Potential Synergies and Collaborations

- Training materials and programs
- Improving report presentation, engaging labs and systems engineers
- Defining educational taxonomy and pedagogy
- Evaluation methods
- Outcome data
- Engaging relevant healthcare professionals including lab scientists, faculty, team approach
- Convincing ministries and funders (“sales”); clinicians and professional societies
- Reaching non-research intensive practices
- Mainstreaming into existing education
Collaborations – Training Materials/Programs

- HEE Masters’ modules and short courses
- UM Masters’ coursework
- NHGRI G2C2, insurers’ webinars
- AGHA Program 4 (and Genome Plus?)
- ASHG Cancer Genetics and virtual meetings
- G2MC webex
- Geisinger and similar local case-based modules
Collaborations – Pedagogy and Taxonomy

- Identify and capitalize on teachable moments
- Involve pedagogical experts
- Case study approach—substantial time involvement (~20 hrs), have elements of cases rather than fully formed
- Framework that can drop in specifics
- Start case with specific point you’re trying to teach
- Consider repurposing existing texts with publishers’ permission
- Engage professional societies in case review or collegial specialists
Collaborations – Improving Report Presentation

- Experts in form design and presentation
- Clinical decision support and alert fatigue
- Open InfoButton
- Effort for standardizing reports within UK
- ACMG has defined elements but not format
- Compare best practices (?CDC), engage labs, choose key components and complements
- Tiers: known variants, unknown variants related to symptoms, incidental findings
Collaborations – Evaluation Methods

• Standardized plans for evaluation with common outcomes defined similarly

• Creation of templates for
  – Workshops
  – Online tools

• Consensus options for research designs in evaluation of education methods
Collaborations – Outcomes of Training

• Knowledge, attitudes, behaviors
• Processes of practice
• Cost and reimbursement success
• Morbidity, mortality, disability
Inter-Professional Collaborations

• Parallel information to physicians’
  – Different emphases or levels?
• More than facilitator model
Collaborations – Convincing Ministers and Funders (“sales”)

- Evidence of need for education
  - Cases of misinterpretation
  - Cases of inappropriate ordering and costs
- Minimizing cost
- Evidence of effectiveness of education
Collaborations – Convincing Students, Clinicians, Professional Societies

• Competitiveness in residency applications
• Critical need for CME– how similar are requirements internationally, how to meet them
• Engage other professional societies by helping develop educational modules, link with their genomics adopters (TRIG model)
Collaborations – Reaching Non-Research Intensive Practices

- Lack of tertiary care and academic medical centers as leaders
- NCHPEG grant competition for societies to develop their own materials and disseminate them (pediatric neurology, dentist, speech/hearing)
- Museum-like programs, “Unlocking Life’s Code”
- Genomics England’s eligibility materials and recruiting patients, involved in return
- Pint of Science in pubs?
Collaborations – Mainstreaming into Existing Education

• Three-year horizon for HEE genomics education program, only til April 2018
• Mainstreamed into business as usual
• 2020 strategy
• IGNITE model for dissemination
Summary and Overview

- How to
Overview of Primary Care Oriented Education Programs

- Organizing and systematizing available materials, tracks, and summaries
- Exome sign-out rounds by webex
- Geisinger program – provider educational modules for all 27 genetic conditions affected by their 76 actionable genes
Lessons Learned for Meeting Changing Educational Needs

- Value of champions/early adopters
- Include evaluation of burden on instructors
- Add implementation scientists to program
- Have variety of “step-off levels”
- Millennial learners and online formats
- Consider entry points for subspecialists
- Focus on things clinicians are likely to see soon
- Describe future: need to prepare for rapid change by identifying underlying causes of disease, will drive transdisciplinary approaches
Best Practices for Implementation

• Mock genetic counseling sessions at professional societies—involve PharmDs in providing PGx info
• Pairing experienced sites with new adopter sites
• Engaging leadership at highest levels
• Create leaders in other specialties—use them to create network of other champions
• Advocates among junior doctors and patients
• Evidence of ROI—quality and safety
Challenges for Education Programs

- Evidence of effectiveness of genomics
- Evidence of effectiveness of training programs
- Funding
- How to reach non-research intensive health care utilization areas
- How to reach clinicians who’ve finished training—professional societies and accreditation standards
- Improving clinicians’ confidence
- Avoiding over-interpretation of VUS—improving reporting
Synergies and Opportunities to Share and Collaborate

- Education and Training GeCIP to form international network (ISCC, G2MC, IGEN)
  - Broad opportunities to work in reading library
  - Rare disease conditions IRDiRC and UDNI
  - Identify best training methods for training needs
  - Best pedagogic methods
- Agreement on disease gene panels and reportable findings
- Need to “anticipate and prepare for what new technologies will emerge and what will be consigned to history”
“…anticipate and prepare for what new technologies will emerge and what will be consigned to history…”

https://www.healthcare.siemens.com/magnetic-resonance-imaging/0-35-to-1-5t-mri-scanner/magnetom-aera/use
Synergies and Opportunities to Share and Collaborate

• Education and Training GeCIP to form international network
• Agreement on disease gene panels and reportable findings
• Need to “anticipate and prepare for what new technologies will emerge and what will be consigned to history”
• Data deposition into ClinVar and DeCIPHER
• Implement HEE Masters’ in U.S.
• Implement UM Masters’ in Commonwealth
• Templates for evaluation
NCBI’s ClinVar

ClinVar

ClinVar aggregates information about sequence variation and its relationship to human health.

doi:10.1093/nar/gkt1113

ClinVar: public archive of relationships among sequence variation and human phenotype

Melissa J. Landrum, Jennifer M. Lee, George R. Riley, Wonhee Jang, Wendy S. Rubinstein, Deanna M. Church and Donna R. Maglott*

National Center for Biotechnology Information, National Library of Medicine, National Institutes of Health, 8600 Rockville Pike, Bethesda, MD 20894, USA

Received September 13, 2013; Revised October 21, 2013; Accepted October 22, 2013

Courtesy E. Ramos, NHGRI
Sharing Genomic Variation Results - ClinVar

Courtesy Erin Ramos, NHGRI
### Top ClinVar Submitters

<table>
<thead>
<tr>
<th>Category</th>
<th>Submitter</th>
<th># of Variants</th>
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<tr>
<td><strong>Expert Panels and Practice Guidelines</strong></td>
<td>International Society for Gastrointestinal Hereditary Tumours (InSiGHT)</td>
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<td>Evidence-based Network for Interpretation of Germline Mutant Alleles (ENIGMA)</td>
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<td><strong>Clinical Laboratories with ≥1000 interpreted variants</strong></td>
<td>GeneDx</td>
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<td>Emory University Genetics Laboratory</td>
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<td>Ambry Genetics</td>
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<td>International Standards for Cytogenomic Arrays (ISCA) Consortium</td>
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<td><strong>Research Programs and Locus-Specific Databases with ≥500 interpreted variants</strong></td>
<td>Sharing Clinical Reports Project for BRCA1 and BRCA2</td>
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<td>ClinSeq Project, National Human Genome Research Institute, NIH</td>
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<td>Breast Cancer Information Core (BIC)</td>
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<td>Royal Brompton Hospital Cardiovascular Biomedical Research Unit</td>
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<td>RettBASE</td>
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<td>Children's Mercy Hospital and Clinics</td>
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<td>Muilu Laboratory, Institute for Molecular Medicine Finland</td>
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<td>University of Tartu, Institute of Molecular and Cell Biology</td>
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<td><strong>Aggregate Databases</strong></td>
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<td>GeneReviews</td>
<td>5,202</td>
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<td><strong>Submitters from Israel</strong></td>
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<td>Erez Levanon Lab, Bar Ilan University</td>
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<td>Department of Human Genetics, Rambam Health Care Campus</td>
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<td>The Institute of Human Genetics, Galilee Medical Center</td>
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<td>Molecular Metabolic Laboratory, Sheba Medical Center Tel-Hashomer</td>
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<tr>
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<td>Sackler Faculty of Medicine, Tel Aviv University</td>
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Top ClinVar Submitters

ClinVar Submission Portal

V600E (SUB 1)

Variant description

* Accession and version of reference sequence
NM_004333.4

* Description of the sequence change
`c.1799T>A`

Gene (Please start typing and select from the list)
BRAF: B-Raf proto-oncogene, serine/threonine kinase

Alternate designations
V600E

Add another alternate designation

Variation identifiers

dbSNP:rs113488022

Add another variation identifier

Location
NM_004333.4:exon 15

General information

* For which type of variant
Single variant

* Select method of denotation
HGVS

* Assembly
GRCh37

Sackler Faculty of Medicine, Tel Aviv University
