



National Human
Genome Research
Institute



National
Institutes of
Health



U.S. Department
of Health and
Human Services

State of the Committee: ISCC Work Products and Plans

U.S. Department of Health and Human Services
National Institutes of Health
National Human Genome Research Institute

Teri Manolio, M.D., Ph.D.
Inter-Society Coordinating Committee on Practitioner
Education in Genomics

April 23, 2014

GM IV: Physician Education in Genomics

Jan 28-29, 2013

Genomic Medicine Centers Meeting IV: *Physician Education in Genomics*



On January 28-29, 2013, the National Human Genome Research Institute (NHGRI) sponsored its fourth Genomic Medicine Centers Meeting IV: *Physician Education in Genomics* - at the Howard Hughes Medical Institute (HHMI) campus in Chevy Chase, MD. The meeting was co-chaired by Dr. Francis S. Collins, M.D., Ph.D., NHGRI and Marc Williams, M.D., HHMI.

The goals of the meeting were to:

- Identify ongoing efforts and current needs
- Understand processes for guideline development
- Promote collaborations among the societies.
- Learn about ongoing genomic medicine initiatives at centers nationwide and within NIH.

[Genomic Medicine 4 Executive Summary](#) PDF

[Genomic Medicine 4 Full Meeting Minutes](#) PDF

Genomic Medicine IV, Jan 28-29, 2013

Accreditation Council for Graduate Medical Education
Accreditation Council for Continuing Medical Education
American Academy of Pediatrics
American College of Cardiology
American College of Medical Genetics and Genomics
American College of Physicians
American Congress of Obstetrics
American Heart Association
American Society of Clinical Oncology
Association of Professors of Human Medical Genetics

Group Activities to Date

- June 26, 2013 Plenary and WG Webinars
- Sept 19-20, 2013 Meeting
- Sustainability exploration commissioned
- Four working groups established and working

Inter-Society Coordinating Committee White Paper, Feb 2014

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COMMENTARY

Genetics
inMedicine

The growing role of professional societies in educating clinicians in genomics

Teri A. Manolio, MD, PhD¹ and Michael F. Murray, MD²; for the Inter-Society Coordinating Committee
for Practitioner Education in Genomics

Recent identification of numerous genetic variants influencing metabolism of commonly used drugs, responses to chemotherapy, and risk of familial and sporadic diseases, coupled with improved technologies for detecting these variants at continually declining costs, have opened exciting prospects for improving clinical care through genomic applications.¹ Lack of education in genomics among physicians and allied health personnel not only delays appropriate clinical application of these promising approaches but also could lead to erroneous uses with serious consequences.² To address these challenges and keep pace with expanding applications of genomics in fields such as drug selection, cancer treatment, high-risk screening, and undiagnosed dis-

advances on current clinical practices—more of an evolution than a revolution. Assessing a pharmacogenetic variant along with creatinine clearance in drug dosing, for example, or using tumor variants as well as histology in predicting treatment response, can be viewed as expansions of the laboratory-based armamentarium already available to clinicians. In contrast, direct-to-consumer marketing of genomic tests raises quite different challenges, including the daunting proposition of a physician being handed a patient-initiated genome-wide scan and expected to provide an expert interpretation.⁶

Surveys of physicians to date have indicated that many feel unprepared to order and interpret genomic tests and wish to

Entrustable Professional Activities (EPAs)

In press, *Genet Med*

PROPOSED ENTRUSTABLE PROFESSIONAL ACTIVITIES (EPAs) IN GENOMIC MEDICINE FOR GENERALIST PHYSICIANS

INTER-SOCIETY COORDINATING COMMITTEE, COMPETENCIES WORKGROUP

Family History

EPA: Elicit, document, and act on relevant family history pertinent to the patient's clinical status

Patient Care

- Conduct patient interview to assemble family history
- Utilize standard pedigree symbols in assembling family history
- Recognize patterns of Mendelian inheritance and calculate simple Mendelian risks; provide this information to patients and family members as appropriate.
- Use empirical risk figures to provide appropriate information for complex (multifactorial) medical conditions.
- Recognize that traits may cluster in families due to multifactorial rather than Mendelian patterns of inheritance
- Formulate an action plan to address relevant family history information

Educational Products Collected and Available through G2C2!



GENETICS/GENOMICS COMPETENCY CENTER
FOR EDUCATION



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Physician

Find professionally curated resources, classroom materials, and real-world examples every Physician should know.

Change Disciplines



Genetic
Counselor



Nurse



Physician
Assistant



Pharmacist

1

Select a Curricular Area below

- Learning Activities & Resources

Exercises designed to develop professional ability and resources that facilitate competency.

- Competencies & Core Knowledge

A high level educational goal for learners. Specific areas of knowledge learners need to achieve a competency.

- Performance Indicators

A measurable knowledge, skill or ability of a professional that demonstrates competency.

- Assessments

Exercises designed to measure the outcome of a Learning Activity.

Specialty Boards Engaged –Ophthalmology

Pediatric Ophthalmology

- 7.10.1 Congenital Disorders
 - 7.10.1.1 Congenital Ptosis
 - 7.10.1.2 Congenital Ectropion
 - 7.10.1.3 Congenital Entropion
- 7.3 Genetics
 - 7.3.1 Basic Concepts
 - 7.3.1.1 Inheritance
 - 7.3.1.1.1 AD inheritance
 - 7.3.1.1.2 AR inheritance
 - 7.3.1.1.3 X-linked inheritance
 - 7.3.1.2 Non Mendelian inheritance
 - 7.3.1.3 Genetic Counseling
 - 7.3.1.4 Gene Therapy
 - 7.4 Congenital Genetic Disorders
 - 7.4.1 Congenital Disorders
 - 7.4.1.7 Diagnose congenital and genetic disorders
 - 7.4.1.8 Develop patient care plans
 - 7.4.1.9 Identify need for genetic counseling

Retina and Vitreous

- 9.2.7 Genetic Testing
 - 9.2.7.1 Disorders that can be tested
 - 9.2.7.2 Counseling requirements
 - 9.2.7.3 Recommend exam for other family members
- 9.3.11 Metabolic disease affecting the retina
 - 9.3.11.1 Systemic mucopolysaccharidoses
 - 9.3.11.2 Sphingolipidoses
 - 9.3.11.3 Mucopolysaccharidoses
 - 9.3.11.4 Cystinosis
 - 9.3.11.5 Diagnosis of metabolic disease affecting the retina
- 9.3.12 Albinism
 - 9.3.12.1 Oculocutaneous Albinism
 - 9.3.12.2 Ocular albinism
 - 9.3.12.3 Albinoidism
 - 9.3.12.6 Diagnose type of albinism
- 9.3.3.1 Age-related macular degen.

Specialty Boards Engaged –Ophthalmology

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7.10.1.2 Congenital Ectropion

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7.3 **Genetics**

7.3.1 **Basic Concepts**

7.3.1.1 **Inheritance**

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7.3.1.1.2 **AR inheritance**

7.3.1.1.3 **X-linked inheritance**

7.3.1.2 **Non Mendelian inheritance**

7.3.1.3 **Genetic Counseling**

7.3.1.4 **Gene Therapy**

7.4 Congenital Genetic Disorders

7.4.1 Congenital Disorders

7.4.1.7 Diagnose congenital and genetic disorders

7.4.1.8 **Develop patient care plans**

7.4.1.9 **Identify need for genetic counseling**

Retina and Vitreous

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Use Cases Template, Example, Key Topics

Use Case Template for ISCC Use Case Workgroup

Text in **bold** reflects mapping of the use case template to the entrustable professional activities developed by the Competency Workgroup. Using the use cases by definition would meet **competency of Personal and professional development**. Applying information from the use case into a clinician's practice would meet the **practice based-learning and improvement competency**.

- I. Specialty/Professional Society
- II. Type of Use Cases
 - a. Genomic-based therapeutics.
 - i. Pharmacogenomic
 - b. Rare Single Gene Mendelian Disorder
 - c. Family History
 - d. Common Complex Disease with Genetic Component
 - e. Whole exome/genome sequencing
 - i. Incidental findings (**specifically included in EPA for genomic testing**)
 - f. Microbial Genomics (**included as part of the EPA**) Probably few specialties at the present time would create such a use case (Hepatology chronic hepatitis C)

ISCC Over-Arching Goals, 6/23/2013

- Gather and facilitate dissemination of best practices and resources in genomic education and clinical care.
 - Next agenda item: Draft G2C2 physician website!
- Identify advances in genomic science that will require new educational initiatives.
 - *Hmm, haven't started...*
- Identify needs of societies and clinicians in filling gaps in evidence and knowledge and providing effective educational efforts.
 - *Hmm...*
- Identify foundational educational needs common across professions and specialties.
 - Yes! Entrustable Professional Activities (EPAs)

ISCC Over-Arching Goals, 6/23/2013

- Seek the optimal educational balance between competencies and basic knowledge.
 - *Hmm...*
- Design short-, medium-, and long-term work plans with initial focus on producing tangible outcomes within the first year.
 - Tangible outcomes: EPAs, G2C2, Use Case template and example, two high-pri specialty boards engaged
- Assist societies in jointly and separately publishing papers of common interest.
 - White paper, EPAs in *Genetics in Medicine*
 - Societies separately? *Hmm...*

ISCC Process Metrics, 6/23/2013

- Educational best practices identified and disseminated
 - *Hmm...*
- Physician competencies are generated
 - YES!
- ...and estimates of use gathered
 - *Not yet, potential next step*
- Professional society guidelines and other guidance documents are reviewed and improved
 - Potential interest from ophthalmology and family medicine

ISCC Process Metrics, 6/23/2013

- Society-specific use cases are identified and educational materials developed to address them.
 - *Hmm...*
- The number and diversity of participating professional societies grows.
 - YES!

ISCC as of March 27, 2014

Accred Counc Grad Med Ed
Accred Council Cont Med Ed
Am Acad Family Physicians
Am Acad Ophthalmology
Am Acad Pediatrics
Am Assoc Clin Chem
Am Board Family Medicine
Am Board Medical Genetics
Am Board Medical Specialties
Am Board Ophthalmology
Am Coll Cardiology
Am Coll Med Genet Genom
Am Coll Physicians
Am Congress Obstet Gynecol
Am Dental Assoc

Am Dental Education Assoc
Am Heart Assoc
Am Med Assoc
Am Soc Clin Oncol
Am Soc Human Genet
Am Thoracic Soc
Assoc Molec Pathology
Assoc Prof Human Med Genet
Ctrs Medicare Medicaid (CMS)
Coll Am Pathologists
Counc Med Specialty Soc
Hlth Res Serv Admin (HRSA)
Int Assoc Dental Res
Int Soc Psychiatric Genetics
Soc Gen Internal Medicine

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Am Board Medical Specialties

Am Board Ophthalmology

Am Coll Cardiology

Am Coll Med Genet Genom

Am Coll Physicians

Am Congress Obstet Gynecol

Am Dental Assoc

Am Dental Education Assoc

Am Heart Assoc

Am Med Assoc

Am Soc Clin Oncol

Am Soc Human Genet

Am Thoracic Soc

Assoc Molec Pathology

Assoc Prof Human Med Genet

Ctrs Medicare Medicaid (CMS)

Coll Am Pathologists

Counc Med Specialty Soc

Hlth Res Serv Admin (HRSA)

Int Assoc Dental Res

Int Soc Psychiatric Genetics

Soc Gen Internal Medicine

ISCC Process Metrics, 6/23/2013

- Society-specific use cases are identified and educational materials developed to address them.
 - *Hmm...*
- The number and diversity of participating professional societies grows.
 - YES!
- Interactions with other efforts such as NCHPEG and other practitioner communities' educational activities are suitably frequent and productive
 - Growing
- Interactions on educational activities among societies within the ISCC are also suitably frequent and productive
 - *Hmm...*

ISCC Substance Metrics, 6/23/2013

Timing?

- Educational products positively assessed and increasingly used by practicing physicians.
- Surveys reveal improved knowledge and comfort in using genomics in their practices.
- Useful papers are published.
- Genomic medicine content is incorporated into certification examinations.
- ACGME and ACCME report improved educational efforts as measured by improved practice of genomic based medicine.

Redesigning NHGRI Training Programs for in Genomic Medicine

Funding Opportunity Title

Ruth L. Kirschstein National Research Service Award (NRSA) Institutional Research Training Grant (Parent T32)

Notice of NHGRI Participation in PA-14-015 "Ruth L. Kirschstein National Research Service Award (NRSA) Institutional Research Training Grants (Parent T32)" and Restructuring of NHGRI Training Programs Focused on Genomic Sciences

Notice of NHGRI Participation in PA-14-015 "Ruth L. Kirschstein National Research Service Award (NRSA) Institutional Research Training Grants (Parent T32)" and Creation of New NHGRI Postdoctoral Training Programs in Genomic Medicine Research



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Funding Opportunities

E-mail List

Ethical, Legal and Social Implications (ELSI)

Minority and Special Populations

Research

Training and Career Development

NHGRI Funding Opportunities: Training and Career Development

- ↓ [Pre- and Post-Doctoral Training Opportunities](#)
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- ↓ [Expired Announcements Whose Topics Remain of Interest](#)
- ↓ [Program Staff](#)

NHGRI Genomic Medicine Training Programs

- Objective: prepare new generation of leaders in genomic medicine
 - Deep and broad training and experiences
 - Open to MD, PhD, or equivalent
- Institutional programs (T32) will support two career paths:
 - Genomic medicine focus for basic research
 - Genomic medicine focus for clinical research (not for clinical care of patients)
- Individual Mentored Clinical Scientist Career Award in Genomic Medicine (K08)

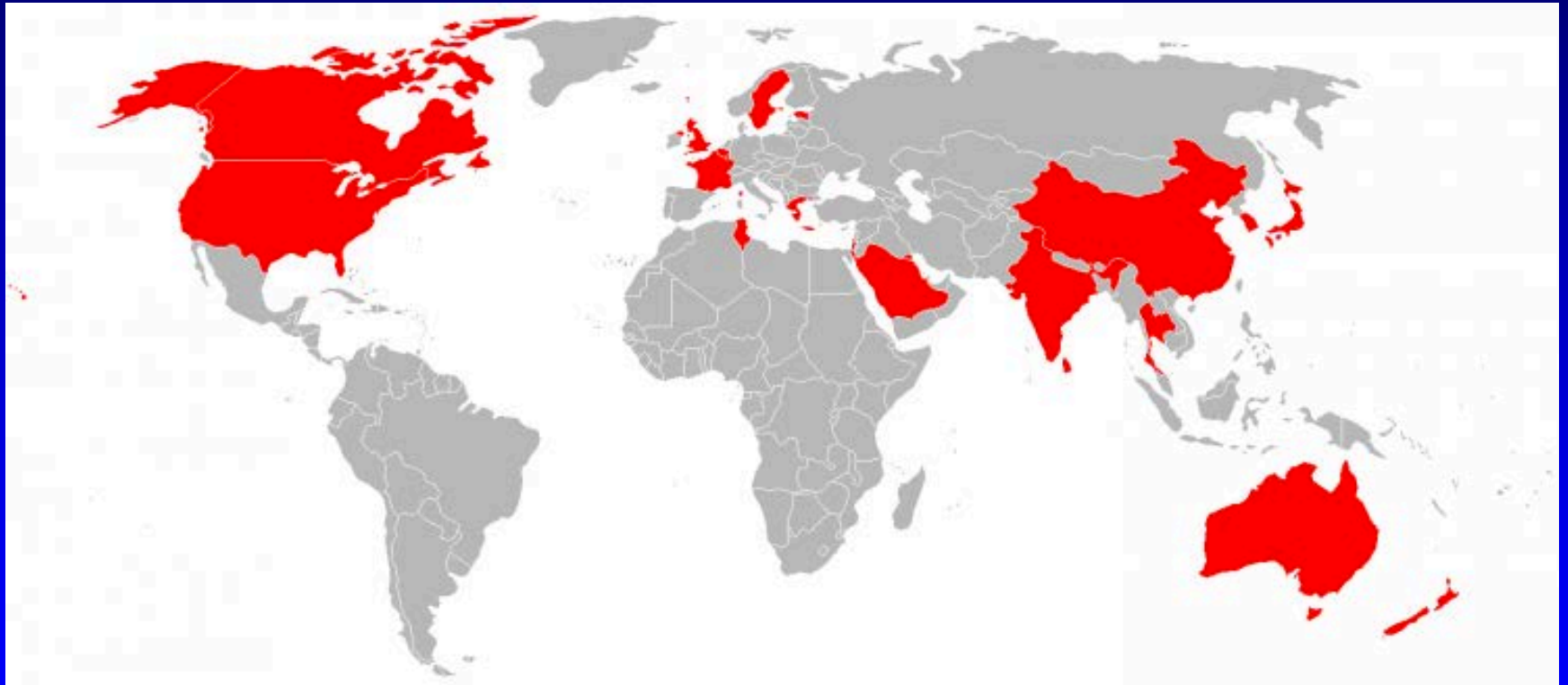
Genomics Training for Non-Geneticist Practitioners

- Am Board Medical Specialties (ABMS)
- Am Coll Med Genet Genom (ACMG)
- Am Soc Human Genetics (ASHG)
- Assoc Prof Human Med Genetics (APHMG)
- Coll Am Pathologists (CAP)
- Jackson Labs/post-NCHPEG



National Academy of Sciences Bldg
2101 Constitution Avenue, NW
Washington, DC

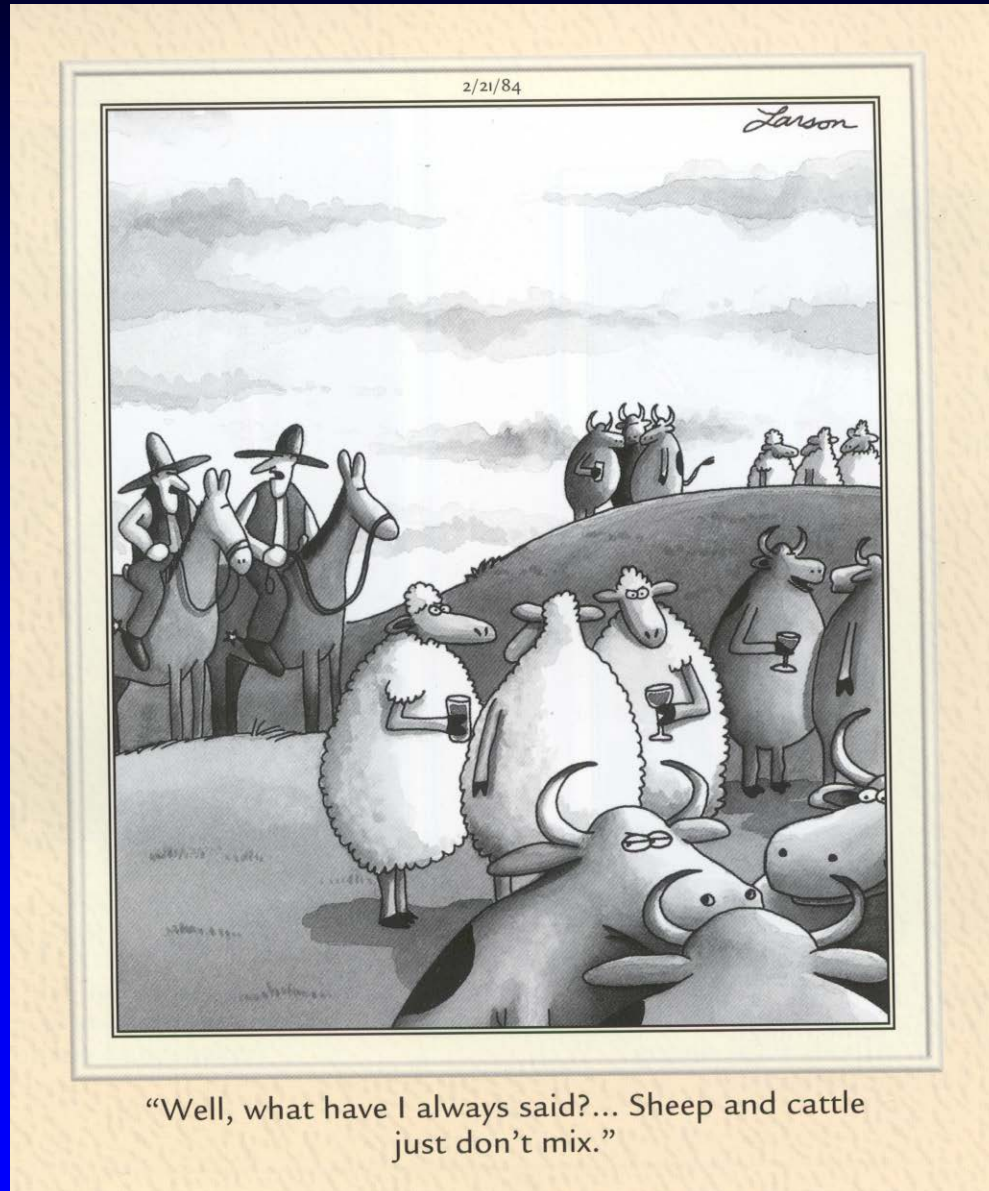
50 International Genomic Medicine Leaders 25 Countries



Courtesy, G Ginsburg, Duke U

Whereto, ISCC?

- More active involvement of societies
- Better engagement of societies' dedicated educational components
- New sources of administrative and fiscal support
- Expand to (and learn from) Canada?
- Expand to (and learn from) the world?



Larson, G. *The Complete Far Side*. 2003.