

Genetic Testing Options

DOWN SYNDROME, TRISOMIES 18/13 AND SPINA BIFIDA

TEST	GESTATIONAL AGE	DEFINITIVE ANSWER	DETECTION RATE	FALSE POSITIVE	LOSS RISK	SPINA BIFIDA	WHAT RISKS ARE EVALUATED?	ELIGIBLE PATIENTS
ULTRASCREEN - NUCHAL TRANSLUCENCY SONO PLUS BLOODWORK²	Blood: 10 weeks Sono: 12 weeks	NO	~95-98%	~0.5-2.3%	NONE	No	CARDIAC DEFECTS, PLACENTAL INSUFFICIENCIES, CHROMOSOMAL RISKS	ALL
SEQUENTIAL – MUST HAVE DONE ULTRASCREEN²	15-21 ⁶ weeks	NO	Trisomy 21/18: 95% Spina Bifida: 90%	~5%	NONE	YES	SPINA BIFIDA, PLACENTAL INSUFFICIENCIES, CHROMOSOMAL RISKS	ALL
AFP TETRA SCREEN	15-21 ⁶ weeks	NO	~80% (~50% Twins)	~5%	NONE	YES	SPINA BIFIDA, PLACENTAL INSUFFICIENCIES	ALL
NIPT – NON-INVASIVE PRENATAL TEST	After 10 weeks	NO	Trisomy 21/18: 99% Trisomy 13: 91%	<1%	NONE	No	CHROMOSOMAL RISKS	ALL ³
SERUM AFP ONLY	15-21 ⁶ weeks	NO	~80% (~50% Twins)		NONE	No	SPINA BIFIDA	ALL
CVS – CHORIONIC VILLI SAMPLING	10-13 weeks	YES	>99%	<1%	1/100-1/200	No	CHROMOSOMAL RISKS	AMA ¹ OR HIGH RISK
AMNIOCENTESIS	≥ 16 weeks	YES	>99%	~0%	1/300-1/500	YES	CHROMOSOMAL RISKS, SPINA BIFIDA	AMA ¹ OR HIGH RISK

Screening Test Diagnostic Test

1. AMA – Advanced Maternal Age (35 years or older at due date)
2. Insurance companies usually provide some coverage for these tests for women 35 and older. Women under 35 may elect to have testing done (potentially at their own expense depending on insurance coverage) and women over 35 may opt not to have testing done due to personal choice.
3. All patients are eligible for insurance submission. Please visit www.integratedgenetics.com/patients/cost-estimator to estimate your personal out of pocket cost prior to having testing done.