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EDITORIAL

Using Genomic Data in Clinical Decision Making: The Opportunities and Challenges

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Introduction

For centuries, medical care has relied on intuitive analysis and pattern recognition to devise diagnoses and provide appropriate remedies. Modern medicine focuses on the delivery of clinical interventions that are generally trialled and tested to suit the average person. Increasingly, public and professional attention is turning away from a traditional one-size-fits-all model of medical practice, towards further tailored, targeted and person-specific healthcare (Evans et al, 2024). The concept of enhanced medical precision for the individual has coincided with rapid advancements in computational, genomic, digital and artificial intelligence (AI) technologies. We are entering an era of healthcare that is progressively data-driven, drawing insights from a multitude of 'big data' modalities, including genomic, environmental and behavioural inputs.

Genomic data represent an important physiological dimension to be considered in pursuit of health optimisation. The objective is a comprehensive analysis of our genes and their physical expression in order to amplify our knowledge of heritability and its consequences for health. As the knowledgebase expands, clinical care can evolve to incorporate genomic information that will improve patient outcomes. There are several established and emerging aspects of healthcare poised to benefit from genome-informed clinical decision making. These opportunities, and the accompanying challenges to widespread adoption are discussed here.

Opportunities

Genomic data have the potential to address and improve the following core objectives for clinical care: (1) early detection, (2) pre-emptive intervention and (3) treatment optimisation.

Early Detection

Advances in genotyping technologies can facilitate screening for rare and common inherited diseases pre- and post-birth. Non-invasive prenatal testing (NIPT)

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analyses foetal cell-free DNA found in maternal blood (Carbone et al, 2021). This can enable the detection of several chromosomal abnormalities and other inherited conditions in early pregnancy.

Over the past decade, Next Generation Sequencing (NGS) techniques have revolutionised the detection of rare inherited diseases, drastically reducing the diagnostic odyssey experienced by patients and families (Stark and Scott, 2023). Additionally, NGS approaches increasingly contribute to routine newborn screening programmes by employing whole-genome or whole-exome sequencing to screen for hundreds of actionable genetic changes (Ziegler et al, 2025; Genomics England, 2025). This can facilitate prompt diagnosis prior to symptoms emerging. This strategy can also be applied to critically unwell newborns when a genetic aetiology is suspected.

Genotyping can also be used to predict the risk of some monogenic and polygenic conditions. For example, genetic testing for Familial Hypercholesterolaemia (FH) can identify individuals at significant risk of developing cardiovascular disease and can stratify hereditary cancer risk by examining relevant genes such as breast cancer (*BRCA*) (Khoury et al, 2016).

Polygenic risk scores (PRS) are an emerging approach for predicting disposition to common conditions such as cardiovascular disease, diabetes, neurodegenerative conditions and some cancers (Polygenic Risk Score Task Force of the International Common Disease Alliance, 2021). PRS are informed by large-scale genome wide association studies (GWAS) analysed at population level to determine which genes are statistically over-represented in those with the disease of interest. However, PRS alone are far less deterministic than monogenic risk and are seldom used to guide clinical care currently. This may progress as the evidence-base grows and probabilistic algorithms improve by better integrating additional patient health-state parameters.

Genomic techniques can also be used to facilitate the detection of pathogens and the surveillance, analysis and treatment of microbes; in turn limiting outbreaks and minimising antimicrobial resistance. The analysis of viral and patient genomic data provided critical epidemiological information during the COVID-19 pandemic (Robishaw et al, 2021).

Pre-Emptive Intervention

Fundamentally, early detection facilitates early intervention. By incorporating genomic data during clinical decision-making, the focus of medical care can shift towards preventative and pre-emptive measures. Established examples of this include initiating lipid-lowering therapy for patients with FH or rigorous surveillance and risk-reducing surgery for those with a *BRCA* gene mutation (Evans et al, 2024). Pre-emptive intervention can also apply to existing family members and future offspring and genomic data might thus be used to inform family planning and genetic testing.

Treatment Optimisation

The application of genomics for treatment optimisation is most advanced in oncology. Genotyping cancer cells enables ultra-targeting and personalisation of chemotherapies (Berger and Van Allen, 2016). Additionally, NGS data can provide diagnostic and prognostic genetic biomarkers to better identify, predict, monitor and treat certain cancers.

Simultaneously, the field of pharmacogenomics i.e., how our genes can affect drug metabolism or drug sensitivity, is expanding towards mainstream implementation (Jordan et al, 2023). Gene-drug relationships for widely prescribed drugs, such as, statins, antidepressants and opiates are well established and peer-reviewed. International guidelines exist to aid the classification of metaboliser status according to genetic variation (Abdullah-Koolmees et al, 2021). Utilising pharmacogenomic data to direct prescribing may reduce the likelihood of adverse reactions, improve therapeutic efficacy and support adherence for many commonly prescribed medications (Swen et al, 2023).

In addition, advances in gene editing hold the potential to definitively treat several chronic and life-limiting conditions such as sickle cell disease (Locatelli et al, 2024).

As outlined here, there are numerous pathways for the application of genomic data for clinical decision-making and improved medical care. By driving early detection, pre-emptive intervention and treatment optimisation, genomics can improve patient health outcomes, increase healthcare efficiency and further facilitate person-centred care. However, to fully realise the benefits of genome-informed healthcare, several challenges require mitigation.

Challenges

Ancestral Representation

Currently, there is significant disparity in ancestral representation across global genomic datasets (Madden et al, 2024). Large-scale studies have predominantly involved populations of European ancestry and as a result, their findings and the clinical benefits derived will be better tailored to the genetic disposition of individuals within those groups. This skew in representation risks deepening existing ethnicity-related health inequity.

Ethical, Legal and Social Implications (ELSIs)

There is a range of ELSIs to consider when generating datasets of genomic information (Roberts et al, 2014). With the expansion of NGS techniques, emerging trends towards newborn sequencing and lifecourse genome biobanks, there are ethical questions surrounding consent, the right-not-to-know and the wider psychosocial effects of genomic information overload. These require further exploration and policy design that is inclusive, prioritises individuals and protects underrepresented groups.

Data Governance

Another challenging element, with potential legal consequences, is the complex governance of big genomic data (Rahimzadeh et al, 2025). As cloud platforms are increasingly used for storage, aspects such as data security, privacy, legislation, usage and ownership, including third party access and commercialisation, require careful consideration.

Data Processing

In order to maximise insights, further development of AI software and bioinformatic tools is needed to support discovery and improve the clinical utility of genomic data. Digital technology solutions integrated within electronic healthcare records are essential to support clinical decision-making at point of care (Sharma et al, 2021). In the absence of such infrastructure, there will be limited capacity for genomic data to impact routine care. However, the adoption of AI tools may in itself create data governance challenges and unforeseen ELSIs.

Cost

There are significant costs associated with implementing and upscaling the use of genomic data. Testing costs can vary substantially between testing methods. The need to expand laboratory, workforce and data storage capacity requires further expense. Quantifying the health benefits of genomic data can be difficult. Therefore, proving cost-effectiveness is a major challenge that limits policymakers from driving widespread implementation (Santos Gonzalez et al, 2025).

Workforce Readiness

A skills-gap must be closed to enable the clinical workforce to deliver high-quality genome-informed healthcare. Awareness, knowledge and perception of genomic clinical utility must be mainstreamed away from clinical geneticists towards broader cohorts of primary and secondary care health professionals, which may include expanding both undergraduate and postgraduate curricula (Mackley et al, 2025).

System Capacity

Clinical pathways and modes of working must adapt to accommodate the use of genomic data at scale (Mackley et al, 2025). This requires the development of national guidelines, information technology infrastructure and data governance policies to direct the safe and effective use of genomic data in practice.

Public Engagement

Facilitating public discourse and literacy about genomics is vital for fostering trust and ensuring adequate and equitable interventional uptake (Roberts et al, 2014). Engaging diverse communities will help address misconceptions and align interventions with societal expectations.

Conclusion

The utility of genomic data for clinical application is greatly expanding and several examples have been briefly discussed here. Genomic knowledge and technology represent an additional pathway for enhancing clinical management. Nonetheless, they should not be viewed as a panacea. The clinical application of genomics should complement other vital modalities of patient data including routine biochemical parameters, personal and family history, environmental influences, socioeconomic factors to name a few. In many cases, these data will remain the most reliable predictive or modifiable factors for illness. However, genomics data can provide an important additional layer of medical insight. Conscious innovation and implementation are required for genome-informed healthcare to grow in its capacity to optimise clinical care. Pertinent challenges must continue to be addressed in order to fully realise the benefits equitably.

Key Points

- The potential utility of genomic data for clinical application is expanding.
- Integrating genomic data into routine practice can improve healthcare outcomes by facilitating early detection, pre-emptive intervention and treatment optimisation.
- Genomic knowledge and technology should be applied to integrate and enhance other effective modalities of clinical management.
- For the benefits of genome-informed healthcare to be realised equitably, several challenges need addressing such as improving ancestral representation, demonstrating cost-effectiveness, developing workforce readiness and building system capacity.
- Certain aspects of genome-informed healthcare such as rare disease diagnostics and cancer therapies have established clinical utility.
- Other emerging domains, such as polygenic risk scores and pharmacogenomics signal potential utility within mainstream healthcare.

Availability of Data and Materials

Not applicable.

Author Contributions

JD, JM and VS substantially contributed to the conception of the paper. JD wrote the original draft. VS and JM reviewed and edited subsequent drafts. All authors contributed to important editorial changes in the manuscript. All authors read and approved the final manuscript. All authors have participated sufficiently in the work and agreed to be accountable for all aspects of the work.

Ethics Approval and Consent to Participate

Not applicable.

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Conflict of Interest

Julia Darko has no conflict of interest to declare. Videha Sharma and John H McDermott are co-founders of Fava Health.

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