CLINICAL SEQUENCING EXPLORATORY RESEARCH CONSORTIUM

CSER Background Document in preparation for the

CSER and Beyond Meeting

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CSER: INTRODUCTIONS

Today, the availability of genomic sequencing for the public has outpaced the evidence base necessary to evaluate its effectiveness in clinical care. Like any new, important medical technology, it is imperative to understand the optimal implementation, advantages and limitations of genomic sequencing in the clinic before it is widely implemented in patient care. Premature implementation using sub-optimal strategies may lead to sub-optimal outcomes. Such an evidence base is also essential to insurers evaluating coverage of this new technology's cost. Before whole exome and whole genome sequencing can become routine practice in patient care, many questions need to be answered, such as:

- How should clinical workflows, institutional policies, standard care procedures and electronic medical records evolve to accommodate genomic sequencing data? What best practices can be derived from a learning health-care system?
- What are the ethical, legal and psychosocial implications (ELSI) related to the use of genomic data? What do providers and/or patients need to optimally use this data? What fail-safes should be implemented to prevent errors, poor integration into care and poor outcomes?
- For what diseases does the addition of genomic sequencing significantly improve patient outcomes at reasonable cost? In what situations is genomic sequencing less impactful? What roles do individual gene and panel sequencing play?

To address these questions and other issues related to the use of genomic data in clinical care, the NHGRI initiated the **Clinical Sequencing Exploratory Research** (CSER) program. CSER is currently comprised of 18 primary research grants (see Appendix A), a coordinating center, a NIH intramural research project (ClinSeq) and other collaborators across the United States, spanning over 300 researchers, clinicians, bioethicists and more. CSER's research includes both children and adults, in areas ranging from the diagnostic needs of children with developmental delay to adults with different forms of cancer, and explores sequencing across the lifespan, from pre-conception to late adulthood. Simultaneously, ethical issues related to these projects, such as the ELSI with the return of genomic sequencing results to children, adults and families, health economics and decision-making related to genomic results are also researched. CSER has diverse experimental approaches, cross-cutting working groups and covers a vast range of issues:

1. Technology: Generate and interpret genomic sequence data on patients in a variety of clinical contexts
2. Clinical Care: Study the challenges of integrating comprehensive, personal genomic sequence data to patient care
3. ELSI: Examine the ethical, psychosocial, and legal implications of bringing genomic sequence data into the clinic

The CSER Consortium seeks to rapidly advance the knowledge necessary to develop best practices for the implementation of genomic sequence data into clinical care. CSER is uniquely positioned among NHGRI programs to answer questions about the clinical implementation of genomic sequencing to meet its growing use in the clinical care of patients with diverse needs.

CSER AT A GLANCE

- **Funding**: 9 U-grants and 1 Coordinating Center grant, $65M from FY11-F16 ($56.2M from NHGRI, $9.4M from NCI). $14.8M NHGRI funds in FY15
- "**U-Grants**" are described in RFAs **HG-10-017**, **HG-12-010** and **HG-12-015** to implement three integrated projects:
    1. Recruitment of patients and/or physicians in clinical settings to study genomic medicine implementation
    2. Generate and interpret genomic sequence data in a clinically useful format
    3. Ethical, legal, and psychosocial implications (ELSI) research
- "**R-grants**" are described in **HG-11-003** and **HG-11-004** and also explore ELSI issues related to the return of genomic results (originally funded as ELSI Return of Results Consortium)
- **Over 200 publications** and **700 presentations and posters** to date, many high impact
- **9 Working Groups** convened to identify and share common challenges and opportunities
- **Identification of high priority cross-study projec**ts, such as clinical exome coverage comparison, analysis of informed consent documents, best practices in variant sign-off, and variant pathogenicity annotation across sites
- **Products from CSER work include** a family pedigree app ***Proband*** (1700 downloads), a **CSER variant database**, 2617 ClinVar submissions, 1149 dbGAP submissions, and numerous other genomic tools like **Atlas 2** and **Cassandra**
- **Data analysis is currently underway and the most important quantitative study outcomes are in progress.**

U-GRANTS: ACCOMPLISHMENTS AND OUTREACH

CSER's **U-Grant Sites**, individually and collaboratively, have made significant advancements in researching the incorporation of genomic sequencing data into clinical care. This research has informed the development of best practices for genomic sequencing in the clinic, and has influenced communities and projects outside of CSER's specific focus. These achievements include:

- Total enrollment of 4979 participants (3984 adults, 995 children)
- Collaborated with the eMERGE Network on a consensus statement on return of genomic research results and with other projects including Children's Oncology Group, NSIGHT, ClinGen, IGNITE and the Undiagnosed Disease Network
- Variant interpretation "bake-offs" refined methodologies for the evaluating genomic variants to reduce discordant classification, and impacting care by influencing ACMG pathogenicity criteria
- Development of bioinformatics analysis tools, variant interpretation tools, patient facing information and other software used collaboratively by CSER projects and in the clinical services offered outside of CSER studies

NCGenes | University of North Carolina

- Discovered that effectiveness of WES varies with clinical presentation: retinal and neurological disorders demonstrate high diagnostic yields while standard clinical approach to familial cancer is less impacted by WES
- Collaborated with Vidant Cardiology (non-profit hospital system) to bring WES into a rural community with a large minority population
- Lessons learned include the need for geographically convenient clinics and offering accessible language resources for non-English speakers

"…through the pursuit of such collaborations that NCGENES accomplished one of its over-arching goals: robust minority participation (~25%)"

University of North Carolina

NEXT Medicine | University of Washington

- Determined the rate of actionable incidental findings and that the return of incidental findings and gene panels for colorectal cancer are cost-effective
- Extensive work on FDA regulation of genomic testing
- Worked with CERNER, EPIC and the Institute of Medicine to prioritize getting genomic information into the medical record in scalable, usable ways

"Our legal and regulatory analyses are widely cited for return of non-CLIA research results and also implications of new genomic medicine regulations."

University of Washington

NEXTGen | Kaiser Foundation Research Institute

- Developed a framework for offering patient choice for results disclosure in the context of preconception carrier screening.
- Developed patient-facing materials, such as consent forms, clinical variant reports and educational materials that is shared with other CSER sites

"...consortium activities have enabled us to address questions that could not be answered by a single site."

Kaiser Foundation Research Institute

MI-ONCOSEQ | University of Michigan

- Clinical sequencing of tumor samples identified actionable mutations in about 60% of patients, many of which aren't routinely screened by standard diagnostic tests
- Developed "Onco1500," a gene panel that targets 1500 genes to efficiently identify genetic aberrations in both highly recurrent cancer genes and additional candidate genes with suggestive links to cancer that expedites turnaround times.

"Some of the treating physicians have pursued therapies informed by sequence analysis, notably for pediatric patients"

University of Michigan

MedSeq | Brigham & Women's Hospital

- Developed and refined a genomic medicine workflow, including iterative interaction between lab and clinic, for both diagnosis and prevention
- Completed pre-planned recruitment of randomized trial of sequencing measuring multi-modal medical, behavioral and economic outcomes
- Created a new method of complex blood serology typing through sequencing
- Contributed, through CSER interactions, to ACMG recommendations for NGS Laboratory Standards, Incidental Findings and Variant Classification

"Despite perceptions to the contrary, we are demonstrating that a pathway exists for physician preparedness to genomic medicine. "

Brigham & Women's Hospital

ClinSeq | National Human Genome Research Institute

- Discovered it was practical and feasible to extract pathogenic variants from exome data for participants unselected for disease
- Contributed to ACMG recommendations for incidental findings that became the standard of care in genomic medicine

"Our work, and all of the CSER medical and genomic research is pointing to the same conclusion - genomics is just medicine"

NHGRI

CanSeq | Dana-Farber Cancer Institute

- Developed processes such as targeted sequencing and innovations such as methods for clinical analysis and interpretation of genomic information, and implemented these developments in Broad Institute's CLIA Sequencing Lab
- Lessons from CanSeq have applied to projects in breast cancer, prostate cancer and other tumor types

"we have learned a great deal about… providers' willingness to encounter large-scale genomic data through the collective efforts of the CSER sites"

Dana-Farber Cancer Institute

BASIC3 | Baylor College of Medicine

- Preliminary research results suggested that exome results were most useful for oncologists when targeted treatment was available for the patient that was consistent with exome results
- Performed clinical exome sequencing of patients as they were diagnosed and disclosed results while patient is under treatment
- Participated in one-on-one consulting with major pediatric oncology institutions about starting their own clinical genome sequencing approaches

"nearly 40% of pediatric solid tumor patients have potentially actionable mutations when combining results of tumor and germline exome sequencing"

Baylor College of Medicine

PediSeq | Children's Hospital of Philadelphia

- Generated open-source software *Proband*, a family history pedigree app
- Created an Individualized Medical Genetics Center that influence approaches to genomic diagnostics clinically across all patients at CHOP
- Algorithms from preliminary investigations of clinical decision support by primary care physicians for incidental findings have been made applicable across non-genetic clinical workflows, such as care coordination

"We have collaborated with two other major pediatric centers based on knowledge gained from this project to extend our work in genomics"

Children's Hospital of Philadelphia

Hudson-Alpha Institute for Biotechnology

- Partnered with University of South Alabama and multiple regional clinics with the goal of using genomic sequencing in diagnosing children in underrepresented and underserved populations
- Data has been used to diagnose children with otherwise unexplained conditions and to get patients into appropriate specialty clinics

"We have also shown that genomic testing can be done with minimal adjustments or alterations to current clinical workflows."

Hudson-Alpha Institute for Biotechnology

R-GRANTS: ACHIEVEMENTS AND ACCOMPLISHMENTS

The introduction of new, important medical technology raises questions of the ethical, legal and psychosocial implications (ELSI) of its implementation. ELSI issues such as informed consent, return of results and methods of returning actionable incidental findings were researched. Nine R-Grant projects were initiated in parallel to the work done by the U-Grant sites to study ELSI issues. Highlights of these R-Grant projects include:

- Developing legal and ethical frameworks if newborn screening will expand to use residual dried blood samples in genomic research (*John Hopkins University*)
- Investigating whether research participants want incidental findings returned and how participants respond to the return of incidental findings; develop a menu of approaches for informed consent for return of incidental findings (*Columbia University)*
- Studying the ethical and legal issues when returning genomic results to participants of pediatric research (*Vanderbilt University*)
- Ethical issues related to the implementation of large-scale mutation testing in the clinic (*Mayo Clinic*)
- Philosophical evaluation to critically analyze the moral obligation of researchers who study biorepository samples to return individual research results to participants (*The Children's Mercy Hospital*)
- Whether or not participant preferences can reliably guide the return of research results (*Boston Children's Hospital*)

"…preliminary analyses suggest there are no adverse reactions from use of My46 for return of results, when compared to return through a genetic counselor"

Seattle Children's Hospital

"…there is considerable interest in staged consent, assuming the infrastructure to support it can be provided."

Columbia University

"Survey responses generally affirm that debate about return of results and incidental findings must include consideration of return to family."

University of Minnesota

CSER METRICS

Total Enrollment

As of September 2015, total enrollment is 4979 participants (1282 from ClinSeq). 3984 participants are adults and 995 participants are children.

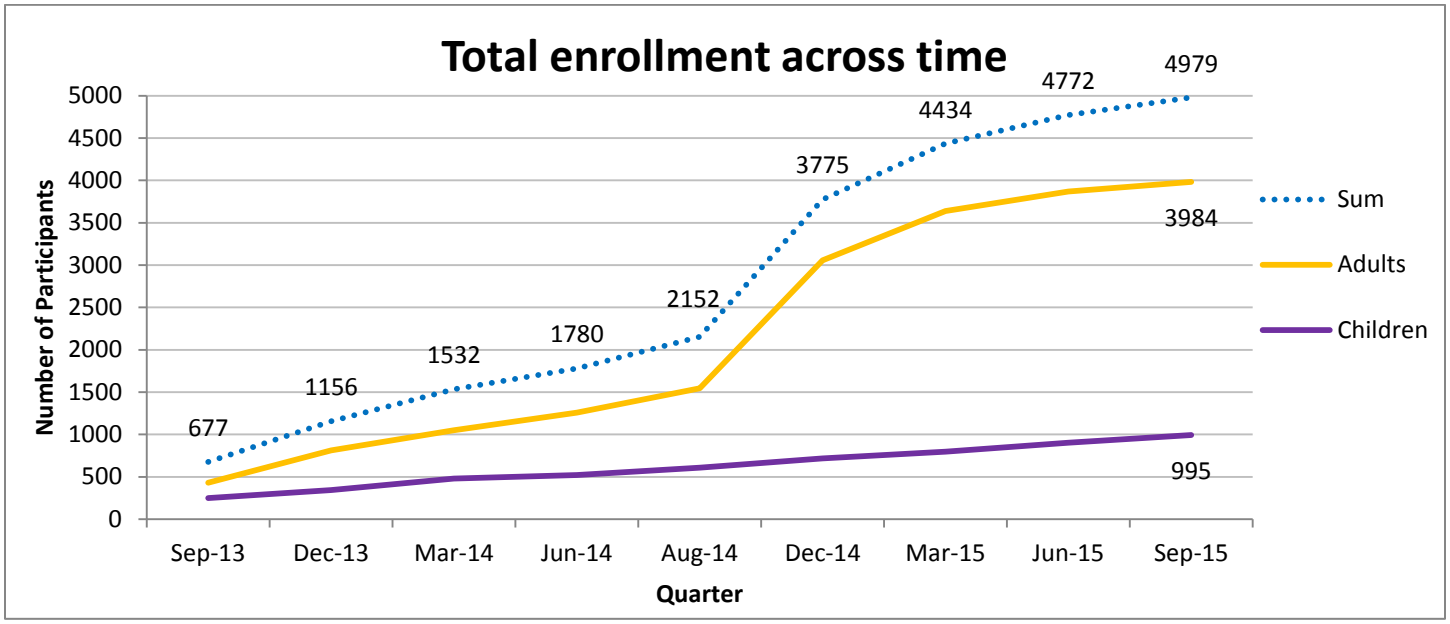

Figure 1: *Number of participants enrolled by CSER sites, including NHGRI ClinSeq (total = 4979)*

Germline Sequencing Progress

As of September 2015, 3989 participants had whole exome or whole genome sequencing completed (not all participants in CSER projects will receive sequencing). Of this number, 3047 participants are adults and 942 participants are children.

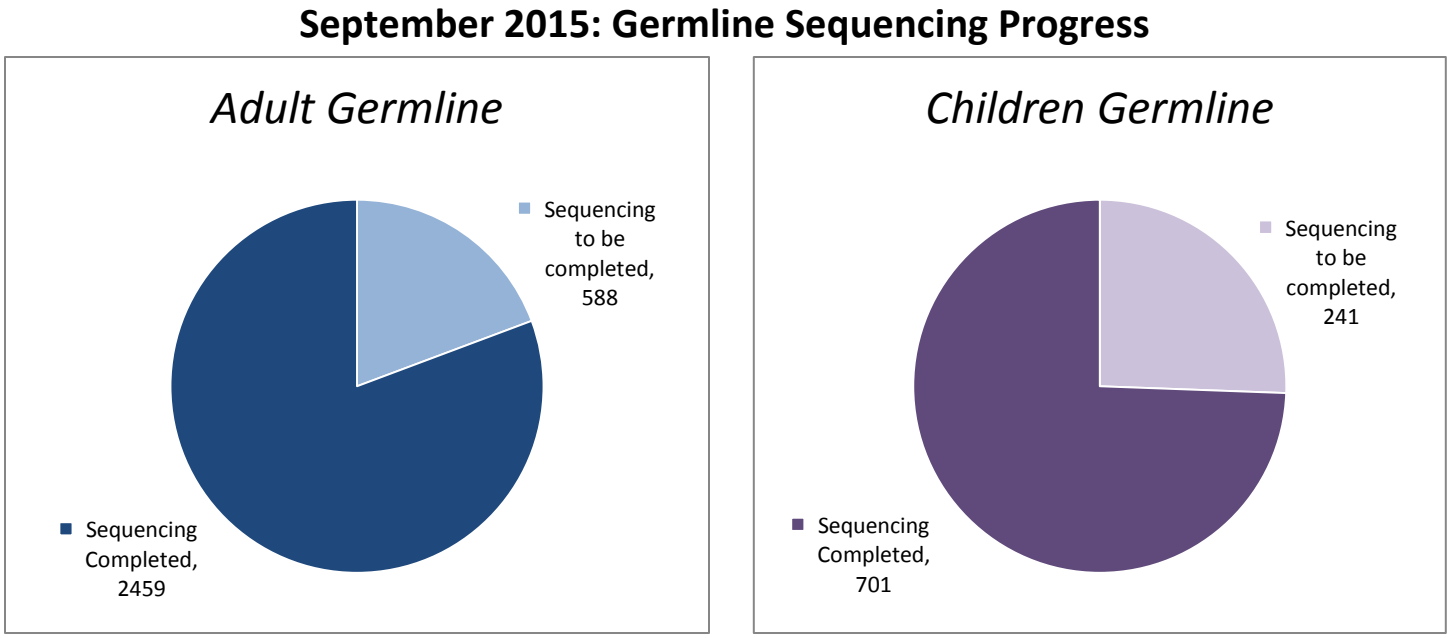

Figure 2a: *Number of adult participants who have had germline sequencing*

Figure 2b: *Number of child participants who have had germline sequencing*

Publications

As of September 2015, CSER Consortium membership has published over 200 publications, with at least 19 publications currently in press and has written many textbook chapters, op-eds, journal commentaries and online news articles. These publications have appeared in *JAMA, NEJM, Genome Research, AJHG, Genetics in Medicine*, *Cell*, and other numerous journals. See Appendix B for a selected list and Appendix C for a full list. Listed below is a selection of publications from CSER working groups.

Working Group Publications

Actionability & Return of Results (ROR)

- Berg et al., "Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequencing data in the CSER Consortium." *Genet Med.* 2013; 15(11):860-7. PMID: Berg et al., "Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequencing data in the CSER Consortium." *Genet Med.* 2013; 15(11):860-7. PMID: 24195999
- Jarvik et al., "Return of Genomic Results to Research Participants: The Floor, the Ceiling, and the Choices in Between. *Am J Hum Genet.* 2014; 94(6):818-26. PMID: 24814192
- Amendola et al., "Challenges of variant classification: Pathogenicity classification from 6503 participant's exomes." *Genome Research* 2015. PMID: 25637381 [Epub ahead of print]

Pediatrics

- Clayton EW et al., "Addressing the ethical challenges in genetic testing and sequencing of children." *Am J Bioeth.* 2014 Mar;14(3):3-9. PMID: 24592828
- Brothers KB, et al., "When Participants in Genomic Research Group Up: Contact and Consent at the Age of Majority." (submitted to *Journal of Pediatrics*)
- Mccullough LB, Brothers KB, Chung WK, et al. Professionally Responsible Disclosure of Genomic Sequencing Results in Pediatric Practice. Pediatrics. 2015. PubMed PMID: 26371191

Electronic Health Records

- Tarczy-Hornoch et al., "A survey of informatics approaches to whole-exome and whole-genome clinical reporting in the electronic health record." *Genet Med.* 2013; 15(10):824-32. PMID: 24071794
- Shirts BH, Salama JS, Aronson SJ, et al. CSER and eMERGE: current and potential state of the display of genetic information in the electronic health record. J Am Med Inform Assoc. 2015. PMID 26142422

Genetic Counsellors

- Amendola LM, Dautenbach D, Scollon S ,Bernhardt B, Biswas S, East K, Everett J, Gilmore MJ, Himes P, Raymond VM, Wynn J, Hart R for the CSER Genetic Counseling Working Group, and Jarvik GP. Illustrative Case Studies in the Return of Exome and Genome Sequencing Results. Personalized Medicine. Vol. 12, No. 3, Pages 283-295.

Informed Consent and Governance

- Henderson GE, et al., "The challenge of informed consent and return of results in translational genomics: empirical analysis and recommendations." *J Law Med Ethics.* 2014 Sep;42(3). PMID: 25264092

Outcomes and Measures

- Gray SW, et al. "Social and behavioral research in genomic sequencing: approaches from the Clinical Sequencing Exploratory Research Consortium Outcomes and Measures Working Group." *Genet Med.* 2014 Mar 13. PMID: 24625446

Presentations and Outreach

As of September 2015, CSER Consortium membership has given over 700 presentations and posters. CSER membership has presented at the ACMG, ASHG, ASBH, AACR, ASCO, PAS and NSGC, as well as at numerous other national and international meetings and conferences. Many presentations have occurred at large public events, such as TedMed and the Smithsonian Institute. Here is a small snapshot of the myriad of places where CSER membership has presented at:

- **Beijing Genomics Institute**; China, 2015
- **Festival of Genomics**; Boston 2015
- **International Symposium on Genome Science**; Japan, 2015 and 2013
- **TedMed** "There is no genome for the human spirit"; 2014
- **Forbes Healthcare Summit**; 2014;
- **European Society of Human Genetics**; Italy, 2014
- **Cambridge Healthtech Institute's Second Annual Advances in Prenatal Molecular Diagnostics Conference**; Boston, 2014
- **ScienceFriday Podcast**, "Does your genome belong to your family, too?"; 2014
- **International Rare Disease Research Symposium;** China, 2014
- **26th Pezcoller Symposium;** Italy, 2014
- **Human Genetics Organization (HUGO);** Switzerland, 2014
- **Israeli Society of Medical Genetics Annual Meeting;** 2013
- **Brocher Foundation Workshop "Returning Genetic Results in Biobanks: Opening an International Dialogue";** Switzerland**,** 2013
- **Shanghai Jiao Tong University Seminar**; China, 2013
- **World Congress of Psychiatric Genetics;** Boston, 2013
- **PBS Radio and Television Documentaries**

CSER: FUTURE OPPORTUNITIES

Selected opportunities where CSER can uniquely contribute to the field of genomic medicine are:

Clinical & Translational

- **Clinical diagnosis of unsolved cases. Genome and exome sequencing currently solve approximately 25% of suspected genetic conditions.** It is important for CSER 2.0 to improve diagnostic yield and explore the interface between clinical testing and discovery research by utilizing novel analysis (e.g., CNV analyses) of patients who had normal sequencing results in CSER 1.0.
- **Evaluation of downstream health and economic outcomes of clinical genome or exome sequencing**. CSER 2.0 would have the collective data across the funded sites that could serve as the foundation for continued development of evidence-based practices in the emerging area of clinical sequencing. This evidence base is needed to standardize care, give less experienced physicians a structure for best practices, and provide insurers and other health care reimbursement systems (payers) with the information they need to determine whether and under what circumstances their enrollees should have access to these genomic tests. Building upon current results, it will be critical to evaluate clinical outcomes along with comparative and cost effectiveness analyses.
- **Optimization of the delivery system to integrate genomic data into clinical workflows including 1) Best practices and scope of practice (workforce, licensing), 2) EHR integration, and 3) Insurance coverage.** Continuation of support through CSER 2.0 would demonstrate and guide how results are integrated into clinical care after they are received. This Consortium is prepared to tackle the refinement of clinical sequencing pipelines including: inter-operability of EMR systems, a focus on rapid turnaround time for urgent clinical scenarios (e.g., newborn in the NICU, unexplained heart failure, or tumor progression through initial treatment), automation of data sharing from labs to national databases, and establishing clinical decision support systems.
- **Investigation of the use of clinical sequencing within larger, more diverse populations.** The CSER research program would benefit from larger sample sizes, including a special focus on recruiting patients from diverse and underserved ancestry groups, more diverse regional and clinical settings, and healthy populations. Anticipation of population-scale clinical sequencing requires efforts to evaluate “healthy” populations and discovery of medically relevant variants in the absence of specific phenotypic indications. It is also important to expand access to genomic testing well beyond the confines of large academic medical centers in major population centers and to form partnerships.
- **Determination of appropriate use of genome and exome sequencing.** There is a need to understand how large-scale sequencing (either panel-based, exome or genome) are currently being used (or misused) in clinical settings and identify areas to innovate that optimize benefits and reduce unnecessary/inappropriate use.
- **Iterative phenotyping.** Sequencing provides an opportunity to perform hypothesis-generating research through iterative phenotyping. A key flaw in clinical genomics is incomplete phenotyping of patients studied for one specific indication. We suffer from a lack of a full understanding of the phenotypic spectrum of variants in the genes that are already associated with disease and the association of other genes with phenotypes may not be known. Current and future CSER cohorts could be utilized to deeply phenotype patients who are identified to have mutations in particular genes, including the use of longitudinal phenotyping and wearable sensors.

ELSI, Return of Results and Regulatory Efforts

- **Conduct biopsychosocial research.** Further biopsychosocial research is needed to ensure that the adoption of clinical sequencing tests takes place while maximizing benefits and mitigating harm to patients. Examples include improving the communication about the biopsychosocial dimensions of sequencing and supporting education and decision-making for both practitioners and patients during informed consent and when delivering results, especially uncertain test results.
- **Continue ELSI investigations.** The ethical, legal, and social implications of genetic testing will remain an important component of the CSER Consortium in the future. Legal and regulatory issues are particularly dynamic given FDA interest and changes to the Common Rule. Possible areas of research include 1) long term outcomes of testing in adolescents transitioning to adulthood, 2) testing new approaches (e.g., ways to give patients and clinicians choices of results) to support patient decision-making while minimizing bias, and 3) understand and impact legal ramifications of wide-spread genomic testing and the evolving regulatory climate.
- **Legal and Regulatory efforts**. With changes in the Common Rule and FDA regulation of genomic testing, CSER’s continued efforts to analyze, educate, and inform these changes are essential.

APPENDIX A: CSER PROJECTS

A more detailed description of all CSER projects can be found on the **CSER Consortium website**.

CSER U-Grants

BASIC³: Baylor College of Medicine | Sharon Plon & Will Parsons**

Examining the diagnostic yield, clinical utility and impact of whole exome sequencing (WES) on physicians and families of newly diagnosed pediatric cancer patients

MedSeq: Brigham & Women's Hospital | Robert C. Green

Whole genome sequencing (WGS) targeting healthy adults from primary care and cardiomyopathy patients to evaluate WGS-derived information in patient care compared against current care standards

PediSeq: Children's Hospital of Philadelphia | Ian Krantz & Nancy Spinner

WES in children with one of four conditions: intellectual disability, hearing loss, sudden cardiac arrest/death and mitochondrial disorders

CanSeq: Dana-Farber Cancer Institute | Levi Garraway & Pasi Jane

Introduction of WES into the care of lung and colorectal cancer

NCGENES: University of North Carolina | James P. Evans

Evaluating the diagnostic usefulness of WES in diverse clinical domains, such as dysmorphology, neurological disorders and familial cancer

NEXT Medicine: University of Washington | Gail Jarvik**

Investigating if WES can identify the genetic causes of colorectal cancer/polyps more often than usual clinic care

Hudson-Alpha Institute for Biotechnology | Greg Cooper &* Richard Myers*

Introducing WES into diagnostic needs of children with developmental delay and/or intellectual disability

NEXTGen: Kaiser Foundation Research Institute | Katrina Goddard & Benjamin Wilfond*

Introducing WGS into pre-conception carrier status testing for women and their partners

MI-ONCOSEQ: University of Michigan | Arul Chinnaiyan*

Application of high throughput sequencing towards precision cancer medicine

** Added in 2013*

*** Co-funded by the National Cancer Institute*

NIH Intramural Research

ClinSeq: National Human Genome Research Institute | Leslie Biesecker

WGS in health adults and adults with heart disease

CSER R-Grants

Mayo Clinic | Richard Sharp

Presenting diagnostic results from large-scale clinical mutation testing

Columbia University | Paul S. Appelbaum

Developing a menu of potential approaches to informed consent for the return of incidental findings

Columbia University | Wendy K. Chung

Impact of return of incidental genetic test results to research participants in the genomic era

Children's Hospital Boston | Ingrid A. Holm

Returning research results in children: Parental Preferences and Expert Oversight

Children's Mercy Bioethics Center | Jeremy R. Garrett

The presumptive case against returning individual results in biobanking research

John Hopkins University | Michelle Lewis

Return of research results from samples obtained for newborn screening

University of California, San Francisco, Mayo Clinic & University of Minnesota | Gloria Petersen, Barbara Koenig & Susan Wolf**

Disclosing incidental findings in a cancer biobank

Seattle Children's Hospital | Holly K. Tabor

Innovative approaches to returning results in exome and genome sequencing studies

Vanderbilt University | Ellen Wright Clayton

Returning research results of pediatric genomic research to participants

Coordinating Center

University of Washington | Gail Jarvik, Debbie Nickerson, Wylie Burke & Peter Tarcyz-Hornoch*

The CSER Program's Coordinating Center

APPENDIX B: SELECTED CSER PUBLICATIONS

Site-specific Publications

These are the three papers which the U-Grant sites deemed most impactful from their work:

CanSeq | Dana-Farber Cancer Institute

- Gray SW, Martins Y, Feuerman LZ, Bernhardt BA, Biesecker BB, Christensen KD, et al. Social and behavioral research in genomic sequencing: approaches from the Clinical Sequencing Exploratory Research Consortium Outcomes and Measures Working Group. *Genetics in medicine : official journal of the American College of Medical Genetics*. 2014;16(10):727-35. Epub 2014/03/15. doi: 10.1038/gim.2014.26. PubMed PMID: 24625446;PMC4163120.
- Van Allen EM, Wagle N, Stojanov P, Perrin DL, Cibulskis K, Marlow S, et al. Whole-exome sequencing and clinical interpretation of formalin-fixed, paraffin-embedded tumor samples to guide precision cancer medicine. *Nature medicine*. 2014;20(6):682-8. Epub 2014/05/20. doi: 10.1038/nm.3559. PubMed PMID: 24836576;PMC4048335.
- Gray SW, Hicks-Courant K, Cronin A, Rollins BJ, Weeks JC. Physicians' attitudes about multiplex tumor genomic testing. Journal of clinical oncology : official journal of the American Society of Clinical Oncology. 2014;32(13):1317-23. Epub 2014/03/26. doi: 10.1200/jco.2013.52.4298. PubMed PMID: 24663044;PMC3992721.

NEXT Medicine | University of Washington

- Amendola LM, Dorschner MO, Robertson PD, Salama JS, Hart R, Shirts BH, et al. Actionable exomic incidental findings in 6503 participants: challenges of variant classification. *Genome research*. 2015;25(3):305-15. Epub 2015/02/01. doi: 10.1101/gr.183483.114. PubMed PMID: 25637381; PMC4352885.
- Gallego CJ, Shirts BH, Bennette CS, Guzauskas G, Amendola LM, Horike-Pyne M, et al. Next-Generation Sequencing Panels for the Diagnosis of Colorectal Cancer and Polyposis Syndromes: A Cost-Effectiveness Analysis. *Journal of clinical oncology : official journal of the American Society of Clinical Oncology*. 2015;33(18):2084-91. Epub 2015/05/06. doi: 10.1200/jco.2014.59.3665. PubMed PMID: 25940718; PMC4461806.
- Evans BJ, Burke W, Jarvik GP. The FDA and genomic tests--getting regulation right. *New England Journal of Medicine*. 2015;372(23):2258-64. doi: 10.1056/NEJMsr1501194. PubMed PMID: 26014592PMCID: PMC4464691

MedSeq | Brigham & Women's Hospital

- McLaughlin HM, Ceyhan-Birsoy O, Christensen KD, Kohane IS, Krier J, Lane WJ, Lautenbach D, Lebo MS, Machini K, MacRae CA, Azzariti DR, Murray MF, Seidman CE, Vassy JL, Green RC, Rehm HL, MedSeq P: A systematic approach to the reporting of medically relevant findings from whole genome sequencing. BMC Med Genet. 2014; 15(1):134-46.
- Biesecker LG, Green RC. Diagnostic clinical genome and exome sequencing. *The New England journal of medicine*. 2014;370(25):2418-25. Epub 2014/06/19. doi: 10.1056/NEJMra1312543. PubMed PMID: 24941179
- Vassy JL, Korf BR, Green RC: How will we know when doctors are ready for genomic medicine? Sci Trans Med. 2015, May 7(287):287fs19. doi: 10.1126/scitranslmed.aaa2401.

NCGenes | University of North Carolina

- Berg JS, Adams M, Nassar N, Bizon C, Lee K, Schmitt CP, et al. An informatics approach to analyzing the incidentalome. *Genetics in medicine : official journal of the American College of Medical Genetics*. 2013;15(1):36-44. Epub 2012/09/22. doi: 10.1038/gim.2012.112. PubMed PMID: 22995991;PMC3538953.
- Henderson GE, Wolf SM, Kuczynski KJ, Joffe S, Sharp RR, Parsons DW, et al. The challenge of informed consent and return of results in translational genomics: empirical analysis and recommendations. *The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics*. 2014;42(3):344-55. Epub 2014/09/30. doi: 10.1111/jlme.12151. PubMed PMID: 25264092;PMC4262925.

PediSeq | Children's Hospital of Philadelphia

- Li MH, Abrudan JL, Dulik MC, Sasson A, Brunton J, Jayaraman V, et al. Utility and limitations of exome sequencing as a genetic diagnostic tool for conditions associated with pediatric sudden cardiac arrest/sudden cardiac death. *Human genomics*. 2015;9:15. Epub 2015/07/19. doi: 10.1186/s40246-015-0038-y. PubMed PMID: 26187847;PMC4506570.
- Bernhardt BA, Roche MI, Perry DL, Scollon SR, Tomlinson AN, Skinner D. Experiences with obtaining informed consent for genomic sequencing. *American journal of medical genetics Part A*. 2015. Epub 2015/07/23. doi: 10.1002/ajmg.a.37256. PubMed PMID: 26198374
- Levenseller BL, Soucier DJ, Miller VA, Harris D, Conway L, Bernhardt BA. Stakeholders' opinions on the implementation of pediatric whole exome sequencing: implications for informed consent. Journal of genetic counseling. 2014;23(4):552-65. Epub 2013/07/13. doi: 10.1007/s10897-013-9626-y. PubMed PMID: 23846343;PMC3849137.

Hudson-Alpha Intsitute for Biotechnology

- Kircher M, Witten DM, Jain P, O'roak BJ, Cooper GM, Shendure J. A general framework for estimating the relative pathogenicity of human genetic variants. Nat Genet. 2014;46(3):310-5. PubMed PMID: 24487276
- Mccullough LB, Brothers KB, Chung WK, et al. Professionally Responsible Disclosure of Genomic Sequencing Results in Pediatric Practice. Pediatrics. 2015. PubMed PMID: 26371191
- Brothers KB, Rothstein MA. Ethical, legal and social implications of incorporating personalized medicine into healthcare. Per Med. 2015;12(1):43-51. PubMed PMID: 25601880

ClinSeq | National Human Genome Research Institute

- Johnston JJ, Lewis KL, Ng D, Singh LN, Wynter J, Brewer C, et al. Individualized iterative phenotyping for genome-wide analysis of loss-of-function mutations. *American journal of human genetics*. 2015;96(6):913-25. Epub 2015/06/06. doi: 10.1016/j.ajhg.2015.04.013. PubMed PMID: 26046366;PMC4457956.
- Green RC, Berg JS, Grody WW, Kalia SS, Korf BR, Martin CL, et al. ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(7):565-74. Epub 2013/06/22. doi: 10.1038/gim.2013.73. PubMed PMID: 23788249;PMC3727274.
- Johnston JJ, Rubinstein WS, Facio FM, Ng D, Singh LN, Teer JK, et al. Secondary variants in individuals undergoing exome sequencing: screening of 572 individuals identifies high-penetrance mutations in cancer-susceptibility genes. *American journal of human genetics*. 2012;91(1):97-108. Epub 2012/06/19. doi: 10.1016/j.ajhg.2012.05.021. PubMed PMID: 22703879;PMC3397257.

MI-ONCOSEQ | University of Michigan

- Robinson DR, Wu YM, Vats P, Su F, Lonigro RJ, Cao X, et al. Activating ESR1 mutations in hormone-resistant metastatic breast cancer. *Nature genetics*. 2013;45(12):1446-51. Epub 2013/11/05. doi: 10.1038/ng.2823. PubMed PMID: 24185510;PMC4009946.
- Robinson D, Van Allen EM, Wu YM, Schultz N, Lonigro RJ, Mosquera JM, et al. Integrative clinical genomics of advanced prostate cancer. *Cell*. 2015;161(5):1215-28. Epub 2015/05/23. doi: 10.1016/j.cell.2015.05.001. PubMed PMID: 26000489;PMC4484602.
- Mody RJ, Wu YM, Lonigro RJ, Cao X, Roychowdhury S, Vats P, Frank KM, Prensner JR, Asangani I, Palanisamy N, Dillman JR, Rabah RM, Kunju LP, Everett J, Raymond VM, Ning Y, Su F, Wang R, Stoffel EM, Innis JW, Roberts JS, Robertson PL, Yanik G, Chamdin A, Connelly JA, Choi S, Harris AC, Kitko C, Rao RJ, Levine JE, Castle VP, Hutchinson RJ, Talpaz M, Robinson DR, Chinnaiyan AM. Integrative Clinical Sequencing in the Management of Refractory or Relapsed Cancer in Youth. JAMA. 2015 Sep 1;314(9):913-25. doi: 10.1001/jama.2015.10080. PubMed PMID: 26325560.

BASIC3 | Baylor College of Medicine

- Scollon S, Bergstrom K, Kerstein RA, Wang T, Hilsenbeck SG, Ramamurthy U, et al. Obtaining informed consent for clinical tumor and germline exome sequencing of newly diagnosed childhood cancer patients. *Genome medicine*. 2014;6(9):69. Epub 2014/10/16. doi: 10.1186/s13073-014-0069-3. PubMed PMID: 25317207;PMC4195891.
- Parsons DW, Roy A, Plon SE, Roychowdhury S, Chinnaiyan AM. Clinical tumor sequencing: an incidental casualty of the American College of Medical Genetics and Genomics recommendations for reporting of incidental findings. Journal of clinical oncology : official journal of the American Society of Clinical Oncology. 2014;32(21):2203-5. Epub 2014/06/25. doi: 10.1200/jco.2013.54.8917. PubMed PMID: 24958819;PMC4188159.
- Yang Y, Muzny DM, Reid JG, Bainbridge MN, Willis A, Ward PA, et al. Clinical whole-exome sequencing for the diagnosis of mendelian disorders. *The New England journal of medicine*. 2013;369(16):1502-11. Epub 2013/10/04. doi: 10.1056/NEJMoa1306555. PubMed PMID: 24088041;PMC4211433.

NEXTGen | Kaiser Foundation Research Institute

- Amendola LM, Dautenbach D, Scollon S ,Bernhardt B, Biswas S, East K, Everett J, Gilmore MJ, Himes P, Raymond VM, Wynn J, Hart R for the CSER Genetic Counseling Working Group, and Jarvik GP. Illustrative Case Studies in the Return of Exome and Genome Sequencing Results. Personalized Medicine. Vol. 12, No. 3, Pages 283-295.
- Schneider JL, Goddard KAB, Davis J, Wilfond B, Kauffman TL, Reiss JA, Gilmore M, Himes P, Lynch FL, Leo MC, McMullen C. “Is it worth knowing? Focus group participants’ perceived utility of genome-scale preconception carrier screening.” Journal of Genetic Counseling. Epub 2015, June 21.
- Amendola LM, Dorschner MO, Robertson PD, Salama JS, Hart R, Shirts BH, Murray ML, Tokita MJ, Gallego CJ, Kim DS, Bennett JT, Crosslin DR, Ranchalis J, Jones KL, Rosenthal EA, Jarvik ER, Itsara A, Turner EH, Herman DS, Schleit J, Burt A, Jamal SM, et al. Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome Res. 2015 Mar;25(3):305-15. doi: 10.1101/gr.183483.114. Epub 2015 Jan 30.

APPENDIX C: CSER PUBLICATIONS

The following list includes all peer-reviewed CSER publications, publications currently in press, along with textbook chapters, op-eds, online news articles, journal commentaries and other forms of publications.

1. Zhou S, Hertel PM, Finegold MJ, Wang L, Kerkar N, Wang J, et al. Hepatocellular carcinoma associated with tight-junction protein 2 deficiency. Hepatology (Baltimore, Md). 2015. Epub 2015/04/30. doi: 10.1002/hep.27872. PubMed PMID: 25921221
2. Wynn J, Martinez J, Duong J, Zhang Y, Phelan J, Fyer A, et al. Association of Researcher Characteristics with Views on Return of Incidental Findings from Genomic Research. Journal of genetic counseling. 2015;24(5):833-41. Epub 2015/01/17. doi: 10.1007/s10897-014-9817-1. PubMed PMID: 25592144;PMC4506262.
3. Wynn J. Genomic Testing: a Genetic Counselor's Personal Reflection on Three Years of Consenting and Testing. Journal of genetic counseling. 2015. Epub 2015/08/06. doi: 10.1007/s10897-015-9868-y. PubMed PMID: 26242468
4. Wilfond BS, Goddard KA. It's complicated: criteria for policy decisions for the clinical integration of genome-scale sequencing for reproductive decision making. Molecular genetics & genomic medicine. 2015;3(4):239-42. Epub 2015/08/08. doi: 10.1002/mgg3.130. PubMed PMID: 26247041;PMC4521960.
5. Wassif CA, Cross JL, Iben J, Sanchez-Pulido L, Cougnoux A, Platt FM, et al. High incidence of unrecognized visceral/neurological late-onset Niemann-Pick disease, type C1, predicted by analysis of massively parallel sequencing data sets. Genetics in medicine : official journal of the American College of Medical Genetics. 2015. Epub 2015/03/13. doi: 10.1038/gim.2015.25. PubMed PMID: 25764212;PMC4486368.
6. Waisbren SE, Back DK, Liu C, Kalia SS, Ringer SA, Holm IA, et al. Parents are interested in newborn genomic testing during the early postpartum period. Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(6):501-4. Epub 2014/12/05. doi: 10.1038/gim.2014.139. PubMed PMID: 25474344;PMC4452417.
7. Vassy JL, McLaughlin HL, MacRae CA, Seidman CE, Lautenbach D, Krier JB, et al. A one-page summary report of genome sequencing for the healthy adult. Public health genomics. 2015;18(2):123-9. Epub 2015/01/24. doi: 10.1159/000370102. PubMed PMID: 25612602;PMC4348325.
8. Vassy JL, Korf BR, Green RC. How to know when physicians are ready for genomic medicine. Science translational medicine. 2015;7(287):287fs19. Epub 2015/05/15. doi: 10.1126/scitranslmed.aaa2401. PubMed PMID: 25971999;PMC4519005.
9. Vassy JL, Christensen KD, Slashinski MJ, Lautenbach DM, Raghavan S, Robinson JO, et al. 'Someday it will be the norm': physician perspectives on the utility of genome sequencing for patient care in the MedSeq Project. Personalized medicine. 2015;12(1):23-32. Epub 2015/02/03. doi: 10.2217/pme.14.68. PubMed PMID: 25642274;PMC4306284.
10. Tomlinson AN, Skinner D, Perry DL, Scollon SR, Roche MI, Bernhardt BA. "Not Tied Up Neatly with a Bow": Professionals' Challenging Cases in Informed Consent for Genomic Sequencing. Journal of genetic counseling. 2015. Epub 2015/04/26. doi: 10.1007/s10897-015-9842-8. PubMed PMID: 25911622
11. Taber JM, Klein WM, Ferrer RA, Lewis KL, Harris PR, Shepperd JA, et al. Information Avoidance Tendencies, Threat Management Resources, and Interest in Genetic Sequencing Feedback. Annals of behavioral medicine : a publication of the Society of Behavioral Medicine. 2015;49(4):616-21. Epub 2015/01/15. doi: 10.1007/s12160-014-9679-7. PubMed PMID: 25582989;PMC4498968.
12. Taber JM, Klein WM, Ferrer RA, Lewis KL, Biesecker LG, Biesecker BB. Dispositional optimism and perceived risk interact to predict intentions to learn genome sequencing results. Health psychology : official journal of the Division of Health Psychology, American Psychological Association. 2015;34(7):718-28. Epub 2014/10/15. doi: 10.1037/hea0000159. PubMed PMID: 25313897;PMC4395524.
13. Taber JM, Klein WM, Ferrer RA, Han PK, Lewis KL, Biesecker LG, et al. Perceived ambiguity as a barrier to intentions to learn genome sequencing results. Journal of behavioral medicine. 2015;38(5):715-26. Epub 2015/05/25. doi: 10.1007/s10865-015-9642-5. PubMed PMID: 26003053;PMC4568142.
14. Shirts BH, Salama JS, Aronson SJ, Chung WK, Gray SW, Hindorff LA, et al. CSER and eMERGE: current and potential state of the display of genetic information in the electronic health record. Journal of the American Medical Informatics Association : JAMIA. 2015. Epub 2015/07/05. doi: 10.1093/jamia/ocv065. PubMed PMID: 26142422
15. Schneider JL, Goddard KA, Davis J, Wilfond B, Kauffman TL, Reiss JA, et al. "Is It Worth Knowing?" Focus Group Participants' Perceived Utility of Genomic Preconception Carrier Screening. Journal of genetic counseling. 2015. Epub 2015/06/22. doi: 10.1007/s10897-015-9851-7. PubMed PMID: 26093606
16. Evans BJ, Burke W, Jarvik GP. The FDA and genomic tests--getting regulation right. New England Journal of Medicine. 2015;372(23):2258-64. doi: 10.1056/NEJMsr1501194. PubMed PMID: 26014592PMCID: PMC4464691
17. Scheuner MT, Peredo J, Benkendorf J, Bowdish B, Feldman G, Fleisher L, et al. Reporting genomic secondary findings: ACMG members weigh in. Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(1):27-35. Epub 2014/11/14. doi: 10.1038/gim.2014.165. PubMed PMID: 25394173
18. Aronson SJ, Rehm HL. Building the foundation for genomics in precision medicine. Nature. 2015;513:1-7
19. Robinson D, Van Allen EM, Wu YM, Schultz N, Lonigro RJ, Mosquera JM, et al. Integrative clinical genomics of advanced prostate cancer. Cell. 2015;161(5):1215-28. Epub 2015/05/23. doi: 10.1016/j.cell.2015.05.001. PubMed PMID: 26000489;PMC4484602.
20. Rinaldi C, Schmidt T, Situ AJ, Johnson JO, Lee PR, Chen KL, et al. Mutation in CPT1C Associated With Pure Autosomal Dominant Spastic Paraplegia. JAMA neurology. 2015;72(5):561-70. Epub 2015/03/10. doi: 10.1001/jamaneurol.2014.4769. PubMed PMID: 25751282
21. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(5):405-24. Epub 2015/03/06. doi: 10.1038/gim.2015.30. PubMed PMID: 25741868;PMC4544753.

22. Richards EG, Sangi-Haghpeykar H, McGuire AL, Van den Veyver IB, Fruhman G. Pregnant patients' risk perception of prenatal test results with uncertain fetal clinical significance: ultrasound versus advanced genetic testing. Prenatal diagnosis. 2015. Epub 2015/08/20. doi: 10.1002/pd.4680. PubMed PMID: 26287960
23. Rehm HL, Berg JS, Brooks LD, Bustamante CD, Evans JP, Landrum MJ, et al. ClinGen--the Clinical Genome Resource. The New England journal of medicine. 2015;372(23):2235-42. Epub 2015/05/28. doi: 10.1056/NEJMsr1406261. PubMed PMID: 26014595;PMC4474187.
24. Regier DA, Peacock SJ, Pataky R, van der Hoek K, Jarvik GP, Hoch J, et al. Societal preferences for the return of incidental findings from clinical genomic sequencing: a discrete-choice experiment. CMAJ : Canadian Medical Association journal = journal de l'Association medicale canadienne. 2015;187(6):E190-7. Epub 2015/03/11. doi: 10.1503/cmaj.140697. PubMed PMID: 25754703;PMC4387060.
25. Prince AE, Berg JS, Evans JP, Jonas DE, Henderson G. Genomic screening of the general adult population: key concepts for assessing net benefit with systematic evidence reviews. Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(6):441-3. Epub 2014/09/19. doi: 10.1038/gim.2014.129. PubMed PMID: 25232850;PMC4387105.
26. Peters TL, Kumar V, Polikepahad S, Lin FY, Sarabia SF, Liang Y, et al. BCOR-CCNB3 fusions are frequent in undifferentiated sarcomas of male children. Modern pathology : an official journal of the United States and Canadian Academy of Pathology, Inc. 2015;28(4):575-86. Epub 2014/11/02. doi: 10.1038/modpathol.2014.139. PubMed PMID: 25360585;PMC4385430.
27. Nishimura AA, Shirts BH, Dorschner MO, Amendola LM, Smith JW, Jarvik GP, et al. Development of clinical decision support alerts for pharmacogenomic incidental findings from exome sequencing. Genetics in medicine : official journal of the American College of Medical Genetics. 2015. Epub 2015/03/06. doi: 10.1038/gim.2015.5. PubMed PMID: 25741865
28. Mody RJ, Wu YM, Lonigro RJ, Cao X, Roychowdhury S, Vats P, et al. Integrative Clinical Sequencing in the Management of Refractory or Relapsed Cancer in Youth. Jama. 2015;314(9):913-25. Epub 2015/09/02. doi: 10.1001/jama.2015.10080. PubMed PMID: 26325560
29. McCullough LB, Brothers KB, Chung WK, Joffe S, Koenig BA, Wilfond B, et al. Professionally Responsible Disclosure of Genomic Sequencing Results in Pediatric Practice. Pediatrics. 2015. Epub 2015/09/16. doi: 10.1542/peds.2015-0624. PubMed PMID: 26371191
30. May T, Strong KA, Khoury MJ, Evans JP. Can targeted genetic testing offer useful health information to adoptees? Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(7):533-5. Epub 2015/04/24. doi: 10.1038/gim.2015.58. PubMed PMID: 25905442
31. Lewis KL, Han PK, Hooker GW, Klein WM, Biesecker LG, Biesecker BB. Characterizing Participants in the ClinSeq Genome Sequencing Cohort as Early Adopters of a New Health Technology. PloS one. 2015;10(7):e0132690. Epub 2015/07/18. doi: 10.1371/journal.pone.0132690. PubMed PMID: 26186621;PMC4506048.
32. Lee K, Berg JS, Milko L, Crooks K, Lu M, Bizon C, et al. High Diagnostic Yield of Whole Exome Sequencing in Participants With Retinal Dystrophies in a Clinical Ophthalmology Setting. American journal of ophthalmology. 2015;160(2):354-63.e9. Epub 2015/04/26. doi: 10.1016/j.ajo.2015.04.026. PubMed PMID: 25910913;PMC4506879.
33. Lazaro-Munoz G, Conley JM, Davis AM, Van Riper M, Walker RL, Juengst ET. Looking for Trouble: Preventive Genomic Sequencing in the General Population and the Role of Patient Choice. The American journal of bioethics : AJOB. 2015;15(7):3-14. Epub 2015/07/07. doi: 10.1080/15265161.2015.1039721. PubMed PMID: 26147254;PMC4493927.
34. Kong SW, Lee IH, Leshchiner I, Krier J, Kraft P, Rehm HL, et al. Summarizing polygenic risks for complex diseases in a clinical whole-genome report. Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(7):536-44. Epub 2014/10/24. doi: 10.1038/gim.2014.143. PubMed PMID: 25341114;PMC4547452.
35. Kohane IS. An autism case history to review the systematic analysis of large-scale data to refine the diagnosis and treatment of neuropsychiatric disorders. Biological psychiatry. 2015;77(1):59-65. Epub 2014/07/19. doi: 10.1016/j.biopsych.2014.05.024. PubMed PMID: 25034947;PMC4260993.
36. Trinidad SB, Fullerton SM, Burke W. Looking for Trouble and Finding It. The American Journal of Bioethics 2015 Jul;15(7):15–17.
37. Roche MI, Berg JS. Incidental Findings with Genomic Testing: Implications for Genetic Counseling Practice [Internet]. Current Genetic Medicine Reports 2015 Aug;
38. Amendola LM, Lautenbach D, Scollon S, Bernhardt B, Biswas S, East K, Everett J, Gilmore MJ, Himes P, Raymond VM, Wynn J, Hart R, CSER Genetic Counseling Working Group, Jarvik GP. Illustrative case studies in the return of exome and genome sequencing results. Personalized Medicine 2015 Jun;12(3):283–295.
39. Khoury MJ, Evans JP. A public health perspective on a national precision medicine cohort: balancing long-term knowledge generation with early health benefit. Jama. 2015;313(21):2117-8. Epub 2015/06/04. doi: 10.1001/jama.2015.3382. PubMed PMID: 26034952
40. Khan CM, Rini C, Bernhardt BA, Roberts JS, Christensen KD, Evans JP, et al. How can psychological science inform research about genetic counseling for clinical genomic sequencing? Journal of genetic counseling. 2015;24(2):193-204. Epub 2014/12/10. doi: 10.1007/s10897-014-9804-6. PubMed PMID: 25488723
41. Keppler-Noreuil KM, Rios JJ, Parker VE, Semple RK, Lindhurst MJ, Sapp JC, et al. PIK3CA-related overgrowth spectrum (PROS): diagnostic and testing eligibility criteria, differential diagnosis, and evaluation. American journal of medical genetics Part A. 2015;167a(2):287-95. Epub 2015/01/06. doi: 10.1002/ajmg.a.36836. PubMed PMID: 25557259;PMC4480633.
42. Katsanis SH, Minear MA, Vorderstrasse A, Yang N, Reeves JW, Rakhra-Burris T, et al. Perspectives on genetic and genomic technologies in an academic medical center: the duke experience. Journal of personalized medicine. 2015;5(2):67-82. Epub 2015/04/10. doi: 10.3390/jpm5020067. PubMed PMID: 25854543;PMC4493486.
43. Johnston JJ, Lewis KL, Ng D, Singh LN, Wynter J, Brewer C, et al. Individualized iterative phenotyping for genome-wide analysis of loss-of-function mutations. American journal of human genetics. 2015;96(6):913-25. Epub 2015/06/06. doi: 10.1016/j.ajhg.2015.04.013. PubMed PMID: 26046366;PMC4457956.
44. Janssens AC, Evans JP. Returning pharmacogenetic secondary findings from genome sequencing: let's not put the cart before the horse. Genetics in medicine : official journal of the American College of Medical Genetics. 2015. Epub 2015/05/08. doi: 10.1038/gim.2015.59. PubMed PMID: 25950734

45. Howard HC, Knoppers BM, Cornel MC, Wright Clayton E, Senecal K, Borry P. Whole-genome sequencing in newborn screening? A statement on the continued importance of targeted approaches in newborn screening programmes. European journal of human genetics : EJHG. 2015. Epub 2015/01/30. doi: 10.1038/ejhg.2014.289. PubMed PMID: 25626707
46. Harms PW, Vats P, Verhaegen ME, Robinson DR, Wu YM, Dhanasekaran SM, et al. The Distinctive Mutational Spectra of Polyomavirus-Negative Merkel Cell Carcinoma. Cancer research. 2015;75(18):3720-7. Epub 2015/08/05. doi: 10.1158/0008-5472.can-15-0702. PubMed PMID: 26238782
47. Haase R, Michie M, Skinner D. Flexible positions, managed hopes: the promissory bioeconomy of a whole genome sequencing cancer study. Social science & medicine (1982). 2015;130:146-53. Epub 2015/02/24. doi: 10.1016/j.socscimed.2015.02.016. PubMed PMID: 25697637;PMC4363274.
48. Green RC, Lautenbach D, McGuire AL. GINA, genetic discrimination, and genomic medicine. The New England journal of medicine. 2015;372(5):397-9. Epub 2015/01/30. doi: 10.1056/NEJMp1404776. PubMed PMID: 25629736
49. Gray SW, Cronin A, Bair E, Lindeman N, Viswanath V, Janeway KA. Marketing of personalized cancer care on the web: an analysis of Internet websites. Journal of the National Cancer Institute. 2015;107(5). Epub 2015/03/07. doi: 10.1093/jnci/djv030. PubMed PMID: 25745021
50. Garrett SB, Dohan D, Koenig BA. Linking Broad Consent to Biobank Governance: Support From a Deliberative Public Engagement in California. The American journal of bioethics : AJOB. 2015;15(9):56-7. Epub 2015/08/26. doi: 10.1080/15265161.2015.1062177. PubMed PMID: 26305757
51. Garrett JR. Collectivizing rescue obligations in bioethics. The American journal of bioethics : AJOB. 2015;15(2):3-11. Epub 2015/02/13. doi: 10.1080/15265161.2014.990163. PubMed PMID: 25674948
52. Garrett JR. Beyond Harms and Benefits: Rethinking Duties to Disclose Misattributed Parentage. The Hastings Center report. 2015;45(4):37-8. Epub 2015/07/15. doi: 10.1002/hast.472. PubMed PMID: 26152387
53. Gambin T, Jhangiani SN, Below JE, Campbell IM, Wiszniewski W, Muzny DM, et al. Secondary findings and carrier test frequencies in a large multiethnic sample. Genome medicine. 2015;7(1):54. Epub 2015/07/22. doi: 10.1186/s13073-015-0171-1. PubMed PMID: 26195989;PMC4507324.
54. Gallego CJ, Shirts BH, Bennette CS, Guzauskas G, Amendola LM, Horike-Pyne M, et al. Next-Generation Sequencing Panels for the Diagnosis of Colorectal Cancer and Polyposis Syndromes: A Cost-Effectiveness Analysis. Journal of clinical oncology : official journal of the American Society of Clinical Oncology. 2015;33(18):2084-91. Epub 2015/05/06. doi: 10.1200/jco.2014.59.3665. PubMed PMID: 25940718;PMC4461806.
55. Evans JP, Watson MS. Genetic testing and FDA regulation: overregulation threatens the emergence of genomic medicine. Jama. 2015;313(7):669-70. Epub 2015/01/07. doi: 10.1001/jama.2014.18145. PubMed PMID: 25560537
56. Evans BJ. The Limits of FDA's Authority to Regulate Clinical Research Involving High-Throughput DNA Sequencing. Food and drug law journal. 2015;70(2):259-87, ii. Epub 2015/08/26. PubMed PMID: 26302600
57. De Castro M, Johnston J, Biesecker L. Determining the prevalence of McArdle disease from gene frequency by analysis of next-generation sequencing data. Genetics in medicine : official journal of the American College of Medical Genetics. 2015. Epub 2015/03/06. doi: 10.1038/gim.2015.9. PubMed PMID: 25741863;PMC4561039.
58. Couser NL, Masood MM, Strande NT, Foreman AK, Crooks K, Weck KE, et al. The phenotype of multiple congenital anomalies-hypotonia-seizures syndrome 1: Report and review. American journal of medical genetics Part A. 2015;167(9):2176-81. Epub 2015/04/30. doi: 10.1002/ajmg.a.37129. PubMed PMID: 25920937
59. Cieslik M, Chugh R, Wu YM, Wu M, Brennan C, Lonigro R, et al. The use of exome capture RNA-seq for highly degraded RNA with application to clinical cancer sequencing. Genome research. 2015;25(9):1372-81. Epub 2015/08/09. doi: 10.1101/gr.189621.115. PubMed PMID: 26253700;PMC4561495.
60. Christensen KD, Vassy JL, Jamal L, Lehmann LS, Slashinski MJ, Perry DL, et al. Are physicians prepared for whole genome sequencing? a qualitative analysis. Clinical genetics. 2015. Epub 2015/06/18. doi: 10.1111/cge.12626. PubMed PMID: 26080898
61. Christensen KD, Roberts JS, Zikmund-Fisher BJ, Kardia SL, McBride CM, Linnenbringer E, et al. Associations between self-referral and health behavior responses to genetic risk information. Genome medicine. 2015;7(1):10. Epub 2015/02/03. doi: 10.1186/s13073-014-0124-0. PubMed PMID: 25642295;PMC4311425.
62. Carere DA, Kraft P, Kaphingst KA, Roberts JS, Green RC. Consumers report lower confidence in their genetics knowledge following direct-to-consumer personal genomic testing. Genetics in medicine : official journal of the American College of Medical Genetics. 2015. Epub 2015/03/27. doi: 10.1038/gim.2015.34. PubMed PMID: 25812042
63. Burke W, Korngiebel DM. Closing the gap between knowledge and clinical application: challenges for genomic translation. PLoS genetics. 2015;11(2):e1004978. Epub 2015/02/27. doi: 10.1371/journal.pgen.1004978. PubMed PMID: 25719903;PMC4342348.
64. Burke W, Appelbaum P, Dame L, Marshall P, Press N, Pyeritz R, et al. The translational potential of research on the ethical, legal, and social implications of genomics. Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(1):12-20. Epub 2014/06/20. doi: 10.1038/gim.2014.74. PubMed PMID: 24946153;PMC4272334.
65. Brothers KB, Rothstein MA. Ethical, legal and social implications of incorporating personalized medicine into healthcare. Personalized medicine. 2015;12(1):43-51. Epub 2015/01/21. doi: 10.2217/pme.14.65. PubMed PMID: 25601880;PMC4296905.
66. Bonora M, Wieckowsk MR, Chinopoulos C, Kepp O, Kroemer G, Galluzzi L, et al. Molecular mechanisms of cell death: central implication of ATP synthase in mitochondrial permeability transition. Oncogene. 2015;34(12):1608. Epub 2015/03/20. doi: 10.1038/onc.2014.462. PubMed PMID: 25790189
67. Blumenthal-Barby JS, McGuire AL, Green RC, Ubel PA. How behavioral economics can help to avoid 'The last mile problem' in whole genome sequencing. Genome medicine. 2015;7(1):3. Epub 2015/01/24. doi: 10.1186/s13073-015-0132-8. PubMed PMID: 25614766;PMC4302430.

68. Bernhardt BA, Roche MI, Perry DL, Scollon SR, Tomlinson AN, Skinner D. Experiences with obtaining informed consent for genomic sequencing. American journal of medical genetics Part A. 2015. Epub 2015/07/23. doi: 10.1002/ajmg.a.37256. PubMed PMID: 26198374
69. Berkman BE, Hull SC, Biesecker LG. Scrutinizing the Right Not to Know. The American journal of bioethics : AJOB. 2015;15(7):17-9. Epub 2015/07/07. doi: 10.1080/15265161.2015.1039733. PubMed PMID: 26147256
70. Berg JS, Foreman AK, O'Daniel JM, Booker JK, Boshe L, Carey T, et al. A semiquantitative metric for evaluating clinical actionability of incidental or secondary findings from genome-scale sequencing. Genetics in medicine : official journal of the American College of Medical Genetics. 2015. Epub 2015/08/14. doi: 10.1038/gim.2015.104. PubMed PMID: 26270767
71. Bennette CS, Gallego CJ, Burke W, Jarvik GP, Veenstra DL. The cost-effectiveness of returning incidental findings from next-generation genomic sequencing. Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(7):587-95. Epub 2014/11/14. doi: 10.1038/gim.2014.156. PubMed PMID: 25394171;PMC4430464.
72. Bacon PL, Harris ED, Ziniel SI, Savage SK, Weitzman ER, Green RC, et al. The development of a preference-setting model for the return of individual genomic research results. Journal of empirical research on human research ethics : JERHRE. 2015;10(2):107-20. Epub 2015/03/06. doi: 10.1177/1556264615572092. PubMed PMID: 25742675
73. Appelbaum PS, Fyer A, Klitzman RL, Martinez J, Parens E, Zhang Y, et al. Researchers' views on informed consent for return of secondary results in genomic research. Genetics in medicine : official journal of the American College of Medical Genetics. 2015;17(8):644-50. Epub 2014/12/17. doi: 10.1038/gim.2014.163. PubMed PMID: 25503499;PMC4465418.
74. Amendola LM, Dorschner MO, Robertson PD, Salama JS, Hart R, Shirts BH, et al. Actionable exomic incidental findings in 6503 participants: challenges of variant classification. Genome research. 2015;25(3):305-15. Epub 2015/02/01. doi: 10.1101/gr.183483.114. PubMed PMID: 25637381;PMC4352885.
75. Adams MC, Berg JS, Pearlman MD, Vora NL. Look before you leap: genomic screening in obstetrics and gynecology. Obstetrics and gynecology. 2015;125(6):1299-305. Epub 2015/05/23. doi: 10.1097/aog.0000000000000871. PubMed PMID: 26000500
76. Ziniel SI, Savage SK, Huntington N, Amatruda J, Green RC, Weitzman ER, et al. Parents' preferences for return of results in pediatric genomic research. Public health genomics. 2014;17(2):105-14. Epub 2014/03/20. doi: 10.1159/000358539. PubMed PMID: 24642506;PMC4073487.
77. Yu JH, Harrell TM, Jamal SM, Tabor HK, Bamshad MJ. Attitudes of genetics professionals toward the return of incidental results from exome and whole-genome sequencing. American journal of human genetics. 2014;95(1):77-84. Epub 2014/07/01. doi: 10.1016/j.ajhg.2014.06.004. PubMed PMID: 24975944;PMC4085580.
78. Yang Y, Muzny DM, Xia F, Niu Z, Person R, Ding Y, et al. Molecular findings among patients referred for clinical whole-exome sequencing. Jama. 2014;312(18):1870-9. Epub 2014/10/19. doi: 10.1001/jama.2014.14601. PubMed PMID: 25326635;PMC4326249.
79. Wang C, Gordon ES, Stack CB, Liu CT, Norkunas T, Wawak L, et al. A randomized trial of the clinical utility of genetic testing for obesity: design and implementation considerations. Clinical trials (London, England). 2014;11(1):102-13. Epub 2013/11/13. doi: 10.1177/1740774513508029. PubMed PMID: 24216219;PMC3946398.
80. Walford GA, Porneala BC, Dauriz M, Vassy JL, Cheng S, Rhee EP, et al. Metabolite traits and genetic risk provide complementary information for the prediction of future type 2 diabetes. Diabetes care. 2014;37(9):2508-14. Epub 2014/06/21. doi: 10.2337/dc14-0560. PubMed PMID: 24947790;PMC4140156.
81. Wagner JK, Mozersky JT, Pyeritz RE. "Use it or lose it" as an alternative approach to protect genetic privacy in personalized medicine. Urologic oncology. 2014;32(2):198-201. Epub 2014/01/22. doi: 10.1016/j.urolonc.2013.09.016. PubMed PMID: 24445287;PMC3970576.
82. Vassy JL, Lautenbach DM, McLaughlin HM, Kong SW, Christensen KD, Krier J, et al. The MedSeq Project: a randomized trial of integrating whole genome sequencing into clinical medicine. Trials. 2014;15:85. Epub 2014/03/22. doi: 10.1186/1745-6215-15-85. PubMed PMID: 24645908;PMC4113228.
83. Vassy JL, Hivert MF, Porneala B, Dauriz M, Florez JC, Dupuis J, et al. Polygenic type 2 diabetes prediction at the limit of common variant detection. Diabetes. 2014;63(6):2172-82. Epub 2014/02/13. doi: 10.2337/db13-1663. PubMed PMID: 24520119;PMC4030114.
84. Van Allen EM, Wagle N, Stojanov P, Perrin DL, Cibulskis K, Marlow S, et al. Whole-exome sequencing and clinical interpretation of formalin-fixed, paraffin-embedded tumor samples to guide precision cancer medicine. Nature medicine. 2014;20(6):682-8. Epub 2014/05/20. doi: 10.1038/nm.3559. PubMed PMID: 24836576;PMC4048335.
85. Van Allen EM, Mouw KW, Kim P, Iyer G, Wagle N, Al-Ahmadie H, et al. Somatic ERCC2 mutations correlate with cisplatin sensitivity in muscle-invasive urothelial carcinoma. Cancer discovery. 2014;4(10):1140-53. Epub 2014/08/07. doi: 10.1158/2159-8290.cd-14-0623. PubMed PMID: 25096233;PMC4238969.
86. Triedman JK, MacRae C. Searching for a Rosetta Stone: genetic data and clinical patient management. Heart rhythm : the official journal of the Heart Rhythm Society. 2014;11(10):1714-5. Epub 2014/07/22. doi: 10.1016/j.hrthm.2014.07.023. PubMed PMID: 25046859
87. Teutsch SM, Fielding JE, Khoury MJ, Evans JP. Utility before business. Genetics in medicine : official journal of the American College of Medical Genetics. 2014;16(12):869-70. Epub 2014/07/11. doi: 10.1038/gim.2014.71. PubMed PMID: 25010054
88. Sparks JA, Iversen MD, Miller Kroouze R, Mahmoud TG, Triedman NA, Kalia SS, et al. Personalized Risk Estimator for Rheumatoid Arthritis (PRE-RA) Family Study: rationale and design for a randomized controlled trial evaluating rheumatoid arthritis risk education to first-degree relatives. Contemporary clinical trials. 2014;39(1):145-57. Epub 2014/08/26. doi: 10.1016/j.cct.2014.08.007. PubMed PMID: 25151341;PMC4175164.
89. Scollon S, Bergstrom K, Kerstein RA, Wang T, Hilsenbeck SG, Ramamurthy U, et al. Obtaining informed consent for clinical tumor and germline exome sequencing of newly diagnosed childhood cancer patients. Genome medicine. 2014;6(9):69. Epub 2014/10/16. doi: 10.1186/s13073-014-0069-3. PubMed PMID: 25317207;PMC4195891.
90. Raghavan S, Vassy JL. Do physicians think genomic medicine will be useful for patient care? Personalized medicine. 2014;11(4):424-33. Epub 2014/01/01. doi: 10.2217/pme.14.25. PubMed PMID: 26161114;PMC4494677.
91. Savage SK, Ziniel SI, Stoler J, Margulies DM, Holm IA, Brownstein CA. An assessment of clinician and researcher needs for support in the era of genomic medicine. Personalized Medicine 2014 Aug;11(6):569–579.

92. Prince AE, Roche MI. Genetic information, non-discrimination, and privacy protections in genetic counseling practice. Journal of genetic counseling. 2014;23(6):891-902. Epub 2014/07/27. doi: 10.1007/s10897-014-9743-2. PubMed PMID: 25063358;PMC4233176.
93. Poggesi C, Ho CY. Muscle dysfunction in hypertrophic cardiomyopathy: what is needed to move to translation? Journal of muscle research and cell motility. 2014;35(1):37-45. Epub 2014/02/05. doi: 10.1007/s10974-014-9374-0. PubMed PMID: 24493262;PMC3982612.
94. Peterson D, Munger C, Crowley J, Corcoran C, Cruchaga C, Goate AM, et al. Variants in PPP3R1 and MAPT are associated with more rapid functional decline in Alzheimer's disease: the Cache County Dementia Progression Study. Alzheimer's & dementia : the journal of the Alzheimer's Association. 2014;10(3):366-71. Epub 2013/06/04. doi: 10.1016/j.jalz.2013.02.010. PubMed PMID: 23727081;PMC3809344.
95. Parsons DW, Roy A, Plon SE, Roychowdhury S, Chinnaiyan AM. Clinical tumor sequencing: an incidental casualty of the American College of Medical Genetics and Genomics recommendations for reporting of incidental findings. Journal of clinical oncology : official journal of the American Society of Clinical Oncology. 2014;32(21):2203-5. Epub 2014/06/25. doi: 10.1200/jco.2013.54.8917. PubMed PMID: 24958819;PMC4188159.
96. Overby CL, Rasmussen LV, Hartzler A, Connolly JJ, Peterson JF, Hedberg RE, et al. A Template for Authoring and Adapting Genomic Medicine Content in the eMERGE Infobutton Project. AMIA Annual Symposium proceedings / AMIA Symposium AMIA Symposium. 2014;2014:944-53. Epub 2014/01/01. PubMed PMID: 25954402;PMC4419923.
97. Nishimura AA, Tarczy-Hornoch P, Shirts BH. Pragmatic and Ethical Challenges of Incorporating the Genome into the Electronic Medical Record. Current genetic medicine reports. 2014;2(4):201-11. Epub 2015/07/07. doi: 10.1007/s40142-014-0051-9. PubMed PMID: 26146597;PMC4489153.
98. Murray MF. Educating physicians in the era of genomic medicine. Genome medicine. 2014;6(6):45. Epub 2014/07/18. doi: 10.1186/gm564. PubMed PMID: 25031626;PMC4075231.
99. McLaughlin HM, Ceyhan-Birsoy O, Christensen KD, Kohane IS, Krier J, Lane WJ, et al. A systematic approach to the reporting of medically relevant findings from whole genome sequencing. BMC medical genetics. 2014;15:134. Epub 2015/02/26. doi: 10.1186/s12881-014-0134-1. PubMed PMID: 25714468;PMC4342199.
100. McGuire AL, Knoppers BM, Zawati MH, Clayton EW. Can I be sued for that? Liability risk and the disclosure of clinically significant genetic research findings. Genome research. 2014;24(5):719-23. Epub 2014/03/29. doi: 10.1101/gr.170514.113. PubMed PMID: 24676095;PMC4009601.
101. Masino AJ, Dechene ET, Dulik MC, Wilkens A, Spinner NB, Krantz ID, et al. Clinical phenotype-based gene prioritization: an initial study using semantic similarity and the human phenotype ontology. BMC bioinformatics. 2014;15:248. Epub 2014/07/23. doi: 10.1186/1471-2105-15-248. PubMed PMID: 25047600;PMC4117966.
102. MacRae CA, Vasan RS. Clinically relevant functional annotation of genotype. Circulation Cardiovascular genetics. 2014;7(1):2-3. Epub 2014/02/20. doi: 10.1161/circgenetics.114.000506. PubMed PMID: 24550428;PMC4188549.
103. MacArthur DG, Manolio TA, Dimmock DP, Rehm HL, Shendure J, Abecasis GR, et al. Guidelines for investigating causality of sequence variants in human disease. Nature. 2014;508(7497):469-76. Epub 2014/04/25. doi: 10.1038/nature13127. PubMed PMID: 24759409;PMC4180223.
104. Lewis MH. Newborn screening controversy: past, present, and future. JAMA pediatrics. 2014;168(3):199-200. Epub 2014/01/08. doi: 10.1001/jamapediatrics.2013.4980. PubMed PMID: 24395113
105. Levenseller BL, Soucier DJ, Miller VA, Harris D, Conway L, Bernhardt BA. Stakeholders' opinions on the implementation of pediatric whole exome sequencing: implications for informed consent. Journal of genetic counseling. 2014;23(4):552-65. Epub 2013/07/13. doi: 10.1007/s10897-013-9626-y. PubMed PMID: 23846343;PMC3849137.
106. Lee IH, Lee K, Hsing M, Choe Y, Park JH, Kim SH, et al. Prioritizing disease-linked variants, genes, and pathways with an interactive whole-genome analysis pipeline. Human mutation. 2014;35(5):537-47. Epub 2014/01/31. doi: 10.1002/humu.22520. PubMed PMID: 24478219;PMC4130156.
107. Koenig BA. Have we asked too much of consent? The Hastings Center report. 2014;44(4):33-4. Epub 2014/07/22. doi: 10.1002/hast.329. PubMed PMID: 25043364;PMC4249719.
108. Koay PP, Sharp RR. Managing Expectational Language: Translational genetic professionals consider the clinical potential of next-generation sequencing technologies. New genetics and society. 2014;33(2):126-48. Epub 2014/06/03. doi: 10.1080/14636778.2014.910448. PubMed PMID: 24883042;PMC4038681.
109. Klitzman R, Buquez B, Appelbaum PS, Fyer A, Chung WK. Processes and factors involved in decisions regarding return of incidental genomic findings in research. Genetics in medicine : official journal of the American College of Medical Genetics. 2014;16(4):311-7. Epub 2013/09/28. doi: 10.1038/gim.2013.140. PubMed PMID: 24071801;PMC3966970.
110. Kircher M, Witten DM, Jain P, O'Roak BJ, Cooper GM, Shendure J. A general framework for estimating the relative pathogenicity of human genetic variants. Nature genetics. 2014;46(3):310-5. Epub 2014/02/04. doi: 10.1038/ng.2892. PubMed PMID: 24487276;PMC3992975.
111. Jarvik GP, Amendola LM, Berg JS, Brothers K, Clayton EW, Chung W, et al. Return of genomic results to research participants: the floor, the ceiling, and the choices in between. American journal of human genetics. 2014;94(6):818-26. Epub 2014/05/13. doi: 10.1016/j.ajhg.2014.04.009. PubMed PMID: 24814192;PMC4121476.
112. Hwang KB, Lee IH, Park JH, Hambuch T, Choe Y, Kim M, et al. Reducing false-positive incidental findings with ensemble genotyping and logistic regression based variant filtering methods. Human mutation. 2014;35(8):936-44. Epub 2014/05/16. doi: 10.1002/humu.22587. PubMed PMID: 24829188;PMC4112476.
113. Holm IA, Savage SK, Green RC, Juengst E, McGuire A, Kornetsky S, et al. Guidelines for return of research results from pediatric genomic studies: deliberations of the Boston Children's Hospital Gene Partnership Informed Cohort Oversight Board. Genetics in medicine : official journal of the American College of Medical Genetics. 2014;16(7):547-52. Epub 2014/01/11. doi: 10.1038/gim.2013.190. PubMed PMID: 24406460

114. Holm IA. Clinical Management of Pediatric Genomic Testing. Current genetic medicine reports. 2014;2(4):212-5. Epub 2014/11/15. doi: 10.1007/s40142-014-0052-8. PubMed PMID: 25396100;PMC4226515.
115. Hivert MF, Vassy JL, Meigs JB. Susceptibility to type 2 diabetes mellitus--from genes to prevention. Nature reviews Endocrinology. 2014;10(4):198-205. Epub 2014/02/19. doi: 10.1038/nrendo.2014.11. PubMed PMID: 24535206;PMC4058643.
116. Henderson GE, Wolf SM, Kuczynski KJ, Joffe S, Sharp RR, Parsons DW, et al. The challenge of informed consent and return of results in translational genomics: empirical analysis and recommendations. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics. 2014;42(3):344-55. Epub 2014/09/30. doi: 10.1111/jlme.12151. PubMed PMID: 25264092;PMC4262925.
117. Hardart GE, Chung WK. Genetic testing of children for diseases that have onset in adulthood: the limits of family interests. Pediatrics. 2014;134 Suppl 2:S104-10. Epub 2014/10/03. doi: 10.1542/peds.2014-1394F. PubMed PMID: 25274875;PMC4258839.
118. Green RC, Christensen KD, Cupples LA, Relkin NR, Whitehouse PJ, Royal CD, et al. A randomized noninferiority trial of condensed protocols for genetic risk disclosure of Alzheimer's disease. Alzheimer's & dementia : the journal of the Alzheimer's Association. 2014. Epub 2014/12/17. doi: 10.1016/j.jalz.2014.10.014. PubMed PMID: 25499536;PMC4461546.
119. Gray SW, Martins Y, Feuerman LZ, Bernhardt BA, Biesecker BB, Christensen KD, et al. Social and behavioral research in genomic sequencing: approaches from the Clinical Sequencing Exploratory Research Consortium Outcomes and Measures Working Group. Genetics in medicine : official journal of the American College of Medical Genetics. 2014;16(10):727-35. Epub 2014/03/15. doi: 10.1038/gim.2014.26. PubMed PMID: 24625446;PMC4163120.
120. Gray SW, Hicks-Courant K, Cronin A, Rollins BJ, Weeks JC. Physicians' attitudes about multiplex tumor genomic testing. Journal of clinical oncology : official journal of the American Society of Clinical Oncology. 2014;32(13):1317-23. Epub 2014/03/26. doi: 10.1200/jco.2013.52.4298. PubMed PMID: 24663044;PMC3992721.
121. Grady WM, Pritchard CC. Molecular alterations and biomarkers in colorectal cancer. Toxicologic pathology. 2014;42(1):124-39. Epub 2013/11/02. doi: 10.1177/0192623313505155. PubMed PMID: 24178577;PMC3947043.
122. Gallego CJ, Bennette CS, Heagerty P, Comstock B, Horike-Pyne M, Hisama F, et al. Comparative effectiveness of next generation genomic sequencing for disease diagnosis: design of a randomized controlled trial in patients with colorectal cancer/polyposis syndromes. Contemporary clinical trials. 2014;39(1):1-8. Epub 2014/07/06. doi: 10.1016/j.cct.2014.06.016. PubMed PMID: 24997220;PMC4175052.
123. Ferrer RA, Taber JM, Klein WM, Harris PR, Lewis KL, Biesecker LG. The role of current affect, anticipated affect and spontaneous self-affirmation in decisions to receive self-threatening genetic risk information. Cognition & emotion. 2014:1-10. Epub 2014/12/09. doi: 10.1080/02699931.2014.985188. PubMed PMID: 25482843;PMC4503523.
124. Fan Z, Greenwood R, Felix AC, Shiloh-Malawsky Y, Tennison M, Roche M, et al. GCH1 heterozygous mutation identified by whole-exome sequencing as a treatable condition in a patient presenting with progressive spastic paraplegia. Journal of neurology. 2014;261(3):622-4. Epub 2014/02/11. doi: 10.1007/s00415-014-7265-3. PubMed PMID: 24509643;PMC4000021.
125. Everett JN, Gustafson SL, Raymond VM. Traditional roles in a non-traditional setting: genetic counseling in precision oncology. Journal of genetic counseling. 2014;23(4):655-60. Epub 2014/03/01. doi: 10.1007/s10897-014-9698-3. PubMed PMID: 24578120;PMC4096055.
126. Evans BJ, Dorschner MO, Burke W, Jarvik GP. Regulatory changes raise troubling questions for genomic testing. Genetics in medicine : official journal of the American College of Medical Genetics. 2014;16(11):799-803. Epub 2014/09/26. doi: 10.1038/gim.2014.127. PubMed PMID: 25255365;PMC4308037.
127. Evans BJ. Mining the human genome after Association for Molecular Pathology v. Myriad Genetics. Genetics in medicine : official journal of the American College of Medical Genetics. 2014;16(7):504-9. Epub 2013/12/21. doi: 10.1038/gim.2013.186. PubMed PMID: 24357850;PMC4063888.
128. Evans BJ. Economic regulation of next-generation sequencing. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics. 2014;42 Suppl 1:51-66. Epub 2014/10/10. doi: 10.1111/jlme.12162. PubMed PMID: 25298291;PMC4460792.
129. Evans BJ. The First Amendment Right to Speak About the Human Genome. University of Pennsylvania journal of constitutional law. 2014;16(3):549-636. Epub 2014/12/05. PubMed PMID: 25473380;PMC4249692.
130. Eckstein L, Garrett JR, Berkman BE. A framework for analyzing the ethics of disclosing genetic research findings. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics. 2014;42(2):190-207. Epub 2014/07/22. doi: 10.1111/jlme.12135. PubMed PMID: 25040383;PMC4142504.
131. Dorschner MO, Amendola LM, Shirts BH, Kiedrowski L, Salama J, Gordon AS, et al. Refining the structure and content of clinical genomic reports. American journal of medical genetics Part C, Seminars in medical genetics. 2014;166c(1):85-92. Epub 2014/03/13. doi: 10.1002/ajmg.c.31395. PubMed PMID: 24616401;PMC4077592.
132. Clayton EW, McCullough LB, Biesecker LG, Joffe S, Ross LF, Wolf SM. Addressing the ethical challenges in genetic testing and sequencing of children. The American journal of bioethics : AJOB. 2014;14(3):3-9. Epub 2014/03/07. doi: 10.1080/15265161.2013.879945. PubMed PMID: 24592828;PMC3950962.
133. Caskey CT, Gonzalez-Garay ML, Pereira S, McGuire AL. Adult genetic risk screening. Annual review of medicine. 2014;65:1-17. Epub 2013/11/06. doi: 10.1146/annurev-med-111212-144716. PubMed PMID: 24188662
134. Burke W, Evans BJ, Jarvik GP. Return of results: ethical and legal distinctions between research and clinical care. American journal of medical genetics Part C, Seminars in medical genetics. 2014;166c(1):105-11. Epub 2014/03/13. doi: 10.1002/ajmg.c.31393. PubMed PMID: 24616381;PMC4078921.
135. Brownstein CA, Beggs AH, Homer N, Merriman B, Yu TW, Flannery KC, et al. An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome biology. 2014;15(3):R53. Epub 2014/03/29. doi: 10.1186/gb-2014-15-3-r53. PubMed PMID: 24667040;PMC4073084.
136. Brothers KB, Lynch JA, Aufox SA, Connolly JJ, Gelb BD, Holm IA, et al. Practical guidance on informed consent for pediatric participants in a biorepository. Mayo Clinic proceedings. 2014;89(11):1471-80. Epub 2014/09/30. doi: 10.1016/j.mayocp.2014.07.006. PubMed PMID: 25264176;PMC4254313.

137. Botkin JR, Lewis MH, Watson MS, Swoboda KJ, Anderson R, Berry SA, et al. Parental permission for pilot newborn screening research: guidelines from the NBSTRN. Pediatrics. 2014;133(2):e410-7. Epub 2014/01/08. doi: 10.1542/peds.2013-2271. PubMed PMID: 24394680;PMC3904278.
138. Blumenthal-Barby JS, McGuire AL, Ubel PA. Why information alone is not enough: behavioral economics and the future of genomic medicine. Annals of internal medicine. 2014;161(8):605-6. Epub 2014/10/21. doi: 10.7326/m14-2074. PubMed PMID: 25329206
139. Biesecker LG, Green RC. Diagnostic clinical genome and exome sequencing. The New England journal of medicine. 2014;371(12):1170. Epub 2014/09/18. doi: 10.1056/NEJMc1408914. PubMed PMID: 25229935
140. Bernhardt B. Genetic counselors and the future of clinical genomics. Genome medicine. 2014;6(7):49. Epub 2014/07/22. doi: 10.1186/gm565. PubMed PMID: 25045402;PMC4100319.
141. Arndt AK, MacRae CA. Genetic testing in cardiovascular diseases. Current opinion in cardiology. 2014;29(3):235-40. Epub 2014/04/11. doi: 10.1097/hco.0000000000000055. PubMed PMID: 24717670;PMC4129647.
142. Appelbaum PS, Waldman CR, Fyer A, Klitzman R, Parens E, Martinez J, et al. Informed consent for return of incidental findings in genomic research. Genetics in medicine : official journal of the American College of Medical Genetics. 2014;16(5):367-73. Epub 2013/10/26. doi: 10.1038/gim.2013.145. PubMed PMID: 24158054;PMC3999314.
143. Appelbaum PS, Parens E, Waldman CR, Klitzman R, Fyer A, Martinez J, et al. Models of consent to return of incidental findings in genomic research. The Hastings Center report. 2014;44(4):22-32. Epub 2014/06/13. doi: 10.1002/hast.328. PubMed PMID: 24919982;PMC4107028.
144. Allen NL, Karlson EW, Malspeis S, Lu B, Seidman CE, Lehmann LS. Biobank participants' preferences for disclosure of genetic research results: perspectives from the OurGenes, OurHealth, OurCommunity project. Mayo Clinic proceedings. 2014;89(6):738-46. Epub 2014/06/20. doi: 10.1016/j.mayocp.2014.03.015. PubMed PMID: 24943692;PMC4148696.
145. Alford RL, Arnos KS, Fox M, Lin JW, Palmer CG, Pandya A, et al. American College of Medical Genetics and Genomics guideline for the clinical evaluation and etiologic diagnosis of hearing loss. Genetics in medicine : official journal of the American College of Medical Genetics. 2014;16(4):347-55. Epub 2014/03/22. doi: 10.1038/gim.2014.2. PubMed PMID: 24651602
146. Zhang Z, Leipzig J, Sasson A, Yu AM, Perin JC, Xie HM, et al. Efficient digest of high-throughput sequencing data in a reproducible report. BMC bioinformatics. 2013;14 Suppl 11:S3. Epub 2014/02/26. doi: 10.1186/1471-2105-14-s11-s3. PubMed PMID: 24564231;PMC3846741.
147. Zaidi S, Choi M, Wakimoto H, Ma L, Jiang J, Overton JD, et al. De novo mutations in histone-modifying genes in congenital heart disease. Nature. 2013;498(7453):220-3. Epub 2013/05/15. doi: 10.1038/nature12141. PubMed PMID: 23665959;PMC3706629.
148. Yu JH, Jamal SM, Tabor HK, Bamshad MJ. Self-guided management of exome and whole-genome sequencing results: changing the results return model. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(9):684-90. Epub 2013/04/27. doi: 10.1038/gim.2013.35. PubMed PMID: 23619276;PMC4010112.
149. Yang Y, Muzny DM, Reid JG, Bainbridge MN, Willis A, Ward PA, et al. Clinical whole-exome sequencing for the diagnosis of mendelian disorders. The New England journal of medicine. 2013;369(16):1502-11. Epub 2013/10/04. doi: 10.1056/NEJMoa1306555. PubMed PMID: 24088041;PMC4211433.
150. Wolf SM, Annas GJ, Elias S. Point-counterpoint. Patient autonomy and incidental findings in clinical genomics. Science (New York, NY). 2013;340(6136):1049-50. Epub 2013/05/21. doi: 10.1126/science.1239119. PubMed PMID: 23686341;PMC3721305.
151. Wolf SM. Return of results in genomic biobank research: ethics matters. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(2):157-9. Epub 2013/02/07. doi: 10.1038/gim.2012.162. PubMed PMID: 23386184;PMC3703930.
152. Wolf SM. Return of individual research results and incidental findings: facing the challenges of translational science. Annual review of genomics and human genetics. 2013;14:557-77. Epub 2013/07/24. doi: 10.1146/annurev-genom-091212-153506. PubMed PMID: 23875796;PMC4452115.
153. Vassy JL, Green RC, Lehmann LS. Genomic medicine in primary care: barriers and assets. Postgraduate medical journal. 2013;89(1057):615-6. Epub 2013/10/17. doi: 10.1136/postgradmedj-2013-132093. PubMed PMID: 24129030;PMC3951755.
154. Tarczy-Hornoch P, Amendola L, Aronson SJ, Garraway L, Gray S, Grundmeier RW, et al. A survey of informatics approaches to whole-exome and whole-genome clinical reporting in the electronic health record. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(10):824-32. Epub 2013/09/28. doi: 10.1038/gim.2013.120. PubMed PMID: 24071794;PMC3951437.
155. Sunyaev SR, Roth FP. Systems biology and the analysis of genetic variation. Current opinion in genetics & development. 2013;23(6):599-601. Epub 2013/12/03. doi: 10.1016/j.gde.2013.11.010. PubMed PMID: 24291212;PMC4116483.
156. Shulman MB, Harkins K, Green RC, Karlawish J. Using AD biomarker research results for clinical care: a survey of ADNI investigators. Neurology. 2013;81(13):1114-21. Epub 2013/08/24. doi: 10.1212/WNL.0b013e3182a55f4a. PubMed PMID: 23966249;PMC3795601.
157. Ross LF, Rothstein MA, Clayton EW. Premature guidance about whole-genome sequencing. Personalized medicine. 2013;10(6). Epub 2013/12/19. doi: 10.2217/pme.13.51. PubMed PMID: 24348686;PMC3859526.
158. Ross LF. Predictive genetic testing of children and the role of the best interest standard. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics. 2013;41(4):899-906, Table of Contents. Epub 2014/01/23. doi: 10.1111/jlme.12099. PubMed PMID: 24446947
159. Robinson DR, Wu YM, Vats P, Su F, Lonigro RJ, Cao X, et al. Activating ESR1 mutations in hormone-resistant metastatic breast cancer. Nature genetics. 2013;45(12):1446-51. Epub 2013/11/05. doi: 10.1038/ng.2823. PubMed PMID: 24185510;PMC4009946.
160. Rehm HL, Bale SJ, Bayrak-Toydemir P, Berg JS, Brown KK, Deignan JL, et al. ACMG clinical laboratory standards for next-generation sequencing. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(9):733-47. Epub 2013/07/28. doi: 10.1038/gim.2013.92. PubMed PMID: 23887774;PMC4098820.
161. Rehm HL. Disease-targeted sequencing: a cornerstone in the clinic. Nature reviews Genetics. 2013;14(4):295-300. Epub 2013/03/13. doi: 10.1038/nrg3463. PubMed PMID: 23478348;PMC3786217.
162. Parens E, Appelbaum P, Chung W. Incidental findings in the era of whole genome sequencing? The Hastings Center report. 2013;43(4):16-9. Epub 2013/07/12. doi: 10.1002/hast.189. PubMed PMID: 23842918;PMC3767184.

163. Overby CL, Kohane I, Kannry JL, Williams MS, Starren J, Bottinger E, et al. Opportunities for genomic clinical decision support interventions. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(10):817-23. Epub 2013/09/21. doi: 10.1038/gim.2013.128. PubMed PMID: 24051479;PMC3858176.
164. McGuire AL, Joffe S, Koenig BA, Biesecker BB, McCullough LB, Blumenthal-Barby JS, et al. Point-counterpoint. Ethics and genomic incidental findings. Science (New York, NY). 2013;340(6136):1047-8. Epub 2013/05/21. doi: 10.1126/science.1240156. PubMed PMID: 23686340;PMC3772710.
165. McGowan ML, Glinka A, Highland J, Asaad G, Sharp RR. Genetics patients' perspectives on clinical genomic testing. Personalized medicine. 2013;10(4):339-47. Epub 2014/06/24. doi: 10.2217/pme.13.32. PubMed PMID: 24955098;PMC4061708.
166. McGowan ML, Cho D, Sharp RR. The changing landscape of carrier screening: expanding technology and options? Health matrix (Cleveland, Ohio : 1991). 2013;23(1):15-33. Epub 2013/07/03. PubMed PMID: 23808097;PMC4120890.
167. MacRae CA. Mendelian forms of structural cardiovascular disease. Current cardiology reports. 2013;15(10):399. Epub 2013/09/21. doi: 10.1007/s11886-013-0399-6. PubMed PMID: 24046092;PMC3884814.
168. Lewis MH. Laboratory specimens and genetic privacy: evolution of legal theory. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics. 2013;41 Suppl 1:65-8. Epub 2013/04/23. doi: 10.1111/jlme.12042. PubMed PMID: 23590744
169. Lautenbach DM, Christensen KD, Sparks JA, Green RC. Communicating genetic risk information for common disorders in the era of genomic medicine. Annual review of genomics and human genetics. 2013;14:491-513. Epub 2013/09/06. doi: 10.1146/annurev-genom-092010-110722. PubMed PMID: 24003856;PMC3862080.
170. Klitzman R, Appelbaum PS, Fyer A, Martinez J, Buquez B, Wynn J, et al. Researchers' views on return of incidental genomic research results: qualitative and quantitative findings. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(11):888-95. Epub 2013/06/29. doi: 10.1038/gim.2013.87. PubMed PMID: 23807616;PMC3823712.
171. Klitzman R, Appelbaum PS, Chung W. Return of secondary genomic findings vs patient autonomy: implications for medical care. Jama. 2013;310(4):369-70. Epub 2013/08/07. doi: 10.1001/jama.2013.41709. PubMed PMID: 23917282;PMC3800693.
172. Hazin R, Brothers KB, Malin BA, Koenig BA, Sanderson SC, Rothstein MA, et al. Ethical, legal, and social implications of incorporating genomic information into electronic health records. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(10):810-6. Epub 2013/09/14. doi: 10.1038/gim.2013.117. PubMed PMID: 24030434;PMC3926430.
173. Green RC, Berg JS, Grody WW, Kalia SS, Korf BR, Martin CL, et al. ACMG recommendations for reporting of incidental findings in clinical exome and genome sequencing. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(7):565-74. Epub 2013/06/22. doi: 10.1038/gim.2013.73. PubMed PMID: 23788249;PMC3727274.
174. Gonzalez-Garay ML, McGuire AL, Pereira S, Caskey CT. Personalized genomic disease risk of volunteers. Proceedings of the National Academy of Sciences of the United States of America. 2013;110(42):16957-62. Epub 2013/10/02. doi: 10.1073/pnas.1315934110. PubMed PMID: 24082139;PMC3801013.
175. Gliwa C, Berkman BE. Do researchers have an obligation to actively look for genetic incidental findings? The American journal of bioethics : AJOB. 2013;13(2):32-42. Epub 2013/02/09. doi: 10.1080/15265161.2012.754062. PubMed PMID: 23391059;PMC4138545.
176. Garrett JR. Reframing the ethical debate regarding incidental findings in genetic research. The American journal of bioethics : AJOB. 2013;13(2):44-6. Epub 2013/02/09. doi: 10.1080/15265161.2013.757972. PubMed PMID: 23391061
177. Evans BJ. Minimizing liability risks under the ACMG recommendations for reporting incidental findings in clinical exome and genome sequencing. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(12):915-20. Epub 2013/09/14. doi: 10.1038/gim.2013.135. PubMed PMID: 24030435;PMC3892767.
178. Duzkale H, Shen J, McLaughlin H, Alfares A, Kelly MA, Pugh TJ, et al. A systematic approach to assessing the clinical significance of genetic variants. Clinical genetics. 2013;84(5):453-63. Epub 2013/09/17. doi: 10.1111/cge.12257. PubMed PMID: 24033266;PMC3995020.
179. Dorschner MO, Amendola LM, Turner EH, Robertson PD, Shirts BH, Gallego CJ, et al. Actionable, pathogenic incidental findings in 1,000 participants' exomes. American journal of human genetics. 2013;93(4):631-40. Epub 2013/09/24. doi: 10.1016/j.ajhg.2013.08.006. PubMed PMID: 24055113;PMC3791261.
180. Clayton EW, Kelly SE. Let us ask better questions. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(11):871-2. Epub 2013/05/25. doi: 10.1038/gim.2013.68. PubMed PMID: 23703684
181. Clayton EW, Haga S, Kuszler P, Bane E, Shutske K, Burke W. Managing incidental genomic findings: legal obligations of clinicians. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(8):624-9. Epub 2013/03/02. doi: 10.1038/gim.2013.7. PubMed PMID: 23448723;PMC3805501.
182. Christensen KD, Green RC. How could disclosing incidental information from whole-genome sequencing affect patient behavior? Personalized medicine. 2013;10(4). Epub 2013/12/10. doi: 10.2217/pme.13.24. PubMed PMID: 24319470;PMC3852635.
183. Cho D, McGowan ML, Metcalfe J, Sharp RR. Expanded carrier screening in reproductive healthcare: perspectives from genetics professionals. Human reproduction (Oxford, England). 2013;28(6):1725-30. Epub 2013/04/17. doi: 10.1093/humrep/det091. PubMed PMID: 23589535;PMC3657126.
184. Caulfield T, Evans J, McGuire A, McCabe C, Bubela T, Cook-Deegan R, et al. Reflections on the cost of "low-cost" whole genome sequencing: framing the health policy debate. PLoS biology. 2013;11(11):e1001699. Epub 2013/11/14. doi: 10.1371/journal.pbio.1001699. PubMed PMID: 24223516;PMC3818164.
185. Burke W, Trinidad SB, Clayton EW. Seeking Genomic Knowledge: The Case for Clinical Restraint. The Hastings law journal. 2013;64(6):1650-64. Epub 2014/04/02. PubMed PMID: 24688162;PMC3969739.
186. Burke W, Antommaria AH, Bennett R, Botkin J, Clayton EW, Henderson GE, et al. Recommendations for returning genomic incidental findings? We need to talk! Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(11):854-9. Epub 2013/08/03. doi: 10.1038/gim.2013.113. PubMed PMID: 23907645;PMC3832423.
187. Botkin JR, Goldenberg AJ, Rothwell E, Anderson RA, Lewis MH. Retention and research use of residual newborn screening bloodspots. Pediatrics. 2013;131(1):120-7. Epub 2012/12/05. doi: 10.1542/peds.2012-0852. PubMed PMID: 23209103;PMC3529945.

188. Berg JS, Amendola LM, Eng C, Van Allen E, Gray SW, Wagle N, et al. Processes and preliminary outputs for identification of actionable genes as incidental findings in genomic sequence data in the Clinical Sequencing Exploratory Research Consortium. Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(11):860-7. Epub 2013/11/08. doi: 10.1038/gim.2013.133. PubMed PMID: 24195999;PMC3935342.
189. Bennette CS, Trinidad SB, Fullerton SM, Patrick D, Amendola L, Burke W, et al. Return of incidental findings in genomic medicine: measuring what patients value--development of an instrument to measure preferences for information from next-generation testing (IMPRINT). Genetics in medicine : official journal of the American College of Medical Genetics. 2013;15(11):873-81. Epub 2013/06/01. doi: 10.1038/gim.2013.63. PubMed PMID: 23722871;PMC3823641.
190. Wolf SM. The Role of Law in the Debate over Return of Research Results and Incidental Findings: The Challenge of Developing Law for Translational Science. Minnesota journal of law, science & technology. 2012;13(2). Epub 2012/01/01. doi: 10.2139/ssrn.2117289. PubMed PMID: 24379751;PMC3874275.
191. Uhlmann WR, Sharp RR. Genetic testing integration panels (GTIPs): a novel approach for considering integration of direct-to-consumer and other new genetic tests into patient care. Journal of genetic counseling. 2012;21(3):374-81. Epub 2012/01/17. doi: 10.1007/s10897-011-9468-4. PubMed PMID: 22246561;PMC4125014.
192. McGuire AL, Wang MJ, Probst FJ. Currents in contemporary bioethics. Identifying consanguinity through routine genomic analysis: reporting requirements. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics. 2012;40(4):1040-6. Epub 2013/01/08. doi: 10.1111/j.1748-720X.2012.00731.x. PubMed PMID: 23289705;PMC4030722.
193. MacRae CA. Pattern recognition: combining informatics and genetics to re-evaluate conduction disease. Heart (British Cardiac Society). 2012;98(17):1263-4. Epub 2012/08/10. doi: 10.1136/heartjnl-2012-302408. PubMed PMID: 22875820
194. Lewis MH, Scheurer ME, Green RC, McGuire AL. Research results: preserving newborn blood samples. Science translational medicine. 2012;4(159):159cm12. Epub 2012/11/09. doi: 10.1126/scitranslmed.3004474. PubMed PMID: 23136040;PMC3763707.
195. Holm IA, Taylor PL. The Informed Cohort Oversight Board: From Values to Architecture. Minnesota journal of law, science & technology. 2012;13(2):669-90. Epub 2012/04/01. PubMed PMID: 24371432;PMC3872117.
196. Harris ED, Ziniel SI, Amatruda JG, Clinton CM, Savage SK, Taylor PL, et al. The beliefs, motivations, and expectations of parents who have enrolled their children in a genetic biorepository. Genetics in medicine : official journal of the American College of Medical Genetics. 2012;14(3):330-7. Epub 2012/01/14. doi: 10.1038/gim.2011.25. PubMed PMID: 22241099;PMC3763713.
197. Goldenberg AJ, Sharp RR. The ethical hazards and programmatic challenges of genomic newborn screening. Jama. 2012;307(5):461-2. Epub 2012/02/03. doi: 10.1001/jama.2012.68. PubMed PMID: 22298675;PMC3868436.
198. Sharp RR. Downsizing genomic medicine: approaching the ethical complexity of whole-genome sequencing by starting small. Genetics in medicine : official journal of the American College of Medical Genetics. 2011;13(3):191-4. Epub 2011/02/12. doi: 10.1097/GIM.0b013e31820f603f. PubMed PMID: 21311340
199. Fabsitz RR, McGuire A, Sharp RR, Puggal M, Beskow LM, Biesecker LG, et al. Ethical and practical guidelines for reporting genetic research results to study participants: updated guidelines from a National Heart, Lung, and Blood Institute working group. Circulation Cardiovascular genetics. 2010;3(6):574-80. Epub 2010/12/16. doi: 10.1161/circgenetics.110.958827. PubMed PMID: 21156933;PMC3090664.
200. Foster MW, Mulvihill JJ, Sharp RR. Evaluating the utility of personal genomic information. Genetics in medicine : official journal of the American College of Medical Genetics. 2009;11(8):570-4. Epub 2009/05/30. doi: 10.1097/GIM.0b013e3181a2743e. PubMed PMID: 19478683
201. Scollon S, Bergstrom K, McCullough LB, McGuire AL, Gutierrez S, Kerstein R, Parsons DW, Plon SE (2015). Pediatric cancer genetics research and an evolving preventive ethics approach for return of results after death of the subject. J Law Medicine and Ethics
202. Wolf S, Burke W, Koenig B. Mapping the full ethics of translational genomics: situating return of results and navigating the research-clinical divide. J Law Med Ethics
203. Posey JE, Rosenfeld JA, James RA, Bainbridge M, Niu Z, Wang X, Dhar S, Wiszniewski W, Akdemir ZHC, Gambin T, Xia F, Person RE, Walkiewicz M, Shaw CA, Sutton VR, Beaudet AL, Muzny D, Eng CM, Yang Y, Gibbs RA, Lupski JR, Boerwinkle E, Plon SE (2015). Molecular diagnostic experience of whole exome sequencing in adult patients. Genet Med
204. Wolf SM, et al. Returning a Research Participant's Genomic Results to Relatives: Analysis and Recommendations. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696543.
205. Daly M. A Family-Centered Model for Sharing Genetic Risk. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in--press). NIHMSID: 696551.
206. Fernandez C, O'Rourke P, & Beskow L. Canadian Research Ethics Board Leadership Attitudes to the Return of Genetic Research Results to Individuals and their Families. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696560.
207. Petersen BM & Van Ness B. Returning a Research Participant's Genomic Results to Relatives: Perspectives from Managers of Two Distinct Research Biobanks. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696564
208. Wilfond B, Fernandez C, & Green RC. Disclosing Secondary Findings from Pediatric Sequencing to Families: Considering the "Benefit to Families". The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696579
209. Wright Clayton E. How much control do children and adolescents have over genomic testing, parental access to their results, and parental communication of those results to others?. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696583
210. Huckaby Lewis M. Return of Results from Research Using Newborn Screening Dried Blood Samples. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in press). NIHMSID: 696584

211. Garrison NA. Considerations for Returning Research Results to Culturally Diverse Participants and Families of Decedents. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696586
212. Branum R & Wolf SM. International Policies on Sharing Genomic Research Results with Relatives: Approaches to Balancing Privacy with Access. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696592.
213. Radeck Breitkopf C, Petersen G, Wolf SM, et al. Preferences Regarding Return of Genomic Results to Relatives of Research Participants, Including after Participant Death: Empirical Results from a Cancer Biobank. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696597
214. Wolf SM. Introduction: Return of Research Results: What About the Family?. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696600.
215. Beskow L & O'Rourke P. Return of Genetic Research Results to Participants and Families: IRB Perspectives and Roles. The Journal of law, medicine & ethics : a journal of the American Society of Law, Medicine & Ethics (in-press). NIHMSID: 696555.
216. Brothers KB, Holm IA, Childerhose JE, Antommaria AHM, Bernhardt BA, Clayton EW, Gelb BD, Joffe S, Lynch JA, McCormick JB, McCullough LB, Parsons DW, Sundaresan AS, Wolf WA, Yu JH, Wilfond BS. When Genomic Research Participants Grow Up: Contact and Consent at the Age of Majority. The Journal of Pediatrics. 2014. In Press.
217. Linderman MD, Green RC. Participatory personal genome sequencing in ostensibly healthy individuals. J Pers Med. In press; PMCID: PMC Journal
218. Christensen KD, Siebert U, Green RC. Integrating genomic sequencing into clinical care: is it worth the cost? J Pers Med. In press; PMCID: PMC Journal
219. Amendola LM, Horike-Pyne M, Trinidad SB, Fullerton SM, Evans BJ, Burke W and Jarvik GP. Patients' Choices for Return of Exome Sequencing Results to Relatives in the Event of their Death. Law Med Ethics In press.
220. Janssens ACJW, Evans JP. Returning pharmacogenetic secondary findings from genome sequencing: let's not put the cart before the horse [Internet]. Genet Med 2015 May;
221. Khoury MJ, Evans JP. A public health perspective on a national precision medicine cohort: Balancing long-term knowledge generation with early health benefit. JAMA 2015 Jun;313(21):2117–2118.
222. Wynn J. Genomic Testing: a Genetic Counselor's Personal Reflection on Three Years of Consenting and Testing. J Genet Couns 2015 Aug; PMID: 26242468
223. Christensen KD, Kalia SS, Green RC. Incidental findings from genetic testing: UpToDate; 2014 [cited 2015 Sept 13]. Available from: http://www.uptodate.com/contents/incidental-findings-from-genetic-testing.
224. Couzin-Frankel J. Divulging DNA Secrets of Dead Stirs Debate. Science 2014 Jan;343(6169):356–357.
225. Koenig BA. Have We Asked Too Much of Consent? Hastings Center Report 2014 Jul;44(4):33–34.
226. Roche MI, Palmer CGS. Next Generation Genetic Counseling: Introduction to the Special Issue. Journal of Genetic Counseling 2014 Aug;23(4):439–444.
227. Giovanni, Monica A, and Murray, Michael F(Nov 2014) Use of Personalized Genomic Information. In: eLS. John Wiley & Sons Ltd, Chichester. http://www.els.net [doi: 10.1002/9780470015902.a0024126]
228. Wolf SM. What to do about too much medical information? Minneapolis StarTribuen op-ed, 12/12/2013. Available from: http://www.startribune.com/what-to-do-about-too-much-medical-information/235664471/.
229. Evans JP. When is a medical finding "incidental"? Genet Med 2013 Jul;15(7):515–516.
230. Evans JP. Return of results to the families of children in genomic sequencing: tallying risks and benefits. Genet Med 2013 Jun;15(6):435–436.
231. Evans JP. Finding common ground. Genet Med 2013 Nov;15(11):852–853.
232. Green RC, Rehm HL, Kohane IS. Clinical genome sequencing. In: Ginsburg GS, Willard HF, editors. Genomic and Personalized Medicine. 2nd ed. San Diego: Academic Press; 2013. p. 102-22.
233. Michelle Huckaby Lewis, Ellen Wright Clayton, Genetics and Ethics in Pediatrics, In: Robert Saul, ed., Medical Genetics in Pediatric Practice, AAP: Elk Grove Village, IL 299-308 (2013)
234. Klitzman R, Appelbaum PS, Chung W. Return of secondary genomic findings vs patient autonomy: Implications for medical care. JAMA 2013 Jul;310(4):369–370.
235. Wolf SM. The Role of Law in the Debate over Return of Research Results and Incidental Findings: The Challenge of Developing Law for Translational Science. Minn J Law Sci Technol 2012;13(2) PMID: 24379751 PMCID: PMC3874275