

## AFP Only Patient Information

**This test is offered to Los Olivos Patients as a supplement to the Nuchal translucency screening test which has already been completed. If you are over 35 or are scheduled for a Level II ultrasound, you do not need this test. If you are under 35 and have completed the Nuchal translucency test, please read the following and sign the consent with your physician. You will have the test drawn at a laboratory between 15 and 20 weeks to test for the following birth defects.**

### **NTD/SLOS Only Screening: Second Trimester Screening For Open Neural Tube Defects and Smith-Lemli-Opitz Syndrome**

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The **California** Expanded AFP Screening Program offers an option for a second trimester blood test that screens **only** for Open Neural Tube Defects (NTD) and Smith-Lemli-Opitz Syndrome (SLOS). **This NTD/SLOS Only Screening is only for pregnant women who have already had CVS, Preimplantation Genetic Diagnosis for chromosome abnormalities, or First Trimester Screening AND who do not want additional screening for chromosome abnormalities.**

**Chorionic Villus Sampling (CVS)** is a test done on cells from the placenta between 10 and 12 weeks of pregnancy. It is a diagnostic test for chromosomal abnormalities in the fetus such as Down syndrome and Trisomy 18.

**Preimplantation Genetic Diagnosis (PGD)** is a test done on embryos before implantation during In Vitro Fertilization. When PGD is done to detect chromosome abnormalities, it can diagnose Down syndrome and Trisomy 18.

**First Trimester Screening** is a special ultrasound and blood test combination, usually done between 9 and 13 weeks of pregnancy. It is an early screening test for Down syndrome and Trisomy 18.

Since none of these tests are used to help detect NTD, Abdominal Wall Defects, or SLOS, there is a **Second Trimester blood test that screens just for these birth defects.** A woman who has had CVS, PGD for chromosome abnormalities, or First Trimester Screening can choose to have **NTD/SLOS Only Screening**, which **does not include** results for Down syndrome and Trisomy 18. Women who have had any of these first trimester tests should discuss their options for **NTD/SLOS Only Screening** with their clinician or a genetic counselor.

#### **What Are The Birth Defects That May Be Detected With NTD/SLOS Only Screening?**

**Open Neural Tube Defects (NTD)**, including spina bifida and anencephaly, are birth defects caused by incomplete closing of the Neural Tube (spinal cord) during development. Spina bifida may lead to life long paralysis, but can be helped with special treatments. Anencephaly is caused by an undeveloped brain and is always fatal.

**Abdominal Wall Defects (AWD)** are problems involving the baby's abdomen and intestines.

Surgery after birth often corrects the defect. Abdominal wall defects may be identified during testing for NTD.

**SLOS (Smith-Lemli-Opitz Syndrome)** is a problem with cholesterol production by the fetus. It causes mental retardation and can cause heart, hearing and other problems.

### **The Blood Test for NTD/SLOS Only Screening**

The blood test for NTD/SLOS **Only** Screening is done between 15 and 20 weeks of pregnancy. It tests for levels of Alpha-fetoprotein (**AFP**), and the hormones human chorionic gonadotropin (**hCG**) and unconjugated estriol (**uE3**) which are produced when a woman is pregnant. The test results show what the chances are that the woman is carrying a fetus with an Open Neural Tube Defect or with SLOS. Test results for NTD/SLOS **do not include** results for chromosome abnormalities such as Down syndrome and Trisomy 18.

If the result is “Screen Negative,” follow-up tests through the Program are not considered necessary. If the result is “Screen Positive,” the Program will pay for Genetic Counseling and follow-up tests at a State- approved Prenatal Diagnosis Center. A “Screen Positive” result does not mean that there is a birth defect –it means that follow-up tests are indicated, because the risk for a birth defect is higher than usual. Most women with “Screen Positive” results will have normal follow-up tests and healthy babies.

Not every birth defect will be detected. The Expanded **AFP** Program detects about 80% of spina bifida, 97% of anencephaly, 85 % of abdominal wall defects, and 60% of SLOS. The blood test costs \$162 and is covered by most insurance companies, once the patient has paid any co-payments or deductibles.

**If you choose to have NTD/SLOS Only Screening, please sign the consent form.**

Information provided by  
California Department of Health Services  
Genetic Disease Branch  
850 Marina Bay Parkway Richmond, CA 94804  
866-718-7915 [www.dhs.ca.gov/gdb](http://www.dhs.ca.gov/gdb)