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Position Statement

Title: The responsible use of artificial intelligence by the human genomics workforce

Document Number: 2025PS03

Publication Date: 12 August 2025

Replaces: Not applicable

Review Date: August 2027

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Introduction

In response to the evolving use of artificial intelligence (AI) in human genomics professional practice, this position statement (PS) aims to guide the use of this technology by the workforce in an ethical and responsible manner, aligned with the values and goals of the Human Genetics Society of Australasia. The PS is primarily aimed at professionals working in the Australasian human genetics and genomics field, regarding responsibilities relative to the community they serve. This PS serves as an introductory high-level document, and individuals should be aware of more specific guidance that may apply to their professional circumstances.

The development of the HGSA position statement on artificial intelligence is informed by key guidance documents and emerging literature that address the ethical and practical implications of AI in healthcare. The Royal College of Pathologists of Australasia (RCPA) *Artificial Intelligence in Pathology: Guiding Principles* (2023) serves as a foundational reference, emphasising the importance of transparency, accountability, and stakeholder engagement—principles that are equally critical in the context of genetics and genomics. Informed by these guidelines, the HGSA recognises the need to address data governance and culturally safe practices, particularly in relation to Indigenous data sovereignty. Contemporary literature underscores the transformative potential of AI in clinical practice while cautioning against the erosion of relational and culturally nuanced aspects of care (Meekins-Doherty et al, 2025). In addition, peer-reviewed studies have explored the ethical dimensions of AI in genomics and healthcare (Kearney et al, 2020)), the potential for bias and inequity in algorithmic decision-making (Nazareth et al, 2021), and the importance of integrating Indigenous perspectives into data governance frameworks (Uhlmann et al, 2020). Together, these resources shape a balanced and ethically grounded position that promotes responsible innovation in AI while safeguarding the rights, values, and voices of all communities served by genetic services.

Glossary

Human genomics	Refers to human genetics and human genomics
Human genomics workforce	Refers to HGSA members and non-members across Australasia who work in the field of human genetics and genomics including, but not limited to clinical practice, laboratory practice, informatics, research and education
Consumer	Refers to members of the community that the human genomics workforce serves including, but not limited to, patients, research participants, students, and the lay community.
End user	Refers to the individual/s who will use a product or service for which AI was used in its development.

AI represents a revolutionary and rapidly advancing technology in human genomics. Although difficult to define, this PS considers AI to include computational systems capable of performing complex tasks that appear to simulate human intelligence, such as learning, reasoning, making decisions and creating content (including text, visual, audio, etc.). AI has many potential applications in areas such as clinical practice and genetic counselling, laboratory sciences, research, and education. Examples include integrating AI into bioinformatic pipelines for laboratory analysis (and re-analysis) of genetic variants; predicting therapeutic responses based on genetic markers; real-time clinical decision support systems to consolidate multiple types of clinical data and facial recognition technologies for syndrome diagnosis; generating educational content for school and university students; and AI-supported genetic counselling tools to assist with aspects of appointments such as information provision and obtaining consent. Generative AI can assist in performing literature reviews and can synthesise complex information into accessible, multilingual educational/communication resources. Such resources can be used by clinicians in patient discussions, can be distributed to patients, and may be of value to non-genetic healthcare professionals to support the mainstreaming of genetic testing.

The integration of AI technology promises to revolutionise how we understand and treat genetic conditions but, to ensure it is used responsibly, it is essential to have a clear-eyed view of its strengths, limitations, risks and ethical considerations in human genomics practice.

Potential advantages and limitations of AI use by the human genomics workforce

- The main advantage AI currently has over human intelligence is the ability to analyse large data sets at high speed and significant scale. Most AI systems process these data to identify patterns and statistical associations, which allow them to generate specific outputs such as summaries, predictions, or text, image, and audio content. Each AI system is designed for tasks, and at the time of this position statement's publication (April 2025) there was no evidence of a 'general AI' with broad, human-like reasoning capabilities.

- The capacity for large data-set analysis has potential strengths including rapid and efficient analysis; a reduction in time-consuming repetitive administrative tasks; scalable and cost-effective practices; and perhaps most importantly, the capacity to undertake analyses otherwise unworkable by the human hand or traditional computing methods.

- Conversely, AI has several limitations and risks associated with its implementation – both practical and ethical. These include, but are not limited to, concerns about privacy and data security; the influence of training data on system accuracy, including the perpetuation or exacerbation of biases, and the generation of false or misleading information (often referred to as 'hallucinations'). These potential problems can, in turn, exacerbate or introduce inequalities in professional practices. Additional important considerations include what the impact of using these technologies might be for informed consent and transparency; and how to locate accountability for AI-driven decisions and outputs within workflows. While these concerns apply broadly, "closed system" type AI, developed for specific clinical tasks using validated datasets, may present different challenges, particularly in terms of adaptability and scope of use. Across all types of AI, human oversight is crucial. Appropriate oversight should include mechanisms for expert review and validation, to ensure the reliability and appropriateness of AI-generated outcomes. Finally, the relational nature of many clinical interactions—particularly those involving sensitive discussions, emotional nuance, or value-based decisions—limits the appropriateness of AI in certain patient-facing settings where human presence, empathy and judgement are essential.

Legal considerations regarding of AI use by the human genomics workforce

- Legally, regulations are needed to protect individuals' genomic information and to govern the use of AI in this sensitive field. AI raises challenges for existing regulatory frameworks, especially in relation to its proprietary or 'black box' aspects. In Australia, software-based medical devices integrating AI are regulated by the *Therapeutic Goods Act 1989* (Cth). In New Zealand, a comparable regime was scheduled to come into effect in 2026, until the *Therapeutic Products Act 2023* was repealed in late 2024. Further, AI tools that use patient data need to comply with privacy legislation in the relevant jurisdiction. Users of these tools should know whether the patient data they contribute is added to a larger dataset, where it is stored, who can access that data, and any potential secondary uses it may be put to. Existing legislative and regulatory tools are piecemeal, not well-suited to emerging technologies, and considerable gaps remain. Even where specific regulation applies, the responsibility for the use of AI in the clinical context and with sensitive information remains with the user.

- AI also raises considerable uncertainty relating to liability for negligence. While there is currently a dearth of case law to draw on, the black box nature of AI-based decision-making is expected to complicate patient consent requirements, and to present challenges in establishing necessary elements of negligence, such as breach and causation.

- The governments of Australia and New Zealand recognise the pressing need to develop regulatory frameworks that address the challenges to patient safety, trust and equity raised by the use of AI in healthcare and health research settings. Recent reviews have focussed on clarifying and strengthening legislation and regulations for AI in health. As such, this is a rapidly evolving area, and imminent changes to the legal and regulatory landscape are to be expected, as are new ethical, practical and social challenges. Ongoing dialogue between politicians, ethicists, legal experts, clinicians, scientists, researchers, technologists, consumers, and patient-led organisations is necessary to navigate these complex issues responsibly.

The following table summarises the key professional obligations and principles to guide professional practices of the HGSA workforce in the context of AI use. It is important to note that individual workplaces, universities and other institutions are also likely to have their own policy regarding AI use, including whether the use of particular AI tools is prohibited. Individuals are therefore advised to be familiar with and uphold the particular AI policies within which they are expected to work.

Professional obligations	Professional principles to guide practice
Understanding	● Be aware that AI technologies are prone to systemic biases, due to inherent computing processes or the training data used. ● Assess the accuracy and limitations, before deploying technologies that use AI for clinical or professional use. ● Ensure human involvement within any workflow using AI, particularly before results are issued, to provide a quality-control measure. ● Commit to continuous education and interdisciplinary collaboration, to maintain adequate human-based expertise in the field. ● Acknowledge that AI might impact Indigenous peoples, particularly in the space of data sovereignty. Ensure that Indigenous stakeholders have control over how their data is collected, used, and shared in alignment with their cultural values, rights, and self-determination. ● Ensure consumer/patient-led organisations are part of any discussions which consider how to navigate these complex issues.
Accountability	● Keep abreast of legal and regulatory obligations, which are likely to change within the short to medium term. ● Establish and maintain clear accountability for information given to end-users. ● Integrate oversight mechanisms and quality-control practices to effectively address misuse, unintended consequences, biases, and limitations of AI tools.
Data Security	● Provide clear information to end users regarding where uploaded information is stored, who has access to it, and how it may be used, and ensure that these comply with institutional and professional data management requirements.
Transparency	● Acknowledge that many AI processes are based on proprietary information and therefore end users must have access to transparent assessments of validation, performance, and limitations from the developer. ● Ensure that consumers are informed as to the role of AI in analysing their data, including how and by whom their data will be stored, and any risks or limitations of AI-based analysis.
Informed consent	● Acknowledge that the use of AI tools adds complexity to informed consent. ● Ensure that consumers have the option to revoke their consent from participation in processes that use AI tools.

References

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