

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
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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
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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)
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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

- Organizational track
- Exome
- Geisler for a

The screenshot shows the Baylor Miraca Genetics Laboratories website. At the top left is the logo with a DNA double helix. The text 'Baylor Miraca Genetics Laboratories' is on the left, and 'Baylor Miraca Genetics Laboratories' is on the right. Below the logo is a navigation bar with links: '>BCM Home >BCM Centers >BCM Departments >Find a BCM person >Giving'. Below that is a location bar for 'Houston, Texas'. A large blue DNA double helix image spans the width of the page. On the left is a vertical navigation menu with items: Home, Testing Available, About Us, Billing, Forms, Shipping Information, Training Programs, Resources, and Add-on Test. The main content area is titled 'Educational Videos' and features 'Exome Sign-Out Conference'. A note reads: 'Note: Please click on "Play All" then click on "Playlist" in the gray bar to select the conference date you would like to view.' Below the note is a video player showing a video titled 'Whole Exome Sequencing Sign-out Meeting' with a play button and the date 'April 24, 2015'.

Materials,
Modules
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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...”



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

NCBI Resources How To

ClinVar ClinVar Search

Advanced

AATTGTACTGATGGTATGGGGCCAAGAGA
CCAAGGACAGGTACGGCTGTCATCACTTAC
CAGGAGCCAGGGCTGGGCATAAAAGTCAGC
ACAG
GCCC
TCTG

ClinVar

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

D980–D985 Nucleic Acids Research, 2014, Vol. 42, Database issue *Published online 14 November 2013*
doi:10.1093/nar/gkt1113

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

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

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NCBI Help Manual	Data & Software	Nucleotide	PubMed Health	Research at NCBI
NCBI Handbook	DNA & RNA	BLAST	GenBank	NCBI Newsletter

Courtesy E. Ramos, NHGRI

Sharing Genomic Variation Results - ClinVar

The image shows a screenshot of the ClinVar Variation Viewer interface. The main view displays details for a variant: **Clinical significance: Likely pathogenic** (indicated by two stars), **Review status: ★★☆☆**, and **Number of submission(s): 2**. The variant is identified as **NM_000257.3(MYH7):c.2105T>A (p.Ile702Asn)**. The associated **Condition(s)** include **Primary familial hypertrophic cardiomyopathy** and **Cardiomyopathy**. A table below the variant details shows the clinical significance and review status for multiple submissions, with the most recent being **Likely pathogenic (May 23, 2014)**.

A pyramid diagram illustrates the criteria for clinical significance, with the following levels from top to bottom:

- Multi-Source Consistency** (★★★)
- Single Submitter – Criteria Provided** (★)
- Single Submitter – No Criteria Provided** (No stars)
- No Assertion** (Not applicable)

Top ClinVar Submitters

Category	Submitter	# of Variants
Expert Panels and Practice Guidelines	International Society for Gastrointestinal Hereditary Tumours (InSiGHT)	2,368
	Evidence-based Network for Interpretation of Germline Mutant Alleles (ENIGMA)	1,264
Clinical Laboratories with ≥ 1000 interpreted variants	GeneDx	24,691
	Partners HealthCare Laboratory for Molecular Medicine	16,430
	Emory University Genetics Laboratory	16,047
	Ambry Genetics	16,035
	International Standards for Cytogenomic Arrays (ISCA) Consortium	13,971
Research Programs and Locus-Specific Databases with ≥ 500 interpreted variants	Sharing Clinical Reports Project for BRCA1 and BRCA2	2,221
	ClinSeq Project, National Human Genome Research Institute, NIH	2,137
	Breast Cancer Information Core (BIC)	2,001
	Royal Brompton Hospital Cardiovascular Biomedical Research Unit	1,521
	RettBASE	1,097
	Children's Mercy Hospital and Clinics	1,058
	Muilu Laboratory, Institute for Molecular Medicine Finland	840
University of Tartu, Institute of Molecular and Cell Biology	761	
Aggregate Databases	Online Mendelian Inheritance in Man (OMIM)	26,804
	GeneReviews	5,202
Submitters from Israel	Laboratory of Prof. Karen Avraham, Tel Aviv University	46
	Erez Levanon Lab, Bar Ilan University	4
	Department of Human Genetics, Rambam Health Care Campus	1
	The Institute of Human Genetics, Galilee Medical Center	1
	Molecular Metabolic Laboratory, Sheba Medical Center Tel-Hashomer	1
Sackler Faculty of Medicine, Tel Aviv University	1	

Top ClinVar Submitters

ClinVar Submission Portal

of
Variants

V600E (SUB)

Unfinished at the V

Info Organization Va

Variant

General information

* For which type of va

- Single variant
- Two single variants
- Haplotype

* Select method of de

- HGVS
- Sequence location

* Assembly

GRCh37

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

remove

Add another alternate designation

Variation identifiers

dbSNP:rs113488022

remove

Add another variation identifier

Location

NM_004333.4:exon 15

homer

Sackler Faculty of Medicine, Tel Aviv University

368
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