

Familial Implications of Returning Genome Results

eMERGE III Subgroup

ROR/ELSI

Outcomes

Specific Aim 1:

- Explore attitudes of participants by convening focus groups and/or qualitative interviews.
- Participant perceptions of:
 - importance of sharing information
 - importance of targeted education for family members
 - barriers to effective communication with family
 - preferences/suggestions for methods or strategies to contact relatives

Specific Aim 2:

- Conduct surveys, standardized for many components, across sites to assess family sharing/communication activities.
 - Some sites doing baseline pre-results disclosure survey
 - All sites will include small number of consistent items in a post-results survey.
 - Some sites will survey immediately after return of results
 - Some sites will utilize a more in-depth survey to supplement the items asked in the general survey.

Specific Aim 3:

- Collect and collate points to inform system-level guidance for policy making or best practice development pertaining to family communication of positive variants in actionable genes.
 - Collect site specific activities currently planned to promote family communication
 - Assess factors that lead to variation in methods of communications including materials, approaches and measures of success
 - Test various methods of contact

Geisinger patient experience interviews

DOMAINS

- Initial experience with result
- Medical follow-up
- Communication with family and friends
- Understanding of the results and resource seeking
- ROR procedures
- Psychosocial reactions to result
- Financial implications of result
- Satisfaction with participation in MyCode

Common themes (very preliminary!)

- All participants shared with some family members
- There were some family members that participants chose NOT to share with
 - Family members too old
 - Family members too young
 - Family member discord
 - Family members would not be interested
- Most used letter provided by ROR team, but some called to tell by phone or in person
- Thought result applied only to women (BRCA)

Site Specific Survey DOMAINS

- Intent to share with family members
- Family Communication
 - Conflict
 - Satisfaction
- Empowerment
- Information Sharing
- Language/Literacy
- Utilization
- Life/Health Insurance Issues

HIPAA, Privacy and ROR Familial Communication

- Group Health initiated conversation with legal counsel re: return of results to family members who receive care within healthcare system
- Hope to convene a group to more broadly address these issues
 - S. Malia Fullerton/Nora Henrikson Group Health
 - Robyn Fossey Mayo
 - Jen Wagner Geisinger
 - Bob Wildrin (not confirmed) NIH

Outcomes survey: Proband

- For each participant after an actionable variant has been returned:

1. Mother Alive/Available Y/N Shared Y/N
2. Father Alive/Available Y/N Shared Y/N
3. Sisters # Alive _____ # Available for contact
 - a) Sister 1 shared result Y/N
 - b) Sister 2 shared result Y/N
 - c) Sister 3 shared result Y/N
4. Brothers # Alive _____ # Available for contact
 - a) Brother 1 shared result Y/N
5. Children #Alive _____ # Available for contact
 - a) Child 1

Outcomes System related

2. If possible estimate how many of the relatives are cared for within your healthcare system
3. Number of at-risk relatives seen and counseled as a result of the return of results for this proband
 - By 6 months
 - By 12 months
4. Provide the number of familial mutation tests sent related to this proband
 - # relatives per proband with positive mutation *high utilization*
 - # relatives per proband with negative mutation *usual care*
5. Reach of cascade screening

• 1 st degree relatives	#Alive	#Informed	#Tested
• 2 nd degree relatives	#Alive	#Informed	#Tested
• 3 rd degree relatives	#Alive	#Informed	#Tested