ACMG and G2MC

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Plan

• Revisit ISCC Framework for Competencies in Genomic Medicine
• American College of Medical Genetics and Genomics (ACMG)
• Global Genomic Medicine Collaborative (G2MC)
• Genomic Medicine at UAB
ISCC Competencies in Genomic Medicine

- Elicit, document, and act on relevant family history pertinent to patient’s clinical status
- Utilize genomic testing appropriately to guide patient management
- Utilize genomic information to inform treatment decisions
- Support the use of genomic information to guide the diagnosis and management of cancer and other disorders involving somatic genetic changes
- Utilize genomic tests that identify microbial contributors to human health and disease, as well as genomic tests that guide therapy of infectious diseases

Genomic Testing

EPA: Utilize genomic testing appropriately to guide patient management

Patient Care
- Discuss the indications for genomic testing – specifically the benefits, risks, and alternatives
- Explain the implications of placing genomic test results in the patient’s medical record
- Discuss the possibility of incidental findings and how they will be handled
- Discuss risks of having genomic testing done: e.g., psychological to the individual as well as family; the potential for discrimination; potential effect on insurance coverage, etc.
- Explain to a patient issues of costs and financial coverage of genomic testing
- Order, interpret, and communicate the results of appropriate genomic tests, within the physician’s scope of practice
- Provide referral to an appropriate specialist for genomic testing of a condition outside the physician’s scope of practice
- Respond to the results of an abnormal genetic screening test, such as newborn screening, including immediate management and appropriate referral

Knowledge for Practice
- Describe the major forms of genomic variability
- Explain how different genomic changes may result in different phenotypes
- Recognize that genomic tests require interpretation with respect to the patient’s clinical status (e.g., pathogenic likely pathogenic, benign, etc.)
- Explain the concepts of analytic validity, clinical validity, clinical utility as they relate to genomic testing
- Recognize that medically “non-actionable” genomic results can be useful to the patient and family (i.e., personal utility)

Practice-Based Learning and Improvement
- Incorporate genomic findings into the health record and patient care plan
- Have a method for periodic review of ‘new’ genomic interpretation for clinical applications.

Interpersonal and Communication Skills
- Ensure that undergoing genomic testing is a joint decision of the patient and the physician
- Explain and document findings from genomic testing to patient, including implications for other family members
- Facilitate access to resources to enhance patient learning about the results of genomic testing
- Address the needs of the patient as an individual as well as the needs of family members
Framework for development of physician competencies in genomic medicine: report of the Competencies Working Group of the Inter-Society Coordinating Committee for Physician Education in Genomics

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The practice of genomics to maintain health, facilitate diagnosis, and care or mitigate disease is dependent on the skillful translation of genomic science into meaningful action at the bedside and in the clinic. A survey of both primary-care and specialist physicians found that 90% of primary-care physicians, 80% of specialists, and 70% of pediatric practitioners are increasing their routine diagnosis and treatment, and this trend is likely to expand to other areas of medical practice in the coming years.

Nearly half of practicing physicians in the United States are more than 50 years of age, medical school and residency training for these physicians occurred before the completion of the Human Genome Project and the breakthrough advances in genomic medicine. Current trainees are faced with a rise of progress in genomics that renders much of what they have learned out of date by the time they enter practice. Considering the rapid rate of change, substantial reductions in the cost of genome sequencing, and the increasing relevance of genomic information to the practice of medicine, the barrier to implementing genomic discoveries within medical practice have to be overcome. Moreover, release of genomics by untrained health-care providers may occur without advantage and may result in harm to patients based on inaccurate diagnosis or use of unnecessary or incorrect tests.

The National Human Genome Research Institute, together with 23 professional societies and other organizations interested in physician education, developed the Inter-Society Coordinating Committee for Physician Education in Genomics (ISCC) in the spring of 2010 (see Supplementary Data Si online). ISCC member organizations focus on physician training, starting with medical school matriculation and continuing through residency and fellowship training. The ISCC seeks to improve genomic literacy of physicians and other practitioners and to enhance the practice of genomic medicine through sharing of educational approaches and joint identification of educational needs. The ISCC developed four working groups: Genomic Medicine Competencies, Educational Programs, Use Cases, and Specialty Boards (see Supplementary Data Si online).

The Genomic Medicine Competencies Working Group was charged with the development of a framework whereby
American College of Medical Genetics and Genomics

• Type of Organization: Professional Society

• Membership:
  • Physician medical geneticists
  • Laboratory geneticists
  • Genetic counselors

• Vision: improving health through the integration of genetics and genomics across all of medicine.

• Mission:
  • Clinical and Laboratory Practice
  • Education
  • Advocacy
During this Case Conference, the presenters will review case(s) that illustrate the difficulty of clinical diagnosis of EDS-HT and review case(s) that illustrate challenges in the management of EDS-HT. They will also address areas of clinical uncertainty and common misconceptions.

This activity has been approved for AMA PRA Category 1 Credit™

This activity is supported in part by an unrestricted educational grant from QIAGEN Bioinformatics and the Ingenuity Clinical Decision Support Platform.
• Chair – Geoffrey Ginsburg, MD, PhD
• Working Groups
  • IT/Bioinformatics
  • Education/Workforce
  • Pharmacogenomics
  • Evidence
  • Policy
  • Sequencing
3rd Global Genomic Medicine Collaborative Conference
Implementing Genomic Medicine into Practice
April 27-29, 2017, Athens, Greece
G2MC Education/Workforce Working Group

• Co-chairs – Bruce Korf, Vajira Dissanayake (Sri Lanka)

• Areas of focus
  • Training paradigms in genomic medicine around the world
  • Development of Case Studies
  • G2MC Grand Rounds

• Potential for collaboration with ISCC
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Genomic Medicine at UAB – Educational Activities

• T32 in Genomic Medicine
  • Postdoctoral training for MDs, PhDs, MD/PhDs
  • Research
  • Participation in clinic and/or clinical lab
  • Individualized education program
  • Participation in educational outreach
  • Seminars in Bioethics and Genomic Medicine Conference

• Immersion Course – Genetics & Genomics in Clinical Research
  • 5 consecutive half days – lectures and demonstrations
  • Fall – Introductory course; spring – Bioinformatics course

• Undergraduate Major in Genetics and Genomics

• Graduate and Postdoctoral Residency and Fellowship Program

• UAB Program in Precision Medicine