Incorporating genetic data on PCOR studies: building a road map for stakeholder engagement

PCORI-EAIN 2419 Presentation #5

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Outline

- Overview of the previous PowerPoint presentation (PPT#4)
- Results of the PPT#4 evaluation survey
- Overview of the ethical issues and genetic testing (NIH website)
- Overview of the ACMG and ASHG guidelines on genetic testing
- Stakeholders' (patients, physicians, researchers) comments on using genetic information in health care decision making
  - Comments gathered from reports/publications
  - Feedback collected from our study participants (CAB)
- Roadmap: a tentative draft
- Project website: update
- What is next? Collecting participants' feedback!

Introduction

- Our overall goals are to:
  - (1) Assess IF it is possible to use genetic information to improve patient health outcomes and
  - (2) If YES, then HOW can this be achieved?

- In the PPT#4, we reviewed:
  - (1) examples of existing resources that include patient genetic data and clinical information (PCORnet and eMERGE),
  - (2) technologies facilitating research, and
  - (3) Electronic Medical Record systems

- In the PPT#5:
  - The results of the PPT#4 evaluation survey will be shared.
  - The following items will be reviewed:
    - Potential ethical issues as well as recommendations for interpretation of genetic testing
    - Potential barriers and needs (gathered from publications and our study participants)
    - A suggested Roadmap to promote incorporating genetic information in patient outcomes studies

PPT#4 Evaluation Survey-Results

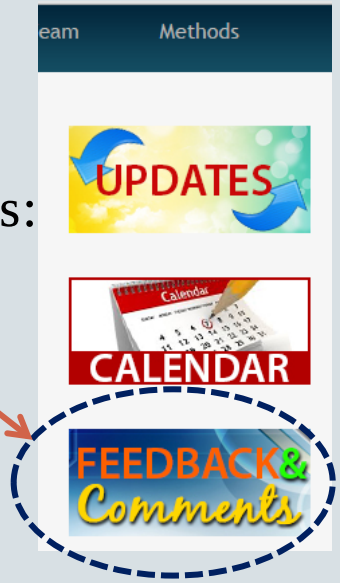
32/33 members responded =97%

For the following statements, please indicate to what extent you disagree or agree with "0" indicating "strongly disagree" and "10" indicating "strongly agree". There are no right or wrong answers, it is important that the responses reflect your individual experience and opinions."The information in the PPT#4 presentation was provided in an easy to understand manner"

1. The information in the PPT#4 presentation was provided in an easy to understand manner	**Rating Average**	**Response Count**
	8.66	**32**
	answered question	**32**
2. Overview of existing resources (PCORnet, eMERGE) was informative and provided in an easy to understand manner	**Rating Average**	**Response Count**
	8.91	**32**
	answered question	**32**
3. Examples of technologies facilitating research (Prometheus, LLC, Pathfinder, Inc.) were informative and provided in an easy to understand manner	**Rating Average**	**Response Count**
	8.59	**32**
	answered question	**32**
4. Examples of Electronic Medical Record systems were informative and provided in an easy to understand manner	**Rating Average**	**Response Count**
	9.16	**32**
	answered question	**32**
5. Would you like to schedule a personal meeting and/or call with the PI for further clarification of materials discussed in the PPT #4 presentation?	**Response Percent**	**Response Count**
Yes, I would like to schedule a meeting	**0%**	**0**
No, not at this point	**100%**	**32**
	answered question	**32**

Feedback & Comments

- To see all Feedback & Comments received from our participants please visit the project website:
  - http://genetics-outcomes.net/EAIN/feedback.html



- Feedback & Comments are listed by the following categories:
  - Meeting#1 presentation (PPT#1)
  - Meeting#2 presentation (PPT#2)
  - Meeting#3 presentation (PPT#3)
  - Meeting#4 presentation (PPT#4)
  - Project website
  - Memorandum of Agreement (MOA)
  - Suggested structure for the meetings
  - Selection of topics to be covered during the course of this project
  - Overall comments/suggestions

PPT#4 Group Discussion

Our PPT#4 group discussion was held on 6/21/2016 with a number of our local members (n=8).
Group discussion notes are posted on the website.

PPT#4 group discussion

Meeting 3 agenda (took place via email on May 9, 2016)

- Overview of the previous PowerPoint (PPT) presentations
- Results of the PPT#2 evaluation survey
- Additional patient/parent personal stories
- Examples of published epidemiological/patient outcomes studies (information
- Examples of genetic research done by our CAB members (Scientists outcomes
- Project website-Update
- What is next?

Meeting 3-PowerPoint Presentation (PPT#3)
Meeting 3-Group Discussion
Meeting 3-Evaluation Survey

If you are interested to participate in upcoming group discussions, please let us know!

Overview of the ethical issues and genetic testing

- In this section, we are reviewing some:
  - Potential ethical issues raised in relation to genetic testing &
  - Suggested guidelines for addressing the ethical issues
- The purpose of sharing these materials is to familiarize our study participants with some potential barriers that need to be considered WHEN designing research studies that include genetic information, which is the ultimate goal of our Engagement project.

- Please note that:
  - These materials were selected for the educational purpose only, and other resources may exist.
  - For transparency, we used either exact original texts from these resources or made minor modifications.

NIH guidelines: using genetic data in research

- **Source:** NIH website
- **Reference:** https://www.genome.gov/27561533/human-subjects-research-in-genomics/

- The use of human subjects in research should follow approved guidelines and regulations to ensure protecting the privacy of subjects and to maintain confidentiality.

- Here is a list of the key topics included in the NIH guidelines about using genetic data in research:
  1. Implications for Relatives
  2. Identifiable Populations
  3. The return of individual research results (IRRs) & incidental findings (IFs)

1. Implications for Relatives (NIH website)

- While genomic research may reveal new information about the research participant's health, the heritable nature of genetic information raises implications for the research participant's relatives.
- Information about family members not involved in the study may be indirectly obtained through the research participant.
- Furthermore, genomic research using family pedigrees can trace disease history, and may reveal family members that are carriers of a disease or will be affected themselves.
- These indirect results pose an ethical conflict between a possible duty to warn family members of research participants and the protection of research participant privacy.
  - The Guidebook from the NIH Office of Protection from Research Risks provides guidance on addressing these issues.

2. Identifiable Populations (NIH website)

- Identifiable populations, which include specific racial or ethnic groups, geographically-defined communities, and members of ultra-rare disease groups, present particular concerns with regard to privacy, stigmatization, and discrimination.
- For example, for some communities, close family relationships may make it nearly impossible to protect participants' privacy.
- Furthermore, genetic information could raise questions around ancestry and family-ties that may disrupt the community structure.
  - Some communities may require the research to obtain community approval before seeking consent from potential participants.
  - For American Indian and Alaska Native communities, for example, the National Congress of American Indians Policy Research Center, in conjunction with NIH, has developed a resource to discuss questions and provide information about how some tribes are thinking about genetics research.

3. The return of individual research results & incidental findings (NIH website)

- The return of individual research results (IRRs) and incidental findings (IFs) from genomics research is an issue of interest among researchers, ethicists, funders of biomedical research, policy makers, research participants, and others.
- At issue is that, when conducting clinical research studies, scientists may discover new health-related information about volunteers who have chosen to participate in the studies.
- Currently, IRRs and IFs from genomic research are not commonly returned to study participants or anyone else outside of the study team (e.g., family members, personal physicians). However, arguments have been raised both for and against sharing this information.
- This raises the question of when and how it is appropriate for the scientists to share such research findings.
  - NIH is currently funding research to examine these questions and inform future policy.

ACMG guidelines: clinical utility of genetic services

- **Source:** Publications
- **References:**
  - Richards et al. GenMed 2015
  - ACMG Board of Directors. GenMed 2015

- The American College of Medical Genetics and Genomics (ACMG) represents the medical genetics professional community. The ACMG routinely develops standards and guidelines primarily as an educational resource for clinical laboratory geneticists to help them provide quality clinical laboratory services.

- A summary of the recent ACMG guidelines for the interpretation of genetic variants is provided in the next 3 slides.

ACMG guidelines (Cont.)

- The ACMG recommends:
  - Specific standard terminology: ["pathogenic," "likely pathogenic," "uncertain significance," "likely benign", and "benign"] should be used to describe variants identified in genes that cause single gene disorders.
  - Clinical molecular genetic testing should be performed in a Clinical Laboratory Improvement Amendments–approved laboratory, with results interpreted by a board-certified clinical molecular geneticist or molecular genetic pathologist or the equivalent.
  - The clinical utility of genetic testing and services should take into account effects on diagnostic or therapeutic management, implications for prognosis, health and psychological benefits to patients and their relatives, and economic impact on health-care systems.

ACMG guidelines (Cont.)

Clinical Utility for Individual Patients

- As increased numbers of individuals are diagnosed with specific genetic disorders, information will be obtained that will help predict future complications and risks, tailor medical interventions, and lead to the development of new specific therapies and management strategies. Examples include:
  - Patients with complex and often poorly understood clinical disorders such as autism spectrum disorders and intellectual disability
  - Patients with rare disorders
  - Patients with genetic conditions such that definitive and specific guidance regarding prognosis and medical management is not yet available

ACMG guidelines (Cont.)

Clinical Utility for Families and Society

- A definitive diagnosis of a genetic disease may provide clinical utility for families and society. Examples include:
  - Enables at-risk family members to obtain testing to determine whether they carry a causative mutation, offering the possibility for early intervention.
  - Enables specific and informed reproductive decision-making and family planning.
  - Brings resolution to the costly and wasteful diagnostic odyssey.
  - Enables involvement in disease support groups and other types of social support for families.
  - A specific diagnosis is commonly required for patients to be eligible to participate in clinical trials.

ASHG recommendations

- **Source:** Publications
- **Reference:**
  - Botkin et al. AJHG 2015

- The American Society of Human Genetics (ASHG) was established in 1948 to provide leadership in research, education and service in human genetics. In 2015, ASHG published a statement representing its current opinion on the ethical, legal, and social issues concerning genetic testing in children and adolescents. This statement includes recommendations relevant to families, clinicians, and investigators.

- A summary of the ASHG guidelines is provided in the next 6 slides.

ASHG recommendations (Cont.)

Predictive Genetic Testing in High-Risk Families

- Unless there is a clinical intervention appropriate in childhood, parents should be encouraged to defer predictive or pre-dispositional testing for adult-onset conditions until adulthood or at least until the child is an older adolescent who can participate in decision making in a relatively mature manner.
- Adolescents should be encouraged to defer predictive or pre-dispositional testing for adult-onset conditions until adulthood because of the complexity of the potential impact of the information at formative life stages.
- Providers should offer to explore the reasons why parents or adolescents are interested in predictive or pre-dispositional testing for adult-onset conditions, such as breast cancer, ovarian cancer, and Huntington disease.

ASHG recommendations (Cont.)

Direct-to-Consumer Testing

- Direct-to-consumer genetic testing (DTC GT) refers to genetic testing that bypasses the involvement of health-care providers and is sold directly to consumers. Several concerns have been raised about DTC GT, and they include the lack of high-quality pre-test and post-test counseling and clinical interpretation of test results, the lack of adequate validation of some tests, and the testing of children for adult-onset conditions.

- The ASHG recommends that DTC GT:
  - be discouraged in children until such a time when companies that provide DTC GT can assure quality, accuracy, and validity of their testing and assure that there is adequate pre- and post-testing counseling.
  - in children be performed with the appropriate informed permission from a parent or legal guardian and the assent of the child when appropriate.
  - not be performed in children for genetic conditions that have onset in adulthood or require surveillance beginning in adulthood.

ASHG recommendations (Cont.)

Pharmacogenomic Testing

- ASHG recommends that when there is a clear evidence base in the literature for clinical utility, pharmacogenetic testing in children might be appropriate.
- ASHG recommends additional evaluation of pharmacogenetic testing opportunities in the pediatric population in order to better demonstrate the utility and limitations of this form of testing.

Adoption

In the US, approximately 2% of children are adopted, and many children are living in foster care.

- The ASHG recommends that both children awaiting adoption and adopted children be given the same consideration in genetic testing as children living with their biological parents.

ASHG recommendations (Cont.)

Newborn Screening

- Newborn screening (NBS) is one of the most effective public-health programs of the last century. The ASHG strongly supports NBS programs and encourages genetic professionals to support NBS in their communication with patients, colleagues, and policy makers.
- NBS is conducted by state-based public-health programs in the US.
  - The ASHG supports conducting outcomes research on NBS and developing infrastructures for conducting outcomes research on these rare conditions. Such infrastructures would support the ability to assess outcomes and to conduct controlled trials of therapeutic options and evaluate support systems required for affected children and their families.
  - The ASHG recommends additional research for improving the quality, delivery, and effectiveness of parental, public, and professional education regarding NBS.

ASHG recommendations (Cont.)

Record and Communication Issues

- The ASHG recommends that:
  - providers of pediatric genetic testing have appropriate training and expertise in the interpretation and communication of genetic information.
  - diagnostic laboratories develop reports that are detailed and accurate but also facilitate comprehension by providers.
  - genetic testing in children should include a long-term communication plan for all results, including consideration of who should be involved in the communication of information and the staging of information sharing on the basis of age, maturity, and capacity to understand.
  - standards be developed for permanent storage of genetic data in electronic health records or other secure electronic systems to facilitate the provision of genetic information in patient portals.
  - the development of mechanisms for sharing family history and genetic results with family members.
  - the development of uniform guidelines to standardize medical-record capabilities and management of interpreted results and raw genetic sequence data.
  - developing novel models for molecular laboratory and interpretive services on the basis of prospects for the re-analysis of genetic information over time.

ASHG recommendations (Cont.)

Professional Education

- If health-care providers are to adhere successfully to the recommendations in this report, they must have appropriate knowledge and skills related to genetic and genomic testing, interpretation of test results, communication of results to patients and families, and basic genetic counseling.
- In addition, the health-care system will require adequate numbers of trained medical geneticists and genetic counselors to assist in the role of specialty testing and interpretation of results.
- With the expected expansion of genetic and genomic testing, all health-care providers will need (1) educational programs that target relevant scientific, clinical, ethical, legal, and social topics and (2) support systems that address structural and systemic barriers to the integration of genetic medicine into clinical practice.

Stakeholders' comments

- In this section, we will review comments, reflecting barriers and needs identified by stakeholders, related to using genetic data in research, which we gathered from:

1. Publications:
   - Study participants
   - Physicians

2. Our study participants:
   - CAB members

Stakeholders' comments (Publications):
Participant attitudes toward genetic testing

- **Source:** Publications
- **Reference:**
  - Lemke et al. Public Health Genomics 2010

- Here we summarize findings from one of the first studies conducted to assess public and biorepository participant views toward
  - genetic research participation and
  - data sharing

- For this study a focus group discussion guide consisting of 8 questions (listed in the next slide) were used.

Questions assessed (n=8)

1. What comes to mind when you think of genetic research data?
2. What might be some reasons that a person would participate in genetic research?
3. What might be some reasons that a person would not participate in genetic research?
4. How do you feel about participating in a genetic research study?
5. What kinds of information would you need to know before participating in a study to store and share your genetic research information?
6. If you agreed to participate in a study in which your genetic research information was stored in a database, what are your thoughts about sharing this data with other investigators?
7. How well do you think the privacy of individual genetic re-search data is protected?
8. What do you think the role of institutions and the government should be in protecting the privacy of those who participate in genetic research?

Responses: theme 1

Five major themes were identified in the focus group data:

1. A wide spectrum of understanding of genetic research
   - Lack of clear understanding about genetic research.
   - Potential for altering human physical characteristics.
   - Opportunity for misuse of genetic research information such as ‘playing god’, or ‘genetic discrimination’ and ‘Big Brother’.
   - Potential for findings not to be fully reported to public.

Responses: theme 2

2. Weighing Pros and Cons of participation in genetic research

Reasons to participate in genetic research:

- the desire "to help"
- having a family member with a genetic condition
- interest in genetic research and wanting to be a part of scientific achievement
- out of curiosity for the science
- bettering society by curing disease
- potential cost savings to society
- ease of participating.

Reasons for *not* participating in genetic research:

- lack of information about and understanding of genetic research
- fear associated with genetic research
- fear of needles related to providing a sample
- fear of discovering a familial disposition toward a disease
- knowledge of genetic diseases in family could influence their decision to conceive
- concern about genetic discrimination, due to the sharing of genetic information as a result of participation in genetic research
- possible discrimination by insurance companies, the government, the health care system, and employers.

Responses: theme 3

3. Influences to participation: credibility, trust, and research integrity matter
   - Reputation and trust are key factors. It is important for participants to know what organization/agency had oversight responsibilities for genetic research data as well as to clearly understand the research goal and motivation. Insurance companies and pharmaceutical companies were noted as generally distrustful by participants, as well as distrust of the government as an oversight body for genetic research data.
   - Separation of research and medicine from profit would be more trusting.

Responses: theme 4

4. Participant requirements in order to feel comfortable having their data shared:
   - similar study purpose
   - adequate security and privacy checks
   - Re-contact to consent to participate in another researcher's study
   - trust in the institution that stored data
   - great detail describing how data will be shared and under what specific circumstances
   - clear explanations of circumstances constituting breach of confidentiality
   - information on specific penalties imposed on researchers who misuse data sharing.

Responses: theme 5

5. More information and education about genetic research needed
   - It is important to provide complete disclosure about genetic research, including to learn more about research progress, to reduce fears and build trust.
   - Providing accurate information on genetic research because of concern about how the media can distort scientific information.
   - Targeting various groups such as perceived disengaged groups (low-income, minority groups), young people, schools, neighborhoods, and disease support groups.
   - Targeting communities where a particular genetic condition occurs more frequently than in the general population.
   - Suggested methods of education: nightly news is a good method of reaching people; through internet, conducting interviews and focus groups with specific disease groups and communities, and educating family members – a primary source for health-related information; separate education approaches for diverse communities, respecting their beliefs; focus group experience itself is a helpful educational experience.

Participant attitudes toward genetic testing (Cont.)

- **Source:** Publications
- **Reference:**
  - Jamal et al. European J of Human Genetics 2014

- In a more recent study, research participants' attitudes towards the confidentiality of genomic sequence information were assessed. Semi-structured phone interviews with participants in research protocols using genomic sequencing were conducted to assess different conceptions of confidentiality, why it is important, and why participants in genomic research care about how their information is used.

- A summary of their findings is described in the next slide.

Responses: Research Participants

- Research participants value confidentiality as a form of control over information about themselves.
- To the individuals interviewed, control was valued as a safeguard against discrimination in a climate of uncertainty about future uses of individual genome data.
- Attitudes towards data sharing were related to the goals of research and details of participants' personal lives.
- Expectations of confidentiality, trust in researchers, and a desire to advance science were common reasons for willingness to share identifiable data with investigators.
- Nearly, all participants were comfortable sharing personal data that had been de-identified.
  - These findings suggest that views about confidentiality and data sharing are highly nuanced and are related to the perceived benefits of joining a research study.

Stakeholders' comments (Publications):
Primary Care Physicians attitudes

- **Source:** Publications
- **Reference:**
  - Haga et al. J General Internal Medicine 2010

- Physicians' attitude and knowledge about genetic information are another important factor in incorporating such data into their clinical practice.
- A group of primary care physicians was surveyed to assess their familiarity with genetic testing, intended use of results, likelihood of future testing, perceived benefits and risks, and their preferences for education/training in genetic testing.
- They also assessed experience with genetic testing, as well as perceived adequacy of genetics training.

- A summary of their findings is provided in the next 4 slides.

Responses: Primary Care Physicians (theme 1)

1. Knowledge of Traditional Genetic Testing & Genomic Risk Testing
   - 45% of respondents strongly or somewhat strongly agreed with the statement that they felt well-informed about genetic testing
   - 52% strongly or somewhat strongly agreed with the statement that they would feel comfortable ordering genetic testing for disease susceptibility.
   - 49% did not believe that their genetics training was adequate.
   - 90% had heard of genomic risk profiling offered by genetic testing companies such as 23andMe.
   - Each additional year since graduation was associated with an 11% reduction in the odds of having heard of genomic risk profiling

Responses: Primary Care Physicians (theme 2)

2. Factors Impacting Use of Traditional Genetic Testing & Personal Genomic Risk Profiling

- Factors considered by respondents who ordered genomic risk profiling for their patients included:
  - patient request or interest (80%),
  - clinical utility (68%),
  - motivating adoption of preventive behaviors (68%),
  - family history (64%), and
  - the cost of testing (58%).

Responses: Primary Care Physicians (theme 3)

3. Preferred Educational Resources

- Respondents preferred similar methods to learn about genomic risk profiling:
  - continuing medical education (CME) courses (69%),
  - medical journals (57%),
  - professional medical meetings (53%), and
  - educational programs offered by testing companies (47%)
- When asked about the best way to educate physicians about genomic risk profiling, respondents most frequently endorsed
  - in-person CME (38%)
  - followed by long-distance CME (10%),
  - grand rounds and other in-house seminars (10%), and
  - educational materials from testing laboratories (8%)

Responses: Primary Care Physicians (Overall)

- Overall, this study demonstrates that
  - "respondent familiarity was a key predictor of physician ordering behavior and clinical utility was a primary concern for genomic risk profiling. Educational and interpretive support may enhance uptake of genomic risk profiling. The results suggest that PCPs who feel well-informed about genomic risk testing are willing to include this type of information in a patient's risk assessment, even while acknowledging the uncertain clinical utility."

EAIN 2419 Study Participants Feedback (CAB)

- We reviewed feedback/comments received from our CAB members throughout the study.
- The general themes identified by our CAB members, reflecting barriers, facilitators and needs, are summarized in the next 3 slides.

- Note:
  - We continue to collect CAB feedback/comments
  - Please send us additional feedback/comments, if any, in the next survey and we will add them into the final study report.

Barriers (EAIN 2419-CAB)

1. Obtaining Meaningful Input from Every Participant (example)
   - I don't understand how I will provide better input back to the team.

2. Including Genetic Information in Clinical Practice (example)
   - I frequently have parents who want to do genetic testing but have trouble obtaining prior authorization from insurance companies to cover testing which is often quite expensive.
   - Currently, there is insufficient number of genetic counselors, particularly in some regions, which results in long wait time for a counseling session for patients.

3. Insufficient Physicians' Training about Genetic Testing (example)
   - Physicians have little if any training on how to identify appropriate testing and how to identify resources to facilitate ordering genetic testing.

4. Insufficient Technical Knowledge Level of non-Scientific Study Participants (example)
   - Some of the provided materials are very informative, but not as easy to understand.
   - For those of us who lack a scientific background, understanding genes and genetic structure is a daunting task.

5. Technical Challenges (example)
   - Patients' data (clinical and genetic) is being collected / stored based on different platforms. The lack of compatibility between different platforms defies data integration and sharing between health care providers.

Facilitators (EAIN 2419-CAB)

1. Meaningful Role for Every Participant
   - Collaborative nature of the project is an attractive element for participants.
   - Sharing patient stories and hearing their concerns are helpful in developing a list of questions about specific diseases.
   - Hearing personal experiences from scientist who also have a family member affected by a genetic condition provides a unique perspective for other participants.

2. Educational Aspects for Participants
   - The patient stories were excellent and illustrated in a concrete way exactly what people. hope to get - and often don't get - from genetic testing and precision medicine today.
   - Learning about PCORnet was helpful, I was unaware of it.
   - Learning about eMERGE was helpful, I was unaware of it.
   - Learning about the resources developed by companies was helpful, I was unaware of them.

3. Stimulating Thoughts for Participants
   - Learning about this study made me think about adoptees, like my own adopted kids, who have little data on their birth family. It reminded me how important this is going to be to find ways to include them in genetic studies.
   - I found the information very interesting - especially in ways that genetic information can be used to improve care, teaching and patient/family knowledge and adherence.
   - Learning about PCORnet encouraged me to explore collaboration opportunity.

Needs (EAIN 2419-CAB)

1. Elements Facilitating Study Participants' Contribution
   - Explaining research data in a non-technical language
   - Designing studies by taking into consideration participants' time-constrains
   - Providing a glossary of terminologies related to the project
   - Providing a lay abstract for technical topics
   - Providing educational materials about the disease condition and genetic risk factors

Needs (EAIN 2419-CAB)

2. Lessons Learned (Future Directions/Recommendations)

- Many useful resources containing genetic/clinical data have already been developed but study participants, including scientific/clinical members, may not be aware of them.
- Providing a framework for how to access existing genetic data would facilitate incorporating it in health outcomes research.
- Setting reasonable expectations for study participants is needed to maintain each member's meaningful contribution without slowing down the overall research process.
- Study participants' motivation and time devotion are essential in reviewing the provided educational materials.
- Reviewing actual research examples would facilitate better understanding of the topic, particularly, for non-technical study participants.
- Visual aids (e.g., videos, webinars, and illustrations) are great educational tools and help with explaining complex contexts.
- Focusing on one disease would be helpful for:
  - (1) developing practical example(s) for incorporating genetic data,
  - (2) identifying more specific barriers and needs, as well as
  - (3) implementing results among the research community

Roadmap (EAIN 2419)

- Based on the feedback collected from our CAB members and lessons learned from the EAIN 2419 project, we drafted a tentative Roadmap (attached PDF).
- The purpose of the Roadmap is to show HOW experiences learned from this engagement project may be used to promote incorporating genetic information in patient outcomes studies.
- Please review it and give us your feedback via PPT#5 survey.

Project Website-Update

- The project website has been updated, including addition of the "Patient Stories" section and Agendas for the remaining presentations (under Methods tab).
- Please visit the website for more information. http://genetics-outcomes.net/EAIN

Methods

Develop Partnerships [Community Advisory Board (CAB) Formation]. Using the principles of community engaged research; our project team will develop and formalize relationships with twenty two stakeholders who will be engaged in every aspects of the project development process. These stakeholders will comprise the members of a formalized genetic concept Community Advisory Board (CAB). Seven of the CAB members have already been recruited by the research team members and engaged from the inception of the project through the application preparation and submission. Additional fifteen CAB members have been recruited at the beginning of the project by the research team members.

Members of the Research Team

Members of the Community Advisory Board-CAB

CAB Meetings. The CAB will convene six meetings at Children's Mercy Hospital or other locations suggested by the stakeholders during the twelve month project period and will work closely with the research team to develop the strategic direction of this engagement/ educational program. CAB members will be strongly encouraged to attend a minimum of 80% of all meetings in person, by telephone or video conference. The specific tasks and activities of each meeting are described below:

Meeting's Agenda (Tentative)

Meeting 1 (click for agenda)

Meeting 2 (click for agenda)

Meeting 3 (click for agenda)

Meeting 4 (click for agenda)

Meeting 5 (click for agenda)

Meeting 6 (click for agenda)

Patient Stories

In this section a number of our patient/parent representatives share their personal stories and why they are interested in genetic information. Some stories have more details to help us grasp the level of health complexity that some patients are dealing with, a long journey they have gone through to find an answer, and how they expect genetic information to help them with better managing health outcomes.

Please note that our Engagement project is not focused on any specific disease. We would be happy to add more patient stories, please feel free to send us your story!

Patient Story 1: Seth Bittker
Patient story 2: Kristen Worden
Patient Story 3: DeeJo Miller
Patient Story 4: Sheryl Chadwick
Patient story 5: Mary Anne Hammond

The following videos provide more information on Precision Medicine Initiative and patient stories.

Personal Genomics: Use of personal genomics in predictive and precision medicine
https://www.youtube.com/watch?v=pSzk-yf6BVU (length: 54 min)

What Is Next? Collecting Participants' Feedback!

- **Survey:** PPT#5 evaluation survey will be sent out to get an overall assessment from all participants about this presentation and the Road Map. Please respond to the survey at your earliest convenience.

- All collected feedback, comments, questions and responses will be summarized and posted on the project website
  - (http://genetics-outcomes.net/EAIN/) and
  - shared in our next/final presentation (PPT #6)