

## *What to Expect . . .* ROUTINE \* **TESTING** TIMELINE & OVERVIEW

(\* additional testing may be required for high risk pregnancies)

**FIRST VISIT:** (see note 1: additional information about Carrier Screening at the end)

- **Blood tests:**
  - blood type
  - verify Rh factor
  - diseases, conditions, and immunities that could affect your baby, pregnancy and delivery,
  - This does not screen for genetic problems,
  - This will screen for SOME genetic carrier states (**see note 1**) in the mother that could effect the baby.
- **PAP:** testing for abnormal cells and STIs
- **Ultrasound:** to confirm due date and viability of your pregnancy

### **EVERY VISIT:**

- **blood pressure,**
- **weight**
- **measure size of the uterus** by measuring OR via an ultrasound per the schedule
- **urine sample** to check for too much protein(preeclampsia) or sugar (gestational diabetes)

### **SCREENING FOR DOWN SYNDROME AND OTHER CHROMOSOMAL ABNORMALITIES**

Screening for the more common chromosomal abnormalities (trisomies 13, 18 and 21) is optional, but is offered to all pregnant women regardless of age.

In most cases the results are reassuring, but some show an elevated risk. This information can be helpful in determining the need for additional testing, as well with management during the pregnancy and after delivery.

It is important to remember that these tests are only screening tests and may carry both false positive or negative results.

**There are three initial screening options:**

### **1. NONINVASIVE PRENATAL TEST (NIPT)**

- This blood test can be drawn in our office as early as 10 weeks.
- The test **evaluates fetal DNA in the mother's blood**
- The detection rate is approximately 98% and false positive rate of 1%.
- May not be covered by insurance, especially if you are younger than 35 years

### **2. 11 ½-13 ½ weeks: NUCHAL TRANSLUCENCY NT.**

- The test involves a finger stick blood test from the mother and an ultrasound that measures the Nuchal Fold "translucency" (NT) in the neck of the fetus.
- **We will give you a referral to a perinatologist for this ultrasound**
- **Most often this test is covered by insurance**
- **You can gain VERY VALUABLE GENETIC information about your baby**
- Detection rate for Down Syndrome is between 82-90%.
- Can also detect other unrelated, but potentially serious abnormalities.

### **3. 16-24 weeks: QUAD SCREEN**

- Done in office
- this test is a blood drawn from the mother to measure certain hormone levels from the baby and placenta which gives a statistical risk factor for genetic abnormalities.
- Detection rates are 80-85% with a higher false positive than the NT.

**For High Risk OB patients:**

- women who are 35 and above
- those who have a pregnancy at increased risk for trisomies based on prior pregnancy outcomes
- those who have a pregnancy at increased risk for trisomies based on abnormal screening test results or abnormal ultrasound results. (If a Nuchal Translucency shows that your baby has a higher than usual risk, we would recommend the NIPT.

*For any woman with abnormal screening test results, we recommend consultation with a high risk pregnancy specialist for further evaluation and to discuss possible confirmatory testing through amniocentesis.*

**ROUTINE TESTS:**

### **16-20 weeks: AFP**

Blood is drawn from the mother looking for increased risk of abnormalities of the Brain and Spine (spina bifida or Neural Tube Defects).

**Anytime after 11 weeks: Doppler** to hear the baby's heartbeat

**20 weeks: Ultrasound** to assess fetal anatomy

**20 weeks: Glucose Screening Test** for patients at higher risk for diabetes.

Higher Risk Patient: Those with diabetes in the family, maternal age 40 and over, increased BMI, previous large baby over 9 pounds and history of gestational diabetes.

**28 weeks: Glucose Screening Test** for all patients:

This screening test determines if your blood sugar is too high.

This might indicate Gestational Diabetes, which is treatable.

- This is a one hour test and involves drinking a sweet drink, waiting an hour and having your blood drawn and tested.
- If the blood sugar level is too high it will be necessary to have the 3 hour **Glucose Tolerance Test**.

### **Glucose Tolerance Test**

This is a three hour version of the Glucose Screening Test to confirm the results of the Screening test and make sure you get the best medical care for you and your baby.

- This is a three hour test and involves a fasting blood sugar level and then drinking a sweet drink, waiting and having your blood drawn hourly an additional three times.

**30 and 36 weeks: Ultrasound**

to monitor baby's growth, check amniotic fluid levels and baby's position

**36 weeks: Group B strep test**

a bacteria in the vagina that is harmless to you but may cause an infection in the baby during a vaginal delivery.

- the vagina and anal area are swabbed at the 36 week OB visit with a pelvic exam.

## **NOTE 1: CARRIER SCREENING FOR GENETIC ABNORMALITIES**

A “**CARRIER**” is a person who has a gene for a medical condition but does not have the actual illness. **If both parents are CARRIERS** of a specific gene for a specific illness, there is a **1 in 4 chance that their child** will have that illness or condition.

- Screening is offered for certain diseases to detect carrier status by blood tests from the mother.
- As insurance coverage varies, there may be additional charges for this testing.

### **■ CARRIER SCREENING INCLUDED IN ROUTINE SCREENING AT YOUR INITIAL VISIT:**

Blood Related Diseases include:

- Sickle Cell
- Thalassemia
- Hemoglobin C

### **■ CARRIER SCREENING OFFERED AND RECOMMENDED FOR ALL AT YOUR INITIAL VISIT:**

- Cystic Fibrosis (CF)
- Fragile X
- Spinal Muscular Atrophy

### **■ CARRIER SCREENING RECOMMENDED FOR PATIENTS WHO ARE OF ASHKENAZI JEWISH DESCENT IN ADDITION TO CF:**

- Tay Sachs
- Bloom syndrome
- Canavan Disease
- Niemann Pick Disease
- Fanconi Anemia
- Gauchers Disease
- Familial Dysautonomia
- Mucopolysaccharidosis.

### **■ Additional testing is available from the outside genetics labs we use.**

**This can detect carrier states for over 100 additional rare diseases.**

We are happy to discuss this testing option further if you are interested or have additional questions.