

Amniotic Fluid Studies for Neural Tube Defects (NTDs)

Amniotic Fluid Alpha-fetoprotein (AFP) assays are done on all amniotic fluid samples. Whenever indicated, acetylcholinesterase and fetal hemoglobin assays are added. These assays are specifically recommended for:

- Couples who have a previous child with an NTD or an affected first degree relative
- One parent has a NTD
- A high maternal serum AFP
- Congenital nephrosis or other leaking fetal defects

Rapid Interphase FISH Analysis

Indicated for urgent second and third trimester study when a fetal defect is present or risk is high. Diagnostic focus is on chromosomes 21, 18, 13, X and Y. A complete routine chromosome analysis is still needed.

Maternal Serum Screening

Neural Tube Defects (e.g. spina bifida):
Recommendations are to test a blood sample from all women in all pregnancies for four biochemical markers (quad-screen) at 16 weeks and/or to have an ultrasound study.

Chromosome defects (e.g. Down syndrome):
Recommendations are to test all women in all pregnancies with two biochemical markers and by ultrasound at 11-13 weeks.

Genetic Counseling and Evaluation

Many indications exist for genetic counseling. Recommendations are especially for couples or individuals concerned about:

- A personal genetic disorder
- A child's or family member's genetic disorder
- Evaluation of developmental delay, intellectual disability, autism, and birth defects
- Early onset of cancer or family history of multiple cancers
- A family history of an hereditary disorder
- A carrier test because the person has a specific ethnic origin (e.g., Italians, Greeks, and others of Mediterranean descent; Blacks, Jews, Asians) or consanguinity
- A previous child with a birth defect or intellectual disability
- Risks of recurrence, or for any treatment or care of a genetic disorder

JEFF MILUNSKY, M.D., F.A.C.M.G.
Co-Director, Center for Human Genetics, Inc.
Director, Clinical Genetics
Senior Director, Molecular Genetics

AUBREY MILUNSKY, M.D., D.Sc., F.R.C.P., F.A.C.M.G.
Founder and Co-Director
Center for Human Genetics, Inc.

Center for Human Genetics, Inc.
Riverside Technology Center
840 Memorial Drive, Suite 101
Cambridge, MA 02139

Tel: (617)-492-7083
Fax: (617)-492-7092

Email: jmilunsky@chginc.org
amilunsky@chginc.org

For appointments, call 617-492-5106

**Free parking and
Phlebotomy on site**



**CENTER FOR HUMAN
GENETICS, INC.**

**AN INTERNATIONAL REFERENCE CENTER
SPECIALIZING IN:**

- Molecular Genetic (DNA) diagnoses
- Clinical diagnosis of genetic disorders and syndromes
- Genetic evaluation for intellectual disability, autism, and birth defects
- Genetic counseling for genetic disorders, pregnancy, and infertility
- Prenatal diagnosis of genetic disorders
- Maternal serum screening for fetal genetic disorders
- Molecular cytogenetics of cancer
- Paternity testing by DNA analysis

For information and the latest available tests, see our website at
www.CHGINC.org

For appointments:
call 617-492-5106

For general inquiries:
call 617-492-7083

Center for Human Genetics, Inc.
Riverside Technology Center
840 Memorial Drive, Suite 101
Cambridge, MA 02139
617-492-7083

Center for Human Genetics, Inc.

A non-profit corporation

This center is an international clinical and reference laboratory for the diagnosis of genetic disorders. Overall experience in the provision of genetic services now exceeds 30 years with a cumulative case-load among the largest in the United States. The specialized services described below include evaluation of patients and employ the latest molecular and cytogenetic techniques, providing diagnostic services to physicians in all specialties.

We are privileged to serve physicians in all states in the USA and more than 44 countries.

DIAGNOSTIC SERVICES

Molecular (DNA) Analyses

Diagnosis, carrier detection, pre-symptomatic and prenatal diagnosis is provided for a rapidly increasing list of genetic disorders. We will extract DNA and arrange for analysis elsewhere for disorders not on our list.

Molecular Diagnosis

Gene sequencing and deletion/duplication analysis as well as direct mutation detection is provided for over 100 genetic disorders. Our extensive multisystem coverage includes neurogenetics, connective tissue syndromes, cancers, ethnic-related disorders, and mitochondrial and metabolic disorders. Examples include Neurofibromatosis, Marfan syndrome, Ehlers-Danlos, Kabuki and other syndromes, colon and other cancers, carrier tests for Ashkenazi disorders, Cystic Fibrosis, and Sickle Cell disease.

Paternity Tests

DNA studies provide certainty (>99.99%) for paternity determination.

DNA Banking

Banking a DNA sample from a family member with a known or as yet unresolved genetic disorder may be extremely important for others in the family to determine precise diagnosis or for future family planning or prenatal diagnosis.

Chromosome Analyses

Blood Chromosome Analysis

Most important indications include:

- Evaluation of birth defects, intellectual disability, autism, and syndromes
- A couple with recurrent miscarriage
- Parents of an offspring with a chromosome defect
- Evaluation for learning disorders
- Evaluation of infertility
- A family history of a chromosome defect

Cancer Cytogenetic Analysis

For determination of characteristic chromosome abnormalities in blood, bone marrow, and tumors for leukemias, lymphomas, myelodysplasia, and other cancers.

Chromosome Analysis of the Products of Conception/Stillbirth

Studies focused on tissue obtained following spontaneous or elective abortion, fetal death, or stillbirth.

Fluorescent in situ hybridization (FISH)

The following are examples among many:

- Angelman syndrome
- Marker chromosomes
- Miller-Dieker syndrome
- Prader-Willi syndrome
- Translocations (Cryptic)
- Velocardiofacial/DiGeorge syndrome [22q-]
- William’s syndrome

SNP Microarray Analysis

Recommended as the first test for diagnosis of intellectual disability, autism, and various birth defects.

- Invaluable for detection of chromosome/gene deletion/duplication disorders.
- Important for prenatal diagnosis if ultrasound reveals a fetal abnormality.

Prenatal Diagnosis

Chromosome study/SNP Microarray

The most important indications for study are:

- advanced maternal age
- Previous offspring with any chromosome defect
- Increased odds for Down syndrome (or other chromosome defect) after maternal serum screening
- A parental carrier or one who has a chromosome defect
- Discovery of a fetal anomaly or intrauterine growth restriction