



[A **technical** review]

First Trimester Serum Screening

Early Screening for Down Syndrome

Introduction

Maternal serum screening is a way to find out information about the developing fetus without invasive procedures. The goal of maternal serum screening is to identify a group of patients who have an increased risk for a fetal disorder and offer that group more specific diagnostic testing. Maternal serum screening has traditionally been offered in the second trimester of pregnancy, but scientific advances now allow health care providers to offer screening for Down syndrome in the first trimester of pregnancy.

Combined Analytes

First trimester screening (FTS) for Down syndrome and trisomy 18 involves combining maternal serum levels of total human chorionic gonadotropin (hCG), pregnancy-associated plasma protein A (PAPP-A), and dimeric inhibin A (DIA) with maternal age risk and a fetal nuchal translucency (NT) measurement.^{1,3} The NT is a collection of fluid behind the fetal neck as visualized by ultrasound.² An increased NT measurement is associated with several fetal conditions, including Down syndrome.² Because this measurement is critical to the interpretation of results, consistent with recommendations of the American College of Obstetricians and Gynecologists (ACOG),² LabCorp will not use NT measurements or provide risk assessments for specimens

submitted without sonographer NT credentialing/certification information.

First trimester screening identifies 86% of Down syndrome and 75% of trisomy 18 pregnancies.^{1,3,4} Prenatal testing options following a positive first trimester screen include chorionic villus sampling and amniocentesis.² Genetic counseling is recommended.² A comparison of screening options for Down syndrome and trisomy 18 is found in Table 1 below.

Limitations

It is important to note that first trimester screening only provides a risk for Down syndrome and trisomy 18. First trimester screening cannot detect other chromosome abnormalities or birth defects nor does it screen for open neural tube defects (ONTD); therefore, women who choose first trimester screening should still receive maternal serum AFP screening for ONTD between 16 and 18 weeks of gestation.⁴

The LabCorp Advantage

- Verbal reports provided on positive results
- Board-certified genetic counselors and geneticists to assist with interpretation
- Specialized genetics customer service staff

Table 1.—Comparison of Screening Options for Down Syndrome and Trisomy 18^{1,3-5}

Test	Analytes	Gestational Age	Detection Rates		
			Down syndrome	Trisomy 18	ONTD
First Trimester Serum Screen	PAPP-A, total hCG, DIA (includes NT)	10-13 weeks	86% ¹	75% ³	N/A
AFP	AFP	15-23 weeks	N/A	N/A	80% ⁴
AFP X-tra	AFP, hCG, uE3	15-21 weeks	60% ⁴	60% ⁴	80% ⁴
AFP Tetra	AFP, hCG, uE3, DIA	15-21 weeks	75%-80% ⁵	60% ⁴	80% ⁴

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CPT 84702; 84163, 86336

Test Includes Human chorionic gonadotropin (hCG); pregnancy-associated plasma protein A (PAPP-A); and dimeric inhibin A (DIA) combined with client supplied maternal age and client determined fetal nuchal translucency (NT) measurement by a sonographer with NT credentialing/certification

Special Instructions The following information must be provided: patient's weight, patient's date of birth, fetal crown rump length (CRL) and nuchal translucency measurement (NT) and the date on which these measurements were taken. Also indicate relevant patient history (eg, previous Down syndrome pregnancy). Complete information is necessary to interpret the test. Patient information may be provided to the laboratory using the Maternal Prenatal Screening test request form (0900). Specimens must be collected before amniocentesis.

Specimen Serum

Volume 3.0 mL

Minimum Volume 1.0 mL

Container Gel-barrier tube

Collection Avoid hemolysis; send complete specimen in the original tube. Do **not** pour off.

Storage Instructions Refrigerate

Causes for Rejection Gross hemolysis; gross lipemia; quantity not sufficient for analysis; fetal nuchal translucency (NT) measurement by a sonographer without NT credentialing/certification

Use Screening test for Down syndrome (detects 86%), and trisomy 18 (detects 75%). First trimester screening cannot detect other chromosome abnormalities or birth defects nor does it screen for open neural tube defects (ONTD). Women who choose first trimester screening, therefore, should still receive maternal serum AFP screening for ONTD between 16 and 18 weeks of gestation.

Methodology PAPP-A and DIA by EIA, hCG by immunochrominometric assay (ICMA).

References

1. Wald NJ, Rodeck C, Hackshaw AK, Chitty L, Mackinson AM. First and second trimester antenatal screening for Down's syndrome: The results of the Serum, Urine and Ultrasound Screening Study (SURUSS). *J Med Screen*. 2003; 10:56-104.
2. American College of Obstetricians and Gynecologists. First-trimester screening for fetal aneuploidy. *Committee Opinion*. 2004; July (296).
3. Tul N, Spencer K, Noble P, Chan C, Nicolaides K. Screening for trisomy 18 by fetal nuchal translucency and maternal serum free beta-hCG and PAPP-A at 10-14 weeks of gestation. *Prenat Diagn* 1999;19:1035-1042.
4. American College of Obstetricians and Gynecologists. Maternal Serum Screening. *Committee Opinion #228*, Sept. 1996.
5. Haddow JE, Palomaki GE, Knight GJ, Foster DL, Neveux LM. Second trimester screening for Down's syndrome using maternal serum dimeric inhibin A. *J Med Screen*. 1998; 5(3):115-119.



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