

Genetic Screening Tests



AVERAGE RISK PREGNANCY

NAME OF SCREEN	TEST TYPE	WHAT DOES IT LOOK FOR
First Trimester Screen (10-13.6 weeks) 82-87% detection rate for Trisomy 21	Blood draw +Ultrasound	Down Syndrome (Trisomy 21) Edward's Syndrome (Trisomy 18) Patau syndrome (Trisomy 13)
Quad Screen (16-21 weeks + 6 days) 81% detection rate for Trisomy 21	Blood draw	Down Syndrome (Trisomy 21) Edward's Syndrome (Trisomy 18) Spina Bifida
MSAFP (16-21 weeks + 6 days) 80-85% detection rate for open neural tube defects	Blood draw	Spina Bifida
Essential Panel – Carrier Screen (any time) 99% detection rate of these three conditions in maternal DNA	Blood draw	Cystic Fibrosis Spinal Muscular Atrophy Fragile X Syndrome

INCREASED RISK PREGNANCY

Age > 35 years old, increased risk by family history

NAME OF SCREEN	TEST TYPE	WHAT DOES IT LOOK FOR
Cell Free - NIPT (after 10 weeks) 99% detection rate for Trisomy 21	Blood draw	Down Syndrome (Trisomy 21) Edward's Syndrome (Trisomy 18) Patau syndrome (Trisomy 13) Sex Chromosome Aneuploidies

These tests may or may not be covered by your insurance and may be subject to a deductible. Please consult your insurance company for more information. These tests are optional. If you have further questions or require more detailed counseling, please feel free to discuss with a provider at your next visit.