussed with a qualified healthcare provider. Pregnancy management decisions, including termination of the pregnancy, should not be based on the results of these screens alone. The healthcare provider is responsible for the use of this information in the management of their patient.

REFERENCES

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