May 2022 Issue 1

# Highlighting Your Lab

### **CYTOGENETICS**

The Cytogenetics laboratory maintains a broad profile of service: prenatal testing, postnatal constitutional analysis and testing for acquired abnormalities (neoplasia). Chromosome disorders are a major category of genetic disease. Chromosome abnormalities account for a large proportion of patients with congenial malformations, development disabilities, and infertility or recurrent pregnancy loss.

For patients with acquired abnormalities or hematologic malignancies, cytogenetic testing plays a key role in the diagnosis, prognosis, and treatment.

#### COMPREHENSIVE TESTING MENU INCLUDES:

- Chromosome analysis on a full range of samples, both neoplastic and constitutional.
  Sample types include peripheral blood, bone marrow, amniotic fluid, products of conception, skin, solid tumors and others as needed.
- FISH (fluorescence in situ hybridization) is available for both metaphase and interphase analysis including panels of probes for pediatric ALL, AML, CLL, MDS, and multiple myeloma (as applied to isolated plasma cells).
- Chromosomal microarray testing is performed for constitutional abnormalities and acquired abnormalities in peripheral blood, bone marrow, products of conception, and formalin-fixed paraffin embedded.

Cytogenetics provides a comprehensive, integrated approach to cytogenetic testing, both diagnostic and prognostic, that focuses on individualized patient care and service to our clinician clients.







#### WHAT YOU MIGHT NOT KNOW

Prior to analysis, the cells that are tested must be cultured and cell division promoted, requiring several days. Analysis results are not generated by an instrument. Analysis of chromosomes requires a specialized technologist expertise. Currently there are 15 technologists and two PRNs in the laboratory, totaling and incredible 272 years of experience. The cytogenetics team is extremely specialized and skilled.

Cytogenetics is part of the MPGG division (Molecular Pathology, Genetics and Genomics) along with the Molecular Diagnostics Laboratory

#### WHATS NEW?

Chromosomal microarray testing has previously been focused on use in understanding constitutional abnormalities. Recently, cytogenetics has expanded testing capabilities to include analysis of paraffin embedding tumors and tissues. Soon, the laboratory plans to expand its testing menu to include chromosomal microarray testing on hematologic malignancies.

## GOOD TO KNOW...

Location: Second floor of the Wadsworth building

Director(s):

Catherine W. Rehder, PhD, FACMG

Kristen L. Deak, PhD, FACMG

Stefan Rentas, PhD, FACMG

**Assistant Director** 

Certified in Laboratory Genetics and Genomics by the AB-MGG

**Manager:** Andrea Fuentes

Administrative Assistant: Marcellina Manzanet-

Schmidt

**Team Members**: 20

Hours of Operation: M-F 5am-8pm (with limited week-

end coverage)