

**Options for Genetic Testing of the Baby**

**Screening Tests**

**Diagnostic Tests**

Name of Testing	NIPT (Non Invasive Prenatal Testing)	Sequential Screen or Integrated	Quad Screen	Chorionic Villus Sampling (CVS)	Amniocentesis
<b>Type of testing</b>	Blood work	Ultrasound and Blood work	Blood work	Small piece of placenta is removed through the vagina or through the abdomen	Small amount of amniotic fluid is removed by inserting a needle into the abdomen
<b>When can the test be done?</b>	From 10 weeks on	Part 1: 11-14 weeks Part 2: 15-20 weeks	15-20 weeks	11-13 weeks	15-20 weeks
<b>What can the test detect?</b>	Trisomy 13, Trisomy 18, Down syndrome, gender	Trisomy 13, Trisomy 18, Down syndrome and open neural tube defects	Trisomy 18, Down syndrome and open neural tube defects	Rules out 99% of all chromosomal abnormalities, including sex chromosomes	Rules out 99% of all chromosomal abnormalities, including sex chromosomes and neural tube defects
<b>How invasive is the testing and what is the detection rate?</b>	<ul style="list-style-type: none"> <li>- Non invasive</li> <li>- 99.9% detection rate</li> <li>- Early detection</li> </ul>	<ul style="list-style-type: none"> <li>- Non invasive; no risk of miscarriage</li> <li>- 85-92% detection rate for Trisomy 13, 18 &amp; 21</li> <li>- Early detection</li> </ul>	<ul style="list-style-type: none"> <li>- Non invasive; no risk of miscarriage</li> <li>- 85% detection rate for Trisomy 13, 18 &amp; 21</li> <li>- More universal insurance coverage</li> </ul>	<ul style="list-style-type: none"> <li>- Comprehensive: rules out 99.9% of all chromosomal abnormalities</li> <li>- Done early in the pregnancy</li> <li>- Is covered by insurance</li> <li>- Definitive</li> </ul>	<ul style="list-style-type: none"> <li>- Comprehensive: rules out 99.9% of all chromosomal abnormalities.</li> <li>- Tests for open neural tube defects</li> <li>- Is covered by insurance</li> <li>- Definitive</li> </ul>
<b>Other information</b>	<ul style="list-style-type: none"> <li>- Does not test for any other abnormalities</li> <li>- Abnormal results require confirmation</li> <li>- May not be covered by insurance</li> </ul>	<ul style="list-style-type: none"> <li>- 5% false position rate</li> <li>- Abnormal test require either an Amnio or CVS to confirm</li> <li>- Does not test for any other chromosomal problems other than Trisomy 18, 13 &amp; 21</li> <li>- Insurance will cover EITHER this ultrasound OR the 18 week ultrasound but NOT both (\$500)</li> </ul>	<ul style="list-style-type: none"> <li>- Lower detection rates</li> <li>- Abnormal tests require an Amnio to confirm</li> <li>- Does not test for any other chromosomal problems other than Trisomy 18, 13 &amp; 21</li> </ul>	<ul style="list-style-type: none"> <li>- Invasive test; has an associated 1% chance of miscarriage</li> <li>- Does not test for open neural tube defects</li> </ul>	<ul style="list-style-type: none"> <li>- Invasive test; has an associated 0.5% chance of miscarriage</li> <li>- Full results might not come back until 22 weeks gestation</li> </ul>