Panel 9: Patient-facing information tools, counseling/consent, reporting results to patients

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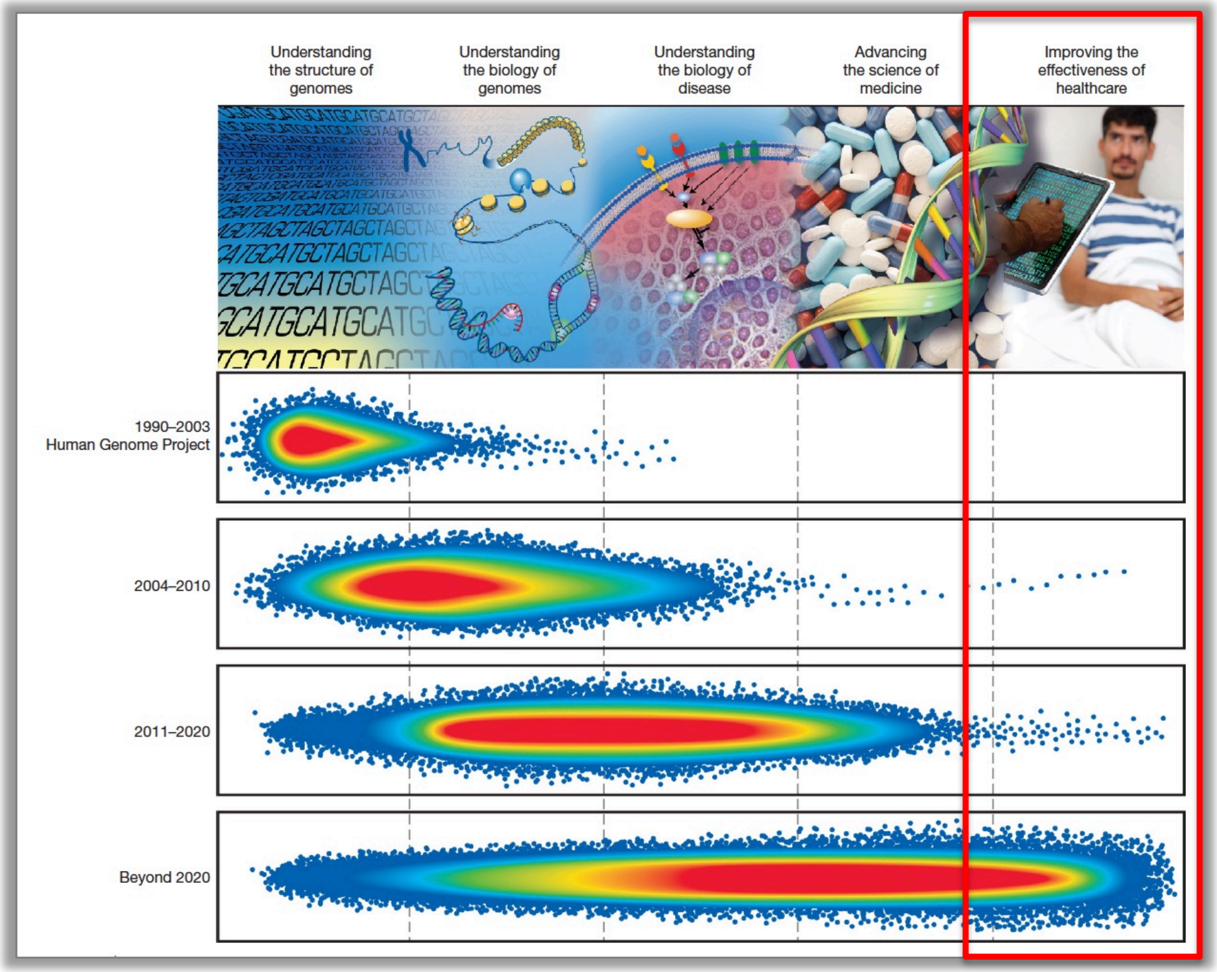
NHGRI

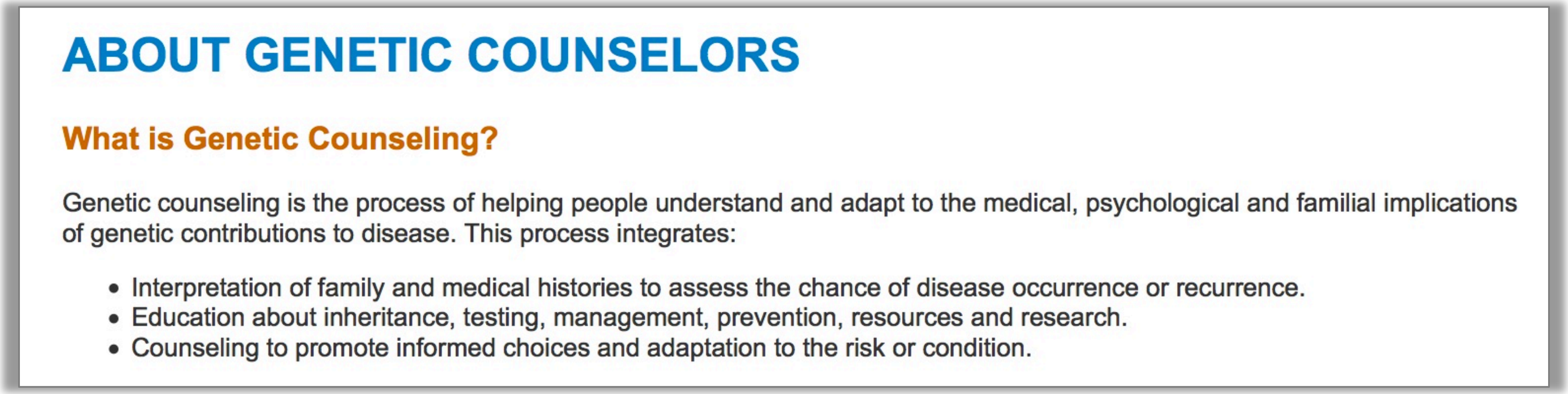
Implementation requires effective clinician-patient communication



Nature 470:204, 2011

Patient-facing tools needed for education, risk assessment & decision support

National Society of
Genetic
Counselors



ABOUT GENETIC COUNSELORS

What is Genetic Counseling?

Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates:

- Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
- Education about inheritance, testing, management, prevention, resources and research.
- Counseling to promote informed choices and adaptation to the risk or condition.

http://nsgc.org/p/cm/ld/fid=175

Challenges for genomic medicine

- Scale
  - workforce demands as genomics infiltrates medical practice
- Scope
  - breadth of potential findings
- Science/genetics literacy
  - clinicians and patients

Functions of patient-facing tools

	Pre-test	Post-test
Education	• Related to indication for test • Possibility of incidental findings	
Risk assessment	• Phenotype • Personal history • Family history	
Decision support	• Whether to test • Preferences for return of incidental/secondary results (?)	

Functions of patient-facing tools

	Pre-test	**Post-test**
Education	• Related to indication for test • Possibility of incidental findings	• Primary result • Secondary/incidental result • VUS • Negative test
Risk assessment	• Phenotype • Personal history • Family history	• Related to secondary/incidental findings
Decision support	• Whether to test • Preferences for return of incidental/secondary results (?)	• Diagnostic, therapeutic, or preventive interventions • Testing of family members

Needs for effective, safe & efficient implementation of genomic medicine

- Strategically designed, evidence-based suite of tools for patients
- Strategically designed, evidence-based suite of tools to support & extend clinicians, including
  - genetic specialists (geneticists, genetic counselors, etc)
  - non-genetic specialists
  - generalists

Google search = *daunting!*

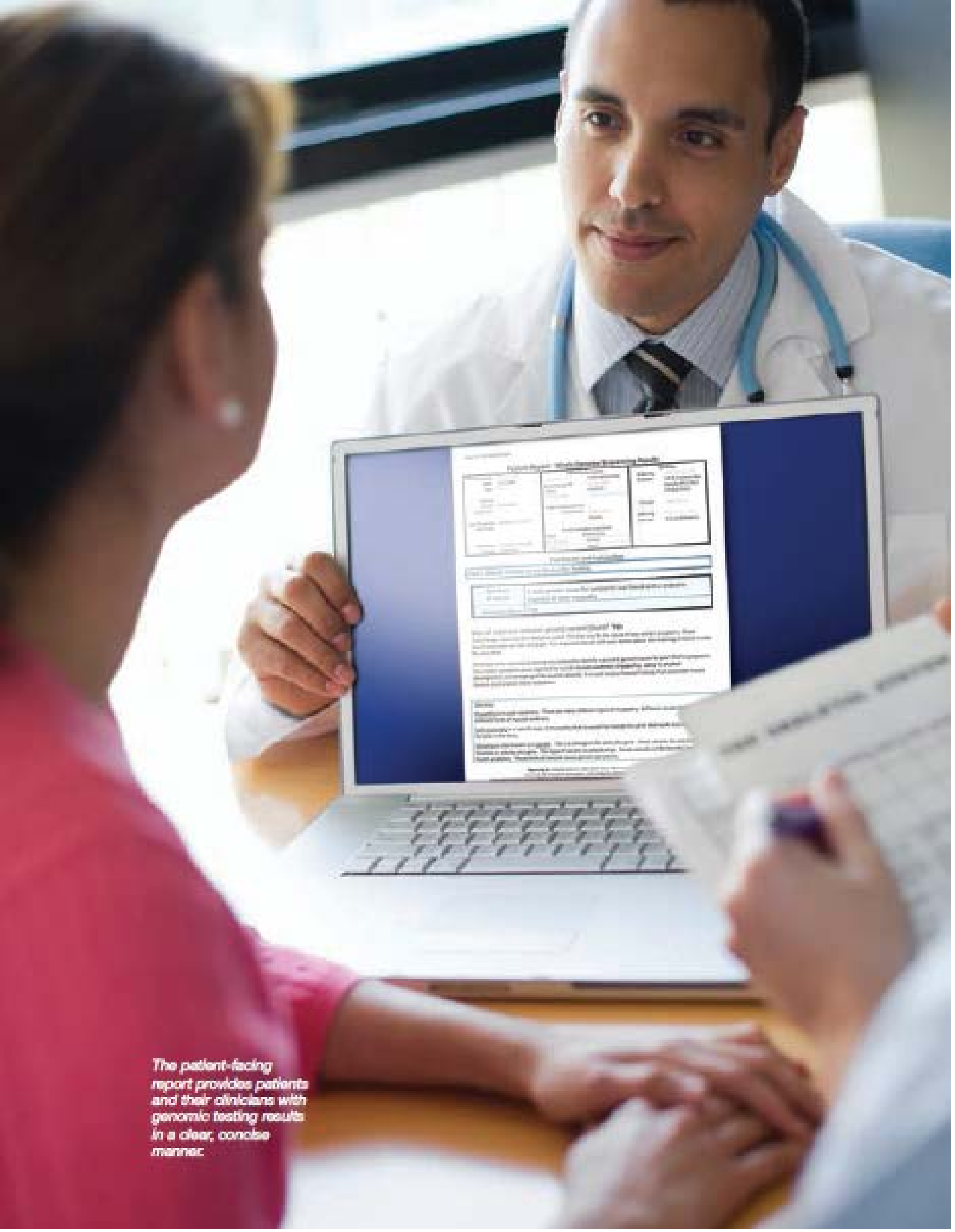
- Patient information about genome results
  - About 51,000,000
- Genome Information for patients
  - About 50,5000,000 results
- Patient information about genome sequencing
  - About 23,500,000 results
- Genome Sequencing for patients
  - About 12,200,000 results
- Genomic Medicine for patients
  - About 3,970,000 results

Patient-facing Resources

Patient Resource	NIH Program
Genetics Home Reference	ClinGen CSER UDN NHGRI
Cancer Genetics PDQ	CSER NHGRI
Genome Connect	ClinGen
Genetic Alliance	UDN
MEDLINEPlus	UDN
NORD/GARD	ClinGen UDN NHGRI
OrphaNet	UDN

Patient-facing results explanations

- www.labtestsonline.org
- www.MyResults.org
- www.yourgenome.org
- www.My46.org
  - *Enables individuals to manage their results from genetic testing, whether it is*
    - *a single result*
    - *exome sequencing*
    - *whole genome sequencing.*
  - *Offers Dynamic Consent*
  - *Allows researchers to study*
    - *how individuals select results that they want returned,*
    - *how they manage those results, and*
    - *how they use the information learned.*



The patient-facing report provides patients and their clinicians with genomic testing results in a clear, concise manner.

Developing genome results reports for families and providers

Presented by:
Janet L. Williams, MS, LGC
Marc S. Williams, MD, FACMG

Funded by PCORI :
Communication and
Dissemination Research

Patient report results: Overall themes

- Parents continually search for valid information and resources regarding child's condition
  - Prior reports do not meet this need
  - All parents expressed this report meets this need

- **Parents would use report to facilitate communication**
  - Described how would use it with family, friends, schools, doctors

- Parents have specific needs from genomic report
  - Understandable language
  - Logical flow
  - Visual appeal
  - Information on what to expect in the future (prognosis)
  - Recommendations for next steps
  - Multiple modes of delivery suggested: paper, computer, thumb drive

Clinical Sequencing Exploratory Research

- **Genetic Counseling**
  Chairs: Denise Lautenbach and Sarah Scollon

- **Mission**: Discuss site-specific experiences with issues related to genetic counseling. Work on publications and educational materials, and function as a sounding board to new groups.

Genetic counselors key

Genetic Alliance UK Patient Charter

- Patients value genetic counselling and are keen for the support of genetic counsellors before and after genome sequencing
  - Recommendation: All patients should be able to access a dedicated genetic counsellor before having their genome sequenced
  - Recommendation: More support should be given to the training of genetic counsellors as the need for their services increases

Seek patient collaboration

Genetic Alliance UK Patient Charter

- Patient Charter February 2015
- Patients welcome the sharing of their genomic data for research purposes
  - Engage with the patient community to develop accurate and comprehensive information on genome sequencing
  - Research studies and clinical care involving genome sequencing should be more closely integrated to reflect the patient experience

http://www.geneticalliance.org.uk/docs/patient-charter-genome-sequencing-what-do-patients-think.pdf

Models of consent in genomic research

- Elements that should be included in informed consent process
- Surveyed 241 genomics researchers
- Interviews of 28 researchers and 20 participants
- Conclude
  - Routine approaches to consent not effective
  - Requires innovative approaches to ensure decisions are informed and meaningful

Hastings Cent Rep. 2014 Jul-Aug;44(4):22-32. doi: 10.1002/hast.328. Epub 2014 Jun 11.

Informed Consent Genetic Alliance UK

- Dynamic consent
  - possible for individuals to continually adjust and re-adjust their preferences
  - allowing the individual to reconsider their preferences whenever they choose or
  - could be solicited after certain periods of time
  - a combination of the two.

PCORI's Broad and Complex Mandate

"The purpose of the Institute is to **assist patients, clinicians, purchasers, and policy-makers in making informed health decisions** by advancing the quality and relevance of evidence concerning the manner in which diseases, disorders, and other health conditions can effectively and appropriately be prevented, diagnosed, treated, monitored, and managed **through research and evidence synthesis...**

… and the dissemination of research findings with respect to the relative health outcomes, clinical effectiveness, and appropriateness of the medical treatments, services..."

--from PCORI's authorizing legislation

Research priorities and fit with genomics

Assessment of Prevention, Diagnosis, and Treatment Options

Improving Healthcare Systems

Communication and Dissemination Research

Addressing Disparities

Accelerating Patient-Centered Outcomes Research and Methodological Research

Communication and dissemination research

Producing information is not enough.

- Clear communication approaches <u>and</u> active dissemination of findings to all audiences, in easy to understand formats, are critical to increasing the awareness, consideration, adoption, and use of the data by patients, caregivers, and healthcare providers
- In other words, information itself is of little use unless:
    - It reaches those who need it
    - It is clear and comprehensible

Towards patient decision-making tools and content development

- Based upon the evidence linking choices to patient-relevant outcomes
- Methodologically rigorous (IPDAS?)
- Involve patients and clinicians throughout the process of developing and evaluating tools
- Accessible to patients
  - Available
  - Understandable
- Responsive to patient needs and preferences
- Evaluated

Moving past development

- Translation into “real-world”: will it get used?
- Is there buy-in from relevant stakeholders?
- Who maintains?
- Does it improve decision making? Does it improve outcomes?

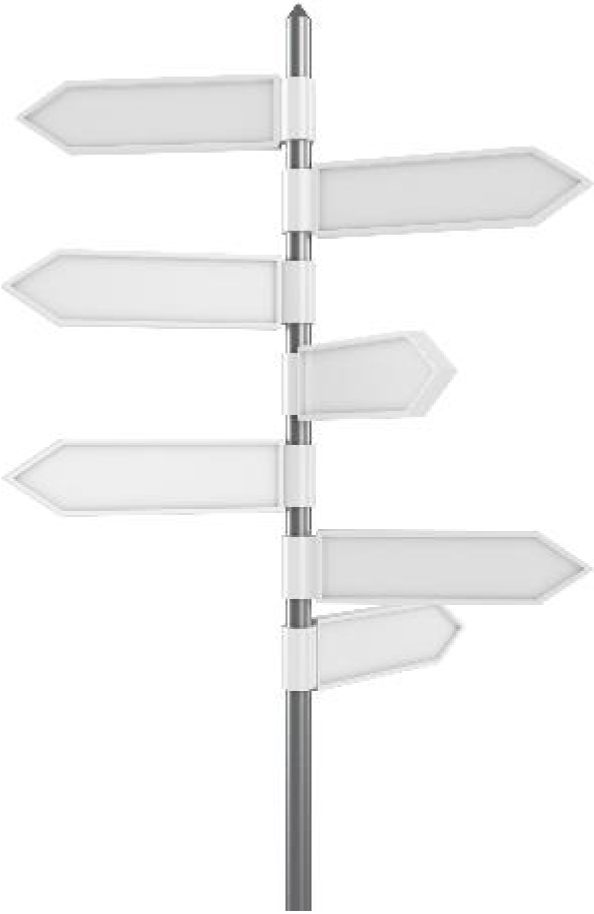
Needs, and questions for discussion

- Fundamental question: is development of patient-facing tools part of NHGRI's mission?
- What sort of evidence do we need to validate tools?
  - How can we incorporate the patient's voice?
- Are we ready to standardize tools?
  - E.g., according to the International Patient Decision Aid Standards (IPDAS)
- What about a clearinghouse of patient-facing tools?

Additional background slides

How is PCORI's Work Different?

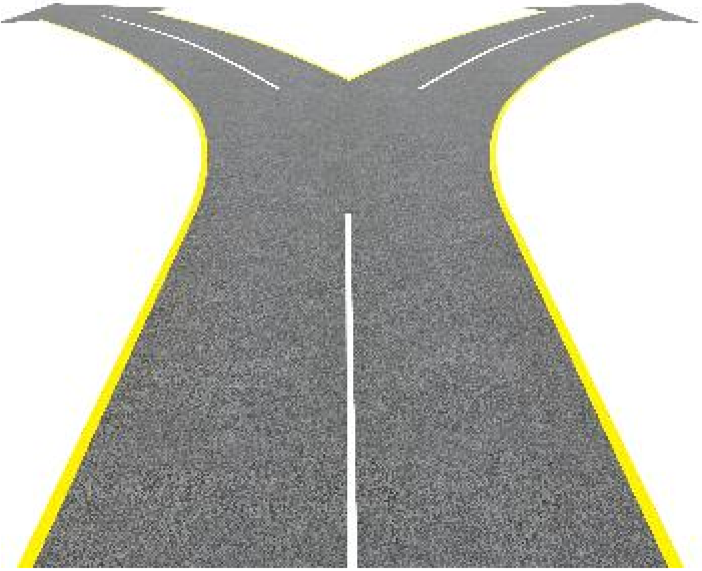
- We fund research on which care options work, for whom, under which circumstances.
- We focus on answering questions most important to patients and those who care for them.
- We aim to produce evidence that can be easily applied in real-world settings.
- We engage patients, caregivers, clinicians, insurers, employers and other stakeholders throughout the research process.
- This makes it more likely we'll get the research questions right and that the study results will be useful and taken up in practice.



We Fund CER

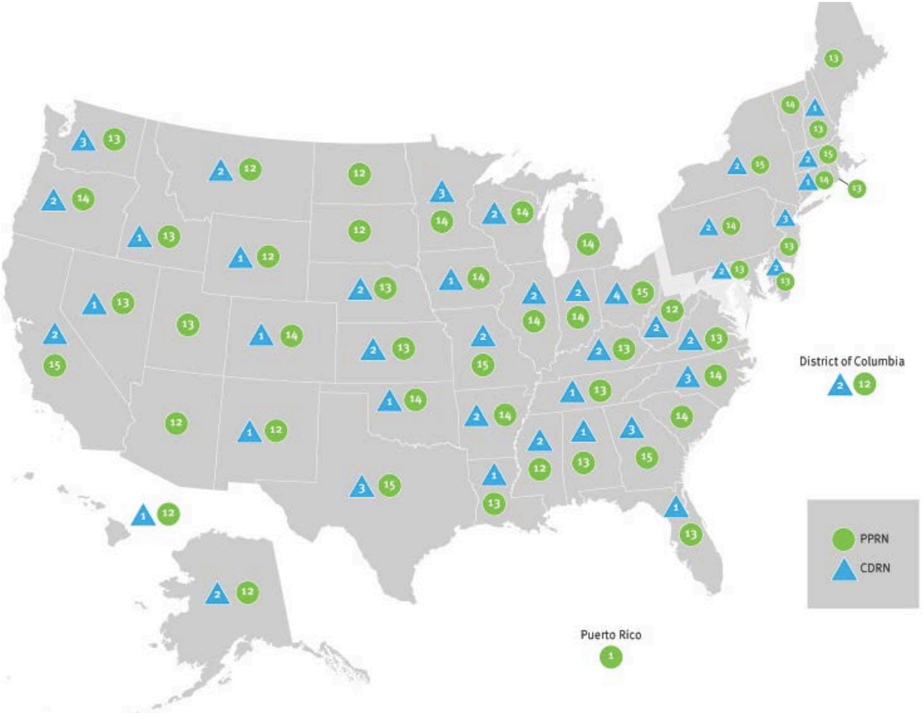
Research that….

- Generates and synthesizes evidence comparing benefits and harms of at least two different methods to prevent, diagnose, treat, and monitor a clinical condition or improve care delivery
- Measures benefits in real-world populations
- Informs a specific clinical or policy decision
- Describes results in subgroups of people
- Applies appropriate methods and data sources
- Helps consumers, clinicians, purchasers, and policy makers make informed decisions that will improve care for individuals and populations



Adapted from *Initial National Priorities for Comparative Effectiveness Research*, Institute of Medicine of the National Academies

PCORnet: 11 CDRNs and 18 PPRNs



This map depicts the number of PCORI funded Patient-Powered or Clinical Data Research Networks that have coverage in each state.

PCORnet Patient-Powered Research Networks (PPRNs)

Common	Rare
American BRCA Outcomes and Utilization of Testing Patient-Powered Research Network (ABOUT Network)	ALD Connect
ARthritis patient Partnership with comparative Effectiveness Researchers (AR-PoWER PPRN)	Community-Engaged Network for All (CENA)
CCFA Partners Patient Powered Research Network	DuchenneConnect Patient-Report Registry Infrastructure Project
COPD Patient Powered Research Network	NephCure Kidney Network for Patients with Nephrotic Syndrome
Health eHeart Alliance	Patients, Advocates and Rheumatology Teams Network for Research and Service (PARTNERS) Consortium
ImproveCareNow: A Learning Health System for Children with Crohn's Disease and Ulcerative Colitis	Phelan-McDermid Syndrome Data Network
Mood Patient-Powered Research Network	PI Patient Research Connection: PI-CONNECT
Multiple Sclerosis Patient-Powered Research Network	Rare Epilepsy Network (REN)
Sleep Apnea Patient Centered Outcomes Network (SAPCON)	Vasculitis Patient Powered Research Network

Critical Knowledge Gaps Impeding Genomic Medicine Implementation

- How best to engage patients & families—upstream from question of tools
- What the key endpoints are
- How to do pretest counseling for exome/genome scale tests
- How to educate patients & families outside of specialty settings
- How to solicit informed preferences

Critical Knowledge Gaps Impeding Genomic Medicine Implementation (2)

- Sensitive, specific & scalable ways to capture family history

Other Key Barriers to Implementation

- Workforce limitations
- Infrastructures for distance education & counseling
- Integration into clinical workflow
- Public literacy
- Public trust (e.g., newborn blood spots)

Recommended Approaches to Addressing Gaps and Barriers

- Engage patients as partners, not subjects, in implementation & communication research
- Integrate tool development (and the necessary infrastructure) into funded implementation projects
- Develop & evaluate tools in clinical settings
- Think dynamic and interactive

Training Needs and Approaches

- Support trainees in communication & decision sciences?