



# Prenatal Diagnosis

Assists in Pregnancy Management

Detects Presence of Genetic Disorders

Provides Information on Recurrence Risk

Predicts Potential High-Risk Deliveries

Prenatal diagnosis encompasses a variety of tests used to monitor the health of the unborn fetus. Information obtained from prenatal testing can affect pregnancy management, indicate the presence of a congenital abnormality or genetic disorder, predict a high-risk delivery or impending fetal demise, and provide information regarding risks in future pregnancies. Testing is generally performed on amniocytes (cells present in the amniotic fluid) or on the amniotic fluid (AF) itself. Certain testing can also be performed on a percutaneous umbilical cord blood sample (PUBS) or on a chorionic villus sample (CVS).

## Chromosome Analysis

### Indications for Prenatal Chromosome Analysis:

Advanced maternal age (mothers 35 years of age and older are at increased risk of having a child with Down syndrome or other chromosome disorders).

Abnormal maternal serum screening

Abnormal fetal ultrasound

Previous pregnancy or child with a chromosome abnormality

Chromosome abnormality in either parent

Family history of a chromosome disorder

History of multiple miscarriages

History of infertility

### Indications for Parental Chromosome Analysis:

Previous pregnancy or child with a structural chromosome abnormality

Family history of a chromosome disorder

Multiple miscarriages

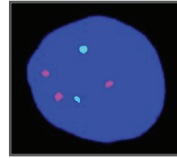
### Indications for Postnatal Chromosome Analysis:

Embryo, fetus, stillborn, or live-born with congenital anomalies

Products of conception

Hydropic placenta

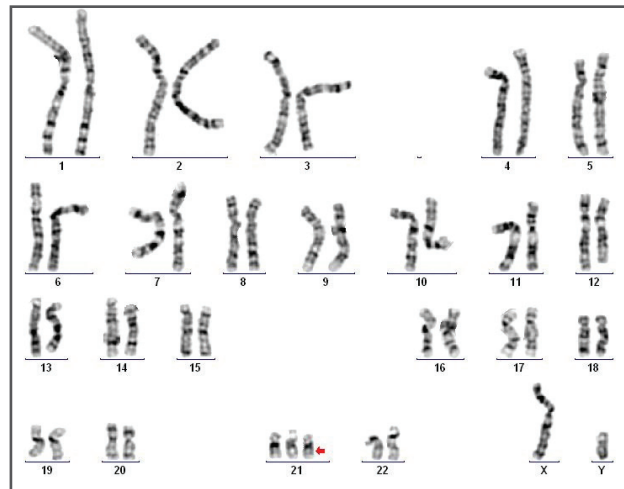
## Fluorescence *in situ* Hybridization (FISH)



FISH is used as an adjunct to conventional cytogenetic analysis and provides a rapid screen for the most common numerical chromosome abnormalities observed in the newborn: trisomy 13, trisomy 18, trisomy 21, aneuploidy for X or Y. Because these studies are performed on uncultured cells, results are available in 1-2 days. FISH is also used to detect cryptic rearrangements and microdeletions that are undetected by routine cytogenetic studies; to identify marker chromosomes; and to rule out chromosomal mosaicism.

## Amniotic Fluid Alpha-fetoprotein Testing (AF-AFP)

Alpha-fetoprotein is produced by the fetal liver and is a normal component of amniotic fluid. However, significant elevations in AF-AFP levels are associated with the presence of neural tube defects, abdominal wall defects, or congenital nephrosis. Reflex testing for the enzyme acetylcholinesterase (AChE) is indicated when AF-AFP is significantly elevated.



Trisomy 21 karyotype consistent with Down syndrome.



## Maternal Serum Screening

Maternal serum (MS) screening is performed on peripheral blood samples typically collected between 15 and 18 weeks of gestation. Screening for MS-AFP (alpha-fetoprotein), beta-HCG (human chorionic gonadotrophin), uE3 (estriol), and Inhibin-A indicates if the fetus is at increased risk of a neural tube defect, an abdominal wall defect, trisomy 21, or other specific chromosome abnormalities. Screening may also indicate the presence of multiple gestation, molar pregnancy, or fetal demise.

## Chromosome Studies for Infertility

In 6-8% of couples with apparent infertility, one member of the couple carries a balanced chromosome rearrangement. Numerical or structural abnormalities of the sex chromosomes are also associated with reduced fertility. For example, Klinefelter syndrome (47,XXY) accounts for 5-15% of male infertility, particularly in males with azoospermia or oligospermia.

## Related Tests

### Cystic Fibrosis (CF)

Guidelines from the American College of Obstetricians and Gynecologists (ACOG) recommend offering CF testing to individuals with a family history of CF, reproductive partners of individuals with CF, and couples who are planning a pregnancy or seeking prenatal care. For couples at risk of producing offspring with CF, prenatal testing can be performed to determine the status of the fetus. The CF panel detects 32 mutations, including the 23 core mutations currently recommended by the American College of Medical Genetics.

### ThromboFLEX Profile

Genetic mutations and polymorphisms in several well-characterized genes are associated with an increased risk for venous thrombotic events (VTEs). Women with increased risk of VTEs may experience recurrent pregnancy loss, unexplained severe pre-eclampsia, placental abruption, intrauterine growth retardation, or stillbirth. Identifying women with genetic risk factors is important for effective management of anticoagulant therapy. Testing includes analysis of the Factor V, Factor II, methylenetetrahydrofolate reductase (MTHFR), and plasminogen activator (PAI) genes for common mutations associated with an increased risk of venous thrombosis.

## About MPLN

Since 1989, Molecular Pathology Laboratory Network, Inc. (MPLN) has offered an expanding selection of tests in molecular oncology, infectious diseases and human genetics to hospitals, medical laboratories and private physician groups nationwide.

Headquartered in Maryville, Tennessee, MPLN is a fully-licensed laboratory, certified by the Clinical Laboratory Improvement Amendment, accredited by the College of American Pathologists and licensed in the states of Tennessee, Florida, New York and Maryland.

At MPLN, our philosophy is simple - we build strong professional relationships, deliver personalized service, and offer advanced diagnostic technology to support high-quality patient care.

For more information, contact your local representative or call MPLN client services at **800.932.2943**.

## References

1. Baschat AA et al. (2003). Prevalence of viral DNA in amniotic fluid of low-risk pregnancies in the second trimester. *J Matern Fetal Neonat Med.* 13(6):381-384.
2. Randolph LM et al. (2005). *Prenatal Diagnosis in The Principles of Clinical Cytogenetics*, 2nd ed, S.L. Gersen and M.B. Keagle, Human Press Inc., Totowa, NJ.