

Ethical, Legal, and Social Issues

Jean Jenkins PhD, RN, FAAN
Clinical Advisor, DPCE, NHGRI





ELSI Program at NHGRI

- McEwen, J. et al. (2014). ELSI Program of the NHGRI. *Annu. Rev. Genomics Hum. Genet.* 15:481–505

Genomic research

The issues that arise in the design and conduct of genomic research, particularly as it increasingly involves the production, analysis, and broad sharing of individual genomic information that is frequently coupled with detailed health information

Genomic health care

How rapid advances in genomic technologies and the availability of increasing amounts of genomic information influence how health care is provided and how it affects the health of individuals, families, and communities

Broader societal issues

The normative underpinnings of beliefs, practices, and policies regarding genomic information and technologies, as well as the implications of genomics for how we conceptualize and understand such issues as health, disease, and individual responsibility

Legal, regulatory, and public policy issues

The effects of existing genomic research, health and public policies and regulations, and the development of new policies and regulatory approaches

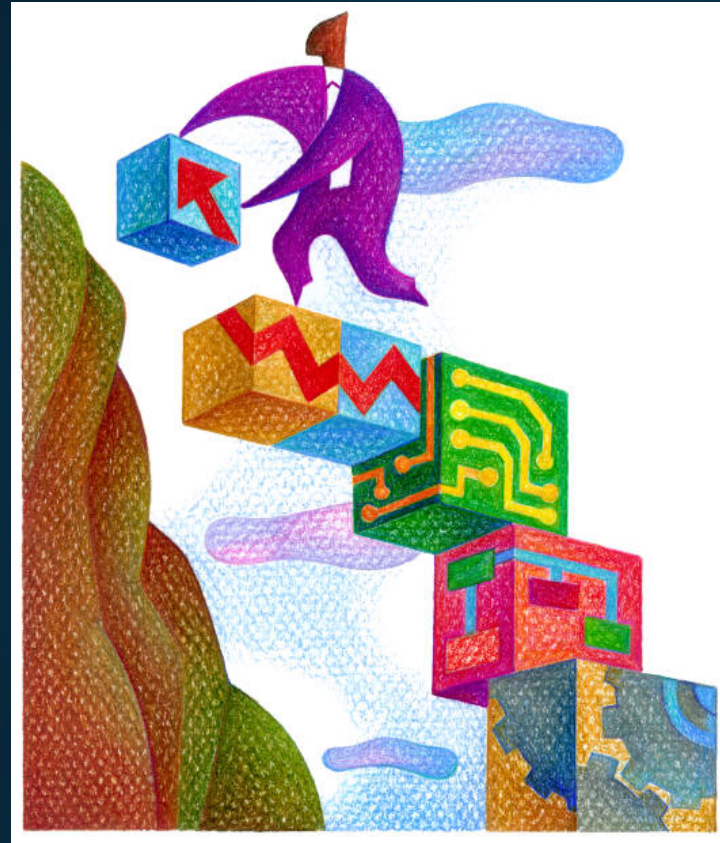


Genetic Influences Across Life's Journey (Relevancy and Options)

- Preimplantation testing
- Prenatal
- Carrier testing
- Diagnostic testing/Children
- Diagnostic testing/Adult Onset
- Treatment decision testing
- Pharmacogenomics
- Recreational (Direct to Consumer)
- Whole Genome Sequencing
- Precision Medicine

BUILDING BLOCKS

- Research
Progress/
Evidence
- Healthcare
Professionals
- Healthcare
Systems
- Resources
- Public
- You





Profound Change

- Genomic technology will allow us to identify those at risk for genetic and genomic conditions
- Infrastructure needed to provide predictive and preventive services is not currently available
- Knowledgeable providers that can interpret test results for individuals and their family are sparse
- Evidence of beneficial outcomes of care still incomplete *Nursing research
- Reimbursement that covers such services is limited



Ethical Implications

- Complexity of information
- Public literacy
- Promise versus reality
- Privacy
- Confidentiality
- Access to health care
- Not everyone wants to know genetic information
- Informed health decisions
- Grey areas of clinical utility and personal utility



Predictive

- Kevin and his family have an extensive history of colon cancer and are seen as part of an HNPCC (Lynch) clinical study. Kevin and his twin brother find out through genetic testing that one is at risk for the hereditary colon cancer and the other is not. This information presents different challenges for each of them as they seek out guidance from their individual care providers. Kevin reports he spends a lot of time explaining why he needs a colonoscopy now even though he's only 30. Kelly works in the state legislature to improve coverage for colonoscopies.

Genetics/Genomics of Common Disease - HNPCC

- IMPLICATIONS FOR INDIVIDUALS AND THEIR FAMILIES
- COMMUNICATION OF TEST RESULTS
- SCREENING BEHAVIORS AND PAYMENT SYSTEMS
- [HTTP://WWW.CANCER.GO
V/TYPES/COLORECTAL/HP
/COLORECTAL-GENETICS-
PDQ](http://www.cancer.gov/types/colorectal/hp/colorectal-genetics-pdq)



HNPPC or Lynch Syndrome



[REGISTER NOW](#)

**PUBLIC HEALTH GENOMICS IN PRACTICE:
Lynch Syndrome: A Public Health Approach**

A webinar presented by
**APHA Genomics Forum Tier 1 Applications and Workforce
Development Workgroup and Genetic Alliance**
Tuesday, April 21, 2015, 12:00 pm - 1:00 pm ET



Psychosocial and Ethical Issues

- Mosen, R. textbook (2008)
- Guilt
- Fear
- Privacy concerns
- Anxiety
- Unable to understand complex information
- Unable to explain complex information to family
- Depression
- Other?



Privacy and Confidentiality

- Individuals who know the information may have difficulty keeping from and/or sharing of genetic information with family members, friends, insurers, and employers
- Genetic tests can be performed on stored samples (e.g. blood or tissue), including samples collected for other purposes
- Medical information is increasingly kept in electronic databases, tied to genomic information, and placed in widely accessible databases (e.g. dbGaP)

Implications Across the Generations

- Gatekeeper
- Communicator
- Decision maker
- Short term and long term implications
- Neurogenethics

Carli, T. (2015).
Applied & Trans-
lational Research
5:18-22.





Barry

- Barry has experienced progressive neurologic symptoms since age 35. He's visited multiple physicians, had expensive work-ups, and been frustrated that he doesn't know why his symptoms or what he has. He was enrolled in the NIH undiagnosed disease program which included whole genome sequencing as part of his work-up.

(<https://www.genome.gov/27550959/undiagnosed-diseases-network-udn/>) Lots of information is now available to share with Barry, including some “incidental findings”. But he's unsure he wants to know all of it.



Incidental Findings

- “A finding concerning an individual research participant that has potential health or reproductive importance and is discovered in the course of conducting research but is beyond the aims of the study.”
 - For analysis on identifiable specimens, establish a plan for handling incidental findings
 - Mechanism for assessing for incidental findings
 - Criteria for offering results
 - Confirmation
 - How long you will look for results

Wolf, S., et al. (2012). Managing incidental findings and research results in genomic research involving biobanks and archived datasets. *Genetics in Medicine*, 4, 361-384.



Return of Incidental Findings ACMG 2013 Policy Statement


- ACMG policy statement is specific to clinical application of whole genome analysis
- Specifies that laboratories should interrogate the genome for a list of 56 specific genes associated with specific phenotype/syndromes.
- Duty to warn surpasses patient autonomy
- Decision to disclose these results should not be restricted to the age of majority but encompass disclosure of results including adult onset disorders to the parents of children
- Option to opt out (update)

Green, R. et al.(2013). ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. *Genetics in Medicine*, 15(7), 565-574.



Patient views on incidental findings (IF)

- Clift, K. et al. (2015). Applied & Translational Genomics 4:38–43 Fifty-five in-depth interviews
- Participants felt it was imperative to include the patient in decisions
- Risks and benefits of knowing genomic information are personal and contextual
- Many voiced desire to received IF
 - Quality of life; finding answers; prep for future
- Many voiced desire not to received IF
 - Religious beliefs; burden of knowledge; future repercussions



Was it worth it? Value of genomic individualized medicine

- Halverson, C. (2016). Journal of Community Genetics. 2:145-152. Thirty-seven interviews
- Types of results

Status

Deleterious mutations related to phenotype

Required to return

Variants of unknown significance suspected to be related to phenotype

Required to return

Medically actionable deleterious mutation unrelated to phenotype

Optional

Carrier status for Mendelian disorders

Optional

Pharmacogenetic variants

Optional



Results were overwhelmingly positive

- Personal utility-felt empowered over their own health
- Felt they were contributing altruistically to genetic technology progress
- Felt their suffering had been legitimized
- Sense of closure in that they had done everything they could

- Lupo, P. (2016). *Per Med.*1; 13(1): 13–20.
- 23% enthusiasts; 60% health conscious-perceived medical utility; 17% skeptics



Research Considerations

- Stability of DNA (i.e., storage and future use)
- Broad sharing of samples/data
- Limited control of downstream use
- Limited right to withdraw
- Identifiability
- Incidental findings and duty to disclose
- Return of results




Return of Research Results

- Process of offering and delivering results
 - Inform subjects how they would be contacted
 - Involve healthcare professionals with the appropriate expertise (i.e., experts in the field, geneticist, genetic nurse, genetic counselor)
 - The research participants' right to not know certain test results
 - The process is in compliance with professional standards related to disclosure of genetic and genomic information for healthcare decision-making
 - Genetic education and counseling



Lauren

Lauren was diagnosed with breast cancer at age 34. She was offered genetic testing and decided this would be a good option for her. She was excited to share this decision with her husband and sister. However, both expressed concerns about her decision. Her husband was worried about what they would do if a mutation was identified – they had a 14 year old daughter and who and when would they tell her? Her sister was concerned because she had no interest in knowing her own risk for cancer. If Lauren knew, that would make it complex for her. Lauren died before getting her genetic test results.




Returning a Research Participant's Genomic Results to Relatives

- Analysis and Recommendations Wolf, S. et al. (2015). *J Law Med Ethics*. 43:3, 440-463
- Discussion as to whether researchers have any responsibility to offer participants results to participants relatives
- Offers ethics review, recommendations, and model pathways



Explore Informed Consent Materials:

- [Process and Special Considerations for Informed Consent in Genomics Research](#)
- [Informed Consent Elements: Considerations for Genomics Research and Sample Language](#)
- [Sample Consent Forms](#)
- [Glossary](#)
- [Additional Resources and Educational Tools](#)
- <http://www.genome.gov/27026588>



In addition to being personal and unique to each individual, genomic data may:

- Be stored and used indefinitely.
- Inform individuals about susceptibility to a broad range of conditions (some unexpected).
- Carry with them risks that are uncertain or unclear.
- Be reinterpreted and change in relevance over time.
- Raise privacy concerns (in part because of the risk of re-identification).
- Be relevant for family members and reproductive decision-making.



Social Implications

- Public perceptions
- Human rights
- Accessibility
- Discrimination

Discrimination

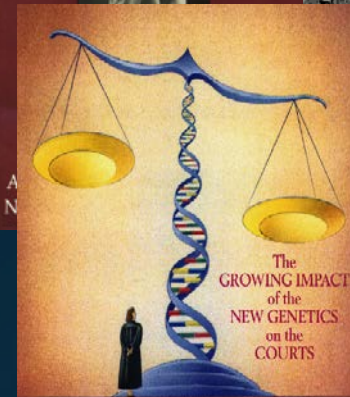
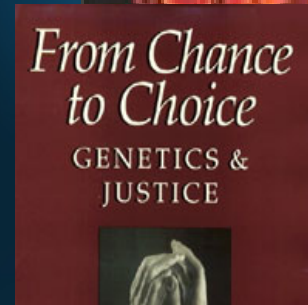
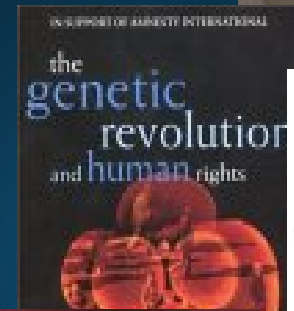
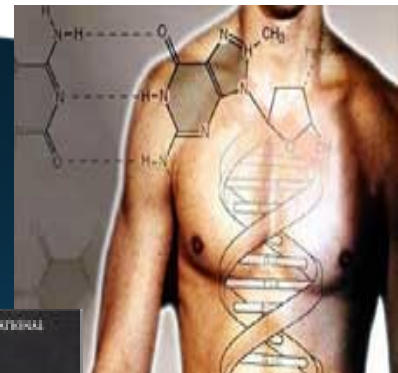
- Stacey's sister is enrolled in a family history study designed to determine if there is an inherited contribution to why her lymphoma occurred at a young age. Stacey is concerned about the potential utilization of this information against her in the workplace and isn't interested in being a part of the study. Is her concern valid?



What Is Genetic Discrimination?

- Social or economic discrimination or stigmatization based on one's genetic information
 - denial of access to or increased cost of insurance
 - loss of employment, educational, or other opportunities
- Insurance considerations
 - Health, Life, Long Term Care, Disability

****Special consideration for the military**



GENES AND JUSTICE



Race, Ethnicity, Culture

- Personal identity
- Community identity
- Past history of abuse
- Health disparities

- * In study by Lupos (2016) TRUST influenced participant perceptions of utility of whole genome sequencing. Recommended research among those with historical mistrust of medicine, physicians, and research



Policy

- Prevention of discrimination
- Accessibility to services
- Return of genetic test results
- Communication of genetic tests results to family members
- Documentation and privacy of health information
- Communication of health information via EHR



What is GINA? (2008)

- A federal law that prevents health insurers and employers from discriminating based on an individual's genetic information
- The law is intended to allow Americans to take advantage of the benefits of genetic testing without fear of losing their health insurance or their jobs
- <http://www.genome.gov/10002328>



GINA Coverage Limitations

- Civil suit is restricted to those that have exhausted all administrative remedies
- Does not prohibit medical underwriting based on current health status
- Does not cover life, disability, and long-term care insurance
- Does not cover employers with <15 employees (those covered by the ADA)



GINA Coverage Exclusions

- Members of the United States military, veterans obtaining health care through the Veteran's Administration, and the Indian Health Service because the laws amended by GINA do not apply to these groups and programs



Legal Implications

- Regulatory oversight
- Reimbursement
- Guidelines



QUALITY OVERSIGHT

- **FDA takes steps to help ensure the reliability of certain diagnostic tests:** Reinforces agency's commitment to fostering personalized medicine Laboratory Developed Tests (LDT)

<http://www.fda.gov/MedicalDevices/ProductsandMedicalProcedures/InVitroDiagnostics/ucm407296.htm>

- Drug Labeling
- Genome sequencing
- NIH Genetic Testing Registry
 - <http://www.ncbi.nlm.nih.gov/gtr/>
- EGAPP (www.cdc.gov)
 - Coordinated, systematic process for evaluating genetic tests



Visioning

- Davey is receiving treatment for cancer. He has a family history of cancer but when tested for known genetic mutations, none have been identified. He is being offered as part of his clinical study to have whole genome sequencing. He has no idea what that is and asks you for more information about whether or not he should do this.

National Institutes of Health (NIH)





Precision Medicine Initiative

- Privacy concerns
- Providers
- Participants
- Diversity
- Sample access and storage
- Big data
- Information provided back to participants
- Communication methodologies and technologies
- Across the generations?
- Other



Religious and Spirituality Values

- Beliefs
- Origins
- Body substances “life”
- <http://genetics.ncai.org/what-do-tribes-think-about-genetics-research.cfm>
- **A Spectrum of Perspectives: Native Peoples and Genetic Research**
- <https://www.youtube.com/playlist?list=PLS6nSmuURFJC6iY2IWMFwfVjXDHfC6AwC>

Positioning for the Future





Resource

- Badzek, L. et al (2013). Ethical, Legal, and Social Issues in the Translation of Genomics Into Health Care. *Journal of Nursing Scholarship*, 2013; 45:1, 15–24. Available at: <http://www.genome.gov/27552093>
- Hammer, M. (2016). Research Ethics Considerations Regarding the Cancer Moonshot Initiative. *ONF*, 43(4), 428–431.
- Hammer, M. (2016). Precision Medicine and the Changing Landscape of Research Ethics. *ONF*, 43(2), 149–150.



ELSI Research

- Pullman, D., & Etchegary, H. (2015). **Clinical Genetic Research 3: Genetics ELSI (Ethical, Legal, and Social Issues) Research** (Chapter 22 in Patrick S. Parfrey and Brendan J. Barrett (eds.), *Clinical Epidemiology: Practice and Methods, Methods in Molecular Biology*, p. 369-382.)
- What is ELSI research? Historical context.
- When and how to engage in ELSI research.

PERSONAL/PROFESSIONAL PERSPECTIVE

- Example
 - NHL

SEMINARS IN
ONCOLOGY
NURSING,
2006, 22(2),
117-125



dnaandu.org

Share and learn from personal stories about using or not using genetic information in personal health care.



I'm Jean Jenkins. I'm a nurse and cancer survivor with a dream of building an archive of stories documenting the use of DNA information in personal healthcare. Your experiences can help others navigate this emerging field. [Browse](#) or [share your story](#).



BROWSE STORIES

See what others say about their experiences.



SHARE YOUR STORY

Write your story, make a video, or connect with Jean for an informal interview.



THE PROJECT & JEAN

Genetic testing helped Jean fight lymphoma. Find out more about her story and what led to this project.



Summary

- This is a pivotal time in history
- ELSI issues are relevant to all healthcare professionals
- Leadership is needed
- You have the ethical foundation necessary to guide translation of genomics
- Each of you are key to bridging the gaps, providing the voice, and moving knowledge forward in a safe and sensitive manner for all “participants/partners”
- It is essential to have diverse populations and voices included

Future Opportunities

Jean.Jenkins@nih.gov
301-547-1897



• OUTCOME INDICATORS PUBLISHED WITH COMPETENCY 2ND EDITION
 K. COLONE, M.D., PH.D., P.F.A.P.A., F.A.A.N.
 1 AND 2
 • EDITORIAL BOARD GENETICS CLERKS 2
 • COVER STORY WATCH...
 • PUBLISHED ONLINE
 • PUBLISHED IN PRINT
 • ACCESS TO FULLTEXT ONLINE
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0 ISSUE # / 0 VOLUME 3 / 0 YEAR 2008

Competency focus

UPDATING THE NURSING COMMUNITY ON THE IMPLEMENTATION OF THE ESSENTIAL NURSING COMPETENCIES AND CURRICULA GUIDELINES IN GENETICS AND GENOMICS.

In September 2005, the Genetics Genomics Nursing Competency initiative finalized Outcome Indicators for each of the individual competencies. They are being published this month with the release of the 2nd Edition of the Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics.

Outcome Indicators Published with Competency Second Edition

After a year long consensus process, we are pleased to announce the availability of the Outcome Indicators that accompany the Essential Competencies. The Outcome Indicators are an adjunct to the Essential Competencies and Curricula Guidelines for Genetics and Genomics. They define the specific areas of knowledge and suggested clinical performance indicators for each specific competence. The Outcome Indicators are not intended to be prescriptive but provide a guide for the user. To be consistent with the Competencies, the definitions utilized are identical to the Essential Competencies. Genetic and genomic information is used as the context for defining knowledge and practice indicators for each competency.

For a portion of the Outcome Indicators, a table of the Professional Responsibilities Competencies is included here to demonstrate how the Outcome Indicators are structured. A copy of the 2nd Edition of the Competencies which include the Outcome Indicators can be found by accessing <http://www.genetics.org/20080404/Cy> request a copy.

Genetic Professional Responsibilities	Genetic Professional Responsibilities
Genetic Professional Responsibilities Recognize when one's own attitude and values related to genetic and genomic science may affect care provided to clients.	Genetic Professional Responsibilities Advocate for clients' access to desired genetic/genomic services and/or resources including support groups.
Genetic Professional Responsibilities Understand the impact of personal values, attitudes and beliefs on genetic and genomic health care.	Genetic Professional Responsibilities Disseminate genetic/genomic information and lay public about disease susceptibility, prognosis (e.g. timing, location, end-of-life), genetic research (e.g. the National Human Genome Research Institute http://www.genome.gov and the Center for Disease Control National Office of Public Health Genomics http://www.cdc.gov/genomics/central/).
Genetic Professional Responsibilities Advocate for genetic testing/genomic medicine when one's competency.	Genetic Professional Responsibilities Help direct appropriate genetic testing/genomic medicine services to those who need them.
Genetic Professional Responsibilities Identify strategies that could be used to build a network of genetic/genomic services.	Genetic Professional Responsibilities Identify strategies that could be used to build a network of genetic/genomic services.

Continued on page 2