Morning day 2

- Paul Ridker: Barriers to getting a test implemented. Importance of basic science links. Can work with industry. Guidelines are very slow to develop (maybe not so in oncology; ?changing landscape)

Morning day 2 summary

Pearl O'Rourke

- Bench versus clinician tensions
- Liability concerns, lack of insights of the "other side", "I already practice personalized medicine"
- Soccer is the game of the future and always will be
- Suggestions:
- How do people decide what to do? Need assessments from clinicians, for the system, patient expectations. Encourage translation & interactions, quite early. Maximize opportunities e.g. make sure genomic data are included in large trials
- Need to create new cadre of translationalists: PhDs? Including informaticians/mathematicians.

Morning day 2

- McLeod
- Do sequencing platforms add value to PGx
  - Technical aspects of variant calling
  - Genotype-phenotype relations
- “meta-meta-analysis”: what is the best practice for assessment. What elements need to be included a PGx analysis or meta-analysis? (dose, indication, etc)
- Compare platforms
- Who is the advocate for PGx? Comparison to imaging
- **What are the right endpoints? Staying on statin? Getting out of hospital?
- What do employers (payers) see as important endpoints?
- Payers will want evidence that acting on any of this will change outcome. Whatever the outcome is.

Rex's barriers for genomic medicine applied to pharmacogenomics

- Biology
- Lack of evidence
- Institution and physician acceptance
- Education patients physicians
- Consents
- Sample availability and biobanking
- recruitment

Updates from working groups

- Sequencing
  - return of reports, focus on quality
  - Build metrics from standard sets; different metrics for 3000 clinical genes?
  - Best practices needed:
    - CAP & CLIA will do this
    - Ensure coverage of target regions and QC
    - Generate standard sample sets; metrics for comparing platforms; data analysis and other software;
  - Target to move to removing need for validation
  - Who curates which set? How are data displayed in EMR?

Updates from working groups

- Family history update
  - EMR integration not yet standardized
    - Encourage patient input
    - Add other risk calculators to FHx
    - Varying display approaches
    - Need concensus with EMR vendors
  - Expanded data capture approaches (web 3.0)
  - Demonstration project: develop and validate tools to inform any system on how to acquire and display FHx in EMRs. Implement in naïve sites, and evaluate performance. Are data valid? Are data used? Change in behaviors or referrals. Create a Spanish version of the tools. Create guest accounts to tools?
  - Use same approaches to study “PGx” or “sequence data” instead of “FHx”

Updates from working groups

- Extend studies of the relationship of the microbiome and human phenotypes to other phenotypes: RA, cancer
- Extend numbers of centers

Physician education

Passamani

- Specialties march through at different tempos
- Professional associations are the ”in” to practitioners: education, guidelines. Timing is key: too early or too late are both undesirable.
- genuine interest & apprehension, NEJM series, NHGRI TV.
- 850,000 licensed MDs, 625,000 do patient care, 209,000 in primary care
- PGx as a first focus. Use case vignettes.

Physician education

Passamani

- Partnerships in education?
  - ACP is now interested
  - Aetna has a budget for education. Figure out how to partner with them
- MDs as patients
- Drivers for MD interest: CME, recertification, patient interest. Web-based short courses for CME credit – downside of “web alert fatigue”
- ??Action item: see how this is done at other sites

Physician education

Passamani

- use informatics approaches
- educational requirements different at different stages.
- genetic testing on yourself as an educational experience.
- Include genetics focus in other subspecialties. Learn about uncertainty.
- ??T32

Idea 1

- Take up the challenge of partnering with payers to identify evidence for outcomes
  - ?Center for Medical Technology Policy ?NHGRI
  - frame as how to design study and metrics to do the right thing for patients in this space, rather than “why aren’t they paying”
  - Have other NIH ICs done this? Partner with … AHRQ? FNIH? PCORI? Need health care economists

Genome Medicine IV

- Maintain momentum by
  - Holding next meeting sooner (September) versus later (December)
- Include focus on standards for sequencing, reporting, documenting in EMR; may be a sub/working group

Genome Medicine IV

- Theme for GM4: continuing to engage stakeholders
  - professional organizations: ASCO, IDSA. Neurology, ~~pharmacy?~~, AAFP, AAP, ACC, ACMG, ACP, AHA, AMP, CAP, CPIC, SGIM, DOs, … Need to hear:
    - relevance of genomics to their field
    - levels of evidence they find convincing
    - how they make decisions on guideline issuance
    - professional education initiatives
    - Case studies
    - Learn from CABig
  - research organizations: PCORI, AHRQ
  - patient groups: Patients Like Me, Genetic Alliance
  - EMRs: Cerner, EPIC, GE
  - FDA, CAP (CLIA), Setting standards. CDC
  - public health depts. ASTHO.
  - DTC and other private genome efforts: search for positive traits
- Workgroup meetings/progress: add one on systems/other “omes”. Where does microbe sequencing fit in?
- Location: Seattle? Bethesda? Dallas?

Next steps/action items

- 2. NHGRI: organize small meeting in July at NHGRI of GMWG, UnitedHealth (perhaps not Reed but “woman who works with Reed” per Marc), Medco (Robert Epstein?), Aetna, BCBSA
- 3. Teri: ask sequencing team about inviting GM3 sequencing wg representatives to Houston NHGRI sequencing meeting in October
- 4. Dan: at “next meeting: session today, propose Stakeholders II meeting in Bethesda or Dallas in fall—if focus on follow-up to July small meeting could be in September, if want to include sequencing wg standards development probably after October sequencing meeting in Houston
- 5. Teri: distribute UnitedHealth report to GMWG (DONE)
- 6: Marc: shower this morning

Idea 1

- - main idea from payers session: Sean Tunis would host payers and us to discuss next steps for designing study(-ies) of endpoints and evidence that would be convincing to them, what outcomes they'd use to make coverage decisions for genomic medicine implementations
- - need not be Tunis group in Baltimore, would be better to convene smaller meeting at NHGRI in July as start
- - frame as how to design study and metrics to do the right thing for patients in this space, rather than "why aren't they paying"
- - include in first meeting the payers who collect and/or have access to data: UnitedHealth, Medco, Aetna, BCBSA; small group ~12 people including these four payers, as many GMWG as can make it, NHGRI, probably not other Institutes/Centers just yet
- - try to use their data collection infrastructure rather than recreating or duplicating it
- - ask other payers regarding interactions with Tunis group
- - find out whether payers have done this kind of work with other NIH IC, AHRQ
- - plan on broader meeting in follow-up including health economists such as Scott Ramsey, David Veenstra
- - if develop public-private partnership could FNIH facilitate
- - include PCORI