CYTOGENETICS

This is a required 2-week rotation. During the two weeks, 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 carrying medium to initiate a successful study, as well as the indications for 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.

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 collects the specimen, cultures the cells, prepares the chromosome spread, stains the slides, analyzes and captures the metaphases, and then constructs the karyotype.

2. Participate in the examination and analysis of karyotypes from patient specimens on a daily basis (typically 40 in a two-week rotation). The resident will work alongside the technologist learning to produce computer-generated karyotypes and to diagnose chromosomal abnormalities, which are then incorporated into a formal written report.

3. List indications for cytogenetic analysis.

4. Tissue cultures: Learn proper procedures for collecting, culturing, and analyzing specimens.

5. Fluorescent in-situ hybridization (FISH): Learn techniques utilized and analysis of specimens

Medical Knowledge

1. Briefly discuss chromosome banding techniques (Q-Banding, C-Banding, G-Banding, and R-Banding) 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's syndrome
   B. Trisomy-13
   C. Trisomy-18
   D. Microdeletion syndrome
   E. Cri du chat
   F. Turner's syndrome
   G. Klinefelter's syndrome
   H. XYZ syndrome
4. 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.

Hobart
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