ge²p² global foundation
governance, ethics, evidence, policy, practice

Center for Genomic Medicine Ethics & Policy

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

March 2023 Number 03

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:

Editor
David R Curry, MS
President/CEO
GE2P2 Global Foundation
david.r.curry@ge2p2global.org
USA

Associate Editor
Daima Bukini, PhD
Associate Fellow
GE2P2 Global Foundation
daima.bukini@ge2p2global.org
Tanzania

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:

Subject Area	Page
EDITOR'S "SHORT LIST"	2
GENOMIC DATA, BIOBANKING, GENOMIC RESEARCH	7
REGULATION, GUIDANCE	12
GOVERNANCE	14
PUBLIC/COMMUNITY ENGAGEMENT	18
ACCESS, EQUITY, BENEFITS SHARING	20
GENETIC SCREENING/GENETIC COUNSELLING	21
PLANTS, CROPS, AGRICULTURE, ANIMALS	26

EDITOR'S SHORT LIST

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

- Of course, we lead our short list with what is arguably the defining meeting of this year [and the next few]: the **Third International Summit on Human Genome Editing** to be convened 6-8 March 2023 at the Francis Crick Institute in London. The agenda for the three-day meeting is now available at the link provided.

- Jonathon Moreno [University of Pennsylvania] provides a very useful historical perspective on the sources and evolution of norms and ethics around biotechnology as a product of the post WWII liberal consensus in his book chapter **Genopolitics: Biotechnology Norms and the Liberal International Order.**

- Governance of biotechnology and ELSI [ethical, legal, and social implications] implications – especially with regard to local institutions and politics – is the focus of **Governing biotechnology to provide safety and security and address ethical, legal, and social implications** by Benjamin Trump [United States Army Corps of Engineers] and collaborators.

- An open letter published in *Wellcome Open Research* by Gerald Michael Ssebunnya [Africa Institute for Human Dignity; Padre Pio Medical Centre, Gaborone, Botswana] explores public engagement – **Towards an appropriate African framework for public engagement with human genome editing: a call to synergistic action.**

- Public engagement is further explored by Jeremy Sugerman [Johns Hopkins University] and collaborators in **Critical considerations for public engagement in stem cell-related research** which addresses prior experiences with engaging various publics.

- Madhumita Dhar Sarkar and Belayet Hussain Mazumder [School of Legal Studies, Assam University, India] provide perspectives on gene editing economics, law, and scientific progress from the India context in **Human Gene Editing and Its Inherent Conundrums: Legal Perspectives.**

- A book chapter by Denise McCurdy (Georgia State University, USA) emphasizes collaboration amongst diverse players in **Ecosystems in Precision Medicine: The Need for Good Governance?**

- A useful Perspective in *Nature Genetics* by Eleanor Wong [Genome Institute of Singapore, Agency for Science, Technology and Research] and collaborators focuses on **The Singapore National Precision Medicine Strategy** – a 10-year, whole-of-government initiative aiming to generate precision medicine data of up to one million individuals, integrating genomic, lifestyle, health, social and environmental data.

- A Nuffield Council on Bioethics blog post by Frances Flinter [Council Member and Emeritus Professor of Clinical Genetics at Guy's & St Thomas NHS Foundation Trust] analyzes **Whole Genome Sequencing in newborns: benefits and risks**.

- Finally, a Comment in *Nature* by Zané Lombard & Guida Landouré [both writing on behalf of the H3Africa Steering Committee] – **Could Africa be the future for genomics research? –** lays out what is needed to ensure that investment in genomics in Africa is not just sustained in a post- H3Africa world but expanded.

The citations/abstracts for our "short list" are below:

Third International Summit on Human Genome Editing

6-8 March 2023 Francis Crick Institute, London UK.

In person tickets are sold out; registration to attend the event virtually is now open

The three-day Summit is being 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. Building on previous events held in Washington, DC (2015) and Hong Kong (2018), the London meeting will continue the global dialogue on somatic and germline human genome editing. Major themes for discussion include 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 agenda for this summit has now been published and is available here.***

Genopolitics: Biotechnology Norms and the Liberal International Order

Book Chapter

Jonathan Moreno

Medical Research Ethics: Challenges in the 21st Century pp 35–45, 2 January 2023

Abstract

What happens in the world's most advanced life sciences laboratories, why those activities are important, and whether and how they can be brought under a uniform governance framework might be considered exquisitely esoteric matters in the context of the great geopolitical questions of our time. Nonetheless, the emerging issues in biotechnology—the use of living organisms to create new products and especially in the control of the human genome—represent a useful stress test for the future of the norms inherent in the liberal international order (LIO). My case study will be the nearly universal public outrage following the announcement by a Chinese scientist that he had engaged in the first gene editing of several embryos that survived to birth, an episode that has created an opportunity to assess the global consensus about the ethics of biotechnology with regard to human DNA. Although not as explicit or well understood or enforced as weapons treaties, trade arrangements, or monetary institutions, the norms around biotechnology are very much a product of the post-World War II liberal consensus.

Governing biotechnology to provide safety and security and address ethical, legal, and social implications

Original Research Article

Benjamin Trump, Christopher Cummings, Kasia Klasa, Stephanie Galaitsi and Igor Linkov

Frontiers in Genetics, 11 January 2023

Open access

Abstract

The field of biotechnology has produced a wide variety of materials and products which are rapidly entering the commercial marketplace. While many developments promise revolutionary benefits, some of them pose uncertain or largely untested risks and may spur debate, consternation, and outrage from individuals and groups who may be affected by their development and use. In this paper we show that the success of any advanced genetic development and usage requires that the creators establish technical soundness, ensure safety and security, and transparently represent the product's ethical, legal, and social implications (ELSI). We further identify how failures to address ELSI can manifest as significant roadblocks to product acceptance and adoption and advocate for use of the "safety-by-design" governance philosophy. This approach requires addressing risk *and* ELSI needs early and often in the technology development process to support innovation while providing security and safety for workers, the public, and the broader environment. This paper identifies and evaluates major ELSI challenges and perspectives to suggest a methodology for implementing safety-by-design in a manner consistent with local institutions and politics. We anticipate the need for safety-by-design approach to grow and permeate biotechnology governance structures as the field expands in

scientific and technological complexity, increases in public attention and prominence, and further impacts human health and the environment.

Towards an appropriate African framework for public engagement with human genome editing: a call to synergistic action

Open Letter

Gerald Michael Ssebunnya

Wellcome Open Research 2022, 7:302, 12 December 2022

Abstract

The CRISPR-Cas9 system has revolutionised the biotechnology of human genome editing. Human germline gene editing promises exponential benefits to many in Africa and elsewhere, especially those affected by the highly prevalent monogenic disorders - for which, thanks to CRISPR, a relatively safe heritable radical therapy is now possible. Africa evidently presents a unique opportunity for empirical research in human germline gene editing because of its high prevalence of monogenic disorders. Critically, however, germline gene editing has raised serious ethical concerns especially because of the significant risks of inadvertent and intentional misuse of its transgenerational heritability. Calls for due prudence have become even more pronounced in the wake of the 2018 case of He Jiankui's 'CRISPR'd babies'. Meanwhile, Africa is seriously lagging in articulating its position on human genome editing. Conspicuously, there has been little to no attempt at comprehensively engaging the African public in discussions on the promises and concerns about human genome editing. Thus, the echoing key question remains as to how Africa should prudently embrace and govern this revolutionary biotechnology. In this article, therefore, I lay the groundwork for the possible development of an appropriate African framework for public engagement with human genome editing and call upon all stakeholders to urgent synergistic action. I particularly highlight the World Health Organisation's possible leadership role in promptly establishing the requisite expert working group for this urgent need.

Critical considerations for public engagement in stem cell-related research

Meeting Report

Jeremy Sugarman, Amander Clark, James Fishkin, Kazuto Kato, Kevin McCormack, Megan Munsie, Michael J. Peluso, Nancy Rene´, and Susan L. Solomon

Stem Cell Reports, 2 February 2023

Open access

Summary

Public engagement is increasingly recognized as being integral to basic and translational research. Public engagement involves effective communication about research along with the mutual exchange of views and opinions among a wide variety of members in society. As such, public engagement can help to identify issues that must be addressed in order for research to be ethically sound and trustworthy. It is especially critical in research that potentially raises ethical concerns, for example research involving embryos, germline genome editing, stigmatized conditions, and marginalized communities. Therefore, it is not surprising that there have been prominent recent calls for public engagement in the emerging sciences. However, given that there is arguably little agreement about how this should be done and the best ways of doing so, those involved with planning and implementing public engagement can benefit from understanding a broad range of prior experiences on related issues.

Human Gene Editing and Its Inherent Conundrums: Legal Perspectives

Journal Article

Madhumita Dhar Sarkar, Belayet Hussain Mazumder

Indian Journal of Law and Justice, Vol. 13 No. 1 Issue March 2022

Open access

Abstract

Gene Editing, as a work of human ingenuity and innovation, opens up a vast range of possibilities for human existence in the future. While Gene Editing, on the surface, opens up the possibility of human perfection, it also raises a slew of ethical, philosophical, economic, and legal difficulties. From the standpoint of India's commitment to ensuring an egalitarian society in which access to the fruits of science and technology is made available to both the rich and the poor, the prospect of Gene Editing raises deep and complex questions about the disparity in the capacity of the less resourceful to reap the benefits of this scientific advancement. The propriety of pushing such a disruptive technology - of men having the potential to fundamentally and dramatically alter nature's systems of creation and sustenance - is also a factor in Gene Editing. Gene Editing also brings up the classic "Frankenstein" question: are we unleashing a beast beyond our control? Is it possible to get a global consensus on Gene Editing's inherent limitations, if there are any? Because Gene Editing involves decrypting the fundamental building components of any human person, it raises the important question of whether such information should be made public, as well as the risks that come with it. Within its limited scope, this study makes a determined effort to address the aforementioned conundrums. It also attempts to provide a glimpse into the future that we are moving towards in terms of Human Gene Editing. While the scope of the various issues relating to Gene Editing is vast, the paper focuses primarily on the dimensions of Gene Editing's economic perspective in India, its ethics, law, and scientific progress, informed consent and counselling in the domain of Gene Editing, and the need for transparency and accountability in the domain of Gene Editing.

Ecosystems in Precision Medicine: The Need for Good Governance?

Book Chapter

Denise McCurdy (Georgia State University, USA)

Digital Identity in the New Era of Personalized Medicine, pages 28-45 - 2023

Abstract

Emerging technology requires participating members to intensely collaborate in fundamentally novel ways. This participation includes established and start-up firms, health professionals, standards bodies, regulatory agencies, and of course, patients. But how to design, implement, and manage emerging technology that cuts across shifting zones? With firm and global boundaries increasingly blurred, uneven regulatory treatment, and evolving standards, how can ecosystem partners collaborate to mitigate the risks to consumers as their data becomes ever more precise and identifiable? This chapter explores the relationships and decisions that ecosystem partners must collaboratively take together in the context of precision medicine and the challenges of working effectively – and ethically - with consumers.

The Singapore National Precision Medicine Strategy

Perspective

Eleanor Wong, Nicolas Bertin, Maxime Hebrard, Roberto Tirado-Magallanes, Claire Bellis, Weng Khong Lim, Chee Yong Chua, Philomena Mei Lin Tong, Raymond Chua, Kenneth Mak, Tit Meng Lim, Wei Yang Cheong, Kwee Eng Thien, Khean Teik Goh, Jin-Fang Chai, Jimmy Lee, Joseph Jao-Yiu Sung, Tien Yin Wong, Calvin Woon Loong Chin, Peter D. Gluckman, Liuh Ling Goh, Kenneth Hon Kim Ban, Tin Wee Tan, SG10K_Health Consortium, …Patrick Tan

Nature Genetics 55, pages 178–186, 19 January 2023

Abstract

Precision medicine promises to transform healthcare for groups and individuals through early disease detection, refining diagnoses and tailoring treatments. Analysis of large-scale genomic–phenotypic databases is a critical enabler of precision medicine. Although Asia is home to 60% of the world's population, many Asian ancestries are under-represented in existing databases, leading to missed opportunities for new

discoveries, particularly for diseases most relevant for these populations. The Singapore National Precision Medicine initiative is a whole-of-government 10-year initiative aiming to generate precision medicine data of up to one million individuals, integrating genomic, lifestyle, health, social and environmental data. Beyond technologies, routine adoption of precision medicine in clinical practice requires social, ethical, legal and regulatory barriers to be addressed. Identifying driver use cases in which precision medicine results in standardized changes to clinical workflows or improvements in population health, coupled with health economic analysis to demonstrate value-based healthcare, is a vital prerequisite for responsible health system adoption.

Whole Genome Sequencing in newborns: benefits and risks

Blog Post

Frances Flinter - Council Member and Emeritus Professor of Clinical Genetics at Guy's & St Thomas NHS Foundation Trust

Nuffield Council on Bioethics, 16 February 2023

Closing summary

…Using whole genome sequencing to screen newborn babies is a step into the unknown. Getting the balance of benefit and harm right will be crucial. The potential benefits are early diagnosis and treatment for more babies with genetic conditions. The potential harms are false or uncertain results, unnecessary anxiety for parents, and a lack of good follow-up care for babies with a positive screening result.

Could Africa be the future for genomics research?

Comment

Zané Lombard & Guida Landouré

Nature, 31 January 2023

Open access

Abstract

In 2020, an analysis of 426 African genomes, involving researchers from 15 African countries, uncovered 3 million new variants in the human genome1. The discovery contributed to the development of a tool that enables researchers to identify genetic associations specifically in African populations — the Infinium H3Africa Consortium genotyping array, produced by the US biotechnology firm Illumina. Although various enterprises have supported cutting-edge human genomics in Africa, the Human Heredity and Health in Africa (H3Africa) initiative2, which supported this work, has probably contributed the most in terms of infrastructure and training. The US$176-million programme began in 2010, funded by the US National Institutes of Health (NIH) and the UK biomedical charity Wellcome (in partnership with the African Society of Human Genetics). Projects have ranged from population-based genomic studies of common disorders, such as heart disease, to investigations of infectious diseases, such as COVID-19. Together, some 51 projects, all led by African scientists and involving researchers from more than 30 African countries, have resulted in 50,000 samples being genotyped and nearly 700 papers being published. Thanks to H3Africa and other genomics initiatives, such as the Nigerian 100K Genome Project3, African genomics is now poised to improve the health of millions of people worldwide, including those across the continent and the African diaspora. But building on the discoveries made so far — and especially applying findings to the clinic — will require several systemic changes, including a major shift in how genomics research in Africa is funded. All remaining projects supported by the H3Africa initiative are expected to wrap up this year. (Although funding formally ended in June 2022, some H3Africa grant recipients were able to obtain extensions because of disruption from the COVID-19 pandemic.) Here, we lay out what is needed to ensure that investment in genomics in Africa is not just sustained in a post-H3Africa world but expanded. In our view, Africa could become the birthplace for a new kind of genomics — one that brings better health to all.

::::::::
::::::::

GENOMIC DATA/BIOBANKING/GENOMIC RESEARCH

Genomics data sharing

Chapter

Judit Kumuthini, Lyndon Zass, Melek Chaouch, Faisal M. Fadlelmola, Nicola Mulder, Fouzia Radouani, Verena Ras, Chaimae Samtal, Milaine S. S. Tchamga, Dassen Sathan, Anisah Ghoorah, Raphael Z. Sangeda, Liberata A. Mwita, Upendo Masamu, Samar Kamal Kassim, Zoe Gill, Zahra Mungloo-Dilmohamud, Gordon Wells

Genomic Data Sharing Case Studies, Challenges, and Opportunities for Precision Medicine, Pages 111-135, 27 January 2023

Abstract

Recent technological advances have allowed the unprecedented generation of large data sets in the biological sciences. Gaining the most value from this generation requires the data to be distributed and shared more widely so that multiple groups may make use of it. This brings about a number of technical and social challenges, and different approaches have been developed to resolve them. In this chapter, we introduce the concept and principles of data sharing, we discuss two data sharing methods, sharing through an archive and sharing through a data commons, we then provide a case example from the Human Heredity and Health in Africa (H3Africa) consortium, sharing data in resource-limited regions. We also discuss the overall challenges associated with data sharing, as well as Beacons and the associated security and privacy concerns.

Towards trust-based governance of health data research

Journal Article

Marieke A. R. Bak, M. Corrette Ploem, Hanno L. Tan, M. T. Blom & Dick L. Willems

Medicine, Health Care and Philosophy, 12 January 2023

Open access

Abstract

Developments in medical big data analytics may bring societal benefits but are also challenging privacy and other ethical values. At the same time, an overly restrictive data protection regime can form a serious threat to valuable observational studies. Discussions about whether data privacy or data solidarity should be the foundational value of research policies, have remained unresolved. We add to this debate with an empirically informed ethical analysis. First, experiences with the implementation of the General Data Protection Regulation (GDPR) within a European research consortium demonstrate a gap between the aims of the regulation and its effects in practice. Namely, strictly formalised data protection requirements may cause routinisation among researchers instead of substantive ethical reflection, and may crowd out trust between actors in the health data research ecosystem; while harmonisation across Europe and data sharing between countries is hampered by different interpretations of the law, which partly stem from different views about ethical values. Then, building on these observations, we use theory to argue that the concept of trust provides an escape from the privacy-solidarity debate. Lastly, the paper details three aspects of trust that can help to create a responsible research environment and to mitigate the encountered challenges: trust as multi-agent concept; trust as a rational and democratic value; and trust as method for priority setting. Mutual cooperation in research—among researchers and with data subjects—is grounded in trust, which should be more explicitly recognised in the governance of health data research.

Protection of genomic data and the Australian Privacy Act: when are genomic data 'personal information'?

Article

Minna Paltiel, Mark Taylor and Ainsley Newson

International Data Privacy Law, 2023, Vol. 00, No. 0, 2023

Abstract

'Personal information', protected under the Australian Privacy Act 1988 (Cth), is 'about an identified individual or an individual who is reasonably identifiable' (S.6), so the legal assessment of 'identifiability' shapes the protection of genomic data under the Privacy Act. Not all genomic data are captured by the statutory definitions of 'genetic information' in the Privacy Act; however, genomic data that do not fit the definition may still be protected if they are about an identifiable individual. In applying the legal test of identifiability to genomic data, the interaction between the data and the data environment must be examined. Overemphasis on particular features of genomic data, such as 'rareness' or 'uniqueness', may lead to a misapplication of the Privacy Act. Whether genomic data are personal information is primarily a matter of the opportunities and likelihood of linking the genomic data in question with other data available in the data environment.

The indigenous African cultural value of human tissues and implications for bio-banking

Original Article

David Nderitu, Claudia Emerson

Developing World Bioethics, 23 January 2023

Abstract

Bio-banking in research elicits numerous ethical issues related to informed consent, privacy and identifiability of samples, return of results, incidental findings, international data exchange, ownership of samples, and benefit sharing etc. In low and middle income (LMICs) countries, the challenge of inadequate guidelines and regulations on the proper conduct of research compounds the ethical issues. In addition, failure to pay attention to underlying indigenous worldviews that ought to inform issues, practices and policies in Africa may exacerbate the situation. In this paper we discuss how the African context presents unique and outstanding cultural thought systems regarding the human body and biological materials that can be put into perspective in bio-bank research. We give the example of African ontology of nature presented by John Samwel Mbiti as foundational in adding value to the discourse about enhancing relevance of bio-bank research in the African context. We underline that cultural rites of passage performed on the human body in majority of communities in Africa elicit quintessential perspective on beliefs about handling of human body and human biological tissues. We conclude that acknowledgement and inclusion of African indigenous worldviews regarding the human body is essential in influencing best practices in biobank research in Africa.

Balancing the safeguarding of privacy and data sharing: perceptions of genomic professionals on patient genomic data ownership in Australia

Journal Article

Yuwan Malakar, Justine Lacey, Natalie A. Twine, Rod McCrea & Denis C. Bauer

European Journal of Human Genetics, 11 January 2023

Open access

Abstract

There are inherent complexities and tensions in achieving a responsible balance between safeguarding patients' privacy and sharing genomic data for advancing health and medical science. A growing body of literature suggests establishing patient genomic data ownership, enabled by blockchain technology, as one approach for managing these priorities. We conducted an online survey, applying a mixed methods approach to collect quantitative (using scale questions) and qualitative data (using open-ended questions). We explored the views of 117 genomic professionals (clinical geneticists, genetic counsellors, bioinformaticians, and researchers) towards patient data ownership in Australia. Data analysis revealed most professionals agreed that patients have rights to data ownership. However, there is a need for a clearer understanding of the nature and implications of data ownership in this context as genomic data often is subject to collective ownership (e.g., with family members and laboratories). This research finds that while the majority of

genomic professionals acknowledge the desire for patient data ownership, bioinformaticians and researchers expressed more favourable views than clinical geneticists and genetic counsellors, suggesting that their views on this issue may be shaped by how closely they interact with patients as part of their professional duties. This research also confirms that stronger health system infrastructure is a prerequisite for enabling patient data ownership, which needs to be underpinned by appropriate digital infrastructure (e.g., central vs. decentralised data storage), patient identity ownership (e.g., limited vs. self-sovereign identity), and policy at both federal and state levels.

Pilot Programs and Postcolonial Pivots: Pioneering "DNA Fingerprinting" on Britain's Borders
Research Article
Roberta Bivins
Comparative Studies in Society and History 1-26, 19 January 2023
Open access
Abstract
Developed in Britain and the United States in the 1980s, genetic profiling has since become a global technology. Today, it is widely regarded as the evidentiary "gold standard" in individual and forensic identification. However, its origins as a technology of post-empire at Britain's externalized borders in South Asia have remained unexamined. This article will argue that the first state-sanctioned use of "DNA fingerprints," a pilot program exploring its value in disputed cases of family reunification migration from Bangladesh and Pakistan to Britain's postcolonial cities, repays closer examination. National and transnational responses to the advent of genetic profiling as an identification technology demonstrate the interplay between imperial and postcolonial models and networks of power and truth production. At the same time, this experiment prefigured and conditioned the wider reception of DNA profiling in matters of kinship. Far from being a footnote, the use of genetic profiling by migrants determined to exercise their legal rights in the face of a hostile state also worked to naturalize genetic ties as the markers of "true" familial relationships.

Exploring the challenges of and solutions to sharing personal genomic data for use in healthcare
Original Research Article
Lasse Parvinen, Ari Alamaki, Heli Hallikainen, Marko Maki
Health Informatics Journal 1-12, 18 January 2023
Open Access
Abstract
Boosted by the COVID-19 pandemic, as well as the tightened General Data Protection Regulation (GDPR) legislation within the European Union (EU), individuals have become increasingly concerned about privacy. This is also reflected in how willing individuals are to consent to sharing personal data, including their health data. To understand this behavior better, this study focuses on willingness to consent in relation to genomic data. The study explores how the provision of educational information relates to willingness to consent, as well as differences in privacy concerns, information sensitivity and the perceived trade-off value between individuals willing versus unwilling to consent to sharing their genomic data. Of the respondents, 65% were initially willing to consent, but after educational information 89% were willing to consent and only 11% remained unwilling to consent. Educating individuals about potential health benefits can thus help to correct the beliefs that originally led to the unwillingness to share genomic data.

Adoptees and their unknown genetic inheritance
Chapter
Gaye Orr
Law, Regulation and Governance in the Information Society, 1st Edition, 2022

Abstract

The principal question in this chapter is whether adopted children (and those children as adults) should themselves have a right to know (or not to know) more about their genetic profile. Such information might be critical to reproductive and relationship decisions made by adoptees in their adult lives. However, there is a great deal that is unclear. In particular, how would an adoptee's right to genetic information sit with the birth parents' interest in confidentiality? How much assistance does the case for recognising such a right derive from ABC v St George's Healthcare NHS Foundation Trust and Montgomery v Lanarkshire Health Board? If such a right is to be recognised, then who would bear the informational responsibility and who, potentially, would be at risk of informational wrongdoing? Is it arguable that there is a common law or statutory duty of care to disclose relevant information about current and future health risks? Recognising that there is a great deal of legal uncertainty as well as many competing and conflicting interests in this regulatory space, this chapter underlines the need for further debate and reflection.

Guidelines for genetic ancestry inference created through roundtable discussions

Journal Article

Jennifer K. Wagner, Joon-Ho Yu, Duana Fullwiley, CeCe Moore, James F. Wilson, Michael J. Bamshad, Charmaine D. Royal, and on behalf of the Genetic Ancestry Inference Roundtable Participants

Human Genetics and Genomics Advances, Volume 4, Issue 2, 13 January 2023

Abstract

The use of genetic and genomic technology to infer ancestry is commonplace in a variety of contexts, particularly in biomedical research and for direct-to-consumer genetic testing. In 2013 and 2015, two roundtables engaged a diverse group of stakeholders toward the development of guidelines for inferring genetic ancestry in academia and industry. This report shares the stakeholder groups' work and provides an analysis of, commentary on, and views from the groundbreaking and sustained dialogue. We describe the engagement processes and the stakeholder groups' resulting statements and proposed guidelines. The guidelines focus on five key areas: application of genetic ancestry inference, assumptions and confidence/laboratory and statistical methods, terminology and population identifiers, impact on individuals and groups, and communication or translation of genetic ancestry inferences. We delineate the terms and limitations of the guidelines and discuss their critical role in advancing the development and implementation of best practices for inferring genetic ancestry and reporting the results. These efforts should inform both governmental regulation and self-regulation.

Attitude Disparity and Worrying Scenarios in Genetic Discrimination—Based on Questionnaires from China

Article

Zhong Wang, Yujun Guo and Rui Xu

Healthcare *2023,* 11*(2), 188,* 8 January 2023

Open access

Abstract

Objectives: As genetic testing is increasingly used in non-medical fields, the judgment of people's potential conditions based on predictive genetic information inevitably causes genetic discrimination (henceforth GD). This article aimed to systematically investigate the disparity in attitudes and worrying scenarios concerning GD in China.

Methods: A questionnaire survey of 555 respondents was conducted. Statistical tests were used to examine disparity in attitudes between gender, age, and education. A descriptive analysis was also conducted to explore other worrying scenarios.

Results: It shows that (1) men are more tolerant of GD compared to women, and (2) participants aged between 18 and 30 years old possess the highest objection to GD. However, (3) no indication can attest to the relationship between educational level and perspective on GD. In addition, (4) the acceptance of gene testing in the three most common scenarios is ranked in descending order as follows: partner choice,

insurance services, and recruitment. Moreover, (5) worrying scenarios relating to GD include: education, social occasions, medical services, fertility, shopping, and so on.
Conclusions: Based on the results, suggestions proposed include developing a blacklist mechanism in the field of genetic data application and strengthening the security regulations for the commercial use of genetic data.

::::::::
::::::::

ETHICAL REGULATION, ETHICAL GUIDANCE

Ethical and Safety Considerations in Stem Cell-Based Therapy for Parkinson's Disease-
Peer-Reviewed Chapter
Fangzhou Li
Parkinson's Disease - Animal Models, Current Therapies and Clinical Trials [Working Title], 30 January 2023
Open access
Abstract
Stem cell-based therapy for Parkinson's Disease (PD) is entering an exciting era with many groups competing to reach the goal of safe and practical clinical application. However, the road to this goal is long and beset by challenging obstacles, among which are Good Manufacturing Practice (GMP) standards, scalability, and regulatory requirements for the final cell product. Of paramount importance is the patient safety of the stem cell-derived dopaminergic neurons, such that each stage of the cell therapy implementation process must be scrutinized for potential safety concerns before introduction to the clinic can be contemplated. In this chapter, we will critically consider the safety regulations and safety strategies of stem cell-based therapy for PD, emphasizing the principal requirements necessary for this new therapeutic approach to benefit PD patients. We will introduce the current safety challenges and the connections between these safety issues and the special characteristics of neural stem cells. In addition, we will summarize the safety standards for stem cell-based therapy currently adopted by leading cell therapy groups and international regulations. Both in vitro and in vivo safety assessment methods will be discussed as they relate to the implementation of these standards. Finally, we will speculate on strategies for further enhancing the safety of stem cell-based therapy for PD.

Revisiting informed consent in forensic genomics in light of current technologies and the times
Original Article
Bruce Budowle & Antti Sajantila
International Journal of Legal Medicine 137, pages 551–565, 16 January 2023
Open access
Abstract
Informed consent is based on basic ethical principles that should be considered when conducting biomedical and behavioral research involving human subjects. These principles—respect, beneficence, and justice—form the foundations of informed consent which in itself is grounded on three fundamental elements: information, comprehension, and voluntary participation. While informed consent has focused on human subjects and research, the practice has been adopted willingly in the forensic science arena primarily to acquire reference samples from family members to assist in identifying missing persons. With advances in molecular biology technologies, data mining, and access to metadata, it is important to assess whether the past informed consent process and in particular associated risks are concomitant with these increased capabilities. Given the state-of-the-art, areas in which informed consent may need to be modified and augmented are as follows: reference samples from family members in missing persons or unidentified human remains cases; targeted analysis of an individual(s) during forensic genetic genealogy cases to reduce an investigative burden; donors who provide their samples for validation studies (to include population studies and entry into databases that would be applied to forensic statistical calculations) to support implementation

of procedures and operations of the forensic laboratory; family members that may contribute samples or obtain genetic information from a molecular autopsy; and use of medical and other acquired samples that could be informative for identification purposes. The informed consent process should cover (1) purpose for collection of samples; (2) process to analyze the samples (to include type of data); (3) benefits (to donor, target, family, community, etc. as applicable); (4) risks (to donor, target, family, community, etc. as applicable); (5) access to data/reports by the donor; (6) sample disposition; (7) removal of data process (i.e., expungement); (8) process to ask questions/assessment of comprehension; (9) follow-up processes; and (10) voluntary, signed, and dated consent. Issues surrounding these topics are discussed with an emphasis on addressing risk factors. Addressing informed consent will allow human subjects to make decisions voluntarily and with autonomy as well as secure the use of samples for intended use.

Comparing regulatory processes in genome editing and autonomous vehicles: How institutional environments shape sociotechnical imaginaries

Original Article

Meghna Mukherjee, Konrad Posch, Santiago J. Molina, Ken Taymor, Ann Keller

Review of Policy Research, 16 January 2023

Abstract

This study compares the regulation of two emerging technologies, the CRISPR genome-editing system and Connected and Autonomous Vehicles (CAV) in the United States. The study draws on 33 in-depth interviews with innovation and governance experts to study the relationship between their regulatory environments and developing beliefs about these technologies. Using sociotechnical imaginaries as a framework, we explore how social actors envision technologically driven futures and the social order that enables them. These imaginaries are essential to emerging technologies, where experts build a framework of potentialities for innovation still underway. While scholarship has documented how sociotechnical imaginaries arise among policymakers, groups of scientists, state and local stakeholders, and public actors in different countries, less has been said about how regulatory organizations and their actors shape expectations around technologies that are in the early and middle stages of development. This article finds that regulatory institutions shape emerging imaginaries along three related axes: the distribution of authority, technological novelty, and risk. Interviewees negotiate these three contingencies differently based on relevant extant regulatory structures and ideologies, resulting in distinct imaginaries around each technology. CRISPR actors envision genome editing as largely diminishing biomedical harm and eventually suitable for health markets, while CAV actors diverge on whether self-driving cars alleviate or exacerbate risk and how they may enter roads. That organizational structures and practices of regulation inform broadly held sociotechnical imaginaries bears significance for studies of innovation trajectories, suggesting regulators can take an active role in shaping how risks and benefits of emerging technology are defined.

::::::::
::::::::

GOVERNANCE FRAMEWORKS

Human genome editing after the "CRISPR babies": The double-pacing problem and collaborative governance

Research Article

Leifan Wang, Lijun Shang, Weiwen Zhang

Journal of Biosafety and Biosecurity, Volume 5, Issue 1, pp 8-13, 2 January 2023

Open access

Abstract

How to ensure the safe, effective, and ethical use of emerging biotechnologies, such as clustered regularly interspaced short palindromic repeats (CRISPR)-based genome editing, is a global challenge. The occurrence

of the “CRISPR babies” in 2018 publicly brought this issue into sharp focus, and led to comprehensive regulatory reforms in China and various countries around the world. The current article analyzes this event-driven regulatory reform in China by elaborating the most salient provisions designed to prevent risk and protect individual rights, public health, and social morality relating to human genome editing in four important sectors of law: biosecurity law, civil code, criminal law and patent law. It highlights that, although regulation is being undertaken, the gaps between the law and advancing technology remain discernible, at both a national and transnational level (i.e., the “double-pacing problem”). Further attention and collaboration will be required to address the ongoing challenges associated with the use of human genome editing.

Modeling policy development: examining national governance of stem cell-based embryo models

Perspective

Morris Fabbri, Margaret Ginoza, Lars Assen, Karin Jongsma & Rosario Isasi

Regenerative Medicine Vol. 18, No. 2, 5 January 2023

Abstract

Researchers can now coax human pluripotent stem cells to imitate the structure and spontaneous self-organization of the developing human embryo. Although these stem cell-based embryo models present an advantageous alternative to embryo research, they also raise ethical and policy challenges. In 2021, the International Society for Stem Cell Research revised its Guidelines for Stem Cell Research and Clinical Translation, providing contemporaneous best practices for ethical conduct in the field. The Guidelines complement national governance frameworks; however, they also contain contentious and aspirational norms that might catalyze change in research practice and in the enactment of national policies. Using a sample of 11 research-intensive countries, the authors compare research policy frameworks against the International Society for Stem Cell Research Guidelines to showcase how developments in global and national policies might affect stem cell-based embryo model research governance and illustrate fertile areas for ethical reflection and policy development.

Challenges of Global Technology Assessment in Biotechnology—Bringing Clarity and Better Understanding in Fragmented Global Governance

Chapter

Sophie van Baalen, Krishna Ravi Srinivas & Guangxi He

Technology Assessment in a Globalized World pp 149–173, 7 January 2023

Open access

Abstract

Biotechnology involves the use and manipulation of living organisms such as plants, animals, humans, and biological systems, or parts of this, to modify their characteristics in order to create desired organisms or products. Biotechnology as a field touches on many aspects that are central to technology assessment (TA), and have been the focus of TA activities since the 1980s. By presenting three key topics in biotechnology—genetically modified food and crops, synthetic biology, and human genome germline editing—we show that a central feature of biotechnology is that the science is evolving globally and the products that it brings forth are traded across the globe. Yet, there are major differences in the regulation and governance of the academic and industrial sectors between countries. These stem from different needs and interests per country, as well as differences in traditions, cultural differences and public perceptions. As global governance is fragmented, with little scope for harmonization, global TA of biotechnology can bring clarity and better understanding, and enable better governance. In order to do so, an integrated global TA framework should consider international trade and differences in risk assessment; cultural variation and different value-systems between countries; and differences in countries’ capacities in R&D and coordination of public engagement efforts.

::::::::
::::::::

PUBLIC AND COMMUNITY ENGAGEMENT

Responsible use of organoids in precision medicine: the need for active participant involvement

Chapter

Michael A. Lensink, Karin R. Jongsma, Sarah N. Boers, Jacquelien J. Noordhoek, Jeffrey M. Beekman and Annelien L. Bredenoord

Effective and sustainable patient participation: The patients' perspective in CF care and CF research, Chapter 9

Open access

Abstract

Organoids are three-dimensional multicellular structures grown in vitro from stem cells and which recapitulate some organ function. They are derivatives of living tissue that can be stored in biobanks for a multitude of research purposes. Biobank research on organoids derived from patients is highly promising for precision medicine, which aims to target treatment to individual patients. The dominant approach for protecting the interests of biobank participants emphasizes broad consent in combination with privacy protection and ex ante FF(predictive) ethics review. In this paradigm, participants are positioned as passive donors; however, organoid biobanking for precision medicine purposes raises challenges that we believe cannot be adequately addressed without more ongoing involvement of patient-participants. In this Spotlight, we argue why a shift from passive donation towards more active involvement is particularly crucial for biobank research on organoids aimed at precision medicine, and suggest some approaches appropriate to this context.

Community partnerships are fundamental to ethical ancient DNA research

Commentary

Emma Kowal, Laura S. Weyrich, Juan Manuel Argüelles, Alyssa C. Bader, Chip Colwell, Amanda Daniela Cortez, Jenny L. Davis, Gonzalo Figueiro, Keolu Fox, Ripan S. Malhi, Elizabeth Matisoo-Smith, Ayushi Nayak, Elizabeth A. Nelson, George Nicholas, Maria A. Nieves-Colón, Lynette Russell, Sean Ulm, Francisco Vergara-Silva, Fernando A. Villanea, Jennifer K. Wagner…Krystal S. Tsosie

Human Genetics and Genomics Advances, 11 January 2023

Open access

Summary

The ethics of the scientific study of Ancestors has long been debated by archaeologists, bioanthropologists, and, more recently, ancient DNA (aDNA) researchers. This article responds to the article "Ethics of DNA research on human remains: five globally applicable guidelines" published in 2021 in *Nature* by a large group of aDNA researchers and collaborators. We argue that these guidelines do not sufficiently consider the interests of community stakeholders, including descendant communities and communities with potential, but yet unestablished, ties to Ancestors. We focus on three main areas of concern with the guidelines. First is the false separation of "scientific" and "community" concerns and the consistent privileging of researcher perspectives over those of community members. Second, the commitment of the guidelines' authors to open data ignores the principles and practice of Indigenous Data Sovereignty. Further, the authors argue that involving community members in decisions about publication and data sharing is unethical. We argue that excluding community perspectives on "ethical" grounds is convenient for researchers, but it is not, in fact, ethical. Third, we stress the risks of *not* consulting communities that have established or potential ties to Ancestors, using two recent examples from the literature. Ancient DNA researchers cannot focus on the

lowest common denominator of research practice, the bare minimum that is legally necessary. Instead, they should be leading multidisciplinary efforts to create processes to ensure communities from all regions of the globe are identified and engaged in research that affects them. This will often present challenges, but we see these challenges as *part of* the research, rather than a distraction from the scientific endeavor. If a research team does not have the capacity to meaningfully engage communities, questions must be asked about the value and benefit of their research.

::::::::
::::::::

ACCESS, EQUITY AND BENEFITS SHARING

Health inequity in genomic personalized medicine in underrepresented populations: a look at the current evidence

Review
Sherouk M. Tawfik, Aliaa A. Elhosseiny, Aya A. Galal, Martina B. William, Esraa Qansuwa, Rana M. Elbaz & Mohamed Salama
Functional & Integrative Genomics 23, Article number: 54, 31 January 2023
Open access
Abstract
Improvements in sequencing technology coupled with dramatic declines in the cost of genome sequencing have led to a proportional growth in the size and number of genetic datasets since the release of the human genetic sequence by The Human Genome Project (HGP) international consortium. The HGP was undeniably a significant scientific success, a turning point in human genetics and the beginning of human genomics. This burst of genetic information has led to a greater understanding of disease pathology and the potential of employing this data to deliver more precise patient care. Hence, the recognition of high-penetrance disease-causing mutations which encode drivers of disease has made the management of most diseases more specific. Nonetheless, while genetic scores are becoming more extensively used, their application in the real world is expected to be limited due to the lack of diversity in the data used to construct them. Underrepresented populations, such as racial and ethnic minorities, low-income individuals, and those living in rural areas, often experience greater health disparities and worse health outcomes compared to the general population. These disparities are often the result of systemic barriers, such as poverty, discrimination, and limited access to healthcare. Addressing health inequity in underrepresented populations requires addressing the underlying social determinants of health and implementing policies and programs which promoted health equity and reduce disparities. This can include expanding access to affordable healthcare, addressing poverty and unemployment, and promoting policies that combat discrimination and racism.

Advancing Precision Medicine in Paediatrics: Past, present and future

Review
Abdelbaset Elzagallaai, Charlotte Barker, Tamorah Lewis, Ronald Cohn and Michael Rieder
Cambridge Prisms: Precision Medicine, Volume 1, 2023 , e11, 10 January 2023
Abstract
Precision Medicine is an approach to disease treatment and prevention taking into account individual genetic, environmental, therapeutic and lifestyle variability for each person. This holistic approach to therapeutics is intended to enhance drug efficacy and safety not only across healthcare systems but for individual patients. While weight and to some extent gestational age have been considered in determining drug dosing in children, historically other factors including genetic variability have not been factored into therapeutic decision making. As our knowledge of the role of ontogeny and genetics in determining drug efficacy and safety has expanded, these insights have provided new opportunities to apply principles of

Precision Medicine to the care of infants, children and youth. These opportunities are most likely to be achieved first in select subgroups of children. While there are many challenges to the successful implementation of Precision Medicine in children including the need to ensure that Precision Medicine enhances rather than reduces equity in children's health care rather, there are many more opportunities. Research, advocacy, planning and teamwork are required to move Precision Medicine forward in children in pursuit of the common goal of safe and effective drug therapy.

::::::::
::::::::

GENETIC SCREENING, COUNSELLING AND DIAGNOSIS

Community Genetics screening in a pandemic: solutions for pre-test education, informed consent, and specimen collection

Viewpoint

Bronwyn Terrill, Lauren McKnight, Angela Pearce, Heather Gordon, William Lo, I-Chieh Jennifer Lee, Monica Runiewicz, Alex Palmer, Lesley Andrews, Edwin Kirk, Daniel Goldberg, John Tucker, David Murray, Warren Kaplan, Sarah Kummerfeld & Leslie Burnett

European Journal of Human Genetics, 11 January 2023

Open access

Abstract

A Community Genetics carrier screening program for the Jewish community has operated on-site in high schools in Sydney (Australia) for 25 years. During 2020, in response to the COVID-19 pandemic, government-mandated social-distancing, 'lock-down' public health orders, and laboratory supply-chain shortages prevented the usual operation and delivery of the annual testing program. We describe development of three responses to overcome these challenges: (1) pivoting to online education sufficient to ensure informed consent for both genetic and genomic testing; (2) development of contactless telehealth with remote training and supervision for collecting genetic samples using buccal swabs; and (3) a novel patient and specimen identification 'GeneTrustee' protocol enabling fully identified clinical-grade specimens to be collected and DNA extracted by a research laboratory while maintaining full participant confidentiality and privacy. These telehealth strategies for education, consent, specimen collection and sample processing enabled uninterrupted delivery and operation of complex genetic testing and screening programs even amid pandemic restrictions. These tools remain available for future operation and can be adapted to other programs.

Informed consent for expanded carrier screening: Past, present, and future

Review

Britton D. Rink

Prenatal Diagnosis, 13 January 2023

Abstract

History, law, bioethics, and geocultural influences all have impacted the modern application of informed consent. It is a complex, multilayered process to communicate information and obtain voluntary patient permission before a health care intervention. Lack of provider education about genetic disorders, complexities of advanced genomic technologies, limited time during patient encounters, and low health literacy within a population all represent challenges to effective communication. There is no consensus on how informed consent in reproductive genetics is optimally obtained. Expanded carrier screening (ECS) is purposed to simultaneously test for a large list of diseases in a pan-ethnic manner. The increased use of ECS is driven by advances in genomic technologies, decreased cost, an improved understanding of single gene disorders, and in support of reproductive autonomy. Academic organizations recommend pretest counseling

when patients consider ECS, yet best practice is not established. Ongoing research is needed to determine how optimally implement informed consent given the increased complexity of ECS.

Legal Regulation of Preimplantation Genetic Diagnosis: A Comparative Analysis of the Baltic Sea Region and the Nordic Countries

Journal Article

Nastė Grubliauskienė

Teisė 2022 Vol. 125, pp. 72–85, Modified 24 January 2023

Open access

Abstract

This paper examines the concept and regulation of some of the latest research in the field of fertility – preimplantation genetic diagnosis – in Lithuania, Poland, the Baltic States, and the Nordic countries. Preimplantation genetic diagnosis raises many legal and ethical questions regarding the protection of embryos, manipulation of the human genome, selection by sex, and the relation of this diagnosis to other similar studies. International legislation or guidelines define genetic testing, including pre-implantation genetic diagnosis, quite broadly, due to the scope of regulation and nuances, leaving the right to decide to the discretion of each state. National regulation of preimplantation genetic diagnosis should be specific, clearly defining cases in which preimplantation genetic diagnosis is applied.

Benefits, Harms and Costs of Newborn Genetic Screening for Hypertrophic Cardiomyopathy: Estimates from the PreEMPT Model

Journal Article

Genetics in Medicine, 30 January 2023

Abstract

Purpose: Population newborn genetic screening for hypertrophic cardiomyopathy (HCM) is feasible, but its benefits, harms, and cost effectiveness are uncertain.

Methods: We developed a microsimulation model to simulate a United States birth cohort of 3.7 million newborns. Those identified with pathogenic/likely pathogenic variants associated with increased risk of HCM underwent surveillance and recommended treatment, compared to usual care, which included surveillance for individuals with family histories of HCM.

Results: In a cohort of 3.7 million newborns, newborn genetic screening would reduce HCM-related deaths through age 20 by 44 (95% uncertainty interval (95% UI): 10 to 103) but increase the numbers of children undergoing surveillance by 8,127 (95% UI, 6,308 to 9,664). Compared to usual care, newborn genetic screening costs $267,000 per life-year saved (95% UI, $106,000 to $919,000 per life-year saved).

Conclusion: Newborn genetic screening for HCM could prevent deaths but at a high cost and would require many healthy children to undergo surveillance. This study demonstrates how modeling can provide insights into the tradeoffs between benefits and costs that will need to be considered as newborn genetic screening is more widely adopted.

Comparing direct-to-consumer genetic testing services in English, Japanese, and Chinese websites

Article Overview

Kentaro Nagai, Mikihito Tanaka, Alessandro R. Marcon, Ryuma Shineha, Katsushi Tokunaga, Timothy Caulfield, Yasuko Takezawa

Anthropological Science

Abstract

Direct-to-consumer genetic testing (DTC-GT) has rapidly become available and affordable throughout developed countries. However, comparative research on DTC-GT services beyond Western countries has remained scarce, particularly in East Asian countries such as Japan and China. Hence, this study's hypothesis

is that although DTC-GT services in three languages might utilize the same underlying testing technology, such services are likely to represent the social, economic, and political characteristics of each country. For the study, a total of 267 websites (182 English, 32 Japanese, and 53 Chinese) were analyzed and coded reflexively into five categories for content analysis before interpretation using cluster and factor analyses. The results demonstrated variation between the three languages that reflected their respective consumer cultures: English, Chinese, and Japanese genetic testing websites focused on empowerment and ancestry; cultural values, especially familism; and health and beauty, respectively.

The impact of genetics on reproductive decisions, such as preimplantation genetic diagnosis.: Pre-implantation genomic diagnosis

Article

Deeblue Musomba, Ji Li, Xingpeng LAN, Wu Liji Ao

Science open, 13 January 2023

Open access

Abstract

Several European nations uphold the requirement of “high risk of a hereditary condition" to restrict the application of pre-implantation genome identification. This constraint about the "front door" ought to be relaxed to provide room for types of implantation genomic diagnosing having different proportionalities. This is true for both the procedure known as "added PGD," which is performed in conjunction with in vitro fertilization, and the procedure known as "combination pre-implantation genomic diagnosis," which is performed for an alternative disorder in addition to the one for which the individuals have an acknowledged pre-implantation genomic diagnosis reason. Both of these procedures fall under the purview of this rule. Therefore, relaxing the rules in the front of pre-implantation genomic diagnosis therapy has ramifications in the back, where a further pre-implantation genomic diagnosis rule states that 'affected conceptus' (meaning fertilized egg with the targeted variation or defect) should not be transplanted to the uterus. This 'rear door' regulation should be unstrained to permit for the implantation of 'last opportunity' affected conceptus in situations of aPGD and cPGD; however, this should only be done if there is not a great danger that the features would have a significantly decreased attribute of life as a result of the procedure.

An European overview of genetic counselling supervision provision

Journal Article

Milena Paneque, Lídia Guimarães, Joana Bengoa, Sara Pasalodos, Christophe Cordier, Irene Esteban, Carolina Lemos, Ramona Moldovan, Clara Serra-Juhé

European Journal of Medical Genetics, Volume 66, Issue 4, 30 January 2023

Abstract

Genetic testing is becoming more commonplace in general and specialist health care, and should always be accompanied by genetic counselling, according to legislation in many European countries and recommendations by professional bodies.

Personal and professional competence is necessary to provide safe and effective genetic counselling. Clinical and counselling supervision of genetics healthcare practitioners plays a key role in quality assurance, providing a safe environment not only for patients but for professionals too. However, in many European countries, genetic counsellors are still an emerging professional group and counselling supervision is not routinely offered and there are no enough evidences on the impact of these insufficiencies. This study aimed to explore the current status of genetic counselling supervision provision across Europe and to ascertain factors that might be relevant for the successful implementation of counselling supervision.

A total of 100 practitioners responded to an online survey; respondents were from 18 countries, with the majority working in France (27%) and Spain (17%). Only 34 participants reported having access to genetic counselling supervision. Country of origin, the existence of a regulation system and years of experience were factors identified as relevant, influencing access and characteristics of counselling supervision.

Although there is a growing number of genetic counsellors trained at European level, just a few countries have implemented and required as mandatory the access to genetic counselling supervision. Nevertheless, this is essential to ensure a safe and effective genetic counselling and should be regulated at the European genetic healthcare services.

Healthy, happy, rational: reflections on genetic counselling in the GDR

Original Research

Susanne Doetz

Med Humanities 0:1, 3 February 2023

Open access

Abstract

The development of genetic counselling in the German Democratic Republic (GDR) was closely connected to a well-established system of prenatal care and a process that placed reproductive decisions in the hands of women. It was embedded in the pronatalist reproductive policy of the GDR and a narrative of medical and (socialist) humanistic progress. As in other countries at that time, it promoted the goal of avoiding the birth of children with disabilities and was hence based on ableist premises. In this paper, I focus on communicative aspects of genetic counselling, as it was established in the 1970s and 1980s in university and district clinics. Thus, on the one hand I explore the communication of genetic counselling to the public; and on the other, I study the communication processes in genetic counselling centres themselves. In contrast to the USA, where the 'genetic counsellor' became established as a professional identity in the 1970s, there was no distinct profession of 'genetic counsellor' in the GDR. Instead, counselling was practised by physicians or biologists with a special interest in human genetics. This resulted in a strong emphasis in these clinical encounters on diagnosis and technical solutions, as well as an educational impetus. I propose that an important goal of genetic counselling in the GDR was to generate a sense of 'rationality' in prospective parents. To achieve this, those advocating and giving counselling explicitly sought to distance this practice from the eugenic ideas of the past, and to dispel superstitious ideas of heredity and religious ideas of fate. In addition, they attempted to alleviate emotions such as fear and guilt. It was in that context that counselling physicians and biologists provided interpretations of genetic findings, risk figures and disease values. I show how different interests and experiences shaped these and how risk evaluations structured counsellor-counsellee communication.

::::::::

::::::::

PLANTS, CROPS, AGRICULTURE, ANIMALS

Cisgenics and Genome Editing or Second Generation Biotechnologies: A Latin America and Caribbean Perspective

Chapter

María Mercedes Roca, José Falck-Zepeda & Paulo Paes-Andrade

Cisgenic Crops: Safety, Legal and Social Issues pp 195–243, 04 January 2023

Abstract

The Latin American and Caribbean (LAC) region must acknowledge both the potential of second-generation biotechnologies and the public's deep mistrust for new, untested technologies they feel are outside their control. The future success of cisgenic and Genome Edited (GEd) crops depend to a large extent on whether public policy is well-crafted. In order to build public confidence in the governance of second-generation biotechnologies, transparency, together with adherence to safety and environmental standards and ethical principles, is essential. However, the all-important safety aspects of policy must be guided by scientifically defensible, risk-based approaches rather than protectionist and restrictive trade and assistance policies, and public opinion, especially when the latter is driven by activist groups and geopolitical agendas. To promote sustainable development and global harmony, industrialized countries also have a moral imperative not to

influence policies that limit the development of other less advanced countries (Lower and Middle Income Countries, LMICs) and learn from the missteps of regulating Genetically Modified Organisms (GMOs) that illustrate that choosing a flawed paradigm has critical implications for a technology. This chapter answers a call from many scientists and stakeholders in the LAC region to policy makers, regulators, science communicators and society, to embrace innovation and seize the opportunity to make second-generation biotechnologies an accessible option for those who could benefit most, contribute to food security and make a circular bio-economy a feasible reality for the region. The agricultural and food systems in the Latin American and Caribbean (LAC) region need urgent reform, as the region is the world's breadbasket and its lungs. To enter the circular bio-economy and contribute to decarbonization, the region needs to enhance its biomass productivity with less inputs (land water, agrochemicals), with environmental responsibility and with social inclusion. Properly used biotechnologies, such as GMOs and New Breeding Techniques (NBTs), also known as Precision Breeding that may include cisgenesis, can contribute to a Climate-Smart-Agriculture and several of the UN's Sustainable Development Goals, especially SDG2. This chapter explores and discusses the adoption of second-generation biotechnologies and discusses mechanisms for crafting coherent science-based and risk-based policy for agriculture biotechnology in the region. Argentina, Brazil, Chile, Uruguay and Paraguay are leading the way in the Southern Cone, and Honduras and Guatemala in Central America, while Mexico, the Caribbean nations and the Andean countries are lagging behind with agricultural policies influenced by political populism and narrow sectorial interests, paradoxically leading to a host of unintended political, social and economic consequences, with the exception of Colombia. As a key agrifood trade partner to the EU, the chapter explores LAC's role and discusses the important influence and the unintended consequences of past and current anti-GMO advocacy, including the damaging EU's Farm-to Fork policies for the region. As a high goal, the chapter proposes policy harmonization on cisgenesis and NBTs in the region, following the lead from the Southern Cone countries, especially Argentina.

Elucidation and Acceptance—Scientific, Legal, and Ethical Aspects of Cisgenesis in Times of an Alleged Dwindling Faith in Science

Chapter

Gregor Becker, Julia Marcińska, Mehmet Nafi Artemel & Anna Juszczyk

Cisgenic Crops: Safety, Legal and Social Issues pp 77–99, 4 January 2023

Abstract

Cisgenics is a type of genetic modification that uses genes from the same or closely related species that can interbreed with the edited organism, opposed with transgenesis, where transferred genes come from outside of the gene pool of the edited species. In this chapter, we discuss cisgenics from the viewpoints of science and law and depict the social issues of genetic modifications struggle nowadays. In the science part, the cisgenesis and gene editing are explained. In the law part, the situation of products of cisgenesis in the eyes of the law in the European Union is described, including the possibility of changes in the near future. In the next part, the problems with acceptance of use and commercialization of genetically modified organisms and biotechnology as such are characterized, both from within the science community and the general public and an opinion whether cisgenics can change something in that matter is given.

Regulation of CRISPR edited food and feed: legislation and future

Chapter

Muhammad Zubair Ghouri, Nayla Munawar, Syed Ovais Aftab, Aftab Ahmad

GMOs and Political Stance, Pages 261-287, 19 January 2023

Abstract

In the modern era of biotechnology, gene editing technologies are pioneering the engineering of genomes of economically important crops in a way other than transgenics. Genome-edited (GenEd) crops are modified with an intention to precisely alter genes for better growth, product quality, nutrition, and sustainability in plants. Waxy corn, nonbrowning mushrooms, low-nicotine tobacco, herbicide-tolerant soybeans, and corn

are all the success stories of genome editing. All countries need to regulate the genome-edited crops before releasing them into the market or environment. There is still a debate on the regulation of GenEd crops in different countries; for example, the United States has excluded GenEd crops from being regulated while the EU has decided to regulate all types of modifications created using GenEd techniques under current genetically modified organism (GMO) regulations of the EU. If the world community remains divided over the regulation of GenEd crops, there is a risk that GenEd crops will soon became part of our food chain with potential risks. Therefore, there is a need to develop a clear strategy that can address all the issues associated with GenEd crops to overcome inconsistencies and ambiguities. In this book chapter, we summarize the current regulatory framework for GMOs and how GMOs' regulations would impact GenEd regulations. We discuss how existing regulations are not enough to accommodate GenEd crops. Additionally, we explain the concerns associated with CRISPR-edited products and current GenEd regulations in different countries. Finally, we give a detailed overview of scalable regulation, public concerns, governance issues, and ethical concerns of genome-edited crops.

An analysis of some ethical argumentation about genetically modified food

Research Article

Green, Nancy L.

Argument & Computation, vol. Pre-press, no. Pre-press, pp. 1-20, 25 January 2023

Abstract

We present an analysis of ethical argumentation and rhetorical elements in an article on the debate about growing genetically modified food (GMF), an issue of current interest in environmental ethics. Ethical argumentation is argumentation that a certain action is permissible, forbidden, or obligatory in terms of ethical intuitions, principles, or theories. Based on analysis of argumentation in the article, we propose several argumentation schemes for descriptive modeling of utilitarian arguments as an alternative to using more general schemes such as practical reasoning and argument from consequences. We also show how the article promoted its pro-GMF stance using rhetorical elements such as quotation, argument from expert opinion, and ad hominem attacks. Pedagogical and computational implications of the analysis of argumentation and rhetoric are discussed.

Governing gene-edited crops: risks, regulations, and responsibilities as perceived by agricultural genomics experts in Canada

Research Article

Sarah-Louise Ruder & Milind Kandlikar

Journal of Responsible Innovation, 24 January 2023

Open access

Abstract

This paper explores the role and responsibilities of agricultural genomics experts in governing gene editing (GE) for food and agriculture, engaging with the frameworks of technological determinism and Responsible Research and Innovation (RRI). We interview agricultural genomics experts in Canada to study expert views on risks, benefits, and regulatory challenges of GE crops and the extent to which agricultural genomics experts exercise the RRI principles of anticipation, reflexivity, deliberative inclusion, and responsiveness. Agricultural genomics experts wield power in food systems both in shaping the applications of technology and as advisers influencing policy and governance. Their resistance to RRI principles, especially deliberative inclusion and responsiveness, and exercises of discursive closure are challenges for responsible governance of GE crops. The study offers empirical and theoretical contributions, working across Science and Technology Studies and food systems research.

CISGENESIS AND CISGENIC CROPS: Need for a Paradigm Shift in Harnessing and Governance

Chapter

Krishna Ravi Srinivas

Cisgenic Crops: Safety, Legal and Social Issues pp 255–268, 4 January 2023

Abstract

Cisgenesis is an emerging technology that has much potential in developing crops and for other innovations. The possibilities of using Cisgenesis are discussed in the literature and there have been experiments to test it and they have proved that it is yet to be commercialized or adopted widely. A major reason is whether to assess it as a Genetically Modified Crop or as a traditionally bred crop or its equivalent. While the opinion on this is divided linking it with the regulation of Genetically Modified Organisms is resulting in controversies although the idea of cisgenesis was advocated by Schouten to overcome this issue. Describing the issue and the entanglements, and, by referring to two concepts 'bio-object' and 'bio-identification', this Chapter takes the view that the controversy will persist in the near future as regulations on genome edited crops are evolving but in a different direction. This chapter proposes a Global Consortium to pursue research and development in Cisgenesis and Cisgenic crops and suggests how this Consortium can use ideas and practices like Responsible Research and Innovation and Participatory Plant Breeding. Similarly, better engagement with stakeholders and dealing with non-technical issues in risk assessment are suggested.

Beyond the Genome: Genetically Modified Crops in Africa and the Implications for Genome Editing

Article

Joeva Sean Rock, Matthew A. Schnurr, Ann Kingiri, Dominic Glover, Glenn Davis Stone, Adrian Ely and Klara Fischer

Development and Change 1-26, 5 January 2023

Open access

Abstract

Genome editing — a plant-breeding technology that facilitates the manipulation of genetic traits within living organisms — has captured the imagination of scholars and professionals working on agricultural development in Africa. Echoing the arrival of genetically modified (GM) crops decades ago, genome editing is being heralded as a technology with the potential to revolutionize breeding based on enhanced precision, reduced cost and increased speed. This article makes two interventions. First, it identifies the discursive continuity linking genome editing and the earlier technology of genetic modification. Second, it offers a suite of recommendations regarding how lessons learned from GM crops might be integrated into future breeding programmes focused on genome editing. Ultimately, the authors argue that donors, policy makers and scientists should move beyond the genome towards systems-level thinking by prioritizing the co-development of technologies with farmers; using plant material that is unencumbered by intellectual property restrictions and therefore accessible to resource-poor farmers; and acknowledging that seeds are components of complex and dynamic agroecological production systems. If these lessons are not heeded, genome-editing projects are in danger of repeating mistakes of the past.

# # # #

# # # #