GM8, Panel 8
Clinician education (including trainees); reporting results to clinicians

Bob Wildin, Mary Relling,
Wendy Rubinstein, Julie Johnson

What is the goal of education?

"..clinicians need not become geneticists to make use of genomic advances any more than they need to become radiologists to make use of imaging."

"The genomics community must align its educational priorities with those of the health professional groups it wishes to educate."

"…all clinicians will need informatics support to interpret and act on genomic information relevant to patient care."

"Ensuring that high-quality software tools are available to clinicians will be more important than forcing them to understand the intricacies of how those tools work."

Feero & Green JAMA, September 7, 2011—

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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nt 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 implications for the individual as well as the family, the potential for discrimination, and the potential effect on insurance coverage;
- Explain to the 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; and
- Respond to the results of an abnormal genetic screening test, such as newborn screening, including immediate management and appropriate referral.

Objectives Related to Genomic Medicine Implementation	Focus Programs									Related Programs							
	UDN	NSIGHT	CSER	eMERGE	IGNITE	ClinGen	GM Mtgs	G2MC	CMG	CPIC	GA4GH	GAPH	IOM	ISCC	LSAC	PAGE	PhenX
Improve genomic diagnostic methods	X	X	X						X								
Facilitate research in undiagnosed and/or Mendelian diseases	X		X						X								
Expand scale of genomic data available in newborns		X															
Advance understanding of disorders of newborns		X															
Research ethical/legal/social issues in genome sequencing		X	X										X				
Interpret sequence data in variety of clinical contexts			X	X													
Integrate sequence data into patient care			X	X	X								X				
Incorporate actionable variants into EMR, develop CDS			X	X	X								X				
Educate clinicians and patients on genomics in clinical care			**X**	**X**	**X**	**X**		**X**		**X**			**X**	**X**			**X**
Develop electronic phenotypes				X													X
Identify variants related to complex traits	X		X	X								X			X	X	
Characterize pharmacogenetic variants and use in care				X	X					X							
Assess outcomes of using genomic information in clinical care				X	X							X					
Assess penetrance of potentially actionable variants				X													
Translate implementation outside highly specialized centers					**X**	**X**											
Define and share processes of implementation, sustainability	X		X	X	X		X	X					X				
Share genotype/phenotype info through open databases						X					X						
Standardize clinical annotation and interpretation						**X**											**X**
Improve understanding of variation in diverse populations				X		X										X	
Assess actionability of genes and variants for clinical use						X				X							
Identify, address barriers to genomic medicine implementation				X			X	X									
Promote interaction and collaboration, reduce duplication	X					X	X	X	X		X		X	X			
Serve as clearinghouse, knowledge base for genomic medicine						X		X		X							
Use genomics to enable new drug development												X	X				
Create genomics-enabled learning health care systems				X	X								X				
Develop evidence base for clinical use of novel diagnostics												X					
	UDN	NSIGHT	CSER	eMERGE	IGNITE	ClinGen	GM Mtgs	G2MC	CMG	CPIC	GA4GH	GAPH	IOM	ISCC	LSAC	PAGE	PhenX

Barriers Identified	Focus Programs								Related Programs												
	UDN	NSIGHT	CSER	eMERGE	IGNITE	ClinGen	GM Mtgs	G2MC	CMG	CPIC	GAPH	GTEx	GS-IT	IOM	ISCC	LSAC	MVP	PAGE	PCORNet	PhenX	PGRN
DATA/INFORMATION NEEDS																					
Evidence base for implement'n incl long-term outcomes			X	X			X	X						X							
Common data elements	X													X					X		
Development, validation of phenotypes				X															X		
Specific drug response phenotypes to add to trials																					X
Publicly available genotype/phenotype info						X			X			X									
Framework for classifying/curating actionable variants			X			X															
Unclear penetrance of actionable genes				X																	
Impact of variants in ancestrally diverse populations																		X			
RAPIDLY EVOLVING NATURE OF GENOMIC INFO																					
Evolving molecular testing panels					X																
Changes in evidence and subsequent treatment					X																
Division between discovery and implementation																X					
Harnessing social media and crowdsourcing methods																			X		
CLINICAL IMPLEMENTATION ISSUES																					
High cost of sequencing, data processing								X									X				
Targeted testing vs genome-scale sequencing			X																		
Limited use of standardized EMR terms, ontologies				X		X															
Concise, comprehensive, interoperable lab reports			**X**							**X**											
Integration of genomic data in learning healthcare system			X																		
Turnaround in clinically emergent settings			X																		
Use cases for genomic CDS development										**X**											
Limited usefulness and interoperability of CDS				X	X		X	X													
Rapidly evolving EMRs					X																
Limited transportability of clinical workflows, protocols					X																
Differing education needs across professional levels					**X**										**X**						
Returning incidental findings												X									
	UDN	NSIGHT	CSER	eMERG E	IGNITE	ClinGen	GM	Mtgs G2MC	CMG	CPIC	GAPH	GTEX	GS-IT	IOM	ISCC	LSAC	MVP	PAGE	PCORN	et PhenX	PGRN

Existing efforts at education of clinicians (1)

- CSER: Dissemination of expertise, protocols and lessons learned via high visibility scientific conferences (ACMG, ASHG, NSGC, ASCO, AACR, ASBH) and expert consultations
- eMERGE: WG on consent, Education, Regulation, and Consultation (CERC)
- IGNITE: Education WG
- ISCC

Existing efforts at education of clinicians (2)

- CPIC: create gene/drug clinical guidelines. Publish widely. Solicit and incorporate feedback from the community. Revise, update.

What is the goal of reporting results to clinicians?

- Interpretable directly by primary care MD?
- Interpretable with help from experts?

Existing efforts for reporting results to clinicians (1)

- eMERGE:
  - Develop, implement, and evaluate the process of clinician-patient education return of results
  - MyResults.org: Patient information about genetic results
  - Genomic Clinical Decision Support Artifact Repository (in development, go live June 2015)

Existing efforts for reporting results to clinicians (2)

- IOM Roundtable:
  - Displaying and Integrating Genetic Information Through the EHR (DIGITizE).
  - Mission: To enable standardized genetic information in the EHR with approach that will ensure interoperability and usability of the data in the clinic and for research applications. https://www.iom.edu/DIGITIZE

Existing efforts for reporting results to clinicians (3)

- CPIC
  - Standardize terms and language for reporting pharmacogenetic results and provide example interpretive language

Challenges, gaps for reporting results (1)

- CSER:
  - Develop lab reports for genomic results and integrate into EHRs
  - Optimize interpretation given inherent clinical time constraints, especially in setting of diseases with poor prognosis and short windows for action. Assess and report on common themes across the CSER sites.

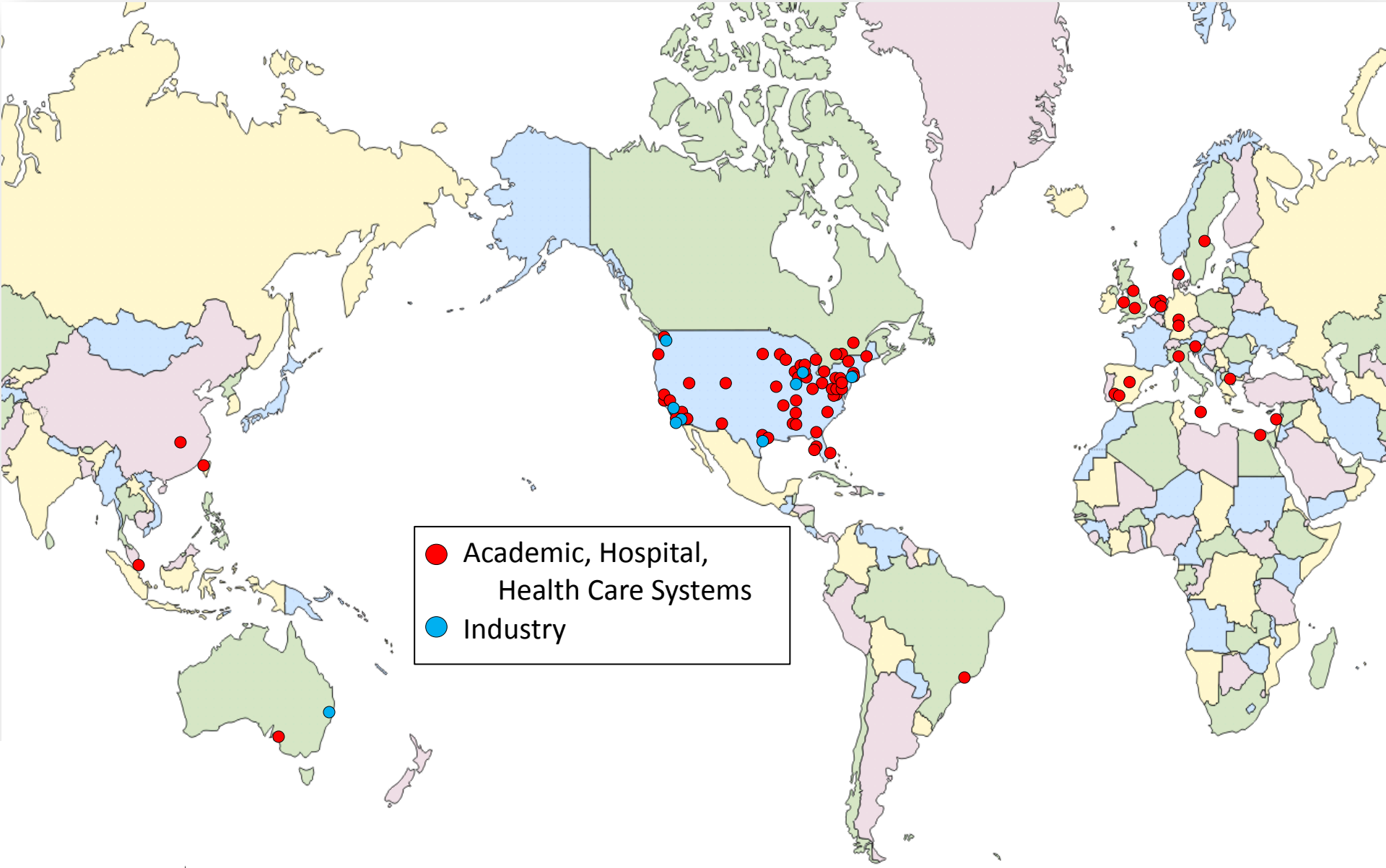
Challenges, gaps for reporting results (2)

- IGNITE:
  - Differing education and training needs for subgroups of professionals involved in clinical implementation; rotating staff. Develop different training modules; continuous education.
- CPIC
  - Limited set of use cases for genomic CDS. CPIC provides vendor-agnostic resources to facilitate implementation of CDS in EHRs

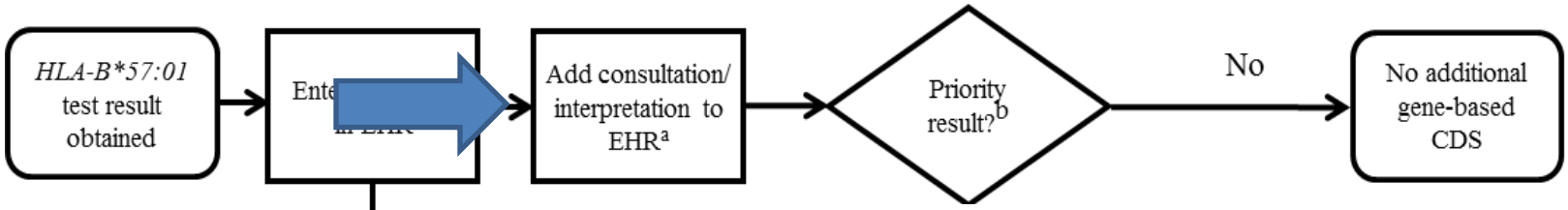
CPIC: Implementing PGx

a PharmGKB & PGRN collaboration

2015 Members



*HLA-B*57:01* Pharmacogenetic Test Result: Clinical Implementation Workflow for EHR



Blue shading indicates

Supplemental Table S7. Example Implementation of this Guideline: Pharmacogenetic Genotype/Phenotype Summary Entries

Test Result for *HLA-B*57:01*[b]	Coded Genotype/Phenotype Summary[c]	EHR Priority Result Notation[d]	Consultation (Interpretation) Text Provided with Test Result[e]
Negative	None	Normal/Low Risk[e]	The *HLA-B*57:01* allele, associated with abacavir hypersensitivity, was not detected in this patient. The patient may be prescribed abacavir. Please refer to the hospital formulary guidelines for specific dosing information. It should be noted that a negative *HLA-B*57:01* result does not absolutely rule out the possibility of some form of abacavir hypersensitivity. Administration of abacavir therapy requires close observation including immediate discontinuation of therapy should any signs or symptoms of hypersensitivity develop.
Positive	*HLA-B*57:01*Carrier	Abnormal/Priority/High Risk[e]	The *HLA-B*57:01* allele, associated with abacavir hypersensitivity, was detected in this patient. *HLA-B*5701* positive patients should **NOT** be prescribed abacavir.

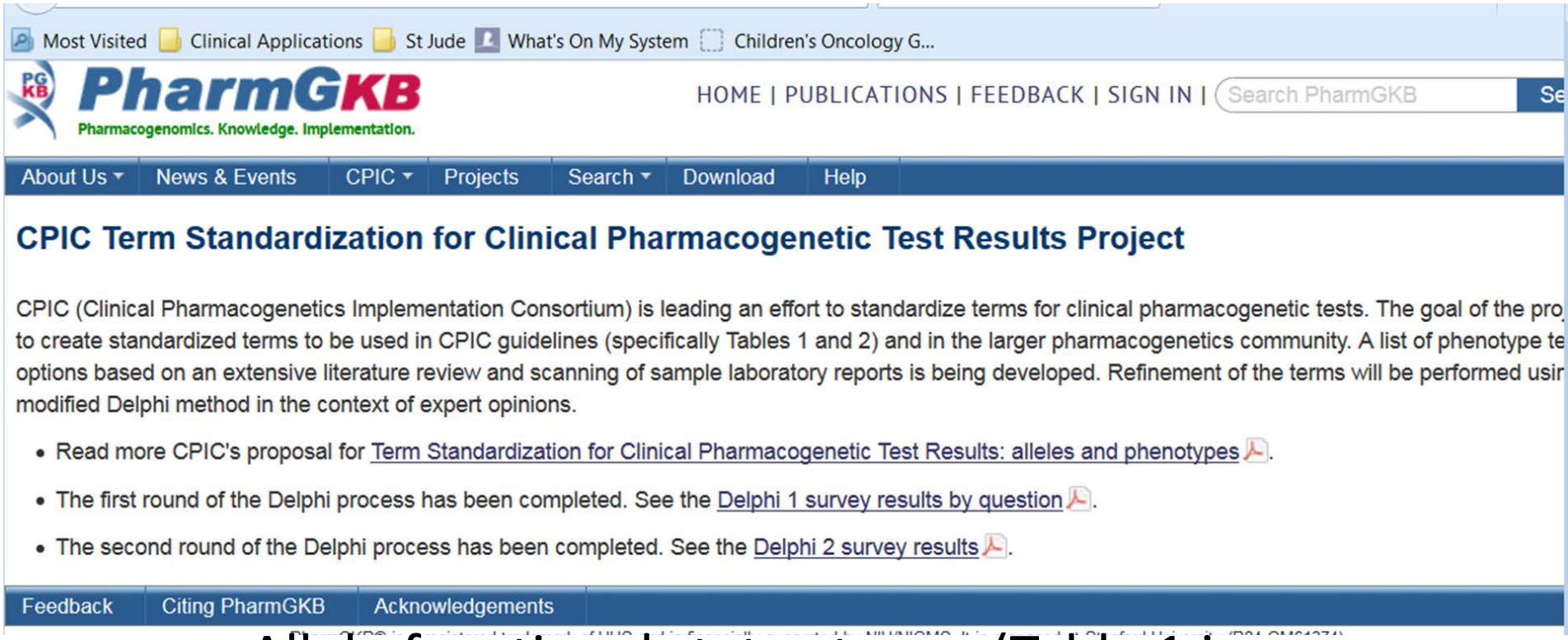
Supplemental Table S12. Example Implementation of this Guideline: Point of Care Clinical Decision Support

Flow Chart Reference Point (See Supplemental Figure S3)	CDS Context, Relative to Genetic Testing	Trigger Condition	CDS Alert Text[a]
1	Pre-Test	No *SLCO1B1* result on file	*SLCO1B1* diplotype may be important for simvastatin side effects. An *SLCO1B1* genotype does not appear to have been ordered for this patient. Use of an alternative statin or dose may be recommended. Please consult a clinical pharmacist[b] for more information.
2	Post-Test	SLCO1B1 - Intermediate Function	Based on the genotype result, this patient is predicted to have intermediate SLCO1B1 function and may be at increased risk for developing simvastatin-associated myopathy. Consider starting with a lower dose of simvastatin (20 mg/day for adults) or choosing an alternate statin agent. Monitor creatine kinase levels routinely. Please consult a clinical pharmacist[b] for more information.
2	Post-Test	SLCO1B1 – Low Function	Based on the genotype result, this patient is predicted to have low SLCO1B1 function and may be at high risk for developing simvastatin-associated myopathy. Consider starting with a lower dose of simvastatin (20 mg/day for adults) or choosing an alternate statin agent. Monitor creatine kinase levels routinely. Please consult a clinical pharmacist[b] for more information.

[a]The specific wording of the alert text may differ among sites.

[b]Pharmacist, pharmacologist, or a clinician with pharmacogenetic expertise/training.

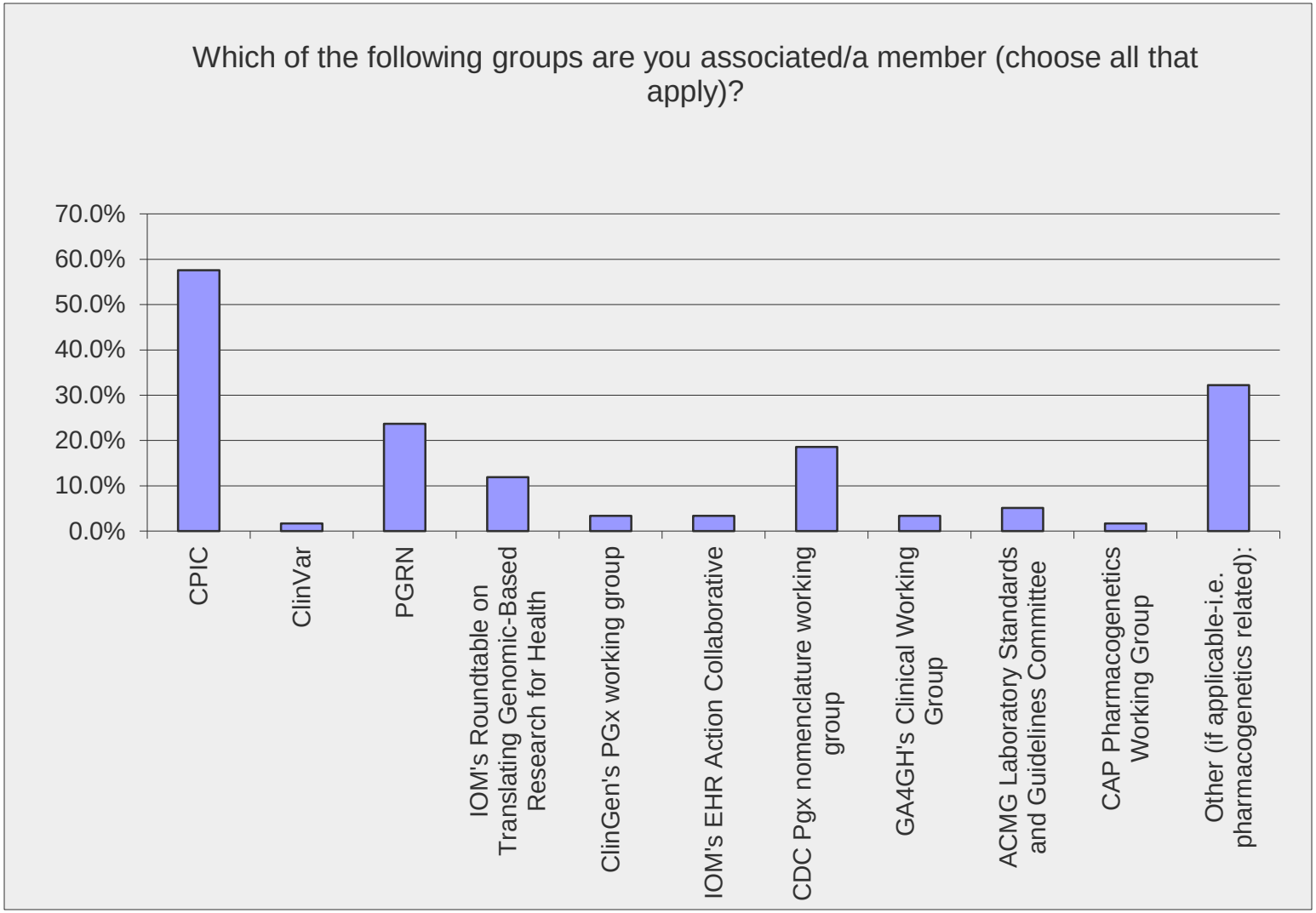
Clinical Pharmacology & Therapeutics (2014); 96 4, 423–428



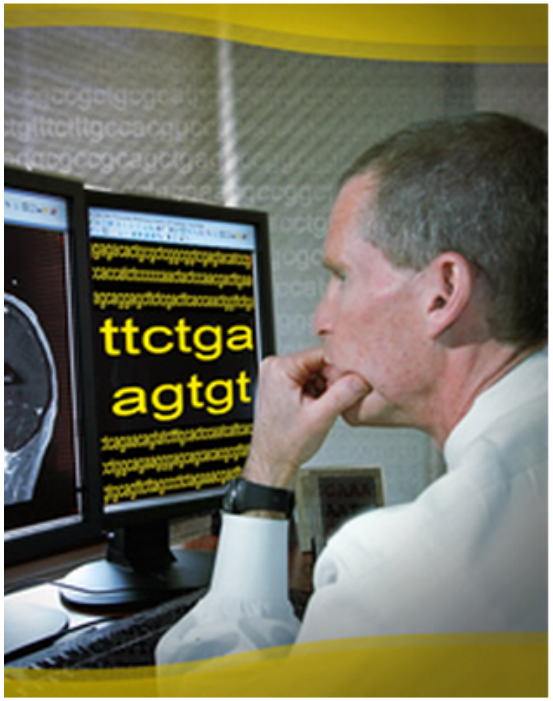
- Allele functional status terms (Table 1 in guideline)
  - E.g. Low, absent, high, intermediate
- Phenotype (i.e. diplotype, Table 2 in guideline)
  - UM, EM, IM, PM

Modified Delphi Process

- **Assessment**
  - Define terms that need to be evaluated and standardized.
- **Development**
  - Create a list of options for terms (literature review and survey to genetic testing labs)
- **Prioritization**
  - Delphi 1: Experts will specify their level of agreement or disagreement on a symmetric agree-disagree scale (1-4) for each set of gene terms. Experts can also list additional terms.
- **Refinement:**
  - Delphi 2: For each gene, retain terms in which 70% of the experts agreed or strongly agreed in Delphi 1*.
  - Experts will pick 1 set of terms per gene/gene group.
  - Results from prior survey will be made available to the experts.
- **Consensus**
  - Delphi 3-?: For each gene/gene group, retain top terms selected by experts.
  - Repeat process until 70% consensus for one set of terms/gene is achieved. Results from prior survey will be made available to the experts.
- **Validation**
  - After 70% consensus reached, terms will be circulated to the experts again for final review and feedback.



Others: ClinGen Data Modeling working group, AMIA Genomics and Translational Bioinformatics working group, HL7 Clinical Genomics working group, eMERGE, PharmGKB, IPWG, ASCPT, AMP, IGNITE, European Medicines Agency, CHMP Pharmacogenomics Working Party; G2MC Pharmacogenomics Working Group, IUPHAR Pharmacogenomics and genetics section



- Overview
- Members
- Working Groups
- ISCC Meetings and Activities
- Links and Resources
- Contact

Overview

The Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC) formed in February 2013 from the Genomic Medicine IV meeting to improve genomic literacy of physicians and other practitioners and to enhance the practice of ne through sharing of educational approaches and joint identification of educational needs. The group actions among medical professional societies and the NIH Institutes & Centers to exchange practices in genomic education and clinical care. By identifying needs of societies and clinicians in filling in gaps in nowledge and in providing effective educational efforts, the ISCC offers partnership and available ese societies to guide development of educational initiatives and applications for clinically relevant nomic science. Incremental evolution in identification of relevant sequence variation will permit gradual ractitioners' knowledge and practice in applying genomics to clinical care.

mation on the ISCC and its mission, refer to the following ISCC Description PDF

http://www.genome.gov/27554614

Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC)

1. Gather and facilitate dissemination of best practices and resources in genomic education and promote their translation into evidence based clinical care.
2. Assist societies and professional organizations in identifying gaps in medical knowledge, procedures, skills, or attitudes related to genomics, including how it relates to patient populations.
3. Assist societies in joint and separate publications of common interest related to genomic medicine.
4. Actively promote agreed-upon milestones and competencies related to genomic medicine.

- Education is the science and process of disseminating evidence and methods of use
- And verifying effect…

Gaps
in Healthcare Provider Education

- Materials
- Methods
- Motivation

Materials

- Collecting and matching quality resources with need, and disseminating them:
  - G2C2
  - MedEd Portal
  - Competencies (outcomes)
- Incentives for creation of resource materials reflecting emerging science for clinical benefit
- Recognition of the need for utilization of resource materials – marketing their availability
- Provider ←→ Patient language and visual tools

Competencies in G-2-C-2.org

Competency Map

Competency Map

Nurse

Genetic Counselor

Physician

Physician Assistant

Pharmacist

View the Competencies Guidelines for these disciplines:

- Genetic Counselors
- Nurses - (Competencies to which resources are currently mapped)
- Nurses - Essential Genetic and Genomic Competencies for Nurses with Graduate Degrees (document provided for your information-resources not yet mapped to these competencies)
- Pharmacists
- Physician: Framework for Physician Competencies
- ISCC Competencies (coded for resource mapping)
- Physician: ACMG Competencies
- Physician: ISCC Membership
- Physician Assistants

Methods

- Timing
  - Academic Training (medical – nursing – pharmacy etc.)
  - Residency
  - CME
  - Maintenance of Certification (Boards)
- Target
  - Physicians
  - Non-physician providers
  - Inter-professional education
- Teachers vs. students
  - Geneticists and Genetic Counselors, others…
- Testing
  - Education research that studies the most effective/efficient education methodology in the current milieu is missing

Pathologists | Laboratory Professionals | Board of Certification | Students

Training Residents In Genomics

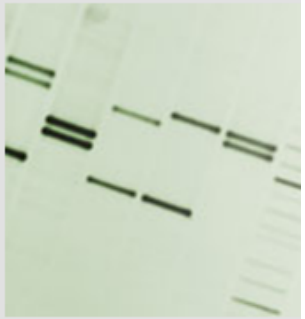
Preparing Pathologists for a Leading Role in Genomics

TRIG Resident Workshop Instructor Materials Now Available!

ABOUT | COURSES/WORKSHOPS | RESOURCES | EVALUATION | LINKS/LITERATURE

About

In 2010, the Training Residents in Genomics (TRIG) Working Group was formed through the Pathology Residency Directors Section (PRODS) of the Association of Pathology Chairs (APC). The goals of this group, made up of experts in medical education, molecular pathology, and clinical genetics, are to develop teaching tools, and promote genomic pathology education. The TRIG Working Group represents a unique collaborative effort in pathology education with members from many major pathology organizations and representatives from the National Society of Genetic Counselors, American College of Medical Genetics and Genomics, and the National Coalition for Health Provider Education in Genetics.



View members of the TRIG Working Group

http://www.pathologylearning.org/trig

Point of Care Education

Motivation (Relevance)

- Right to Practice
  - Board certification
  - Maintenance of Certification
- Value
  - Coverage and Reimbursement
  - Improved patient outcomes
- Evidence
  - Published peer-reviewed
  - Professional practice guidelines
- System Priorities
  - Healthcare System Administrators

ISCC Challenges

- Members are varied
  - Differences in governance, specialty focus
  - Differences in roles and responsibilities as part of the team
- Mission
  - Member societies each have own mission
  - *Not* a research mission
- Money
  - Volunteer organization, no dues, no purse
  - *not* a research organization (no research grants)
  - Approval to seek donor funding being sought
  - c/w UK NHS: spending £20 million on provider education for genomic medicine
- Metrics
  - How do we know if we're having any effect?
  - …when specific education in genomics is no longer needed?

ISCC Opportunities

- GM Programs' Education working groups – overlap with ISCC mission
- Take advantage of Leader of Leaders status
- **Connect researchers with organizations that can facilitate**
  - Provider education
  - Education research
  - *Planning research projects*
  - Implementation research
  - Disseminate evidence

Proposed Discussion Points

- Lots of groups working on education
  - Do we know what “we” want for education?
  - Are all gaps/barriers being addressed?
  - Is coordination across projects adequate?
- Lots of groups working on reporting results to clinicians
  - Is there a need/desire to catalog approaches?
  - To harmonize approaches?
  - How are resulting approaches being shared?