Prenatal Laboratory Testing

First Trimester Blood Tests

Routine blood tests - Hepatitis B, HIV, RPR (state mandated syphilis test), Rubella (German measles), Blood type and antibody screen, CBC (Complete blood count for anemia), TSH (hypothyroidism), and Urinalysis and Urine culture.

Additional optional tests - Toxoplasmosis, Cystic fibrosis testing, Tay-Sachs, Fragile X, Gaucher’s disease screening, Sickle cell, Varicella (Chicken pox), Hemoglobin electrophoresis.

Genetic Testing: California Prenatal Screening Program –
www.cdph.ca.gov/Programs/pns

Screening Test Options: Screening ESTIMATES the risk of certain birth defects.

Quad Marker Screening (single blood test 15-20 weeks)
Serum Integrated Screening (two blood tests 10-13 6/7 weeks and again 15-20 weeks, no nuchal translucency ultrasound)
Full Integrated Screening (serum integrated blood tests and NT ultrasound)
First trimester Preliminary Risk Assessment (previously called nuchal translucency screening – includes first trimester blood test and NT ultrasound) – preliminary risk assessment for chromosomal abnormalities in the first trimester. This preliminary risk will be revised when the second trimester blood specimen is received.

First Trimester Preliminary Risk Assessment (10 – 13 6/7 weeks gestation) – This is an optional non-invasive assessment of the baby’s risk for Down’s syndrome. The test includes both an ultrasound and blood work that you can complete prior to your ultrasound appointment. Please schedule the NT ultrasound at Obstetrix Medical Group (408) 371-7111 if you plan to have the ultrasound associated with the Fully Integrated Screening.

<table>
<thead>
<tr>
<th>Detection Rates</th>
<th>Trisomy 21</th>
<th>Trisomy 18</th>
<th>Anencephaly</th>
<th>Open spina bifida</th>
<th>AWD</th>
<th>SLOS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Quad Marker Screening</td>
<td>80%</td>
<td>67%</td>
<td>90%</td>
<td>80%</td>
<td>85%</td>
<td>60%</td>
</tr>
<tr>
<td>Serum Integrated Screening</td>
<td>85</td>
<td>79</td>
<td>97</td>
<td>80</td>
<td>85</td>
<td>60</td>
</tr>
<tr>
<td>Fully Integrated Screening</td>
<td>90</td>
<td>81</td>
<td>97</td>
<td>80</td>
<td>85</td>
<td>60</td>
</tr>
<tr>
<td>First Trimester blood test plus NT scan</td>
<td>75</td>
<td>59</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
</tr>
</tbody>
</table>

Definitions in the California Prenatal Screening Program Booklet

Trisomy 21 – Down Syndrome
Trisomy 18 – associated with mental retardation and physical defects
NTD – Neural Tube Defects: anencephaly: absence of the brain; spina bifida: opening in the spine
AWD – Abdominal Wall Defects
SLOS – Smith-Lemli-Opitz syndrome, SCD – cannot make cholesterol normally – associated with mental retardation, may be associated with congenital abnormalities and fetal demise.

The state considers a result of 1 in 100 to be indication for further testing and will report a “negative” result. Obstetrix Medical Group and Los Olivos physicians recommend genetic
counseling and possible diagnostic testing for a result of 1 in 1000 or less. Traditionally, women over 35 have a 1 in 350 chance of having a baby with Down Syndrome.

**Second Trimester Testing**

*Diagnostic Testing if Screen “Positive”*

**Diagnostic Test Options:** Diagnostic tests can tell whether or not a fetus actually has a specific birth defect.

**Genetic counseling, Level II ultrasound and genetic amniocentesis** -
If you will be over 35 at delivery, have a history of an inherited genetic condition or screen “positive” through one of the above screening tests, we recommend genetic counseling. CVS, Level II ultrasound and amniocentesis may be offered. These tests are usually done at Obstetrix Medical Group (371-7111).

**Ultrasound** - If your physician recommends an ultrasound, these are usually performed between 18 and 20 weeks of pregnancy. You need to schedule this with your physician’s receptionist at Los Olivos (if under 35 years of age) or at Obstetrix Medical Group (if 35 years of age – Level II)

**Glucola (between 24-28 weeks) and CBC** - You will be asked to take the one-hour glucola test between 24 and 28 weeks of your pregnancy to screen for gestational diabetes. As a screening test, it is used to determine which patients are at risk for gestational diabetes and need the 3-hour glucose tolerance test. See page 23.

**Rhogam** - If you are Rh negative, your partner’s blood type becomes important. If he is Rh positive, you will need Rhogam to prevent Rh incompatibility. Rhogam is administered as an injection after amniocentesis, at 28 weeks of your pregnancy and after delivery if the baby is Rh positive.

**Third Trimester Testing**

**Group B Strep culture (36 weeks)** – This is a vaginal and perineal culture that tests for a bacteria that the baby does not normally have immunity to and may cause an infant infection.

**Ultrasound** – Your physician may recommend additional ultrasounds to check for fetal growth, fluid and position. It is very difficult to see anatomy in the third trimester.