

Chapter 9: Ethical, Legal, and Social Implications (ELSI) of artificial intelligence-driven genomic medicine: Navigating privacy, consent, and bias

9.1 Introduction

As the speed of technology and innovation rapidly progresses on a slope inclined towards vertical, the intersection of these advancements with healthcare is inevitable. Recognized as one of the dynamics that is rapidly transforming the healthcare ecosystem, both the reagents and the instruments used to implement medical practices are continuously under improvement. One of the most in vogue technologies which is part of this transformation is Artificial Intelligence (AI). Since the late 1890s, as technology posed opportunities for better medical diagnosis and treatment, it is observed that physicians and engineers have joined forces to validate the potential of emerging technologies. Media discourse, government and intergovernmental reports, and the growing engagement in certain social spheres influence the envisioning of biotechnologies, but more generally, of genomics as an enabling science. Precision medicine, a promising outcome of the biotechnology revolution, becomes the prime example of how genomics and its offspring shape the directions of scientific research, the economy, public policies, and society at large. AI at the borders of genomics and medicine is envisioned as a new revolutionary science, paving the way to solutions for age-old and large-scale medical problems, as well as remolding patient care by predicting, diagnosing, and treating diseases, and preventing new health concerns. However, the discussions are dominated by the benefits and risks associated with the advance of AI in public arenas with little thought for the interests of future intended stakeholders, namely clinicians and patients. But the diffusion of emerging technologies is systemically complicated and open to interpretation. Ethical worlds are informed mainly by the politically active members of society, with important implications for the technological future. A range of

interpretative frameworks broach the implications of AI practices in genomics and medicine, marginalizing at the outset those concerns that do not align with stated objectives, thus designing the path that biotechnological applications can take and limiting the social forces that can shape it. Social and legal facets pertaining in the advance and infusion of AI in genomics and medicine are finely etched, giving spotlight to the potentiality of lay preferences and concerns to influence the ethical critique of the envisioned science and technology and to offer new ethical and regulatory directions.

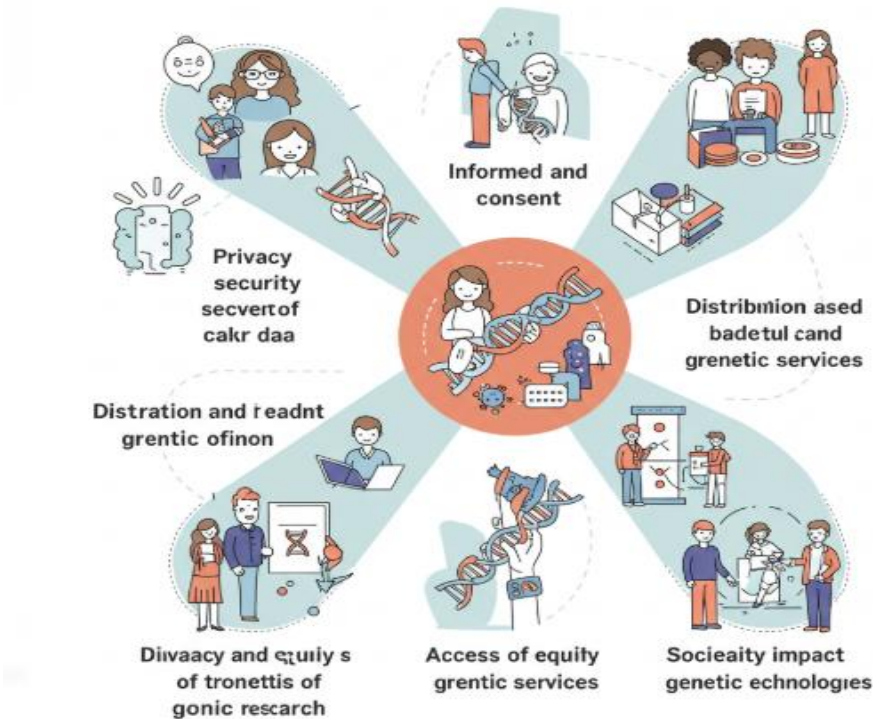


Fig 9.1: Ethical, legal, and social implications (ELSI) of genomics

9.2. Overview of AI in Genomic Medicine

The integration of artificial intelligence (AI) with genomic medicine has been the subject of significant discussion. The promise, challenges, and ethical implications of the AI–genomic medicine relationship are examined. AI technologies are empowering health professionals to more effectively analyze genetic data that is growing ever more complex. Since traditional methods of analyzing genetic data are no longer able to keep up with the mass of information generated, AI tools are expected to grow ever more useful. Through the analysis of large genetic databases and electronic health records, AI can help identify hereditary causes of disease and provide personalized treatment plans.

As such, AI is expected to bring about a transformative revolution in the field of precision medicine, by sifting through complex genetic data in record time and enhancing diagnostic accuracy. It is also anticipated that AI will improve health outcomes and access to medical treatments, high-quality medical care, while also reducing costs. Yet, AI is expected to face a series of challenges as they enter genomics in medicine. A main challenge is the vast quantity of patient-generated genomic data, which will strain even the most robust AI algorithm. Electronic health records will use up too much time to sort through stacked records that go as far back as birth. Another challenge is the slow maturity of the AI-based disease predictive models. Although many chronic conditions can be predicted from an individual's genetics, the complexity of the human genome can also confound such predictions. It is unclear how much the machine will know regarding the genetic underpinnings of the traits it studies. Additionally, there is worry that the data fed into the AI system will contain biases that fail to properly represent the population from which they come. Another problem is that not all people will have access to such technology, meaning the people who benefit the most may not be the people who need it the most. Finally, legal and ethical norms are all but absent in the development, testing, and deployment of AI disease predictors. All these issues are expected to surface as AI comes on-line and promises to complicate their integration into the field of genomic medicine. On the other hand, the potential of AI in disease prediction and prevention is considerable. With the development of AI, it will be more accurate in assessing a person's genetic risk for developing a range of diseases, and more effective at suggesting preventive treatments (Nanan & Chitta, 2022; Recharla et al., 2023).

9.3. Ethical Considerations

AI-driven genomic medicine is full of promise but also brings challenges. On the potential side, advances in genomics, bioinformatics, data science, and artificial intelligence (AI) are driving the creation of sophisticated methods and tools for extraction and analysis of genetic data in their far-reaching complexities. The rapid pace of these advancements is fueling hope that AI analytics will soon translate into clinical benefits, including more precise diagnoses, better prognostics, and more targeted treatment.

In order to deliver cross-disciplinary recommendations, ethical frameworks from bioethics, as well as practices from legally binding regulations in healthcare and public health domain, are invoked. This includes deontological theory and consequentialism from the broader bioethical numinous, but also more specific regulation processes as an outcome and expression of the application of bioethical principles to medical practices. This diversity in perspective is aimed at fostering interdisciplinary understanding of

ethical dilemmas and challenges that would arise (Recharla et al., 2023; Pandiri et al., 2023).

AI-driven genomic medicine needs to ensure fair principles of autonomy, beneficence, non-maleficence, and justice. Transparency and informed consent, and with them patient's autonomy, are the first that springs into light when it comes, on one hand, to distinguishing uncharted black boxes and, on the other, to transformation of data into valuable insights and medical decisions. The challenge is going to be made by complex AI algorithms tangled with medical data, as the data analytics is far from simple with complicated models and algorithms. Fertile grounds for tensions are the rules requiring the detailness of the informed consent forms and at the same time, the ambiguity spoken of the analytics and data processing.



Fig 9.2: Ethical considerations

9.3.1. Autonomy and Informed Consent

Introduction of artificial intelligence (AI) in genomic medicine generates the new ménage à trois of healthcare, consisting of the patient, the physician, and the machines. Physicians traditionally act on behalf of the patient, gaining sufficient medical knowledge to autonomously select the treatments on which they inform the patient. Informed consent to the patient is typically not requested nor pursued. The research of public and big data provides the basis to a new statistical model, also adapted by pharma

companies for a market offer. Learning all available data, the physician does not give rise to informed medical consent but rather acts as an expert salesman, under the appearance of neutrality. This conflict of interests can undermine transparency, patient trust and rights. Therefore, this would lead to an exclusion/selection bias, potentially amplified by the autonomous learning mechanisms of the machine itself. Dissent to the use of AI by patients could reverberate into a systematic selection bias, resulting in potential erroneous conclusions in medical research. As also proven by the legal implications of the negligence of physicians towards AI, it seems appropriate to consider how AI can concretely integrate into the proposal for informed consent to treatment, responding to the dilemma between the patient's self-determination and his right to health and the best available care. The result is a set of key proposals for informed consent in the era of the triangular therapeutic alliance between physician, patient, and artificial intelligence and for its legislative enforcement, raising specific technical issues to currently unaddressed aspects of AI in genomics.

In healthcare, informed consent (IC) is the ethical and legal requirement for transparent communication to the patient practitioner of their health status, diagnoses, risks, and benefits of treatment options as well as the costs and liabilities associated with these treatments prior to them undergoing the selected procedure. However, technology and big data increase the complexity of the treatments proposed, often exceeding the skills really possessed by the physician. Obtaining genuine IC by patients would involve their understanding of such complexity, as well as the interpretability of the mechanisms of artificial intelligence performed by the machines with which they interface. The options proposed involve treatment- (drugs, gene editing), monitoring- (diagnostics, AI, telemedicine), and care- (robotics) devices. It is often difficult to ensure predictable and reproducible plausibility. Moreover, such plausibility could be even less evidently grasped by the patient if the mechanisms involve machine learning mechanisms. Therefore, probability-based decisions by patients could be based on a misleading well-understanding of the device's mechanisms or concepts, or on a wrong well-understanding of the potential benefits/harms. It could reverberate in a perception of non-declared obligation to accept AI interferences. This would not allow the IC to provide free and informed consensus, with the outcome of information-coercion processing instead of genuine education, defense, and compliance. However, if properly educated and empowered, the patient could fully exploit AI assisting strategies even beyond real medical need, on a voluntary and enlightening choice. Nor AI nor medical professionals should promote or hide non-solicited choices, but rather empower the patient to independently and adequately interact with AI. For patient educational and safety purposes, each device should autonomously inform on one another in an unbiased and symmetric balancer way and support the patient in the scrutiny of the medical notes and treatment recommendations. Contrary to this, AI devices often treat complex and technical medical topics in a simplistic and sometimes misleading way. Women are

typically less addressed but a growing numeric illiteracy by males may equally affect both genders. While acknowledging the challenge in translating knowledge to lay subjects, the interpretation should make an effort to unveil the process underneath the decision taken by the machine. Ethical, Legal, and Social Implications (ELSI) research about AI in genomics should pay more attention to the ethical and legal repercussions of healthcare policy implementations and safety issues about AI interactive strategies. Regarding the implementation of AI-based healthcare, poorly regulated environments may undermine the accuracy and security requirements needed for safe and effective medical procedures. On the other hand, the AI operational strategy is confronted with the litigious limits posed by practitioners and institutions who may not easily support the intervention of AI interference. In support of patient rights and safety, the legal liability legislative frame should also be further detailed on these limits. In this context, explanation of operability, transparency and safety mechanisms should be facilitated to patients, professionals, and institutions. This framework would also stimulate a commitment to the safe and smart co-development of AI devices. Due to the growing medical literacy by the patient, the physician should have the legal obligation to monitor algorithms from external providers, providing consent or refusing those methods that reduce the transparency beyond the professionally significant threshold. On the other hand, AI providers should also offer transparency mechanisms and clear protocols in operation to which the physician can also monitor.

9.3.2. Justice and Equity in Access

Disparities in access to the benefits of AI-driven genomic medicine by population groups represent fundamental ethical, legal, and social concerns and have raised interest among ethicists and health policymakers. As efforts are made worldwide to implement genomic medicine and AI technologies at the patient point of care, this interest will grow further. Will there be equal benefit for all or similar burdens and risks befalling those already struggling to obtain access to essential healthcare? This question is particularly timely and important. Marginalized and underserved multicultural and lower socioeconomic status (SES) groups form the majority of patients striving to gain access to healthcare facilities and show the U.K. and U.S perspectives. At the same time, a quantitative method is introduced to facilitate public discussion and to assist policymakers in addressing justice issues in relation to AI-driven genomic medicine. Issues of justice and equity in access to genomics and genomics-based healthcare ought to be high on the agenda, as only a minority of countries worldwide enable citizens access to personalized and genomic healthcare. A challenge arises for bioethicists to question and advise on whether current policies, guidelines, and activities facilitate or impede equitable benefits and harms of AI-driven genomic medicine. Equally, there is a need to question whether bioethics, as an academic field and in its practical activities, is advocating and

emphasizing the right values and principles to promote fairness and equity in the context of (genomic) medicine and healthcare. Five relevant and different philosophical approaches to distributive justice are distinguished and operationalized. The paper argues that more targeted and segmented efforts are needed to address specific and systemic inequalities in healthcare systems. Attention is given to political and practical implications of considerations for a wide range of stakeholders, including patients, healthcare providers and practitioners, advocates, policymakers, society at large, and bioethicists. Amidst all these issues and debates, it is crucial not to lose sight of the subject of the ethical, legal, and social implications (ELSI) of genomic and post-genomic developments themselves. The paper seeks to emphasize the juxtaposition of ELSI and justice concerns and their policy implications, thereby highlighting their relevance to the evolving field of genomic medicine.

9.3.3. Accountability and Responsibility

Because of the increasing infusion of artificial intelligence into clinical and research activities, it is important to have a dedicated discourse on ethical, legal and social implications of AI-driven genomic medicine. The 20 critical viewpoints presented in this article cover privacy and consent issues, accountability and responsibility, and challenges for biased algorithms, among others. Twenty recommendations for appropriate guidelines and accountability frameworks for developing and deploying AI technologies in genetic and vaccine research and their clinical applications are proposed. There is general agreement that AI will have a transformative effect on our work and health care practices, but to realize the potential of AI in a culture of safety, there is a pressing need to advance relevant tools and knowledge in the measurement, prediction, prevention, and/ or minimization of errors, biases in algorithm, alongside adverse advances.

Because of the multifaceted ethical, legal, and social implications of the use of AI in a healthcare environment, the present article should serve as an encouragement to developers and practitioners of AI technologies working in genetics/ genomics/drug fields to seek more effective ways to determine how these technologies can be used safely and how unintentional harm can be reduced and avoided. Stakeholders, including technicians, companies, healthcare providers, public organizations and patient groups, must work together to help the development, testing, management, control, and use of these new tools. Paired with adaptive, stakeholder-driven guidelines, it would also supply impetuses for more inquiring future research and meta-analytic studies on the AiRLIS. All of us will work and live in the inescapable ubiquity of AI, so let us make sure this new world is reasonable, fair, and free of hidden harm.

9.4. Legal Frameworks

Existing and emerging legal frameworks for AI applications in genomic medicine are discussed, and highlights the potential for legal innovations to improve the regulatory environment. AI systems face unique challenges posed by genomic data, meaning traditional regulatory approaches may fall short of legal governance. The focus is thereon, simultaneously complex data protection laws and wide-ranging patient privacy. Nevertheless, these issues are critical for compliance, thereafter considering various intellectual property concerns as well as the numerous complicated questions surrounding the ownership and usage rights to genomic information, which itself is inherently proprietary and valuable. Subsequently, shifting to a discussion of liability, many have raised concerns that current approaches to determining liability in cases of AI-driven decisions, especially in increasingly complex and opaque machine learning algorithms, may not work well for healthcare contexts – the potential legal difficulties in establishing whether liability lies with hardware or software engineers, clinicians or platform developers, or some other actor, are examined. Finally, it is argued that current legal frameworks, even emerging ones, may not be up to the task of addressing the nuanced scenarios involving AI in healthcare, necessitating adaptive frameworks that can respond to changes in technology. The potential for legal innovations to develop in concert with – and even foster – the safe, equitable use of AI in genomic medicine is ultimately underscored. In the words of Pope: “Grant that I may not so much seek to be understood, as to understand” .

9.4.1. Regulatory Approaches to Genomic Data

Since AI-driven applications in genomic medicine largely rely on computing infrastructure to analyze and make use of genomic data, it is significant to discuss regulatory approaches specifically adapted to genomic data. Genomic-based analyses require a different set of laws and guidelines to dictate the process of collecting, storing, and utilizing the data. Genomic data need to be protected throughout the process and in storage to prevent misuse by opponents; therefore, good storage and communication mechanisms should be put in place. Different approaches have been adopted by various governments, reflecting the diverse stages of progression for genetic research and medicine. These approaches could range from governmental agencies participating in national and international collaborations to relatively standard regulatory bodies giving out general guidelines for industry best practices. The road ahead includes international partners focusing on domestic laws to encourage more sharing of data across borders. If these laws can be harmonized, they will thus facilitate the sharing of data across different jurisdictions. The importance of data sharing in genomics is of critical importance for the advancement of the preventive and precision medicine discussed presently. In

modern law-based economies, the regulatory landscape moves at a slower pace than technological innovations. Therefore, the development of AI in genomic and health technologies demands a combination of rigorous care with a vastly flexible but meaningful legal system that can “future proof” a much diverse variety of rapidly and continuously evolving innovations. As genomic technologies evolve at an ever-faster pace, the associated regulatory measures must necessarily follow suit in order to avoid becoming ineffective or hampering legitimate developments. Conditions should be implemented to ensure that AI-driven applications in genomic medicine run to the highest possible technical and ethical standards. This can involve the development of production protocols that must be followed when designing an AI-driven product in the field of genomic measurements. Other actions could include the industry’s commitment to strictly implement the guidelines set out by the law and regulatory agencies.

It is likely that a great number of research projects will have to apply for ethical clearance for each innovation. Indeed, many academic research groups wishing to set foot in this sphere of innovation must either form collaborations with established companies legally certified for such analyses or navigate intricate spot-testing requirements. Though such testing might ensure the security of patients, it could lead to the exclusion of a wide variety of salutary possible future findings. Moreover, it is unlikely that the mentioned testing infrastructure could adapt to the extremely rapid pace of innovation discussed here. The development of AI technologies will be governed by legitimate rules. To this, there are other socioeconomic considerations evolving with the arrival of AI-driven applications in genomic measurements. Many minor regulatory obstacles could slow down growth. I.e., companies are reluctant to disburse the enormous sums necessary for approval for an extensive classification of their innovative results and practices in the form of medical products, or face hurdles during their application for complex medical transfers. Anti-competitive actions might support revenue growth forcefully by inhibiting pricing, or pressuring colleges and commercial companies to maintain exclusive loyalty to providers. Manufacturers may be driven to not clearly expose their predictions or hazard methodologies for fear of legal repercussions. On the contrary, these hidden workings presumably deliberate to survive the rights to full transparency. Medical and Fitness Insurers may profit from genetically primed customers by denying indemnity and protection. Concerns might escalate surrounding data privacy and the potential misuse of information by administration organizations. This is difficult to manage without the cooperation of important stakeholders, such as research, moral and governmental enforcement agencies. It will be crucial to manage the challenges of competitive excitement so that together we can maximize the benefits of AI-driven technologies in genomic medicine for the betterment of societies.

9.4.2. Intellectual Property Issues

The first topic to be discussed is patent issues of AI-based genomic discovery, from algorithm and biological side. It is not uncommon to discover that multiple parties have overlapping claims on the IP. In the consortium trial, researchers released a database for disease studies based on AI derived methods which showed it is feasible to develop gene perturbation predictors based on in vivo data. Subsequently, researchers found that the method could overlap with their pending patent. On the biological side, there might also exist overlapping patents with different claims. For example, one party gets broad rights to restraints of nature and activity for a large group of structures. However, another would-be machine of care getting narrow rights with respect to only specific critical features. If an overlap exists, the competitiveness and access will be influenced, and even hinder further development regarding the relevant project. Despite the great promise to develop innovative solutions and discoveries in various domains, the potential usage of IP still concerns users and leads to a complicated scenario.

The second IP issue discussed challenges in engaging collaboration and sharing data in AI-driven genomic medicine. It is inspiring that an increasing amount of work on AI applications in genomics and medicine have been conducted and many public resources and databases are released by the research community. On one hand, such an open study boost promotes the research of innovative AI and genomic discovery applications. On the other hand, increasing academics' work might lead to more overlaps in IP claims. The repercussions of such overlaps affect public resources and may narrow the usage of some resources. Beyond patents, rapid growth in other licensing forms might include know-how, database rights and software rights. Furthermore, ownership of the genomic dataset collected for specific AI analysis might also involve IP dose issues. Combining these observations, it is implied that the impact of AI IP might be a significant consideration for developing optimized analysis strategies in genomic medicine. On the other hand, it calls for policy regulations that lead to a balanced strategy to protect but also ensure public access and benefits.

9.4.3. Liability in AI-Driven Decisions

The use of artificial intelligence (AI) technologies in genomic medicine raises complex questions regarding liability for the decisions made by AI systems. This is especially challenging given the “black box” nature of AI decision-making. As such, it can be difficult to determine who is responsible for a decision made by an AI system that may lead to an adverse outcome. In relation to liability for decisions made by AI, questions of accountability in health care practice are being raised. Who is responsible for a harmful outcome resulting from a decision made by an artificial intelligence system, the health care provider who recommended that intervention, the individual or team who

developed and trained the algorithm, the institution using that system, or the circumstances that led to the AI decision being made? The connected issues of liability, accountability and therefore obligation, sit uneasily with the therapeutic expectations that often come with medical practice, and underlie broader legal and ethical questions of justice, error and the operation of medical systems. However, while the increasing use of artificial intelligence systems in health care raises concerns about accountability, the legal frameworks to address liability for the decisions taken by artificial intelligence are not yet well developed, risking undermining patient rights and the ability to exercise effective recourse in the case of a decision. Any evolution of artificial intelligence technology in healthcare necessitates clarity around liability guidelines through comprehensive legal frameworks and regulations. Given the unique black box nature of AI decision-making, the standard of care in medical practice may likewise be challenging, in instances where AI systems are used. This can have far-reaching implications regarding the information that must be shared with the patient for obtaining a properly informed consent, if any action or decision from a healthcare provider is the result of an algorithm. It is argued that, in the event of a bad outcome from an intervention, a heightened standard of liability may be imposed on the individual or team that developed and trained the AI, or the team that implemented and disseminated it, thus slowing down the use of these systems and attempting to standardize this responsibility. Major regulatory vaccinations and input patient for its compounded, Cerra, rado-rolos he contramativind, the Non.

9.5. Social Implications

Artificial intelligence (AI) and machine learning (ML) technologies are rapidly moving into healthcare settings with the promise of improving patient care. Nevertheless, public perceptions aiming to reflect broader social acceptance of AI can vary widely. Similarly, while AI and ML technologies promise to revolutionize genomics leading to better possible care, ethical concerns and worries about the privacy and security of patient data could also foster distrust among both patients and healthcare professionals. It is possible that technological advances can also greatly influence social relations and practices that are unfolding at present but which are difficult to interpret only on the basis of the present situation. The relationship between providers and patients can also be affected by AI-driven genomic medicine. Concerning primary care providers, AI is expected to strongly regulate the way in which they initially choose which patients to investigate and consequently also who to treat.

In addition to such basic considerations, a meaningful way to understand the rapid spread of AI technologies is to consider them in relation to public discourses and collective symbolism which frame cultural responses to various types of change.

Altogether, there is a common agreement that trust is critical to the proper functioning of healthcare systems in general and that enhancing trust between patients and healthcare professionals and institutions is crucial for AI-driven genomic medicine to yield positive social outcomes. For this reason, the wider pluralistic sociocultural and institution-specific responses to AI advances in healthcare and determining what types of procedures and practices are used to foster acceptance are the subject of ongoing and wider research, notably in efforts to explore the local response to AI respiratory technologies.

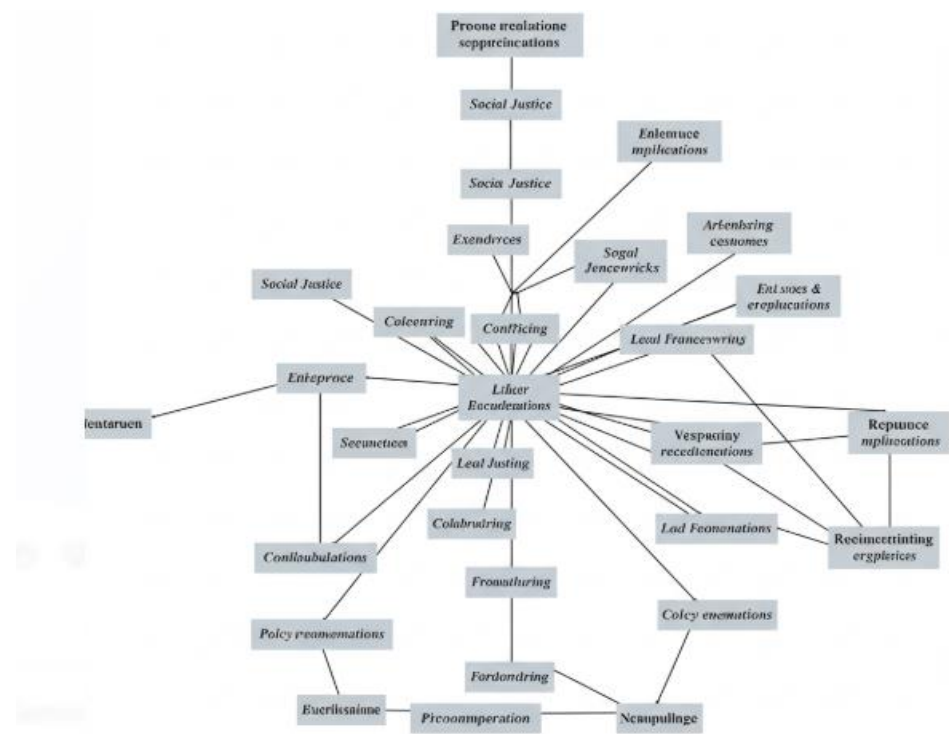


Fig : ELSI concept fields and their interconnections

9.5.1. Public Perception of AI in Healthcare

Artificial intelligence (AI) and genomic medicine technologies are developing at an unprecedented pace. Ethical, Legal, and Social Implications (ELSI) are growing concerns in biomedical research, and genomic medicine is no exception. The successful adoption of AI in genomic medicine requires ongoing public support and engagement, and their views on the ELSI of that use are critical. Here I provide the perspectives of AI experts working on biomedical applications of AI. Health professionals, data scientists

and pharmaceutical researchers highlight widespread concerns, including the protection of personal data, the risk of harm, explainability, transparency, and consent. These ELSI concerns offer crucial guidance to foster public understanding of the opportunities and challenges of AI in genomic medicine.

As AI technologies find increasing applications in healthcare and genomic research, understanding and managing their ethical, legal, and social implications is becoming more critical. Many studies have examined these impacts, including privacy, patient safety and data transparency, which are of concern to multiple stakeholders. AI is expected to transform healthcare delivery and genomic research by facilitating early disease prediction, high-throughput genomic data analysis, drug discovery, treatment personalization, and environmental risk assessment. In consideration of its complex social and ethical implications, researchers have called for the development of a comprehensive regulatory framework to guide the productive and ethical use of AI technologies in these domains. Such a framework would ensure that health systems, institutions and companies implement a set of normative principles and guidelines to ensure the responsible development and deployment of AI applications. Not only does the public need to understand the relevant technology, but it also raises widespread concerns, expectations, hopes and fears about the implementation of AI in health and care. There is also fear. Damage regulation, balancing, managing trust and understanding are critical, if acceptance and support for transformative technology are to be ensured. Not being able to predict and prevent security breaches and data misuse. Uncertainty persists about the quality of existing and to come health care AI applications. Other prominent issues are liability and accountability concerns. The economic interests of the general public are to ensure the accountable use of taxpayer funds and to prevent exploitation. Inequalities can be exacerbated by the implementation of AI technologies as they risk benefiting primarily those who house them. The need to ensure that care providers are fully committed to the safety and well-being of patients and do not rely excessively on machines. Ensuring informed and transparent consent. Patient trust can be eroded if there is suspicion about the use of technology or its role in the decision-making process. Unwillingness also to implement a “black box” technology and support understandable algorithms. Furthermore, the public expects comprehensive information and explanations. Ethical, legal, social and other aspects are considered essential for substantive public discussion.

9.5.2. Impact on Patient-Provider Relationships

Recent developments in the use of data analytics and artificial intelligence (AI) show the potential to revolutionize current medical practices. In the near future, patients may use AI to seek medical advice, monitor their conditions, or carry out routine health checks

independently from healthcare professionals. The implementation of AI in the curation of medical information and delivery of medical advice may have a significant long-term impact on healthcare structures and the work of healthcare professionals. At the same time, current differences in background knowledge and experience among healthcare professionals will affect the capability of integrating AI in daily practice and may exacerbate differences between care provided by different providers. On the other hand, enforcing data and algorithmic transparency in the health domain may facilitate the assessment of AI-generated medical opinions by individuals and professionals. Still, the ethical analysis suggests the need for careful co-development and regulators' supervision to ensure citizens' right to fair and independent healthcare decisions is not compromised.

9.5.3. Community Engagement and Trust

A series of discussions with local communities would help better explain artificial intelligence (AI) and what it might mean for their health care. Engaging a diversity of mutual-networked communities in the design phase may also help identify ethical and social issues that have not been considered, such as algorithms that are racially biased, and establish inclusive strategies and practices to address them. Healthcare institutions should also be transparent about the AI algorithms they implement and what they do, how they are developed, and where they are used, including both clinical applications and administration. A way to foster community trust and gain social licenses for AI may involve establishing partnerships between healthcare institutions and local communities to design, develop, and implement AI technologies mutually. Potential community involvement in AI governance could include participating in advisory groups to decide when, where, and how AI-driven genomic medicine is implemented, and having AI monitored by separate institutions with community involvement.

A set of community engagement strategies can be established to foster trust between public healthcare institutions and local communities while respecting the cultural and regional factors that influence community responses to AI. In addition, AI can be pursued to address unmet community health needs. These may be developed in consultation with local communities and public health consultations, and may include better management of acute public health needs, lifestyle changes that are beneficial according to genomic data, and promoting mental and emotional health. Research and development of interventions may rely on AI to analyze health needs and predict the impact and cost-effectiveness of medical procedures, and local communities may prioritize intervention proposals as decentralized budgets for these purposes. AI-driven advancement in regulatory medical procedures may also develop resource allocation mechanisms. Although the healthcare and research focus would be public, it would be possible to cooperate with a commercially operated social health insurance provider that

specializes in community vigilance, thus enhancing confidence and promoting public acceptance.

9.6. Conclusion

In this essay, the ethical, legal, and social implications of integrating artificial intelligence (AI) with genomic medicine are discussed. An evolving framework of precision medicine, genomic medicine represents a radical shift in the way medicine is practiced. By tapping into the vast amount of genetic and genomic information that illuminates the biological etiology of diseases and the way drugs work with individual patients, genomic medicine promises customized – personal and precise – medical diagnosis and treatment. With the advancement of artificial intelligence (AI), numerous computer algorithms have been developed to help analyze large-scale data in genomic medicine. Genomic medicine will definitely shape up the way medicine is provided to individual patients. There are however numerous emerging challenges facing its wide and safe integration into healthcare, including the interpretation of the massive amount of genomic data and developing genomics professionals in healthcare settings. Japan is not an exception to these problems and the problem must be solved to promote genomic medicine in Japan. While the genomics revolution raises hopes for individualized medicine and improved health outcomes, its clinical application offers many challenges. Despite the rapid technological advancements in AI and readable acquisition of EMR and genome data, it remains difficult to extract meaningful information for clinical practice. The outcomes of independent initiatives or projects to develop AI for genomics have been underwhelming and seldom translate to actionable clinics. To help realize the clinical potential of AI technologies in genomics, several directions in informatics research are discussed in this commentary by members of the AMIA GRAND Workgroup. Most of these technologies are deeply involved in the intersection of genomics and AI, offering a unique perspective on the challenges and opportunities posed.

9.6.1. Future Trends

In the Future Trends subsection, it is explored what may be expected in the converging fields of AI and genomic medicine. As AI technologies continue to develop at a rapid pace, integration with genomic medicine is likely to follow the technological advancements. In the near future, it is expected that AI-driven genome interpretation will further mature and become more prevalent. More sophisticated AI algorithms for genomic interpretation are anticipated to be developed. This development could lead to better understanding of disease mechanisms and—together with public concern—set the

ground for obligations from the part of the scientific community, e.g., to improve hard-to-interpret ('dark') regions of the genome, or to inform patients and the wider public about the risks associated with particular gene variants. Adoption of genome interpretation services into routine clinical care would require further reductions in current barriers related to regulatory, ethical and legal aspects, such as dealing with incidental findings and concerns about insurance. Regulators will need to adapt to rapid technological developments to ensure safe and effective healthcare delivery. Nonetheless, interdisciplinary collaborations will have a primary role in shaping future developments by setting technical standards and regulatory frameworks.

In the longer term, societal perception of AI utilization in medicine will change as a new generation grows up with AI and related technologies. The necessity and/or potential of AI-assisted healthcare and other AI-driven activities, especially in public health domains, will be undertaken by governments, companies and individual citizens. The domain of public health offers a broad range of possibilities for utilizing AI technologies, particularly with respect to setting strategies and tailored actions aimed at improving the overall health of the population. On the other hand, the potential development of such technologies is likely to invite modifications in public life organization and minor adjustments in individual behavior in light of free will, societal control and/or data privacy concerns. Despite the broader interdisciplinary approach in dealing with future societal issues, an open debate in scientific journals should be encouraged, including the involvement of socio-economic researchers, ethicists, legal experts and stakeholders outside the strictly academic community.

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