Glossary of Pregnancy Terms

Abdominal Wall Defects (AWD) – Developmental defects involving the intestines and other organs that form outside the body.

Anencephaly – Anencephaly refers to an incomplete development of the brain that usually results in death.

Amniocentesis – A small amount of amniotic fluid is removed by a needle and is sent to test for chromosomal abnormalities such as Down syndrome and Trisomy 18. Amniotic fluid also screens for neural tube defects such as spina bifida.

Chorionic Villus Sampling (CVS) – This test may be offered at 10-14 weeks of pregnancy. A small number of cells are taken from the placenta and are diagnostic for Down syndrome and Trisomy 18. The advantage over amniocentesis is that it is performed earlier in pregnancy. On the negative side, CVS does not detect neural tube defects.

Detailed or Level II Ultrasound – A specialized ultrasound that includes basic information as well as detailed anatomical information about the fetus in the second trimester. It is recommended for women who will be 35 years or older at delivery, Screen Positive with the Full Integrated or Serum Integrated Screen or who have other high-risk indications. A Level II ultrasound is always performed with an amniocentesis and is performed at a Prenatal Diagnosis Center.

Diagnostic Test – CVS and amniocentesis are invasive tests that obtain amniotic fluid or placental tissue to grow chromosomes from the fetus. The test can tell if the fetus actually has a specific birth defect. Screening tests estimate the risk of certain birth defects.

Down Syndrome – Down syndrome is a chromosome abnormality that causes mental retardation and certain types of birth defects. It is due to an extra copy of chromosome 21, so that, three copies (trisomy) versus the normal two copies of this particular chromosome are present. Down syndrome affects approximately one in every 800 newborns. The chance of having a pregnancy affected with Down syndrome increases with increased maternal age. Women age 35 years and older are more likely to have a child affected with Down syndrome.

First Trimester Testing or Preliminary Risk Assessment – A blood test is drawn between 10 weeks and 13 weeks and 6 days of pregnancy and combined with a nuchal translucency (NT). A positive test results in referral to a Prenatal Diagnosis Center.

Another option is having the second blood test at 15-20 weeks to complete the Full Integrated Screen.

Full Integrated Screen – This combines the First Trimester Screening (blood test and NT) with a second trimester blood test to detect 90 out of 100 with Down syndrome, 81 out of 100 with Trisomy 18, 97 out of 100 with anencephaly, 80 out of 100 with open spina bifida, 85 out of 100 with abdominal wall defects and 60 out of 100 with SLOS.

Genetic Counseling – A genetic counselor reviews test results and family medical history. The counselor explains diagnostic tests, which may be offered.

Neural Tube Defects (NTD) – During the first 5 weeks of fetal development, the neural tube develops into the brain and spinal cord. Abnormalities in development may cause spina bifida or anencephaly.

Nuchal Translucency (NT) – An ultrasound preformed between 11 weeks 2 days and 14 weeks by a perinatologist to measure the back of the fetus' neck. This measurement helps screen for Down syndrome and Trisomy 18. It is used in conjunction with two blood tests to complete the California Full Integrated Screening. Check with your insurance company to determine your benefits.

Prenatal Diagnosis Center – A center that offers genetic counseling, diagnostic testing and detailed ultrasound for screen positive results. Obstetrix Medical Group (408) 371-7111 is the local diagnosis center.

Prenatal Screening Program – The California screening program offers Serum Integrated Screening. With a Screen Positive result, the California Prenatal Screening Program includes referral to a Prenatal Diagnosis Center for the same fee.

Prenatal Screening Test – Screening tests offer risk assessment to determine whether further diagnostic tests should be done. These tests cannot detect 100% of birth defects.
Quad Marker Screen – One blood specimen drawn between 15 and 20 weeks of pregnancy that gives a risk assessment for detection of 80 out of 100 Down syndrome, 67 out of 100 Trisomy 18, 97 out of 100 anencephaly, 80 out of 100 open spina bifida, 85 out of 100 abdominal wall defects and 60 out of 100 SLOS.

Rh Incompatibility – This is due to the mother having Rh negative antibody in the blood and the father of the baby having Rh positive antibody in his blood. If the baby has Rh positive blood type from the father, it can cause the mother to produce an antibody response against the baby. This is prevented by the mother receiving Rhogam after amniocentesis, at 28 weeks and again after delivery.

Rhogam – Rhogam is a shot given to Rh-negative mothers to prevent Rhesus disease in the newborn. IgG antibody (Rhogam) binds to fetal cells in the maternal circulation to prevent the mother from producing antibodies that could harm subsequent pregnancies.

Risk Assessment – An estimate of certain birth defects obtained with the Prenatal Screening Program.

Serum Integrated Screen – Two blood specimens drawn (first and second trimester) to detect 85 out of 100 with Down syndrome, 79 out of 100 with Trisomy 18, 97 out of 100 with anencephaly, 80 out of 100 with open spina bifida, 85 out of 100 with abdominal wall defects and 60 out of 100 with SLOS.

Screen Negative – The screening result shows that the screed for abnormality is unlikely. California reports risk of 1 in 100 or less to be negative. This does not guarantee that there are no birth defects.

Screen Positive – If the test shows a "positive" of 1 in 200 chance of having a baby with Down syndrome, the program authorizes follow-up services at a Prenatal Diagnosis Center which includes genetic counseling, a detailed ultrasound, CVS and amniocentesis. Obstetrix Medical Group offers genetic counseling and diagnostic testing (CVS or amniocentesis) to anyone who screens less than 1 in 1000 chance of Down syndrome or Trisomy 18. A positive screen does not always mean that there is a birth defect. Most women will have normal follow-up diagnostic tests.

SLOS or Smith-Lemli-Opitz syndrome – A very rare metabolic defect in which babies cannot make cholesterol normally and results in mental retardation and physical defects. Screen positive results for SLOS can also indicate increased chances of other congenital abnormalities and fetal demise.

Spina Bifida – When there is an opening in the spine, it is called spina bifida and can cause paralysis in the lower extremities as well as loss of bowel and bladder function.

Trisomy 18 – Trisomy 18 is a fatal chromosome abnormality that causes multiple birth defects and profound mental retardation. Few Trisomy 18 infants survive into childhood. Trisomy 18 results when the fetus has three, instead of the normal two, copies of chromosome 18. Like Down syndrome, the chance of an increased risk for fetal abnormality is determined by the test and then genetic counseling, ultrasound examination, and when needed, amniocentesis will aid in the diagnosis. Having a pregnancy affected with Trisomy 18 increases with increased maternal age.

Ultrasound – A device known as a transducer is used to direct high frequency sound waves to visualize the developing baby. The sound waves create an image of the baby's features and can determine growth and development of the baby.