

# Prenatal Diagnosis and Reproductive Genetics



## **Convenient Locations for Prenatal and Reproductive Genetics Care**

The University of Chicago Medicine's Ultrasound and Reproductive Genetics program offers personalized care and one-stop access to specialists at our Hyde Park campus and three other locations in the Chicagoland area.

#### **HYDE PARK**

#### Family Birth Center Comer Children's Hospital

5721 S. Maryland Ave. 3rd Floor Chicago, IL 60637

**Duchossois Center for Advanced Medicine (DCAM)** 

5758 S. Maryland Ave. 3rd Floor, Suite G Chicago, IL 60637

#### SOUTH LOOP

Center for Advance Care at South Loop 1101 S. Canal St. Level P1, Suite 202B Chicago, IL 60607

#### SCHERERVILLE

222 Indianapolis Blvd. Suite 200 Schererville, IN 46375

### TO MAKE AN APPOINTMENT Call **773-702-6118**

Visit UChicagoMedicine.org/high-risk-ob



## Our Prenatal Diagnosis and Reproductive Genetics Team



Ryan E. Longman, MD Director, Ultrasound Center Director, Fetal and Neonatal Care Center



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Bryanna Cox McCathern, MS, CGC Genetic Counselor



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#### **DIAGNOSTIC GENETIC TESTING**

There are a number of reasons to consider having a diagnostic fetal genetic test: your age, family history, an ultrasound finding, an abnormal blood test or simply because you want more information. If the fetus is found to have a genetic disorder, detailed counseling about the disorder and a discussion of reproductive options can follow. A prenatal diagnosis may also help the pediatricians who will care for the infant.

#### CHORIONIC VILLUS SAMPLING (CVS) (11 TO 13 WEEKS)

A chorionic villus sampling (CVS) procedure helps determine if there are chromosomal abnormalities or other genetic disorders in the fetus. Chorionic villi are tiny projections of placental tissue that have the same genetic material as the fetus. Using ultrasound technology, the physician guides either a needle through the abdomen or catheter through the cervix to obtain a small sample of the placenta. The small sample of placenta tissue is sent to the lab for testing.

#### AMNIOCENTESIS (15 WEEKS AND AFTER)

Amniotic fluid surrounds the fetus in the uterus. During amniocentesis, the physician inserts a long thin needle into the lower part of the abdomen below the navel. An ultrasound assists the physician in guiding the needle to a safe pocket of amniotic fluid, away from the baby. A small amount of fluid is removed, which contains living cells that have been shed from the baby. The fluid is then sent to the lab for testing.

#### SHOULD I HAVE GENETIC COUNSELING?

Genetic counseling can help you learn whether your baby is facing an increased chance of a genetic abnormality or birth defects and take the appropriate next steps. Our genetic counselors will review your family history and genetic test results so they can provide you with clear, condition-specific



genetic counseling to help you understand your baby's condition and plan of care.

## Services Offered

## PRENATAL ANEUPLOIDY SCREENINGS

Ultrasound exams and blood tests can safely determine whether your baby is at risk for genetic abnormalities or specific birth defects such as spina bifida. It's important to note that these routine screening tests only provide a probability that a condition exists. Results can help a couple decide if they want to pursue diagnostic genetic testing.

# FIRST TRIMESTER SCREENING (10 TO 14 WEEKS)

An ultrasound exam and blood test to provide information about the chances for your baby to be affected by either Down syndrome or trisomy 18 (a genetic abnormality of chromosome 18 which results in multiple birth defects).



### SECOND TRIMESTER QUAD SCREENING (15 TO 22 WEEKS)

A blood test, typically performed in your primary obstetrician's office, to measure the levels of four analytes produced by the pregnancy and found in the mother's blood. These analyte levels help calculate the chance of a baby being born with Down syndrome, trisomy 18 or other birth defects.

#### CELL-FREE FETAL DNA (cffDNA) SCREENING (10 WEEKS AND AFTER)

A blood test to give a risk assessment for Down syndrome, trisomy 18, trisomy 13 and sex chromosome abnormalities (extra or missing X or Y chromosomes). The cffDNA testing can be done as a primary screening test or following an abnormal first trimester or quad screening. It is not recommended for women whose previous screenings indicated a low risk (normal).

### WHAT HAPPENS AFTER A POSITIVE RESULT?

Our genetics team will review your screening results and answer all of your questions. Though most of the conditions detected by these screening tests are not inherited, information regarding your family and pregnancy history will be reviewed. In addition, diagnostic testing options such as chorionic villus sampling (CVS), amniocentesis and additional ultrasound exams will be discussed.

# State-of-the art screening and testing for pregnant women and their babies

UChicago Medicine provides the full range of prenatal screening, diagnostic genetic testing and genetic counseling:

- » High-resolution fetal imaging
- » 3D and 4D ultrasound
- » Fetal genetic screening tests
- » Chorionic villus sampling (CVS)
- » Amniocentesis
- » Fetal echocardiogram
- » Genetic consultation

