GOALS AND OBJECTIVES FOR CLINICAL GENETICS

PATIENT CARE: the resident should demonstrate the ability to

- Gather genetic family-history information, including an appropriate multi-generational family history
- Educate clients about availability of genetic testing and/or treatment for genetic conditions. Provide appropriate information about the potential risks, benefits, and limitations of genetic testing
- Seek assistance from and refer to appropriate genetics experts and peer support resources
- Identify clients who would benefit from genetic services

MEDICAL KNOWLEDGE: the resident should demonstrate an understanding of

- Basic human genetics terminology (see Glossary)
- The basic patterns of biological inheritance and variation, both within families and within populations. The patterns of inheritance characteristic of autosomal dominant, autosomal recessive, X-linked dominant, and X-linked recessive traits
- The importance of family history (minimum three generations) in assessing predisposition to disease
- The clinical manifestations of common mendelian diseases (e.g. Marfan syndrome, NF1)
- The definition of variable expressivity and incomplete penetrance
- The clinical features of common numerical, structural and mosaic chromosomal abnormalities (e.g. trisomy 21, Turner syndrome)
- How to recognize and classify congenital anomalies and multiple congenital anomaly syndromes
- The basic principles of inborn errors of metabolism and their general clinical manifestations
- The genetic basis of mitochondrial diseases
- The nature of mutations and premutations and how they contribute to human variability and disease
- The concepts and clinical importance of genetic imprinting and uniparental disomy
- The purpose of genetic counseling. When and how to refer individuals with a genetic disease or congenital anomaly to medical genetics specialists, and why referral is beneficial to the patients.
- The difference between clinical ***diagnosis*** of disease and identification of genetic ***predisposition*** to disease. The concept that genetic variation is not strictly correlated with disease manifestation
- That the frequency of particular mutations varies within populations, the clinical implications of consanguinity, the continuing occurrence of new mutations, and that gene frequencies are not impacted by medical intervention
- Difference between constitutional and acquired genetic alternations in malignant neoplasms (e.g. breast cancer vs. BRCA)
- The potential advantages, limitations, and disadvantages of presymptomatic testing for genetic disease
- Conventional approaches to treatment of genetic diseases and the general status of gene-based therapies

- Common molecular and cytogenetic diagnostic techniques and how they are applied to genetic disorders
- What information can and cannot be predicted from the DNA sequence of a gene
- The advantage of measuring protein or enzyme levels to obtain information that cannot be obtained from the DNA sequence alone

INTERPERSONAL & COMMUNICATION: the resident should demonstrate the ability to

- Apply appropriate techniques for conveying difficult medical information
- Recognize the importance of reiterating information to patients/parents who are anxious or unfamiliar with the concepts being presented
- Educate clients about the range of emotional effects they and/or family members may experience as a result of receiving genetic information

PROFESSIONALISM: the resident should demonstrate the ability to

- Recognize the importance of delivering genetic education and counseling fairly, accurately, and without coercion or personal bias
- Appreciate the importance of sensitivity in tailoring information and services to clients' culture, knowledge and language level
- Provide and encourage use of culturally appropriate, user-friendly, materials/media to convey information about genetic concepts
- Cope emotionally with patient responses
- Recognize the limitations of one's own genetics expertise and seek consultation when necessary
- Recognize when personal values and biases with regard to ethical, social, cultural, religious, and ethnic issues may affect or interfere with care provided to clients. Respect patients' religious, cultural, social, and ethical beliefs, and recognize how their perspectives influence their use of genetic information and services
- Respect the autonomy of all patients, but also provide guidance with their decision-making when requested
- Convey to patients and other professionals the ethical, legal and social issues related to genetic testing and recording of genetic information (e.g., privacy, the potential for genetic discrimination by life insurance policies, the limitations to protection by GINA)
- Provide clients with an appropriate informed consent process to facilitate decision making related to genetic testing
- Safeguard privacy and confidentiality of genetic information of clients to the extent possible
- Inform clients of potential limitations to maintaining privacy and confidentiality of genetic information
- Appreciate the sensitivity of genetic information and the need for privacy and confidentiality
- Comprehend how legal and ethical issues related to genetics affect general medical practice

PRACTICE BASED LEARNING: the resident should demonstrate the ability to

- Obtain credible, current information about genetics for self, clients, and colleagues. Use effectively new information technologies about genetics to obtain current information about genetics

- Demonstrate willingness to update genetics knowledge at frequent intervals
- Effectively use resources such as medical textbooks, research articles, and computer-based systems to obtain information necessary for good patient care
- Apply the principles of evidence-based medicine to clinical practice
- Attend didactic sessions and diagnostic dilemma conferences (when available)
- Present a patient or a topic of interest at morning report

SYSTEM BASED PRACTICE: the resident should demonstrate an understanding of

- The influence of ethnicity, culture, related health beliefs, and economics in the clients' ability to use genetic information and services
- The potential physical and/or psychosocial benefits, limitations, and risks of genetic information for individuals, family members and communities
- The resources available to assist clients seeking genetic information or services, including the types of genetics professionals available and their diverse responsibilities
- Discuss costs of genetic services, benefits and potential risks of using health insurance for payment of genetic services, potential risks of discrimination
- The components of the genetic-counseling process and the indications for referral to genetic specialists
- The importance of coordination and collaboration with interdisciplinary team of health professionals by attending multidisciplinary clinics: Orthopedics-Genetics, Craniofacial clinic, Genodermatoses clinic, Cancer Genetics sessions, Prenatal Genetics clinic.
- How to make appropriate referrals to genetics support groups, community groups, or other resources that can benefit the patient and family
- How appropriate applications of genetic medicine can improve public health, and how to determine whether such interventions are warranted in a particular population
- The alternative approaches and goals of screening programs for genetic diseases in newborn infants, pregnant women, and other adults, and the ethical issues involved in justifying each program. The existence of and justification for screening programs to detect genetic disease, and the difference between screening and more definitive testing

SPECIFIC SKILLS: the resident should demonstrate the ability to

- Elicit a comprehensive family medical history, construct an appropriate medical pedigree, and recognize patterns of inheritance and other signs suggestive of genetic disease in the family history
- Recognize features in a patient's medical history, physical examination or laboratory investigations that suggest the presence of genetic disease
- Identify patients with strong inherited predispositions for common diseases and facilitate appropriate assessment of other at-risk family members
- ***Recognize and initiate the evaluation of patients with inborn errors of metabolism***
- Be able to determine recurrence risks for mendelian disorders in affected families
- Use the information that a patient has a genetic predisposition for a particular disease to help reduce the risk of developing that disease or deal with it more effectively if it does develop
- Communicate genetic information in a clear and **non-directive** manner that is suitable for individuals of different educational, socio-economic, ethnic and cultural backgrounds

- Recognize and accept varying cultural, social, and religious attitudes in relation to issues such as contraception, abortion, prenatal diagnostic techniques and raising a child with physical or mental disability
- Utilize community support services and agencies, in particular, support groups for genetic diseases, appropriately
- Discuss with patients diagnostic and predictive tests that are appropriate for the condition in their family and advise patients of the benefits, limitations, and risks of such tests
- Work with a medical genetics specialist and genetic counselors to develop a comprehensive plan for the evaluation and management of patients with genetic disease
- Introduce to patients with genetic diseases information regarding appropriate treatments, including dietary, pharmacological, enzyme-replacement, transplantation, and gene therapies
- Access anticipatory guidance regarding health screening practices specific to the diagnosis

SPECIFIC BEHAVIORS: the resident should demonstrate the ability to

- Present all relevant options fairly, accurately, non-coercively
- Be aware of the dilemmas posed by confidentiality when relatives are found to be at risk for a serious disease
- Appreciate the implications that information regarding a genetic abnormality can have for a person's self-image, family relationships, and social status and that patients' reactions may differ depending on factors such as gender, age, culture and education
- When appropriate, encourage patient participation in medical research provided the patient and/or family is fully informed and understands the risks and benefits of participation in terms of their own disease, treatment and social context.