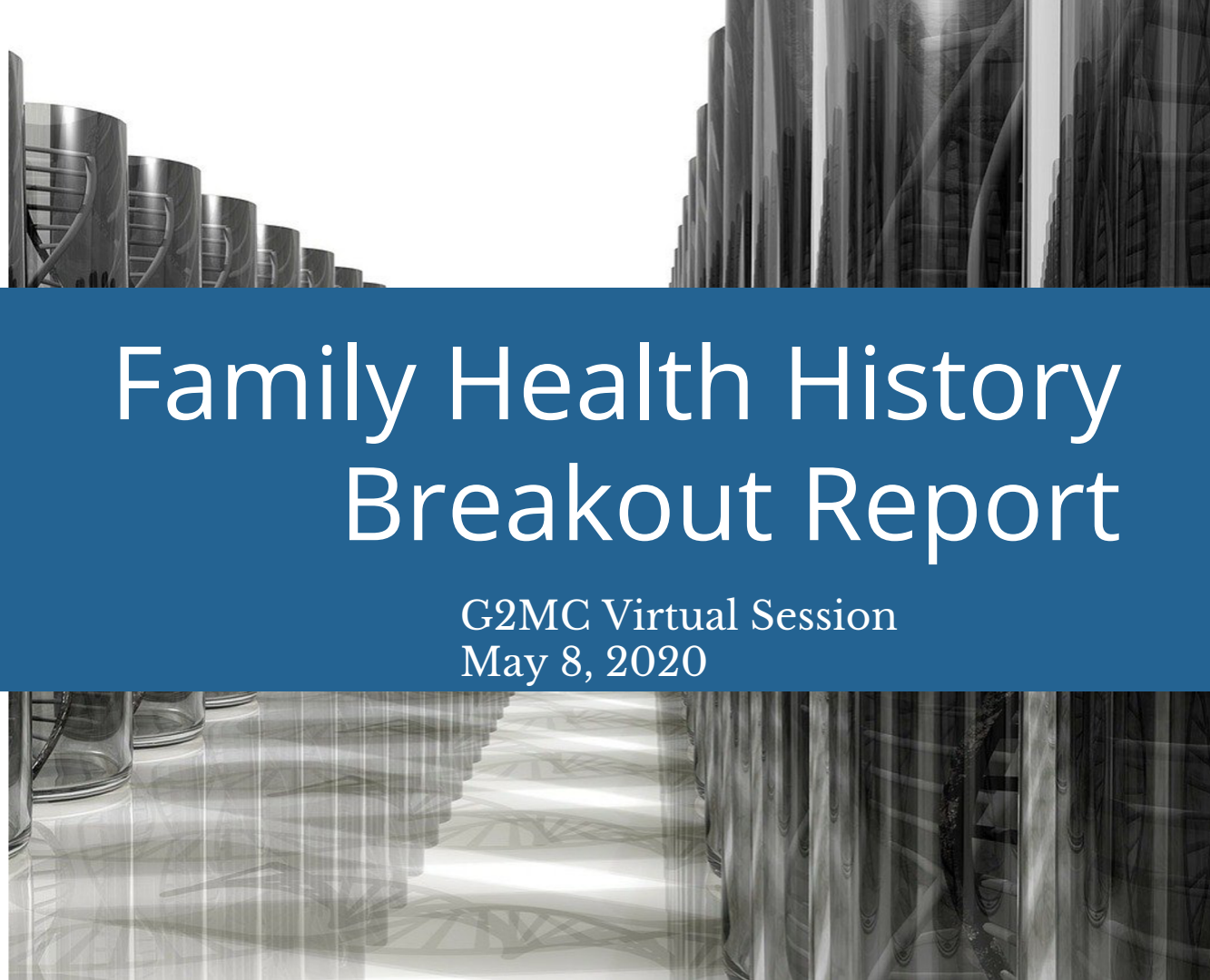


Family Health History Breakout Report

G2MC Virtual Session
May 8, 2020

Global Genomic Medicine Collaborative

Grant Opportunities and other Funding Ideas for a Shared Family Health History Project

- Dr. Theodora Katsila (Greece) outlined EU grants (digital biomarkers for public health, young investigators) postponed until later this year due to COVID-19. She will provide an outline of the funding opportunity and her aims.

- Geoff Ginsburg (USA) would like to see funding that would support G2MC hosting a FHH resource center for members to use: knowledge experts, examples of established implementations and solutions, papers, access to shared FHH tools, grant opportunities, etc.

Grant Opportunities and other Funding Ideas for a Shared Family Health History Project

- Other sources of funding could include –
  - Genealogy companies,
  - Cloud providers and Software tech - could provide in-kind support (personnel, etc) or funding,
  - Genetic testing labs,
  - Direct-to-consumer companies (e.g. 23andme, etc.)
  - Rare disease foundations (need to suggest specific projects closer to their interests)

- Need to create marketing stories for each potential funding partner

GA4GH and HL7 Related Activities

- Global Alliance for Genomics & Health Pedigree subgroup working on a minimum core dataset with common data elements and definitions for collection and sharing of FHH information.

- Reviewed current HL7 FHIR Pedigree standards.

Sharing of Use Cases

FHH Breakout

Common Family Health History Use Cases

- Familial risk assessment for a disease of interest
- Using FHH as an indication for genetic testing, or for genetic analysis post-genotyping/sequencing
- Using FHH, genotype, and phenotype (clinical) data of a patient or relative, to determine if the patient needs further testing or sequence analysis, and/or if a relative needs the same, which is called cascade testing
- Tracking in pedigrees inherited genetic disorders, such as both autosomal recessive and dominant, codominant, x-linked, and mitochondrial

Some shared stories

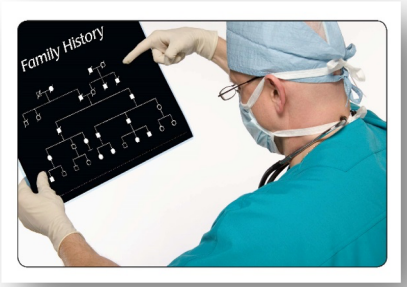
- Tomoharu Tokutomi (Japan) new tools one being in English, French, and Japanese
- Theodora Katsila (Greece) discussed her work in using FHH coupled with PGx for drug toxicity evaluation
- Sonia Margarit (Chile) described how the medical school is teaching student to administer pedigree analysis
- Franciso Domingues (Italy) described population study looked at chronic, age-related diseases. They are still interested in integrating FHH tools into this population. They are conducting a sub-study on COVID-19
- Geoff Ginsburg (USA) suggested that using FHH tools in the realm of infectious diseases
- Anne Ochoa (Philippines) is a Genetic Counseling student from Manila says they take family histories by hand, manually drawing family pedigrees up to third generation.
- Yusuf Danasabe Jobbi (Nigeria) shared that they take a short family history while doing case files and appreciates the chance to learn and have the opportunity to introduce more tools in their system.

Ongoing Collection of FHH Use Cases Using a Webform

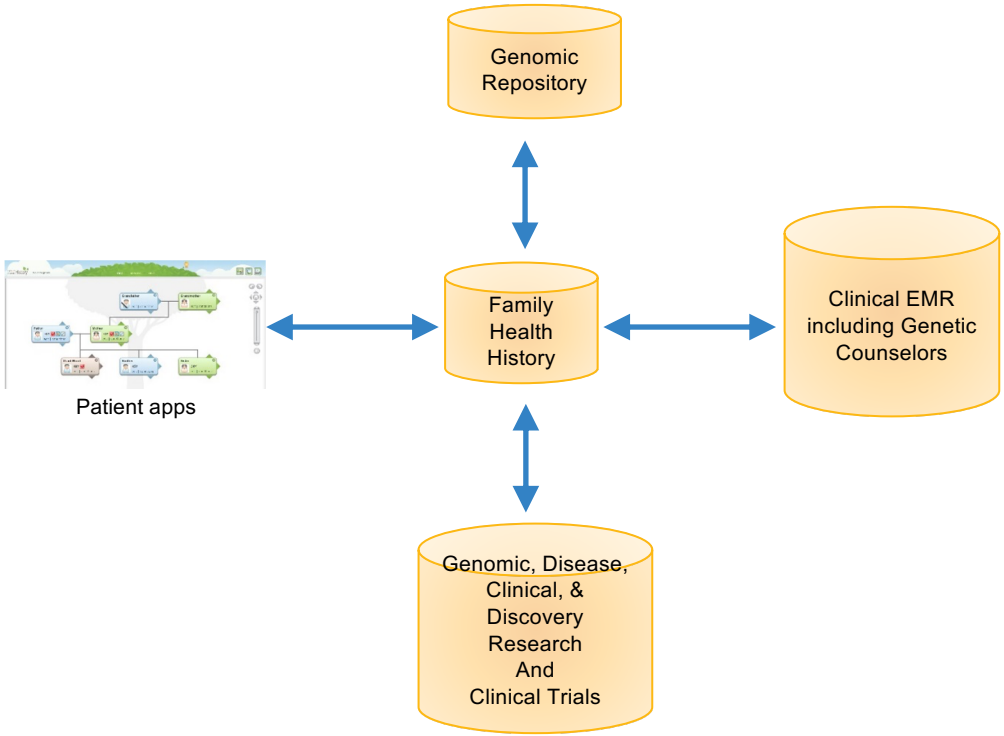
► Describe what FHH collection tools are currently in use and need to be created

► What is the targeted patient population and main data focus (disease, genetic variants, drug, etc.)

► Explain the main goal and high-level clinical outcome you want to achieve



Universal FHH Use Case for Data Sharing



Goals review and discussion

Short term – infrastructure

- Discuss what tools can be shared, or must be local
- Local data storage requirements
- Discuss how data could be accessed/shared across countries
- List other local requirements (language, devices, etc.)

Goals review and discussion

Long term – Desired outcomes

- Utilize common or standardized method/process to capture data
- Generate consistent FHH data across multiple different research and care sites
- Analyze FHH data to develop clinical decision support algorithms