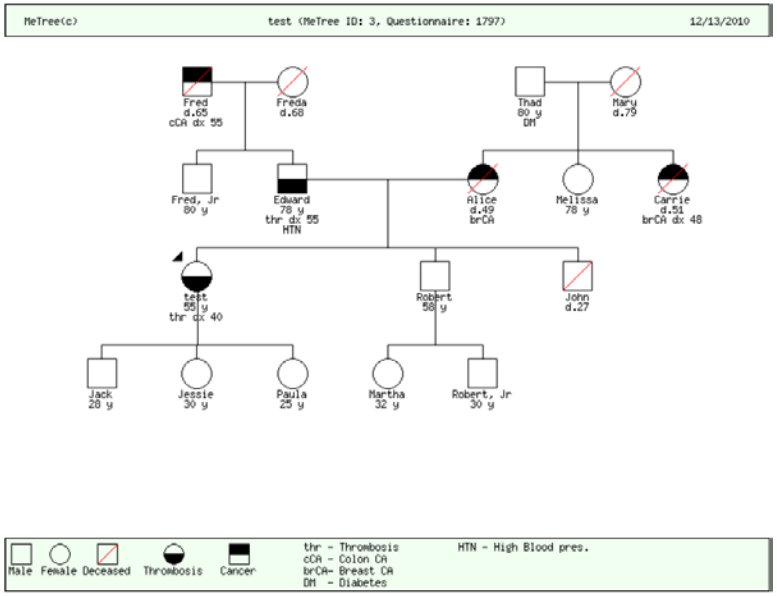

Family History: Breakout Discussion and Demo Projects

Genomic Medicine Meeting II

December 5 2011

Group

- Geoff Ginsburg
- Maren Scheuner
- Jeff Struewing
- Jonas Almeida
- Greg Feero
- David Adam
- Andy Faucett
- Murugu Manickam
- Maureen Smith
- David Dimmock
- David Valle
- Corina Din-Lovinescu

Topics

- Validation of family history information
- Implementation science to integrate FH into the clinical workflow
- Develop the ‘user interface’ for patients and providers for FH
- Develop the outcomes research agenda
- Holy grail (?) – integration of all data and development of disease risk models
- Assemble a group to facilitate incorporation of FH initiatives into large cohort studies

Validation of FH Information

- Goal – Increasing the accuracy of FH information
- Key questions
  - Is patient entered data accurate?
  - How often does a pedigree need to be updated?
- Possible initiatives and Demo projects to
  - Develop sets of iterative questions following baseline info and vary timing
  - Develop adaptive patient questionnaires
- Validation in the context of JHU's Mendelian diseases initiative using OMIM
- Benchmark against information from living affected individuals
- Evaluation of attitudes, beliefs, and cultural norms in obtaining family history

What is the best way to get FH into the clinical workflow?

- 1% of records have an adequate FH (genetic clinic)
- Discussed a series of small studies that interrogate the best/cheapest/least time consuming way to collect family history data
- What is the best way to get collect information?
- What is the optimal time to gather ?
- What are the incentives?
- Evaluate
  - Stand-alone application vs one integrated into the EMR
  - Pt entered vs PA/NP entered
  - Patient derived FH data vs that obtained in a clinical setting
- Explore electronic media tools to help patients create their own family histories and encourage their family members to participate
  - Wiki model for FH
  - Facebook application
  - iPad app
  - Genealogy web page

Information Interface and Education of Providers

- How do we take the information and create teaching tools and clinical decision support?
- What information about family history do PCPs want? What do specialists want?
  - Generalist want ‘flags’
  - Specialists want ‘data’
- Project
  - Survey providers in different settings and understand their needs and desires for optimal use of FH data
  - Create an informatics system that translates family history data for different audiences (ex. for PCPs, geneticists, residents, etc.).
- Build an educational module *into existing tools*
  - Draw pedigrees (we should not lose the genetics !)
  - Most likely diagnosis
  - Alternative diagnoses
  - Targeted and brief
  - Actionable information

Develop an Outcomes Research Agenda

- Desired Outcome - Adherence to evidence based guidelines (USPTF)
  - Provider behavior – compliance to guidelines
  - Patient behavior – compliance to recommendations
- Suggested studies
  - Retrospective analysis of outcomes associated with FH vs no FH
    - Marshfield/Geisinger/IMHC
- Cluster Randomization
  - Usual care vs FH informed care
  - Structured FH vs FH as dictated by the provider
- VA or EMERGE +/-FH collection (prospectively)
  - T2DM, CAD, CRC, BrCA
  - Compliance with guidelines
  - Diet
  - Exercise
  - Med compliance
  - Control of HTN, DM, cholesterol, cancer screening

Holy Grail?
Building models with all the data

- Gather and integrate all relevant risk data
  - FH
  - HRA/environment data
  - Genomic data
  - Other Molecular info
  - Clinical info
- Develop methodology and models that aggregate all info to predict disease risk
- Requires access to population studies
- Recommendation – structured FH data be incorporated into all NIH studies that are collecting genotyping or sequencing data and outcomes
  - National Children's Study
  - 1M Veteran study
  - Kaiser
  - Geisinger
  - IMHC

Advisory Group on FH

- Esteemed colleagues from breakout session (and others interested)
- Identify opportunities in ongoing population studies, longitudinal studies, or GWAS studies at NHGRI or other ICs
- Advise larger studies on implementation of FH and data collection
- Recommend incorporation of FH data
  - Into RFAs
  - Specify or suggest desired outcomes measures