



## CYTOGENETICS

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This is a required rotation of at least 14 days duration, not necessarily all days in a row. During the 14 days, the resident will participate in hands-on laboratory training in basic cytogenetics procedures and will review cytogenetic disorders. The resident will also be able to discuss the type of tissue, the condition of the tissue, and the appropriate transport medium to initiate a successful study, as well as the indications for culture setup and analysis. The resident is expected to augment his/her experience with required reading assignments, which include techniques as well as diseases associated with chromosomal abnormalities. One or two Residents' conferences will also be presented to all Residents on some cytogenetics related topics.

#### **Objectives for Six General Competencies:**

**Patient Care:** Proper specimen collection and processing, gathering pertinent data, and understanding laboratory techniques.

1. Learn the laboratory procedures for karyotype analysis, which includes completing at least one karyotype from start to finish. The resident cultures the cells, prepares the metaphase chromosomes, bands and stains the slides, and then prepares the karyotype.
2. Participate in the review of karyotypes from patient specimens on a daily basis. Current and archival cases of particular interest will be presented.
3. List indications for cytogenetic analysis. Appreciate the need for precise and accurate indications accompanying the specimens.
4. Tissue cultures: Learn proper procedures for collecting, culturing, and analyzing specimens.
5. Fluorescent in-situ hybridization(FISH): Learn techniques used for preparation and analysis of specimens.

#### **Medical Knowledge**

1. Briefly discuss chromosome banding techniques (C-Banding, G-Banding, Q-banding, R-banding and silver staining) including the major differences in techniques used for the different banding procedures and the results that are yielded.
2. Briefly discuss the nomenclature of banded chromosomes and karyotypes.
3. For the following chromosomal abnormalities, be able to recognize each abnormality from banded karyotypes, and give the key associated phenotypic abnormalities for each:
  - A. Down syndrome
  - B. Trisomy-13



- C. Trisomy-18
  - D. Microdeletion syndromes,
  - E. Cri du chat
  - F. Turner syndrome
  - G. Klinefelter syndrome
4. Be familiar with the terms contiguous gene syndrome, genomic disorder ??????????
  5. Understand the qualitative differences between the data collected by the different modes genomic analysis and the appropriate employment of one modality versus another.
  6. The resident should be familiar with chromosome defects commonly associated with the following tumors:
    - A. Childhood tumors including Retinoblastoma, Wilm's tumor, Ewings' sarcoma, Rhabdoid tumor, Alveolar Rhabdomyosarcoma, Desmoplastic small round cell tumor.
    - B. Soft tissue tumors including Clear Cell Sarcoma, Synovial Sarcoma, Myxoid and Round cell Liposarcoma, Hemangiopericytoma.
    - C. Hydatidiform Mole
    - D. Hematologic Disorders: Acute lymphocytic and Non-lymphocytic leukemias, Chronic Myelogenous leukemia, Myelodysplastic syndromes, non-Hodgkin's lymphoma

### **Practice-Based Learning and Improvement**

1. Understand quality assurance and control practices as they relate to cytogenetics techniques and outcomes.
2. Assimilate and review scientific literature and be familiar with the most current techniques being utilized in the field of cytogenetics.
3. M-2 small group sessions: Teach medical students the important aspects of cytogenetics as part of small group sessions.
4. Serve as a consultant to junior residents in the appropriate collection of cytogenetic specimens and in the interpretation of cytogenetic test results.

### **Interpersonal and Communication Skills**

1. Effectively communicate with cytogenetic technologists, supervisors, and other faculty, and, when applicable, serve as a liason when exchanging information with clinicians involved in a particular case.
2. Know the fundamentals of how lab reports are interpreted and written.

**Professionalism:** Demonstrate a commitment to carrying out professional responsibilities and adherence to ethical principles.

### **Systems-Based Practice**



1. Understand the costs of utilizing different methodologies in the cytogenetics laboratory.
2. Be familiar with potential legal ramifications as it relates to cytogenetic testing.
3. Understand how information technology utilized in the lab stores and relays information to other systems.
4. Understand the efficiency of the workflow in the lab and how cases are prioritized.
5. Understand the managerial aspects of the lab and how these practices effect other health systems.

**Outcomes Assessment:** A standard competency-based evaluation form will be completed.