

CYTOGENETICS: Rotation director: Roberto Zori, M.D.

1. Description of the rotations: Training in cytogenetics involves training in **I) prenatal and medical cytogenetics, II) cancer cytogenetics, and III) pediatric pathology**. The dedicated 4-week cytogenetics rotation involves 3 weeks at the R. C. Philips Cytogenetics Laboratory and 1 week of seminars held on the UF Health Science Center campus. At the R.C. Philips Cytogenetics Laboratory at Tacachale State Home (Gainesville, FL) residents are oriented to the basic laboratory methods used to construct and interpret karyotypes. This laboratory focuses on prenatal karyotypes and medical karyotypes for syndromic diagnosis (e.g., Down Syndrome) as well as cancer diagnosis (Competency #2: Medical Knowledge). Molecular techniques are employed such as fluorescent in situ hybridization (FISH). The residents learn to interpret the resulting karyotype and construct consultative reports. Residents will then be able to correlate this data with the clinical history and physical examination through a review of the patient's on-line medical record (OLMR), chart or by contacting the clinical service.

Seminar topics include: Introduction to genetics, modes of inheritance, techniques, syndromes, and cytogenetics (Competency #2: Medical Knowledge). The objective of these seminars is to discuss nomenclature and classic genetic syndromes involving chromosomal disorders. At the conclusion of the rotation, each resident will be expected to present a seminar to the pathology faculty and residents on a topic that the resident found interesting in cytogenetics (Competencies #3: Practice-Based Learning and Improvement, #4: Communication, and #5 Professionalism).

These experiences are supplemented by 1) directed readings, and 2) periodic lectures on the topic cytogenetics. Fellows do not participate in this rotation. As appropriate to the individual case or consultation under review, the ethical, socioeconomic, medicolegal and const-containment issues are reviewed and discussed. As well, research design, statistics and critical review of the literature are discussed. By the use of literature, Medline, and textbooks, the resident is trained to become a life-long learner.

2. Goals of the rotation: the resident will be able to 1) recognize the major features of common chromosomal disorders (e.g., Turner syndrome, Down syndrome, Klinefelter syndrome, 5p-, etc.) 2) know the indications for chromosome analysis in oncology and pediatrics. 3) recount the common chromosomal abnormalities/translocation is common neoplasms and 4) be able to use sophisticated cytogenetic nomenclature.

3. Duration of the rotation: 4 weeks: 3 weeks in the cytogenetics laboratory and 1 week on campus. Cytogenetics is also addressed in hematopathology and perinatal/autopsy pathology.

4. Duties and responsibilities of the resident: the resident will review the clinical history and cytogenetic analysis to assist in the generation of the clinical report.

5: Teaching staff: Robert Zori, M.D.

6.

- i. Resident supervision: Reports are generated in concert with the attending faculty and signed out by the attending faculty.
- ii. Resident Monthly written/electronic evaluation

Curriculum: Cytogenetics, Roberto Zori, M.D.

I) LABORATORY EXPERIENCE

1.A) THE MECHANICS OF PERFORMING A KARYOTYPE ANALYSIS: Basic Laboratory Procedures and Concepts:

Goals:

- Provided a basic introduction to how a karyotype is performed
 - Explain what differences exist in performing a perinatal karyotype versus a clinical karyotype versus ad tumor/cancer karyotype
 - Describe the common sources of failure and error
 - Overview quality assurance/quality control in the cytogenetic laboratory
 - Review the regulatory requirements of the cytogenetics laboratory
 - Who in Florida accredits the cytogenetics laboratory
 - What are the education and licensure requirements need to become a cytogenetics technician, supervisory, or laboratory director
 - How is proficiency testing carried out
 - Can cytogenetics laboratory become CAP accredited
1. Specimen collection and handling: blood, bone marrow, amniotic fluid, chorionic villus samples, skin, abortus material
 2. Culture initiation; culture maintenance; culture harvest
 3. Chromosomal staining and banding: how you identify and count chromosomes; how you recognize abnormalities of chromosome number or structure: G-banding, Q-banding, R-banding, C- banding, T-banding, Cd staining, G-11 banding, NOR staining, DAP1/DA staining
 4. Molecular technologies: FISJ, other molecular genetic techniques used in the R.C. Philips Laboratory
 5. High -resolution studies/special techniques: cell synchronization techniques, chemical elongation, fragile X analysis
 6. Culture failure; Preservation of cells
 7. Microscopy and photomicroscopy

8. Quality control and quality assurance; automation in the cytogenetics laboratory

I.B) Analysis of the Karyotype: chromosome analysis:

Goals:

- Become familiar with cytogenetic nomenclature
- Understand the concepts of aneuploidy, translocation, etc.
- See examples of the chromosomal abnormalities
- Overview the role of the cytogenetic laboratory in genetic counseling

1. Autosomal aneuploidy: Trisomies 13, 14, 15, 16, 18, 21, monosomies 21, 22, polyploidy (triploidy, tetraploidy), partial autosomal aneuploidies (examples), tetrasomy 8p, tetrasomy 9p, tetrasomy 12 p, tetrasomy 18p, other partial autosomal aneuploidies

2. Structural chromosomal rearrangements: deletions, duplications, inversions, dicentric chromosomes, acentric chromosomes, isochromosomes, ring chromosomes, reciprocal autosomal translocations, Robertsonian translocations, jumping translocations, insertions

3. Sex chromosomes and sex chromosome abnormalities: X and Y chromosome structure, organization, numerical abnormalities (Turner syndrome, 47, XXX; 48, XXXX; 49, XXXXY; 47, XYY), structural abnormalities of the X, structural abnormalities of the Y

4. Genomic imprinting and uniparental disomy

II.) Special topics:

Introduction to Genetics (concepts of chromosomal organization, genomic organization, gene structure and gene regulation [transcription factors], modes of inheritance (Mendelian inheritance, monogenetic, polygenetic, mitochondrial inheritance), introduction to cytogenetics (clinical uses of cytogenetics), cytogenetic techniques (banding studies, molecular methods), chromosomal syndrome (clinical description of the classic syndromes), cytogenetics and oncogenetics (leukemias, lymphomas, solid tumors), indications for chromosomal analysis