Implementing GM – Laboratories

- Labs should share genomic data; many clinical labs willing but requires resources
- “Don’t put in clinical lab til well-established” but WGS/WES breaks that rule (66%-80% variants found in only one family)
- Delivering information from available sequence takes resources
- Updating needs resources – changed categories 300 times, ~4% MD reports change per year
- How to enable hospital/academic CLIA-certified labs to move to NG sequencing (resources for infrastructure)
- Much of recent growth in genomic testing is ID (11%), germline and cancer 18%)

Implementing GM – Laboratories

- "Whole genome" may imply complete or infallible
- No CLIA standards yet for nextgen sequencing (though CAP, ACMG, CLSI, and AMP developing checklists)
- Uncertainty of regulatory oversight (i.e., allowed as lab-developed test?)
- Need genomic medicine specialty?
- What is regulated under CLIA vs. what is art of medicine in interpretation of sequences
- Try to capture the marked variability in interpretation to understand it
- Need bioinformaticians incorporated into pathology groups

Implementing GM – Common Criteria for Adopting in Commercial Laboratories

- Well-controlled and adequately powered studies demonstrating analytic validity and clinical utility where feasible)
- Clearly actionable results:
  - Prevent drug toxicity
  - Identify treatment path
  - Diagnosis of rare heritable disorders and carrier testing
- Path to fair reimbursement
- Freedom to operate: patent issue is huge (clearinghouse for patents in development)

Sequencing Working Group

- White paper laying out research and policy agenda for implementation of sequencing
- Focus on those that are gaps, not being done by other groups
- Highest priorities– how to assign clinical relevance to variants?
- Wet lab moving so fast not clearly gap area
- Consider genomic “critical values”
- Determine what legal requirements are for data return – varies by state
- No substitute for knowing what pt and clinician want to know

Implementing GM – Financial Impact and Reimbursement

- Utilization of imaging driven by regulatory approval and reimbursement rather than by evidence they provide benefit
- Evidence evaluation needs to work from clinical problem rather than starting with test
- Coverage policy principles
  - Services related to prevention, dx, tx
  - Info will affect course of treatment
  - Care and/or treatment likely to improve outcome
  - Improvement attainable outside investigational settings
  - Services consistent with plan design

Implementing GM – Financial Impact and Reimbursement

Evidence standards

- Analytic validity, clinical validity, clinical utility
- Final approval from appropriate regulatory bodies helpful, or necessary when required
- Demonstrated benefit

Telephonic genetic counselors substantial advance

Implementing GM – Financial Impact and Reimbursement

- Unit costs are biggest driver of escalation in healthcare costs, not utilization
- Costs for molecular diagnostics have risen much faster than other costs (14%/yr 2008-10)
- Payers should not fear innovation (always seems to cost more) but look for those that disruptively replace more expensive and less effective technologies
- **Public-private-academic collaborations to develop, design, fund, conduct, interpret research to produce decisive information**

Implementing GM – Public Health

- Potential partnerships:
  - Genetics and chronic disease leadership in state health depts
  - Local cancer and heart disease coalitions
  - National professional and disease-related organizations
  - Genetic Alliance, Patients Like Me
- Consider cross-cutting goals, impact on health disparities
- HFE homozygotes with s/s receiving iron

Family History Working Group – Possible Opportunities

- SBIR/STTR with EPIC
- Social network software and infrastructure for collecting/correcting FHH info from relatives
- FHH interventions
  - Optimize in emergency situations, especially regarding potential MI
  - Bring to other environments such as rural, underserved
  - Educational - residents in training
  - Does intervention work in usual care
- Link to sequencing WG on both Mendelian and complex traits

Periodontal Microbiome Working Group

- Management of patients with diabetes and periodontitis
- Management of dental patients
  - Pain
  - Coagulation
- PGx data to dentists with CDS tools
- Oral-systemic personalized medicine model
- Sequencing may replace culture in micro lab – potential for huge impact

Planning Committee

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