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 Hudsen- 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, provides, 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