MEDICAL GENETICS RESIDENCY PROGRAM
Department of Pediatrics

University of Michigan Health Systems
1500 E. Medical Center Drive
D5240 MPB
Ann Arbor, MI 48109-5718

(734) 763-6767
(734) 763-6561 (fax)

Biochemical Genetics Goals and Objectives

Director: Drs. Ayesha Ahmad and Shane C. Quinonez

The goals and objectives of the Biochemical Genetics Clinic rotation in the Medical Genetics Residency Program are to provide the resident with exposure to all aspects of care of metabolic disease and counseling in accordance with the Residency Review Committee for Medical Genetics expectations and to fulfill criteria for board eligibility by the American Board of Medical Genetics.

Patient Care

The resident will become familiar with the evaluation, diagnosis and management of urea cycle disorders, organic acidemias, disorders of carbohydrate and lipid metabolism and numerous other inborn errors of metabolism (IEMs). Residents will gain exposure in performing and expertise in interpreting biochemical analyses relevant to the diagnosis and management of human genetic diseases.

By the end of the rotation the resident should be able to identify signs and symptoms of IEMs, formulate a differential diagnosis, order appropriate tests and recognize normal variants and complex patterns of metabolites. Residents will be able to manage acute metabolic crises and provide chronic management of patients with an IEM.

Residents should be able to interpret NBS results, collaborate with the primary provider to act upon results in a timely manner, develop a differential diagnosis and order appropriate confirmatory testing and communicate results to families.

Medical Knowledge

Through coursework and didactic sessions with attending physicians, residents will become familiar with fundamental concepts, molecular biology and biochemistry relevant to IEMs. Residents will also interpret biochemical genetic assays and be familiar with specific metabolites altered in disorders listed above. Residents will become familiar with laboratory procedures and fundamental biochemistry that encompass screening tests, newborn screening, diagnostic tests, technical procedures, quality assurance and interpretation of biochemical genetics results, including recognition of common laboratory test artifacts and normal variants. Residents will develop broad knowledge of basic biochemistry and biology, the application of biochemical methods through the diagnosis and management of genetic diseases and an understanding of the etiology, pathogenesis, clinical manifestations and management of IEMs.

Residents will be able to formulate and finalize a care plan for a patient with an IEM and apply practice guidelines as appropriate.

Practice-Based Learning & Improvement

Residents will learn to apply appropriate diagnostic and therapeutic modalities in the management of inpatients with metabolic disease crises and to provide clinical care directed at selected biochemical disorders.

Residents will assess self-performance and develop a learning plan.
Residents will critically evaluate and utilize information from diverse sources.
Residents will identify areas for improvement in individual practice.

Interpersonal & Communication Skills

Residents will display effective communication with ordering and consulting physicians and laboratory directors and will have opportunities to improve their communication skills in the laboratory setting.

Residents will display effective communication and relationship development with families and health care team members.

Residents will exhibit the ability to manage conflicts with patients/families and health care teams and participates in team based care (includes dieticians, social workers, nurses, lab personnel, other physicians, etc).

Residents will exhibit the ability to communicate general and sensitive information with awareness of the social context, demonstrate sensitivity to cultural values in communicating information and be able to provide appropriate information and resources to patients and families with an IEM.

Professionalism

Residents will interact closely with laboratory staff and learn their responsibilities and tasks. Residents will learn how laboratory staff communicates with other hospital and laboratory personnel.

Residents will demonstrate caring, honest and sensitive behavior in all relationships and situations.

Residents should display the ability to manage personal beliefs to avoid any negative impact on patient care and be able to recognize and manage ethical issues in genetics practice.

Residents should complete professional responsibilities in a timely manner.

Residents are expected to recognize limits of their own knowledge and ask for assistance when needed.

Residents should be able to identify and manage situations in which maintaining personal emotional, physical and mental health is challenged by common and typical clinical care situations.

Systems Based Practice

Residents will learn costs and benefits of diagnostic and therapeutic modalities in the management of inpatients with IEM crises and how these costs are transferred to insurance providers.
Residents will learn to function effectively within different systems; and be able to incorporate and advocate for genetic services to enhance cost-effectiveness of care. Residents should be able to recognize and manage the variation in access to genetic testing; facilitate management and transitions of care teams as the patient ages and participate in identifying system errors and implementing potential systems solutions.

Residents will be able to document essential elements of genetics encounters for patients with IEMs to enhance the transfer of information and patient safety; and utilize decision support tools.

MEDICAL GENETICS RESIDENCY PROGRAM
Department of Pediatrics

University of Michigan Health Systems
1500 E. Medical Center Drive
D5240 MPB
Ann Arbor, MI 48109-5718

(734) 763-6767
(734) 763-6561 (fax)

Clinical Cancer Genetics Rotation

Director: Elena Stoffel, MD

Goals:

The primary objective of this rotation is to introduce clinical Medical Genetics residents to the principles and practice of clinical cancer genetics. The Clinical Cancer Genetics Rotation consists of individual tutoring, a case-based self-study program and practical experience in the Cancer Genetics Clinic. At the end of the rotation residents should have:1) a working understanding of oncogenes, tumor suppressor genes and "caretaker genes," 2) an introduction to techniques for cancer risk assessment, 3) the ability to recognize classic familial cancer syndromes based on pedigree analysis and clinical phenotype and 4) an appreciation of the role of genetics in cancer treatment and prevention (eg. precision oncology). Clinical cancer genetics will be framed in the context of a medical genetics model as well as multidisciplinary care including cancer risk assessment, cancer screening/surveillance, and therapeutic oncology.

Resident Responsibilities:

Residents will be asked to fully participate in case-preparation for the clinics that they attend. This may include record review, construction of pedigrees, researching the availability of genetic testing, and preparing risk estimates based on empiric models or Mendelian genetics. Residents will be responsible for dictating clinic notes and patient letters for patients / families the resident sees in clinic. Attendance at weekly clinical case conference, reading and case-based self-study are highly recommended. Residents may be invited to present a short talk or review a journal article on a topic in clinical cancer genetics.

Patient Care

Residents will be expected to learn appropriate diagnosis and risk assessment for patients with known or suspected genetic forms of cancer.

Medical Knowledge

Residents will learn from cases and didactic sessions about specific cancer genetic disorders and their underlying mechanisms (see list below for details).

Practice-Based Learning & Improvement

Residents will learn basics of cancer genetics, testing options, and medical management for a variety of cancer predisposition syndromes. Residents will learn to use several different databases and risk models to assess patients' risks for genetic disorders associated with increased risk for cancer. Residents will learn how to calculate carrier risks for Mendelian and complex disorders. Residents will learn about how pathologic features of tumors can be used to identify hereditary cancer syndromes and inform treatment and prognosis.

Interpersonal & Communication Skills

Residents are expected to communicate effectively with other health care providers and with their patients and families. Residents will learn about privacy and confidentiality in cancer genetics.

Professionalism

Residents will learn the importance of accurate communication of test results and counseling information in the clinical setting. They will also learn basics of how to relay test results to patients in stressful situations.

Systems Based Practice

Residents will learn about cost-effectiveness of genetic evaluation, as well as review cost-benefit analyses for various testing strategies (including multigene panel testing), and how these tests influence care of cancer patients and their families.

Clinical Cancer Genetics Rotation Curriculum and Schedule

Clinical Experience
In addition to the didactic component, residents will participate in the evaluation and management of patients scheduled in the Thursday Cancer Genetics Clinic.

Self-Study Program
It is recommended that residents complete 4 cases from the self-study program

Resident Presentation
At the conclusion of the rotation residents will have the opportunity to give a 20-30 minute presentation on a topic in cancer genetics (topic is chosen in conjunction with the course director).

One Month Rotation – *didactic focus*

Week 1:	Oncogenes, Tumor Suppressor Genes, "Caretaker Genes"
	Clinical correl: Familial Adenomatous Polyposis, Lynch
Week 2:	Phenotypic variation and cancer syndromes
	Clinical correl: LFS, PTEN Hamartoma Tumor Syndrome
Week 3	Cancer Risk Assessment

Clinical correl: Hereditary Breast Ovarian Cancer Synd.

Week 4 Managing Individuals at High Risk

Clinical correl: LFS, von Hippel Lindau Syndrome

Recommended Reading

Online Resources:

Elements of Cancer Genetics Risk Assessment and Counseling (PDQ®)
http://www.cancer.gov/cancertopics/pdq/genetics/risk-assessment-and-counseling/healthprofessional

National Comprehensive Cancer Network. Guidelines for detection, prevention, and risk reduction. http://www.nccn.org

Overview of Cancer Genetics:

Fearon, ER Human cancer syndromes: clues to the origin and nature of cancer. Science. 1997 Nov 7;278(5340):1043-50.

Foulkes, WD. Inherited susceptibility to common cancers. NEJM 2008. 359: 2143-2153.

Lindor NM, McMaster ML, Lindor CJ: Concise handbook of family cancer susceptibility syndromes – second edition.. J Natl Cancer Inst Monogr 2008;38:3-93.

Lu KH, Wood ME, Daniels M, Burke C, Ford J, Kauff ND, Kohlmann W, Lindor NM, Mulvey TM, Robinson L, Rubenstein WS, Stoffel EM, Snyder C, Syngal S, Merrill J, Wollins DS, Hughes KS. ASCO Expert Statement: Collection and Utilization of a Cancer Family History for Oncology Providers. J Clinical Oncology 2014 (epublished ahead of print February 3, 2014).

Stoffel EM, Cooney KA eds. Advances in Inherited Cancers. Seminars in Oncology.43(5). 527-622.

Selected References:

Breast/Ovarian Cancer:

Lu K, Kauff N, Powell CB, et al: Hereditary breast and ovarian cancer syndrome. Gynecol Oncol.113:6-11, 2009

Domchek et al Association of Risk Reducing surgery in BRCA1 and BRCA2 mutation carriers with Cancer Risk and Mortality. JAMA. 2010 September 1; 304(9): 967–975.

Domchek SM, Eisen A, Calzone K, et al.: Application of breast cancer risk prediction models in clinical practice. J Clin Oncol 21 (4): 593-601, 2003

Colorectal Cancer:

Stoffel EM, Boland CR. Genetics and Genetic Testing in Hereditary Colorectal Cancer. Gastroenterology. 2015 Oct;149(5):1191-1203.

Precision Oncology:

Mody RJ, Wu YM, Lonigro RJ, Cao X, Roychowdhury S, Vats P, Frank KM, Presner JR, Asangani I, Palanisamy N, Dillman JR, Rabah RM, Kunju LP, Everett J, Raymond VM, Ning Y, Su F, Wang R, Stoffel EM, Innis JW, Roberts JS, Robertson PL, Yanik G, Chamdin A, Connelly JA, Choi S, Harris AC, Kitko C, Rao RJ, Levine JE, Castle VP, Hutchinson RJ, Talpaz M, Robinson DR, Chinnaiyan AM. Integrative Clinical Sequencing in the Management of Refractory or Relapsed Cancer in Youth. JAMA 2015; 314 (9): 913-925. PMC: 4758114

Date last edited: August 9, 2017

Clinical Cancer Genetics Rotation

Three Month Rotation – *didactic focus*

Week 1:	Genetic Testing – Principles and Practice
	Clinical correl: Breast Cancer, Colon Cancer, VHL
Week 2:	Oncogenes, Tumor Suppressor Genes, "Caretaker Genes"
	Clinical correl: Familial Adenomatous Polyposis, HNPCC
Week 3:	Cancer Risk Assessment
	Clinical correl: Hereditary Breast Ovarian Cancer Synd.
Week 4	Managing Individuals at High Risk
	Clinical correl: von Hippel Lindau Syndrome, LFS
Week 5	Genotype-Phenotype Correlations
	Clinical correl: VHL, FAP
Week 6	GeneticRisk Assessment Models
	Clinical correl: BRCApro, MMRPro, PREMM1,2,6
Week 7	Incomplete Penetrance, Variable Expressivity and Phenocopies
	Clinical correl: BRCA, LFS, Prostate cancer
Week 8	Hereditary Pediatric Tumors
	Clinical correl: LFS, VHL, Retinoblastoma, Neuroblastoma
Week 9	MEN and Other Hereditary Cancer Syndromes
	Clinical correl: MEN2A,B, Melanoma

Clinical Cancer Genetics Rotation

Case-Based Self-Study Program

Case 1: FAP – 32 year old woman with a history of thyroid cancer and familial polyposis.

Case 2: Hereditary Breas/Ovarian cancer – 46 year old woman with fibrocystic breast disease and family history of breast cancer in mother (bilateral 40, 60), maternal grandmother (38) and maternal aunt (78).

Case 3: Lynch Syndrome 44 year old healthy male with a family history of colon cancer in his brother (32, 40), father (46), paternal grandmother (63).

Case 4: NF1 – 40 year old woman with segmental neurofibromatosis and a hard mass.

Case 5: PTEN- 62M with multiple renal tumors

Case 6: VHL – 37 year old man with polycythemia, cerebellar hemangioblastoma, and a kidney nodule.

Date last edited: August 9, 2017

MEDICAL GENETICS RESIDENCY PROGRAM
Department of Pediatrics

University of Michigan Health Systems
1500 E. Medical Center Drive
D5240 MPB
Ann Arbor, MI 48109-5718

(734) 763-6767
(734) 763-6561 (fax)

Cytogenetics Rotation for Medical Genetics Residents

Laboratory Director: Lina Shao, M.D., Ph.D., FACMG
Clinical Cytogenetics Laboratory – Pathology
2900 Huron Parkway Rm 1127
Ann Arbor, MI 48109

Goals and objectives for residents in the Clinical Cytogenetics Rotation: To learn fundamental concepts, specific knowledge of the field, and the methods that are used, as well as to acquire basic skills in clinical cytogenetics.

Patient Care

Residents will learn appropriate indications for sending karyotypes from peripheral blood, amniocytes, bone marrow, and other tissues. Residents will learn the implications of test results on health and disease.

Medical Knowledge

Cytogenetics is a cornerstone of Medical Genetics, having application to our understanding of abnormal development at every stage and in virtually every tissue of the human body. As chromosomes are the physical embodiment of our genetic blueprints, cytogenetics intersects with molecular genetics, cell biology and development. As such, a general knowledge of some basic concepts in several related areas is required to understand cytogenetics:

List of related concepts/basic knowledge to cover prior to the rotation:

Cell biology: the cell cycle, meiosis, mitosis, chromosome and chromatin structure, recombination; Embryology: Gametogenesis (male and female), fertilization, embryogenesis, placental development: formation of chorionic villi, amniotic fluid; Human genetics: modes of inheritance, autosomal vs. sex-linked inheritance, linkage vs. synteny, expression of phenotypes—penetrance, variable expressivity, anticipation, heterogeneity; Molecular genetics: DNA probes, hybridization techniques, classes of repetitive sequences, unique DNA sequences.

Major concepts and current topics in cytogenetics can be comprehensively covered by reading recent textbooks, reviews and benchmark journal articles in the field. Some of the basic concepts in cytogenetics to be studied are listed below. Additional topics will be determined by the types of samples submitted to the laboratory and abnormalities detected during the rotation. Then, in practice, one can follow the flow of decision-making in the laboratory and the ultimate interpretation and significance of the results

obtained. In this way, one can better understand the importance of having a clinical history and indication for cytogenetic studies provided with the sample for the proper handling of different types of specimens, the importance of integration of the clinical history for the correct interpretation of results, and the significance of the results for further studies, genetic counseling and involvement of additional family members. A reference list and copies of selected references will be provided (see attached).

Basic concepts in cytogenetics and molecular cytogenetics to be acquired by the end of the rotation:

Properties of autosomes vs. sex chromosomes, nondisjunction, trisomy, monosomy, ploidy, mosaicism, X-inactivation, structural rearrangements, meiotic segregation behavior, microdeletions, uniparental disomy, loss of heterozygosity, imprinting, fragile sites, the pseudoautosomal region, polymorphisms, variants, copy number variants, NORs, euchromatin, heterochromatin (facultative, constitutive), constitutional vs. acquired abnormalities.

In addition to general concepts, specific knowledge of the field will be covered, including historical developments, the common cytogenetic abnormalities and syndromes, and the various syndromes that have associated cytogenetic manifestations, as outlined below.

A. Historical landmarks in cytogenetics: use of mitotic inhibitors, mitogens, hypotonic solution, fixative, discovery of the human chromosome number, Lyon hypothesis, chromosome banding methods, prenatal diagnosis, fluorescence in situ hybridization (FISH), molecular cytogenetics, genomic and SNP microarrays

B. The most common constitutional cytogenetic abnormalities and their association with congenital syndromes and spontaneous abortions, especially with regard to phenotype and recurrence risks:
   1) Numerical abnormalities
      a) Autosomal trisomies: +21, +13, +18
      b) Sex chromosome abnormalities: Turner syndrome, including X/XY mosaicism, XXX, XXY, XYY
      c) Mosaicism: confined placental mosaicism, trisomy rescue
      d) Uniparental disomy, copy-neutral loss of heterozygosity
   2) Structural rearrangements
      a)Translocations: Robertsonian, reciprocal, balanced, unbalanced, 3:1 segregation -- eg: the constitutional t(11;22)
      b) Inversions: the inversion loop
      c) Duplication/Deletion syndromes
      d) Microdeletion syndromes: eg. Prader-Willi and Angelman
      e) Copy number variants, variants of uncertain significance; pathogenic vs benign

   3) Chromosome breakage/DNA repair/other syndromes with cytogenetic manifestations: Fragile X, Fanconi anemia, Bloom syndrome, ataxia telangiectasia, xeroderma pigmentosum, Werner syndrome, Roberts syndrome

Practice-Based Learning & Improvement

Methods in cytogenetics and molecular cytogenetics:

An important aspect of the rotation is observation of the methods that are used to obtain a cytogenetic result. Knowledge of routine laboratory procedures used to obtain karyotypes from amniotic fluid, chorionic villus, PHA-stimulated peripheral blood, and bone marrow samples is best obtained by watching and discussing the significance of each step of the process. The specific methods to be observed and to be discussed are outlined below.

A. Cell culture methods and chromosome preparation:
   1) Sample procurement, triage, sample preparation for adherent cell cultures (especially in situ cultures), and suspension cultures: Amniocentesis samples, chorionic villus samples (CVS), mitogen-stimulated peripheral blood samples (including prophase), bone marrow samples.
   2) Slide preparation, banding techniques (G-banding), special stains
   3) Analysis at the microscope, photomicrography, automated scanning, automated karyotyping
   4) Instrumentation: sterile laminar flow hoods, 5% CO_2 incubators, inverted microscopes, robotic harvester, microscopy (phase contrast, brightfield, fluorescence), automated karyotyping system, automated scanning system for metaphase cells

B. Fluorescence in situ hybridization (FISH):
   1) DNA Probes: chromosome painting, repetitive sequence probes, unique sequence probes, MFISH/SKY, limitations.
   2) Slide preparation, hybridization, detection (counterstains)
   3) Instrumentation: hybridization apparatus, fluorescence microscopy and color imaging
   4) Test Development and Validation:
      a) Metaphase vs interphase FISH analysis
      b) Probe validation, test validation (analytic sensitivity and specificity)
      c) Reportable ranges, cut-offs for an abnormal result

C. Cancer Cytogenomic Microarray and SNP analysis
   1) Indications for microarrays with actionable results
   2) Microarray platforms: Affymetrix Cytoscan HD platform; contains more than 2.6 million copy number markers, including 750,000 SNPs, and median spacing of 0.88 kb within genes.
   3) DNA extraction, QA, chip hybridization and washes
   4) Analysis software, resolution, making calls, QA
   5) Interpretation and reporting: databases, CNVs, runs of homozygosity

D. Analysis, Interpretation and Reporting:

1) Chromosome identification (karyotyping)
2) Levels of banding resolution for metaphase chromosomes
3) Number of cells, colonies appropriate for cytogenetic analysis
4) Mosaicism issues, chimerism
5) Maternal cell contamination
6) FISH analysis (metaphase vs. interphase)
7) Nomenclature: ISCN 2016, FISH ("ish"), microarray nomenclature

Interpersonal & Communication Skills

Residents will gain an appreciation for the necessity of accurate communication among lab staff and between lab staff and medical professionals.

Professionalism

Residents will learn to interact closely with laboratory technicians, the laboratory director, and other lab staff. Residents will also learn guidelines for sample handling and processing across departments and medical facilities.

Systems Based Practice

Residents will learn multiple aspects of lab management, including turn-around times, costs of tests, sample volumes, and quality control. They will become acquainted with ACMG Practice Guidelines and Laboratory Standards and Guidelines for Clinical Cytogenetics. Residents will also learn about CLIA and other certification.

References with handouts provided:
Medical genetics textbooks with introductory cytogenetics chapters:

Strachan, T., and Read, A.P. *Human Molecular Genetics,* 4th Ed. Garland Science, NY. 2011. (Chapter 2 provided)
Vogel, F, and A.G. Mitulsky. 1997. *Human Genetics: Problems and Approaches,* 3rd Ed. Springer-Verlag, New York. (Chapter 2 provided)

Confined Placental Mosaicism:

Lestou VS and Kalousek DK. Confined placental mosaicism and intrauterine fetal growth. *Arch Dis Child Fetal Neonatal Ed* 1998; 79:F223-F226. (provided)
Kalousek DK. Confined placental mosaicism and genomic imprinting. *Bailliere's Clinical Obstetrics and Gynaecology* 2000; 14:723-730. (provided)

General References
Counseling issues, recurrence risks:

Gardner, RJM, and Sutherland, GR. *Chromosome Abnormalities and Genetic Counseling*, 3rd Ed. Oxford University Press, NY. 2012.

Cytogenetic methodology:

Barch, MJ, Ed. *The AGT Cytogenetics Laboratory Manual*, 3rd Ed. Lippincott-Raven, NY. 1997.
Gersen SL, Keagle MB, Eds. *The Principles of Clinical Cytogenetics,* 3rd ed. Springer, New York. 2013.

Nomenclature:

ISCN (2016): An International System for Human Cytogenetic Nomenclature. McGown-Jordan J, Simons A, and Schmid M (Eds.). S. Karger, Basel. 2016.

Constitutional abnormalities:

Schinzel, A. *Catalogue of Unbalanced Chromosome Aberrations in Man*, 2nd Ed. deGruyter: New York. 2001.
Borgoankar, DSl *Chromosomal Variation in Man*, 8th Ed. Wiley-Liss: New York. 1997.
Milunsky A, Ed. *Genetic Disorders and the Fetus, 5th Ed.* Johns Hopkins Univ Press: Baltimore. 2004. Esp. chapter by Lillian Hsu.

Cancer cytogenetics (all neoplasia):
Heim, Sverre and Mitelman, Felix. *Cancer Cytogenetics*, 4th Ed. John Wiley & Sons, Hoboken, NJ. 2015. Chapters 1-4.

Websites
"Atlas of genetics and cytogenetics in oncology and haemotology"
http://atlasgeneticsoncology.org/

MEDICAL GENETICS RESIDENCY PROGRAM
Department of Pediatrics

University of Michigan Health Systems
1500 E. Medical Center Drive
D5240 MPB
Ann Arbor, MI 48109-5718

(734) 763-6767
(734) 763-6561 (fax)

Molecular Genetics and Genomics Laboratory Rotation Goals and Objectives

Director: Marwan Tayeh, Ph.D.

The goals and objectives of the Molecular Genetics and Genomics Laboratory rotation in the Medical Genetics Residency Program are to provide the resident with exposure to all aspects of care of molecular genetic diagnostics in accordance with the Residency Review Committee for Medical Genetics expectations and to fulfill criteria for board eligibility by the American Board of Medical Genetics.

Patient Care

The residents will become familiar with the evaluation, diagnosis and management of genomic disorders, chromosomal deletions, duplications, and copy number variation; the residents will also become familiar with the methods involved in DNA sequencing based diagnostic protocols, methylation analysis, Multiplex Ligation-dependent Probe Amplification (MLPA), and bioinformatics relevant to genetics diagnostics. Residents will gain expertise in performing and interpreting molecular analyses relevant to the diagnosis and management of human genetic diseases and will develop broad knowledge of basic molecular biology, the application of molecular methods through the diagnosis and management of genetic diseases and an understanding of the etiology, pathogenesis, clinical manifestations and management of human inherited genetic disorders.

Medical Knowledge

Through coursework and didactic sessions with attending physicians, residents will become familiar with fundamental concepts, molecular biology and genetics relevant to Mendelian single gene disorders, imprinting disorders, and to genomic disorders. Residents will also be required to interpret molecular genetic assays and distinguish de novo genomic disorders from copy number variation. Residents will also learn appropriate nomenclature for genetic mutations. Details about the curriculum are attached below.

Practice-Based Learning & Improvement

Residents will learn to apply appropriate diagnostic modalities in the management of patients with genomic and genetic disorders. Residents will also become familiar with laboratory procedures and fundamental molecular biology that encompasses diagnostic

tests, including PCR, sequencing, Southern blotting, and array comparative genomic hybridization and SNP based copy number and allele-specific assays.

Interpersonal & Communication Skills

Residents will learn how laboratory results are communicated to other physicians and laboratory directors, and will have opportunities to improve their own communication skills in the laboratory setting.

Professionalism

Residents will interact closely with laboratory staff and learn their responsibilities and tasks. Residents will learn how laboratory staff communicates with other hospital and laboratory personnel.

Systems Based Practice

Residents will learn the costs and benefits of diagnostic and therapeutic modalities in the evaluation of patients with genetic disorders, and how these costs are transferred to patients and insurance providers.

Curriculum:

Week 1

I. Discussion topics:
   - A. Pre-analytical issues: Specimen and Patient demographic requirements - Anticoagulants, fixatives, handling, storage, stability
   - B. Specimen processing from blood, other types of samples
   - C. Nucleic acid extraction, quantitation
   - D. Quality control of nucleic acid extractions

II. Laboratory Procedures:
   - A. Blood sample processing for nucleic acid extraction
   - B. Extraction, Quantitation of nucleic acid
     - Automated “robotic” procedure
     - Manual procedure
   - C. Spectrophotometric quantitation of nucleic acid
   - D. Quality assessment of extracted nucleic acids – gel electrophoresis

III. Discussion topics:
   - A. Nucleic acid analysis: Electrophoresis
     - Agarose gels
     - Acrylamide gels
     - Capillary electrophoresis
   - B. Nucleic acid detection methods
   - D. Nucleic acid amplification:
     - Polymerase Chain Reaction (PCR)
     - Whole genome amplification

IV. Laboratory Procedures
   - A. PCR based assays

Week 2

I. Discussion topics:
   A. DNA sequencing – Sanger methods and Next-Gen Sequencing

II. Laboratory procedures:
   A. Sequencing-based assays:

III. Discussion topics:
   A. DNA microarrays

IV. Laboratory procedures:
A. Array CGH and SNP-based arrays
B. Methyl- Sensitive PCR
C. Multiplex Ligation-dependent Probe Amplification (MLPA)

MEDICAL GENETICS RESIDENCY PROGRAM
Department of Pediatrics

University of Michigan Health Systems
1500 E. Medical Center Drive
D5240 MPB
Ann Arbor, MI 48109-5718

(734) 763-6767
(734) 763-6561 (fax)

Medical Genetics Clinic Goals and Objectives

Director: Wendy Uhlmann, MS, CGC and Shane C. Quinonez, MD

The primary objective of the Medical Genetics Clinic rotation is to introduce residents to the principles and practice of adult medical genetics. Our Medical Genetics Clinic has a rich history as the first genetics clinic in the country, established in 1941, and currently is one of the few adult medical genetics clinics nationwide. In this rotation, residents will work closely with genetic counselors to learn case preparation skills, pedigree construction and family history taking skills, risk assessment and risk communication skills and components of genetic counseling. The clinical geneticists will work with residents on understanding key concepts in adult genetics, evaluation and work-up of patients for genetic conditions, approach to the physical exam and assessment of dysmorphic features and generating and considering differential diagnoses. Both the genetic counselors and the clinical geneticists will work with residents on how to select and order genetic testing, understand insurance coverage issues and implications, interpret and communicate genetic test results.

Patient Care

Residents will provide care in our Medical Genetics Clinic to patients with known or suspected genetic conditions and/or positive family histories. Responsibilities will include case preparation, management of the clinic visit and follow-up post clinic.
Case prep and follow-up skills will include:

- Construct pedigree from family history form or at appointment (if family history form is not returned)
- Review medical records (including MiChart and any outside records received)
- Perform literature search to obtain articles about the genetic condition
- Generate differential diagnoses (when indicated)
- Perform risk assessment (think about risks for patient, children/future children, siblings etc.)
- Conduct search of genetic testing laboratories
- Select laboratory and complete paperwork
- Develop Powerpoint counseling aids to use for provision of genetic counseling and develop a plan for the session
- Obtain pertinent patient literature and contact information for national and Michigan chapters of support groups
- Track results of tests/evaluations

- Phone out results
- Write clinic visit summary note/letter
- Present summary of patients seen at post-clinic conference

Medical Knowledge

Residents will learn about adult medical genetics from reading, individual weekly meetings with genetic counselors and clinical geneticists, care of patients in clinic and post-clinic conferences. A goal of this rotation is to recognize issues specific to adult patients with a personal and/or family history of a wide spectrum of conditions including:

- Connective tissue disorders (e.g. Ehlers-Danlos syndrome, Marfan syndrome, Osteogenesis imperfecta)
- Aneurysm syndromes
- Neurocutaneous conditions (e.g. Neurofibromatosis, Tuberous sclerosis)
- Classic genetic conditions (e.g. Hereditary Hemorrhagic Telangiectasia, hemochromatosis, alpha-1 antitrypsin deficiency, hemoglobinopathies)
- Sex chromosome abnormalities (e.g. Turner syndrome, Klinefelter syndrome)
- Chromosome abnormalities and rearrangements (e.g. Translocations)
- Neurological conditions (e.g. Alzheimer's disease, ALS, muscular dystrophies, myotonic dystrophy, Charcot-Marie-Tooth syndrome)
- Intellectual disability and/or congenital anomalies

In addition, residents generally will have the opportunity to see patients considering predictive genetic testing for Huntington disease. The resident will learn about the international predictive genetic testing guidelines, pre and post-test counseling considerations, insurance implications, familial implications and potential ethical issues. Residents generally will also see some patients for preconception counseling and risk assessment.
If patients are not seen with some of the above conditions during the resident's rotation, the resident will be encouraged to read about the conditions.

Practice-Based Learning & Improvement

Residents will learn how to work-up, evaluate and provide care for patients seen in an adult medical genetics clinic. Residents will learn to provide risk assessment and risk communication for Mendelian and complex inherited conditions.

Interpersonal & Communication Skills

Residents will continue to build on their interpersonal and communication skills by working with a multidisciplinary team that includes clinical geneticists, genetic counselors and other rotators (genetic counseling students, medical students). Residents will be expected to communicate effectively with adult patients ranging from late teens to 70s, sometimes with multiple family members present. Residents will have opportunities to communicate with other clinicians and genetic testing laboratories. Through their work with genetic counselors, residents will learn about wording, tone and nuances in providing genetic information and assessing patient's understanding and psychological responses.

Professionalism

Residents will be expected to be professional in their interactions, to assume full responsibility for the tasks described above and to comply with HIPAA guidelines in their interactions with patients and the clinical team. Specifically, residents will be expected to be attuned to the privacy and confidentiality issues that genetic information poses, particularly an issue when multiple family members are seen unbeknownst to each other.

Systems Based Practice

Residents will learn about the different databases to access 1) information about genetic conditions (e.g. MedGen, GeneReviews, OMIM) 2) information about genetic testing (e.g. Genetic Testing Registry, GeneTests) and 3) support group information for patients (e.g. Genetic Alliance, NORD, Michigan Genetics Resource Center). Residents will also learn the different tasks needed to select and order genetic tests and find out about insurance coverage.

MEDICAL GENETICS RESIDENCY PROGRAM
Department of Pediatrics

University of Michigan Health Systems
1500 E. Medical Center Drive
D5240 MPB
Ann Arbor, MI 48109-5718

(734) 763-6767
(734) 763-6561 (fax)

Neurogenetic Disorders Residency Training Goals and Objectives

Director: John Fink, M.D.

Patient Care

The goals of the Neurogenetic Disorders rotation for Medical Genetics Residents are to develop clinical skills in the principles and practice of genetics related to neurological inherited syndromes. The objectives of the rotation in Neurogenetics are to develop familiarity with diagnosis, management and genetic counseling of patients (children, adolescents, and adults) with inherited neurologic disorders. The Resident will learn to build a differential diagnosis and to order and interpret appropriate diagnostic testing. The resident will learn the natural history, presentation, complications, molecular basis, inheritance pattern and prevailing treatment for neurogenetic syndromes.

Medical Knowledge

This rotation will consist of primarily outpatient consultations and evaluations, self-study, genetic counseling and development of interdisciplinary team approaches to the care of families with genetic neurologic conditions including herediatry spastic paraplegia, spinocerebellar ataxias, Charcot-Marie-Tooth, dystonia, familial dementia, Tourette's syndrome, Wilson's disease, Niemann-Pick disease, Von-Hipple-Lindau disease, Fabry disease, CADASIL, episodic ataxias, and mitochondrial encephalomyopathies.

Practice-Based Learning & Improvement

The resident is required to calculate recurrence risks, order and interpret the results of diagnostic tools, and preparation of correspondence between referring physicians and families including clinic notes and letters for families. Reading and case-based self study is recommended.

Interpersonal & Communication Skills

Residents will work closely with Dr. Fink and other physicians and nurses in the Neurogenetics clinic. Residents will learn appropriate diagnosis, treatment, and recurrence risk counseling for a variety of neurogenetic diseases.

Professionalism

Medical Genetics Residents will be responsible for attending clinic and inpatient consults. They will work with the attending physician in management of patient inquiries. They are responsible for obtaining the medical history for referred patients in combination with the attending

physician, performing a relevant neurological physical examination and providing appropriate genetic counseling when required.

Systems Based Practice

Residents will learn the costs and benefits of diagnostic tests and studies, and their relevance to care and management of patients with Neurogenetic disorders. Residents will become familiar with the clinical and economic burden of neurodegenerative diseases on patients, families, and society.

MEDICAL GENETICS RESIDENCY PROGRAM
Department of Pediatrics

University of Michigan Health Systems
1500 E. Medical Center Drive
D5240 MPB
Ann Arbor, MI 48109-5718

(734) 763-6767
(734) 763-6561 (fax)

Pediatric Genetics Residency Training Goals and Objectives

Director: Catherine E. Keegan, MD, PhD

The purpose of the Pediatric Genetics Rotation is designed to provide the resident with exposure to all aspects of Pediatric medical genetics in accordance with the Residency Review Committee for Medical Genetics and will aid in fulfillment of criteria for board eligibility by the American Board of Medical Genetics. Residents will gain expertise in performing a competent medical genetics diagnostic evaluation, genetic counseling and case preparation skills, DNA, cytogenetic and metabolic diagnostic testing, and evaluation and management of inpatients and outpatients with known or suspected genetic disorders.

Patient Care

Residents will participate actively in the inpatient and outpatient evaluation and management of patients with a variety of known or suspected genetic and biochemical genetic disorders. This includes follow-up of test results, putting together an appropriate differential diagnosis, understanding treatment modalities and options, and managing inpatients on the Pediatric Genetics Service.

Medical Knowledge

Conditions and major concepts that residents are expected to gain familiarity with during their Pediatric Genetic rotation:

1. Multiple Anomalies and/or Intellectual Disability

Objectives:

- Know the basic underlying mechanisms of normal and abnormal morphogenesis.
- Distinguish between syndromes, field defects, and associations.
- Understand how to do an appropriate work up in a patient who presents with multiple anomalies.
- Become familiar with the terminology used in dysmorphology.
    - (see handout)
- Learn how to take a family history, analyze the pedigree and assess risks.
- Understand the concepts of minor and major anomalies and know what kinds of studies/evaluations are available (MRI, echocardiogram, etc.) and may be required to determine the extent of anomalies. Understand how to coordinate a cost-effective evaluation of a pediatric patient who presents with dysmorphic features and/or cognitive delays.

- Know that improvements in resolution of chromosomal microarray testing augmented by FISH necessitates that consideration of a chromosomal abnormality must be entertained in these individuals even if they had normal karyotypes before.
- Know that the features of a syndrome may change with age. Different medical problems may occur as an infant grows.
    - Some examples:
        - Williams syndrome
            - ✓ Facial features coarsen with age, joint contractures develop
        - Down syndrome
            - ✓ As individuals with Down syndrome age, there is an increased risk of seizures and early onset Alzheimer's disease
        - Prader-Willi syndrome
            - ✓ Weight management is of paramount importance and obese adults with PWS are at significant risk for heart disease and diabetes
- Recognize psychosocial problems of pediatric patients with birth defects.
- Be aware of services available for children with special needs due to physical disabilities and/or cognitive impairment.
- Apply appropriate counseling for known/unknown diagnoses.
- Learn how to use the medical specialists in the evaluation and management of cases.

2. Approach to Multifactorial Diseases in Pediatrics

Objectives:

- Understand the concepts of multifactorial inheritance.
- Understand the concept of empiric recurrence risks.
- Familiarize yourself with genetic counseling issues as related to the following common birth defects:
    - Isolated, non-syndromic cleft lip and/or cleft palate
    - Neural tube defects
    - Congenital heart defects
    - Congenital hip dysplasia
    - Pyloric stenosis
    - Etc.
- Understand the risks to fetuses of the above conditions and the medications to treat them, and surveillance during pregnancy (e.g. echocardiograms for congenital heart disease, ultrasound for NTD). Know how to use Reprotox and related services to assess teratogenicity of various medications that are commonly used in the adult population.

3. Teratogenesis

Objectives:

- Understand the principles of dose, agent, and time of exposure.
- Understand the molecular effects of certain teratogens.
- Understand and learn to recognize the consequences of teratogenic exposures.
- Know the risks of agents known to be teratogenic and those that are not.
- Know how to appropriately counsel women who may have a pregnancy wherein teratogenic exposure has or potentially has occurred.

4. Chromosome Disorders I - Aneuploidies

Objectives:

- Become familiar with general clinical presentations of common autosomal and sex chromosome aneuploidies including Trisomy 21, Turner syndrome, Klinefelter syndrome, and other sex chromosome abnormalities.
- Understand the concepts of non-disjunction, translocation and counseling/evaluation for translocation carriers.
- Familiarize yourself with counseling issues for Trisomy 21 at birth and in adulthood.
- Understand reproductive issues for affected individuals including the potential application of assisted reproduction technologies.
- Understand the indications for ordering chromosomal microarrays and how this technology differs from standard karyotypes
- Become familiar with copy number variations and counseling issues that arise in de novo vs. inherited copy number variants

5. Chromosomal Disorders II – Structural Abnormalities

Objectives:

- Know the different types of structural chromosome alterations: deletions, inversions, translocations, duplications.
- Become familiar with the phenotypic characteristics, medical management, and counseling issues of patients with common microdeletion and microduplication syndromes. (see your handbook)
- Understand the reproductive risk of having a "balanced translocation" and related genetic counseling issues.
- Understand the reproductive risk of having a Robertsonian translocation, and related genetic counseling issues.

6. Neurocutaneous Disorders

Objectives:

- Know general characteristics of neurocutaneous disorders and understand how to evaluate individuals with these conditions including physical exams, imaging studies, and laboratory studies.
- Become familiar with the diagnostic criteria for, management and counseling issues in Pediatric patients for:
    - Neurofibromatosis Type I and II
    - Tuberous sclerosis
    - Von Hippel Lindau
- Know the value and application of DNA diagnostic studies in these conditions.

7. Connective Tissue Disorders

Objectives:

- Become familiar with the general features and learn how to evaluate individuals with suspected connective tissue disorders.
- Know how to do appropriate measurements and specific clinical tests as they relate to diagnostic criteria.
- Understand how to classify connective tissue disorders and what type of laboratory testing is indicated.
- Familiarize yourself with management and genetic counseling issues for the following connective tissue disorders:
    - Osteogenesis Imperfecta
    - Marfan syndrome and other fibrillinopathies
    - Ehlers-Danlos syndromes
    - Stickler syndrome

8. Disorders of Coagulation

Objectives:

- Understand the molecular genetic basis, genetic counseling, and genetic testing, of:
    - Von Willebrand disease
    - Hemophilia A & B
    - Inherited thrombophilias

9. Hemoglobinopathies

Objectives:

- Understand the molecular genetic basis, genetic counseling, and genetic testing, of:
    - Sickle Cell Anemia
    - Thalassemia (a and B)

10. Craniosynostosis Syndromes

Objectives:

- Familiarize yourself with the general approach to the evaluation of craniosynostosis, understanding the distinction between primary and secondary forms.
- Understand the importance of making a specific diagnosis and of arranging for specific molecular testing if indicated.

- Familiarize yourself with the following craniosynostosis syndromes (be able to compare and contrast):
    - Apert syndrome
    - Crouzon syndrome
    - Carpenter syndrome
    - Pfeiffer syndrome
    - Non-syndromic coronal synostosis (Muenke syndrome)

11. Neurogenetic Syndromes

Objectives:

- Familiarize yourself with the general approach to evaluating neurological regression and/or deterioration in both children and adults.
- Understand the importance of making a specific diagnosis and of using appropriate metabolic and/or molecular testing.
- Understand the class of trinucleotide repeat disorders.
- Understand the concept of predictive genetic testing for adult onset neurological conditions and be aware of ethical issues and management issues surrounding predictive testing.
- Familiarize yourself with the following neurological disorders:
    - Hereditary Motor and Sensory Neuropathy (Charcot-Marie-Tooth Disease)
    - Inherited Ataxias with late adolescent, adult onset
    - Huntington Disease
    - Myotonic Dystrophy
    - Spinal Muscular Atrophy (Types I, II, III)

12. Principles of Metabolic Abnormalities: Biochemical Genetics

Residents will gain expertise in performing and interpreting biochemical analyses relevant to the diagnosis and management of human genetic diseases and will develop broad knowledge of basic biochemistry and biology, the application of biochemical methods through the diagnosis and management of genetic diseases and an understanding of the etiology pathogenesis clinical manifestations and management of human inherited biochemical disorders. Residents will learn to apply appropriate diagnostic and therapeutic modalities in the management of inpatients with metabolic disease crises and to provide clinical care directed at selected biochemical disorders.

Objectives:

- Know what is meant by an inborn error of metabolism.
- Understand how to approach and manage infants and children with metabolic disorders in both emergent and chronic situations.
- Become familiar with initial and long-term management issues for the following metabolic disorders including issues related to pregnancy.
- Be familiar with the symptoms, referral basis, history, diagnosis and management of:
    - Phenylketonuria
    - Galactosemia
    - Homocystinuria
    - Lysosomal Storage Disorders
    - Wilson's Disease
    - Tay Sachs
    - Gaucher Disease
    - Hemochromatosis
    - Maple syrup urine disease
    - Biotinidase deficiency
    - Glycogen storage diseases
    - Disorders of glycoprotein metabolism (CDG)
    - Disorders of peroxisomal biogenesis
    - Urea cycle disorders
    - Disorders of fatty acid oxidation or lipid cholesterol metabolism
    - Organic acidemias
    - Disorders of creatine synthesis

- Understand teratogenic risks of maternal PKU.

13. Skeletal Dysplasias

Objectives:

- Understand how to clinically classify and approach the diagnosis and management of skeletal dysplasias.
- Understand the diagnostic distinctions of the numerous skeletal dysplasias and the chondrodysplasias.
- Become familiar with management, DNA testing, and genetic counseling for:
    - Achondroplasia
- Know what resources are available for individuals with a skeletal dysplasia.

14. Imprinting

Objectives:

- Understand the concept of imprinting and the consequences of mutations involving imprinted genes or chromosomal regions.
- Be familiar with the diagnostic criteria, molecular causes and management of the following imprinted conditions:
    - Prader-Willi Syndrome
    - Angelman Syndrome
    - Beckwith-Wiedemann Syndrome

15. Mitochondrial Genetics

Objectives:

- Understand the essential nature and functions of mitochondria in metabolism.
- Understand the concept of heteroplasmy.
- Understand the inheritance pattern of mitochondrial diseases.
- Be able to recognize pedigrees and symptoms that may suggest mitochondrial disorders.
- Know how to work-up and manage mitochondrial disorders.
- Learn the counseling issues related to inheritance, penetrance, and expressivity of mitochondrial diseases.
- Know the common mitochondrial diseases and their varied presentations.

16. Predictive and Presymptomatic Risk Analysis: Application of Bayes' Theorem

- Become familiar with the general principles of calculating recurrence risk.
- Understand how to utilize Bayes Theorem and conditional probabilities.
- Know how to modify risks based on genetic testing.
- Understand concepts of sensitivity, specificity, positive predictive value and negative predictive value.

Practice-Based Learning & Improvement

Residents will learn how to use Genetic Databases for Clinical Genetics

London/Oxford Medical Databases, located in MPB 5th floor

Syndroc, **http://syndroc.hospvd.ch:8004/**

GeneTests, **http://www.genetests.org**

GeneReviews, **http://www.genereviews.org**

Online Mendelian Inheritance in Man, OMIM (TM). Center for Medical Genetics, Johns Hopkins University (Baltimore, MD) and National Center for Biotechnology Information, National Library of Medicine (Bethesda, MD), 1996. World Wide Web URL: **http://www.ncbi.nlm.nih.gov/Omim/**

Information for Genetic Professionals through the University of Kansas Medical Center (with links to other sites, support groups)
http://www.kumc.edu/gec/geneinfo.html

Human-Mouse Homology Database
http://www.hgmp.mrc.ac.uk/DHMHD/dysmorph.htm/

Taubman Medical Library
http://www.lib.umich.edu/libhome/Taubman.lib/

"University of Michigan Pediatric Genetics video and photograph database of genetics syndromes, please see Shane C. Quinonez MD for access"

Interpersonal & Communication Skills

Residents will learn appropriate preconception counseling for a variety of conditions, including:

- Family history Duchenne/Becker Muscular Dystrophy
- Family history Cystic Fibrosis
- Consanguinity
- Recurrent pregnancy loss
- Preconception counseling

Professionalism

Residents will learn basic ethical frameworks and applications to genetic cases such as privacy and confidentiality of genetic information.

Systems Based Practice

Residents will learn multiple aspects involved in the interpretation of Laboratory Genetic tests, including:

- Cytogenetics
- Molecular Diagnostics
- Biochemical Tests

Residents will also learn how to find and utilize information and services provided by Support Groups/ Foundations to provide information to patients. They will also learn how to access information about Genetic counseling paradigms ("A Guide to Genetic Counseling". *Edited by Diane Baker, Jane Schuette, and Wendy Uhlmann*), about gene therapy development/trials (understand the rationale for, developing modalities, and limitations of gene therapy for genetic

disease). They will also learn about Clinical trials and research - Informed consent issues, dissemination of information issues (i.e. the General Clinical Research Center, University of Michigan) and ACGME issues for Medical Genetics Residents (ACGME Homepage http://www.acgme.org)

MEDICAL GENETICS RESIDENCY PROGRAM
Department of Pediatrics

University of Michigan Health Systems
1500 E. Medical Center Drive
D5240 MPB
Ann Arbor, MI 48109-5718

(734) 763-6767
(734) 763-6561 (fax)

Prenatal Genetics Residency Training Goals and Objectives

Prenatal Genetics

Marjorie C. Treadwell, M.D., Director
Clark Nugent, M.D.
Deborah Berman, M.D.
Lori Day, M.D.
Mark Chames, M.D.
Cosmas VandeVen, M.D.
Elizabeth Langen, MD
Lauren Mohnach, MS, CGC
Audrey Norby, MS, CGC
Beth Dugan, MS, CGC

The goals of the Prenatal Genetics rotation for Medical Genetics Residents are to introduce the Medical Genetics resident to the principles and practice of Prenatal Genetics. This rotation consists primarily of outpatient consultations and evaluations. Residents are involved in genetic counseling and development of interdisciplinary team skills relative to the care of families with prenatal fetal malformations and genetic conditions. The patient population for which prenatal genetics care is provided primarily includes pregnant women for whom genetic counseling is provided due to advanced maternal age, family history that increases the risk of genetic disease, maternal diseases that place a fetus at increased risk, or identification of fetuses with major and minor anomalies detected or suspected by ultrasound, or increased risk based on biochemical or karyotype abnormalities. Some counseling is also provided to couples presenting for certain assisted reproductive technologies. For conditions not presenting during the rotation, it is expected the information will be learned through the independent study opportunities available.

Medical Genetic Resident Responsibilities

The resident will become familiar with evaluating the potential for DNA-based genetic testing or other testing or screening, both non-invasive and invasive, involving amniocentesis or chorionic villus sampling and non-invasive prenatal screening with cell-free DNA, preparing risk estimates for recurrence risk, presenting diagnostic tools, evaluating diagnostic test results and preparing of correspondence between referring physicians and families. Reading and case-based self study is recommended. Residents are also responsible for attending the Fetal Development Conference (a multi-disciplinary conference reviewing cases with abnormalities presenting during the preceding week) and may present cases in which they were involved.

Date last edited: June 2, 2016

Patient Care

Medical Genetics residents will be responsible for obtaining the medical history for referred patients in combination with the attending physician or genetic counselor, participating in a relevant physical examination and providing appropriate genetic counseling when required. The resident will become familiar with the evaluation, diagnosis and management of prenatally diagnosed genetic disorders, including metabolic diseases. Residents will gain experience in counseling pregnant women and their partners for recurrence risks, amniocentesis, chorionic villus sampling, and prenatal ultrasound as well as other screening or diagnostic tests available.

Medical Knowledge

Residents will develop familiarity with diagnosis, management and genetic counseling related to the following ultrasound findings and medical conditions:

- Ultrasound markers for aneuploidy
- Increased nuchal lucency
- Cystic hygroma
- Congenital heart defects
- Multiple congenital anomalies
- Renal anomalies
- CNS anomalies
- Skeletal anomalies
- Abnormalities of placenta development or function and effects on the fetus
- Chromosome anomalies
    - Common (Tris 21, 18, 13)
    - Confined placental mosaicism
    - Balanced translocations
    - Robertsonian translocations
- Advanced maternal age
- Hydrops (immune and non-immune)
- Fetal demise
- Oligohydramnios
- Polyhydramnios
- Twins and related problems

Residents will also understand the appropriate counseling and use of:

- Chorionic villus sampling
- Amniocentesis
- Cordocentesis
- Non-invasive prenatal testing
- First trimester screening
- Maternal serum quad screening

Practice-Based Learning & Improvement

Residents will learn to apply appropriate diagnostic and therapeutic modalities in the prenatal diagnosis. Residents will also become familiar with laboratory tests and surgical procedures that encompass prenatal screening tests.

Interpersonal & Communication Skills

Residents will learn appropriate communication between prenatal genetic counselors and physicians and laboratory directors, and will have opportunities to improve their own communication skills in the prenatal clinic setting. They will also participate and have the opportunity to increase their awareness of counseling patients regarding unanticipated abnormalities in the fetus.

Professionalism

Residents will interact closely and appropriately with prenatal staff, and learn their responsibilities and tasks.

Systems Based Practice

Residents will learn the costs and benefits of diagnostic and therapeutic prenatal modalities in the management of patients with known or suspected genetic disorders. Residents will also learn how these costs are transferred to patients and insurance providers.