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Center for Genomic Medicine Ethics & Policy

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

May 2023 Number 05

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:

EDITOR'S SHORT LIST

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.

:: We lead this edition's short list with a milestones open access article – **A draft human pangenome reference** – published today in *Nature* by Wen-Wei Liao and collaborators. The article reports on the Human Pangenome Reference Consortium's "first draft of the human pangenome reference. The pangenome contains 47 phased, diploid assemblies from a cohort of genetically diverse individuals1. These assemblies cover more than 99% of the expected sequence in each genome and are more than 99% accurate at the structural and base pair levels." The authors observe that "A richer human reference map promises to improve our understanding of genomics and our ability to predict, diagnose and treat disease. A more diverse human reference map should also help ensure that the eventual applications of genomic research and precision medicine are effective for all populations…"

In a *Nature* news/analysis piece on the Consortium's report, Layal Liverpool inventories a number of ethical concerns associated with the project and others striving for pangenome reference tools.

:: Also from *Nature,* we note an important editorial – **The gene-therapy revolution risks stalling if we don't talk about drug pricing** – which echoes pricing and access issues in LMICs raised at the Third International Summit on Human Genome Editing, held in London in March. The editorla properly recognizes that beyond access and pricing issues,"… gene-therapy technologies are mired in debates around regulation and intellectual property…It's important that scientists have an active role in these debates, and that they push such discussions to the fore sooner rather than later."

:: Leonard Kimura et al. present some very interesting implementations of emerging technologies in their article **Amazon Biobank: a collaborative genetic database for bioeconomy development.** The authors lay out a "proposal on community-based genetic databases to facilitate a just distribution of benefits, employing blockchain to build a transparent and verifiable log of transactions involving genomic data, and smart contracts to implement an internal monetary system for all participants who collect, insert, process, store, and validate genomic data."

:: We also take note of a paper by Yusuke et al, on **Restrictions on monetary payments for human biological substances in Japan: The mu-shou principle and its ethical implications for stem cell research.** The paper examines some 28 Japanese laws and governmental guidelines around the "no payment" or the *mu-shou* principle in stem-cell research over the years and challenges in its implementation. The authors assess that inconsistencies in the monetary payment requirements, unless resolved, could hinder future research and development.

:: We conclude our short list with an important Hastings Center Report – **Wrestling with Social and Behavioral Genomics: Risks, Potential Benefits, and Ethical Responsibility.** Meyer and co-authors survey what they term the "often ugly" history of the field, discuss responsible behavior in the context of SBG research, and argue that future research "…that compares individuals within a group according to a "sensitive" phenotype requires extra attention to responsible conduct and to responsible communication about the research and its findings."

SHORT LIST ABSTRACTS/TEXT

A draft human pangenome reference

Article

Wen-Wei Liao…Benedict Paten

Nature, volume 617, pages 312–324 (2023) Published: 10 May 2023

Open Access

Abstract

Here the Human Pangenome Reference Consortium presents a first draft of the human pangenome reference. The pangenome contains 47 phased, diploid assemblies from a cohort of genetically diverse individuals[1]. These assemblies cover more than 99% of the expected sequence in each genome and are more than 99% accurate at the structural and base pair levels. Based on alignments of the assemblies, we generate a draft pangenome that captures known variants and haplotypes and reveals new alleles at structurally complex loci. We also add 119 million base pairs of euchromatic polymorphic sequences and 1,115 gene duplications relative to the existing reference GRCh38. Roughly 90 million of the additional base pairs are derived from structural variation. Using our draft pangenome to analyse short-read data reduced small variant discovery errors by 34% and increased the number of structural variants detected per haplotype by 104% compared with GRCh38-based workflows, which enabled the typing of the vast majority of structural variant alleles per sample.

Discussion Excerpt: Concluding Text

….We acknowledge that references generated from the 1KG samples alone are insufficient to capture the extent of sequence diversity in the human population. To ensure that we are able to maximize our surveys of sample diversity while abiding by principles of community engagement and avoiding extractive practices[14,15], we will broaden our efforts to recruit new participants to improve the representation of human genetic diversity. A richer human reference map promises to improve our understanding of genomics and our ability to predict, diagnose and treat disease. A more diverse human reference map should also help ensure that the eventual applications of genomic research and precision medicine are effective for all populations. We recognize that the value of this project will partly be in the future establishment of new standards for how we capture variant diversity, the opportunity to disseminate science into diverse communities and continued efforts to engage with diverse voices in this ambitious goal to build a common global reference resource. The methods we are developing should prove valuable for other species. Indeed, other groups are pioneering such efforts[69,70]. In parallel with our efforts to obtain a more comprehensive collection of diverse and highly accurate human reference genomes, we anticipate further optimization and rapid improvement of the pangenome reference, enabling an increasingly broad set of applications and use cases for both the research and clinical communities.

First human 'pangenome' aims to catalogue genetic diversity

Researchers release draft results from an ongoing effort to capture the entirety of human genetic variation

NEWS

Layal Liverpool

Nature, volume 617, Published: 10 May 2023

[Excerpt; Concluding text]

…Ethical considerations

However, some researchers — including Fox — are concerned that the project risks repeating ethically questionable practices from other large-scale genetic-diversity projects. For instance, the Human Genome Diversity Project in the 1990s and the ongoing All of Us Research Program received criticism, including from US-based tribes, for failing to engage sufficiently with members of the communities whose DNA they were sampling. These included people belonging to marginalized groups usually under-represented in human genetic research.

"We will, of course, advance knowledge of human structural variation with new data sets and new tools. The progress we should be striving for, however, is the equitable engagement of under-represented communities in this work from the ground up," says Krystal Tsosie, a genetic epidemiologist and bioethicist at Arizona State University in Tempe. Tsosie is also co-founder of the Native BioData Consortium, a non-profit research institute in Eagle Butte, South Dakota, led by Indigenous scientists and tribal members. "If the research is not benefiting the diverse communities first and foremost, then we are doing something fundamentally wrong here," she says.

Fox, who is on the board of the Native BioData Consortium, agrees. He is concerned that data from the Human Pangenome Reference Project, which is funded by the US National Institutes of Health, could be used by the pharmaceutical industry for commercial purposes — as has happened in the past3 — without tangible benefits to the study participants or their communities.

Revisiting consent

Latifa Jackson, a geneticist at Howard University in Washington DC, points out that the 1000 Genomes Project relied partly on samples collected many years before it launched. "I am concerned that many of the participating pangenome locations have samples that were collected in the 1980s under very different political and social structures," she says. "We need to revisit ideas of consent, especially for samples collected 30–40 years ago under very different power structures."

"We recognize that this work is at the forefront of genomic research and has specific features, including open access of data, that warrant a great deal of consideration, and that the applications can raise ethical, legal and social issues," Kenny said at the press conference. "We have drawn not only from our own expertise, but also built on the work of scholars and organizations throughout the world, to be aware of the many pitfalls and to systematically review other efforts for lessons learned, that we can bring into this new initiative."

Kenny added that the pangenome consortium is hoping to recruit new study participants as part of its effort to maximize diversity among the planned 350 genomes. These will include participants obtained "through a large health system in an urban city like New York, which has people from almost every country of the world through processes of diaspora and migration", said Kenny.

The gene-therapy revolution risks stalling if we don't talk about drug pricing

Editorial

Nature 616, 629-630 (2023), 25 April 2023

Introduction

Regulation and new intellectual property laws are needed to reduce the cost of gene-editing treatments and fulfil their promise to improve human health. "We wish to suggest a structure for the salt of deoxyribose nucleic acid (D.N.A.)," wrote James Watson and Francis Crick in this journal in 1953 (J. D. Watson and F. H. C. Crick Nature 171, 737–738; 1953). "This structure has novel features which are of considerable biological interest." In the 70 years since those famous words were published, researchers have poured huge effort into unravelling those features and harnessing them for medicine. The result is a flourishing understanding of the genetic causes of diseases — and a host of therapies designed to treat them.

Seventy years from now, the world might look back on 2023 as a landmark, as well. This year could see the first authorization of a therapy based on CRISPR–Cas9 gene editing, that involves tweaking the DNA in the body's non-reproductive (somatic) cells. Gene editing allows scientists — and could soon permit clinicians — to make changes to targeted regions in the genome, potentially 'correcting' genes that cause disease. Regulators in the United States, the European Union and the United Kingdom are evaluating a therapy that uses this approach to treat sickle-cell disease, and a decision could be made in the next few months. But even as such advances accrue, researchers are worrying about the future role of gene editing — as well as other, more established forms of gene therapy — in treating disease.

Gene therapies currently carry eye-watering price tags, putting them out of the reach of many who need them. High prices could diminish the willingness of government funders to pay for gene-therapy research. And that, in turn, would make it harder for research institutions to continue to attract top talent to the field.

Researchers, especially health economists, must work urgently with industry and governments to find a more affordable funding model…

Million-dollar treatments

…Even in wealthy countries, health-care systems are ill-equipped to shoulder the high initial costs associated with gene therapies. In 2021, therapeutics developer Bluebird Bio in Somerville, Massachusetts, withdrew plans to market a gene therapy for β-thalassaemia — another blood disorder — in Europe, after failing to reach an agreement with European authorities over the price. It said it would focus its sales efforts on the United States, where there has been comparatively little regulation of drug costs.

But even in the United States, costs matter. US health insurance is often subsidized by employers, and some are already saying that they will probably restrict their coverage of gene therapies in the next year, says Steven Pearson, president of the Institute for Clinical and Economic Review, a health-economics think tank in Boston, Massachusetts.

Low- and middle-income countries, meanwhile, are left entirely in the lurch. This is especially painful given that some of the diseases under consideration, such as β-thalassaemia and sickle-cell disease, are more common in poorer parts of the world than in wealthy nations. In some sub-Saharan regions, for example, it is estimated that about 2% of children are born with sickle-cell disease. This is likely to be an underestimate, given how little screening is taking place...

Improving access

…At the Third International Summit on Human Genome Editing, held in London in March, much of the discussion centred on making gene-editing therapies accessible, particularly to low- and middle-income countries. The focus was on technological approaches to streamline the production and testing of such treatments. The sickle-cell treatment, for example, requires clinicians to isolate and edit blood-forming stem cells, destroy those that remain in the body, and then reinfuse the edited cells. Converting this to a genome-editing procedure that could be performed directly in the body rather than in isolated cells could make the treatment cheaper and more accessible…

…How much that price will drop in other countries could be limited by intellectual property rights and hindered by the complexities of making generic copies of biological drugs such as gene therapies. Some academic centres are trying to develop and deploy gene therapies without relying on pharmaceutical companies, but it is unclear how far such efforts can stretch without the financial resources and regulatory expertise found in industry.

In addition to pricing, gene-therapy technologies are mired in debates around regulation and intellectual property. How each of these plays out will determine how far researchers can go in capitalizing on Watson and Crick's initial discovery. It's important that scientists have an active role in these debates, and that they push such discussions to the fore sooner rather than later.

Amazon Biobank: a collaborative genetic database for bioeconomy development

Original Article

Leonardo T. Kimura, Ewerton R. Andrade, Ismael Nobre, Carlos A. Nobre, Bruno A. S. de Medeiros, Diego M. Riaño-Pachón, Felipe K. Shiraishi, Tereza C. M. B. Carvalho & Marcos A. Simplicio Jr.

Functional & Integrative Genomics, Volume 23, Article number: 101 (2023), 25 March 2023

Abstract

Biodiversity is proposed as a sustainable alternative for the economic development of high-biodiversity regions. Especially in the field of biodiversity genomics, the development of low-cost DNA sequencing opens an opportunity for new actors beyond academia to engage in genomic sequencing. However, it is challenging to adequately compensate non-academic actors such as local populations for their contribution to the innovation process, preventing better bioeconomy development. Although many repositories register genomic data to support biodiversity research, they do not facilitate the fair sharing of economic benefits. In this work, we propose the creation of the Amazon Biobank, a community-based genetic database.

We employed blockchain to build a transparent and verifiable log of transactions involving genomic data, and we used smart contracts to implement an internal monetary system for all participants who collect,

insert, process, store, and validate genomic data. We also used peer-to-peer solutions to allow users with commodity computers to collaborate with the storage and distribution of DNA files.

By combining emerging technologies, Amazon Biobank provides adequate benefit-sharing among all participants that collaborate with data, knowledge, and computational resources. It also provides traceability and auditability, allowing easy association between biotechnological research and DNA data. In addition, the solution is highly scalable and less dependent on the trust deposited in any system player. Therefore, Amazon Biobank can become an important steppingstone to unlock the potential of bioeconomy in rich ecosystems such as the Amazon Rainforest.

Restrictions on monetary payments for human biological substances in Japan: The mu-shou principle and its ethical implications for stem cell research

Original Article

Yusuke Inoue, Tohru Masui, Kana Harada, Hyunsoo Hong, Minori Kokado

Regenerative Therapy, Volume 23, 2023, Pages 1-7, 09 March 2023

Open access

Abstract

Introduction

Restrictions on financial gains from the sale of human body parts is a leading policy issue surrounding the use of human tissues and cells. However, discrepancies exist between regulations and reality. In stem cell research, in which diverse sources of tissues and cells can be used, unclear regulations can impede research. Thus, using the Japanese system as a case study, we examined the challenges in the implementation of the "no payment" or the *mu-shou* principle in stem-cell research over the years.

Methods

We reviewed 28 Japanese laws and governmental guidelines and summarized the scope of restrictions on payments for the donation and supply of human biological samples (HBS).

Results

As part of restrictions on financial rewards, the *mu-shou* principle emerged in Japanese laws and administrative documents in the 1990s. Although the Japanese *mu-shou* generally means "free" or "gratis" in English, its interpretation in research and development settings remains ambiguous. Traditionally, this principle was used to deny remuneration to donors. However, it is also inconsistently applied while processing and transferring human tissue after donation, which creates confusion among the various stakeholders. Recent policies have interpreted the principle in multiple ways: (1) treating the use of HBS for cell-processing as a non-profit activity; (2) a flexible interpretation of the principle to broaden the scope of user payments; and (3) removal of the principle itself to allow for commercial use.

Conclusions

The inconsistencies in the monetary payment requirements for HBS could hinder research and development. After scrutinizing the principle's background, an effective approach is needed that considers the concerns of the providers, users, and society alike.

Wrestling with Social and Behavioral Genomics: Risks, Potential Benefits, and Ethical Responsibility

Article Free Access

Michelle N. Meyer, Paul S. Appelbaum, Daniel J. Benjamin, Shawneequa L. Callier, Nathaniel Comfort, Dalton Conley, Jeremy Freese, Nanibaa' A. Garrison, Evelynn M. Hammonds …

The Hastings Center Report, First published: 20 April 2023 https://doi.org/10.1002/hast.1477

Abstract

In this consensus report by a diverse group of academics who conduct and/or are concerned about social and behavioral genomics (SBG) research, the authors recount the often-ugly history of scientific attempts to understand the genetic contributions to human behaviors and social outcomes. They then describe what the

current science—including genome-wide association studies and polygenic indexes—can and cannot tell us, as well as its risks and potential benefits. They conclude with a discussion of responsible behavior in the context of SBG research. SBG research that compares individuals within a group according to a "sensitive" phenotype requires extra attention to responsible conduct and to responsible communication about the research and its findings. SBG research (1) on sensitive phenotypes that (2) compares two or more groups defined by (a) race, (b) ethnicity, or (c) genetic ancestry (where genetic ancestry could easily be misunderstood as race or ethnicity) requires a compelling justification to be conducted, funded, or published. All authors agree that this justification at least requires a convincing argument that a study's design could yield scientifically valid results; some authors would additionally require the study to have a socially favorable risk-benefit profile.

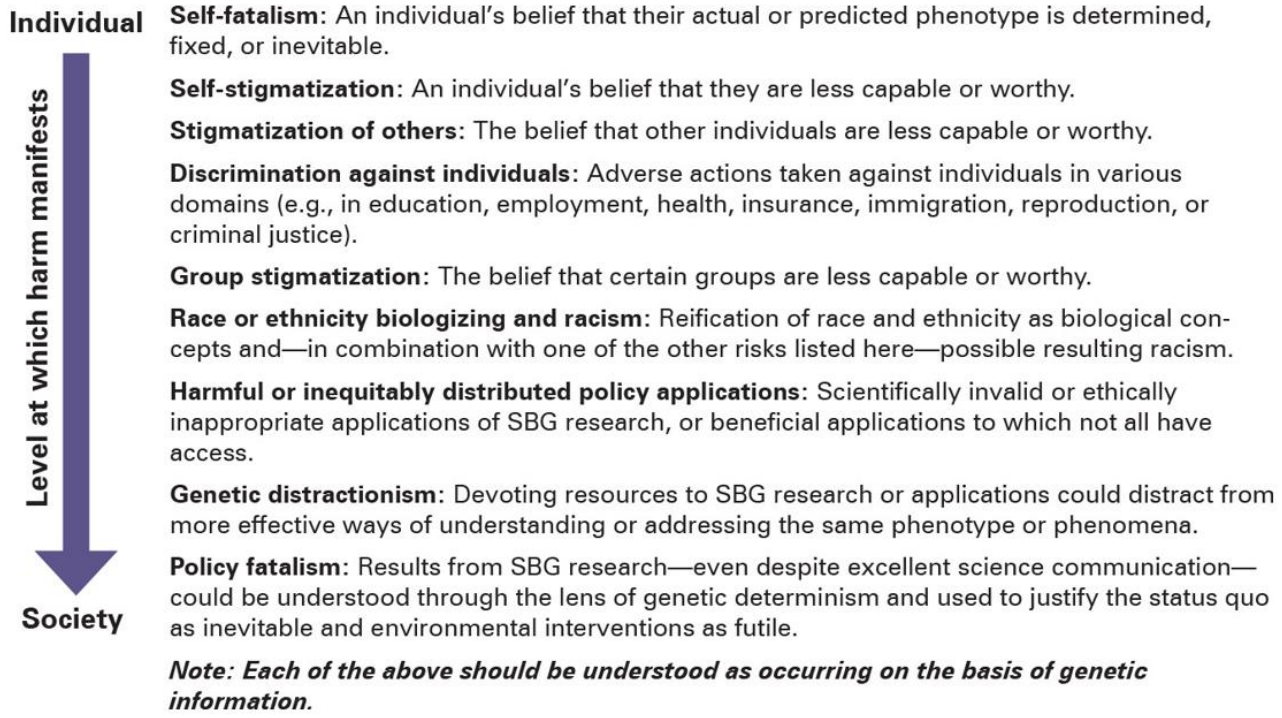

Figure 3

Risks of Sociobehavioral Genomics Research

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GENOMIC DATA, BIOBANKING, GENOMIC RESEARCH

Return of Results in Genomic Research Using Large-Scale or Whole Genome Sequencing: Toward a New Normal

Susan M. Wolf and Robert C. Green

Annual Review of Genomics and Human Genetics, Vol 24, 13 March 2023

Abstract

Genome sequencing is increasingly used in research and integrated into clinical care. In the research domain, large-scale analyses, including whole genome sequencing with variant interpretation and curation, virtually guarantees identification of variants that are pathogenic or likely pathogenic and actionable.

Multiple guidelines recommend that findings associated with actionable conditions be offered to research participants in order to demonstrate respect for autonomy, reciprocity, and participant interests in health and privacy. Some recommendations go further and support offering a wider range of findings, including

those that are not immediately actionable. In addition, entities covered by the US Health Insurance Portability and Accountability Act (HIPAA) may be required to provide a participant's raw genomic data on request. Despite these widely endorsed guidelines and requirements, the implementation of return of genomic results and data by researchers remains uneven.

This article analyzes the ethical and legal foundations for researcher duties to offer adult participants their interpreted results and raw data as the new normal in genomic research.

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

Journals of Medical Ethics, 20 March 2023

Open access

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.

Editor's note: from the discussion section the main recommendation provided is "…..for future studies, consent forms should explicitly address the secondary use of research data and/or samples by commercial entities. Ideally, if commercial entities are involved in the research from the outset, all material agreements should be adequately documented and related to participants. The nature of the commercial entities' involvement, what data they will have access to, and how they will use such data should be clarified during the informed consent process".

A practical checklist for return of results from genomic research in the European context

Article

Danya F. Vears, Nina Hallowell, Heidi Beate Bentzen, Bridget Ellul, Therese Haugdahl Nøst, Angeliki Kerasidou, Shona M. Kerr, Michaela Th. Mayrhofer, Signe Mežinska, Elizabeth Ormondroyd, Berge Solberg, Birgitte Wirum Sand & Isabelle Budin-Ljøsne

European Journal of Human Genetics (2023), 22 March 2023

Open access

Abstract

An increasing number of European research projects return, or plan to return, individual genomic research results (IRR) to participants. While data access is a data subject's right under the General Data Protection Regulation (GDPR), and many legal and ethical guidelines allow or require participants to receive personal data generated in research, the practice of returning results is not straightforward and raises several practical and ethical issues. Existing guidelines focusing on return of IRR are mostly project-specific, only discuss which results to return, or were developed outside Europe.

To address this gap, we analyzed existing normative documents identified online using inductive content analysis. We used this analysis to develop a checklist of steps to assist European researchers considering whether to return IRR to participants. We then sought feedback on the checklist from an interdisciplinary panel of European experts (clinicians, clinical researchers, population-based researchers, biobank managers, ethicists, lawyers and policy makers) to refine the checklist.

The checklist outlines seven major components researchers should consider when determining whether, and how, to return results to adult research participants: 1) Decide which results to return; 2) Develop a plan for return of results; 3) Obtain participant informed consent; 4) Collect and analyse data; 5) Confirm results; 6) Disclose research results; 7) Follow-up and monitor.

Our checklist provides a clear outline of the steps European researchers can follow to develop ethical and sustainable result return pathways within their own research projects. Further legal analysis is required to ensure this checklist complies with relevant domestic laws.

Researchers' Perspectives Regarding Ethical Issues of Biobank Research in the Arab Region

Article

Maha E. Ibrahim, Latifa Adarmouch, Alya Elgamri, Samar Abd ElHafeez, Zeinab Mohammed, Fatma Abdelgawad, Eman H. Elsebaie, Ahmed Samir Abdelhafiz, Ehsan Gamel, Karima El Rhazi, Asmaa Abdelnaby, Mamoun Ahram, and Henry Silverman

Biopreservation and Biobanking, 23 March 2023

Abstract

Background

The recent expansion of genomic biobank research in the Arab region in the Middle East North Africa has raised complex ethical and regulatory issues. However, there is a lack of studies regarding the views of Arab researchers involved in such research. We aimed to assess the perceptions and attitudes of Arab researchers regarding these issues in biobank research.

Methods

We developed a questionnaire to assess the perceptions and attitudes regarding genetic research of researchers from Egypt, Sudan, Morocco, and Jordan. The questionnaire requested demographic data, perceptions, and attitudes regarding the collection, storage, and use of biospecimens and data, the use of broad consent, data security, data sharing, and community engagement. We used multiple linear regressions to identify predictors of perceptions and attitudes.

Results

We recruited 383 researchers. Researchers favored equally the use of broad and tiered consent (44.1% and 39.1%, respectively). Most respondents agreed with the importance of confidentiality protections to ensure data security (91.8%). However, lower percentages were seen regarding the importance of community engagement (64.5%), data sharing with national colleagues and international partners (60.9% and 41.1%, respectively), and biospecimen sharing with national colleagues and international partners (59.9% and 36.2%, respectively). Investigators were evenly split on whether the return of individual research results should depend on the availability or not of a medical intervention that can be offered to address the genetic anomaly (47.5% and 46.4%, respectively). Predictors of attitudes toward biospecimen research included serving on Research Ethics Committees, prior research ethics training, and affiliation with nonacademic institutions.

Conclusions

We recommend further exploratory research with researchers regarding the importance of community engagement and to address their concerns about data sharing, with researchers within and outside their countries.

Public attitudes toward cloud computing and willingness to share personal health records (PHRs) and genome data for health care research in Japan

Article

Mayumi Kusunose & Kaori Muto

Human Genome Variation volume 10, Article number: 11 (2023), 30 March 2023

Open access

Abstract

Japan's government aims to promote the linkage of medical records, including medical genomic testing data and personal health records (PHRs), via cloud computing (the cloud). However, linking national medical records and using them for health care research can be controversial. Additionally, many ethical issues with using cloud networks with health care and genome data have been noted. However, no research has yet explored the Japanese public's opinions about their PHRs, including genome data, being shared for health care research or the use of the cloud for storing and analyzing such data.

Therefore, we conducted a survey in March 2021 to clarify the public's attitudes toward sharing their PHRs, including genome data and using the cloud for health care research. We analyzed data to experimentally create digital health basic literacy scores (BLSs).

Our results showed that the Japanese public had concerns about data sharing that overlapped with structural cloud computing issues. The effect of incentives on changes in participants' willingness to share data (WTSD) was limited. Instead, there could be a correlation between WTSD and BLSs.

Finally, we argue that it is vital to consider not only researchers but also research participants as value co-creators in health care research conducted through the cloud to overcome both parties' vulnerability.

Data sharing in the context of community-engaged research partnerships

Article

Karen M. Emmons, Samuel Mendez, Rebekka M. Lee, Diana Erani, Lynette Mascioli, Marlene Abreu, Susan Adams, James Daly, Barbara E. Bierer, For the RADx-MA Partners

Social Science & Medicine, Volume 325, May 2023, 115895, 14 April 2023

Abstract

Over the past 20 years, the National Institutes for Health (NIH) has implemented several policies designed to improve sharing of research data, such as the NIH public access policy for publications, NIH genomic data sharing policy, and National Cancer Institute (NCI) Cancer Moonshot public access and data sharing policy. In January 2023, a new NIH data sharing policy has gone into effect, requiring researchers to submit a Data Management and Sharing Plan in proposals for NIH funding (NIH. Supplemental information to the, 2020b; NIH. Final policy for data, 2020a).

These policies are based on the idea that sharing data is a key component of the scientific method, as it enables the creation of larger data repositories that can lead to research questions that may not be possible in individual studies (Alter and Gonzalez, 2018; Jwa and Poldrack, 2022), allows enhanced collaboration, and maximizes the federal investment in research. Important questions that we must consider as data sharing is expanded are to whom do benefits of data sharing accrue and to whom do benefits not accrue? In an era of growing efforts to engage diverse communities in research, we must consider the impact of data sharing for all research participants and the communities that they represent.

We examine the issue of data sharing through a community-engaged research lens, informed by a long-standing partnership between community-engaged researchers and a key community health organization (Kruse et al., 2022). We contend that without effective community engagement and rich contextual knowledge, biases resulting from data sharing can remain unchecked. We provide several recommendations that would allow better community engagement related to data sharing to ensure both community and researcher understanding of the issues involved and move toward shared benefits.

By identifying good models for evaluating the impact of data sharing on communities that contribute data, and then using those models systematically, we will advance the consideration of the community perspective and increase the likelihood of benefits for all.

Cloud-based biomedical data storage and analysis for genomic research: landscape analysis of data governance in emerging NIH-supported platforms

Article

Jacklyn M. Dahlquist, Sarah C. Nelson, and Stephanie M. Fullerton

Human Genetics and Genomics Advances 4, 100196, July 13, 2023, 27 April 2023

Summary

The storage, sharing, and analysis of genomic data poses technical and logistical challenges that have precipitated the development of cloud-based computing platforms designed to facilitate collaboration and maximize the scientific utility of data. To understand cloud platforms' policies and procedures and the implications for different stakeholder groups, in summer 2021, we reviewed publicly available documents (N ¼ 94) sourced from platform websites, scientific literature, and lay media for five NIH-funded cloud platforms (the All of Us Research Hub, NHGRI AnVIL, NHLBI BioData Catalyst, NCI Genomic Data Commons, and the Kids First Data Resource Center) and a pre-existing data sharing mechanism, dbGaP.

Platform policies were compared across seven categories of data governance: data submission, data ingestion, user authentication and authorization, data security, data access, auditing, and sanctions. Our analysis finds similarities across the platforms, including reliance on a formal data ingestion process, multiple tiers of data access with varying user authentication and/or authorization requirements, platform and user data security measures, and auditing for inappropriate data use. Platforms differ in how data tiers are organized, as well as the specifics of user authentication and authorization across access tiers.

Our analysis maps elements of data governance across emerging NIH-funded cloud platforms and as such provides a key resource for stakeholders seeking to understand and utilize data access and analysis options across platforms and to surface aspects of governance that may require harmonization to achieve the desired interoperability.

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GENE THERAPY/ GENOME EDITING

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

Article

Houria Bachtarzi

Human Gene Therapy. Volume: 34 Issue 5-6: 171-176. 20 March 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.

Ethical and Legal Aspects of Cloning the Human Body: Current Challenges

Article

Tetiana Tarasevych, Serhii Grynko, Oleksandr Ostapenko, Maryna Dzeveluk, Nataliia Chernyshchuk

Lex Humana, v. 15, n.2, 2023, 14 March 2023

Abstract

Legal regulation in the field of genetic engineering is becoming more and more relevant. Therefore, the purpose of the academic paper is to clarify the development of ethical and legal aspects in cloning the human body and determine the features of this process and the current challenges. Such methods as analysis, synthesis, generalization, explanation and qualification of data were used in the process of writing the scientific work.

The primary sources for clarifying our research purpose were the fundamental Convention on Human Rights and Biomedicine as of 1997 and international legal and domestic regulations of the issue outlined. The problem of human cloning has not only technological and moral aspects. It should be considered more broadly in the context of the science progress, the possibility and necessity of its regulation, including the legal and ethical aspects of any research paper. Moreover, it is about the legal regulation of human cloning at the national and international levels. This is the case when the legal influence has a precautionary nature and makes it possible to trace the emergence and development of a new field of legal regulation.

Editor's note: Lex Humana is a journal hosted by the Universidade Catolica de Petropolis, Rio de Janeiro, Brasil

Research Integrity in Emerging Technologies: Gene Editing and Artificial Intelligence (AI) Research in Medicine

Chapter

Barbara Redman

Reconstructing Research Integrity pp 153–171, 24 March 2023, Springer

Abstract

Emerging technologies challenge research integrity in several important ways. Technical research and product development usually precede (often by several years) attention to social and ethical issues, leading to a need to discover and "clean up" harmful effects that should have been anticipated. A frequent assumption that the science/research community should direct development of the technology, is badly mistaken. Issues of justice related to access to or freedom from the technology, and to participate in its development and balance of burdens versus access to its benefits at various stages of development must be engaged.

From test tube babies to human clones: salient issues in the international law of biomedicine

Chapter

Matthias Herdegen

Law 2023, pp 86–112, 18 April 2023, Elgar Online

Abstract

Socio-political, ethical and legal debates have always surrounded biomedicine. Reproductive medicine and, particularly, in vitro fertilisation, remain fields of legal and ethical controversy. Other contested areas are the use of human embryos for scientific research or therapeutic purposes, somatic gene therapy, and interventions in the germ line. Categoric bans on germline therapy come under pressure as control of undesirable collateral effects increases. Particular concern has arisen from the possibility of human cloning. In this regard, many jurisdictions distinguish between "reproductive" cloning leading to the creation of an identical human being, on the one hand, and "therapeutic" cloning on the other hand. These areas provide ample material for the delicate interplay between domestic and international law. The Coronavirus pandemic

has posed a challenge for the international community and triggered unprecedented cooperation and new approaches to vaccine regulation.

Current Applications of Gene Manipulation and the Associated Ethical Considerations

Student publications

Cassandra Poole (Faculty Advisor: Dr. Wade Znosko)

The Journal of Undergraduate Scholarship, Longwood University, Volume 14, 2023, pp 35-55

Abstract

Over the past decade, there have been rapid advances in the field of genetic editing and gene therapy as techniques are becoming more precise and more reliable. Currently, there are gene therapies in use or development to treat a wide variety of human diseases, including cancer, hemophilia, retinal dystrophy, severe combined immunodeficiency, and spinal muscular atrophy.

The use of gene therapy and gene editing can be a controversial topic. There are a variety of potential side effects, which raise questions of safety. Informed consent is another consideration, as is the potential stigmatization of those with disabilities linked to genetic causes. While most gene therapy applies to somatic cells, germline gene manipulation can involve editing the genetic material in reproductive cells, zygotes, and embryos, causing effects which are inheritable by future generations.

Therefore, germline gene manipulation raises its own set of ethical questions. Germline gene manipulation could have unpredictable effects on the human gene pool. Additionally, some detractors are concerned that informed consent cannot be obtained from future offspring, who would also be affected by any change in germline cells. Given the rapid pace of scientific development, regulations must be put into place as soon as possible to ensure the ethical use of gene therapy and gene editing technologies.

Status of Human Embryo in vitro as Ethical and Legal Issue: Religious Roots of Diverging Approaches

Research Article

Valentina V. Lapaeva

Legal Issues in the Digital Age, vol. 4, no. 1, pp. 4–23, 01 April 2023

Abstract

The paper is focused on the ontological status of the human embryo in vitro, a question that determines its ethical and legal status that is in turn of exceptional importance for ethical and legal regulation of manipulations with the embryo in the course of academic research as well as in clinical practice of assisted reproductive technologies.

The author discusses different approaches (Roman Catholic, Protestant, Greek Orthodox, etc.) to the issue of embryo status that have emerged in different parts of the world in the course of history from the perspective of religious anthropology. It is argued thesis that the idea of God-likeness of human person in the Christian culture giving a powerful impetus to the scholar and technological change originally contained profound ideological premises capable of inhibiting the most dangerous intrusions into the nature of human nature created after the likeness of God. One such premise is the idea that the human embryo is attributed with a soul from the moment of its conception.

Those countries, whose cultural matrix does not provide for such moral, religious constraints, have a competitive advantage in the globalized research and technological context that in a sense concerns the human civilization as such. This circumstance has become a contributing factor in the emerging change in the international ethical and legal regulation setting the limits to genetic research of the embryonic development of human person.

The main vector of the change has been determined by liberalization of former constraints date back to the dogmatic Christian view of the world. Moreover, the latest innovations in this area demonstrate an intention of the medical and biological academic community to share the responsibility for the development of

regulatory policies concerning human embryo research with specialists of other branches of sciences and with public at large.

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GOVERNANCE AND REGULATORY FRAMEWORKS

Governing emerging technologies of global significance in the developing countries: the case for synthetic biology regulation in Kenya

Thesis

Odhiambo Alphonce Kasera

A Thesis Submitted in Partial Fulfillment for the Requirement of the Degree of Masters of Arts in International Relations School of Development and Strategic Studies, Maseno University, 2023

Abstract

Technologies with transnational impact can no longer be relegated as 'mundane artefacts' in International Relations (IR). Two recent events validate this assertion. In 2014 world found itself in stress and confusion due to unprecedented Ebola attack. Very recently the novel COVID-19 threatened the very existence of mankind. In both events synthetic biology (SynBio) techniques saved the world, by enabling scientists to study and imitate the genetic make-up of the viruses and create a vaccine. Despite such immense value of SynBio, the field remains dominated by developed nations.

Additionally, effective global governance of SynBio requires proper regulation in all countries including developing countries like Kenya. Against this backdrop, and motivated by the fact that despite the Government of Kenya (GoK) commissioning a synthetic biology (SynBio) project in 2020 in line with her Vision 2030, it remains blurred the extent to which Kenya's current biotechnology regulatory frameworks are sufficient to the regulation of SynBio. This study explored Kenya's biotechnology regulatory environment. Study specific objectives explored: Kenya's biotechnology-related policy frameworks; biotechnology-related legislations; the extent to which the theme of Science, Technology and Innovation (ST&I) is embedded into selected national development plans (NDPs) and; key expert stakeholders' perceptions and expectations on the adoption of SynBio technologies in Kenya.

A conceptual framework derived from the concept of national power as used in International Relations and the theory of adaptive anticipatory governance guided the collection, analysis and interpretation of findings. Exploratory sequential mixed-method design was utilized. Study locations were Nairobi, Kisumu, Kakamega, and Kisii Counties and on Zoom, Google Meet and Gmail platforms. Study population composed 83 purposively sampled experts stratified into academia, research, industry, medical, and policy, governance and regulatory and media & communication sectors. Data collection was done through documentary analysis of 6 policies, 8 legislations, and 5 NDPs; survey questionnaires, 4 Focus Group Discussions (FGDs), and 22 key informant interviews (KIs). Quantitative data was analyzed through simple descriptive statistics while qualitative through thematic analysis.

The study established, a) while the biotechnology development policy 2006 and Biosafety Act 2009 are the main policy and legislations, respectively, governing biotechnology in Kenya, their scopes do not however cover biosecurity, ethical, social and economic issues that come handy with SynBio regulation; b) Kenya Vision 2030 and the Big Four Agenda place ST&I at the core of national development, but the ST&I theme is not emphasized in other NPDs and the place of biotechnology in these two key NDPs and other relevant NDPs is not properly spelt out; c) there is above average national capacity to adopt and implement SynBio in terms of requisite human expertise (90%); further, key regulatory and research institutions were rated above average: NACOSTI-86%; NBA-60%; KALRO-67% and KEMRI-60% except for NEMA-46%. These findings lead to the conclusion that Kenya has a robust biotechnology regulatory system but to optimally gain from SynBio technologies, the biotech governance frameworks will have to be tailor-made to cover the unique SynBio regulatory issues.

The study thus recommends to the GoK and concerned stakeholders to ensure the establishment of clearly spelt-out SynBio policy, legislation and an overarching NDP. The findings of this study thus revealed the extent to which current biotech governance in Kenya can regulate SynBio. Such evidence is relevant to IR debates insofar as it will inform debates around global governance of SynBio. The evidence is also locally relevant as it showcases to policy makers and other concerned stakeholders the underlying limitations to utilizing SynBio as an engine to revitalizing Kenya's bio-economy, and consequently assert herself as a regional SynBio powerhouse. Such include political economy challenges emanating from an almost fully donor-funded approach which permeates current biotechnology development in Kenya.

Stakeholder Involvement in the Governance of Human Genome Editing in Japan

Article

Tatsuki Aikyo, Atsushi Kogetsu & Kazuto Kato

Asian Bioethics Review (2023), 26 April 2023

Abstract

Genome editing is a technology that can accurately and efficiently modify the genome of organisms, including the human genome. Although human genome editing (HGE) has many benefits, it also involves technical risks and ethical, legal, and social issues. Thus, the pros and cons of using this technology have been actively debated since 2015. Notably, the research community has taken an interest in the issue and has discussed it internationally. However, for the governance of HGE, the roles of government agencies and the general public are also important for an effective regulatory system. Here, we examine the roles of the research community, government, and public in the governance of HGE through an analysis of discussions in the Japanese Expert Panel on Bioethics. During the discussion of the research ethics review system, the professionalism of the research community and the pros and cons of state oversight have become issues for debate.

Furthermore, through an examination of the overall policy-making process, three stakeholders are clearly involved in the governance of emerging medical technologies in the Expert Panel on Bioethics, a discussion forum established by government agencies. The contrast among these roles provides insight into the positive roles of government agencies and the research community and the conditions under which these roles are played. We also note that there are diverse actors in the public, which may have an impact on their participation. Our results may serve as a guide for countries and organizations to establish governance on emerging medical technologies

Governance of Heritable Human Genome Editing: Developing a Regulatory Framework for a Transformative Technology

Report

Natacha Tang

University of Zurich- University Research Priority Program (URPP) Human Reproduction Reloaded H2R Working Paper (1), 2023, 1–44, 15 March 2023

Abstract

With the discovery of CRISPR and its potential uses in genome editing of the human germline, the end to hereditary diseases and infertility seems closer than ever before. However, as is always the case with emerging transformative technologies, there are caveats. This paper seeks to explore possibilities and pitfalls in the regulation of and policymaking around heritable human genome editing, with a focus on the international (bioethics) law and human rights law perspective.

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GENETIC TESTING AND SCREENING

"Overestimated technology–underestimated consequences" – reflections on risks, ethical conflicts, and social disparities in the handling of non-invasive prenatal tests (NIPTs)

Article

Marion Baldus

Medicine, Health Care and Philosophy (2023), 18 March 2023

Open access

Abstract

New technologies create new complexities. Since non-invasive prenatal tests (NIPTs) were first introduced, keeping pace with complexity constitutes an ongoing task for medical societies, politics, and practice. NIPTs analyse the chromosomes of the fetus from a small blood sample. Initially, NIPTs were targeted at detecting trisomy 21 (Down syndrome): meanwhile there are sequencing techniques capable of analysing the entire genome of the unborn child. These yield findings of unclear relevance for the child's future life, resulting in new responsibility structures and dilemmas for the parents-to-be.

The industry's marketing strategies overemphasize the benefits of the tests while disregarding their consequences. This paper chooses the opposite path: starting with the underestimated consequences, it focuses on adverse developments and downsides. Disparities, paradoxes, and risks associated with NIPTs are illustrated, ethical conflicts described. Indications that new technologies developed to solve problems create new ones are examined.

In the sense of critical thinking, seemingly robust knowledge is scrutinized for uncertainties and ambiguities. It analyses how the interplay between genetic knowledge and social discourse results in new dimensions of responsibility not only for parents-to-be, but also for decision-makers, authorities, and professional societies, illustrated by a review of different national policies and implementation programmes. As shown by the new NIPT policy in Norway, the consequences can be startling. Finally, a lawsuit in the United States illustrates how an agency can risk forfeiting its legitimation in connection with the inaccuracy of NIPTs.

The clinical application of polygenic risk scores: A points to consider statement of the American College of Medical Genetics and Genomics (ACMG)

ACMG Statement

Aya Abu-El-Haija, Honey V. Reddi, Hannah Wand, Nancy C. Rose, Mari Mori, Emily Qian, Michael F. Murray on behalf of the ACMG Professional Practice and Guidelines Committee

Genetics in Medicine, Volume 25, Issue 5, 100803, 03 May 2023

Introduction

This document offers guidance to the health care provider who seeks to understand the challenges and limitations of applying PRS testing in patient care. The ACMG has developed this Points to Consider document to address the potential value of PRS given the limited evidence-base for clinical utility. Table 1 outlines the general considerations for the clinical application of PRS. An accompanying ACMG Points to Consider document addresses considerations for the development, implementation, and reporting of PRS from a laboratory perspective. A third ACMG document addresses the issues related to prenatal clinical applications of PRS testing.

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, Volume 25, Issue 5, 100017, 03 May 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.

Decision-Making About Newborn Screening Panels in Canada: Risk Management and Public Participation

Chapter

Marisa Beck, Brendan Frank, Sara Minaeian & Stuart G. Nicholls

Democratizing Risk Governance, pp 217–243, 29 April 2023 [Palgrave Macmillan]

Open access

Abstract

Newborn Bloodspot Screening (NBS) enables diagnosis and early treatment of rare diseases in non-symptomatic neonates. NBS has well-documented benefits for babies, their families, and the healthcare system at large. In recent decades, rapid advances in screening technologies enabled the proliferation of testable diseases. This has led to increased discussion of both the benefits relevant to decision-making but also the health, economic and ethical challenges associated with the expansion of NBS panels.

However, technological capability is not the sole driver of panel expansions, and we suggest that decisions to add a condition to the screening panel constitute exercises in risk management. Using a risk governance lens, this chapter examines procedures that govern decision-making concerning screening panel additions in several Canadian NBS programs. Specifically, we draw on an analysis of documents in the public domain and interviews with individuals associated with Canadian NBS programs to identify the risk management tools that are applied.

Our analysis indicates that there is a reliance on the advice of experts and economic controls but limited public participation in decisions about screening panels. We conclude with a discussion of why democratization might strengthen decision-making and offer recommendations to practitioners and scholars regarding next steps and future research.

The influence of Expanded Carrier Screening in Assisted Reproductive Techniques: Changed the "game"? Review.

Article

Petros Drettas, Vasileios Tatanis, Chara Spiliopoulou, Georgios Adonakis, Evangelos Liatsikos

Annals of Medicine & Surgery, Published Ahead of Print, 2023

Abstract

Expanded carrier screening constitutes a new scientific tool able to detect conditions that can be treated immediately after birth or during pregnancy. Its implementation could affect both the prenatal period and assisted reproductive techniques. It is strongly beneficial as provides much useful information to future parents concerning the medical status of their offspring. In addition, the definition of “serious/severe”, regulating preimplantation diagnosis, donor insemination, and even the definitions of prerequisites for abortion diseases, should be reformed including all clinically severe diseases. On the other hand, controversies may arise especially regarding gamete donation. Future parents and offspring may be informed regarding donors' demographic and medical characteristics. This study aims to investigate the effects of the implementation of expanded carrier screening in the reformation of the definition of “severe/serious” disease, the decision-making of future parents, gamete donation and the possible new moral dilemmas that may arise.

Polygenic Scores in the Direct-to-Consumer Setting: Challenges and Opportunities for a New Era in Consumer Genetic Testing

Review

Jin K. Park & Christine Y. Lu

Journal of Personalized Medicine, 223, 13(4),573, 23 March 2023

Open access

Abstract

Direct-to-consumer (DTC) genetic tests have generated considerable scholarly attention and public intrigue. Although the current consumer genetic testing regime relies on the reporting of individual variants of interest to consumers, there has recently been interest in the possibility of integrating polygenic scores (PGS), which aggregate genetic liability for disease across the entire genome. While PGS have thus far been extensively explored as clinical and public health tools, the use of PGS in consumer genetic testing has not yet received systematic attention, even though they are already in use for some consumer genetic tests. In this narrative review, we highlight the ethical, legal, and social implications of the use of PGS in DTC genetic tests and synthesize existing solutions to these concerns. We organize these concerns into three domains: (1) industry variation; (2) privacy and commercialization; and (3) patient safety and risk. While previously expressed concerns in these domains will remain relevant, the emergence of PGS-based DTC genetic tests raises challenges that will require novel approaches.

Genomic-Based Newborn Screening for Inborn Errors of Immunity: Practical and Ethical Considerations

Opinion

Jovanka R. King, Kalle Grill and Lennart Hammarström

International Journal of Neonatal Screening 2023, 9(2), 22, 11 April 2023

Open access

Abstract

Inborn errors of immunity (IEI) are a group of over 450 genetically distinct conditions associated with significant morbidity and mortality, for which early diagnosis and treatment improve outcomes. Newborn screening for severe combined immunodeficiency (SCID) is currently underway in several countries, utilising a DNA-based technique to quantify T cell receptor excision circles (TREC) and kappa-deleting recombination excision circles (KREC). This strategy will only identify those infants with an IEI associated with T and/or B cell lymphopenia. Other severe forms of IEI will not be detected. Up-front, first-tier genomic-based newborn screening has been proposed as a potential approach by which to concurrently screen infants for hundreds of monogenic diseases at birth. Given the clinical, phenotypic and genetic heterogeneity of IEI, a next-generation sequencing-based newborn screening approach would be suitable. There are, however, several ethical, legal and social issues which must be evaluated in detail prior to adopting a genomic-based newborn screening approach, and these are discussed herein in the context of IEI.

Never “totally prepared”: Support groups on helping families prepare for a child with a genetic condition

Research Article

Kaitlynn P. Craig, Kirsten A. Riggan, Sabina Rubeck, Stephanie H. Meredith, Megan A. Allyse & Marsha Michie

Journal of Community Genetics (2023), 12 April 2023

Abstract

A rapid increase in the reach and breadth of prenatal genetic screening and testing has led to an expanding need for prenatal support of families receiving this genetic information. As part of a larger study investigating prenatal preparation for a child with a genetic condition, we interviewed representatives of patient advocacy groups (PAGs) who support parents post-diagnosis.

Groups supporting families with Down syndrome were often local or regional, while other groups were often national or international in scope. Groups varied in their willingness or ability to support families prior to making a pregnancy continuation decision, and participants reflected on ways they addressed these needs with individual counseling and referrals, if needed. Participants described supporting parents with information about conditions and a range of lived experiences for families, while referring families to healthcare professionals for technical questions and additional medical needs. PAGs also prioritized connecting parents experiencing a new diagnosis with other families for peer support and community-building, both in person and on social media. Participants discussed limitations, such as a lack of racially-concordant support, ability to offer resources in languages other than English, and a lack of funding to meet the expressed needs of families post-diagnosis. Overall, participants emphasized that the parenting experience of each child is unique, irrespective of a genetic diagnosis, an experience for which parents can never be “totally prepared.”

The Ethical Challenges of Newborn Screening Programs in the United States

Thesis

Devin Donovan

University of South Carolina – Columbia, Senior Thesis. 598. (2023)

Thesis Summary

Newborn screening programs have been mandated throughout the United States since the 1960s, and technological advancements have allowed for their evolution into the essential public health entities they are today (Arnold 558). These programs screen newborns for a variety of congenital and genetic conditions in all states, but each state varies in conditions screened and policies for collecting and using samples. Residual blood spots are a key component of these programs because they are often used for secondary purposes, such as for quality assurance and public health or biomedical research (Botkin et al. 121). Ethical challenges have arisen related to consent and privacy policies employed by states. Legal cases have arisen surrounding the accessibility of these blood spots by outside entities, such as law enforcement agencies. Problems with informed consent, privacy protections, and transparency within many states’ programs call for the alteration of policies so that citizens’ autonomy and privacy are upheld and so that trust and support for these public health initiatives are sustained.

A Review of the Legislation of Direct-to-Consumer Genetic Testing in China

Article

Chen Yichao, Li Wei, and Chen Jiajv

Human Gene Therapy, Ahead of Print, 18 April 2023

Open access

Abstract

Direct-to-consumer genetic test (DTC-GT) has been developing into an enormous market in China. Though no existing laws are directly applicable to DTC-GT, relevant laws and regulations are gradually being improved. In this study, we discuss how China's legislative and juridical practices in the field of DTC-GT have led to it being strictly constrained. The continuous improvement of relevant private and public laws is increasingly strengthening the informed consent and data protection issues that are involved with DTC-GT.

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PLANTS, CROPS, AGRICULTURE, ANIMALS

International agreements and the plant genetics research community: A guide to practice

Perspective

Emily Marden, Ruaraidh Sackville Hamilton, Michael Halewood, and Susan McCouch

PNAS Vol. 120 | No. 14 e2205773119, March 27, 2023

Abstract

Plant genetic resources (PGR), including collections held in national and international gene banks, provide access to a wide array of genetic diversity and are critical to genomics research, conservation efforts, and applied breeding. Yet, there is a general lack of awareness in the research community about the rules and treaties that govern the use of PGR, about access and benefit sharing obligations contained in international treaties and/or national laws, and about how best to comply with potentially applicable requirements. This article provides a brief history and overview of three key international agreements, namely the Convention on Biological Diversity, the Nagoya Protocol, and the International Treaty on Plant Genetic Resources for Food and Agriculture, which collectively address responsibilities and obligations related to the use of much of the world's PGR. By highlighting the coverage and key considerations of each agreement, the article provides a guide for those who use PGR in plant genetics research to better understand when and how international agreements apply, and—where the rules are unclear—to suggest best practices for compliance with existing agreements.

Our shared global responsibility: Safeguarding crop diversity for future generations

Perspective

Hannes Dempewolf, Sarada Krishnan, and Luigi Guarino

PNAS Vol. 120 | No. 14 e2205768119, March 27, 2023

Abstract

The resilience and sustainability of food systems depend on crop diversity. It is used by breeders to produce new and better varieties, and by farmers to respond to new challenges or demands and to spread risk. However, crop diversity can only be used if it has been conserved, can be identified as the solution for a given problem, and is available. As the ways in which crop diversity is used in research and breeding change and expand, the global conservation system for crop diversity must keep pace; it must provide not only the biological materials themselves, but also the relevant information presented in a comprehensive and coherent way—all while ensuring equitable access and benefit sharing. Here we explore the evolving priorities for global efforts to safeguard and make available the diversity of the world's crops through ex situ genetic resource collections. We suggest that collections held by academic institutions and other holders that are not standard gene banks should be better integrated in global efforts and decision-making to conserve genetic resources. We conclude with key actions that we suggest should be taken to ensure that crop diversity collections of all types are able to fulfill their role to foster more diverse, equitable, resilient, and sustainable food systems globally.

Customizing ethical tools for Malaysian farmers: a case for GM crops technology

Conference Paper

H Omar, S H Idris, I M Nashir, S. Jayabalan, A B A Majeed, L Amin and H Omar

IOP Conference Series: Earth and Environmental Science, Volume 1151, The 1st 2022 International Conference on Sustainable Environment, Development, and Energy (CONSER 2022) 05/12/2022 - 06/12/2022 Denpasar, Bali, Indonesia

Open access

Abstract

The socio-economic and cultural factors are rarely at the centre stage in the discourse on genetically modified organisms (GMOs) compared to the scientific and technological aspects. This study described the indicators for the ethical principles of genetically modified (GM) crops that can protect the rights of Malaysian farmers. Respect for life and the need to balance the benefits and harms are the indicators of general principles of ethics. It is vital for a decision to comprise of both scientific evidence and ethical consideration. Ethics focuses on what we should or should not do and locality-based. Therefore, ethical principles must be part of the evaluation criteria of policy practices. They must also be custom-made to suit Malaysian farmers. Ethical decision-making regarding genetic modification is complicated since it encompasses many ethical aspects of our lives. This paper aims to analyze and customize the indicators of ethical principles and guidelines on using GM crops in Malaysia to protect Malaysian farmers. A meta-analysis exploration comprised of systematic review of established research within GM crops' agronomic, socio-economic, cultural, and environmental effects is employed to consolidate the evidence. The outcome is the formation of an ethical tool comprised of nine indicators that suits farmers in Malaysia.

Ethical tools of genetically modified (GM) crops technology for farmers' protections

Conference paper

Siti Hafsyah Idris, Habibah Omar, Irdayanti Mat Nashir

International Conference on Mathematical and Statistical Physics, Computational Science, Education, and Communication (ICMSCE 2022); 126160T (2023), 10 April 2023, 8-9 December 2022, Istanbul Turkey

Abstract

Genetically modified organisms (GMOs) created through genetic engineering have dominated the headlines since their initial scientific discovery. Humanity continues to be in awe of how biotechnological progress can help ensure human survival, primarily through food security. However, GMOs' future benefits have rarely been enough to outweigh the ethical and socio-economic concerns. These elements were not previously thought to be equally significant. This study aims to identify possible methods to incorporate ethical considerations into the decision-making process of GM technology for the protection of farmers' rights and to present indicators to aid policymakers in assessing the ethical issues in GM technology. Indicators for the ethical tools of genetically modified (GM) crops that can protect the rights of Malaysian farmers were described in this study. General ethical principles can be seen in the reverence for life and the requirement to weigh benefits and harms. Therefore, a decision-making process must consider ethical issues and scientific evidence. The results of this study revealed that despite this, farmers' rights to a living and contractual justice had been ethically ignored. This is because ethical principles are typically descriptive and challenging to implement. Therefore, its ethical implications must be carefully considered to ensure that the use of GM crops technology does not violate the fundamental rights of farmers. Hence, additional research is needed to hone and broaden this framework to ensure sustainable modern biotechnology and the protection of farmers' rights. The study's output will be a legal framework to evaluate the ethical implications of GM crops in preserving farmers' rights. Government regulators and other pertinent stakeholders can use the recommendations.

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