



Cytogenetics

A complete range of cytogenetic services for oncology, prenatal diagnosis and postnatal analysis.

The cytogenetics laboratory at Molecular Pathology Laboratory Network, Inc. (MPLN) provides a full range of constitutional and cancer cytogenetic services. Dr. David DeBauche, the laboratory director, has more than 20 years experience and is a founding fellow of the American College of Medical Genetics.

The laboratory is staffed by highly skilled cytogenetic technologists who are certified as clinical specialists in cytogenetics by the American Society Clinical Pathologists (ASCP). The cytogenetics laboratory takes pride in accuracy, efficiency, short turn-around time, competitive prices, and providing the highest quality of service.

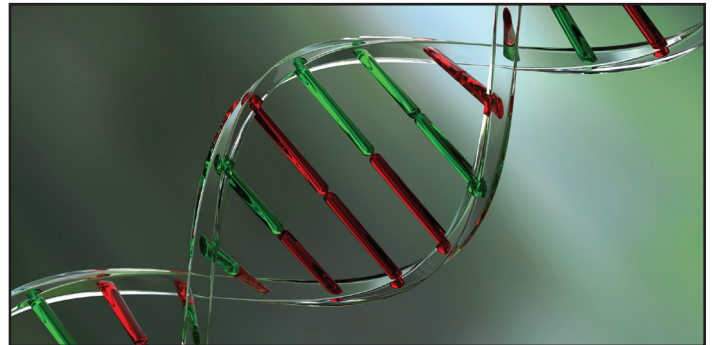
Cancer Chromosome Studies

Cytogenetic analysis in neoplastic diseases involve the study of the cancer cells themselves. In leukemia, a bone marrow aspirate is usually obtained for study. In some cases, peripheral blood is used in place of the bone marrow, particularly if the white blood cell count is >10,000. The purpose of the cytogenetic study in hematological disorders is to detect the presence of acquired chromosome changes, i.e. those aberrations that have arisen secondary to the disease state.

The study of chromosomes in leukemia serves two functions:

- to assist in a more accurate diagnosis, thereby providing important prognostic information
- to identify the sites of consistent rearrangements

Specific chromosome abnormalities often correlate with particular subtypes of disease. Serial samples from the patient permit the study of cytogenetic patterns during the various stages of a patient's clinical course.

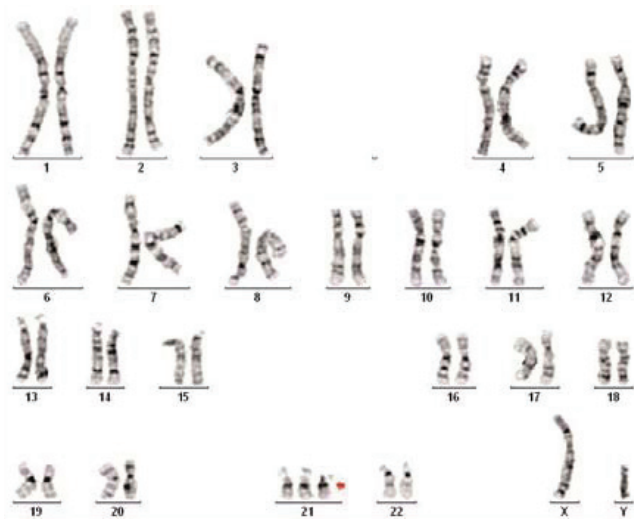


Constitutional Chromosome Studies

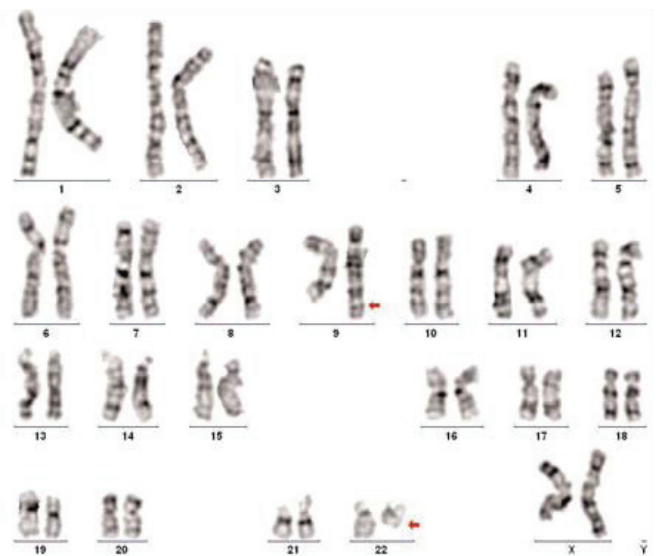
Cytogenetic analysis to determine if constitutional abnormalities are present is performed for a variety of indications including multiple congenital abnormalities, mental retardation of unknown etiology, abnormalities of growth, features of a recognized genetic syndrome, recurrent pregnancy loss, prenatal diagnosis via amniocentesis, mosaicism, stillbirth, fetal loss, or molar pregnancy. Adjunct studies such as FISH or other molecular and biochemical testing can be performed in addition to chromosomal analysis.

FISH

In addition to offering high quality chromosome analysis, the cytogenetics laboratory also specializes in fluorescence in situ hybridization (FISH). FISH, a molecular cytogenetic technique, enables the analysis of disease specific abnormalities. It is offered for the detection of cryptic rearrangements, microdeletion syndromes, aneuploidy, and marker chromosome identification.



Karyotype showing trisomy 21 consistent with a clinical diagnosis of Down syndrome



Karyotype showing the Philadelphia translocation involving chromosomes 9 and 22

Test Name	Specimen Requirements
Cancer cytogenetics (CYTO BM, CYTO UPB, CYTO ST)	7mL (min. 5mL) whole blood or 3mL (min. 1mL) bone marrow in sodium heparin, 5mm ³ bone marrow core biopsy or fresh tissue in transport media, 4mL (min. 2mL) fine needle aspiration in tissue transport media, 15mL ascites, gastric pleural effusions in plain tube
Prenatal chromosome analysis (CYTO PN, CYTO AF)	30mL (min 20mL) amniotic fluid in 2-3 sterile tubes
Constitutional tissue chromosome analysis (CYTO TC)	Products of conception, fetal tissue, 1cm skin biopsy or other solid tissue in sterile tissue transport media
Constitutional peripheral blood chromosome analysis (CYTO PB)	7mL (min. 5mL) whole blood or 1ml newborn blood or 2mL (min. 1mL) percutaneous umbilical cord blood in sodium heparin
Fluorescence <i>in situ</i> hybridization	Peripheral blood, amniotic fluid, solid tissue, and bone marrow samples with or without routine chromosome studies; please see catalogue for more detailed information