Session 1 – Basics of implementation science

- Defined implementation science, dissemination research, implementation research
- Timing of implementation research when evidence still evolving– when are we ready
- Lessons from Pronovost central line infection checklist– needs to be modified for each site
- Dozens of models and frameworks– can we develop specialized implementation framework for genomic medicine?
- Novel ways to fund this research outside slow NIH processes, similar to clinical trials networks?

Session 1 – Basics of implementation science (cont)

- Could we expand IS into larger sphere than just existing medical care systems (DTC?)
- Can we leverage payers' decisions (reimbursing for MSI testing) to accomplish other goals (Lynch syndrome identification)

Session 2 – Resources for genomic medicine implementation

- Identify common threads of genom med implementation to build implementation guides
- Need some de-implementation guidance as for long QT variants no longer deemed pathogenic, removing codeine from pediatric formulary
- Payers paying for testing is tiny fraction of cost, need broader view of real cost of implementation
- Do we need a CPIC for non-PGx genes, especially for non-Mendelian conditions?
- Resource needs in informatics esp accessing pt-level pheno/genotype data; standards established when trying to share or submit to useful resource

Session 3 – Novel models of genomic medicine implementation

- Phenotype risk scores use EHR phenotype data to identify probable Mendelians– how to develop and disseminate
- Comparison of weekly expenditures in advanced cancer care is useful metric, nearly every category reduced except drug cost
- Can we develop and share attractive data portals for clinicians and patients– EHR interfaces are dreary
- Incentivize standardization of APIs
- Develop “genomed” patient registry similar to cardiovascular data registry
- Do more with patient-reported outcomes– CEOs pay attention to these

Primary Care Debate

- Geneticists still largely make diagnosis and refer back for management so PCP has to be involved
- Rare serious disorders– realm of geneticists
- PGx– not realm of geneticists (? PCP vs pharm)
- Shift more genetics care to genetic counselors (find ways to bill for this)
- Develop limited training for majority of common complex diseases, certify as consultant
- Seed relevant specialties with needed info
- Can some diagnostics be done by AI– often genome reveals answer after long line of clinicians including geneticists

Primary Care Debate

- Geneticists still largely make diagnosis and refer back for management so PCP has to be involved
- How to ensure clinicians adequately exposed and trained throughout professional lifespan
- Rare serious disorders– realm of geneticists
- PGx– not realm of geneticists (? PCP vs pharm)
- Shift more genetics care to genetic counselors (find ways to bill for this)
- Need innovative care delivery models
- Develop limited training for majority of common complex diseases, certify as consultant
- Seed relevant specialties with needed info
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Session 4 – EHRs in implementation

- Improving CDS is limited by a lack of institutional acceptance of supporting evidence
- CDS architecture varies creating a barrier
- Tension between operational IT and “research” use of the EHR
- Defined framework: stakeholders, transactions and clinical systems
- CDS for Patient Screening is a great opportunity
- HL7 domain analysis document provides use cases; establish community process for validating it
- Take necessary time to develop enduring standards

Session 4 – EHRs in implementation

- Need to standardize technology and variant specifications
- FHIR is an important resource
- HL7 clinical genomics workgroup is likely to have a huge impact
- ClinGen/NCBI allele registry is a great resource
- Embrace a common data model (CDM)
- One barrier is everyone wants to use their own
- FHIR provides more flexibility than standard CDMs as is pluripotent data model; use it as pt-engagement tool
- Support tools for sharing lessons learned and for trying out CDS in test platform

Session 5 – What evidence is needed

- Evidence exists, stop focusing on it (don’t be held hostage by “not enough evidence),” get on with it
- Employers may have lower threshold than payers; consumers/employees are going to be important drivers, increase focus on them
- Reduce emphasis on educating everyone involved
- Employers in the service of their employees are going to be an important force in moving genomic medicine forward
- Need research showing employers benefit from adoption; better utilize evidence being generated even if imperfect

Session 5 – What evidence is needed

- We should convene a group of employers, work with employers to ensure rolling out in way evidence captured and publishable; provide consulting help on ground
- Employers self-aggregating into consortia, take advantage of for research
- Need to develop a basic genomics formulary
- Public payers still major tough nut to crack

NHGRI Strategic Plan

- Can we develop partnerships with regulatory agencies and payers to get clearer needs and priorities for evidence generation
- Are we shifting goals of research from high-value publications to convincing payers
- Define what NHGRI can own
- Need economic studies for pre-emptive testing; geneticists not trained in this
- Need improved standardization of genome-related phenome

NHGRI Strategic Plan

- ` Increase emphasis on last-mile problem of clinician and patient interacting
- Be more pt-focused, involve them more in care processes
- Don’t forget the babies…
- Capture medicine-based evidence
- Capture longer-term outcomes