VisibiliT™ Risk Score Test Lab Report

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CLIA #: 05D2015356 CAP #: 7527138
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Final Report

Ordering Provider:	**Doe, John, MD**	Patient:	**Sample, Jane**
Provider Location:	**Grand Rapids**	DOB:	**09/13/1970**
Provider Phone:	**555-555-5555**	Patient ID:	**12345-01234**
Date Ordered:	**11/28/2014**	Specimen:	**1035600024**
Date Collected:	**11/29/2014**	Referral Clinician:	**Smith, Jane, GC**
Date Received:	**11/30/2014**	Lab Director:	**Juan-Sebastian Saldivar, MD**
Order ID:	**ORD12345-01234**	Date Reported:	**04/29/2013 6:00 PM PT**

Test Result for Chromosomes 21, 18 and Y

Risk Cutoff: 1/100

High Risk

Trisomy 21: 99/100

	Age-related risk[1]	Post-NIPT risk
Trisomy 21	**1/1,200**	▶ **99/100**
Trisomy 18	1/800	1/10,000

Y chromosomal material detected.
Consistent with a male fetus.

Lab Director Comments
Fetal Fraction: 13%

Interpretation

These results are consistent with an increased relative amount of chromosome 21 material, indicating an increased risk of Trisomy 21. Clinical correlation is suggested. Genetic Counseling is recommended.

Test Method

Circulating cell-free DNA was purified from the plasma component of anti-coagulated maternal whole blood. It was then converted into a genomic DNA library for the determination of chromosome 21, 18, and Y.[1]

About the Test

The VisibiliT test analyzes circulating cell-free DNA extracted from a maternal blood sample. Maternal age, fetal fraction and the relative amount of chromosome material for chromosome 21 and 18 are used to generate a risk score. This test is indicated for use in singleton pregnancies only.[2]

Performance

This blinded analytical validation study was designed to be representative of a general pregnancy population cohort, of ten weeks gestation or greater.[2]

Chromosome	Performance	Confidence Interval (95% CI)
Trisomy 21	Sensitivity: > 99%	80.8–100%
	Specificity: > 99.9%	99.5–100%
Trisomy 18	Sensitivity: > 99%	65.6–100%
	Specificity: > 99.9%	99.5–100%
Y chromosome	Accuracy: 99.3%	98.6–99.7%

Limitations of the Test

The VisibiliT test reports a risk score result. Cell-free DNA does not replace the accuracy and precision of prenatal diagnosis with CVS or amniocentesis. A patient with a high risk result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results. A low risk result does not ensure an unaffected pregnancy. While results of this testing are highly accurate, not all chromosomal abnormalities may be detected due to placental, maternal or fetal mosaicism, or other causes. The health care provider is responsible for the use of this information in the management of their patient.

Note

This test was developed and its performance characteristics determined by Sequenom Laboratories. It has not been cleared or approved by the U.S. FDA. This test is used for clinical purposes. It should not be regarded as investigational or for research. This laboratory is certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing and accredited by the College of American Pathologists.

References

1. Savva GM, et al. The maternal age-specific live birth prevalence of trisomies 13 and 18 compared to trisomy 21 (Down syndrome). *Prenat Diagn*. 2010 Jan; 30(1):57-64.
2. Jensen TJ, et al. High-throughput massively parallel sequencing for fetal aneuploidy detection from maternal plasma. *PLoS One*. 2013;8(3):e57381.
3. Kim, S et al. Application of risk-score analysis to low-coverage sequencing data for noninvasive detection of trisomy 21 and trisomy 18. Poster presented at the 18th International Conference on Prenatal Diagnosis and Therapy; July 2014; Brisbane, Australia.

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xx/xx/2014

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