

Discussion: Return of Genetic Results

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Major themes:

- Research vs. Implementation
 - GWAS vs. Sequence
- Incidental Findings:
 - Research vs. Clinical application
- Consent and Patient Preference
 - Duty to warn- patient, family, population?
- CLIA approved lab results
- EMR and CDS, duty to reinterpret VUS

ROR for discovery by GWAS

- Gene discovery- association studies
 - Very little data actionable
 - 23 and me model- well liked
- Most research subjects very interested and would want individual data
 - Are we too paternalistic?
 - Could release of this information increase genetic literacy?
 - Return info on a group level?
 - Risk score data for macular degeneration and other eMERGE projects a good star
 - Can f/u subjects on preference over time and literacy

ROR for research sequencing studies

- WGS/Exome
 - Sequencing provides better data with firmer disease associations, but still problems
 - Read length, repeats, patient and provider comprehension
- CLIA issue
 - Institutional requirements for research data may differ
- Selected genes for study vs. IFs
 - ACMG list, expanded? Who curates?

Consent

- Research Informed Consent vs. Consent to treat
- What Clinical tests require specific consent?
 - Range of clinical tests
 - TPMT to Huntington disease
 - Incidental findings (IF) in clinical setting- consent for release?
- Research-
 - Only 10% of research studies mention IF (Lawrenz and Sobotka, J Law, Med and Ethics, 2008)
 - Some research studies state only subjects will not be informed
 - Most not in CLIA certified labs
 - Often need to retest in approved lab

Patient Preferences: Issues

- Many subjects unclear on research vs. clinical care
- Most patients/subjects state they want all results returned
 - K Hudson- 96% of veterans want results
 - But must honor the wishes of those that don't
 - Don't test, don't return results??
 - Often preference changes at time of test
- Consent documents vs. *a la carte* menu
 - What education is required?
- Return to Provider/EMR vs. to subject/patient and others
- Good opportunity for health services research-
 - Patient preferences and reactions over different models

“Fostering a culture of fear”

- Virginia Hughes, Slate.com
 - Incidental findings vs. “Dark DNA secrets” (Time)
- Many genetic tests require consent and counseling- where should this line be drawn?
 - CA?, PGX?
 - Most millennials not greatly concerned
 - GINA and ACA offer some protection
- What are community preferences for consent for genetic testing in practice?

Computerized decision support:

- Much genetic CDS effort focused on interpretation, data curation, technical areas
- Relatively little on human factors and implementation
 - Standard drug interaction only notifies after order complete
- What methods work to bring genetics into clinic workflow?
 - What education is needed, and how to deliver?

eMERGE Opportunities

- How can ROR provide genetic education?
 - Patients, providers, community
- Patient and population preferences over time
 - Targeted education, time on web
 - Tools used, questions posed
- Human factors in engaging EMR CDS
 - Pop up errors vs. intelligent drug choice
 - PGX obvious good start