

Physicians and Midwives Collaborative Practice

Genetic Testing Options

Test	Timing
Ashkenazi Jewish Panel Cystic Fibrosis Hemoglobinopathy Screen All of the above are genetic conditions which can be detected by blood tests done ideally before pregnancy	Pre Pregnancy Early Pregnancy
Nuchal Translucency An ultrasound measuring the thickness of the neck of the fetus in combination with a blood test from the mother. This gives an estimate of the risk of Down's Syndrome, Trisomy 13/18. Has 90% detection rate with 5% false positive	11.0-13.6 weeks
Chorionic Villi Sampling (CVS) A diagnostic test done by inserting a needle into the placenta. This is the earliest diagnostic test. This test will detect chromosomal abnormalities. There is a 1-2% risk of miscarriage from this procedure.	10-13 weeks
Quadruple Screen (AFP) A screening test done by taking the mother's blood. Reports the risk of Down's syndrome, Trisomy 13/18 and Spina Bifida. It is done later in pregnancy and is an alternative to the earlier Nuchal Translucency test. Has 85% detection rate with 5% false positive.	16-21 weeks
Genetic Ultrasound A targeted ultrasound looking for markers associated with Down's syndrome and other genetic conditions. An ultrasound may also detect birth defects.	16-18 weeks
Amniocentesis An invasive diagnostic test done by inserting a needle into the amniotic fluid. This test will detect chromosomal abnormalities. The risk of miscarriage is 1 in 400-700.	16-23 weeks

I understand the risks, benefits, and alternatives to the genetic tests described above.

Print Name

Date

Signature

Date