ge²p² global foundation

governance, ethics, evidence, policy, practice

Center for Genomic Medicine Ethics & Policy

Genomics: Governance, Ethics, Policy, Practice – A Monthly Digest

January 2025 – Number 11

Genomics – spanning discovery, preclinical, clinical and translation to daily patient interventions – continues to evolve at an extraordinary pace. Advances in the scientific and technical dimensions of genomics overall are extensively communicated through the peer-reviewed journal literature and supporting grey literature.

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

Further, we intend this digest to provide a useful summary of key strategic and programmatic announcements from across genomics as issued by multilateral agencies, INGOs, governments/regulatory bodies, academic and research institutions, consortiums and collaborations, foundations, investors, and commercial organizations.

Given the complexity and velocity of the field, we are striving to make this digest comprehensive – but we acknowledge it is not exhaustive. We invite suggestions and ideas on how it can evolve to be more useful.

The digest is a program of the GE2P2 Global Foundation which is solely responsible for its content. Questions and comments should be directed to the Editors as below:

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HPE – Heritable Polygenic Editing

We lead this edition with an important emerging debate triggered by Nature's decision to publish the analysis by Wisscher et al. [just below] on heritable polygenic editing [HPE]. We include further below an excerpt from the accompanying Nature Editorial [which seems to anticipate the debate now emerging].

We also include the analysis of the paper by Carmi et al. in the same issue of Nature which observes: "…Although the authors' claims are logical and thought-provoking, their model relies on several speculative assumptions and glosses over unknown, but predictable, serious risks. Given the broad interest in this topic, the work will probably be discussed widely and might ultimately affect policy. It raises both scientific and ethical issues.." Indeed it does.

Shortly after the Nature issue was released, ARRIGE issued this statement – Heritable Polygenic Editing, January 2025 – which vigorously argues against the paper's integrity noting "… In this contemporary era of fake news and alternative truth, such a publication is not only a provocation, it will also feed distrust in scientific activities and remains highly irresponsible."

We will monitor and report on the ensuing conversation!

[Note: The GE2P2 Global Foundation is an institutional member of ARRIGE and Co-Editor David Curry serves on its Board]

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Heritable polygenic editing: the next frontier in genomic medicine?

Peter M. Visscher, Christopher Gyngell, Loic Yengo & Julian Savulescu

Analysis Open access Published: 08 January 2025
Nature, Volume 637 Issue 8045, 9 January 2025

Abstract

Polygenic genome editing in human embryos and germ cells is predicted to become feasible in the next three decades. Several recent books and academic papers have outlined the ethical concerns raised by germline genome editing and the opportunities that it may present1,2,3. To date, no attempts have been made to predict the consequences of altering specific variants associated with polygenic diseases. In this Analysis, we show that polygenic genome editing could theoretically yield extreme reductions in disease susceptibility. For example, editing a relatively small number of genomic variants could make a substantial difference to an individual's risk of developing coronary artery disease, Alzheimer's disease, major depressive disorder, diabetes and schizophrenia. Similarly, large changes in risk factors, such as low-density lipoprotein cholesterol and blood pressure, could, in theory, be achieved by polygenic editing. Although heritable polygenic editing (HPE) is still speculative, we completed calculations to discuss the underlying ethical issues. Our modelling demonstrates how the putatively positive consequences of gene editing at an individual level may deepen health inequalities. Further, as single or multiple gene variants can increase the risk of some diseases while decreasing that of others, HPE raises ethical challenges related to pleiotropy and genetic diversity. We conclude by arguing for a collectivist perspective on the ethical issues raised by HPE, which accounts for its effects on individuals, their families, communities and society4.

…

Concluding remarks

Advances in technology21 have already led to the birth of at least two genetically edited children and children screened (before birth) for polygenic conditions. Over the coming decades, it may become possible to make multiple edits to the DNA sequence of human embryos and germ cells, potentially targeting dozens to hundreds of variants involved in the development of complex traits. In this

Analysis, we demonstrated that editing a relatively small number of variants could make dramatic changes to an individual's risk of disease, and if widely and safely used, it may substantially reduce the incidence of polygenic diseases among those with edited genomes.

From the modelling results, it appears that editing only a few variants would maximize benefit and minimize risk, so that an 'oligogenic' approach may be preferred to an approach with many loci. However, there are still too many unknowns to draw such a strong conclusion. For example, we do not know how gene editing technologies will develop with respect to precision (that is, risk of deleterious off-target effects) in the next 30 years. There may be diseases and their risk factors that do not have rare protective variants with large effects, so that change may only be achieved by editing many loci. Rare large-effect variants may also show deleterious epistatic effects when homozygous.

Gene editing techniques applied to non-disease traits may deepen inequalities and raise the spectre of eugenics. It is vital for governments and the international community to carefully consider how to regulate HPE to best manage the ethical challenges. In doing so, it is important to consider the risk of deciding not to use HPE. Polygenic diseases are a leading cause of premature death worldwide, strain the health system and reduce people's freedom by making them reliant on medical resources. Successful management of the risks posed by HPE will likely require strong international cooperation, which is particularly challenging in the face of globally competing interests, priorities and conflicting values. There is good reason to start exploring the challenges and opportunities that HPE provides now, well before it becomes a practical possibility, and our modelling serves as a foundation for an informed and balanced discussion on the potential use of gene editing to reduce the genomic contribution to common diseases or traits

Nature Editorial

We need to talk about human genome editing

08 Jan 2025

Nature, Volume 637 Issue 8045, 9 January 2025

In a few decades, gene-editing technologies could reduce the likelihood of common human diseases. Societies must use this time to prepare for their arrival.

"…The study reveals that polygenic genome editing in human embryos could substantially reduce the likelihood of certain diseases occurring, but it raises concerns, not least the renewed threat of eugenics. There are other caveats too, the researchers report. *Nature* is publishing this work because it is important to start a conversation about what could happen if more-sophisticated gene-editing technologies become available, which could be the case within 30 years, the authors say. Societies need to consider relevant benefits and risks before that day comes…"

Human embryo editing against disease is unsafe and unproven — despite rosy predictions

Mathematical modelling suggests that it is theoretically possible to reduce risk of common diseases using heritable genome editing. Scientists argue that the technology involves considerable risk and uncertain benefits.

By Shai Carmi, Henry T. Greely & Kevin J. Mitchell

NEWS AND VIEWS 08 January 2025

Nature, Volume 637 Issue 8045, 9 January 2025

The possibility of editing the genomes of human embryos has been widely discussed, particularly since the discovery of the CRISPR–Cas9 gene-editing tool. The revelation in late 2018 that a Chinese scientist, He Jiankui, had edited embryos that became living babies created a huge wave of controversy[1]. Writing in *Nature*, Visscher *et al.*[2] describe a mathematical model that argues that just a handful of edits could reduce the risk of various disorders dramatically — in a theoretical scenario in which heritable, large-scale genome editing is feasible and safe. Although the authors' claims are logical and thought-provoking, their model relies on several speculative assumptions and glosses over unknown, but predictable, serious risks. Given the broad interest in this topic, the work will probably

be discussed widely and might ultimately affect policy. It raises both scientific and ethical issues, which we discuss here.

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Genomics and Justice

We continue to review and reflect on the important Hastings Center Report [Vol 54, Issue S2] which presents 14 essays and other analyses. We include the Introduction and two examples of essay content below. We urge readers to review the full Report!

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Expanding the Agenda for a More Just Genomics

Hastings Center Report, Volume 54, Issue S2 December 2024 Published January 2, 2025
https://onlinelibrary.wiley.com/doi/10.1002/hast.4924
Issue Edited by: Josephine Johnston, Deanne Dunbar Dolan, Danielle M. Pacia, Sandra Soo-Jin Lee, Mildred K. Cho

This Report – "Envisioning a More Just Genomics" is a collaboration between The Hastings Center, a bioethics research institute, and the Center for ELSI Resources and Analysis (CERA), which focuses on the ethical, legal, and social implications of genetics and genomics. It was funded by the National Human Genome Research Institute.

Abstract

The integration of genomics into public health and medicine is happening at a faster rate than the accrual of the capabilities necessary to ensure the equitable, global distribution of its clinical benefits. Uneven access to genetic testing and follow-up care, unequal distribution of the resources required to access and participate in research, and underrepresentation of some descent groups in genetic and clinical datasets (and thus uncertain genetic results for some patients) are just some of the reasons to center justice in genomics. A more just genomics is an imperative rooted in the ethical obligations incurred by a publicly funded science that is reliant on human data. These features of genomics indebt the genomics enterprise and compel the expanded scope of responsibility proposed by the authors of this special report. The report begins to define justice in genomics for different stakeholder groups and proposes substantial shifts in power, resource distribution, scientific practice, and governance that could enable genomics to meet its obligations to humanity.

[Excerpt from concluding section]

A New Agenda

Massive public investment in genomics research obligates everyone involved in the enterprise to reflect on how to equitably distribute its benefit and mitigate the risk of further exacerbating inequities. A more just genomics requires substantive shifts of power and resources from the genomics research enterprise and its agents in government and industry toward initiatives that empower research participants and communities and build capacity for underresourced institutions in the United States and globally. A just genomics should advance global health equity,[64] not widen the health gap between haves and have-nots. Power sharing is an essential demonstration of dedication to a more just genomics, alongside substantial investments in infrastructure and processes for equity monitoring. To center justice in genomics, these investments must be grounded in the moral responsibility to address and rectify inequity…

The imperative to center justice in genomics requires creating a robust and equitable system to address and mitigate injustices that result from the concentration of power manifested in racialized thinking, inequitable access to both genomics research and its clinical applications, and structures that create barriers to benefit. Fundamentally, this imperative is rooted in the ethical obligations inherent to publicly funded scientific endeavors and the debt the genomics enterprise owes to the public sphere to ensure all members of our shared human community have the right to partake in the benefits derived from genomic advancements.

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Genomics and Biodiversity: Applications and Ethical Considerations for Climate-Just Conservation

Research

Skye A. Miner, Timothy J. Thurman

Posted on rand.org Dec 26, 2024 Published in: Hastings Center Report, Volume 54, Issue S2 (Supplement: Envisioning a More Just Genomics, December 2024), Pages S114-S119. DOI: 10.1002/hast.4936

Genomics holds significant potential for conservationists, offering tools to monitor species risks, enhance conservation strategies, envision biodiverse futures, and advance climate justice. However, integrating genomics into conservation requires careful consideration of its impacts on biodiversity, the diversity of scientific researchers, and governance strategies for data usage. These factors must be balanced with the varied interests of affected communities and environmental concerns. We argue that conservationists should engage with diverse communities, particularly those historically marginalized and most vulnerable to climate change. This inclusive approach can ensure that genomic technologies are applied ethically and effectively, aligning conservation efforts with broader social and environmental justice goals. Engaging diverse stakeholders will help guide responsible genomic integration, fostering equitable and sustainable conservation outcomes.

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The UN Declaration on the Rights of Indigenous People and Genomics: Ethical Complementarity for Just Research

Expanding the Agenda

Ibrahim Garba, Stephanie Russo Carroll

Hastings Center Report, Volume 54, Issue S2 December 2024 Published January 2, 2025 S120–S125. DOI: 10.1002/hast.4937

Abstract

Governance of biomedical research in the United States has been characterized by ethical individualism, a mode of reasoning that treats the individual person as the center of moral concern and analysis. However, genomics research raises ethics issues that uniquely affect certain genetically related communities as collectives, not merely as aggregates of individuals. This is especially true of identifiable populations—including Indigenous Peoples—that are often minoritized, socially marginalized, or geographically isolated. We propose an alternative, complementary framework based on the United Nations Declaration on the Rights of Indigenous Peoples (UNDRIP) (2007), which explicitly recognizes both individual and collective rights. We use the CARE Principles for Indigenous Data Governance as a case study to show how this UNDRIP-based framework can complement the individual-focused national standard for research oversight represented by the *Belmont* principles, thereby better protecting Indigenous Peoples' rights and interests in genomic data.

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Governance and Oversight Frameworks – Genomics, Emerging Technologies

We present below highlights from two reports developed in tandem around "oversight frameworks" for emerging technologies, commissioned by Wellcome and developed by Rand Europe.

We assess these reports to make a significant contribution to analysis of the complex governance challenges involved across the intriguing span of emerging technologies addressed – Genomics [engineering biology], Organoids, Human Embryology, Nanotechnology and AI Research Tools/Data Platforms.

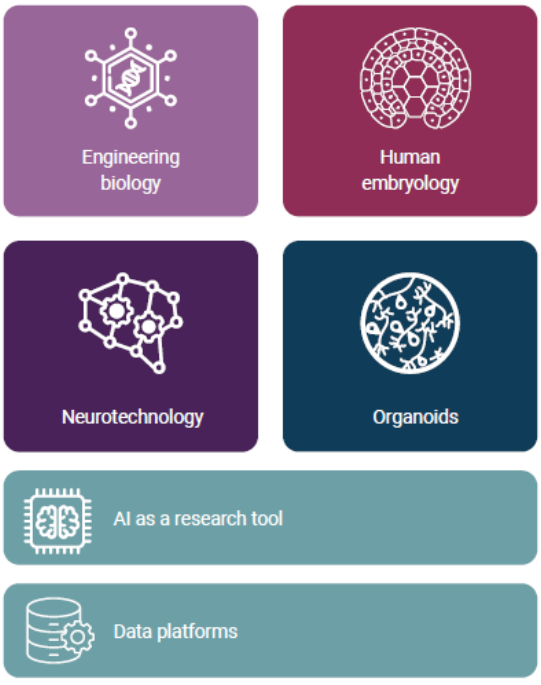
We recommend that readers review and act on these important reports. More reflection in our next edition.

State-of-play and future trends on the development of oversight frameworks for emerging technologies: Part 1: Global landscape review of emerging technology areas
Rand *Report* Dec 16, 2024
Sana Zakaria, Ioli Howard, Eva Coringrato, Anna Louise Todsen, Imogen Wade, Devika Kapoor, Alec Ross, Katarina Pisani, Chryssa Politi, Martin Szomszor, Salil Gunashekar

…**Wellcome commissioned RAND Europe to undertake a study on the state-of-play and future trends on the development of oversight frameworks for emerging technologies**. The specific objective of the study is to identify and analyse a suite of oversight frameworks and mechanisms (including associated emerging trends and novel approaches) that are in use, in development or under debate in different jurisdictions across the globe for a set of emerging technologies.

The technologies of interest include genomics (specifically engineering biology), human embryology, organoids, neurotechnology, artificial intelligence (AI) (specifically its application and use as a research tool) and data platforms.

Figure 1. Technology areas covered by this study



Source: RAND Europe analysis.

The study findings are presented in two related documents: the global technology landscape review report and the technology oversight report (this report). The two reports should be read alongside each other. This report examines notable oversight mechanisms that are either established or under development across a selection of global jurisdictions, offering key learning and insights that could inform future technology oversight discussions.

Key Findings

Diverse range of oversight frameworks

For each technology, the study maps and examines a variety of oversight frameworks across multiple jurisdictions – covering a spectrum of options with differing levels of accountability, obligation and enforcement – to assess how technology is being used, while keeping issues such as safety, privacy and risk mitigation at the forefront.

Lack of specific frameworks for organoids

There is an absence of specific regulatory frameworks for organoids, with current oversight relying on broader stem cell and biomedical regulations. Emerging mechanisms, such as Japan's consent-to-govern approach, are gaining traction to address ethical challenges, particularly around donor consent and privacy concerns.

Challenges in human embryology oversight

Existing frameworks, like the UK's Human Fertilisation and Embryology Act, are outdated and not designed for new technologies such as AI in embryo selection. Disparate national regulations complicate international collaboration, and there is a need for frameworks to adapt to scientific advancements and public interest.

Fragmented oversight in engineering biology

The global landscape features disparate oversight mechanisms, creating obstacles for international collaboration. There is a need for alignment across diverse applications and jurisdictions, with potential solutions including cross-sector collaboration and international biosecurity measures.

Neurotechnology oversight gaps

Current regulations do not address the unique challenges posed by neurotechnologies, such as data privacy and dual-use concerns. Ethical guidelines, like Chile's neurorights, offer proactive models, but there is a need for stronger post-market surveillance and international guidelines to prevent misuse.

Figure 2. Spectrum of oversight approaches

Source: RAND Europe analysis.

Recommendation

This report proposes eight priority considerations for stakeholders engaged in technology R&I, to support the development of the broader R&I and technology oversight ecosystem in the future. These cross-cutting actions cover topics such as

[i] developing networks of interconnected oversight mechanisms;
(ii) ensuring equity considerations are prioritised;
(iii) establishing actionable international alignment to harmonise governance practices;
(iv) developing internationally coordinated risk mitigation strategies;
(v) supporting the implementation and scaling of innovative oversight mechanisms;
(vi) facilitating proactive public involvement in the development of oversight frameworks;
(vii) incorporating adaptive practices into oversight processes; and
(viii) integrating anticipatory strategies into oversight frameworks.

[Excerpt]

Key takeaways: Genomics

Genomics (focusing on engineering biology) is a field of biology that focuses on the study of an organism's complete set of DNA. Engineering biology applies the tools and techniques of engineering to biology, enabling novel biological system design, or redesign of existing systems.

Exemplar trends: There has been a rapid growth of engineering biology infrastructure, research and applications that span biomanufacturing, net-zero and climate mitigation, and agriculture security.

Exemplar opportunities: Innovations in healthcare, agriculture and industrial biotechnology are leading to sustainable solutions and new bio-based products.

Exemplar challenges: These include biosafety concerns given the dual-use nature of biological tools and outputs, ethical implications of synthetic organisms, and public acceptance.

Exemplar oversight mechanisms: Given its varied applications, diverse policies, laws and frameworks govern this field, such as biosafety standards and public contracts, as well as international conventions such as the Biological Weapons Convention (BWC).

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State-of-play and future trends on the development of oversight frameworks for emerging technologies Part 2: Technology oversight report

Sana Zakaria, Ioli Howard, Eva Coringrato, Anna Louise Todsen, Imogen Wade, Devika Kapoor, Alec Ross, Katarina Pisani, Chryssa Politi, Martin Szomszor, Salil Gunashekar

Research Published Dec 16, 2024

…. **Part 2 highlights the challenges and opportunities in regulating these technologies, emphasising the need for updated frameworks that address ethical, privacy, and collaboration issues**. In Part 2, we use a mixed-methods approach, including desk research, interviews, SWOT analysis and expert elicitation, to examine existing and developing oversight mechanisms. We provide insights into legislative and non-regulatory standards, ethical guidelines and self-regulatory frameworks relevant to key debates on oversight of emerging technologies, including on the lack of specific regulatory frameworks for organoids, ethical challenges in human embryology, fragmented oversight in engineering biology, and privacy concerns in neurotechnology. The study also discusses the potential for dual-use scenarios in neurotechnology and the need for international collaboration in managing biosecurity threats in engineering biology.

The report proposes eight priority considerations for future oversight, emphasising the importance of equity, international alignment, risk mitigation, public involvement and adaptive practices. These considerations aim to enhance the global oversight ecosystem and ensure ethical and beneficial applications of emerging technologies. The study underscores the importance of developing comprehensive oversight frameworks that can adapt to rapid technological advancements and address global challenges.

Key Findings

Lack of specific frameworks for organoids

There is an absence of specific regulatory frameworks for organoids, with current oversight relying on broader stem cell and biomedical regulations. Emerging mechanisms, such as Japan's consent-to-govern approach, are gaining traction to address ethical challenges, particularly around donor consent and privacy concerns.

Challenges in human embryology oversight

Existing frameworks, like the UK's Human Fertilisation and Embryology Act, are outdated and not designed for new technologies such as AI in embryo selection. Disparate national regulations complicate international collaboration, and there is a need for frameworks to adapt to scientific advancements and public interest.

Fragmented oversight in engineering biology [Genomics]

The global landscape features disparate oversight mechanisms, creating obstacles for international collaboration. There is a need for alignment across diverse applications and jurisdictions, with potential solutions including cross-sector collaboration and international biosecurity measures.

Neurotechnology oversight gaps

Current regulations do not address the unique challenges posed by neurotechnologies, such as data privacy and dual-use concerns. Ethical guidelines, like Chile's neurorights, offer proactive models, but there is a need for stronger post-market surveillance and international guidelines to prevent misuse.

Priority considerations for future oversight

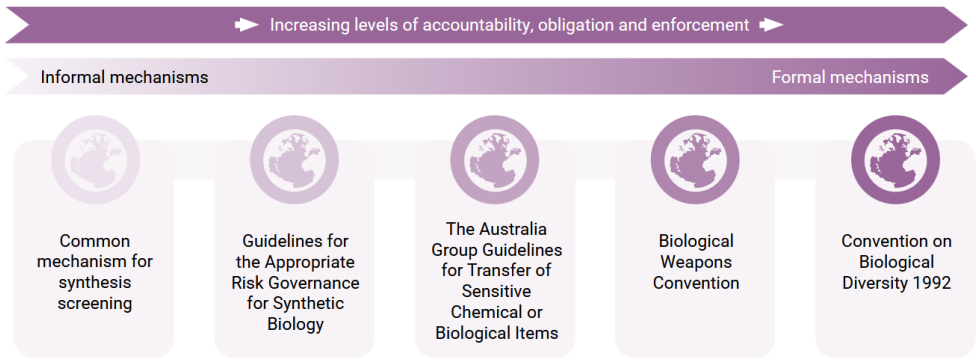
The report outlines eight priority considerations, including developing interconnected oversight networks, ensuring equity, harmonizing international governance, fostering public involvement, and integrating adaptive and anticipatory strategies into oversight frameworks.

Recommendations

- Develop comprehensive process maps and establish networks of interconnected oversight mechanisms to support stakeholders in effectively navigating the labyrinth of relevant mechanisms in the technology oversight landscape.
- Ensure that equity considerations are prioritised and integrated into all aspects of technology oversight to promote fairness and inclusivity.
- Identify and establish common ground for practical and actionable international alignment to harmonise governance practices across borders.
- Intensify efforts to develop internationally coordinated risk mitigation strategies as part of implementing oversight mechanisms to address global challenges posed by emerging technologies.
- Support the implementation and scaling of innovative oversight mechanisms to effectively manage the complexities and dynamics of emerging technologies.
- Facilitate proactive public involvement in the development of oversight frameworks to ensure transparency and accountability.
- Incorporate adaptive practices into oversight processes to foster continuous learning, flexibility and agility in response to technological advancements.
- Integrate anticipatory strategies into oversight frameworks to prepare for and address future developments in emerging technologies.

5.6. Oversight of engineering biology in international forums

Figure 16. Illustrative oversight examples of engineering biology in international forums



Source: RAND Europe analysis.

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Cell & Gene Therapy Sector: Regulatory Landscape/Market Dynamics

We include below the latest in the series of cell-gene "sector snapshots" published by the Alliance for Regenerative Medicine [ARM]. Our Foundation is a non-voting, nonprofit member of ARM.

December 2024 Sector Snapshot: This Year's Cell and Gene Thrapy Milestones
Alliance for Regenerative Medicine [ARM] December 2024 :: 8 pages
PDF: https://alliancerm.org/wp-content/uploads/2024/12/20241223-2024-Sector-Snapshot.pdf

This report recaps this year's milestones and provides an early picture of what could potentially be in store for the 2025 clinical pipeline of cell and gene therapies.
:: Review of approved cell and gene therapy products in the United States and European Union
:: A look at ARM's Q3 2024 Sector Data
:: 2024 milestones in clinical innovation, oncology, frontiers in disease treatment, and patient access
:: A look into the 2025 clinical pipeline

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FDA: Developing CGT Products – Draft Guidance for Industry/Public Comment Invited

The FDA has developed over 30 guidances for industry in the CGT space, most in the last several years and aggregated here: FDA Cellular & Gene Therapy Guidances. A new draft guidance as below is now available for public comment. Our Foundation will respond to this public comment opportunity

Frequently Asked Questions-Developing Potential Cellular and Gene Therapy Products; Draft Guidance for Industry; Availability
A Notice by the Food and Drug Administration on 11/19/2024 **Comment period ends 02/18/2025**
SUMMARY:
The Food and Drug Administration (FDA or Agency) is announcing the availability of a draft document entitled "Frequently asked Questions—Developing Potential Cellular and Gene Therapy Products." The **draft guidance document provides industry with answers to frequently asked questions**

(FAQs) and commonly faced issues that arise during the development of cellular and gene therapy (CGT) products. The FAQs represent common questions directed to the Agency and span multiple disciplines, including regulatory review; chemistry, manufacturing, and controls (CMC); pharmacology/toxicology; clinical; and clinical pharmacology.
PDF: https://www.fda.gov/media/183631/download
SUPPLEMENTARY INFORMATION:
I. Background
FDA is announcing the availability of a draft document entitled "Frequently asked Questions—Developing Potential Cellular and Gene Therapy Products." The draft guidance document provides industry with answers to FAQs and commonly faced issues that arise during the development of CGT products. The FAQs represent common questions directed to the Agency and span multiple disciplines, including regulatory review; CMC; pharmacology/toxicology; clinical; and clinical pharmacology.
The guidance was created as part of FDA's response to the PDUFA VII commitment to increase efficiency and to support development of CGT products by providing a repository of common questions posed to the Office of Therapeutic Products by sponsors and other key stakeholders. The Agency compiled FAQs received from a variety of sources, including FDA interactions with sponsors in development programs.
The guidance covers relevant, current, and timely topics related to the development of CGT products, which may be updated to include additional FAQs as appropriate. Sponsors are encouraged to visit the Cellular and Gene Therapy Guidances web page on the FDA website for a full list of finalized as well as draft guidances relevant to the development of CGT products…

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DSI and Benefits Sharing: Public Consultations [COP16]

In our December edition, we reported on key outcomes from COP16 around DSI benefits sharing and the establishment of the Cali Fund [see media release below]. Post COP16, the CBD secretariat has released two public consultations to support refinement and implementation details of these outcomes as below. Our Foundation intends to respond to these calls and invites interested individuals and institutions/organizations to advise us if interested in collaborating on a response.

The multilateral mechanism for the fair and equitable sharing of benefits from the use of digital sequence information on genetic resources, including a global fund ("The Cali Fund"): Submission of views on possible additional modalities of the multilateral mechanism [Public Consultation]

CBD Convention on Biological Diversity, Notification 2024-114 2024-12-10 **Comments no later than 21 March 2025**

As noted in notification 2024-113, at its sixteenth meeting, by decision 16/2, the Conference of the Parties adopted the modalities for operationalizing the multilateral mechanism for benefit-sharing from the use of digital sequence information on genetic resources, including the global fund, which are set out in the annex to the decision, and decided that the global fund will be known as the Cali Fund for the Fair and Equitable Sharing of Benefits from the Use of Digital Sequence Information on Genetic Resources. By the same decision, Parties also set out some intersessional work.
While the Conference of the Parties, in decision 16/2, adopted the modalities of the multilateral mechanism, it also decided (in paragraph 3 of the decision) to explore possible additional modalities, including, in the context of paragraph 7 of decision 15/9 and the annex to decision 16/2, to take products and services into account.
Parties, other Governments, indigenous peoples and local communities, and relevant organizations are invited to submit their views on this issue as soon as possible …

The multilateral mechanism for the fair and equitable sharing of benefits from the use of digital sequence information on genetic resources, including a global fund ("The Cali Fund"): Submission of views on possible new tools and models, such as databases, for making digital sequence information on genetic resources publicly available and accessible [Public Consultation]

CBD Convention on Biological Diversity, Notification 2024-115 2024-12-10 **Comments no later than 4 April 2025**

As noted in notification 2024-113, at its sixteenth meeting, by decision 16/2, the Conference of the Parties adopted the modalities for operationalizing the multilateral mechanism for benefit-sharing from the use of digital sequence information on genetic resources, including the global fund, which are set out in the annex to the decision, and decided that the global fund will be known as the Cali Fund for the Fair and Equitable Sharing of Benefits from the Use of Digital Sequence Information on Genetic Resources. By the same decision, Parties also set out some intersessional work.

In particular, the Conference of the Parties, in decision 16/2, decided to explore possible new tools and models, such as databases, for making digital sequence information on genetic resources publicly available and accessible in a transparent and accountable manner to all Parties.

Parties, other Governments, indigenous peoples and local communities, and relevant organizations are invited to submit their views on this issue as soon as possible…

Biodiversity COP 16: Important Agreement Reached Towards Goal of "Making Peace with Nature"

November 2, 2024, CBD Secretariat.

[Excerpt]

…Call Fund is Launched: Sharing the Benefits of Digital Genetic Information

Having agreed at COP 15 to establish a multilateral mechanism, including a global fund, to share the benefits from uses of digital sequence information on genetic resources (DSI) more fairly and equitably, delegates at COP 16 advanced its operationalization – a historic decision of global importance.

This complex decision addresses how pharmaceutical, biotechnology, animal and plant breeding and other industries benefiting from DSI should share those benefits with developing countries and Indigenous Peoples and local communities.

Under the agreed guidelines, large companies and other major entities benefiting commercially from DSI uses should **contribute to "the Cali Fund," based on a percentage of their profits or revenues**. The model targets larger companies most reliant on DSI and exempts academic, public research institutions and other entities using DSI but not directly benefiting.

Developing world countries will benefit from a large part of this fund, with allocations to support implementation of the KMGBF, according to the priorities of those governments.

At least half of the funding is expected to support the self-identified needs of Indigenous peoples and local communities, including women and youth within those communities, through government or by direct payments through institutions identified by Indigenous peoples and local communities. Some funds may support capacity building and technology transfer.

Strong monitoring and reporting will ensure industries see the impact of their contributions in a transparent and open way, and regular reviews will build the mechanism's efficiency and efficacy over time.

This agreement marks a precedent for benefit-sharing in biodiversity conservation with a fund designed to return some of the proceeds from the use of biodiversity to protect and restore nature where help is needed most….

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Heritable Human Genome Editing [HHGE] – South Africa Law/Regulation/Ethics Guidance 2.0

In our December edition, we provided a high-level summary of responses to the startling inclusion of HHGE – as a "matter-of-fact" topic area – in the 2024 South Africa Ethics in Health Research Guidelines. We include below selected text from the Guidelines and from the important editorial by Ramsey et al. arguing that the "…current wording for HHGE for research purposes in the guidelines should be deleted in its entirety." We are not aware of any new action by the South Africa NDoH but assess that this issue deserves close monitoring and further engagement by the global community.

> **South African Ethics in Health Research Guidelines: Principles, Processes and Structures**
> 2024 Third Edition
> **4.3.2 Heritable human genome editing**
> Research on heritable human genome editing (HHGE) holds significant potential for addressing genetic diseases and improving human health. It also presents ethical challenges that require careful consideration and deliberation. A framework for analysing the protocol emphasises responsible and cautious practices.
> **a) Scientific and medical justification**
> HHGE research must have a clear and compelling scientific and medical rationale, focusing on the prevention of serious genetic disorders and immunity against serious diseases. The potential benefits to individuals and society should outweigh the risks and uncertainties associated with HHGE.
> **b) Transparency and informed consent**
> Researchers must maintain transparency throughout the research process, ensuring that participants and stakeholders are well-informed about the goals, methods, and potential implications of HHGE. Informed consent must be obtained from all parties involved, including prospective parents and individuals whose genetic material is used in the research.
> **c) Stringent ethical oversight**
> HHGE research should be subject to rigorous ethical review by health research ethics committees to evaluate its ethical implications. Ethical oversight should especially be illuminated by the right to freedom of scientific research, the right to access to healthcare, the best interests of prospective children, and the dignity of all individuals involved.
> **d) Ongoing ethical evaluation and adaptation**
> Ethical scrutiny of HHGE research projects should be a continuous process that adapts to evolving circumstances. This entails regular re-evaluation as new information emerges and as the technology progresses.
> **e) Safety and efficacy**
> Researchers must prioritize safety in all HHGE experiments, with thorough assessments of potential risks and strategies for mitigating them. The research should demonstrate a high level of scientific rigor and provide evidence of the technique's efficacy.
> **f) Long-term Monitoring**
> Researchers should commit to ongoing monitoring of individuals born as a result of HHGE interventions to assess their health, wellbeing, and potential unforeseen consequences.
> **g) Legal compliance**
> Researchers must adhere to all relevant laws governing HHGE research. In particular, researchers must adhere to the fourteen-day rule, and must obtain the necessary ministerial permission to conduct research on embryos.

In the context of a number of other commentaries and analyses, **we note again the editorial titled Heritable human genome editing in South Africa – time for a reality check** [*South*

Africa Medical Journal, Early Online -2024-11-29] by Michele Ramsay, Michael Pepper, Jantina de Vries, Safia Mahomed and Eleni Flack-Davison.

The editorial is well structured and provides discussion of the "origins of the current controversy" and its consequences. One contextual clarification in the piece is around public consultation processes leading to the inclusion of the HHGE language in SA guideline referenced above:

> "…A draft of the guidelines released for public comment in 2023 did not include text on HHGE. A subsequent draft released early in 2024 included the section on HHGE with a brief window for comment. It is unclear why the National Health Research Ethics Council (NHREC) chose not to consult widely with topic experts to ensure that the text of the guidelines was appropriate, considering the national legal context, ethical concerns and international recommendations and guidelines for HHGE. It is unfortunate that this situation has arisen. The misguided wording in the current guidelines may serve to precipitate a troubling change in opinions in favour of permitting or promoting HHGE in SA…"

More important is the editorial's clear-thinking conclusion:

> ***…What should happen next?***
> Revision of the NHREC guidelines
> The NHREC must clarify its view on HHGE. We note the recent press release that was circulated to ethics committees around the country by the chair of the NHREC on 8 November 2024. But this does not sufficiently address nor resolve the controversy, and the press release has no legal standing. **The current wording for HHGE for research purposes in the guidelines should be deleted in its entirety**. The underlying premise of section 4.3.2 of the guidelines appears to rest on the fact that there will be prospective parents, prospective children and individuals born because of HHGE research interventions specifically, which is problematic and inconsistent with the law. In addition, this current ambiguity in wording blurs the line between HHGE research and clinical application

In summary, we assess this to be an extremely important, troubling and unresolved matter with global implications. We will continue to monitor for any further analysis, and, hopefully, action!

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Featured Journal Content

We present here four current articles which we assess to be best aligned to our digest focus around genomics governance, ethics, policy, and practice.

We invite readers to review our full selection of articles and commentaries in the Journal Watch section below, aggregated from our monitoring list and google scholar.

We lead this section with a useful "systematic scoping review" by Geuverink, et al. writing in the *European Journal of Human Genetics* focused on **public engagement around human germline gene editing.** The review assesses some 36 studies from 2012-2023 and concludes that "co-created efforts are needed to engage underrepresented groups as well as to yield values rather than acceptance levels, and to concretize how engagement might result in societal impact."

Second, we highlight **a report on The Clinical Genome Resource (ClinGen)** by Andersen et al. describing this NIH-funded program founded 10 years ago that "defines the clinical relevance of genes and variants for medical and research use" and whose working groups "develop standards for data sharing and curating genomic knowledge."

Third, we present an **important consensus statement from a number of Spanish medical/scientific societies addressing population-based genetic carrier screening** and treats program design and implementation strategies, as well as key technical, ethical, and legal issues.

Finally, we include a research paper from the *Journal of Community Genetics* by Berlincourt, et al. **assessing "expert" perspectives on human gene editing in Switzerland**. The author's reference "the unique Swiss context that is shaped by cultural diversity, conservative attitudes towards new medical technologies, and a democratic system that engages the public in policy and law making" and urge that similar research be undertaken in different country contexts to better assess global attitudes and their diversity.

A decade of public engagement regarding human germline gene editing: a systematic scoping review

Review Article
Geuverink, W.P., Houtman, D., Retel Helmrich, I.R.A. et al.
European Journal of Human Genetics (2024). 28 November 2024
https://doi.org/10.1038/s41431-024-01740-6**.**
Abstract
Following the discovery of the CRISPR-Cas technology in 2012, there has been a growing global call for public engagement regarding the potential use of human germline gene editing (HGGE). In this systematic scoping review, we aim to evaluate public engagement studies considering the following questions based on three points of attention: 1) Inclusion of underrepresented groups: who have been engaged? 2) Gathering values: what output has been reported? 3) Reaching societal impact: what objectives of public engagement have been reported? A systematic literature search from 2012 to 2023 identified 3464 articles reporting on public engagement studies regarding HGGE retrieved from 12 databases. After screening, 52 full-text articles were assessed for eligibility, resulting in 36 articles that cover 31 public engagement studies. We conclude that co-created efforts are needed to engage underrepresented groups as well as to yield values rather than acceptance levels, and to concretize how engagement might result in societal impact.

The Clinical Genome Resource (ClinGen): Advancing genomic knowledge through global curation

Research article Open access
Erica F. Andersen, Danielle R. Azzariti, Larry Babb, Jonathan S. Berg, ... Joannella Morales
Genetics in Medicine, Volume 27, Issue 1, January 2025, 101228, Article 101228
Abstract
The Clinical Genome Resource (ClinGen) is a National Institutes of Health-funded program founded 10 years ago that defines the clinical relevance of genes and variants for medical and research use. ClinGen working groups develop standards for data sharing and curating genomic knowledge. Expert panels, with >2500 active members from 67 countries, curate the validity of monogenic disease relationships, pathogenicity of genetic variation, dosage sensitivity of genes, and actionability of gene-disease interventions using ClinGen standards, infrastructure, and curation interfaces. Results are available on clinicalgenome.org and classified variants are also submitted to ClinVar, a publicly available database hosted by the National Institutes of Health. As of January 2024, over 2700 genes have been curated (2420 gene-disease relationships for validity, 1557 genes for dosage sensitivity, and 447 gene-condition pairs for actionability), and 5161 unique variants have been classified for pathogenicity. New efforts are underway in somatic cancer, complex disease and pharmacogenomics, and a systematic approach to addressing justice, equity, diversity, and inclusion. ClinGen's knowledge can be used to build evidence-based genetic testing panels, interpret copy-number variation, resolve

discrepancies in variant classification, guide disclosure of genomic findings to patients, and assess new predictive algorithms. To get involved in ClinGen activities go to https://www.clinicalgenome.org/start.

Population-based genetic carrier screening. A consensus statement from the Spanish societies: AEGH, AEDP, ASEBIR, SEAGEN, SEF and SEGCD

Review Article

Xavier Vendrell, Anna Abulí, Clara Serra, Juan José Guillén, Joaquín Rueda, Javier García-Planells, Fernando Santos-Simarro, Ramiro Quiroga, Fernando Abellán, Raluca Oancea-Ionescu & Encarna Guillén-Navarro

European Journal of Human Genetics (2024). 2 December 2024
https://doi.org/10.1038/s41431-024-01751-3**.**

Abstract

Autosomal recessive or X-linked disorders are passed from parents to offspring through Mendelian inheritance patterns and may lead to severe clinical manifestations in early childhood development. Together, the Spanish Association of Human Genetics (AEGH), Association for the Study of Reproductive Biology (ASEBIR), Spanish Association of Genetic Counselling (SEAGEN), Spanish Fertility Society (SEF), Spanish Society of Clinical Genetics and Dysmorphology (SEGCD), and the Spanish Association of Prenatal Diagnostics (AEDP) developed a consensus statement for population-based genetic carrier screening (GCS). The presented opinion statement recommends that preconception GCS services be included in the public healthcare system to support couples' reproductive autonomy and timely medical decision-making**.** Program design and implementation strategies, as well as key technical, ethical, and legal considerations are discussed.

Experts' perspectives on human gene editing in Switzerland

Research Article

Jade Berlincourt, Sumanie Gächter, Effy Vayena & Kelly E Ormond

Journal of Community Genetics (2024). 19 December 2024 https://doi.org/10.1007/s12687-024-00757-0**.**

Abstract

Despite many specialized studies on the views of the public or stakeholders who face inherited conditions that may be treatable by HGE, limited studies have focused on experts' views towards Human Gene Editing (HGE). Therefore, in this study we conducted exploratory interviews with 14 experts (scientists, clinicians, social scientists, lawyers) in Switzerland to assess their views towards HGE and how they expect the Swiss public to view HGE. We found general acceptance of Somatic Gene Editing (SGE), but opinions towards Germline Gene Editing (GGE) were more divided. Participants emphasized patient autonomy and informed decision-making in pursuing gene editing treatments, and described a need for regulation, as with any other new therapy. Only a few participants (mostly lawyers and ethicists) described the regulations that currently prohibit GGE in Switzerland. Some expressed concern that restrictive regulations would lead to healthcare outsourcing and medical tourism to other nearby countries, as it has in the past with other restricted technologies. The analysis explored the unique Swiss context that is shaped by cultural diversity, conservative attitudes towards new medical technologies, and a democratic system that engages the public in policy and law making. Given that our findings identify areas of difference from that published in other countries, we emphasize the value in conducting similar research across different countries in order to achieve a global sense of attitudes towards HGE, so that regulations can be tailored to the diverse needs of citizens around the world.

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Organization Watch – *Selected Events*

See list of monitored organizations here

African Society of Human Genetics
https://www.afshg.org/about/
Upcoming Event
15th African Society of Human Genetics Conference and the 1st Ugandan Society of Human Genetics and Bioinformatics
3 to 7 February 2025
Entebbe, Uganda
Theme: Harnessing Data Science and Artificial Intelligence for African Genomics

U.S. Department of Health and Human Services
Meeting
Meeting of the Advisory Committee on Heritable Disorders in Newborns and Children
Health Resources and Services Administration (HRSA),
DATES: Thursday, February 13, 2025, from 10 a.m. to 4 p.m. eastern time (ET) and Friday, February 14, 2025, from 10 a.m. to 4 p.m. ET.
SUMMARY:
…During the February 13-14, 2025, meeting, ACHDNC will hear from experts in the fields of public health, medicine, heritable disorders, rare disorders, and newborn screening. Possible agenda items may include the following topics:
(1) An interim update on the evidence review of metachromatic leukodystrophy,
(2) A presentation on the Newborn Screening Contingency Plan framework,
(3) A panel discussion on genomic sequencing in newborns,
(4) A presentation on the National Institutes of Health Newborn Screening by Whole Genome Sequencing Collaboratory, and
(5) An update from the ACHDNC Naming and Counting Condition ad hoc topic group and potential Committee vote on a list of conditions that should be considered for evidence review…

Human Genome Organization (HUGO)
https://www.hugo-international.org/
Upcoming Event
HGM2025 – HUGO Annual Meeting
March 12 - 14, 2025
Southern Sun Elangeni Maharani Hotel, Durban, South Africa
Our scientific program covers a wide range of topics, spanning from system biology and epigenomics to genomic technologies; from drug discovery to gene therapy, pharmacogenomics and genomic medicine; from computation genomics and bioinformatics to genetic and genomic databases; aiming to share most up-to-date research trends, results, information and databases which often sparks off new collaboration opportunities. Apart from generic studies, HGM also provides an arena for presentation and discussion of more focused studies in human genetics and genomics.

ARM [Alliance for Regenerative Medicine]
https://alliancerm.org/press-releases/
Upcoming Event
Cell & Gene Meeting on the Mediterranean
April 15–17, 2025, Rome

The Cell & Gene Meeting on the Mediterranean is the leading conference bringing together the entire cell and gene therapy community from Europe and beyond. Covering a wide range of commercialization topics from market access and regulatory issues to manufacturing and financing the sector, this program features expert-led panels, extensive one-on-one partnering capabilities, exclusive networking opportunities, and 60+ dedicated presentations by leading publicly traded and privately held companies in the space. Join ARM for Europe's premier conference for advanced therapies. Visit the program's website at www.meetingonthemed.com for additional details!

Global Observatory for Genome Editing
https://global-observatory.org/
Upcoming Event
Global Observatory International Summit
May 21 – 23, 2025
The Global Observatory will convene an international summit on May 21-23, 2025 at the American Academy of Arts and Sciences in Cambridge, Massachusetts. Please check back soon for more details…

Global Genomic Medicine Consortium [G2MC]
https://g2mc.org/
Upcoming Event
G2MC 8th International Conference, 2025
DATES TO BE CONFIRMED
The Global Genomic Medicine Consortium (G2MC) 8th International Conference will be held in 2025 in Colombo, Sri Lanka [dates to be confirmed]. The theme of this year's conference is "Collaboration Beyond Borders for Global Implementation of Genomic Medicine" and aims to bring together key stakeholders in the field of genomic medicine to discuss best practices and strategies for implementation, with a particular focus on under-represented regions and low-resource settings.

American Society for Gene and Cell Therapy [ASGCT]
https://asgct.org/
Upcoming Event
ASGCT 28th Annual Meeting
May 13-17, 2025 | New Orleans
Abstract submission is open!
Submit your research for the opportunity to present your cutting-edge work to leaders and colleagues in the field in person during the 28th Annual Meeting in New Orleans, May 13-17, 2025. Submit your research by Jan. 31, 2025, at 11:59 p.m. ET.

NIH National Human Genome Research Institute (NHGRI)
https://www.genome.gov/
Past Event – Documentation/Recordings
Genomic Medicine XVI: Host Genomics and Infectious Disease
December 12-13, 2024
YouTube Channel for Session Recordings:
https://www.youtube.com/playlist?list=PL1ay9ko4A8sllNYK4dC6EbFDixnVS5xJv
The meeting aims to identify needs, opportunities, and challenges for applying a patient's genomic information (genome sequence, transcriptomic, epigenomic, etc. data) in the diagnosis, prevention, and treatment of infectious diseases. Persistent barriers and evidence gaps will be examined as opportunities for additional research.
Meeting Objectives
The objectives of the meeting were to:

- Define currently available approaches for using host genomic information in the diagnosis, prevention, and treatment of infectious diseases
- Examine obstacles and potential solutions to incorporating these and on-the-horizon approaches in clinical care (e.g., cost, reimbursement, regulatory, access, education, insufficient guidelines, and sparse ascertainment of underrepresented groups)
- Identify research opportunities for increasing implementation of host genomic information in clinical care of infectious diseases

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Organization Watch – *Selected Announcements*

See list of monitored organizations here.

Academy of Medical Sciences [UK]
https://acmedsci.ac.uk/
News
Academy welcomes Government's AI Opportunities Action Plan
Monday 13th January 2025
The Government has unveiled its AI Opportunities Action Plan, which aims to ramp up AI adoption across the UK to boost economic growth, provide jobs for the future and improve people's everyday lives.

The plan is a roadmap for government to capture the opportunities of AI.
Professor Andrew Morris CBE FRSE PMedSci, President of the Academy of Medical Sciences, said: "We welcome the Government's bold and ambitious AI Opportunities Action Plan which marks a significant step forward in harnessing AI's potential and transforming the healthcare sector in the UK. As we enter a new era where AI increasingly shapes how we live and work, the plan recognises both its transformative potential and the need for thoughtful implementation.

Paul G. Allen Frontiers Group
https://alleninstitute.org/news-press/
News
CryoSCAPE: Allen Institute scientists develop 'suspended animation' technique for blood draws that will aid research for underserved populations
01.06.2025
A new approach that keeps blood cells alive in deep freeze promises to expand reach of cutting-edge single-cell technologies to underserved...

American Board of Medical Genetics and Genomics (ABMGG)
http://www.abmgg.org/pages/resources_appeal.shtml
Latest Announcements
ABMGG Appoints Azra Ligon, PhD, as its Next Chief Executive Officer
December 20, 2024
The American Board of Medical Genetics and Genomics Board of Directors is pleased to announce the appointment of Azra Ligon, PhD, FACMG, as its new Chief Executive Officer, effective January 1, 2025.

ARM [Alliance for Regenerative Medicine]
https://alliancerm.org/press-releases/
News, Information
The Sector Snapshot: December 2024
A recap of 2024's milestones and an early picture of what could potentially be in store for the 2025 clinical pipeline of cell and gene therapies.
[See Events listing above]

ARRIGE
https://www.arrige.org/
News, Statements, Events
ARRIGE STATEMENTS
Heritable Polygenic Editing
January 2025

Broad Institute of MIT and Harvard
https://www.broadinstitute.org/
Latest
News 01.14.2025
Gene editing extends lifespan in mouse model of prion disease
A single-letter edit in DNA reduces levels of the disease-causing prion protein in the brain and could lead to a preventative, one-time treatment for the deadly neurodegenerative disorder.

News 01.13.2025
Broad Institute and Manifold collaborate to build AI-enabled life sciences research platform to accelerate global discoveries

News 01.09.2025
Scientists engineer CRISPR enzymes that evade the immune system

Center for ELSI Resources and Analysis (CERA)
https://elsihub.org/about/our-mission
Spotlight
News: Announcement
New Special Report of the Hastings Center Report
Jan 7, 2025
We are pleased to announce the publication of a new Hastings Center Report, special report, "Envisioning a More Just Genomics". The report, a collaboration between The Hastings Center and CERA, outlines opportunities to enhance justice in genomics,…

Francis Crick Institute
https://www.crick.ac.uk/news-and-reports
News and Reports
Tumour DNA in the blood can predict lung cancer outcome
Type: News 13 January 2025

Lung cancer test predicts survival in early stages better than current methods
Type: News 9 January 2025

Over 40% of variation in kidney cancer behaviour is not due to changes in DNA
Type: News 8 January 2025

Genetic cause for main type of hearing loss in children with Down syndrome identified
Type: News 7 January 2025

Surprising 'two-faced' cancer gene role supports paradigm shift in predicting disease
Type: News 3 January 2025

Global Alliance for Genomics and Health
https://www.ga4gh.org/
News
Model Data Access Agreement Clauses have been approved as an official GA4GH Product
5 Dec 2024
Following a successful vote by the Global Alliance for Genomics and Health (GA4GH) Product Steering Committee, the Model Data Access Agreement Clauses have been approved as an official GA4GH product to help researchers access data more efficiently and ethically in line with local legislation.

Genomics England
https://www.genomicsengland.co.uk/
Latest
13 Jan 2025
Researchers uncover what drives aggressive bone cancer

Genetics Society of America (GSA)
http://genetics-gsa.org/
News, Events
GSA News
New members of the GSA Board of Directors: 2025–2027
We are pleased to announce the election of six new leaders to the GSA Board of Directors, including 2026 President Cassandra Extavour.
December 20, 2024

GSA Journals
January 2025 issue of GENETICS published
January 10, 2025

NIH National Human Genome Research Institute (NHGRI)
https://www.genome.gov/
News & Events
NHGRI appoints Erin Ramos as the next director of the Division of Genome Sciences
January 07, 2025
Ramos to lead the institute's extramural programs in basic genomics research

National Organization for Rare Disorders (NORD)

https://rarediseases.org/news/
News
Tracey Sikora Joins the National Organization for Rare Disorders (NORD) as Vice President of Research and Clinical Programs
Published December 12, 2024 by NORD

Nuffield Council on Bioethics
https://www.nuffieldbioethics.org/news
News
News 27th November 2024
Nuffield Council on Bioethics sets out proposals to bolster governance of stem cell-based embryo models including a call for legislation to ensure that research does not cross ethical 'red lines'
The Nuffield Council on Bioethics (NCOB) has published proposals for the governance of research using stem cell-based embryo models (SCBEMs).

UNESCO–The World Academy of Sciences
https://twas.org/
Press Room
19 December 2024
Editorial: Science is our greatest hope for a brighter future
TWAS is a beacon of hope for scientists in the developing world

Wellcome Sanger Institute
https://www.sanger.ac.uk/
News
13 Jan 2025
Bacterial survival genes uncovered using evolutionary map
The most detailed study to date on the mechanisms by which a common type of bacterium, Staphylococcus aureus, adapts to living ...

8 Jan 2025
Human 'Domainome' reveals root cause of inherited conditions
The largest catalogue of human protein variants to date has revealed that protein destabilisation is the main driver of inherited genetic ...

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Journal Watch

In preparing *Journal Watch*, we formally monitor a broad range of academic journals [listed here] and, in parallel, utilize Google Scholar to identify articles aligned with our areas of focus. After careful consideration, a selection of these results appear in the digest, organized under thematic areas to help readers navigate.

Thematic Areas

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GENOME EDITING

Prime time for gene editing

Year in Review 12 Dec 2024
Karen O'Leary
Nature Medicine, Volume 30 Issue 12, December 2024

Gene-editing tools are evolving rapidly, from the first CRISPR–Cas systems to base editing and, more recently, prime editing. Unlike CRISPR–Cas systems, base editing does not require double-strand breaks; neither does prime editing, and it supports greater editing power — including targeted insertions and deletions. In April, the US Food and Drug Administration cleared the investigational new drug application for PM359 (from Prime Medicine), making it the first prime editor to enter the clinic. Designed as a one-time treatment to correct a mutation that causes chronic granulomatous disease, PM359 will first be evaluated in adult patients in a phase 1/2 study (NCT06559176) — with the aim of then testing it in adolescents and children, if it is deemed safe. Another technique, epigenome editing, enables gene silencing without any alteration to the gene itself. Instead, the editing machinery targets the heritable (but reversible) structures…

When promising therapies are out of reach: Ethical responsibilities of stakeholders in gene therapy trials for rare disorders

Opinion
Rami M. Major, Zollie Yavarow
Molecular Therapy, Jan 08, 2025 Volume 33 Issue 1 p1-422

When a gene therapy enters the clinic, a nuanced conversation must be conducted with patients who will ultimately decide whether or not to enroll in a clinical trial. A thin line exists between pitching the prospect of a therapeutic benefit to patients and emphasizing the "trial" aspect of a therapy itself. But what does it mean to create a gene therapy that is a success or a failure? An initially promising gene therapy for giant axonal neuropathy was dropped by Taysha Gene Therapies after the FDA recommended a double-blind, placebo-controlled trial with extensive long-term follow-up to demonstrate efficacy.[1] A gene therapy clinical trial by Editas Medicine for a rare eye disorder also showed substantial benefits in an open-label, phase 1/2 clinical trial, but it was abandoned after the company realized that these benefits only occurred for a small subset of patients and would therefore

have a smaller market.[2] These cases illustrate some of the difficult conundrums of gene therapies: clinical benefits do not necessarily guarantee that a company will continue to invest in the cost-intensive process of advancing a therapy through a lengthy clinical trial process.

An assessment of the current state of interdisciplinary CRISPR research

Report based on the workshop: The Technology and Ethics of CRISPR [Spring 2024]

Franziska Bächler and Anina Meier

H2R Working Paper 01/2024 :: 36 pages Seismo: https://doi.org/10.33058/wpuzh.2024.7110

University of Zurich

Abstract:

CRISPR/Cas genome editing is a dynamic field of biotechnology that is evolving and establishing itself in diverse fields such as agriculture and human medicine. The rapid development of this technology is accompanied by public debate about its various applications. This article sets out the opportunities and risks of CRISPR/Cas9 technology from an interdisciplinary perspective. The occasion and inspiration for the article was a workshop entitled 'The Technology and Ethics of CRISPR', which was organised in collaboration between the University of Zurich's Human Reproduction Reloaded (H2R) research programme and the University of Basel's Centre for Life Sciences Law (ZLSR) and was held in spring 2024 with presentations by renowned experts from various scientific disciplines.

The article outlines the current state of research on CRISPR with an overview of the natural and social sciences, legal and patent law as well as ethical aspects that appear to be essential for the interdisciplinary negotiation of the technology.

This review of the application of CRISPR in medicine highlights the first approved CRISPR-based gene therapy for sickle cell anaemia and addresses the challenges of access and regulation that such medical breakthroughs face. The somatic application of gene editing is contrasted with its use in the human germline. The technological uncertainties and open ethical and socio-political questions regarding the latter are also summarised.

Key findings from the workshop presentations are embedded in a discussion and analysis of the CRISPR ecosystem, which is characterised by public-private partnerships and a complex patent situation.

9 Conclusion [excerpt]:

…The diversity of perspectives and questions raised in this paper make it clear that CRISPR is far more than just a biotechnological tool. The opportunities and dangers arising from the use of CRISPR comprise a societal challenge that calls for an interdisciplinary dialogue in which various, sometimes contradictory perspectives, must be taken into account. How should these diverse concerns be evaluated? Should priority be given to those who want to advance CRISPR research, or to those who object to the possibility of modifying the genome at will, or to the patients who could benefit from new therapies, or to the parents of future generations?

This endeavour is necessary because a responsible approach to CRISPR can only emerge if all scientific and social perspectives are taken into account, weighing up both the possibilities and the risks of the technology. The expert discussions at the workshop showed that to achieve such a goal, it is important to develop a common understanding of the technology in order to facilitate an evidence-based discussion between the disciplines. To this end, this article contributes an informative basic resource to institutionalise such interdisciplinary exchange and to engage the public in discussions about the future possibilities of CRISPR.

Franziska Bächler is a PostDoc researcher and the Scientific Manager at the Centre for Life Sciences Law (ZLSR) at the University of Basel.

Anina Meier is a PhD candidate at the URPP Human Reproduction Reloaded | H2R, University of Zurich.

Advances in the use of Genome Editing Tools in Africa: A Review

Review Article

Ogbu, Angela Chinenye, Okpaga Austine Ume, Olando Cletus Nwogiji, Samson Olumide Akeredolu, Ofobuike Godson Eze, Chikezie Victor Onwe, Oluwasegun Ifeoluwa Oduguwa, and Uchenna Victor Chigozie.
Asian Journal of Biochemistry, Genetics and Molecular Biology 16 (12):48-67.
https://doi.org/10.9734/ajbgmb/2024/v16i12422. 11 December 2024
Abstract
Africa is grappling with various challenges, particularly in agricultural production and disease prevention affecting humans, animals, and crops. Gene editing, or genome editing (GE), involves modifying, adding, or removing nearly any DNA sequence in various cells and organisms. Due to new techniques, GE is now quicker, less expensive, and more effective. The CRISPR/Cas9 system is based on RNA and has been more effectively tweaked than protein-based methods, enabling multi-site manipulation. The Type II CRISPR system protects DNA from plasmids and viruses that invade it through RNA-guided DNA cleavage by Cas proteins. Several diseases are currently being treated with CRISPR-based GE technology. The raising of livestock is vital to modern society, and it is directly impacted by disease resistance. Here, we provided a comprehensive review of how these GE tools have enhanced resilience against biotic and abiotic stresses, leading to increased yields. We elaborated on how GE has also facilitated the development of disease-resistant varieties of bananas, cassava, and maize, effectively addressing plant diseases like cassava mosaic and brown streak by targeting specific genes. We further emphasized the application of GE in animal breeding, exploring the successful creation of disease-resistant livestock and developing vaccines against diseases. Our findings explored the applications of GE in tackling human health challenges, including artemisinin resistance and hepatitis B treatment. Our summary highlighted limited adoption of GE technologies only in a few African countries such as Kenya, South Africa, Nigeria, Ethiopia, Egypt, Uganda, Burkina Faso, Ghana, and Rwanda. We further reported the persistence of societal issues despite its advancement, including religious beliefs and concerns about the implications of GE in homes, leading to fear and discrimination against its use. We finally reported the efforts of scientists in advocating for policies and consensus on implementing GE in Africa to address these challenges.

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PRECISION MEDICINE

Cost-Effectiveness and Policy Implications of Personalised Medicine

Review Article
Syed Sibghatullah Shah
Premier Journal of Science. 2024:1;100018 DOI: https://doi.org/10.70389/ **PJS.100018. 14 October 2024**
Abstract
PURPOSE
Personalised medicine's integration into healthcare systems around the world, as well as its policy implications and economic impact, are all thoroughly examined in this article. Personalised medicine holds great potential for better patient outcomes through pharmacogenomic-guided therapies; however, there are substantial financial and regulatory hurdles to overcome.
METHODS
Our extensive work included cost-utility analyses and cost-effectiveness analyses. The long-term economic impacts of various scenarios were modelled using Monte Carlo simulations. These scenarios included increasing the scale of genetic testing in high-risk populations and expanding the use of pharmacogenomic treatments for cardiovascular disease. Additionally, data about ethical considerations were discarded from case studies and regulatory documents.
RESULTS

For high-risk cancer patients, the incremental cost-effectiveness ratio (ICER) for expanding genetic testing averaged $58,500 per quality life years (QALY), and it was likely cost-effective 75% of the time. Cardiovascular pharmacogenomic testing had a higher economic benefit, with an 88% likelihood of cost effectiveness and an ICER of $42,000 per QALY. With a 40% likelihood of cost-effectiveness and an ICER of $115,000 per QALY, personalised cancer immunotherapies were less cost-effective.
CONCLUSIONS
Despite obstacles such as high initial costs and disjointed regulatory frameworks, personalised medicine shows great promise for improved outcomes and cost savings through genetic testing and pharmacogenomics.

Recent Advances in Personalized Medicine and Their Key Role in Public Health Systems
Book Chapter
Frank Abimbola Ogundolie, Ale Oluwabusolami, Akinmoju Olumide Damilola
Health Technologies and Informatics, 1st Edition 2024, CRC Press, Pages 13. eBook ISBN9781003309468
Abstract
Across the various health systems, the desire to ensure more efficient treatments through techniques tailored to people's individual health needs based on their genomic information has been a ground-breaking area in the health sector today. Advancement in genomics and understanding of the genetic basis of diseases have made personalized medicine an emerging medical practice for easier diagnosis and treatment. Today, more specific medical approaches have been engaged which are largely based on individuals, therapies ranging from diets, nutraceuticals, nutrigenomics, dosage among others has been improving the field of medicine. This chapter looks at personalized medicine as a whole and the role of public health systems in personalized medicine.

Health equity innovation in precision medicine: data stewardship and agency to expand representation in clinicogenomics
Comment
Patrick J. Silva, Vasiliki Rahimzadeh, Reid Powell, Junaid Husain, Scott Grossman, Adam Hansen, Jennifer Hinkel, Rafael Rosengarten, Marcia G. Ory & Kenneth S. Ramos
Health Research Policy and Systems 22, 170 (2024). https://doi.org/10.1186/s12961-024-01258-9**. 19 December 2024**
Abstract
Most forms of clinical research examine a very minute cross section of the patient journey. Much of the knowledge and evidence base driving current genomic medicine practice entails blind spots arising from underrepresentation and lack of research participation in clinicogenomic databases. The flaws are perpetuated in AI models and clinical practice guidelines that reflect the lack of diversity in data being used. Participation in clinical research and biobanks is impeded in many populations due to a variety of factors that include knowledge, trust, healthcare access, administrative barriers, and technology gaps. A recent symposium brought industry, clinical, and research participants in clinic-genomics to discuss practical challenges and potential for new data sharing models that are patient centric and federated in nature and can address health disparities that might be perpetuated by lack of diversity in clinicogenomic research, biobanks, and datasets. Clinical data governance was recognized as a multiagent problem, and governance practices need to be more patient centric to address most barriers. Digital tools that preserve privacy, document provenance, and enable the management of data as intellectual property have great promise. Policy updates realigning and rationalizing clinical data governance practices are warranted.

Health data analytics for precision medicine: A review of current practices and future directions
Article

Vyvyenne Michelle Chigboh, Stephane Jean Christophe Zouo, & Jeremiah Olamijuwon
International Medical Science Research Journal, Volume 4, Issue 11, November 2024
Abstract
This review paper explores the role of health data analytics in advancing precision medicine, highlighting current practices, challenges, and future directions. Precision medicine aims to tailor medical treatment to individual characteristics, and health data analytics plays a critical role in achieving this goal by leveraging diverse data sources, including genomic, clinical, and lifestyle information. Key insights reveal the transformative impact of artificial intelligence (AI) and machine learning in analyzing complex datasets and integrating multi-omics data and wearable health devices. However, challenges such as data privacy and security concerns, issues of standardization and interoperability, and barriers to implementation in clinical settings persist. The paper concludes with recommendations to enhance data protection measures, promote interoperability, provide training for healthcare providers, foster collaborative research, and advocate for supportive regulatory reforms. By addressing these challenges, stakeholders can unlock the full potential of health data analytics, ultimately improving patient care and outcomes in precision medicine.

Deep learning in predictive medicine: Current state of the art

Book Chapter
Manoj Kumar Yadav, Manish Kumar Tripathi, Navaneet Chaturvedi, Abhigyan Nath, Upendra Kumar
Deep Learning in Genetics and Genomics. Volume 1: Foundations and Introductory Applications 2025, Pages 261-291. 29 November 2024
Abstract
Predictive medicine is an emerging field that utilizes various data sources and advanced technologies to predict disease occurrence, patient outcomes, and responses to treatment. Integrating predictive medicine with AI-based techniques has the potential to transform traditional healthcare into a more patient-centric, preventive, and efficient model. By analysing clinical and nonclinical data patterns, healthcare predictive analytics can forecast future health events and outcomes. Recently, deep learning applications have become prevalent in pharmaceutical research, helping address drug discovery issues and improving patient treatment by analysing medical data. The transformative role of deep learning in predictive medicine is further explored, covering data acquisition, preprocessing, and fundamental deep learning concepts in healthcare. The discussion includes the use of deep learning in disease diagnosis, early detection, medical imaging diagnostics, and risk assessment models. This chapter concludes by exploring current challenges, future trends, and unexplored potentials, supported by case studies demonstrating successful real-world applications.

Challenges of Clinical Pharmacogenomics Implementation in the Era of Precision Medicine

Research Article
Sherin Shaaban, Yuan Ji
Medical Research Archives, [S.l.], v. 12, n. 11, nov. 2024. ISSN 2375-1924. doi: https://doi.org/10.18103/mra.v12i11.5955**. 29 November 2024**
Abstract
While pharmacogenomics (PGx) presented to many as the poster child of personalized medicine, in the context of moving away from the "one size fits all" model of pharmacological therapy of diseases to a more tailored approach addressing the individuality of each person, the application of PGx has been hindered by numerous challenges. These challenges range from issues with study designs in scientific research, both clinical and for discovery, policy and regulatory hurdles affecting insurance coverage, to the very fundamental need for adequate training for laboratorians, physicians, and pharmacists. Moreover, access to services and addressing health disparities in personalized medicine generally and PGx specifically remain a complicated endeavor. PGx-related ethical, legal, and social issues continue to be a point of concern for those looking to implement PGx in clinical practice.

Additionally on the technical side, the speed with which next generation sequencing (NGS) technologies have evolved, generating tens of thousands of rare PGx variants adds a new layer of challenges requiring accurate interpretation and assessment of functional role of novel variants to determine their impacts on drug response and possible toxicities. Without consensus and standardized approaches to testing and interpretation, integration of PGx into routine clinical care becomes an unattainable task. In this article we aim to address some of the challenges that impede broad adoption of clinical PGx testing, and to shed the light on needed steps towards a successful implementation of PGx, with the goal of improving health outcomes individually and for the general population.

Harnessing the Power of Precision Medicine and AI: Success Stories and Ethical Considerations

Book Chapter

Latika Sahni, Rishi Prakash Shukla

Driving Global Health and Sustainable Development Goals with Smart Technology. Copyright: © 2025|Pages: 46. DOI: 10.4018/979-8-3373-0240-9.ch003

Abstract

This chapter explores the transformative potential of precision medicine and AI in revolutionizing healthcare delivery. By tailoring treatments to individual patient profiles through genomic insights and advanced analytics, personalized healthcare optimizes therapeutic efficacy, particularly in oncology, rare diseases, and chronic conditions. Case studies illustrate the economic advantages of targeted interventions, highlighting cost efficiency through reduced trial-and-error treatments and improved patient outcomes. However, the integration of AI introduces ethical dilemmas, including data privacy concerns, algorithmic biases, and the implications of genetic editing technologies. Global examples of healthcare systems implementing AI-driven personalized medicine provide valuable insights into best practices and lessons learned from early adopters. As the landscape of healthcare evolves, the chapter emphasizes the need for interdisciplinary collaboration, robust data infrastructure, and active patient engagement to navigate the complexities of precision medicine.

Global Perspectives on Pharmacogenomics and Drug Discovery

Book Chapter

K. Rajakumari, K. Kavinaya Shri, R. Logesh, S. S. Meenambiga, P. Vivek, S. Ivo Romauld

Genomics-Driven Drug Discovery Through Pharmacogenomics. Copyright: © 2025 |Pages: 44. DOI: 10.4018/979-8-3693-6597-7.ch005

Abstract

Pharmacogenomics, the study of how genes influence an individual's response to drugs, is changing the scene of medication disclosure and improvement by joining pharmacology and genomics to make powerful, custom-made to an individual's hereditary profile. This arising field is vital in customized medication, where medicines are upgraded in light of individual hereditary varieties. This chapter offers an in-depth exploration of the global impact of pharmacogenomics, underscoring key examination foundations and undertakings, moral contemplations, difficulties and future headings. The joint efforts exhibit the capability of joint endeavor to defeat difficulties and advance the field by fitting worldwide guidelines and creating strategies that help even-handed admittance to pharmacogenomic progressions are significant for the worldwide reconciliation of customized medication.

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GENOMICS RESEARCH ETHICS, INTEGRITY

Ethical challenges of conducting and reviewing human genomics research in Malaysia: An exploratory study

ORIGINAL ARTICLE

Teong Win Zee, Mohammad Firdaus Bin Abdul Aziz, Phan Chia Wei

Developing World Bioethics, Volume 24, Issue 4 Pages: 263-342 December 2024 Pages: 331-341

First Published: 23 November 2023

Abstract

Even though there is a significant amount of scholarly work examining the ethical issues surrounding human genomics research, little is known about its footing in Malaysia. This study aims to explore the experience of local researchers and research ethics committee (REC) members in developing it in Malaysia. In-depth interviews were conducted from April to May 2021, and the data were thematically analysed. In advancing this technology, both genomics researchers and REC members have concerns over how this research is being developed in the country especially the absence of a clear ethical and regulatory framework at the national level as a guidance. However, this study argues that it is not a salient issue as there are international guidelines in existence and both researchers and RECs will benefit from a training on the guidelines to ensure genomics research can be developed in an ethical manner.

Ethical, legal, and social issues related to genetics and genomics in cancer: A scoping review and narrative synthesis

Review article Open access

Amelia K. Smit, Akira Gokoolparsadh, Rebekah McWhirter, Lyndsay Newett, ... Ainsley J. Newson

Genetics in Medicines, Volume 26, Issue 12 December 2024, Article 101270

Abstract

Genomics is increasingly being incorporated into models of care for cancer. Understanding the ethical, legal, and social issues (ELSI) in this domain is important for successful and equitable implementation. We aimed to identify ELSI scholarship specific to cancer control and genomics. To do this, we undertook a scoping literature review and narrative synthesis, identifying 46 articles that met inclusion criteria. Eighteen ELSI themes were developed, including (1) equity of access, which included structural barriers to testing and research, access to preventive and follow-up care, and engagement with health systems; (2) family considerations, such as an ethical obligation to disseminate relevant genomic information to at-risk family members; (3) legal considerations, including privacy and confidentiality, genetic discrimination, and the prospective duty to reclassify variants; and (4) optimizing consent processes in clinical care and research. Gaps in the literature were identified with respect to equity for people living in rural or remote areas, and how to provide ethical care within culturally, linguistically, and ethnically diverse communities, including First Nations peoples. Our findings suggest a need for a multidisciplinary approach to examining ELSI in cancer genomics beyond initial test indication and within the broader context of the mainstreaming of genomics in health care.

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GENOMIC DATA, BIOBANKING

SHARE: An ethical framework for equitable data sharing in Caribbean health research

Article

Michael H Campbell, Natalie S Greaves

Rev Panam Salud Publica. 2024 Dec 16;48:e97. doi: 10.26633/RPSP.2024.97

Abstract

Data sharing increasingly underpins collaborative research to address complex regional and global public health problems. Advances in analytic tools, including machine learning, have expanded the potential benefits derived from large global repositories of open data. Participating in open data collaboratives offers opportunities for Caribbean researchers to advance the health of the region's population through shared data-driven science and policy. However, ethical challenges complicate these efforts. Here we discuss fundamental challenges that threaten to impede progress if not strategically addressed, including power dynamics among funders and researchers in high-income countries and Caribbean stakeholders; research and health equity; threats to privacy; and risk of stigma. These challenges may be exacerbated by resource and infrastructure limitations often seen in small island developing states (SIDS) and low- and middle-income countries. We propose a framework for **S**afeguarding **H**ealth **A**nd **R**esearch data sharing by promoting **E**quity (SHARE) for Caribbean researchers and communities participating in shared data science. Using the SHARE framework can support regionally relevant and culturally responsive work already underway in the region and further develop capacity for intentional sharing and (re)use of Caribbean health data.

Genetic Data in Insurance Underwriting: Considerations in the Thai Context

Article

Punnathorn Noramut

This article is summarized and rearranged from the thesis "Problems Related to Genetic Data and Life Insurance", Faculty Law, Thammasat University, 2023.

Thammasat Business Law Journal Vol. 14 2024

Abstract

Whole-genome sequences contribute significantly to our understanding of human development and functions. Various abnormalities can result in diseases or reduce individuals' life expectancy. Utilizing genetic data for predicting an individual's health status is a consideration in insurance underwriting assessments. However, there is an ongoing debate about the appropriateness of using this information, given its unchangeable nature since birth. Discussions often center around the principles of the insurance premium setting and underwriting, which vary based on the applicant's health status and risk assessment.

There are two potential approaches. The first option is enacting laws. This would ensure that individuals are not discriminated against based on their genetic data. One consequence of this approach might be a significant increase in premium pricing for the overall portfolio. This could be due to the increased uncertainty for insurers when they cannot use genetic data to assess risks. Another option is discussion and agreement among concerned parties. This approach involves engaging in open discussions and reaching agreements among the relevant stakeholders. It allows for a more flexible and adaptable approach, where policies can be revised based on outcomes and experiences, especially if adverse events occur during a trial period.

Both options have their advantages and disadvantages. The best choice for Thailand will depend on what is considered most important and what aligns best with the country's specific circumstances and priorities. It's crucial to carefully weigh the implications and make a decision that serves the interests of all stakeholders involved.

Genomics and Health Data Governance in Africa: Democratize the Use of Big Data and Popularize Public Engagement

Data

Nchangwi Syntia Munung, Charmaine D. Royal, Carmen De Kock, Gordon Awandare, Victoria Nembaware, Seraphin Nguefack, Marsha Treadwell, and Ambroise Wonkam

Special report, Hastings Center Report 54, no. S2 (2024): S84–S92.
DOI:10.1002/hast.4933. 21 December 2024

Abstract

Effectively addressing ethical issues in precision medicine research in Africa requires a holistic social contract that integrates biomedical knowledge with local cultural values and Indigenous knowledge systems. Drawing on African epistemologies such as *ubuntu* and *ujamaa* and on our collective experiences in genomics and big data research for sickle cell disease, hearing impairment, and fragile X syndrome and the project Public Understanding of Big Data in Genomics Medicine in Africa, we envision a transformative shift in health research data governance in Africa that could help create a sense of shared responsibility between all stakeholders in genomics and data-driven health research in Africa. This shift includes proposing a social contract for genomics and data science in health research that is grounded in African communitarianism such as solidarity, shared decision-making, and reciprocity. We make several recommendations for a social contract for genomics and data science in health, including the coproduction of genomics knowledge with study communities, power sharing between stakeholders, public education on the ethical and social implications of genetics and data science, benefit sharing, giving voice to data subjects through dynamic consent, and democratizing data access to allow wide access by all research stakeholders. Achieving this would require adopting participatory approaches to genomics and data governance.

Health equity innovation in precision medicine: data stewardship and agency to expand representation in clinicogenomics

Comment

Patrick J. Silva, Vasiliki Rahimzadeh, Reid Powell, Junaid Husain, Scott Grossman, Adam Hansen, Jennifer Hinkel, Rafael Rosengarten, Marcia G. Ory and Kenneth S. Ramos

Health Research Policy and Systems, 2024 22:170 Published on: 19 December 2024

Abstract

Most forms of clinical research examine a very minute cross section of the patient journey. Much of the knowledge and evidence base driving current genomic medicine practice entails blind spots arising from underrepresentation and lack of research participation in clinicogenomic databases. The flaws are perpetuated in AI models and clinical practice guidelines that reflect the lack of diversity in data being used. Participation in clinical research and biobanks is impeded in many populations due to a variety of factors that include knowledge, trust, healthcare access, administrative barriers, and technology gaps. A recent symposium brought industry, clinical, and research participants in clinicogenomics to discuss practical challenges and potential for new data sharing models that are patient centric and federated in nature and can address health disparities that might be perpetuated by lack of diversity in clinicogenomic research, biobanks, and datasets. Clinical data governance was recognized as a multiagent problem, and governance practices need to be more patient centric to address most barriers. Digital tools that preserve privacy, document provenance, and enable the management of data as intellectual property have great promise. Policy updates realigning and rationalizing clinical data governance practices are warranted.

A framework for sharing of clinical and genetic data for precision medicine applications

Article Open Access 03 Sept 2024

Ahmed Elhussein, Ulugbek Baymuradov, Gamze Gürsoy

Nature Medicine, Volume 30 Issue 12, December 2024

Abstract

Precision medicine has the potential to provide more accurate diagnosis, appropriate treatment and timely prevention strategies by considering patients' biological makeup. However, this cannot be realized without integrating clinical and omics data in a data-sharing framework that achieves large sample sizes. Systems that integrate clinical and genetic data from multiple sources are scarce due to their distinct data types, interoperability, security and data ownership issues. Here we present a secure framework that allows immutable storage, querying and analysis of clinical and genetic data using blockchain technology. Our platform allows clinical and genetic data to be harmonized by combining them under a unified framework. It supports combined genotype–phenotype queries and

analysis, gives institutions control of their data and provides immutable user access logs, improving transparency into how and when health information is used. We demonstrate the value of our framework for precision medicine by creating genotype–phenotype cohorts and examining relationships within them. We show that combining data across institutions using our secure platform increases statistical power for rare disease analysis. By offering an integrated, secure and decentralized framework, we aim to enhance reproducibility and encourage broader participation from communities and patients in data sharing.

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GENETIC SCREENING/ GENETIC COUNSELLING

Who has the responsibility to inform relatives at risk of hereditary cancer? A population-based survey in Sweden

Original Research

Grill K, Phillips A, Numan Hellquist B, et al

BMJ Open 2024;14:e089237. doi: 10.1136/bmjopen-2024-089237. 27 November 2024

Abstract

Objectives

Hereditary cancer has implications not only for patients but also for their at-risk relatives (ARRs). In current clinical practice, risk disclosure to ARRs involves collaboration between patients and healthcare providers (HCPs). However, the specific responsibilities of each party are intertwined and at times unclear. In this study, we explored public attitudes regarding moral and legal responsibilities to disclose familial risk information to uninformed ARRs.

Design

In an online cross-sectional survey, participants were prompted with a hypothetical scenario where a gender-neutral patient learnt about their familial risk of colorectal cancer. The patient was advised to regularly undergo colonoscopy screening, and this recommendation was extended to both their siblings and cousins. While the patient informed their siblings, they had not spoken to their cousins in 20 years and did not want to contact them. The survey assessed respondents' views on the patient's and HCPs' ethical responsibility and legal obligation to inform the cousins (ARRs).

Participants

A random selection of 1800 Swedish citizens 18–74 years of age were invited. Out of those, 914 (51%) completed the questionnaire.

Results

In total, 75% believed that HCPs had a moral responsibility to inform ARRs, while 59% ascribed this moral responsibility to the patient. When asked about the ultimate responsibility for risk disclosure to ARRs, 71% placed this responsibility with HCPs. Additionally, 66% believed that HCPs should have a legal obligation to inform ARRs, while only 21% thought the patient should have such an obligation. When prompted about a scenario in which the patient actively opposed risk disclosure, a majority believed that HCPs should still inform the ARRs.

Conclusion

Our study indicates that the Swedish public ascribes moral responsibility for informing ARRs to both the patient and HCPs. However, contrary to current practice, they believe HCPs hold the ultimate responsibility. The majority of respondents support disclosure even without patient consent.

Preconception carrier screening among assisted reproduction patients: insights from a monocentric survey in France

Article

Mario Abaji, Arnold Munnich, Catherine Racowsky, Camille Fossard, Jessica Vandame, Mathilde Labro, AchrafBenammar, Jean-Marc Ayoubi, Marine Poulain

Reproductive BioMedicine. https://doi.org/10.1016/j.rbmo.2024.104757. 21 December 2024

Abstract

Research Question

What are the opinions of individuals undergoing assisted reproduction in France on preconception genetic carrier screening for severe genetic diseases?

Design

This prospective monocentric study utilized an anonymous online questionnaire targeting patients who consulted at the Assisted Reproductive Technology (ART) service of the Obstetrics and Reproductive Medicine Department, Foch Hospital between January 2021 to October 2023.

Results

Out of the 7,793 patients successfully contacted by email, 1,206 responded to the survey, yielding a 15.5% response rate. The majority of participants (90.8%) agreed or strongly agreed that preconception genetic carrier screening represents a significant advancement in the field of Assisted Reproductive Technology, 41.9% documented that such screening should be available to all couples planning to have children and 43.1% supported such screening being available to any adult wishing to know their carrier status.

Conclusions

There is strong support for preconception genetic carrier screening among patients undergoing assisted reproduction. This patient population is ideal for initiating essential pilot studies in France concerning such screening. These studies are crucial for understanding and addressing economic and technological challenges, as well as advancing ethical considerations in this emerging field.

Global Perspectives on Returning Genetic Research Results in Parkinson Disease

Research Article

Ai Huey Tan, Paula Saffie-Awad, Artur F. Schumacher Schuh, et al.

Neurology Genetics, December 2024 issue. 10 (6). 5 December 2024

https://doi.org/10.1212/NXG.0000000000200213.

Abstract

Background and Objectives

In the era of precision medicine, genetic test results have become increasingly relevant in the care of patients with Parkinson disease (PD). While large research consortia are performing widespread research genetic testing to accelerate discoveries, debate continues about whether, and to what extent, the results should be returned to patients. Ethically, it is imperative to keep participants informed, especially when findings are potentially actionable. However, research testing may not hold the same standards required from clinical diagnostic laboratories and hold significant psychosocial implications. The absence of universally recognized protocols complicates the establishment of appropriate guidelines.

Methods

Aiming to develop recommendations on return of research results (RoR) practice within the Global Parkinson's Genetics Program (GP2), we conducted a global survey to gain insight on GP2 members' perceptions, practice, readiness, and needs surrounding RoR.

Results

GP2 members (n = 191), representing 147 institutions and 60 countries across 6 continents, completed the survey. Access to clinical genetic testing services was significantly higher in high-income countries compared with low- and middle-income countries (96.6% vs 58.4%), where funding was predominantly covered by patients themselves. While 92.7% of the respondents agreed that genetic research results should be returned, levels of agreement were higher for clinically relevant results relating to pathogenic or likely pathogenic variants in genes known to cause PD or other neurodegenerative diseases. Less than 10% offered separate clinically accredited genetic testing before returning genetic research results. A total of 48.7% reported having a specific statement on RoR policy in their ethics consent form, while 53.9% collected data on participants' preferences on

RoR prospectively. 24.1% had formal genetic counselling training. Notably, the comfort level in returning incidental genetic findings or returning results to unaffected individuals remains low.
Discussion
Given the differences in resources and training for RoR, as well as ethical and regulatory considerations, tailored approaches are required to ensure equitable access to RoR. Several identified strategies to enhance RoR practices include improving informed consent processes, increasing capacity for genetic counselling including providing counselling toolkits for common genetic variants, broadening access to sustainable clinically accredited testing, building logistical infrastructure for RoR processes, and continuing public and health care education efforts on the important role of genetics in PD.

Promises and pitfalls of preimplantation genetic testing for polygenic disorders: a narrative review
Review Article
Roura-Monllor, Jaime A. et al.
F&S Reviews, Volume 0, Issue 0, 100085. 19 December 2024
Abstract
Preimplantation genetic testing for polygenic disorders (PGT-P) has been commercially available since 2019. PGT-P makes use of polygenic risk scores for conditions which are multifactorial and are significantly influenced by environmental and lifestyle factors. If current predictions are accurate, then absolute risk reductions range from about 0.02% to 10.1%, meaning that between 10 and 5,000 in vitro fertilization patients would need to be tested with PGT-P to prevent one offspring from becoming affected in the future, depending on the condition and the number of embryos available. Survey and interview data reveal that patients and the public have largely favorable views regarding the use of PGT-P for disease prevention; however, clinicians and professional organizations have many reservations. The use of PGT-P raises multiple social and ethical concerns including the need for adequate counseling, the setting of realistic expectations, the application of distributive justice, the impact of environmental and social determinants of health, and the potential exacerbation of health inequities. Clinicians expressed significant concerns relating to the cost of PGT-P, the potential time-consuming counseling for reproductive endocrinologists and genetic counselors, the intentional creation of supernumerary embryos, and patients' unrealistic expectations regarding "healthiest disease-free" embryos. Furthermore, current evidence lacks long-term outcome data and generalizability. Prior to offering PGT-P to patients, additional clinical validation studies are needed. Also, ethical and social considerations raised by PGT-P should be carefully delineated. Systemic practices to increase equitable access to unbiased genetic counseling and reproductive services would be desirable prior to the ethical implementation of PGT-P.

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DISEASE-SPECIFIC GENOMICS

Researchers' perspectives on the integration of molecular and genomic data into malaria elimination programmes in Africa: a qualitative study
Research Article
Tindana, P., Sekwo, D.E., Baatiema, L. *et al.*
Malaria Journal volume 23, Article number: 385 (2024). 18 December 2024
Abstract
Background
Malaria remains a significant public health concern, despite global efforts to combat the disease with highest burden in Africa. Reports of emerging artemisinin partial- resistance in East Africa emphasize the importance of molecular data to guide policy decisions. Hence the need for researchers to

collaborate with National control programmes to conduct genomics surveillance of malaria to inform malaria control and elimination policies. This study explored genomic researchers' views on engaging with national control programmes to aid malaria elimination efforts in Africa.
Methods
This research employed an exploratory qualitative approach to investigate the views and experiences of malaria genomics researchers across 16 member countries of the Pathogen Genomic Diversity Network Africa (PDNA). In-depth interviews were conducted with each PDNA Principal Investigator, which were recorded, and transcribed verbatim. Subsequently, the data were analysed thematically with NVivo 12 qualitative data analysis software.
Results
The study revealed that majority of malaria genomics researchers focused on understanding the genetic composition and adaptation of the malaria parasite, its vector, and human host. Their investigations delved into areas such as drug and insecticide resistance, parasite evolution, host interactions, human host susceptibility to malaria, diversity of vaccine candidates, and molecular surveillance of malaria. Challenges included limited funding, lack of interest and capacity among National Malaria Control Programmes (NMCP) to use research evidence effectively, and difficulties in communicating data implications to policymakers due to the absence of WHO-certified use cases. Despite these obstacles, researchers expressed a keen interest in forming partnerships with NMCPs to integrate genetic data into malaria control efforts in Africa. They also stressed the importance of enhancing researchers' ability to communicate findings to policymakers and local communities through policy briefs and innovative communication strategies.
Conclusion
The study underscores the need to strengthen partnerships between genomic researchers and NMCPs to support malaria elimination in Africa. Furthermore, researchers should create practical frameworks for easy integration into WHO reporting formats to facilitate the use of molecular and genomic data in malaria control programme decision-making.

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ANIMALS, PLANTS, MICROORGANISMS

Assessing risks associated with large-scale adoption of CRISPR gene-edited crops

Review Article
Priya Bansal & Navjot Kaur
Journal of Crop Science and Biotechnology, (2024). 27 November 2024
https://doi.org/10.1007/s12892-024-00273-0**.**
Abstract
CRISPR gene-edited crops have demonstrated promising potential to ensure food security in a world witnessing extreme climate change and exponential population growth. This study assesses the risks associated with the widespread adoption of CRISPR crops in agricultural fields. Major ecological risks include the evolution of superweeds, rise in resistant pest populations and monocultures, loss of biodiversity, food chain disruption, and the possibility of gene flow. The authors identify four key factors to ensure sustainable intensification with CRISPR crops: (i) unconventional approaches to design and develop new gene-edited crop strains that incorporate fail-safe mechanisms, (ii) early stakeholder engagement on technology development and deployment built on the foundation of effective science communication to identify and address pain-points, (iii) strategically planned and monitored deployment of new crop strains to prevent identified risks, and (iv) regulatory frameworks to prevent international and national monopoly of few entities in gene-edited crop development and distribution.

Early Detection of Wildlife Disease Pathogens Using CRISPR-Cas System Methods

Review Articles
Adam A. Pérez, Guelaguetza Vazquez-Meves, and Margaret E. Hunter
The CRISPR Journal, Volume 7, Issue 6 / December 2024, Pages:327–342 Published Online:31 October 2024
Abstract
Wildlife diseases are a considerable threat to human health, conservation, and the economy. Surveillance is a critical component to mitigate the impact of animal diseases in these sectors. To monitor human diseases, CRISPR-Cas (clustered regularly interspaced short palindromic repeats-CRISPR-associated protein) biosensors have proven instrumental as diagnostic tools capable of detecting unique DNA and RNA sequences related to their associated pathogens. However, despite the significant advances in the general development of CRISPR-Cas biosensors, their use to support wildlife disease management is lagging. In some cases, wildlife diseases of concern could be rapidly surveyed using these tools with minimal technical, operational, or cost requirements to end users. This review explores the potential to further leverage this technology to advance wildlife disease monitoring and highlights how concerted standardization of protocols can help to ensure data reliability.

Enhancing Animals is "Still Genetics": Perspectives of Genome Scientists and Policymakers on Animal and Human Enhancement
Research Article
Rebecca L. Walker, Zachary Ferguson, Logan Mitchell & Margaret Waltz
AJOB Empirical Bioethics, DOI: 10.1080/23294515.2024.2441688. 18 December 2024
Abstract
Background: Nonhuman animals are regularly enhanced genomically with CRISPR and other gene editing tools as scientists aim at better models for biomedical research, more tractable agricultural animals, or animals that are otherwise well suited to a defined purpose. This study investigated how genome editors and policymakers perceived ethical or policy benefits and drawbacks for animal enhancement and how perceived benefits and drawbacks are alike, or differ from, those for human genome editing. *Methods:* We identified scientists through relevant literature searches as well as conference presentations. Policymakers were identified through rosters of genome editing oversight groups (e.g., International Commission on the Clinical Use of Human Germline Genome Editing, World Health Organization) or efforts aimed at influencing policy (e.g., deliberative democracy groups). Interviews covered participants' views on ethical differences between interventions affecting somatic or germline cells and distinctions between using gene editing for disease treatment, prevention, and enhancement purposes. *Results:* Of the 92 participants interviewed, 81 were genome editing scientists, and 33 were policymakers, with 22 interviewees being both scientists and policymakers. Multiple areas were identified in which the ethical implications of genomic enhancements for nonhuman animals differ from those for human animals including with respect to experiential welfare; germline edits; environmental sustainability; and justice. *Conclusions:* Overall, respondents viewed that animal enhancement is unburdened by the ethical complexities of human enhancement. These views may be related to participant perceptions of animals' lesser moral status and because germline editing in animals is common practice.

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Month in Review – *Milestones, Strategic Announcements, Analysis, Guidance*
Organization Watch – *Selected Events*
Organization Watch – *Selected Announcements*
Journal Watch – *Spotlight Articles, Thematic Sections*
Journals/Pre-Print Sources Monitored
Institutions/Organizations Monitored

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Institutions/Organizations – Monitored

We recognize this listing is incomplete, unbalanced and skewed to the Global North…please help us make it more complete, more inclusive, and more useful by recommending additional organizations/ institutions/programs to monitor.

Academy of Medical Sciences [UK]
https://acmedsci.ac.uk/

Africa CDC - Institute of Pathogen Genomics [IPG]
https://africacdc.org/institutes/ipg/

African Society of Human Genetics
https://www.afshg.org/

Paul G. Allen Frontiers Group
https://alleninstitute.org/news-press/

American Board of Medical Genetics and Genomics (ABMGG)
http://www.abmgg.org/pages/resources_appeal.shtml

American College of Medical Genetics and Genomics
https://www.acmg.net/

American Society for Gene and Cell Therapy [ASGCT]
https://asgct.org/

American Society of Human Genetics (ASHG)
http://www.ashg.org/

ARM [Alliance for Regenerative Medicine]
https://alliancerm.org/press-releases/

ARRIGE
https://www.arrige.org/

Australian Genomics
https://www.australiangenomics.org.au/

Bespoke Gene Therapy Consortium (BGTC)
https://ncats.nih.gov/research/research-activities/BGTC

BMGF - Gates Foundation [
https://www.gatesfoundation.org/ideas/media-center

Bill & Melinda Gates Medical Research Institute
https://www.gatesmri.org/news

Broad Institute of MIT and Harvard
https://www.broadinstitute.org/

CDC – Office of Genomics and Precision Public Health
https://www.cdc.gov/genomics/default.htm

Center for Genetics and Society [USA]
www.geneticsandsociety.org

Center for the Ethics of Indigenous Genomic Research [CEIGR] – University of Oklahoma
https://www.ou.edu/cas/anthropology/ceigr

Center for ELSI Resources and Analysis (CERA)
https://elsihub.org/about/our-mission

Chan Zuckerberg Initiative [to 18 Jan 2025]
https://chanzuckerberg.com/newsroom/

Francis Crick Institute
https://www.crick.ac.uk/news-and-reports

FDA Cellular & Gene Therapy Guidances
https://www.fda.gov/vaccines-blood-biologics/biologics-guidances/cellular-gene-therapy-guidances

The Genomic Medicine Foundation
https://www.genomicmedicine.org

Global Alliance for Genomics and Health
https://www.ga4gh.org/

Genetic Alliance
https://geneticalliance.org/about/news

Genomics England
https://www.genomicsengland.co.uk/

Genetics Society of America (GSA)
http://genetics-gsa.org/

Global Genomic Medicine Consortium [G2MC]
https://g2mc.org/

Global Observatory for Genome Editing
https://global-observatory.org/

HHMI - Howard Hughes Medical Institute [to 30 Aug 2023]
https://www.hhmi.org/news

H3Africa
https://h3africa.org/

Human Genome Organization (HUGO)
https://www.hugo-international.org/

ICH
https://www.ich.org/

Innovative Genomics Institute
https://innovativegenomics.org/about-us/

INSERM [to 30 Aug 2023]
https://www.inserm.fr/en/home/

Institut Pasteur [to 30 Aug 2023]
https://www.pasteur.fr/en/press-area

NIH [to 30 Aug 2023]
http://www.nih.gov/

NIH National Human Genome Research Institute (NHGRI)
https://www.genome.gov/

NIH – All of Us Research Program
https://allofus.nih.gov/news-events/announcements

National Organization for Rare Disorders (NORD)
https://rarediseases.org/news/

Nuffield Council on Bioethics [to 30 Aug 2023]
https://www.nuffieldbioethics.org/news

Penn Center for Global Genomics & Health Equity [University of Pennsylvania]
https://globalgenomics.med.upenn.edu/index.php

PHG Foundation
https://www.phgfoundation.org

The Royal Society
https://royalsociety.org/

UNESCO–The World Academy of Sciences
https://twas.org/

Wellcome Sanger Institute
https://www.sanger.ac.uk/

WHO
https://www.who.int/news

WHO - Human genome editing
https://www.who.int/teams/health-ethics-governance/emerging-technologies/human-genome-editing
[last update on page - July 2021]

World Organisation for Animal Health [OIE]
https://www.oie.int/

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Journals/Pre-Print Sources – **Core/Penumbra Journals Monitored**

If you would like to suggest other journal titles to include in this service, please contact David Curry at: david.r.curry@centerforvaccineethicsandpolicy.org

AJOB Empirical Bioethics
https://www.tandfonline.com/toc/uabr21/current

AMA Journal of Ethics
https://journalofethics.ama-assn.org/issue/peace-health-care

American Journal of Human Genetics

https://www.cell.com/ajhg/current

American Journal of Infection Control
http://www.ajicjournal.org/current

American Journal of Preventive Medicine
https://www.ajpmonline.org/current

American Journal of Public Health
http://ajph.aphapublications.org/toc/ajph/current

American Journal of Tropical Medicine and Hygiene
https://www.ajtmh.org/view/journals/tpmd/111/4/tpmd.111.issue-5.xml

Annals of Internal Medicine
https://www.acpjournals.org/toc/aim/current

Artificial Intelligence – An International Journal
https://www.sciencedirect.com/journal/artificial-intelligence/vol/336/suppl/C

BMC Cost Effectiveness and Resource Allocation
http://resource-allocation.biomedcentral.com/

BMC Health Services Research
http://www.biomedcentral.com/bmchealthservres/content

BMC Infectious Diseases
http://www.biomedcentral.com/bmcinfectdis/content

BMC Medical Ethics
http://www.biomedcentral.com/bmcmedethics/content

BMC Medicine
http://www.biomedcentral.com/bmcmed/content

BMC Pregnancy and Childbirth
http://www.biomedcentral.com/bmcpregnancychildbirth/content
(Accessed 16 Nov 2024)

BMC Public Health
http://bmcpublichealth.biomedcentral.com/articles

BMC Research Notes
http://www.biomedcentral.com/bmcresnotes/content

BMJ Evidence-Based Medicine
https://ebm.bmj.com/content/29/5

BMJ Global Health
https://gh.bmj.com/content/9/10

Bulletin of the World Health Organization
https://www.ncbi.nlm.nih.gov/pmc/issues/471305/

Cell
https://www.cell.com/cell/current

Clinical Pharmacology & Therapeutics
https://ascpt.onlinelibrary.wiley.com/toc/15326535/current

Clinical Therapeutics
http://www.clinicaltherapeutics.com/current

Clinical Trials
https://journals.sagepub.com/toc/ctja/21/6

Contemporary Clinical Trials
https://www.sciencedirect.com/journal/contemporary-clinical-trials/vol/146/suppl/C

The CRISPR Journal
https://www.liebertpub.com/toc/crispr/7/5

Current Genetic Medicine Reports
https://link.springer.com/journal/40142/volumes-and-issues/11-3

Current Medical Research and Opinion
https://www.tandfonline.com/toc/icmo20/current

Current Opinion in Infectious Diseases
https://journals.lww.com/co-infectiousdiseases/pages/currenttoc.aspx

Current Protocols in Human Genetics
https://currentprotocols.onlinelibrary.wiley.com/journal/19348258

Developing World Bioethics
https://onlinelibrary.wiley.com/toc/14718847/current

EMBO Reports
https://www.embopress.org/toc/14693178/current

Emerging Infectious Diseases
http://wwwnc.cdc.gov/eid/

Ethics & Human Research
https://onlinelibrary.wiley.com/toc/25782363/current

Ethics & International Affairs
https://www.cambridge.org/core/journals/ethics-and-international-affairs/latest-issue

Ethics, Medicine and Public Health
https://www.sciencedirect.com/journal/ethics-medicine-and-public-health/vol/31/suppl/C

The European Journal of Public Health
https://academic.oup.com/eurpub/issue/34/5

Expert Review of Vaccines
https://www.tandfonline.com/toc/ierv20/current

Frontiers in Medicine
https://www.frontiersin.org/journals/medicine/volumes?volume-id=1237

Gene Therapy – Nature
https://www.nature.com/gt/volumes/31/issues/11-12

Genetics in Medicines
https://www.sciencedirect.com/journal/genetics-in-medicine/vol/26/issue/11

Genome Medicine
https://genomemedicine.biomedcentral.com/articles

Global Health Action
https://www.tandfonline.com/toc/zgha20/current?nav=tocList

Global Health: Science and Practice (GHSP)
http://www.ghspjournal.org/content/current

Global Public Health
http://www.tandfonline.com/toc/rgph20/current

Globalization and Health
http://www.globalizationandhealth.com/

Health and Human Rights
https://www.hhrjournal.org/volume-26-issue-1-june-2024/

Health Economics, Policy and Law
https://www.cambridge.org/core/journals/health-economics-policy-and-law/latest-issue

Health Policy and Planning
https://academic.oup.com/heapol/issue/39/9

Health Research Policy and Systems
http://www.health-policy-systems.com/content

Human Gene Therapy
https://www.liebertpub.com/toc/hum/35/19-20

Human Vaccines & Immunotherapeutics (formerly Human Vaccines)
https://www.tandfonline.com/toc/khvi20/20/1?nav=tocList

Immunity
https://www.cell.com/immunity/current

Infectious Agents and Cancer
http://www.infectagentscancer.com/

Infectious Diseases of Poverty
http://www.idpjournal.com/content

International Health
https://academic.oup.com/inthealth/issue/16/6

International Human Rights Law Review
https://brill.com/view/journals/hrlr/13/1/hrlr.13.issue-1.xml

International Journal of Community Medicine and Public Health
https://www.ijcmph.com/index.php/ijcmph/issue/view/118

International Journal of Epidemiology
https://academic.oup.com/ije/issue/53/5

International Journal of Human Rights in Healthcare
https://www.emerald.com/insight/publication/issn/2056-4902/vol/17/iss/4

JAMA
https://jamanetwork.com/journals/jama/currentissue

JAMA Health Forum
https://jamanetwork.com/journals/jama-health-forum/issue

JAMA Pediatrics
https://jamanetwork.com/journals/jamapediatrics/currentissue

JBI Evidence Synthesis
https://journals.lww.com/jbisrir/Pages/currenttoc.aspx

Journal of Adolescent Health
https://www.jahonline.org/current

Journal of Artificial Intelligence Research
https://www.jair.org/index.php/jair

Journal of Community Health
https://link.springer.com/journal/10900/volumes-and-issues/49-5

Journal of Current Medical Research and Opinion
https://www.cmro.in/index.php/jcmro/issue/view/75

Journal of Empirical Research on Human Research Ethics
http://journals.sagepub.com/toc/jre/current

Journal of Epidemiology & Community Health
https://jech.bmj.com/content/78/11

Journal of Evidence-Based Medicine
https://onlinelibrary.wiley.com/toc/17565391/current

Journal of Global Ethics
http://www.tandfonline.com/toc/rjge20/current

Journal of Health Care for the Poor and Underserved (JHCPU)
https://muse.jhu.edu/issue/52935

Journal of Immigrant and Minority Health
https://link.springer.com/journal/10903/volumes-and-issues/26-5

Journal of Medical Ethics
http://jme.bmj.com/content/current

Journal of Patient-Centered Research and Reviews
https://institutionalrepository.aah.org/jpcrr/

The Journal of Pediatrics
https://www.sciencedirect.com/journal/the-journal-of-pediatrics/vol/274/suppl/C

Journal of Pharmaceutical Policy and Practice
https://www.tandfonline.com/toc/jppp20/17/1

Journal of Public Health Management & Practice
https://journals.lww.com/jphmp/pages/currenttoc.aspx

Journal of Public Health Policy
https://link.springer.com/journal/41271/volumes-and-issues/45-3

Journal of the Royal Society – Interface
https://royalsocietypublishing.org/toc/rsif/current

Journal of Virology
http://jvi.asm.org/content/current

The Lancet
https://www.thelancet.com/journals/lancet/issue/current

The Lancet Child & Adolescent Health
https://www.thelancet.com/journals/lanchi/issue/current

Lancet Digital Health
https://www.thelancet.com/journals/landig/issue/current

Lancet Global Health
https://www.thelancet.com/journals/langlo/issue/current

Lancet Infectious Diseases
https://www.thelancet.com/journals/laninf/issue/current

Lancet Public Health
https://www.thelancet.com/journals/lanpub/issue/current

Lancet Respiratory Medicine
https://www.thelancet.com/journals/lanres/issue/current

Maternal and Child Health Journal
https://link.springer.com/journal/10995/volumes-and-issues/28-11

Medical Decision Making (MDM)
http://mdm.sagepub.com/content/current

Molecular Therapy
https://www.cell.com/molecular-therapy/current

Nature
https://www.nature.com/nature/volumes/633/issues/8038

Nature Biotechnology
https://www.nature.com/nbt/volumes/42/issues/11

Nature Genetics
https://www.nature.com/ng/volumes/56/issues/11

Nature Human Behaviour
https://www.nature.com/nathumbehav/volumes/8/issues/10

Nature Medicine
https://www.nature.com/nm/volumes/30/issues/11

Nature Reviews Drug Discovery
https://www.nature.com/nrd/volumes/23/issues/11

Nature Reviews Genetics
https://www.nature.com/nrg/volumes/25/issues/11

Nature Reviews Immunology
https://www.nature.com/nri/volumes/24/issues/11

New England Journal of Medicine
https://www.nejm.org/toc/nejm/medical-journal

NEJM Evidence
https://evidence.nejm.org/toc/evid/current

njp Vaccines
https://www.nature.com/npjvaccines/

Pediatrics
https://publications.aap.org/pediatrics/issue/154/Supplement%203

PharmacoEconomics
https://link.springer.com/journal/40273/volumes-and-issues/42-11

PLoS Biology
https://journals.plos.org/plosbiology/

PLoS Genetics
https://journals.plos.org/plosgenetics/

PLoS Global Public Health
https://journals.plos.org/globalpublichealth/search?sortOrder=DATE_NEWEST_FIRST&filterStartDate=2021-10-01&filterJournals=PLOSGlobalPublicHealth&q=&resultsPerPage=60

PLoS Medicine
https://journals.plos.org/plosmedicine/

PLoS Neglected Tropical Diseases
http://www.plosntds.org/

PLoS One
http://www.plosone.org/

PLoS Pathogens
http://journals.plos.org/plospathoge ns/

PNAS - Proceedings of the National Academy of Sciences of the United States
https://www.pnas.org/toc/pnas/121/46

PNAS Nexus
https://academic.oup.com/pnasnexus/issue/3/10

Preventive Medicine
https://www.sciencedirect.com/journal/preventive-medicine/vol/187/suppl/C

Proceedings of the Royal Society B
https://royalsocietypublishing.org/toc/rspb/current

Public Health
https://www.sciencedirect.com/journal/public-health/vol/236/suppl/C

Public Health Ethics
http://phe.oxfordjournals.org/content/current

Public Health Genomics
https://karger.com/phg/issue/27/1

Public Health Reports
https://journals.sagepub.com/toc/phrg/139/6

Qualitative Health Research
https://journals.sagepub.com/toc/QHR/current

Research Ethics
http://journals.sagepub.com/toc/reab/current

Reproductive Health
http://www.reproductive-health-journal.com/content

Revista Panamericana de Salud Pública/Pan American Journal of Public Health (RPSP/PAJPH)
https://www.paho.org/journal/en

Risk Analysis
https://onlinelibrary.wiley.com/toc/15396924/current

Risk Management and Healthcare Policy
https://www.dovepress.com/risk-management-and-healthcare-policy-archive56

Science
https://www.science.org/toc/science/current

Science and Engineering Ethics
https://link.springer.com/journal/11948/volumes_-and-issues/30-6

Science Translational Medicine
https://www.science.org/toc/stm/current

Social Science & Medicine
https://www.sciencedirect.com/journal/social-science-and-medicine/vol/360/suppl/C

Systematic Reviews
https://systematicreviewsjournal.biomedcentral.com/articles

Theoretical Medicine and Bioethics
https://link.springer.com/journal/11017/volumes-and-issues/45-5

Travel Medicine and Infectious Diseases
https://www.sciencedirect.com/journal/travel-medicine-and-infectious-disease/vol/61/suppl/C

Tropical Medicine & International Health
https://onlinelibrary.wiley.com/toc/13653156/current

Vaccine
https://www.sciencedirect.com/journal/vaccine/vol/42/issue/25

Vaccines
https://www.mdpi.com/journal/vaccines

Value in Health
https://www.valueinhealthjournal.com/current

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Pre-Print Servers

arxiv
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[Filters: Emerging Technologies; Neural and Evolutionary Computing; Computers and Society; Genomics; Neurons and Cognition; Populations and Evolution; Other Quantitative Biology; General Economics]

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https://gatesopenresearch.org/browse/articles

medRxiv
https://www.medrxiv.org/content/about-medrxiv
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OSF Pre-prints
https://osf.io/preprints/discover?provider=OSF&subject=bepress%7 CLife%20Sciences
[Provider Filter: OSF Pre-prints Subject filters: Medicine and Health Sciences Format Filter: Pre-

Wellcome Open Research
https://wellcomeopenresearch.org/browse/articles

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