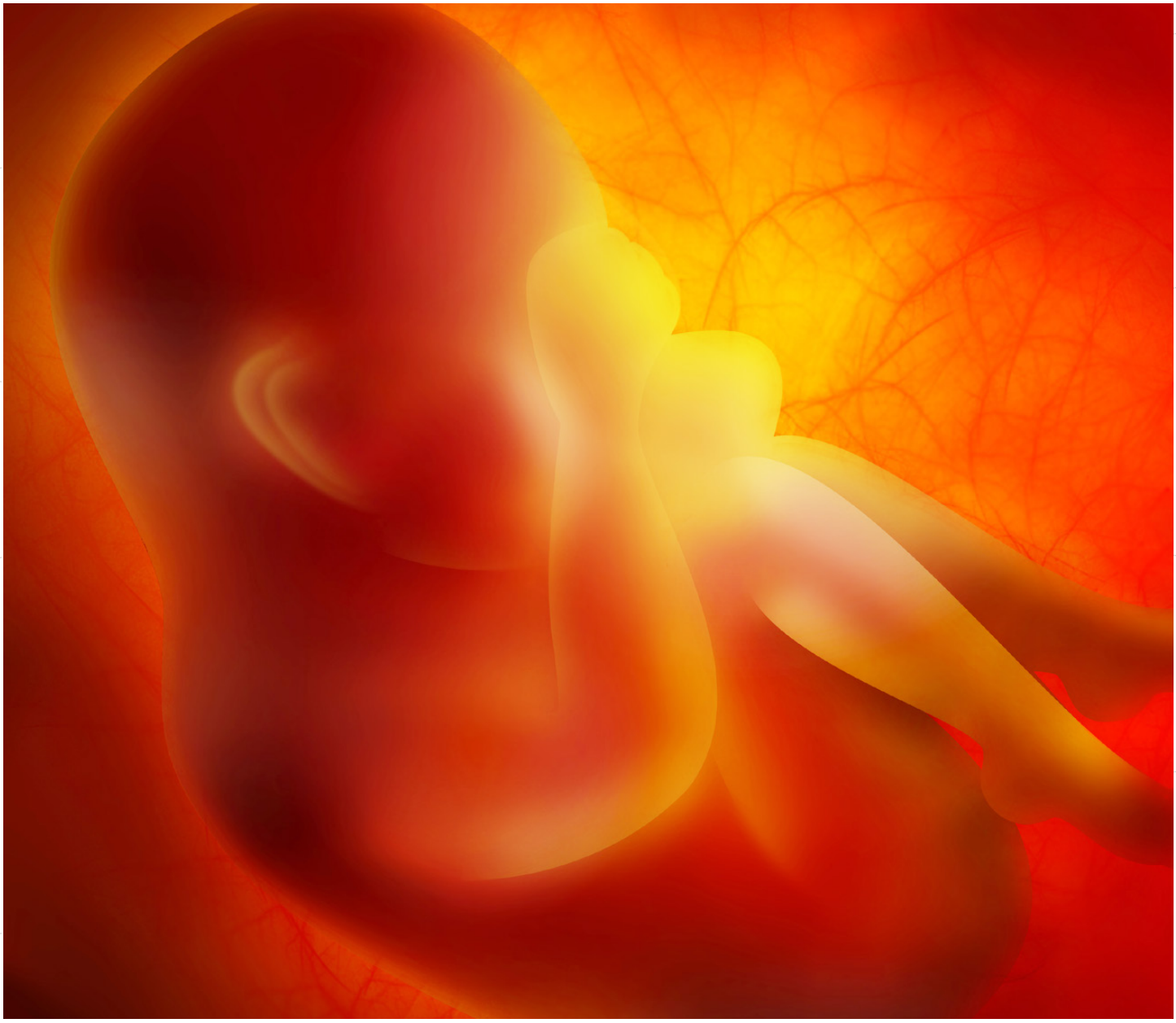
MAYO CLINIC
Mayo Medical Laboratories

PRENATAL TESTING

COMPREHENSIVE SCREENING AND DIAGNOSTIC TESTING FOR FETAL ABNORMALITIES



PRENATAL TESTING AT MAYO CLINIC

Prenatal testing includes both screening and diagnostic tests to identify certain birth defects and inherited disorders. Mayo Medical Laboratories offers a comprehensive test menu with more than 70 prenatal offerings from noninvasive screening to invasive diagnostic testing. We offer sequencing, mutation analysis, and known mutation testing for many inherited disorders. Cytogenetic analysis including karyotype, targeted fluorescent *in situ* hybridization (FISH), and microarray testing is also available for chromosome aneuploidy and several microdeletion/microduplication syndromes.

BY THE NUMBERS

85,000+
Tests Performed Annually

70
Tests in our Prenatal Specialty Catalog

8
Testing Laboratories

IDENTIFY AT-RISK PATIENTS THROUGH NONINVASIVE PRENATAL SCREENING

CELL-FREE DNA SCREENING

Prenatal cell-free DNA screening is a type of noninvasive prenatal screening that detects certain chromosome abnormalities in the fetus. The test examines small pieces of fetal DNA circulating in maternal blood and provides patients with information regarding the chance that their fetus has a chromosome abnormality. While this screening test is more sensitive and specific than traditional maternal serum screening, the results are not diagnostic. Our cell-free DNA prenatal screen is available to women after 10 weeks of pregnancy and used to detect trisomies 21 (Down syndrome), 18 (Edwards syndrome), and 13 (Patau syndrome) and aneuploidies of the X and Y chromosomes such as monosomy X (Turner syndrome) and XXY (Klinefelter syndrome).

MATERNAL SERUM SCREENING

Maternal serum screening is used to identify pregnancies that may have an increased risk for certain birth defects such as trisomy 21 (Down syndrome), trisomy 18, and open neural tube defects. Although a positive screen is not a diagnosis, pregnancies at higher risk are identified, and these patients can be offered diagnostic testing. Multiple screening options are available, including first trimester screening, second trimester screening, and cross-trimester screening.

SCREENING GUIDELINES

American Congress of Obstetricians and Gynecologists (ACOG) recommends the following:

- All patients receive detailed counseling regarding the benefits and limitations of current prenatal screening options including cell-free DNA screening.
- Cell-free DNA screening is the recommended screening test for pregnancies at high risk of aneuploidy. Risk factors include advanced maternal age, certain ultrasound abnormalities, family history of aneuploidy, and positive maternal serum screening results.
- Maternal serum screening is still the recommended test for pregnancies that are not at an increased risk for aneuploidy.
- Patients may opt for the screening method of their choice, including the option of not undergoing screening.

PROVIDE DIAGNOSTIC RESULTS FOR PATIENTS SELECTING INVASIVE TESTING

If a screening test or ultrasound indicates a possible problem, or the patient's age, family history, or medical history increases the risk of having a baby with a genetic abnormality, an invasive prenatal diagnostic test should be considered. Invasive prenatal cytogenetic tests include karyotype, FISH, and microarray analysis performed on amniotic fluid or chorionic villus sampling.

In situations where one or more structural abnormalities are detected on ultrasound, microarray is the recommended test. Chromosomal microarray provides a high-resolution assessment of copy number changes, including deletions and duplications, as well as detection of regions of homozygosity, which are not detectable with traditional cytogenetic analysis. For diagnosis of specific known or suspected disorders, targeted FISH or molecular analysis can be performed.

IDENTIFY CAUSES OF PREGNANCY LOSS

Chromosomal abnormalities may result in spontaneous miscarriages, stillbirths, or neonatal deaths. About 10 percent of known pregnancies end in miscarriage of which an estimated 50% may be due to chromosomal abnormalities.[1] Laboratory testing to identify causes of miscarriage can provide useful information when there is a personal history of two or more miscarriages or when fetal malformations are evident. Cytogenetic testing, including karyotype, FISH, and microarray analysis, on early miscarriage, fetal demise, and still birth can provide important information on the cause as well as the risk of recurrence in future pregnancies.

1. American College of Obstetricians and Gynecologists (ACOG) Committee on Practice Bulletins—Obstetrics. ACOG Practice Bulletin No. 150: Early pregnancy loss. Obstetrics & Gynecology. 2015;125:1258.

FEATURED TESTS

CELL-FREE DNA & MATERNAL SERUM SCREENING

- Alpha-Fetoprotein, Single Marker Screen, Maternal Serum (Mayo ID: MAFP)
- Cell-free DNA Prenatal Screen (Mayo ID: NIPS)
- First Trimester Screen, Maternal Serum (Mayo ID: 1STT)
- Quad Screen, Maternal Serum (Mayo ID: QUAD)
- Sequential Maternal Screen, Parts 1 and 2 (Mayo ID: SEQU/SEQF)

CARRIER SCREENING

- Ashkenazi Jewish Mutation Analysis Panel without Cystic Fibrosis (CF) (Mayo ID: AJPO)
- Cystic Fibrosis Mutation Analysis, 106-Mutation Panel (Mayo ID: CFP)
- Fragile X Syndrome Molecular Analysis (Mayo ID: FXS)
- Hexosaminidase A and Total, Leukocytes/Molecular Reflex (Mayo ID: NAGR)

INVASIVE FOLLOW-UP TESTING

- Chromosome Analysis, Amniotic Fluid (Mayo ID: CHRAF)
- Chromosome Analysis, Chorionic Villus Sampling (Mayo ID: CHRCV)
- Chromosomal Microarray, Prenatal, Amniotic Fluid/ Chorionic Villus Sampling (Mayo ID: CMAP)
- Prenatal Aneuploidy Detection, FISH (Mayo ID: PADF)

SPONTANEOUS MISCARRIAGE

- Aneuploidy Detection, FISH Analysis, Products of Conception, Fresh Tissue (Mayo ID: POCRF)
- Aneuploidy Detection, FISH Analysis, Products of Conception, Paraffin-Embedded Tissue (Mayo ID: POCF)
- Chromosome Analysis, Autopsy, Products of Conception, or Stillbirth (Mayo ID: CHRPC)
- Chromosomal Microarray, Autopsy, Products of Conception, or Stillbirth (Mayo ID: CMAPC)

TAP INTO THE EXPERTISE OF MAYO CLINIC

When you partner with Mayo Clinic, you gain access to leading experts in the field of prenatal diagnostics. Our comprehensive prenatal testing spans eight laboratories, each with dedicated teams of laboratory directors, genetic counselors, and scientists working in a state-of-the-art environment. This team provides testing and result interpretation of the highest quality for the diagnosis and clinical care of prenatal disorders.

Experienced clinicians and genetics counselors are available for result interpretation and case review.

FOR MORE INFORMATION ABOUT PRENATAL TESTING, VISIT

MayoMedicalLaboratories.com

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