Building a Family Network
GHC/GHRI/UW

GHC = Kaiser Permanente of Washington
GHRI = Kaiser Permanente Washington Health Research Institute

Kathleen A. Leppig, MD
February 2, 2017
Family Network

• Providing information to family members at risk for their own health care
• Sharing information because of looking for support
  – Correlated with the severity of disease?
• Barriers of sharing genetic information
  – Family dynamics
  – HIPAA vs. Duty to Warn
Three families from eMERGEII with PGRNSeq actionable variants

Molecular Genetics & Genomic Medicine

Building a family network from genetic testing

Kathleen A. Leppig,1,2 Heidi A. Thiese,1, David Carrel,2, David R. Crosslin,4, Michael O. Dorschner,2, Adam S. Gordon,3, Andrea Hartzler,1, James Ralston,3, Aaron Scrol,3, Eric B. Larson,4 & Gail P. Jarvik,5

1Genetic Services, Group Health Cooperative, Seattle, WA 98112, USA
2Department of Pathology, University of Washington, Seattle, WA 98195, USA
3Group Health Research Institute, Group Health Cooperative, Seattle, WA 98101, USA
4Department of Biomedical Informatics and Medical Education, University of Washington, Seattle, WA 98195, USA
5Department of Medicine (Medical Genetics) and Genomic Sciences, University of Washington, Seattle, WA 98195, USA

Three families from eMERGEII with PGRNSeq actionable variants

Family 1
Brugada/sudden death
German, English, Dutch

Family 2
RYR1 c.1840 C>T p.Cys614Arg
Malignant hyperthermia
Irish, German, English, French

Family 3
Familial hypercholesterolemia
Ashkenazi Jewish, Russia, Poland, Austria
Specific Aim: To explore the feasibility of health systems-led identification and communication with family members of eMERGE participants

- Patients to be included are members of GHC/KPW and enrolled in eMERGE
- GHC/KPW will be returning pathogenic, likely pathogenic, VUS for CRC, and negative results to eMERGE participants
- GHC/KPW is an integrated healthcare system
To explore social, ethical and legal feasibility:

We will conduct semi-structured interviews with approximately 20 eMERGE participants before results have returned with:

• Topics:
  – Family definition
  – Preferred role (if any) in GHC/KPW for sharing actionable results with likely affected family members who are also GHC/KPW members
    • GHC/KPW duty to contact relatives directly with the proband’s consent
    • Use of EMR for sharing information, particularly between providers
    • Special considerations for minors
      – Hypothetical scenarios: clinical vignettes (CRC and Marfan syndrome) followed by questionnaire
Building a Family Structure

• We will also collect contact information for relatives identified by each participant
  • We will attempt to identify relatives based on first name/last name/DOB provided by eMERGE participant
  • We will NOT access relatives’ EMR or contact relatives directly

Currently questionnaire and vignettes are at IRB for approval
Legal Ramifications

• In contact with GHC/KPW legal consultants regarding the process within our institution

• Consultation with Bob Wildin, MD at NHGRI regarding Family History Tool

• In Process: Assembling a team of legal experts from the eMERGE sites to address issues including duty to warn vs. HIPAA
Ongoing Work

• Family Talk trial to improve CRC genetic results communication headed by Deb Bowen.
• Development of DOT phrases to be used in all eMERGE return of results appointments for consistent collection of information
• Exploration of family network development in other patient populations with a genetic diagnosis
• Development of eMERGE wide Family Network project, in conjunction with ROR
eMERGE KPW/KWHRI/UW

Family Network Supplement Team at Kaiser Washington Health and U. Washington:
PI: Nora Henrikson, PhD, MPH
Gail Jarvik, MD, PhD; multi-PI
S. Malia Fullerton, PhD
Kathleen Leppig, MD
Aaron Scrol, MS

The rest of the Seattle team:
Kaiser Washington Health (formerly Group Health) Research Institute
Eric Larson, MD MPH; multi-PI
David Carrell, PhD
Jane Grafton, MS
Andrea Hartzler, PhD
James Ralston, MD MPH

University of Washington
David Crosslin, PhD
Michael Dorschner, PhD
Adam Gordon, PhD
David Veenstra, Pharm D, PhD
Elisabeth Rosenthal, PhD
Melody Palmer, PhD
Parimala Devi, MS

• Funded by a supplement to the Electronic Medical Records and Genomics (eMERGE) Network, Phase III (NHGRI U01 HG008657)