



## **GENERAL INFORMATION ABOUT THE UNITED SCREENING APPROACH**

### **What is the United Screening Approach?**

The United Screening Approach (USA) is a combined first and second trimester prenatal screening program that provides patient-specific risk assessments for Down syndrome, trisomy 18, trisomy 13, Turner syndrome, triploidy, and sex chromosome abnormalities (XXX, XXY, XYY). It also provides an overall risk for any chromosome abnormality in the pregnancy.

The United Screening Approach combines information from the First Trimester Screen (nuchal translucency, free beta hCG, and PAPP-A) with information from the second trimester maternal serum screen (AFP, hCG, estriol, and inhibin). It is currently the most reliable chromosome screening program available, and detects at least 97% of cases of Down syndrome with an overall false positive rate of 5%.

### **Who should be screened?**

Women age 35 and older and those with a family history of a chromosomal condition are more likely to have an affected pregnancy. However, most cases occur in women younger than 35 with no family history of chromosomal conditions. The USA is offered to all women.

### **At what gestational age should the United Screening Approach be performed?**

The First Trimester Screen consists of two parts: a blood test that can be done between 9-13 weeks, and an ultrasound examination performed between 11-14 weeks in pregnancy. We generally schedule our patients for their blood draw during their 11<sup>th</sup> week in pregnancy and for their ultrasound during their 12<sup>th</sup> week. *The First Trimester Screen is performed at Northwestern Reproductive Genetics and can be scheduled by calling our office at 312-981-4400.*

The second trimester maternal serum screen (also called the “quad screen” or “triple screen”) is a simple blood test that can be performed between 15-22 weeks in pregnancy, but is ideally drawn between 15-18 weeks. *The second trimester screen is performed at your obstetrician’s office, and is usually part of one of your regularly scheduled appointments.*

Once your second trimester screen result is complete, your obstetrician’s office will fax the result to Northwestern Reproductive Genetics. Your USA result will then be calculated using the information from your First Trimester Screen combined with the information from your second trimester screen. A USA report will be generated and will be faxed to your obstetrician and sent to you by mail. If your result is “screen positive” (i.e. high risk), you will be called to discuss the results as well as your options for further testing.

### **What are the advantages of doing the United Screening Approach?**

- For those shown to be at high risk, the United Screening Approach gives the patient and physician time to consider follow-up diagnostic options, such as amniocentesis
- For those shown to be at low risk, the United Screening Approach reduces the use of amniocentesis, a procedure that increases risk to the patient and fetus
- The United Screening Approach is safe, simple, and non-invasive

## **CONDITIONS INCLUDED IN THE UNITED SCREENING APPROACH**

### **Down syndrome**

Down syndrome, also known as trisomy 21, is a congenital disorder caused by the presence of an extra copy of chromosome 21, in which the affected person has mild to moderate mental retardation, characteristic physical appearance, and often has congenital malformations such as heart defects.

### **Trisomy 18**

Trisomy 18 is a congenital disorder caused by the presence of an extra copy of chromosome 18, occurring in about 1 out of 3,000 to 1 out of 6,000 live births. Trisomy 18 is generally fatal, with 50% of babies dying within the first week of life and an additional 40% dying within the first year of life. The disorder is characterized by profound mental retardation, heart defects, and central nervous system defects.

## **CONDITIONS INCLUDED IN THE UNITED SCREENING APPROACH (CONTINUED)**

### **Trisomy 13**

Trisomy 13 is a **congenital disorder** caused by the presence of an extra copy of chromosome 13, occurring in about 1 out of 10,000 live births. Trisomy 13 is generally fatal, with more than 80% of babies dying in the first month of life. The disorder is characterized by profound mental retardation, cardiac problems, and congenital abnormalities.

### **Turner syndrome**

Turner syndrome is a congenital disorder seen in females who have inherited only one X chromosome instead of the usual two X chromosomes. It occurs in approximately 1 out of 2,000 live births. The features of Turner syndrome are quite variable, including short stature and developmental delay. Many times, the diagnosis is suspected before birth due to certain ultrasound findings, and Turner syndrome can lead to miscarriage in many pregnancies. Sometimes the features are noticeable at birth; other times they are not. Girls and women with Turner syndrome can have such mild features that the diagnosis is not made until adolescence or adulthood.

### **Triploidy**

Triploidy is a congenital disorder caused by the presence of an entire extra set of chromosomes. Instead of the usual 46 chromosomes, pregnancies with triploidy have a total of 69 chromosomes. This chromosome composition is not compatible with survival. Almost every triploid pregnancy ends in loss by miscarriage or stillbirth. Rarely, babies are born with triploidy, but they generally do not survive beyond infancy.

### **Sex Chromosome Abnormalities (XXX, XXY, XYY)**

There are several different types of sex chromosome changes, including an extra or missing copy of the entire X or Y chromosome. Changes in the number of sex chromosomes result from an error in the formation of the egg or sperm cell. Most babies with changes in their sex chromosomes are healthy at birth, without serious birth defects. Some people with sex chromosome changes never develop symptoms or learn they have a sex chromosome abnormality. However, learning difficulties and fertility problems are more common in people with a change in their sex chromosomes. These problems generally cannot be identified prenatally or early in life.

*It is important to note that First Trimester Screening cannot detect all birth defects and genetic syndromes.*

## **UNDERSTANDING YOUR RESULTS**

### **What does a “screen negative” result mean?**

A screen negative result means that the risk for chromosome abnormalities is low enough that diagnostic testing (amniocentesis) is *not* indicated. The majority of pregnant women have a “screen negative” result. Although a “screen negative” result greatly reduces the risk for chromosome abnormalities, it does not completely rule out the possibility that the fetus is affected.

If your USA is “screen negative”, your obstetrician will continue to provide you with routine obstetrical care. This will likely include undergoing an anatomy scan or level II ultrasound at approximately 20 weeks.

### **What does a “screen positive” result mean?**

A screen positive result means that there is an increased risk for a chromosome abnormality in the pregnancy. A screen positive result does *not* mean that the fetus is affected with the condition, only that a pregnancy has an increased risk for the condition. Most women with “screen positive” results have healthy babies.

When the result is “screen positive”, the following options are available:

- Diagnostic testing by amniocentesis performed after 15 weeks
- Level II ultrasound performed at approximately 20 weeks

## **QUESTIONS?**

If you have any questions concerning this information, please do not hesitate to contact us at 312-981-4400.