



Genetic Testing Specifications

Test	Gestational Age Parameters	Detection Rate*	Procedure-Related Risk of Miscarriage	Screen Positive Rate	Sensitivity	Specificity
Karyotype by Amniocentesis	15 0/7 - 22 6/7	99.9%	1/200 (1/340 at NWP)			
Karyotype by Chorionic Villus Sampling	11 0/7 - 13 6/7	99.9%	1/100 - 1/150			
FISH for 21, 18, 13, X, Y (Fluorescent In-Situ Hybridization)		99.8%			99.3%	99.9%
Sequential Screening	11 3/7 - 13 6/7		N/A			
Trisomy 21		92%		4%		
Trisomy 18		90%		4%		
Open Neural Tube Defect		60-80%**		1-3%		
Serum Integrated Screening	11 3/7 - 13 6/7		N/A			
Trisomy 21		88%		6%		
Trisomy 18		90%		6%		
Open Neural Tube Defect		60-80%**		1-3%		
Quad Screening	15 0/7 - 22 6/7		N/A	5%		
Trisomy 21		79%				
Trisomy 18		73%		0.20%		
Open Neural Tube Defect		60-80%**		1-3%		
Non-invasive Prenatal Testing (NIPT)	>10 0/7		N/A	<0.5%		
Trisomy 21		99%			99.9%	99.8%
Trisomy 18		97%			97.4%	99.6%
Trisomy 13		85%			87.5%	99%
Sex chromosome aneuploidy		95%			95%	97.6%
XX		97%			97%	99.2%
XY		98%			99%	99%

*Numbers may vary by laboratory. **95% for anencephaly