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Testing Services by Ariosa Diagnostics

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PRENATAL TEST

For You

It's about understanding your prenatal testing options for your pregnancy

Answers that matter

Screening Options

Non-Invasive Prenatal Screening (NIPT)
NIPT is a blood test that looks at fetal DNA found in the mother's bloodstream. This screening test helps determine if your fetus is at increased risk for Down syndrome (trisomy 21), trisomy 18, and trisomy 13. It may also screen for fetal sex and differences in the number of sex chromosomes. NIPT has a higher detection rate and lower false positive rate than conventional screening.[1] Results are typically returned in less than one week. Less than 5 in 1,000 women will receive a false positive* result for trisomies 21, 18, 13.[1] In order to confirm a high risk NIPT screening result during pregnancy, your physician may recommend invasive diagnostic testing (chorionic villus sampling (CVS) or amniocentesis). Alternatively, this diagnostic testing can be done after delivery.

First Trimester Combined Screening (FTS)
FTS involves Nuchal Translucency (NT) ultrasound and blood work done between 11-14 weeks. The nuchal translucency is a pocket of fluid behind the fetal neck. It can be seen by ultrasound in the first trimester of pregnancy. NT screening measures the size of this pocket. Fetuses with larger NT's have a higher risk for Down syndrome and other genetic conditions.1 FTS can detect up to 82-87% of fetuses with Down syndrome (trisomy 21).1 Depending on the laboratory, the test may also screen for trisomies 18 and 13. Results take approximately five days. 1 in 20 women will receive a false positive* result.1 If prenatal diagnosis is desired, invasive diagnostic testing (CVS or amniocentesis) may be recommend by your physician to confirm the result.

Second Trimester Serum Screening (Quad/Triple)
Quad/Triple screen involves a blood draw at 15-22 weeks gestation. This testing can detect 69-81% of fetuses with Down syndrome (trisomy 21).[1] This testing will also provide a risk assessment for trisomy 18 and open spina bifida (a type of open neural tube defect). Results are reported 1-2 weeks after the blood draw. 1 in 20 women will received a false positive* result.[1] If prenatal diagnosis is desired, amniocentesis may be recommend by your physician to confirm the result.

Sequential Screening
This type of screening involves NT screening and two blood draws, one at 11-14 weeks and the second at 15-22 weeks. If both blood draws are completed, sequential screening can detect up to 95% of fetuses with Down syndrome (trisomy 21).[1] This testing will also provide a risk assessment for trisomy 18 and open spina bifida (a type of open neural tube defect). Some results are available in the first trimester, but the final results are reported in the second trimester. Approximately 1 in 20 of women will receive a false positive* result.[1] If prenatal diagnosis is desired, invasive diagnostic testing (CVS or amniocentesis) may be recommend by your physician to confirm the result.

Integrated Screening
Integrated screening involves NT screening and two blood draws, one at 11-14 weeks and the second at 15-22 weeks. Integrated screening can detect 96% of fetuses with Down syndrome (trisomy 21).[1] Integrated screening will also provide a risk assessment for trisomy 18 and open spina bifida (a type of open neural tube defect). Results are not reported until the second trimester when all parts of the test have been compoleted. 1 in 20 women will receive a false positive* result.[1] If prenatal diagnosis is desired, amniocentesis may be recommend by your physician to confirm the result.

Diagnostic Testing

Chorionic villus sampling (CVS) can be done between 10-13 weeks of pregnancy. A small sample of cells that form the placenta are collected and the chromosomes within those cells are studied.[3] This procedure under ultrasound guidance can be done through the cervix using a catheter or through the abdomen using a needle. CVS is 98-99% accurate for the detection of chromosome conditions.[4] It has a risk of miscarriage of <0.5%.[3]

Amniocentesis is generally done between 15-20 weeks of pregnancy although it can be performed later in gestation.[3] Using ultrasound guidance a needle is inserted through the belly (not through the belly button) into the amniotic sac. A small amount of amniotic fluid surrounding the fetus is taken out. Inside the fluid are cells from the fetus. Those cells contain the chromosomes from the fetus, which are studied. Amniocentesis is 98-99% accurate for the detection of chromosome conditions.[5] It has a risk of miscarriage of 0.1-0.3%.[3] Amniocentesis can also detect neural tube defects like spina bifida by measuring alpha-fetoprotein (AFP) in the amniotic fluid.[5]

The following are SCREENING options. Screening options provide a risk assessment, but do not diagnose (tell you 'yes' or 'no') a fetus with a chromosome condition. Screening options pose no risk to the pregnancy, but invasive, diagnostic testing may be recommended by your physician to confirm whether a fetus is affected.

	First Trimester Screening	Sequential Screening	Integrated Screening	Second Trimester Serum Screening (Quad/Triple)	Harmony (NIPT)
Procedure Description	A blood draw that measure chemical levels and a specialized fetal nuchal translucency ultrasound that measures the pocket of fluid behind the fetal neck	Two blood draws that measure chemical levels and a specialized fetal nuchal translucency ultrasound that measures the pocket of fluid behind the fetal neck	Two blood draws that measure chemical levels and a specialized fetal nuchal translucency ultrasound that measures the pocket of fluid behind the fetal neck	A blood draw that measures chemical levels at 15-22 weeks	A blood draw that measures DNA from the pregnancy
Gestational Age	11-14 weeks	11-14 weeks for first blood draw 15-22 weeks for second blood draw	11-14 weeks for first blood draw 15-22 weeks for second blood draw	15-22 weeks	Consider 10 weeks until delivery
Detection Rate for Trisomy 21	82-87%[1]	95%[1]	96%[1]	69-81%[1]	>99%[1]
False Positive Rate for Trisomy 21	5%[1]	5%[1]	5%[1]	5%[1]	<0.1%[2]
Other Conditions included in screening	trisomy 18, trisomy 13	trisomy 18, open spina bifida	trisomy 18, open spina bifida	trisomy 18, open spina bifida	trisomy 18, trisomy 13, sex chromosome aneuploidy, fetal sex. 22q

**False positive - A result that indicates that a given condition is present when it is not. Example: Screen result is positive for Down syndrome, but the baby does not have Down syndrome.*

*** False negative - A result that appears negative when it should not be negative. Example: Screen result is negative for Down syndrome, but the baby has Down syndrome.*

	Chorionic Villus Sampling	Amniocentesis
Procedure Description	Sample of chorionic villi, cells that form the placenta, are removed under ultrasound guidance either using a catheter through the cervix or using a needle through the abdomen. The placental cells are grown and studied in a laboratory.	Under ultrasound guidance, a needle is inserted into the abdomen and into the amniotic sac. Amniotic fluid is taken from around the fetus which contains fetal cells. The fetal cells are grown and studied in a laboratory.
Gestational Age	10-13 weeks	Typically 15-20 weeks, but can be done later
Risk of Miscarriage	<0.5%	0.1%-0.3%
Results TAT	7-14 days for conventional karyotype	7-14 days for conventional karyotype

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Why Choose Harmony?

The Harmony prenatal test is a screening test. It can tell if there is a high or low chance for the common chromosomal conditions: Down syndrome (trisomy 21), trisomy 18 and trisomy 13. It can also tell the chance of having an extra or missing sex chromosome (X or Y), and the sex of your baby.

Three Simple Steps to Clarity

1
A blood sample is taken at 10 weeks or later in pregnancy.

2
Your sample is sent to the laboratory where it is analyzed.

3
Results are sent to your healthcare provider within 3-5 days from sample receipt.

For assistance email **sjc.clientservices@roche.com** or call **1-855-927-4672.**

The Harmony prenatal test measures a woman's risk of carrying a baby with chromosomal conditions such as Trisomies 21, 18 and 13. If your test indicates a high probability result, talk with your healthcare provider about what further diagnostic testing is needed to confirm the result. The Harmony Prenatal Test was developed by Ariosa Diagnostics, a laboratory certified by the federal government to perform highly complex tests. This test is a lab–developed test and has not been reviewed or approved by the FDA.

1. ACOG Practice Bulletin No. 163, May 2016.
2. Stokowski R, Wang E, White K et al. *Prenatal Diagn.* 2015;35(12):1243-1246
3. ACOG Practice Bulletin No. 162, May 2016
4. American Pregnancy Association, http://americanpregnancy.org/prenatal-testing/chorionic-villus-sampling Accessed November 9, 2018
5. American Pregnancy Association, http://americanpregnancy.org/prenatal-testing/amniocentesis Accessed November 9, 2018

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