

# GENETIC TESTING

## DOWN SYNDROME, TRISOMIES 13/18 AND SPINA BIFIDA

TEST	GESTATIONAL AGE	DEFINITIVE ANSWER	DETECTION RATE	FALSE POSITIVE	LOSS RISK	SPINA BIFIDA TEST	ELIGIBLE PATIENTS
<b>**Ultrascreen</b> -Nuchal translucency and first trimester blood test	Blood: 9 <sup>3</sup> weeks Sono: 12 weeks	NO	~93%	~5%***	None	NO	ALL
<b>Sequential</b> - Nuchal translucency – first and second trimester blood tests (If NIPT done)	15-21 <sup>6</sup> weeks	NO	95%	~5%	None	YES	ALL
<b>AFP Tetra Screen</b>	15-21 <sup>6</sup> weeks	NO	~80% (~50% twins)	~5%***	None	YES	ALL
<b>**NIPT</b> -Non Invasive Prenatal Test	10 weeks	NO	99%	<1%	None	NO	ALL****
Serum <b>AFP</b> only	15-21 <sup>6</sup> weeks	NO	~80% (~50% twins)		None	YES	ALL
<b>CVS</b> -Chorionic Villi Sampling	10-13 weeks	YES	>99%	<1%	1/100- 1/200	NO	AMA* or HIGH RISK
<b>Amniocentesis</b>	≥ 16 weeks	YES	>99%	~0%	1/300-1/500	YES	AMA* or HIGH RISK

\* AMA = Advance Maternal Age (35 years old or greater at delivery)

\*\* Insurance companies usually cover these tests for women 35 and older. However, women under 35 may elect to have testing (potentially at their own expense depending on insurance coverage) and women 35 and older may opt not to have testing due to personal choice.

\*\*\* False positive percentage increases with maternal age.

\*\*\*\* All patients are eligible for insurance submission. Please visit [www.integratedgenetics.com/patients/cost-estimator](http://www.integratedgenetics.com/patients/cost-estimator) to estimate your personal out-of-pocket expense prior to having testing done.