RWJ OB/GYN associates

Screening for Down Syndrome

Down syndrome, also called Trisomy 21, is a condition which affects a child's development both mentally and physically. It may result in mental retardation, abnormal features of the face, and medical problems such as heart defects. A screening test can give information about a pregnant woman's risk of having a baby with Down syndrome.

PLEASE READ THE FOLLOWING AND CHECK THE BOX WHICH CORRESPONDS TO YOUR CHOICE.

☐ **1) FIRST TRIMESTER SCREENING (PAPP-A, hCG & Nuchal Translucency)**

This is done **BETWEEN 11 TO 14 WEEKS** of pregnancy. A **BLOOD TEST** and **ULTRASOUND** examination which measures the thickness of the back of the neck of the fetus are combined to detect 82 to 87% of Down syndrome cases.

☐ **2) SECOND TRIMESTER SCREENING (AFP, Estriol, hCG & Inhibin A)**

If your prenatal care begins **AFTER 14 WEEKS** (too late for the First Trimester Screening), this **BLOOD TEST** can be performed at 15 to 20 weeks and detect 81% of Down syndrome cases.

☐ **3) NON-INVASIVE PRENATAL TESTING USING CELL-FREE FETAL DNA (HIGH RISK PATIENTS)**

Cell-free fetal DNA is released into the mother's blood and can be detect through a simple blood test. It can be analyzed to identify about 99% of Down syndrome affected fetuses.

This prenatal test is offered to women with pregnancies of at least 10 weeks and **ONE OF THE FOLLOWING CONDITIONS**:

- Maternal age of 35 or older at delivery
- Fetal ultrasound findings suggest an increased risk
- History of prior pregnancy with Down syndrome
- Abnormal first or second trimester screening
- Parents with balanced Robertsonian translocation with increased risk of Down syndrome

Although this test has a high detection rate, it is still a **SCREENING TEST** and a negative test does not ensure an unaffected pregnancy.

☐ **4) I am not interested in any screening for Down syndrome.**

NAME:_____ SIGNATURE:_____ DATE:_____

***If the results of any screening tests or other factors raise concerns about your pregnancy, diagnostic tests (invasive testing, such as amniocentesis or chorionic villus samplings) can be done to provide more information.