Specialties of Genetics

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To be an active candidate for certification by the ABMGG, an individual must meet the criteria in the area of desired certification and provide the required supporting documentation. Certification is offered in Clinical Genetics and Genomics, Clinical Biochemical Genetics, Clinical Cytogenetics and Genomics, and Clinical Molecular Genetics and Genomics.

Clinical Geneticist

A clinical geneticist is an individual who holds a U.S. or Canadian earned or the equivalent of an earned M.D. or D.O. degree, has had 1 year in an ACGME-accredited primary care residency (pediatrics, internal medicine, obstetrics and gynecology, and/or family medicine) and 2 years in an ACGME-accredited residency in clinical genetics categorical residency or completion of a 4-year combined residency training program (Pediatrics/Genetics, Internal Medicine/Genetics or Maternal-Fetal Medicine/Genetics), a valid and unrestricted medical license, and demonstrates competence to provide comprehensive genetic diagnostic, management, therapeutic, and counseling services.

These requirements imply that the individual possesses:

- broad knowledge in human and medical genetics, 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 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 Geneticist

A clinical biochemical geneticist is an individual with a U.S. or Canadian earned or the equivalent of an earned doctoral degree (M.D., D.O., Ph.D.) who can correctly perform 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;

- 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 Geneticist/Genomicist (check out the Laboratory Genetics and Genomics FAQs)

A laboratory geneticist is an individual with a U.S. or Canadian earned doctoral degree (M.D., D.O., Ph.D.), or equivalent, who is certified by the ABMGG and 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 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 Cytogeneticist

A clinical cytogeneticist is an individual with a U.S. or Canadian earned or the equivalent of an earned doctoral degree (M.D., D.O., Ph.D.) who can correctly perform and interpret cytogenetic analyses relevant to the diagnosis and management of human genetic diseases, and who acts as a consultant regarding laboratory diagnosis for a broad range of cytogenetic disorders, including inherited 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 Geneticist

A clinical molecular geneticist is an individual with a U.S. or Canadian earned or the equivalent of an earned doctoral degree (M.D., D.O., Ph.D.) 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 generaltion 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.