

Overview

Useful For

Helpful to identify pregnancies at increased risk of having a child with Down Syndrome (DS), open neural tube defects (ONTD) and trisomy 18 (T18). This test is not diagnostic.

Profile Information

| Test Id | Reporting Name        | Available Separately | Always Performed |
|---------|-----------------------|----------------------|------------------|
| FPATI   | Patient Information   | No                   | Yes              |
| FMAS1   | Maternal Screen INT-1 | No                   | Yes              |

Special Instructions

- [PATIENT HISTORY FOR MATERNAL SERUM TESTING](#)

Method Name

Quantitative Chemiluminescent Immunoassay

NY State Available

Yes

Specimen

Specimen Type

Serum

Specimen Required

Specimen #1 collection must occur between 10 weeks, 0 days and 13 weeks, 6 days gestation. (If gestational age is based on Crown-Rump length (CRL), the specimen must be collected when the CRL is between 32.4 - 83.9 mm)

Draw blood in a plain red-top tube(s), serum gel tube is acceptable. Spin down and send 0.5 mL serum refrigerated in a plastic vial.

Separate from cells ASAP or within 2 hours of collection.

Note:

**Submit with order:** Patient's date of birth, current weight, number of fetuses present, patient's race, if the patient was diabetic at the time of conception, if there is a known family history of neural tube defects, if the patient has had a previous pregnancy with a trisomy, if the patient is currently smoking, if the patient is taking valproic acid or

carbamazepine (Tegretol), if this is a repeat sample, and the age of the egg donor if in vitro fertilization.

In addition to the above:

**If a NT measurement is performed:** the date of ultrasound, the CRL measurement, the nuchal translucency (NT) measurement and the name and certification number of the sonographer is required. NT must be measured when the CRL is between 38-83.9mm.

Or

**If no NT measurement is performed:** a due date or CRL measurement with the date of ultrasound is required.

The NT measurement must also be performed by an ultrasonographer that is certified by one of the following agencies: Fetal Medicine Foundation (FMF) or Nuchal Translucency Quality Review (NTQR).

Specimen Minimum Volume

0.3 mL

Reject Due To

|           |                          |
|-----------|--------------------------|
| Hemolysis | Mild reject/Gross reject |
| Other     | Plasma                   |

Specimen Stability Information

| Specimen Type | Temperature              | Time     | Special Container |
|---------------|--------------------------|----------|-------------------|
| Serum         | Refrigerated (preferred) | 14 days  |                   |
|               | Frozen                   | 90 days  |                   |
|               | Ambient                  | 72 hours |                   |

Clinical & Interpretive

Clinical Information

This test combines a first- and second-trimester specimen to screen low-risk pregnancies for Down syndrome (DS), open neural tube defects (ONTD) and trisomy 18 (T18).

Collection of two blood samples is required for this test. A first trimester ultrasound to measure the fetal nuchal translucency (NT) is optional (see special instructions).

Patient demographics and analyte/ultrasound measurements are used to calculate multiple of the median (MoM) values for each of the laboratory analytes and the NT. The pattern of the MoM values is used to calculate post-test risks of ONTD, DS and T18.

Markers used for assessment of risk include first-trimester PAPP-A with or without NT and second-trimester AFP, hCG, unconjugated estriol (uE3) and dimeric Inhibin A.

A DS risk of 1 in 110 or worse is reported as abnormal. This risk cutoff predicts a detection rate of 87 percent at a screen positive rate of 1.0%.

A T18 risk of 1 in 100 or worse is reported as abnormal. This risk cutoff predicts a detection rate of 90 percent at a screen positive rate of <0.5%.

ARUP uses a singleton AFP MoM cut off of  $\geq 2.5$ . If the interpretation is "high AFP," there is an increased risk of an ONTD in the pregnancy. This cutoff value predicts a detection rate of 80% at a screen positive rate of 1.5%. High AFP also occurs in unrecognized twin pregnancies and with underestimated gestational age.

Pregnancies at an increased risk for ONTD with an AFP MoM  $< 2.5$ , but a risk of 1 in 250 or worse, are also reported as abnormal. This is usually due to a family history of ONTD, the use of certain seizure medications by the patient during pregnancy, or the presence of maternal insulin-dependent diabetes, any of which increases a patients risk for ONTD.

An increased risk of congenital steroid sulfatase deficiency or Smith-Lemli-Opitz syndrome ( $uE3 \leq 0.14$  MoM) and poor fetal outcome ( $hCG \geq 3.5$  MoM) is reported as "see note".

**Reference Values**

An interpretive report will be provided.

Part 2 must be completed in order to receive an interpretable result.

If the second specimen is not received for sequential screening, the results are uninterpretable and no maternal risk will be provided.

**Interpretation**

The first specimen of an integrated Maternal Serum Screening is used to measure PAPP-A. A second sample must be submitted for a final interpretive report. Acceptable date ranges to draw the second samples will be provided in the Integrated-1 report. Final interpretive report will be available when the second specimen test results are complete.

**Cautions**

A screen interpreted as "normal" misses approximately 15% of Down Syndrome, 20% of open neural tube defects and 10% of trisomy 18 cases.

Abnormal results require follow-up with targeted ultrasound, genetic counseling and consideration of fetal diagnostic testing.

**Performance**

**Method Description**

PAPP-A is pregnancy associated plasma protein A and is a sequential immunoenzymatic assay that uses two monoclonal antibodies and external calibrators.

AFP and hCG are both measured using a non-competitive immunoassay that uses one antibody to capture the protein to a solid phase, another antibody to detect the protein, and external calibrators.

The estriol assay is a solid phase competitive immunoassay that uses an anti-estriol polyclonal antibody, labeled estriol, a solid phase antibody directed against the estriol antibody, and external calibrators.

Inhibin-A is measured using a non-competitive microtiter immunoassay that uses a detection antibody to subunit *α*, a capture antibody to inhibin subunit *BA*, and external calibrators.

Calculation of post-test risks uses a multivariate log Gaussian model. Risk estimates for DS and T18 are influenced strongly by maternal age.

PDF Report

No

Day(s) Performed

Sunday through Saturday

Report Available

6 to 10 days

Performing Laboratory Location

ARUP Laboratories

Fees & Codes

Fees

- Authorized users can sign in to [Test Prices](#) for detailed fee information.
- Clients without access to Test Prices can contact [Customer Service](#) 24 hours a day, seven days a week.
- Prospective clients should contact their account representative. For assistance, contact [Customer Service](#).

CPT Code Information

84163

LOINC® Information

| Test ID | Test Order Name                 | Order LOINC® Value |
|---------|---------------------------------|--------------------|
| FFMSS   | Maternal Serum Screen INT, Sp-1 | Not Provided       |

| Result ID | Test Result Name         | Result LOINC® Value |
|-----------|--------------------------|---------------------|
| Z5958     | PAPP-A Maternal          | 48407-1             |
| Z5959     | Nuchal Translucency (NT) | 12146-7             |

Test Definition: FFMSS

Maternal Serum Screening, Integrated,  
Specimen #1, PAPP-A, NT

|       |                                     |         |
|-------|-------------------------------------|---------|
| Z5960 | Nuchal Translucency (NT), Twin B    | 12146-7 |
| Z5961 | Maternal Screen Interpretation      | 49586-1 |
| Z5962 | Maternal Age At Delivery            | 21612-7 |
| Z5963 | Maternal Weight                     | 29463-7 |
| Z5964 | Estimated Due Date                  | 11778-8 |
| Z5965 | Gestational Age Calculated at Coll. | 18185-9 |
| Z5966 | Dating                              | 21299-3 |
| Z5967 | Number of Fetuses                   | 11878-6 |
| Z5968 | Maternal Race                       | 21484-1 |
| Z5969 | Smoking                             | 64234-8 |
| Z5970 | Family History of Aneuploidy        | 32435-0 |
| Z5971 | Specimen                            | 19151-0 |
| Z5972 | Crown Rump Length                   | 11957-8 |
| Z5973 | Crown Rump Length, Twin B           | 11957-8 |
| Z5974 | Sonographer Certification Number    | 49089-6 |
| Z5975 | Sonographer Name                    | 49088-8 |
| Z5976 | Ultrasound Date                     | 34970-4 |
| Z5977 | Best date to draw sample nmb 2 by   | 33882-2 |
| Z5978 | EER Maternal Serum, Integrated, Sp1 | 11526-1 |