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Training Options

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The American Board of Medical Genetics and Genomics (ABMGG) certifies individuals in various specialty areas of medical genetics and genomics. Medical genetics and genomics encompasses the application of genetics and genomics to medical care and as such is recognized by the American Board of Medical Specialties (ABMS) [www.abms.org] as one of the primary medical specialty certification boards.

The field of medical genetics and genomics is comprised of several different specialties. Of these, ABMGG currently offers certification in five specialties and two subspecialties, as outlined below. To be eligible for certification by the ABMGG, an individual must complete all training requirements, meet the specified criteria in the area of desired certification and provide the required supporting documentation.

Specialties in Medical Genetics and Genomics

Clinical Genetics and Genomics

A clinical geneticist is a physician who holds a U.S. or Canadian earned M.D. or D.O. degree, or equivalent*, who has trained a minimum of one year in an ACGME-accredited residency (that includes 12 months of direct patient care), and then has completed an ACGME-accredited residency in medical genetics and genomics. This can be done in a two-year categorical residency or in one of the 4-year combined residency training programs (combined programs with Medical Genetics and Genomics are offered with Pediatrics, Internal Medicine, Maternal Fetal Medicine, or Reproductive Endocrinology & Infertility). A clinical geneticist demonstrates competence to provide comprehensive genetic diagnostic, management, therapeutic, and counseling services.

These requirements imply that the individual possesses:

- broad knowledge in medical genetics and genomics, including an understanding of heterogeneity, variability, and natural history of genetic disorders;
- diagnostic and therapeutic skills in a wide range of genetic disorders;
- the ability to elicit and interpret individual and family histories;
- the ability to integrate clinical and genetic and genomic information and understand the uses, limitations, interpretation, and significance of specialized laboratory and clinical procedures;
- the expertise in genetic and mathematical principles to perform risk assessment;
- the skills in interviewing and counseling techniques required to: (1) elicit from the patient or family the information necessary to reach an appropriate conclusion; (2) anticipate areas of difficulty and conflict; (3) help families and individuals recognize and cope with their emotional and psychological needs; (4) recognize situations requiring psychiatric referral; and (5) transmit pertinent information in a way that is comprehensible to the individual or family; and
- knowledge of available health care resources (community, regional, and national) required for appropriate referral or support.

Clinical Biochemical Genetics

A diplomate certified in clinical biochemical genetics is an individual with a U.S. or Canadian earned doctoral degree (M.D., D.O., Ph.D.), or equivalent*, who can direct and interpret biochemical analyses relevant to the diagnosis and management of human genetic diseases, and who acts as a consultant regarding laboratory diagnosis of a broad range of biochemical genetic disorders.

These requirements imply that the individual possesses:

- the ability to supervise and direct the operations of a clinical biochemical genetics diagnostic laboratory, including technical expertise and knowledge in quality control and quality assessment procedures and adherence to regulatory requirements (e.g., CLIA, CAP);
- broad knowledge of (1) basic biochemistry and genetics, (2) the application of biochemical techniques to the diagnosis and management of genetic diseases, and (3) the etiology, pathogenesis, clinical manifestations, and management of human inherited biochemical disorders;
- an understanding of the heterogeneity, variability, and natural history of biochemical genetic disorders;
- diagnostic and interpretive skills in a wide range of biochemical genetic problems; and
- the ability to communicate biochemical laboratory results in the capacity of consultant to medical genetics professionals and other clinicians, and directly to patients in concert with other professional staff.

Laboratory Genetics and Genomics (check out the Laboratory Genetics and Genomics FAQs)
A diplomate certified in Laboratory Genetics and Genomics is an individual with a U.S. or Canadian earned doctoral degree (M.D., D.O., Ph.D.), or equivalent*, who can direct and interpret both clinical cytogenetic and molecular genetic analyses relevant to the diagnosis and management of human genetic disease. These individuals act as consultants in laboratory diagnoses for a broad range of molecular and chromosomal-based disorders, including both inherited and acquired conditions.
These requirements imply that the individual must possess:

- the ability to supervise and direct the operations of a clinical molecular genetics or clinical cytogenetics diagnostic laboratory, including requisite technical expertise, understanding of quality control and quality assessment procedures and adherence to regulatory requirements (e.g., CLIA, CAP);
- an understanding of the etiology, heterogeneity, variability, natural history and clinical management of disorders with a cytogenetic or molecular basis;
- an understanding of basic molecular biology, chromosome biology and genomic mechanisms of disease;
- the ability to select and apply the most appropriate current and evolving technologies, including karyotype, FISH, microarrays and next-generation DNA sequencing methodologies, to diagnose human disease.
- the ability to guide the development and validation of appropriate assays used in the evaluation of disorders with a chromosomal or molecular basis;
- the ability to interpret a broad range of genetic diagnostic tests, including methodologies that assess for chromosomal aneuploidies, structural chromosome rearrangements, genomic copy number variants (CNVs), single nucleotide variants (SNVs), structural changes within single genes, and absence or loss of heterozygosity (AOH/LOH);
- the ability to analyze various tissue types such as: tumors, cell free DNA, bone marrow, fibroblasts, and saliva.
- the ability to integrate clinical data (e.g., family history, physical examination, results of any diagnostic testing) into an individualized interpretation of laboratory results;
- strong communication skills that facilitate discussion of laboratory results with referring providers, and that allow a team approach to guiding further testing and clinical management;
- the ability to communicate cytogenetic and molecular genetic laboratory results directly to patients, when necessary, and in conjunction with others members of the clinical team.

Clinical Cytogenetics and Genomics
After July 1, 2017, no new trainees may begin training solely in Clinical Cytogenetics & Genomics or Clinical Molecular Genetics & Genomics. These specialties are now merged into Laboratory Genetics and Genomics.
A diplomate certified in Clinical Cytogenetics and Genomics is an individual with a U.S. or Canadian earned doctoral degree (M.D., D.O., Ph.D.), or equivalent, who can correctly perform and interpret cytogenetic analyses relevant to the diagnosis and

acquired conditions.

These requirements imply that the individual possesses:

- the ability to supervise and direct the operations of a clinical cytogenetic diagnostic laboratory, including technical expertise and knowledge in quality control and quality assessment procedures;
- broad knowledge in human cytogenetics, including prenatal and postnatal cytogenetic diagnosis, infertility, recurrent pregnancy loss, and of various cancers;
- an understanding of the heterogeneity, variability, and natural history of cytogenetic disorders; the analyses of various tissue types to diagnose and manage suspected or known diseases such as leukemias, lymphomas, and various solid tumors;
- diagnostic and interpretive skills in a wide range of cytogenetic and molecular cytogenetic and CGH microarray problems; and
- the ability to communicate cytogenetic laboratory results in the capacity of consultant to medical genetics professionals and other clinicians, and directly to patients in concert with other professional staff.

Clinical Molecular Genetics and Genomics
After July 1, 2017, no new trainees may begin training solely in Clinical Cytogenetics & Genomics or Clinical Molecular Genetics & Genomics. These specialties are now merged into Laboratory Genetics and Genomics
A diplomate certified in Clinical Molecular Genetics and Genomics is an individual with a U.S. or Canadian earned doctoral degree (M.D., D.O., Ph.D.), or equivalent*, who can correctly perform and interpret molecular analyses relevant to the diagnosis and management of human genetic diseases, and who can act as a consultant regarding laboratory diagnosis of a broad range of molecular genetic disorders.

These requirements imply that the individual possesses:

- the ability to supervise and direct the operations of a clinical molecular genetics diagnostic laboratory, including technical experience and knowledge in quality control and quality assessment procedures;
- the ability to perform a variety of molecular diagnostic assays;
- an understanding of the heterogeneity, variability, and natural history of molecular genetic disorders;
- a broad knowledge of evolving technology including CGH microarray and next generation DNA sequencing, and their application to genomic medicine, (1) basic molecular biology and genetics, (2) the application of molecular genetic techniques to the diagnosis of genetic diseases, and (3) the etiology, pathogenesis, clinical manifestations, and management of human genetic disorders;
- diagnostic and interpretive skills in a wide range of clinical molecular genetics problems; and
- the ability to communicate molecular diagnostic laboratory results in the capacity of a consultant to medical genetics professionals and other clinicians, and directly to patients in concert with other professional staff.

SUBSPECIALTIES OF GENETICS

Medical Biochemical Genetics encompasses the evaluation, prevention, treatment, and discovery of diseases due to genetic defects in human biochemical processes. This area of genetics is recognized as a subspecialty by the American Board of Medical Specialties (ABMS) [www.abms.org]. Residency training programs in this subspecialty are accredited by the Accreditation Council of Graduate Medical Education (ACGME) [www.acgme.org], are one year in duration, and are available only to licensed physicians who are board-certified by the ABMGG as clinical geneticists. This subspecialty is also available as a two-year training program for licensed physicians certified by other appropriate ABMS member boards. Training in medical biochemical genetics focuses on the diagnosis and treatment of patients with metabolic disorders. Training does not include those skills and knowledge necessary to direct a clinical laboratory. A medical biochemical geneticist deals with the evaluation and treatment of patients with biochemical genetic disorders, defined as inborn errors of metabolism at any age of onset.

Molecular Genetic Pathology focuses on diagnosing and confirming diagnoses of Mendelian genetic disorders, diseases of human development, infectious diseases, and malignancies, as well as assessing the natural history of those disorders. Molecular Genetic Pathology is recognized as a subspecialty by the American Board of Medical Specialties (ABMS) [www.abms.org]. Residency training programs in this subspecialty

geneticists who are board certified by ABMGG or diplomates of the American Board of Pathology. These training programs instruct molecular genetic pathologists in the principles, theory, and technologies of molecular biology and molecular genetics. Upon graduation, molecular genetic pathologists can provide information about gene structure, function, and alteration, as well as apply laboratory techniques for diagnosis, treatment, and prognosis for individuals with related disorders.

***NOTE TO GRADUATES OF NON-U.S., PUERTO RICO, OR CANADIAN ACADEMIC OR MEDICAL PROGRAMS:**

- Individuals with a doctoral degree earned outside of the U.S., Canada or Puerto Rico are strongly **urged** to have their international credentials reviewed by the ABMGG Credentials Committee for equivalency of their doctoral degree **prior** to entering medical genetics and genomics training in an ABMGG-accredited fellowship or ACGME-accredited clinical genetics and genomics residency program. For more information, review the Foreign Medical Graduates Credentials Review page.