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

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NHGRI
Implementation requires effective clinician-patient communication
Patient-facing tools needed for education, risk assessment & decision support

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

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

<table>
<thead>
<tr>
<th>Patient Resource</th>
<th>NIH Program</th>
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<tbody>
<tr>
<td>Genetics Home Reference</td>
<td>ClinGen, CSER, UDN, NHGRI</td>
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<tr>
<td>Cancer Genetics PDQ</td>
<td>CSER, NHGRI</td>
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<tr>
<td>Genome Connect</td>
<td>ClinGen</td>
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<tr>
<td>Genetic Alliance</td>
<td>UDN</td>
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<tr>
<td>MEDLINEPlus</td>
<td>UDN</td>
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<tr>
<td>NORD/GARD</td>
<td>ClinGen, UDN, NHGRI</td>
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<tr>
<td>OrphaNet</td>
<td>UDN</td>
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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.
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.
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

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

• 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.
“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 and 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)

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