

ISCC Case Studies Working Group

Co-chairs

Reed Pyeritz, MD, PhD
University of Pennsylvania



Wendy Rubinstein, MD, PhD
NIH/NLM/NCBI



Membership

Louis Cantor

Amy Cronister

Alix D'Angelo

Katrina Gwinn

Emily Harris

Thomas Hart

Richard Haspel

Becki Hulinsky

Thomas Lehner

Flora Lum

Frank Ong

George Papanicolaou

Carsten Rosenow

Robert Saul

Surya Singh

Myra Wick

Bob Wildin

Richard Zorab



ISCC Case Studies Working Group

Charge

Develop educational materials based on scenarios that health care professionals are likely to encounter in practice

Brief history

Under Marc Williams' leadership developed general case study framework, translated to competencies, and created two products

<http://www.genome.gov/27560578>

- Aminoglycoside-induced deafness related to mitochondrial-inherited genetic variation
- Utilizing family history to identify Lynch syndrome



Challenges

Engagement!

- Identifying cases studies to develop
- Sustaining the enterprise with writers who have domain knowledge
- Developing a cogent plan for disseminating materials

Current activities

Perform a needs assessment for professional genetics education among ISCC member organizations

- Quickly answer whether there is sufficient engagement to continue to develop case studies
- Identify perceived educational gaps, for this WG and ISCC overall
- Survey tool developed

Thanks for your help developing the survey!

Amy Cronister

Kathy Calzone

Alix D'Angelo

Becki Hulinsky

Ann Karty

Jean Jenkins

Karen Novak

Vicky Pratt

Steven Singer

Bob Wildin



'The Ask'

Your participation in a needs assessment for professional genetics education

Distribute survey

Member organizations

→ to your education committee and/or your membership

Accreditation and

→ to leaders



WE WANT YOU!

Survey Questions



Approx. 15 minute completion time

8 questions

2 elicit case studies for development by workgroup (shown on next slide)

- 1 with categories to check and examples to support understanding
- 1 open-ended response

2 elicit opinions about educational needs, formats

1 invitation to help create materials

1 offers assistance in developing relationships with regional genetics professionals

1 for open-ended comments

1 to categorize respondents by professional affiliation

→ 2. Do you need additional knowledge in any of the following categories?

Pharmacogenetic testing, for example:

- Genetic susceptibility to drug-induced Stevens-Johnson syndrome
- Genetically influenced response to drugs such as antidepressants, pain medications, or cardiac medications

Mendelian disorders, for example:

- Diagnosis and management of Marfan syndrome, cystic fibrosis, Ehlers-Danlos syndrome, Charcot-Marie-Tooth disease, inborn errors of metabolism

Strong family history of disease, for example:

- Early-onset breast cancer, colorectal cancer, heart disease, or stroke

Common complex disease with a genetic component, for example:

- Intellectual disability, autism, dementia, ataxia, neuropathy, epilepsy, hearing or visual loss, dental anomalies
- Cardiomyopathy, arrhythmia, aneurysms, sudden death
- Diabetes, bleeding disorders, autoimmune disease, nephrotic syndrome, infertility, immunodeficiency, congenital anomalies

Either providing results of whole exome sequencing / whole genome sequencing or a question of whether to order one of these tests, for example:

- Non-invasive prenatal testing (NIPT) to screen for chromosomal disorders
- Evaluation of undiagnosed disease with a suspected genetic component
- Genomic testing for asymptomatic, preventable disease

→ 3. Are there clinical genetics topics you have encountered in practice that would make good case studies?
Please describe any case studies you think would be useful.

Open-ended response



Reed and Wendy will contact you

DRAFT QUESTIONNAIRE – ISCC Case Studies Work Group – version.052015

The purpose of this brief, voluntary, 15 minute questionnaire is to learn about the needs of your organization for improving genomic literacy of your membership. Your participation is optional.

The Inter-Society Coordinating Committee for Practitioner Education in Genomics ([ISCC](#)) is a partnership between the National Institutes of Health and medical professional societies to identify educational needs and support educational initiatives for genomic medicine. The ISCC Case Studies Working Group develops educational materials based on scenarios that health care professionals are likely to encounter in practice. For example, the Working Group has developed [two learning scenarios](#) (<http://www.genome.gov/27560578>) – one about aminoglycoside-induced deafness related to mitochondrial-inherited genetic variation, and another about utilizing family history to identify Lynch syndrome which predisposes to colorectal, endometrial and other cancers.

We wish to know if other case studies would be useful for and and/or your specialty practice.

1. What percentage of clinical cases in your practice have a genetic issue that plays a role in the diagnosis and management of your patient and/or their relatives?

- 0% (none)
- 1-5%
- 6-15%
- Over 15%

4. Do you believe that educational activities in genetics are needed for clinicians in your specialty?

Yes

➤ What are your preferred educational formats for learning?

live / didactic

online

written

other [*open-ended response*]

➤ How important is it that educational activities offer continuing medical education (CME) credit?

Usually essential for me to participate

An important factor, but not crucial for my participation

Not an important factor for my participation

No

➤ If you don't believe educational activities in genetics are needed in your specialty, please share feedback regarding why not.

Open-ended response

5. Would you/others in your organization have interest in working with ISCC to help create educational materials based on scenarios that health care professionals are likely to encounter in practice?

Yes

If yes, provide a name of a contact for assistance if possible:

Open-ended response

No, not at this time

6. Would it be helpful for ISCC to assist you/your membership in developing relationships with professionals in your region who are credentialed in genetics?

Genetic counselors, genetic nurses, and medical geneticists may be able to provide educational support, accept patient referrals, and provide information about insurance coverage for genetic testing.

Yes

No

7. Please provide any additional comments.

Open-ended response

8. Please share your professional affiliations so that the ISCC can target our efforts more effectively.

Check one or more:

- Accreditation Council for Continuing Medical Education (ACCME)
- Accreditation Council for Graduate Medical Education (ACGME)
- American Academy of Family Physicians (AAFP)
- American Academy of Ophthalmology (AAO)
- American Academy of Pediatrics (AAP)
- American Association for Clinical Chemistry (AACC)
- American Association for Dental Research (AADR)
- American Board of Family Medicine (ABFM)
- American Board of Medical Specialties (ABMS)
- American Board of Ophthalmology (AAO)
- American College of Cardiology (ACC)