

 CLINICAL VALIDATION STUDY		POSITIVE RESULTS	SENSITIVITY	SPECIFICITY
1,696 SAMPLES	DNA Sequencing of Maternal Plasma to Detect Down syndrome: An International Clinical Validation Study <i>Genet Med.</i> 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 <i>Genet Med.</i> 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 <i>Prenat Diagn.</i> 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. <i>Prenat Diagn.</i> 2013;33(6):591-597	25 OF 26	96.2%	99.7%

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