



CLINICAL VALIDATION STUDY

POSITIVE RESULTS

SENSITIVITY

SPECIFICITY

1,696
SAMPLES

DNA Sequencing of Maternal Plasma to Detect Down syndrome: An International Clinical Validation Study

Genet Med. 2011;13(11):913-920.

210 OF **212**
Trisomy 21

99.1%

99.9%

1,988
SAMPLES

DNA Sequencing of Maternal Plasma Reliably Identifies Trisomy 18 and Trisomy 13, as well as Down syndrome: An International Collaborative Study

Genet Med. 2012;14(3):296-305.

59 OF **59**
Trisomy 18

>99.9%

99.6%

11 OF **12**
Trisomy 13

91.7%

99.7%

2,015
SAMPLES

DNA Sequencing of Maternal Plasma to Identify Down syndrome and Other Trisomies in Multiple Gestations

Prenat Diagn. 2012;32(8):730-734.

8 OF **8**
7 OF Trisomy 21
1 OF Trisomy 13

DETECTION RATE:
>99.9%



Y CHROMOSOME

2,107
SAMPLES

Accuracy of noninvasive prenatal fetal sex determination
Poster presented at American Society of Human Genetics Meeting (ASHG);
2012; San Francisco.

ACCURACY:
99.4%



COMBINED SEX ANEUPLOIDIES

POSITIVE RESULTS

SENSITIVITY

SPECIFICITY

420
SAMPLES

Noninvasive prenatal detection of sex chromosomal aneuploidies by sequencing circulating cell-free DNA from maternal plasma.

Prenat Diagn. 2013;33(6):591-597

25 OF **26**

96.2%

99.7%

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M Carnoy



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