



Global Genomic  
Medicine Collaborative

# ACMG and G2MC

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Knowledge that will change your world

# 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

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

# ISCC Competencies in Genomic Medicine

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- 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

## 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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Completion of the Human Genome Project, in conjunction with dramatic reductions in the cost of DNA sequencing and advances in translational research, is gradually ushering genomic discoveries and technologies into the practice of medicine. The rapid pace of these advances is opening up a gap between the knowledge available about the clinical relevance of genomic information and the ability of clinicians to include such information in their medical practices. This educational gap threatens to be rate limiting to the clinical adoption of genomics in medicine. Solutions will require not only a better understanding of the clinical implications of genetic discoveries but also training in genomics at all levels of professional development,

including for individuals in formal training and others who long ago completed such training. The National Human Genome Research Institute has convened the Inter-Society Coordinating Committee for Physician Education in Genomics (ISCC) to develop and share best practices in the use of genomics in medicine. The ISCC has developed a framework for development of genomics practice competencies that may serve as a starting point for formulation of competencies for physicians in various medical disciplines.

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**Key Words:** competencies; education; genomic medicine; genomics

The promise of genomics to maintain health, facilitate diagnosis, and cure or mitigate disease is dependent on the skillful translation of genomic science into meaningful action at the bedside and in the clinic.<sup>1,2</sup> Surveys of both primary-care and specialist physicians, often by their professional societies, reveal unease, and even unwillingness, to use genomic data.<sup>1,4</sup> The use of genomics in caring for patients with certain cancers and for some pediatric patients is increasing in routine diagnosis and treatment, and this trend is likely to expand to other areas of medical practice in the coming years.<sup>3,6</sup>

Nearly half of practicing clinicians 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.<sup>7</sup> Current trainees are faced with a rate of progress in genomics that renders much of what they have learned out of date by the time they enter practice. Considering this 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 barriers to implementing genomic discoveries within medical practice have

to be overcome. Moreover, misuse of genomics by untrained health-care providers may incur cost 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, 15 other institutes at the National Institutes of Health, and other organizations interested in physician education, developed the Inter-Society Coordinating Committee for Physician Education in Genomics (ISCC) in the spring of 2013 (see **Supplementary Data S1** online). ISCC member organizations focus on physician training, starting with medical school matriculation and continuing through residency and fellowship, for active clinicians.<sup>9</sup> 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 Products, Use Cases, and Specialty Boards (see **Supplementary Data S1** online).

The Genomic Medicine Competencies Working Group was charged with the development of a framework whereby

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# American College of Medical Genetics and Genomics

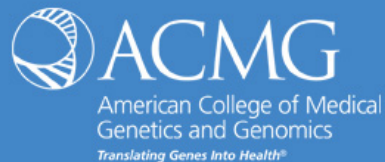
- Type of Organization: Professional Society
- Membership:
  - Physician medical geneticists
  - Laboratory geneticists
  - Genetic counselors
- Vision: improving health through the integration of genetics and genomics across all of medicine.
- Mission:
  - Clinical and Laboratory Practice
  - Education
  - Advocacy



# 2017 Genetics and Genomics Review Course

Center for Advanced Medical Learning and Simulation (CAMLS)  
Tampa, Florida

May 4-7, 2017



March 21-25 | Exhibit Dates  
March 22-24  
Phoenix Convention Center  
Phoenix, AZ

# 2017

## ACMG Annual Clinical Genetics Meeting

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Session Title	Event Type	Room	Day	Time	CE Units	CEU Type
<a href="#">NAMA at the ACMG 2.0</a>	Short Course		Tue., Mar. 21	12:30 PM - 5:30 PM	4.5	CME,NSGC,P.A.C.E.
<a href="#">Variant Interpretation from the Clinician's Perspective</a>	Short Course		Tue., Mar. 21	12:30 PM - 5:30 PM	4.5	CME,NSGC,P.A.C.E.

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### Mark Your Calendar

Ehlers-Danlos Syndrome, Hypermobility Type, in Children: Differential Diagnosis and Recommendations for Management

Hosted by the ACMG Professional Practice and Guidelines Committee

**Wednesday, January 18, 2017**  
**2:00 pm – 3:00 pm EST**

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.*

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American College of Medical Genetics Foundation

## Summer Genetics Scholars Program

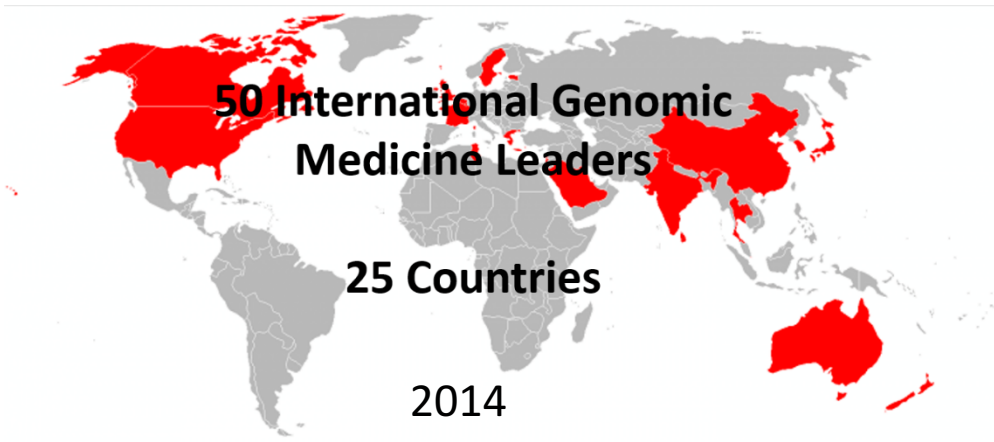


Better Health Through Genetics®



## Global Genomic Medicine Collaborative

- 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

**Grand Rounds**  
 4 May 2016 - GMT 04:00 am, New Delhi 8:30 am, Manila 11:00 am  
 3 May 2016 - EST 11:50 pm, CST 10:50 pm

University of Alabama in Birmingham (UAB)  
 Undiagnosed Disease Programme









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Visit: [bit.ly/aehinhour](http://bit.ly/aehinhour) to register

## G2MC Grand Rounds

Global Genomic Medicine Collaborative Grand Rounds • 8 videos • 91 views • Last updated on Dec 8, 2016

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1		<b>G2MC Grand Rounds - Session 01</b> by Global Genomic Medicine Collaborative Grand Rounds	50:38
2		<b>G2MC Grand Rounds - Session 02</b> by Global Genomic Medicine Collaborative Grand Rounds	1:03:45
3		<b>G2MC Grand Rounds - Session 03</b> by Global Genomic Medicine Collaborative Grand Rounds	54:26
4		<b>G2MC Grand Rounds - Session 04</b> by Global Genomic Medicine Collaborative Grand Rounds	1:02:12
5		<b>G2MC Grand Rounds - Session 05</b> by Global Genomic Medicine Collaborative Grand Rounds	46:24
6		<b>G2MC Grand Rounds - Session 06</b> by Global Genomic Medicine Collaborative Grand Rounds	53:35
7		<b>G2MC Grand Rounds - Session 07</b> by Global Genomic Medicine Collaborative Grand Rounds	56:35
8		<b>G2MC Grand Rounds - Session 08</b> by Global Genomic Medicine Collaborative Grand Rounds	53:32

# G2MC Grand Rounds

# 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