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governance, ethics, evidence, policy, practice

Center for Genomic Medicine Ethics & Policy

Genomic Medicine Governance, Ethics, Policy, Practice – A Monthly Digest

April 2023 Number 04

Genomic medicine – spanning pre-clinical basic science through clinical development and translation into daily patient interventions – continues to evolve at an extraordinary pace. Advances in the scientific and technical dimensions of genomic medicine are extensively communicated through the peer-reviewed journal ecology and supporting grey literature.

Complementing this technical literature is a growing body of commentary, analysis and research around the governance, ethics, regulation, and policy dimensions of genomic medicine. Much of this content is communicated through academic journals and grey literature, but is also appearing in the general media. This digest intends to capture and curate the most substantive examples of this non-technical content.

In aggregating and editing this digest, we directly review a broad spectrum of peer-reviewed journals and grey literature, as well as announcements and strategic actions from various practice domains and organization types including international agencies, INGOs, governments/regulatory bodies, academic and research institutions, consortiums and collaborations, foundations, and commercial organizations. More broadly, we utilize *Google Scholar's* alert capability to scan current literature. We acknowledge that this approach and scope yields an indicative and not an exhaustive digest product.

This digest is a service of the GE2P2 Global Foundation and its newly formed Center for Genomic Medicine Governance, Ethics & Policy. The Foundation is solely responsible for its content. Comments and suggestions should be directed to the Editor or Associate Editor as below:

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We organize digest content in each edition using subject categories to help readers navigate to areas of interest. We expect that these categories will evolve over time. Active categories in this edition include:

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EDITORS SHORTLIST

We lead each edition of this digest with a "short list" of entries that we assess to be strategically important and well aligned to our thematic focus areas of governance, ethics, policy and [clinical] practice. The full citation/abstract for each item appears just below this summary.

:: The statement issued by organizers of the Third International Summit on Human Genome Editing deserves our primary attention. The Summit [6-8 March 2023 at the Francis Crick Institute, London UK] was extraordinary and we urge readers to view the recorded sessions at the links provided. **The full statement below addresses somatic human genome editing, equitable access for somatic human genome editing, human germline genome editing for research (not for reproduction), heritable human genome editing, and ongoing international collaboration and discussions.**

Other shortlist entries related to the Summit and human genome editing include:
:: An editorial by *The Lancet* – Human genome editing: ensuring responsible research – argues "… a better international consensus is essential on how to advance gene editing while safeguarding humanity's collective gene pool. There is broad agreement that altering embryo DNA for reproductive purposes should remain forbidden; a 2020 study showed that 75 of 96 surveyed countries have banned it. However, many do not have effective oversight and governance mechanisms to enforce existing regulations. In some countries, although altering embryo DNA is generally forbidden, exceptions are allowed, making more likely the possibility that the technology is used for a banned purpose. The lack of policy alignment between countries raises the possibility of scientists exporting their research to evade constraints established in their home jurisdictions…"

:: A literature review – **Advances in Genetic Editing of the Human Embryo –** by Astarăstoae, Vasile et al. in the *American Journal of Therapeutics* urges global dialogue to achieve a broad social consensus around the ethics involved here..

:: **Germline Gene Editing: The Gender Issues** by Iñigo de Miguel Beriain et al in the *Cambridge Quarterly of Healthcare Ethics* argues that the introduction of gene editing in human reproduction will have an undetermined and perhaps double-edged effect on the position of women seeking to conceive.

:: Vanessa Godwin's thesis – **Law, bioethics, and society: Jewish and Islamic approaches to fertility treatments and human germline genome editing** – takes note of "a religious narrative which endorses the deselection of pre-embryos as an ethical alternative to abortion and explores how this may impact future debates about the permissibility of HGGE. Given this context it becomes apparent how Jewish and Muslim jurists debate the fundamental questions about the creation of human life and why their divergent legal judgements, which are generated for the moral good of their respective societies, matter in the global debate."

:: Finally, we are now monitoring for important meetings/symposia/conferences and will add them to each edition. On the immediate horizon are two meetings organized by NASEM [National Academies of Science, Engineering, Medicine, USA]:

> **NASEM - The Potential Contribution of Cancer Genomics Information to Community Investigations of Unusual Patterns of Cancer: A Workshop**
> Apr 13, 2023 8:30AM - 3:00PM ET

The National Academies of Sciences, Engineering, and Medicine will hold a virtual, 1-day public workshop on the Potential Contribution of Cancer Genomics Information to Community Investigations of Unusual Patterns of Cancer on April 13, 2023 to explore the state of the science with regard to the identification of genomic and epigenomic biomarkers of environmental exposures associated with cancers, with emphasis on pediatric cancers.

NASEM - In Vitro Derived Human Gametes as a Reproductive Technology: Scientific, Ethical, and Regulatory Implications: A Workshop

April 19, 2023 | 9:15 a.m.-5 p.m. ET
April 20, 2023 | 9:30 a.m.-5 p.m. ET
April 21, 2023 | 9:30 a.m.-2:30 p.m. ET

The National Academies will convene a workshop to explore the in vitro derivation of human gametes (eggs and sperm) from embryonic or induced pluripotent stem cells, and its potential impact on research and reproductive medicine.

Invited speakers will address the scientific milestones observed in other mammalian model species, the scientific and technical roadblocks that challenge this method of producing human gametes from stem cells, the ethical and sociocultural issues raised by potential uses of this technology, and related legal and regulatory considerations.

SHORTLIST ABSTRACTS

Third International Summit on Human Genome Editing

The Third International Summit on Human Genome Editing took place on 6-8 March 2023 at the Francis Crick Institute, London UK.

Building on previous events held in Washington, DC (2015) and Hong Kong (2018), the London meeting continued the global dialogue on somatic and germline human genome editing. Major themes for discussion included developments in clinical trials and genome editing tools such as CRISPR/Cas9, as well as social, ethical and accessibility considerations these scientific developments entail.

The three-day Summit was organised by the Royal Society, the UK Academy of Medical Sciences, the US National Academies of Sciences and Medicine and The World Academy of Sciences. The Summit's Organising Committee, chaired by Professor Robin Lovell-Badge FMedSci FRS, released a statement [full text below] based on the Summit discussions.

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Statement from the Organising Committee of the Third International Summit on Human Genome Editing

Issued 08 March 2023

The Third International Summit on Human Genome Editing, convened by the UK Royal Society, UK Academy of Medical Sciences, US National Academies of Sciences and Medicine, and The World Academy of Sciences, was held to discuss progress, promise, and challenges in research, regulation, and equitable development of human genome editing technologies and therapies.

After listening to three days of thoughtful and inclusive discussion, the members of the Organising Committee offer the following conclusions:

:: Remarkable progress has been made in somatic human genome editing, demonstrating it can cure once incurable diseases. To realise its full therapeutic potential, research is needed to expand the range of diseases it can treat, and to better understand risks and unintended effects. The extremely high costs of current somatic gene therapies are unsustainable. A global commitment to affordable, equitable access to these treatments is urgently needed.

:: Heritable human genome editing remains unacceptable at this time. Public discussions and policy debates continue and are important for resolving whether this technology should be used. Governance frameworks and ethical principles for the responsible use of heritable human genome editing are not in place. Necessary safety and efficacy standards have not been met.

:: Governance mechanisms for human genome editing need to protect ongoing, legitimate research, while preventing clinics or individuals from offering unproven interventions in the guise of therapies or ways to avoid disease.

Somatic human genome editing[1]

Numerous clinical trials using somatic human genome editing are in progress or soon to be initiated, with preliminary but encouraging results that point to future therapies. The dramatic improvement following CRISPR-based research interventions for sickle cell disease offers hope for patients. Many techniques, including base, prime, and epigenetic editing, may also prove to be useful interventions for a broad range of both genetic and acquired diseases and disorders. However, as with other gene therapies, extended long-term follow-up is essential to fully understand the consequences of an edit and to identify any unanticipated effects, should they occur.

Improved techniques have enhanced the efficiency, precision, and accuracy of the editing process, yet effective delivery and editing remains difficult for many tissues of the body. Further research to diversify and increase the efficiency, specificity, and safety of editing-delivery systems is essential for improving potential treatment options and promoting equitable access.

Equitable access for somatic human genome editing

As interventions based on somatic genome editing become more widespread, a commitment to equitable, financially sustainable, and accessible treatments becomes more urgent. In many cases, costs and infrastructure needs of current gene therapy treatments are not manageable for either patients or healthcare systems. Correcting this will require appropriate planning from the earliest stages of the research and development for each potential application. Ensuring research includes more genetically diverse populations and expanding the range of those who conceive and conduct the research, play a vital role in achieving equitable outcomes.

With sickle cell disease (as well as other genetic diseases), a large percentage of patients live in underserved countries and communities or in settings without adequate infrastructure. Moving from ex-vivo to “one and done” in-vivo somatic human genome editing can partially address this problem. But knowledge transfer between nations, improved clinics and research facilities, and strong oversight are also needed to establish sustainable access to safe interventions for research participants and patients.

Health care systems and the global health community should prepare to provide patients with cost-effective, affordable, proven therapies. Therapies based on somatic genome editing that could help meet these needs should be a priority for research investment.

Human germline genome editing for research (not for reproduction)[2]

Basic research using genome editing in human embryos has continued, with the aim of either understanding aspects of early human development or exploring how the methods might be used to correct gene variants leading to genetic disorders. There has also been significant progress in basic research on deriving functional gametes from stem cells. Basic research in this field should continue.

Heritable human genome editing

Preclinical evidence for the safety and efficacy of heritable human genome editing has not been established, nor has societal discussion and policy debate been concluded. (In some cases, preimplantation genetic testing is among the alternatives.) Heritable human genome editing should not be used unless, at a minimum, it meets reasonable standards for safety and efficacy, is legally sanctioned, and has been developed and tested under a system of rigorous oversight that is subject to responsible governance. At this time, these conditions have not been met.

Ongoing international collaboration and discussions

The Organising Committee calls for on-going dialogue and continued international collaboration on innovative approaches to governance and regulation of human genome editing technologies, the state of the science, and innovation in the treatment of genetic diseases.

> *1. Somatic human genome editing refers to the editing of somatic cells, which are non-reproductive cells, and changes made in these cells affect only the person who receives the genome editing.*
> *2. In this statement, germline human genome editing refers to the editing of human embryos or gametes in a research setting, with no plans for those embryos or gametes to be used for human reproduction. Heritable human genome editing refers to the editing of human embryos or gametes to be used for human reproduction.*

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Summit resources

- The Summit booklet, which includes the agenda, is available to download.
- The report of a research project aiming to survey, document, catalogue and analyse empirical information regarding regulatory capacity and governance approaches for somatic genome editing research interventions in different countries, which was commissioned ahead of the Summit, is available to download. Findings from the report were presented by Piers Millett on Wednesday 8 March at the Summit.
- A recorded presentation on 'CRISPR and Human Genome Editing: Progress & Opportunities' by Jennifer Doudna, Li Ka Shing Chancellor's Chair Professor in the Department of Chemistry and the Department of Molecular and Cell Biology, University of California, Berkeley, USA, is available to watch.
- A handout for the agenda session on the morning of Wednesday 8 March 'Civil Society and Human Genome Editing: roles and challenges in public engagement' is available to download.
- In preparation for the Summit, a three-part series of online events was held in 2022 'Looking Ahead to the Third Human Genome Editing Summit', which focused on scientific developments, equity and access, and governance of human genome editing. Recordings of all the presentations are available to watch.

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Human genome editing: ensuring responsible research

Editorial

The Lancet, Volume 401, Issue 10380, P877, 18 March 2023

Article Info

In 2018, during the Second International Summit on Human Genome Editing in Hong Kong, Jiankui He shocked the world by announcing the birth of two children whose genomes he had edited using CRISPR technology. Following widespread condemnation and a criminal investigation, he was sentenced to 3 years in prison. The case caused international outcry and brought to the fore the need to reconsider the serious ethical, scientific, and social issues of heritable human genome editing. As science advances, especially in non-heritable, somatic gene editing for treatment of previously incurable diseases, regulatory gaps are

becoming exposed. Governance of gene editing research was a major discussion point at the Third International Human Genome Editing Summit in London, on March 6–8, with widespread recognition for the need to build on existing guidelines to develop global standards for governance and oversight of human genome editing. As He's unconscionable actions showed, the ethical and scientific risks are substantial.

Gene editing regulations must consider the aims and consequences of the different practices involved. Somatic genome editing interventions (eg, targeted therapies such as chimeric antigen receptor T cells or small interfering RNA gene therapies) are not transmitted to offspring and are widely used. Heritable genome editing—also called germline editing—is aimed at research on human fertilisation and embryology or for reproductive purposes. From a genetic point of view, germline editing is of most concern because alterations are passed to offspring, with the risk of perpetuating unexpected and undesired changes through generations. It is impossible for our unborn descendants to give consent.

Loopholes and ambiguities in regulation need to be closed urgently to enable scientists to be held to account. In China, He's prosecution was based on practising medicine without a licence, rather than specifically based on a provision governing assisted reproduction or genome editing. China has since instituted new regulations, widely seen as a response to the He case, but they have been criticised in press reports for not doing enough to cover private companies. Wording of legislation needs to be explicit and clear. In the USA, use of funds by the FDA for the purpose of accepting and reviewing any application to begin a clinical trial for heritable germline editing is prohibited. While this, in effect, makes some reproductive editing illegal, it falls short of a ban on the practice itself. Similar ambiguities exist in many countries, and as the technologies involved become cheaper and more widely available, the risk increases.

A better international consensus is essential on how to advance gene editing while safeguarding humanity's collective genepool. There is broad agreement that altering embryo DNA for reproductive purposes should remain forbidden; a 2020 study showed that 75 of 96 surveyed countries have banned it. However, many do not have effective oversight and governance mechanisms to enforce existing regulations. In some countries, although altering embryo DNA is generally forbidden, exceptions are allowed, making more likely the possibility that the technology is used for a banned purpose. The lack of policy alignment between countries raises the possibility of scientists exporting their research to evade constraints established in their home jurisdictions.

How will a global consensus be enforced? The UN is the only body in a position to do so, and the prospect of an international legally binding treaty to govern genome editing was raised at the Second International Summit in 2018, but has seemingly proceeded no further. The Oviedo Convention, a legally binding instrument established by the European Council, permits somatic genome modifications for preventive, diagnostic, or therapeutic purposes, and prohibits germline editing, but only 29 countries have enacted it into law.

It is almost 20 years since scientists announced the mapping of the human genome. Now they are editing it, and the promise of a truly personalised medicine, tailored to an individual's genetic makeup, is becoming reality. The first CRISPR-based technology, for sickle cell disease, is expected to soon be approved by US regulators. Such advances have the potential to bring enormous benefits for humankind, but also bring unique social and ethical challenges. Resolving these challenges will involve ongoing conversations within and outside the scientific community. To protect legitimate genetic research—to close loopholes in regulations and establish a global consensus on oversight and regulation—will require governance that is as dynamic as the science.

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Advances in Genetic Editing of the Human Embryo

Journal article

Astarăstoae, Vasile MD, PhD; Ioan, Beatrice Gabriela MD, PhD; Rogozea, Liliana M. MD, PhD; Hanganu, Bianca MD, PhD

American Journal of Therapeutics 30(2): p e126-e133, March/April 2023

Abstract

Background

Genetic engineering has allowed a major development of research in this field, with specialists attempting to edit the human genome, after the successful editing of the genomes of plants and animals. However, human gene editing technologies are at the center of ethical debates around the world.

Areas of Uncertainty

Ethical concerns about genetic editing of the human embryo raise several issues that can be viewed through the prism of optimism and reluctance leading to a number of recommendations regarding the acceptance of what may soon become a reality.

Data Sources

A literature search was conducted through PubMed, MEDLINE, Plus, Scopus, and Web of Science (2015–2022) using combinations of keywords, including: human genome or gene editing plus ethics.

Ethics and Therapeutic Advances

Gene therapy is seen by researchers as a way to solve congenital diseases, multifactorial diseases in general or specific diseases such as cystic fibrosis, muscular dystrophy, or can increase resistance to HIV infection. Genome editing technologies, germline gene editing, clustered regularly interspaced short palindromic repeats gene editing technology, technologies such as zinc finger nucleases are not only advanced gene therapies that require solving technical problems, but also techniques that require complex and complete analysis of ethical problems. Genetic engineering raises many ethical concerns such as: safety concerns especially the risk of off-target effects; autonomy of the individual—with the limitation of the future generations to consent for an intervention over their genome; social justice—keeping in mind the costs of the procedures and their availability to the general population. Discussions can go further from questions such as "How can we do this?" to questions such as "Should we do this?" or "Is society ready to accept this technology and is it able to manage it rationally?"

Conclusions

The ethics of biomedical research should be based on global dialogue, on the involvement of experts and the public, to achieve a broad social consensus. The fundamental review of the ethics of genetics is a desire and an opportunity of the current period.

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Germline Gene Editing: The Gender Issues

Research Article

Iñigo de Miguel Beriain, Ekain Payán Ellacuria and Begoña Sanz

Cambridge Quarterly of Healthcare Ethics (2023), 1–7, 27 February 2023

Open access

Abstract

Human germline gene editing constitutes an extremely promising technology; at the same time, however, it raises remarkable ethical, legal, and social issues. Although many of these issues have been largely explored by the academic literature, there are gender issues embedded in the process that have not received the attention they deserve. This paper examines ways in which this new tool necessarily affects males and females

differently—both in rewards and perils. The authors conclude that there is an urgent need to include these gender issues in the current debate, before giving a green light to this new technology.

Conclusions
From the discussion above, we may conclude that the introduction of gene editing in human reproduction would have an undetermined and perhaps double-edged effect on the position of women seeking to conceive. On the one hand, it could substantially reduce physical suffering, due to the improvements in IVF techniques that would be introduced. On the other, however, the extension of this technology requires the use of female oocytes that can only be obtained by submitting some women to a huge cost.43 Moreover, the success of this technology could in the more distant future introduce a separation between sex and human reproduction, triggering the use of IVF and gene editing technologies. In these circumstances, women could be forced to expose themselves to the discomforts and risks of a technology that has, as we have shown, serious drawbacks. These considerations imply the existence of a gender bias that should be included in the discussions on gene editing.

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Law, bioethics, and society: Jewish and Islamic approaches to fertility treatments and human germline genome editing

Thesis
Vanessa Goodwin
Doctor of Philosophy, The University of Edinburgh 2022
Abstract

In 2018, the Chinese biophysicist He Jiankui announced the birth of the first genetically modified babies; this broke an international moratorium. When the Russian biologist Denis Rebriko reported similar intentions, ethicists and legislators began working with an increased sense of urgency towards a global framework to define the limits of Human Germline Genome Editing (HGGE). This is because HGGE poses concerns for the safety of future generations, and reproductive tourism has the potential to undermine the legislation of any one country. Whilst secular international bioethics councils aim to find global consensus on this matter, religious jurists and bioethicists, though influential in their own communities, are not necessarily part of the same international debate.

The thesis proposes that the influence of religious law is considerable on societies, legislation and on fertility practices, especially in Israel and the Muslim Middle East, so the inclusion of religious legal viewpoints is an important aspect of any global consensus on bioethical issues. In Judaism and Islam the field of bioethics is a subcategory of contemporary religious law. As the legal narratives of Jewish law (Halakha) and Islamic law (Shari'a) are complex, the legal reasoning and influence of religious jurists has to be understood within their religious paradigm if they are to be successful integrated in the international debate. The thesis investigates the process by which contemporary Orthodox Jewish and Muslim jurists engage with the bioethical questions of reproductive medicine in general in order to understand their specific response to the potential permissibility of HGGE. It enquires how the religious legal systems have already adapted to reproductive technologies and how the legislation of fertility treatments in Israel and the Muslim Middle East incorporates the values and legal guidelines of Halakha (Jewish law) and Shari'a (Muslim law).

The thesis is divided into three parts. Parts I and II introduce the mechanisms by which Halakha and Shari'a respectively engage with new legal cases as a result of medical and scientific advancements and then explore the sociological context of applied Jewish and Muslim bioethics in the Middle East. Part III charts the development of the key legal debates concerning fertility treatments from the late twentieth century onwards in Orthodox Judaism and in Islam. It focuses on three 4 reproductive technologies: 1.) Artificial Insemination with Donor sperm (AID) 2.) In Vitro Fertilisation (IVF) and 3.) Pre-Implantation Genetic Diagnosis

(PGD). It finds that the legal opinions of prominent jurists in the late 20th century set precedents for all subsequent debates in both religious legal traditions including the current debate about the permissibility of HGGE. This evolving engagement of scientists and religious jurists demonstrates how Halakha and Shari'a both have normative legal principles that are rooted in the Torah, the Qur'an and in the wider scriptural tradition in both faiths. Legitimate conception and lineage retain their central importance in all debates about fertility treatments. However, the legal traditions have adapted significantly in the face of emerging reproductive medicine and the wider societal and ethical implications for the rights of the parent and the child.

Finally, this research studies the rapid acceptance of genetic screening programs in Israel and the Middle East and highlights the different approaches to the genetic improvement of societies. It finds a religious narrative which endorses the deselection of pre-embryos as an ethical alternative to abortion and explores how this may impact future debates about the permissibility of HGGE. Given this context it becomes apparent how Jewish and Muslim jurists debate the fundamental questions about the creation of human life and why their divergent legal judgements, which are generated for the moral good of their respective societies, matter in the global debate.

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ETHICAL ISSUES IN RELATION TO HUMAN GENOME EDITING

Points to consider in the Development of National Human Genome Editing Policy

Research Article

Dianne Nicol, Simon Niemeyer, Rebecca Paxton and Christopher Rudge

Cambridge University Press, accepted Manuscript (March, 2023), Open access

Abstract

CRISPER and other genome editing technologies have the potential to transform the lives of people affected by genetic disorders for the better. However, it is widely recognised that they also raise large ethical and policy questions. The focus of this article is on how national genome editing policy might be developed in ways that give proper recognition to these big questions. The article first considers some of the regulatory challenges involved in dealing these big ethical, social questions, and also economic issues. It then reviews the outcomes of a series of major reports on genome editing from international expert bodies, with particular focus on the work of the World Health Organisation's expert committee on genome editing. The article then summarises five policy themes that have emerged from this review of the international reports together with a review of other literature, and the authors' engagement with members of the Australian public and with a wide range of experts across multiple disciplines. Each theme is accompanied by one to three pointers for policy makers to consider in developing genome editing policy.

Genome Editing: Moving Toward a New Era of Innovation, Development, and Approval

Journal article

Houria Bachtarzi

Human Gene Therapy, Volume 34, Issue 5-6, Pages:171–176, Published Online:10 February 2023

Abstract

Therapeutic genome editing is currently reshaping and transforming the development of advanced therapies as more ex vivo and in vivo gene editing-based technologies are used to treat a broad range of debilitating and complex disorders. With first-generation gene editing modalities (notably those based on ZFNs, TALENs and CRISPR/Cas9), comes a new second-generation of gene editing-based therapeutics including base editing, prime editing and other nuclease-free genome editing modalities. Such ground-breaking innovative

products warrant careful considerations from a product development and regulatory perspective, that take into account not only the common development considerations that apply to standard gene and cell therapy products, but also other specific considerations linked with the technology being used. This article sheds light into specific considerations for developing safe and effective in vivo and ex vivo genome editing medicines that will continue to push barriers even further for the cell and gene therapy field.

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GENOMIC DATA/ BIOBANKING/GENOMIC RESEARCH

Data Protection in Healthcare-Integrated Biobanking
Research Article
Petra Duhm- Harbeck, Jens K. Habermann
Innovations in Digital Health, Diagnostics, and Biomarkers 2023; 3:1-7, 22 February 2023
Abstract
Introduction
Development of personalized medicine depends on research using clinical biospecimens and data. This interface between clinical care and translational research is increasingly served by hospital-integrated biobanks; yet their implementation is hampered by complex data regulations.
Methods
A generic data protection concept with a decision and application matrix was developed addressing five criteria: (1) organizational integration into university medicine, (2) biobank governance, (3) ethical and legal aspects, (4) specifications of the BSI (Bundesamt fu¨r Sicherheit in der Informationstechnik [Federal Office for Information Security]), and (5) FAIR (findable, accessible, interoperable, and reusable) principles for research data. Applicability was tested for the highest complexity level at Campus Lu¨beck.
Results
The data protection concept was approved by the local ethics committee as well as local and national data protection authorities. The concept allows an automated research-guided patient recruitment and data protection compliant information technology (IT) in connection to national and international research networks. It ensures university and hospital conformity with the EU Data Protection Regulation. Consent behavior of 277,766 patients over five years proved routine practicability (error rate 0.0013%; withdrawals 0.09%). Clinical staff obtained higher consent rates (85.6%) compared with consent rates for use of data only at central patient admission (56.1%); even though consents in central patient admission increased constantly during observation time.
Conclusion
The generic data protection concept can legitimately enable personalized medicine through biobanking in the clinical context.

REVISED - Respecting values and perspectives in biobanking and genetic research governance: Outcomes of a qualitative study in Bengaluru, India [version 2; peer review: 1 approved, 1 approved with reservations]
Journal article
Manjulika Vaz, Prasanna Warrier, Calvin Wai-Loon Ho, Susan Bull
Wellcome Open Research, accessed on 06 March 2023
Abstract
Background: The promise of biobanking and genetic research (BGR) in the context of translational research towards improving public health and personalised medicine has been recognised in India. Worldwide experience has shown that incorporating stakeholders' expectations and values into the governance of BGR

is essential to address ethical aspects of BGR. This paper draws on engagement with various stakeholders in the South Indian city of Bengaluru to understand how incorporating people's values and beliefs can inform policy making decisions and strengthen BGR governance within India.
Methods: We adopted a qualitative research approach and conducted six focus group discussions with civil society members and seven in-depth interviews with key informants in BGR, identified through a targeted web search and snowballing methods, until data saturation was reached. Data were thematically analysed to identify emergent patterns.
Results: Specific themes relating to the ethics and governance of BGR emerged. Fears and uncertainty about future sample and data use, possibilities of discrimination and exploitation in the use of findings and the lack of comprehensive data protection policies in India along with expectations of enhanced contributor agency, control in future use of samples and data, benefit sharing, enhanced utility of samples, sustained BGR and public good, reflected tensions between different stakeholders' values and beliefs. Fair governance processes through an independent governance committee for biobanks and a system of ongoing engagement with stakeholders emerged as best practice towards building trust and respecting diversity of views and values.
Conclusions: Ensuring public trust in BGR requires listening to stakeholders' voices, being open to counter narratives, and a commitment to long term engagement embedded in principles of participatory democracy. This is central to a 'people-centred governance framework' involving a negotiated middle ground and an equilibrium of governance which promotes social justice by being inclusive, transparent, equitable, and trustworthy.

Patients and Members of the Public's Wishes Regarding Transparency in the Context of Secondary Use of Health Data: A Scoping Review

Peer-reviewed preprint

Annabelle Cumyn, Jean-Frédéric Ménard, Adrien Barton, Roxanne Dault, Frédérique Lévesque, Jean-François Ethier

Journal of Medical Internet Research; 45002, 03 March 2023

Abstract

Background

Learning Health Systems rely on the secondary use of health data to improve care. Transparency regarding this secondary use is frequently cited as necessary to increase patient awareness, support alternative approaches to consent, and foster trust.

Objective

To review the current published literature to identify different stakeholders' perspectives and recommendations on what exactly should be communicated to members of the public regarding the secondary use of health data for research, how and at what conditions.

Methods

Using PRISMA-ScR guidelines, we conducted a scoping review through several bibliographic databases (Medline, CINAHL, PsycINFO, Scopus, Cochrane Database of Systematic Reviews, and PubMed) to locate a broad range of studies published in English or French up to November 2022. We included articles that reported a stakeholder's opinion or recommendations of what should be communicated to patients or members of the public regarding the secondary use of health data for research, how to communicate the information or at what conditions. Data were collected and analyzed using an iterative thematic approach with NVivo software.

Results

A total of 178 articles was included in this scoping review. Communication was deemed crucial for many purposes including: a) educating patients and members of the public on the potential benefits; b) giving some control over data use c) as a form of reciprocity and, d) as a condition to build and maintain trust. Elements that should be communicated include generic content such as governance and regulatory

frameworks, scientific aims and potential future uses of the data and specific content that is relevant to each person with regards to the use of their data. Methods for communication generally favored broad approaches such as nationwide publicity campaigns, mainstream and social media for generic content and mixed approaches for specific content including websites, patient portals and face-to-face encounters. Content should be tailored to the individual as much as possible with regards to length, avoidance of technical terms, cultural competence, and level of detail.

Conclusions

This review can serve as a foundation for evaluating current communication approaches with regards to secondary use of health data or designing future strategies. Future work will be needed to assess which strategies achieve the greatest outreach while striking a balance between transparency and utilization of resources.

Using Population Descriptors in Genetics and Genomic Research: A New Framework for an Evolving Field

Consensus Study Report

National Academics of Sciences, Engineering, and Medicine, 2023

Description

Genetic and genomic information has become far more accessible, and research using human genetic data has grown exponentially over the past decade. Genetics and genomics research is now being conducted by a wide range of investigators across disciplines, who often use population descriptors inconsistently and/or inappropriately to capture the complex patterns of continuous human genetic variation.

In response to a request from the National Institutes of Health, the National Academies assembled an interdisciplinary committee of expert volunteers to conduct a study to review and assess existing methodologies, benefits, and challenges in using race, ethnicity, ancestry, and other population descriptors in genomics research. The resulting report focuses on understanding the current use of population descriptors in genomics research, examining best practices for researchers, and identifying processes for adopting best practices within the biomedical and scientific communities.

Security and Sharing of NIPT Data Are the Basis of Ethical Decision-Making Related to Non-Medical Traits

Open Peer Commentaries

Wenke Yang, Zhenglong Guo, Weili Shi, Litao Qin, Xiaoliang Xia, Bingtao Hao & Shixiu Liao

The American Journal of Bioethics, Volume 23 (3), Pages 29-31, 15 March 2023

Article Info

Bowman-Smart et al. (2023) outlined the scenario that with the content expansion of noninvasive prenatal testing (NIPT) in the future, its application is not limited to the screening for aneuploidies, microdeletions and single-gene disorders of the fetus. More comprehensive genomic data and polygenic risk scores can provide information on "sub-clinical"" or "non-medical" traits of fetuses in early pregnancy. It also presents significant challenges for ethical decision-making, and data management. Thus, an ethical framework needs to be clearly defined to preemptively address possible inconsistencies in ethical decision making related to "non-medical" traits.

The genetic data generated by cell-free NIPT con-sists of maternal and fetal genomes, which involves prominent privacy concerns (Liu et al. 2018), so that the data security protection is necessary. However, excessive security protection will confine data sharing, which is not beneficial to the development of NIPT. Therefore, the challenges of NIPT data security and sharing need to be considered when developing an ethical framework. The security of genome data generated in NIPT involves fetal medical experts, genetic counselors, ethics committees, and relevant legal provisions. Any improper management or misuse of genomic data will lead to privacy leakage, which may cause discrimination and unequal risk to personal social

activities, such as employment, medicare, or commercial insurance. Thus, data security is fundamental to an ethical decision-making framework and should be given due attention.

Data security management runs throughout the process of data collection, transmission, storage, access, analysis, provision, and deletion to ensure effective protection and legal use of data. First, partici-pants, investigators, and institutes should collect genetic information in a way that conforms to laws and ethics, respect the privacy and autonomy of the sub-jects, and keep the use of genetic information within the scope of informed consent. Second, the security management in genomic data transmission should be comprehensive, including not only managers but also networks, software, equipment. According to the June 2022 report of the U.S. Food and Drug Administration and Cybersecurity and Infrastructure Security Agency (ICSA-22-153-02), a network security vulnerability exists in the Local Run Manager software of some sequencing platforms of Illumina company, a popular NIPT sequencing platform. This may lead to tampering of clinical sequencing results or leakage of medical data and private information. Illumina explained the risk of this vulnerability and released a repair patch in time. Third, although anonymization of clinical specimens and encryption of genomic data can avoid privacy attacks to some extent (Bonomi et al. 2020), there are still loopholes in the de-identified data, which is not enough to fully protect…..

Genetic determinism, essentialism and reductionism: semantic clarity for contested science

Perspective

Nature Reviews Genetics volume 24, pages197–204 (2023), 31 October 2022

Abstract

Research linking genetic differences with human social and behavioural phenotypes has long been controversial. Frequently, debates about the ethical, social and legal implications of this area of research centre on questions about whether studies overtly or covertly perpetuate genetic determinism, genetic essentialism and/or genetic reductionism. Given the prominent role of the '-isms' in scientific discourse and criticism, it is important for there to be consensus and clarity about the meaning of these terms. Here, the author integrates scholarship from psychology, genetics and philosophy of science to provide accessible definitions of genetic determinism, genetic reductionism and genetic essentialism. The author provides linguistic and visual examples of determinism, reductionism and essentialism in science and popular culture, discusses common misconceptions and concludes with recommendations for science communication.

Future on a Flashdrive: Timely Considerations for the Imminent Adoption of Whole Genome Sequencing in Pediatric Healthcare

Article

Ella Hohmann, Brian King, Robert A.S. Laroche, Adam M. Navara, Alexis F. Wilkinson

Journal of Science Policy & Governance, Volume 21, Issue 03, 23 January 2023

Open access

Executive Summary

In just twenty years, humanity has progressed from the first sequenced human genome to the ability to sequence one in a matter of hours and for only hundreds of dollars. This rise in affordability and speed has enabled physicians to use whole genome sequencing (WGS) as a diagnostic tool, particularly in cases of rare disease in pediatric patients where it has already demonstrated immense potential. However, such a rapid development in technology powerful enough to unlock a person's genetic information has also led to necessary questions regarding when and how it is applied. In this assessment, we discuss the implications of WGS adoption in pediatric healthcare, focusing specifically on ensuring ethical and equitable collection and communication of genomic data as well as the need for secure and accessible data storage methods. We identify several key areas where further policy is most pressing and provide value-driven recommendations

centered on guaranteeing pediatric patient safety, equity, and empowerment during the broader introduction of WGS tools. In particular, we advocate for legal frameworks that limit present usage of WGS to only those patients with a clear and present need, guidelines that expand the labor force that can conduct WGS, increasing access and equity, improved standards for storage, access, and sharing of WGS data, and finally expanding Medicaid coverage to include WGS use in critical care settings.

Realizing the Potential of Genomics across the Continuum of Precision Health Care - Proceedings of a Workshop

Workshop report

National Academies of Sciences, Engineering, and Medicine; Health and Medicine Division; Board on Health Sciences Policy; Roundtable on Genomics and Precision Health;

Theresa M. Wizemann, Kathryn Asalone, Meredith Hackmann, and Sarah Beachy, Rapporteurs

NASEM – National Academies of Science, Engineering, Medicine (2023)

The National Academies Roundtable on Genomics and Precision Health, in collaboration with the National Cancer Policy Forum, hosted a public workshop that examined how genomic data are used in health care, outside of the traditional settings for clinical genetics. The workshop identified opportunities for advancement of precision health care delivery. The event also explored how patients, clinicians, and payers assess and act upon the risks and benefits of genomic screening and diagnostic testing. Discussions focused on strategies to ensure that genomic applications are responsibly and equitably adopted to benefit populations as well as individuals over time. This Proceedings of a Workshop summarizes content from the event. PDF: https://nap.nationalacademies.org/login.php?record_id=26917

Webinar report: stakeholder perspectives on informed consent for the use of genomic data by commercial entities

Short report

Baergen Schultz, Francis E Agamah, Cornelius Ewuoso, Ebony B Madden, Jennifer Troyer, Michelle Skelton, Erisa Mwaka On behalf of H3Africa Ethics and Community Engagement Working Group

Journal of Medical Ethics, Published Online First: 20 March 2023.

Abstract

In July 2020, the H3Africa Ethics and Community Engagement (E&CE) Working Group organised a webinar with ethics committee members and biomedical researchers from various African institutions throughout the Continent to discuss the issue of whether and how biological samples for scientific research may be accessed by commercial entities when broad consents obtained for the samples are silent. 128 people including Research Ethics Committee members (10), H3Africa researchers (46) including members of the E&CE working group, biomedical researchers not associated with H3Africa (27), representatives from the National Institutes of Health (16) and 10 other participants attended the webinar and shared their views. Several major themes emerged during the webinar, with the topics of broad versus explicit informed consent, defining commercial use, legacy samples and benefit sharing prevailing in the discussion. This report describes the consensus concerns and recommendations raised during the meeting and will be informative for future research on ethical considerations for genomic research in the African research context.

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ETHICAL REGULATION AND GUIDANCE

China Engages in International Regulation of Disclosure Obligation

Book Chapter

Wenting Cheng

China in Global Governance of Intellectual Property, Palgrave Macmillan, pp 81–113, 11 February 2023

Abstract

This chapter explores how China has engaged in international regulation concerning the disclosure obligation in patent applications. “Disclosure obligation” refers to the mandatory requirement to disclose the origins/sources of genetic resources in patent applications. This chapter first analyses polarised positions in post-TRIPS multilateral negotiations on this issue. Despite that no international consensus has been reached, China actively learned from international proposals submitted to the WTO TRIPS Council and the WIPO IGC, engaged in international negotiation, and created a disclosure mechanism in its Patent Law (2008) with its own characteristics. Generally, China took a pro-development position to support the disclosure obligation in multilateral negotiations after its patent law and incorporated provisions on the protection of genetic resources in its FTAs. By supporting negotiations towards a binding disclosure obligation in the TRIPS Agreement, China’s activities contribute to enhancing global distributive justice from a cosmopolitan perspective.

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GOVERNANCE FRAMEWORK

Global health and global governance of emerging biomedical technologies

Journal Article

Bryan Cwik

Journal of medical ethics, 07 February 2023

Abstract

Global governance of emerging, disruptive biomedical technologies presents a multitude of ethical problems. The recent paper by Shozi et al raises some of these problems in the context of a discussion of what could be the most disruptive (and most morally fraught) emerging biomedical technology—human germline genome editing. At the heart of their argument is the claim that, for something like gene editing, there is likely to be tension between the interests of specific states in crafting regulation for the technology, and disagreement about what would be necessary to meet the requirements for responsible translation of gene editing into the clinic. This complicates hopes for a tidy, algorithmic process of crafting global governance via frameworks for regulation built around core ‘ethical values and principles’ (as they are called in the WHO Framework), and also forces us to confront deeper philosophical questions about biotechnology and global health.

Editor’s note: The referenced paper by Shozi et al: Future of global regulation of human genome editing: A south African perspective on the WHO Draft Governance Framework on Human Genome Editing

Ethical governance model for the data economy ecosystems

Journal Article

Jani Koskinen, Sari Knaapi-Junnila, Ari Helin, Minna Marjaana Rantanen, Sami Hyrynsalmi

Digital Policy, Regulation and Governance, 10 February 2023

Abstract

Purpose

Data economy is a recent phenomenon, raised by digital transformation and platformisation, which has enabled the concentration of data that can be used in economic purposes. However, there is a lack of clear

procedures and ethical rules on how data economy ecosystems are governed. As a response to the current situation, there has been criticism and demands for the governance of data use to prevent unethical consequences that have already manifested. Thus, ethical governance of the data economy ecosystems is needed. The purpose of this paper is to introduce a new ethical governance model for data economy ecosystems. The proposed model offers a more balanced solution for the current situation where a few global large-scale enterprises dominate the data market and may use oligopolistic power over other stakeholders.

Design/methodology/approach

This is a conceptual article that covers theory-based discourse ethical reflection of data economy ecosystems governance. The study is based on the premise of the discourse ethics where inclusion of all stakeholders is needed for creating a transparent and ethical data economy.

Findings

This article offers self-regulation tool for data economy ecosystems by discourse ethical approach which is designed in the governance model. The model aims to balance data "markets" by offering more transparent, democratic and equal system than currently.

Originality/value

By offering a new ethically justified governance model, we may create a trust structure where rules are visible and all stakeholders are treated fairly.

Ethics governance development: The case of the Menlo Report

Research Article

Megan Finn and Katie Shilton

Social Studies of Science, 19 February 2023

Open access

Abstract

The 2012 Menlo Report was an effort in which a group of computer scientists, US government funders, and lawyers produced ethics guidelines for research in information and communications technology (ICT). Here we study Menlo as a case of what we call ethics governance in the making, finding that this process examines past controversies and enrols existing networks to connect the everyday practice of ethics with ethics as a form of governance. To create the Menlo Report, authors and funders relied on bricolage work with existing, available resources, which significantly shaped both the report's contents and impacts. Report authors were motivated by both forward- and backward-looking goals: enabling new data-sharing as well as addressing past controversies and their implications for the field's body of research. Authors also grappled with uncertainty about which ethical frameworks were appropriate and made the decision to classify much network data as human subjects data. Finally, the Menlo Report authors attempted to enrol multiple existing networks in governance through appeals to local research communities as well as taking steps towards federal rulemaking. The Menlo Report serves as a case study in how to study ethics governance in the making: with attention to resources, adaptation, and bricolage, and with a focus on both the uncertainties the process tries to repair, as well as the new uncertainties the process uncovers, which will become the site of future ethics work.

Editor's note; The referenced Menlo Report: The Menlo Report: Ethical Principles Guiding Information and Communication Technology Research

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PUBLIC AND COMMUNITY ENGAGEMENT/ EDUCATION

Victims of eugenic sterilisation in Utah: cohort demographics and estimate of living survivors

Article

James Tabery, Nicole L. Novak, Lida Sarafraz, Aubrey Mansfield

The Lancet Regional Health-Americas, Volume 19, 100436, 15 February, 2023

Open access

Summary

Background

Eugenicists at the beginning of the twentieth century feared that the "unfit" were outbreeding the "fit" and promoted interventions like sterilisation as a solution to the perceived problem. Over 60,000 people were sterilised across the United States, victims of eugenic programs implemented in 32 states. Utah had a particularly aggressive eugenic sterilisation program, hailed by eugenicists for sterilising such a large proportion of its population, and lasting well into the 1970s. The goal of the present study was to determine who, at the demographic level, was targeted by this eugenic practice in Utah, and to also estimate how many survivors of the program might still be alive in 2023.

Methods

We used archival records and data abstracted from charts at the Utah State Developmental Center to construct an observational cohort of people sterilised under Utah's coercive, eugenic sterilisation program. We described the demographics of the cohort and presented a life table analysis to estimate the number of survivors still living in 2023.

Findings

At least 830 men, women, and children (modal age of 15–19, 53.6% female) were sterilised in Utah institutions under a program that was launched in 1925, peaked in the 1940s, and concluded in the 1970s. The life table analysis predicts approximately 54 survivors (36 women, 18 men), with an average age of 78.

Interpretation

Many people sterilised under Utah's eugenics law are likely living today. While some states have taken steps to reckon with their roles in depriving people of their reproductive rights, Utah lacks even an official acknowledgment of this shameful, medical history. Given the advanced age of the potential survivors, time is running out for a reconciliation that can be experienced by those who were most harmed by the practice.

Do genomic passports leave us more vulnerable or less vulnerable? Perspectives from an online citizen engagement

Article

Humanities and Social Sciences Communications, Volume 10 (83), 04 March 2023

Open access

Abstract

Since genomics is becoming commonplace in healthcare for the diagnosis, treatment, and prevention, the prospect of generating a genomic passport for all citizens is gaining traction. While this would have many advantages, it raises ethical issues requiring societal debate alongside academic reflection. Hence, Sciensano—the Belgian Scientific Institute of Public Health—organised an online citizen engagement on genomic information usage, including a question on a genomic passport for all. The inductive thematic analysis of participants' contributions highlighted vulnerability as a fundamental concern, while this has not received sufficient attention so far in genomics. Participants expressed their vulnerability in two ways. First, the genomic passport would inform them about their ontological vulnerability. By revealing their constitutional weaknesses (predisposition to diseases), it reminds them that everyone is unavoidably and perennially at risk of being harmed. Second, the misuse of the genomic passport can add situational

vulnerabilities (e.g., discrimination causing psychological and economic harm). Moreover, the fundamental uncertainty in genomics—how will such sensitive information be used, and how will the science evolve? — exacerbates these vulnerabilities. This article ends with recommendations to alleviate these vulnerabilities in genomics now and in the future in which the genomic passport may become a reality.

…Conclusion

Confronted with the idea of a genomic passport for all citizens, participants from the Belgian online DNA debate expressed a sense of vulnerability in two ways. First, in their view, the genomic passport is a personal code embodying their ontological vulnerability. It reminds them that they are unavoidably vulnerable due to constitutional factors, such as psychological frailty (e.g., anxiety), physical weakness (e.g., diseases), and finitude (death). Second, genomic data misuse can exacerbate this first perennial vulnerability by adding situational vulnerabilities, such as exploitation, discrimination, inequalities, exclusion, and stigmatisation based on genetic makeup.

We propose some policy recommendations to alleviate these vulnerabilities inherently linked to genomic technologies usage in a public setting. These recommendations combine individuals' empowerment, protective measures, and trust:

- Any implementation of a genomic passport should aim at improving preventive and curative health care provision for the citizens whose genomic information is catalogued to avoid genetic vulnerabilities (e.g., predispositions) becoming concrete harms (e.g., diseases). However, public health policies should not fall into a health imperative imposing paternalistic measures, for instance, pressuring individuals to know and take their genomic information into account. Adequate information, education, and respect for autonomy could empower individuals dealing with their genomic information throughout life.
- All genomic data users should also identify external factors causing situational vulnerability to erase, avoid or minimise them. They should ensure that protective and preventive measures are implemented—such as data security, prohibiting access to non-legitimate actors, and legal sanctions in the case of infringements.
- Organising public engagement initiatives supported by policymakers and genomic experts have two main advantages. First, they ensure that citizens' values and concerns, such as vulnerability, are understood and considered in the public health policy agenda and daily medical practice. Second, they could improve citizens' trust in genomics and thus relieve part of their sense of vulnerability.

Defining epigenetic literacy: How to integrate epigenetics into the biology curriculum

Research Article

Niklas Gericke, Birgitta Mc Ewen

Journal of Research in Science Teaching, 02 March 2023

Open access

Abstract

The aim of this study is to define epigenetic literacy and describe how it can be included in school biology. Epigenetics is a new field of research in biology with abundant societal consequences and conceptual implications on how genetics is understood. Epigenetics explains how genes are regulated, thereby clarifying cell differentiation, and providing an understanding of how the environment interacts with genes. Students are bound to encounter epigenetic knowledge and applications related to issues such as health, food, and exercise in the media and their everyday lives. Consequently, there is a need to develop epigenetic literacy. Nevertheless, epigenetics is missing in biology curricula and is almost unknown among teachers and students. Research on epigenetics in science education is scarce, and we do not know what and how to teach. Therefore, we conducted a policy Delphi study with a panel of experts to define an epigenetic literacy framework for teaching in secondary education in relation to Robert's Vision I and Vision II perspectives on epigenetic literacy. Participants were 41 recognized international experts representing 11 countries and five

areas of expertise. The experts suggested that epigenetics should be introduced in the lower secondary genetics course (students aged 13–15 years), but also addressed in other relevant areas of biology. The study generated six content themes: epigenetics as a metaphor; epigenetics connecting nature with nurture; epigenetics as a dynamic process; epigenetic mechanisms; epigenetics and inheritance; and epigenetics and nature of science, and five sociocultural themes for contextualization: epigenetics and lifestyle; epigenetics and diseases; epigenetics and ethics; epigenetics and policies; and epigenetics and forensics. Taken together, these themes constitute the epigenetic literacy framework. Further, we uncover divergent meanings in the expert panel—as is typical of policy Delphi studies—and connect the framework to genetic literacy and learning progressions in genetics education.

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ACCESS, EQUITY, BENEFITS SHARING

Global Oncology Medical Diplomacy Working Group Inaugural Meeting: Defining Worldwide Barriers to Germline Genomics in Cancer Prevention and Management

Viewpoints

Abou-Alfa, Ghassan K., Larry Norton, The Global Oncology Medical Diplomacy Working Group

Annals of Global Health, 89(1), 21 February 2023

Abstract

We convened an international working group to examine the issues that challenge equity and inclusion in genetic medicine. Specifically, 72 internationally known experts in oncology and cancer genetics from 34 countries (the Global Oncology Medical Diplomacy Working Group), gathered virtually on January 4–5, 2022, for the “Humanity Cancer Germline Convergence and Divergence Cancer Predispositions” conference hosted by Memorial Sloan Kettering Cancer Center, in collaboration with the United Arab Emirates Ministry of Health and the Al Jalila Foundation. The goal of the conference was to broaden transnational understanding of the current state of genetics in preventive and therapeutic cancer medicine, and to define barriers to increased uptake of germline genomics to decrease the international burden of cancer. Here, we highlight the overarching barriers that were defined through this effort. These global barriers to incorporating germline genomics into optimal cancer care can inform ongoing research, collaboration, and advocacy for equitable, cost-effective genomic medicine for populations worldwide.

Personalised Medicine—Implementation to the Healthcare System in Europe (Focus Group Discussions)

Article

Dorota Stefanicka-Wojtas and Donata Kurpas

Journal of Personalized Medicine, 13, 380, 21 February 2023

Abstract

Background

Personalized medicine (PM) is an approach based on understanding the differences between patients with the same disease and represents a change from the “one size fits all” concept. According to this concept, appropriate therapies should be selected for specific groups of patients. PM makes it possible to predict whether a particular therapy will be effective for a particular patient. PM will still have to overcome many challenges and barriers before it can be successfully implemented in healthcare systems. However, it is essential to remember that PM is not a medical revolution but an evolution.

Methods

Three focus groups were conducted, to achieve the purpose of this study, which was to identify the barriers and facilitators existing to the implementation of PM and to highlight existing practices in European

countries. Focus group discussions covered the areas of barriers and facilitators to the implementation of personalized medicine.
Results
This section describes the results of the focus groups that covered the areas of barriers and facilitators of personalized medicine implementation.
Conclusions
Personalized medicine faces many challenges and barriers before it can be successfully implemented in health systems. The translation of PM to European countries, differences in regulations, high costs of new technologies, and reimbursement are the reasons for the delay in PM implementation.

Precision health now and in the future

Report
Fay Sowerby
Breast Cancer Aotearoa Coalition, 27 January 2023, 48 pages
Executive Summary [Excerpt]
The Breast Cancer Aotearoa Coalition (BCAC) represents over 30 breast cancer charities and groups across Aotearoa, as well as individual members. Our purposes are to support, inform and represent those diagnosed with breast cancer in Aotearoa from an evidence basis. We agree that precision health, including precision medicine and integral components including tools and technologies, require urgent focus to enable innovation in the near and longer term. Bringing focus to the various elements of precision health will lead to development and improvements across our health system, through ongoing research and clinical trials.
We see New Zealand's purposeful adoption of precision health, including precision medicine, as vital to improving New Zealanders' health outcomes and we welcome this opportunity to contribute to this consultation as Aotearoa progresses from the status quo…

Editor's note: Aotearoa is an indigenous name for people in New Zealand

Facing our history—Building an equitable future

ASHG Report
Chazeman S. Jackson, Daria Turner, Maya June, and Mona V. Miller
The American Journal of Human Genetics 110, 377–395, 02 March 2023
Open access
Summary
This report is the product of a major year-long initiative to acknowledge and reckon with past injustice, as well as progress toward justice, within the American Society of Human Genetics (ASHG) and the broader field of human genetics. Approved by the ASHG Board of Directors and launched in 2021, the initiative was sparked by the social and racial reckonings in 2020. The ASHG Bboard of Directors asked ASHG to acknowledge and provide examples of how human genetics theories and knowledge ''have been used to feed and justify racism, eugenics, and other systemic forms of injustice, and to focus specifically on examples of ASHG's role in fostering or failing to rebuke harms and on steps the Society could take to address findings.'' The initiative was undertaken with support and input from an expert panel of human geneticists, historians, clinician-scientists, equity scholars, and social scientists and included a research and environmental scan, four expert panel meetings, and a community dialogue as its main activities.

Equity of Access and Return in Global Genomics

Report

Global insights, Public Policy Projects 2022, 32 pages

Key Insights

- Countries are in completely different stages of readiness for the implementation of genomics programmes focused on human healthcare. This may not be best achieved by focusing on rare disease and personalized medicine, which can be interpreted as western luxuries.
- It is no longer good enough to say that communities should participate in genomics research for the global good – much greater focus needs to be placed on creating an equitable approach to benefit sharing, recognising the barriers to participation that communities face and acknowledging and accepting them, rather than blaming excluded cohorts for not participating.
- The challenges of creating a more equitable approach to access and return in global genomics is not solely an issue of creating a more diverse dataset, nor is it a challenge faced exclusively by low-and-middle-income countries (LMICs). Every jurisdiction in the world has much to do to address these challenges for all of their populations, including their Indigenous and minority communities.
- The WHO has a huge role to play in helping to articulate and support global participation in genomics. The recommendations of the WHO Science Council Report are far-reaching and implementation of all may be challenging.
- Industry in all of its guises has a significant role to play in bringing the benefits of genomics equitably to the world but lacks a coherent means to engage. This means that the benefits of what industry can do are often not widely known, particularly in those jurisdictions which arguably have the greatest need. Different elements of industry will have different considerations and different means of making a contribution but the concept of a Voluntary Code of Conduct merits further consideration.

Conclusion

It is morally imperative that the genomics ecosystem collaborates to facilitate equitable global access to genomics. While achieving this in isolation is a difficult task, cross-sectoral coordination alongside support from multilateral organisations such as the WHO can mobilise international decisionmakers to implement strategies that will seek to provide genomics-derived health interventions to every individual that needs them.

This report acknowledges that altruism alone is an insufficient motivator for participation in genomics research and tackling this requires a better means of delivering benefits to individuals, in a way that is cognisant of their needs. Furthermore, providing equitable access to genomics is a global problem that every country in the world struggles with; individual countries must endeavour to make genomics accessible to their entire population, with particular consideration shown for minority and Indigenous Communities. Unfortunately, there is no one-size-fits-all approach to articulating the benefits of genomic sequencing and countries must adopt bespoke approaches when articulating the healthcare and economic rationale behind implementing genomics within their healthcare systems.

This report addresses that the ongoing and potential roles of industry in making genomics equitably accessible is not widely discussed, nor is it consistently demonstrated by the many actors within sub-sectors of the genomics industry. Contributors suggested that industry representatives should consider signing a Voluntary Code of Conduct in which their individual means of making a contribution is considered. Continued support from the WHO has the potential to catalyse global action towards making genomics equitably accessible across the world and the same principles of cross sectoral collaboration will be essential in enabling this.

Many of these issues are exacerbated by complex social challenges including public trust in science and global health and economic inequality. Addressing these insights requires collaboration at a global scale

between all components of the genomics ecosystem. While suggesting specific strategies to address these challenges falls outside the scope of this work, PPP's Global Genomics 2023 Programme aims to continue facilitating relationships between all stakeholders in a manner that aims to benefit both the genomics ecosystem and the global population.

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GENETIC SCREENING/ GENETIC COUNSELLING

Prenatal Genetic Testing, Abortion, and Disability Justice

Book

Amber Knight and Joshua Miller

Oxford University Press, 2023

Scope

This book analyses women's reproductive autonomy in the age of genomic medicine, at a time when prenatal genetic screening is the new normal in obstetric practice. Why women's reproductive autonomy? Throughout the writing process, we have debated which language to use in the book.

"Expectant mothers" or "prospective parents"? "Pregnant women," "pregnant persons," or "gestating persons"? After all, it is not just cisgendered women who experience pregnancy, since transmen and non-binary individuals also give birth and raise children. The transgender rights movement has urged the use of gender-neutral language, such as "pregnant people" and "parent." Although there are inclusionary benefits to speaking about parenting in gender-neutral terms, such language also masks the ways in which reproduction and parenthood are deeply gendered experiences.

A binary understanding of gender is an indispensable part of the infrastructure of the public policy, political economy, and social norms that inform contemporary reproductive politics. To avoid the risk of making sexism and the specific oppressions of women invisible, we frequently employ the terms "woman" and "mother," although we do so with the recognition that the boundaries of these terms are socially contingent, fluid, and open to contestation and revision. Moreover, our focus on women's reproductive autonomy has obvious limitations, since the book says little about how reproductive decision-making might play out for cismen, transmen, or non- binary individuals…

Points to consider in the practice of postmortem genetic testing: A statement of the American College of Medical Genetics and Genomics (ACMG)

ACMG statement

Joshua L. Deignan, Mauricio De Castro, Vanessa L. Horner, Tami Johnston, Daniela Macaya, Joseph J. Maleszewski, Honey V. Reddi, Marwan K. Tayeh, ACMG Laboratory Quality Assurance Committee

Genetics in Medicine, 16 February 2023

Introduction

A traditional autopsy involves both histopathological examination of tissues and toxicology studies and is often used to help obtain a postmortem diagnosis in cases of sudden death. More recently, molecular technologies including next-generation sequencing are being used to assist in establishing or supporting a diagnosis when traditional autopsies fail to uncover a cause. Next-generation sequencing methods can also be used to more fully characterize a variety of conditions identified at autopsy that are suspected of having a heritable cause. For specific clinical indications such as sudden arrhythmic death syndrome, postmortem genetic testing has a relatively high diagnostic yield, leading to a molecular diagnosis in approximately 30% of traditional autopsy-negative cases. As "molecular autopsies" involving postmortem genetic testing become more common, there is a need to address the unique set of challenges and issues inherent in postmortem

testing. Challenges with postmortem genetic testing include difficulties in obtaining appropriate specimens for testing, a lack of insurance reimbursement for genetic testing of deceased individuals, the often limited availability of complete phenotypic information to help guide interpretation of genetic test results, and concerns around obtaining appropriate consent for the individual and/or family members. In addition, there is no consensus guidance for laboratories regarding how to approach variant reporting for postmortem diagnostic testing (including whether it should be treated differently than routine genetic testing). This new points to consider statement will address these and other concerns related to the use of "molecular autopsies" from the perspective of laboratories, genetic counselors, and clinicians.

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PLANTS, CROPS, AGRICULTURE, ANIMALS

CRISPR Crops and Sustainable Agriculture

Editorial
Rodolphe Barrangou
The CRISPR Journal, Volume 6, Issue 1 / February 2023, Published Online:9 Feb 2023
Executive summary

For most of the past decade, the CRISPR community has focused primarily on the deployment of the genome editing toolbox in therapeutics. Many companies have advanced preclinical programs and the pioneers have ushered in a new era of gene and cell therapies, with milestones already reached in the clinic. This will have tangible benefits for patients and significant consequences for sponsoring biopharmaceutical companies.

But there is so much more to CRISPR than just the clinic. As the genome editing field progresses, the disruptive toolbox is increasingly being deployed across the tree of life. The plant kingdom especially stands to benefit tremendously from lessons learned in therapeutics and biotechnology—at speed and at scale. With less fanfare but huge potential for impact, genome editing technologies are poised to revolutionize agriculture at a time when sustainability is a global priority.

Molecular breeders have been waiting for a game changer, and the emergence of CRISPR technologies has delivered. Decades of investments in plant genetics, trait mapping, and hybrid breeding have yielded tremendous gains in productivity and pest resistance. We are now transitioning to a greater focus on crop quality and sustainability, with plant resilience on top of mind. The farmer spirit of doing a lot with relatively little is the ethos of ag-focused companies committed to providing a safer, healthier, and more sustainable food supply.

Even foresters have embarked on enhanced carbon capture and next-generation breeding practices, although with longer timelines in play. This quest is already attracting talent and investments, whereas climate change awareness provides timely visibility and overdue attention. Adventurous investors and strategic partners have started to commit to these endeavors.

From Clinical to Field Trials

As featured on the cover of this issue, the seeds for CRISPR crops have been planted. Edited rice is moving from the laboratory to the field. In the article on page 62 of this issue, Gurel et al. illustrate how Cas12-based multiplexed editing can be deployed safely, specifically and inheritably in plants to alter agronomically relevant traits.

Encouragingly, the growth rate for the genome editing literature in plants and ag-relevant organisms is increasing, as promising greenhouse results get harvested across the planet. The global industrial

commitment in this area is poised to reach commercial milestones in 2023, with the anticipated launch of edited crops, fruits, and vegetables in several markets. As the farming landscape visually and genetically evolves, progress continues to be made regarding public awareness and governmental regulation of edited foods.

Against a backdrop of widespread enthusiasm, leading regulatory agencies are (re)opening the dossiers and their minds, even in Europe (as discussed in these pages before). Notwithstanding pockets of antiscience skepticism, CRISPR awareness in combination with our need to more responsibly and sustainably manage our agricultural practices presents unique opportunities to shift attention from editing in the clinic to the farm.

As 2023—our sixth year of publishing—unfolds, we look forward to covering advances in the deployment of genome editing technologies in the field and on the farm. We anticipate considerable progress in the editing of crops and livestock as well as in the forestry space during the year. The geopolitical stakes of the bioeconomy in the context of an intercontinental food supply chain are material, especially given ever-increasing pressure from population, land usage, and globalization trends. Exactly how fast and wide CRISPR crops will grow remains to be seen.

Gene Drive: Past, Present and Future Roads to Vertebrate Biocontrol

Review

Gus R. McFarlane, C. Bruce A. Whitelaw, and Simon G. Lillico

Applied Biosciences; 2(1):52-70, 13 February 2023

Open access

Abstract

Scientists have long sought a technology to humanely control populations of damaging invasive pests in a species-specific manner. Gene drive technology could see this become a reality. This review charts the twists and turns on the road to developing gene drives in vertebrates. We focus on rodents, as these will likely be the first targets, and trace the journey from the early understanding of selfish genetic elements to engineering gene drives in mice; before discussing future research focuses and the crucial role that public perception and governance will play in the application of this technology. The realization of robust gene drive strategies in vertebrate pests has the potential to revolutionize biocontrol.

Gene-Edited Food Adoption Intentions and Institutional Trust in the United States: Benefits, Acceptance, and Labeling

Original Article

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Open access

Abstract

New gene editing techniques, such as CRISPR-Cas9, have created the potential for rapid development of new gene-edited food (GEF) products. Unlike genetically modified organism foods, there is limited research and literature on U.S. public opinions about GEFs. We address this knowledge gap by examining how crop-based GEF adoption is linked to public trust in institutions and values using the Theory of Planned Behavior. We employ ordinal regression models to predict adoption intentions (direct benefits, acceptability, willingness to eat, and labeling) using a unique and nationally representative survey of n = 2,000 adults in the United States. We find that adoption hinges on public trust in institutions overseeing GEF development, especially trust in university scientists. The 29 percent of Americans likely to adopt GEFs highly trust government food regulators and the biotech industry. A nearly equal number of likely non-adopters distrust current regulatory systems in favor of consumer and environmental advocacy groups. However, most Americans (41 percent) are uncertain about GEF adoption and whom to trust. Although 75 percent of Americans want GEFs labeled,

few trust government agencies who have authority to issue labels. Our findings suggest public trust in GEFs and labels can only be obtained by tripartite oversight by universities, advocacy groups, and government food regulators.

Genome-edited salmon: a sustainable and socially acceptable solution to aquaculture?

Thesis

Torill Blix

The Artic University of Norway, A dissertation for the degree of Philosophiae Doctor, December 2022

Abstract

The Norwegian Atlantic salmon farming industry is halted by challenges related to environmental impact and fish welfare. Some of the issues have been suggested solved by the use of novel genome editing technologies, such as CRISPR, which allows for targeted mutations and speeding up fish breeding. For successful introduction, applications of the technology need to be socially acceptable and contribute to sustainability.

In this dissertation, I study the technological potential and challenges, the sustainability issues, and conditions for social acceptance of introducing CRISPR in salmon farming, in three papers, respectively. In paper I, a systematic literature review was conducted to identify and categorize publications that have used genome editing in aquaculture finfish species. The search was designed according to relevant PRISMA elements. Results shows that a wide variety of aquaculture species have been used, salmonids being the second most studied group, with a broad specter of potential for future application in aquaculture such as sterility, disease resistance and increased growth.

Paper II and III are both based on a qualitative study of semi-structured stakeholder interviews and citizen focus group interviews. The interviews were conducted in video calls and included three main topics: the salmon as an animal, genome editing, and sustainability. For paper II, considerations and conditions related to aquaculture, sustainability and genome editing were identified and merged with data from an analysis of international and national policy and strategy documents, to inform a biosphere-based sustainability assessment framework.

For paper III, general considerations, and conditions for social acceptance of genome-edited salmon were identified. Main finding where that across all interviews, considerations to the wild salmon viability and the farmed salmon welfare, are widely shared and seems to be of main concern to the study participants. Further, several conditions to the industry and products were raised, such as unintended consequences being unacceptable, and the editing must contribute to improve welfare above increasing profit.

The papers show that there are potential applications of genome editing under research which might be considered socially acceptance and sustainable for salmon farming. However, this seem to depend on social acceptance to the salmon farming industry in general, and on genome editing being applied in concert with other measurements that improve salmon health and welfare, and that reduces environmental effects.

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