GOALS & OBJECTIVES: Medical Genetics Training Program

The overall goal of the clinical genetics training program is to provide the trainee with the skills they will need to provide appropriate diagnostic testing and management for a wide variety of genetic disorders, inborn errors of metabolic disease and congenital anomalies. This includes providing family centered patient care that is developmentally and age appropriate, compassionate, and effective for treatment and promotion of health in individuals with genetic disorders as well as using a logical and appropriate clinical approach to the care of patients presenting for a medical genetics evaluation, applying principles of evidence-based decision-making and problem solving. In addition, the trainee will also be prepared to describe indications for procedures used in medical genetics and interpret results for families.

The specific skills to master are outlined below:

Patient Care:

1. Diagnosis of genetic disorders: Gather essential and accurate information about patients in whom a genetic disorder is being considered
    - Elicit present and past medical history, habits, exposures, medications, allergies, contraceptives, pregnancy, prevention and screening activities
    - Record developmental milestones, loss of milestones, and neurodegenerative symptoms
    - Construct at least a detailed three generation pedigree and document ancestry, consanguinity
    - Gather information about social history
    - Perform a thorough physical examination on a child suspected of a genetic disorder, identifying major and minor anomalies that could be signs of a specific genetic disorder or malformation syndrome
    - Note pertinent positives and negatives, and display sensitivity and respect privacy
    - Explain the findings on clinical history and examination that suggest a known or potential genetic disorder, inborn error of metabolism, or malformation syndrome
    - Be able to use printed, computer-based, and internet resources to aid in generating a differential diagnosis and diagnosing genetic disorders, using physical findings and laboratory examination. The resources may include:
        - Smith's recognizable patterns of human malformation
        - Pictures of standard syndromes and undiagnosed malformations (POSSUM)
        - Inborn errors of metabolism
        - Online Mendelian Inheritance in Man (OMIM)
        - GeneTests
        - Pubmed
    - Use appropriate laboratory tests to help identify genetic diseases, inborn errors of metabolism, or malformation syndromes. Explain the reason for the test to the family and interpret the results. The tests may include:
        - Chromosome analysis
        - FISH testing for specific disorders and commonly encountered aneuploidies, and subtelomeric analysis
        - Biochemical screens (amino acids, organic acids, ammonia, lactate, pyruvate, acylcarnitine profiles)
        - DNA testing for selected disorders, including hereditary cancer syndromes

        - Newer and future genetic technologies (e.g., comparative genomic hybridization (CGH) and microarray technologies)
    - Discuss the ethical, legal, and social issues involved in genetic testing, such as testing children for incurable genetic disorders, especially if they present in adulthood
2. Management of genetic disorders: Learn to provide treatment and promote health for individuals with genetic conditions
    - Implement treatment plans for genetic disorders, understanding principles of management and practice guidelines
    - Understand tests used to monitor treatment and/or natural history of genetic disorders
    - Describe how well-child care differs in a child with certain genetic conditions, e.g., use of different growth charts, different developmental milestones, and guidelines for monitoring
    - Understand how management plan is influenced by environmental and lifestyle factors
3. Screening for genetic disorders: Gather essential and accurate information about patients who are at risk for a genetic disorder
    - Identify individuals for whom genetic screening is appropriate, based on family history
    - Screen for known familial genetic disorder processes using appropriate tests
    - Understand prenatal screening methods, such as amniocentesis, chorionic villus sampling, maternal serum markers, and high resolution ultrasound
    - Understand state mandated newborn screening programs and indications for follow-up testing
4. Genetic counseling: Understand the role of the medical geneticist in informing families about genetic disorders, inborn errors of metabolism and/or malformations, including diagnosis, laboratory results, family history, natural history, treatment, risk for occurrence in relatives, and screening
    - Be able to provide counseling to parents and patients that addresses:
        - Folic acid supplementation before and during pregnancy
        - Avoidance of known teratogens during pregnancy (e.g., isotretinoin and alcohol)
        - Possible genetic screening for disease during pregnancy
        - Possible genetic screening for certain ethnic groups
        - Disorders identified in the state newborn screening program
        - Possible genetic screening for cancer and adult diseases
    - Be able to provide prenatal and postnatal genetic counseling to parents and patients with specific genetic disorders, addressing: genetic disorders with known inheritance patterns, based on the family's pedigree. Expected course of known genetic disorders. Risk factors, such as parental age and family history. Internet and other resources and support groups for known genetic disorders
5. Procedures: Describe the procedures used frequently by medical geneticists, including how and when they should be used. Competently perform the procedures
    - Wood's lamp examination of skin
    - Punch biopsy of the skin
    - Clinical photography
    - Phlebotomy
6. Documentation: Prepare medical record report, progress report, laboratory results, and letters
    - Write relevant information about medical history, physical exam, family history, laboratory data, diagnosis, treatment, management, and risk assessment to health professionals and families

Medical Genetic Knowledge:

1. Continually expand your knowledge of genetic disorders, metabolic disease, and/or congenital anomalies and malformations, including information on important concepts, natural history of disorders, mechanisms of disease, and molecular testing
    - Understand the scope of established and evolving biomedical, clinical, epidemiological, and social-behavioral knowledge needed by the medical geneticist
    - Acquire, interpret, and apply the knowledge appropriate for the care of individuals with genetic disorders
    - Critically evaluate current medical information and scientific evidence related to medical genetics and expand your knowledgebase accordingly
2. Concepts in medical genetics: Understand basic concepts in medical genetics and apply them to diagnosis, counseling and screening
    - Understand general concepts related to chromosome structure and function, gene structure, mutations, molecular and biochemical genetic techniques commonly used in diagnosis of genetic diseases
    - Describe common patterns of Mendelian and non-Mendelian inheritance, such as dominant, recessive, X-linked, and multifactorial. Demonstrate the ability to construct and analyze pedigrees.
    - Discuss unusual patterns of inheritance, such as mitochondrial inheritance, anticipation, and imprinting
    - Learn other principles of clinical genetics including preimplantation diagnosis, cancer genetics, population genetics, pharmacogenetics
3. Knowledge of specific genetic disorders: Develop habits and attitudes that promote expansion of your knowledge of genetic disorders, including information on natural history, mechanisms of disease, and molecular testing
    - Identify commonly encountered diseases and know their modes of inheritance, including cystic fibrosis, sickle cell anemia, Marfan syndrome, Huntington disease, neurofibromatosis, and familial cancer syndromes
    - Identify common disorders with unusual inheritance patterns and describe their modes of inheritance, including: a triplet repeat disorder (Fragile X syndrome) and a mitochondrial disorder (MELAS)
    - Thoroughly review current information on specific genetic disorders as they are encountered in your practice. Use such knowledge in patient care, teaching, and improving your overall understanding of genetic diseases
    - Develop strategies to learn about advances in medical genetics, in order to incorporate into one's practice improved screening, identification, counseling and management of such disorders
4. Evaluation of signs and symptoms: Know how to evaluate, treat, and/or refer patients with presenting signs and symptoms that suggest a genetic disorder
    - Create strategies to determine if the following presenting signs and symptoms are caused by a genetic condition and determine if the patient needs treatment or referral:
        - Miscarriage, stillbirth or neonatal death
        - Dysmorphic features
        - Multiple malformations
        - Cleft lip/palate
        - Ophthalmologic abnormalities
        - Hearing loss
        - Cardiac diseases
        - Respiratory problems
        - Endocrine problems

        - Hematologic disorders
        - Gastointestinal problems
        - Urogenital disorders
        - Skin lesions
        - Skeletal dysplasias
        - Limb defects
        - Connective tissue disorders
        - Vomiting
        - Poor feeding
        - Failure to thrive
        - Obesity
        - Hypotonia
        - Developmental delays
        - Seizures
        - Autism
        - Neurodegenerative and muscular diseases
        - Aging disorders
    - Identify criteria that warrant genetics consultation
    - Know how to distinguish among normal variation, genetic diseases, and acquired disorders

Practice-Based Learning

1. Show knowledge, skills, and attitudes needed for continuous self-assessment, using scientific methods and evidence to investigate, evaluate, and improve one's patient care practice
    - Identify standard guidelines for diagnosis and treatment of conditions commonly encountered in medical genetics and adapt them to the individual needs of specific patients
    - Identify personal learning needs related to medical genetics. Systematically organize relevant information resources for future reference. Plan for continuing acquisition of knowledge and skills
    - Thoroughly review current information on specific genetic disorders as they are encountered in your practice. Use such knowledge in patient care, teaching, and improving your overall understanding of genetic diseases
2. Commonly encountered genetic disorders, inborn errors of metabolism and/or congenital anomalies that are not considered urgent: Assist in diagnosis of genetic disorders and counseling of parents, with input from a geneticist.
    - Discuss the presenting signs and symptoms for commonly encountered genetic disorders and identify accepted guidelines for care:
        - Down syndrome in older children (not newborns)
        - Turner syndrome
        - Fragile X syndrome
        - Neurofibromatosis
        - Spina bifida
        - Marfan syndrome
        - Achondroplasia
    - Develop management plans for commonly encountered genetic disorders, identifying principles of long-term management, including use of disorder-specific growth charts and practice guidelines
    - Identify resources in your community for diagnosis, genetic counseling, therapy, and psychosocial support of children with genetic disorders and/or birth defects

3. Conditions requiring urgent management: Recognize and respond to urgent problems related to genetic and inherited metabolic disorders, such as:
    - Dysmorphic features found in chromosomal abnormalities that require prompt diagnosis in the newborn period, such as trisomies 13, 18, and 21
    - Infants presenting with symptoms that indicate the possibility of a severe inborn error of metabolism, for example: metabolic acidosis, hyperammonemia, unexplained seizures, ketosis or hypoketosis, profound hypoglycemia
    - Unexplained critical illness or death suggestive of metabolic disorder, requiring collection of tissue samples before or at time of death
    - Developmental delay with signs or symptoms suggesting an underlying metabolic or genetic disorder
    - Physiologic changes or regression of milestones that suggest a possible metabolic etiology, for example certain urea cycle disorders, mitochondrial disorders, lysosomal storage diseases, and certain organic acidoses or amino acid disorders
4. Commonly used diagnostic and screening procedures: Learn how to appropriately use and gain skill in interpreting the following tests
    - Radiologic tests: plain film examinations, such as chest films, spine films, skeletal surveys
    - Radiologic tests: ultrasound examinations, such as abdominal ultrasound
    - Imaging radiologic tests: CT and MRI (including spectroscopy (MRS)) of the brain
5. Molecular medicine in clinical practice: Recognize that genetic factors may be important in common diseases of childhood and adulthood
    - Identify standard guidelines for diagnosis and treatment of conditions commonly encountered in medical genetics and adapt them to the individual needs of specific patients
    - Discuss current knowledge regarding the molecular basis of common childhood and adult conditions
    - Recognize current and future uses of DNA tests in the office setting, such as identification of pharmacogenetic variants and testing for genetic predisposition to complex disorders
    - Develop a strategy to incorporate concepts of molecular medicine into the identification, treatment, counseling, and prevention of certain disease processes
7. Quality improvement: Reflect on areas of uncertainty to identify improvement needs and implement effect changes.
    - Critique research evidence and review accepted guidelines
    - Seek feedback and review evaluations from mentors, colleagues, and staff

Interpersonal & Communication Skills

1. Develop and practice good interpersonal and communication skills.
    - Use responsive, attentive, and reassuring behavior and provide psychosocial support
    - Show interpersonal and communication skills that lead to useful information exchange and partnering with patients, families, and professional associates
    - Provide effective patient education for conditions encountered in medical genetics
    - Maintain accurate, legible, timely, and legally appropriate medical records, including referral forms and letters, in the outpatient and inpatient setting.

Professionalism

1. Develop and practice professional attitudes and conduct skills.
    - Show personal accountability to the well being of patients (e.g., following-up lab results, writing careful notes, and seeking answers to patient care questions)
    - Show commitment to professional responsibilities, ethical and legal principles, and sensitivity to individuals

System-Based Practice

1. Understand how to practice high quality health care and advocate for patients within the context of the health care system
    - Identify key aspects of health care systems as they apply to medical genetics, including the referral and/or consultation process
    - Show sensitivity to the costs of clinical care in medical genetics, and take steps to minimize costs without compromising quality
    - Recognize and advocate for families who need assistance to deal with problems in the health care system, such as referrals, lack of insurance, pharmacy problems, multiple appointments with long transport times, or inconvenient hours of service
    - Recognize one's limits and those of the system
    - Take steps to avoid medical errors
    - Recognize the important role genetic counselors, nurses, nurse practitioners, nutritionists and social workers play in the care of patients with genetic disorders

Research Related Activities

1. Learn about IRB and HIPAA protocols
2. Learn skills to review the literature to determine significance of clinical findings by studying cases and cohorts of patients from our clinics and hospital for new symptoms, laboratory data, diagnosis, and treatment
3. Learn skills to report and write about new clinical findings by preparing powerpoint presentations and write abstracts and manuscripts for publications