Minnesota Perinatal Physicians

Prenatal Genetic Tests

First Trimester Screening (Nuchal Translucency)	Second Trimester Screening (Sequential or Quadruple Screen)	Noninvasive Prenatal Testing	Chorionic Villi Sampling (CVS) and Amniocentesis
Screening test This test can help identify if you are at an increased risk of carrying a baby with a chromosome condition such as Down syndrome or trisomy 18.	**Screening test ◊** This test can help identify if you are at an increased risk of carrying a baby with a chromosome condition such as Down syndrome, trisomy 18 or spina bifida.	**Screening test** This test can help identify if you are at an increased risk of carrying a baby with one of the most common chromosome conditions including Down syndrome, trisomy 18, trisomy 13 and sometimes sex chromosome conditions.	**Diagnostic tests** These are the only tests that can say with certainty if your baby has a chromosome condition. It is also possible to test for other genetic conditions such as cystic fibrosis and spinal muscular atrophy, if needed. There is a small chance of miscarriage (less than 0.5 percent).
How the test is done ■ ultrasound ■ blood test	**How the test is done ◊** ■ ultrasound ■ two separate blood tests	**How the test is done** ■ blood test	**How the test is done** ■ CVS: A catheter or needle is used to remove a small amount of chorionic villi (cells that form the placenta). ■ Amniocentesis: Ultrasound and a thin needle are used to remove a small amount of amniotic fluid.

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When is the test done It is usually done between 11 and 14 weeks of pregnancy.	**When is the test done ◊** The first blood test is done between 11 and 14 weeks of pregnancy. The second blood test is done between 15 and 21 weeks of pregnancy.	**When is the test done** It can be done any time after 10 weeks of pregnancy.	**When is the test done** CVS is usually done between 10 and 13 weeks of pregnancy An amniocentesis is usually done after 15 weeks of pregnancy.
Test results Test results are available in 1 to 2 weeks	**Test results ◊** Final test results are available 1 to 2 weeks after your second blood test.	**Test results** Test results are available in 1 to 2 weeks	**Test results** Final test results are available in 1 to 2 weeks. Optional: You can be given initial results in 1 to 2 days.
Detection rate* ■ Down syndrome: 82 to 87 percent ■ Trisomy 18: 80 percent	**Detection rate* ◊** ■ Down syndrome: 95 percent ■ Trisomy 18: 90 percent ■ Spina bifida: 80 percent	**Detection rate*** ■ Down syndrome: more than 99 percent ■ Trisomy 18: 99 percent ■ Trisomy 13: 79 to 92 percent	**Detection rate*** ■ Down syndrome: 99.99 percent ■ Trisomy 18: 99.99 percent ■ Trisomy 13: 99.99 percent ■ Other major chromosome conditions: 99.99 percent

***The detection rate is how accurate the test is in finding these conditions. No prenatal test is 100 percent accurate. The higher the percent, the more likely the condition will be found.**

◊ The quadruple screen is one blood test done between 15 and 22 weeks of pregnancy. The detection rate for this test alone is lower compared to the sequential screen.

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