TEMPLATE 1: 21/18/Yes Gender

VisibiliT™ Risk Score Test Lab Report

Sequenom Laboratories
3595 John Hopkins Court
San Diego, CA 92121
CLIA #: 05D2015356 CAP #: 7527138
877.821.7266

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

Low Risk

	Age-related risk	Post-NIPT risk
Trisomy 21	1/1,200	1/10,000
Trisomy 18	1/800	1/10,000

Y chromosomal material present.
Consistent with a male fetus.

Interpretation

These results are consistent with a normal amount of chromosome 21 and 18, indicating a low risk of Trisomy 21 and 18.

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

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Test Result for Chromosomes 21 and 18
Risk Cutoff: 1/100

Low Risk

	Age-related risk	Post-NIPT risk
Trisomy 21	1/1,200	1/10,000
Trisomy 18	1/800	1/10,000

Interpretation

These results are consistent with a normal amount of chromosome 21 and 18, indicating a low risk of Trisomy 21 and 18.

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 and 18.[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%

Limitations of the Test

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Test Result for Chromosomes 21, 18 and Y
Risk Cutoff: 1/100

High Risk

Trisomy 21: 1/10

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

Y chromosomal material present.
Consistent with a male fetus.

Interpretation

These results are consistent with an increased relative amount of chromosome 21, 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.

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Test Result for Chromosomes 21, 18 and Y
Risk Cutoff: 1/100

High Risk

Trisomy 21: 99/100

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

Y chromosomal material present.
Consistent with a male fetus.

Interpretation

These results are consistent with an increased relative amount of chromosome 21, 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.

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Test Result for Chromosomes 21, 18 and Y
Risk Cutoff: 1/100

High Risk

Trisomy 18: 50/100

	Age-related risk	Post-NIPT risk
Trisomy 21	1/1,200	1/10,000
Trisomy 18	**1/800**	▶ **50/100**

Y chromosomal material present.
Consistent with a male fetus.

Interpretation

These results are consistent with an increased relative amount of chromosome 18, indicating an increased risk of Trisomy 18. 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

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Test Result for Chromosomes 21, 18 and Y
Risk Cutoff: 1/100

Non-reportable	Quantity of DNA not sufficient.

Test Method

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About the Test

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Performance

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Trisomy 21	Sensitivity: > 99%	80.8–100%
	Specificity: > 99.9%	99.5–100%
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Y chromosome	Accuracy: 99.3%	98.6–99.7%

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Test Result for Chromosomes 21, 18 and Y
Risk Cutoff: 1/100

Non-reportable	Due to technical or sample-related issues, data failed to meet quality standards for interpretation.

Test Method

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