Precision medicine in Malaysia: Aligning genomics research, policy and translation

Wan Nazatul Shima Shahidan, PhD[1], Mohd Nor Azim Ab Patar, PhD[2], Sarina Sulong, PhD[3], Mohd Nazri Hassan, MPath (Haematology)[4], Irfan Mohamad, MMed (ORL-HNS)[5], Bin Alwi Zilfalil, PhD[3]

[1]Department of Pharmacology, [2]Department of Neurosciences, [3]Human Genome Centre, [4]Department of Haematology, [5]Department of Otorhinolaryngology-Head & Neck Surgery, School of Medical Sciences, Universiti Sains Malaysia, Health Campus, Kubang Kerian, Kelantan, Malaysia

The Integration of Genomics into Malaysian Healthcare

The transition of clinical medicine toward precision medicine represents a fundamental shift in how we conceptualize patient care, moving from empirical, population-level protocols to strategies tailored to individual genetic, clinical and environmental profiles. Following the completion of the Human Genome Project, the clinical focus has shifted from the basic generation of raw sequence data to the practical application of this molecular information at the bedside.[1] Today, genomics is no longer a peripheral research interest, as it is increasingly integrated into routine diagnostics, disease risk stratification and targeted therapeutic selection.

In Malaysia, incorporating genomic medicine into public healthcare is supported by key national strategies, including the 12th Malaysia Plan and the Ministry of Health's Health White Paper.[2] These national policy frameworks are actively translated into clinical action through major nationwide initiatives, including the Academy of Sciences Malaysia Position Paper on the Precision Medicine Initiative published in 2020,[3] the National Policy for Rare Diseases and the MyGenom Project.[4]

The formal development of the National Precision Medicine Implementation Plan commenced in 2025, establishing the essential clinical, laboratory and ethical guidelines required for the clinical application of genomic medicine. This clinical blueprint was recently reinforced by national consultation workshops involving genomic experts and stakeholders across key academic centres, which were jointly organised and co-chaired by the Ministry of Health, the Ministry of Science, Technology and Innovation and the Ministry of Higher Education. Alongside this administrative effort, the MyGenom Project serves as the critical sequencing vehicle focused on mapping local genomes to build a comprehensive, high-resolution national reference database. The completion of Phase 1 of the MyGenom Project from 2024 to 2025 has laid a solid foundation, with Phase 2 planned to run from 2026 to 2028. Engaging with all relevant stakeholders remains essential for the successful execution of the Precision Medicine Action Plan in Malaysia, ensuring a unified direction for genomics-driven healthcare.

The Genomics and Life Sciences (GLS) Research Cluster

The GLS Research Cluster at the School of Medical Sciences, USM, was established in 2023 to support this clinical translation. Although still relatively new, the cluster has developed a clear strategic direction to strengthen genomics and life sciences research within the institution. Based at the Health Campus in Kubang Kerian, Kelantan, this cluster brings together researchers and clinicians from various basic science and clinical departments, such as pharmacology, neurosciences, haematology and paediatrics, alongside the Human Genome Centre and other allied medical and life science disciplines, to study the genetic factors of diseases common in Malaysia.[5]

Since its establishment, the cluster has focused on building a structured research ecosystem through strategic planning, subcluster development, research profiling, enhanced research visibility and interdisciplinary project matching. Through digital platforms like ThinkLab (www.thinklab.my), the cluster provides an interactive portal to highlight research expertise and promote collaboration opportunities, hosting major events such as the Science, Technology, Engineering and Mathematics Next Generation (STEMNEXT) webinar, the Fakultas Kedokteran Universitas Padjadjaran (FK-UNPAD) and GLS-USM Joint Webinar Series and collaborative webinars with the Human Genome Centre and the Malaysian Society of Human Genetics. This early strategic planning ensures that genomic and life sciences research within USM is coordinated, visible and aligned with both institutional priorities and Malaysia's national precision medicine agenda.

By working closely with Hospital Pakar USM (HPUSM), the cluster uses a translational research model that allows laboratory findings to be evaluated in clinical settings, which helps to develop local diagnostic services and support targeted clinical research. In alignment with Malaysia's broader national health agenda, this integrated ecosystem of clinicians, scientists and genomic researchers is well positioned to contribute strategically to national initiatives such as the National Precision Medicine Project and the MyGenom Project. Through its research initiatives, the cluster supports the generation of locally relevant genomic data, facilitates translational and implementation research and strengthens national capacity. This close linkage between academic research, clinical services and patient cohorts enables the development of evidence-based approaches

Corresponding Author: Irfan Mohamad
Email: irfankb@usm.my

tailored to Malaysia's multi-ethnic population, positioning the cluster as an important academic and clinical partner in advancing a sustainable national genomic healthcare framework.

Addressing Rare Diseases, Oncology, Thalassaemia and Neurodevelopmental Disorders

A major clinical objective of this collaborative work is reducing the diagnostic delay for patients with rare genetic conditions. Using whole exome sequencing and whole genome sequencing, USM researchers identify molecular causes for these conditions, which helps clinicians manage patients and advise families on risk.[6]

In oncology, precision medicine has progressed through the clinical application of molecular profiling alongside standard histopathology, which allows clinicians to match specific genetic alterations within a tumour to regulatory-approved targeted therapies.[7] Research within the cluster focuses on identifying and analysing these actionable mutations within the local population, ensuring that treatment decisions are guided by data that accurately represent Malaysian patients. Similarly, in the neurosciences, genomic research is closely aligned with established institutional expertise in molecular genetics, neurogenetics, neuro-oncology and cellular and molecular neuroscience. This includes active research directions involving clinically relevant genes and molecular markers such as SCN1A in epilepsy-related neurogenetic disorders, *IDH1, MGMT, BAX* and *CCND1* (cyclin D1) in glioma and other central nervous system tumours, as well as mitochondrial DNA alterations in brain tumour biology.[8] By linking these molecular findings with clinical phenotypes, neuroimaging features and disease progression, genomic research in neuroscience strengthens its contribution to precision medicine and supports more targeted diagnostic and translational approaches for Malaysian patients.

Thalassaemia remains one of the most important inherited disorders in Malaysia and continues to represent a major public health challenge. Research within the cluster has focused on improving molecular characterisation, carrier detection and genotype–phenotype correlation studies among the local population. These efforts not only strengthen early diagnosis and genetic counselling services but also support the development of more personalised approaches to disease monitoring and management for affected patients and families.[5]

Collectively, the multidisciplinary expertise within the USM GLS Research Cluster places it in a strong position to support Malaysia's broader national precision medicine agenda. By integrating genomic research, clinical data and translational implementation strategies, the cluster contributes to national initiatives aimed at expanding genomic databases, strengthening local diagnostic capabilities and improving equitable access to precision medicine services. Such efforts are essential for ensuring that future national genomic policies and healthcare strategies are informed by evidence that reflects the diversity of the Malaysian population.

Pharmacogenomics and Patient Safety

Pharmacogenomics, which studies how genetic variations influence drug efficacy and adverse reactions, represents one of the most clinically impactful applications of genomic medicine because it provides a clear pathway for translating genetic discovery into daily patient care. Identifying patients at high risk of severe drug reactions or treatment failure is critical for improving therapeutic outcomes across various medical fields. For instance, incorporating pharmacogenomic testing in clinical settings helps predict individual drug responses, which optimises dosing regimens, minimises adverse effects and significantly improves drug safety profiles.[9]

Implementing pre-emptive genetic screening allows clinicians to choose alternative drugs for high-risk patients. This clinical practice protects patients from preventable harm and reduces the hospital costs associated with treating severe drug reactions, which contributes to a more efficient healthcare system.

As one of the emerging centres advancing translational genomic research in Malaysia, the USM GLS Research Cluster is well positioned to support patients within the Ministry of Health Malaysia healthcare system. Through collaborative precision medicine initiatives, the cluster aims to expand access to pharmacogenomic-guided therapies and genomic-informed clinical decision-making in line with ongoing global developments in personalised healthcare.

Bioinformatics, Diagnostics and Data Sovereignty

Managing large-scale genomic data requires secure computing systems and specialised analysis. The bioinformatics resources at USM support the storage, quality control and interpretation of clinical sequencing data.

Integrating next-generation sequencing technologies into clinical workflows is essential for advancing precision medicine across Asia, as demonstrated by regional frameworks that highlight the clinical transition toward genomic-based diagnostics.[10] Through initiatives like the Malaysian Node of the Human Variome Project, USM researchers help build a representative national reference database.[11] This leadership in genetic research is further highlighted by international appointments, such as the designation of local experts to lead global research on diseases like thalassaemia.[5] This database helps clinicians distinguish actual disease-causing variants from harmless genetic variations unique to our population.

Additionally, combining genomic data with routine pathology, haematology and biochemistry improves diagnostic precision. For example, in blood cancers, combining genetic profiling with flow cytometry and cytogenetics provides a clearer prognosis and helps monitor treatment response.

Taken together, these capabilities position the USM GLS Research Cluster as an important contributor to Malaysia's evolving national precision medicine ecosystem. The cluster's strengths in genomic sequencing, bioinformatics infrastructure, clinical data integration and translational research provide a strong platform for supporting nationwide efforts in genomic-based diagnostics and personalised

healthcare implementation. Through collaborations involving clinical institutions, research networks and national genomic initiatives, the cluster contributes towards developing harmonised genomic databases, strengthening local analytical capacity and establishing evidence-based frameworks that facilitate the integration of precision medicine into routine healthcare practice across the country.

Cultivating Genomic Literacy in Medical Education

As genomic tools become part of clinical workflows, medical training must adapt to prepare future clinicians for the demands of personalised medicine. To address existing curricular gaps and the divide between laboratory technology and clinical application, there is a clear need to bridge pharmacogenomics with core medical genetics education. At the School of Medical Sciences in USM, this integration focuses on establishing competency-based frameworks and interprofessional, case-based learning to equip students with practical skills in interpreting genetic variation and navigating genomic technologies.[12]

Specialised programmes such as the Master of Pathology (Medical Genetics) offered by the Human Genome Centre, case-based clinical discussions and DNA Day celebrations further support genomic literacy in medical education. These activities expose medical students and young clinicians to real-world examples of how genomic data are generated, interpreted and applied in clinical decision-making, thereby strengthening their understanding of personalised medicine beyond the formal curriculum.

The goal is to ensure that future doctors can interpret molecular test results, understand their limitations and explain them clearly to patients. Communicating genetic risk requires both scientific understanding and cultural sensitivity. These educational initiatives are directly aligned with Malaysia's national precision medicine agenda, helping to ensure a sustainable pipeline of clinicians who are competent in applying genomic information in routine care. By embedding genomics and pharmacogenomics training within competency-based medical education, the USM GLS Research Cluster contributes directly to build a workforce capable of supporting the implementation of the National Precision Medicine Project, accelerating the transition toward precision medicine at scale.

Ethical, Legal and Social Implications (ELSI)

We must also address the ethical, legal and social challenges associated with genomic data, particularly concerning the practicalities of conducting and reviewing human genomics research within the country. In Malaysia's multi-ethnic society, these ethical oversight processes are crucial for managing data privacy, preventing genetic discrimination and establishing robust institutional frameworks that protect participant autonomy while supporting scientific progress.[13] It is important that these diagnostic advances are accessible to all patient groups, regardless of socioeconomic background, to avoid widening existing health disparities.

At present, the Malaysian Personal Data Protection Act (PDPA) does not explicitly recognise genetic data as sensitive personal data. This regulatory gap leaves the public vulnerable to potential discriminatory misuse, such as employer coercion for genetic testing or the exploitation of genetic results by the insurance industry. As national genomic databases expand, it is critical for Malaysia to establish a comprehensive legislative blueprint. This framework must introduce explicit prohibitions against genetic discrimination, expand statutory definitions to protect genomic data and establish strong supervisory mechanisms to govern research, insurance underwriting, employment and the commercialisation of genetic information.

Furthermore, given that individual institutions often operate under their own research ethics committees and governance structures, greater harmonisation of ethical review processes and genomic consent frameworks is needed among centres actively involved in genomic and precision medicine research. Establishing more standardised approaches to genomic consent, data sharing, and secondary data use would facilitate multicentre collaboration while ensuring consistency in participant protection and regulatory oversight across institutions. It is essential that these diagnostic advances are accessible to all patient groups, regardless of socioeconomic background, to avoid widening existing health disparities.

CONCLUSION

The integration of genomics into Malaysian healthcare represents a fundamental shift toward precision medicine, where clinical care is increasingly guided by molecular and patient-specific data rather than population-based approaches. Supported by national initiatives such as the National Precision Medicine Project and the MyGenom programme, Malaysia is progressively building the infrastructure, policy framework and clinical capacity required for genomics-driven healthcare.

Within this context, the USM GLS Research Cluster serves as a key translational platform, linking genomic research, bioinformatics and clinical practice to generate locally relevant evidence that supports improved diagnosis, risk stratification and therapeutic decision-making across diseases including rare disorders, cancer, thalassaemia and pharmacogenomics. Strengthened by integrated medical education, strategic institutional frameworks like ThinkLab and evolving ethical governance, these efforts collectively reinforce Malaysia's trajectory toward an equitable and sustainable precision medicine ecosystem that is responsive to its diverse population.

REFERENCES

1. Green ED, Gunter C, Biesecker LG, et al. Strategic vision for improving human health at the forefront of genomics. Nature 2020; 586(7827): 1-16.
2. Ministry of Health Malaysia. Health White Paper: A Future-Proof Healthcare System. Putrajaya: MOH; 2023.
3. Academy of Sciences Malaysia. Position Paper on Precision Medicine Initiative in Malaysia. Kuala Lumpur: Academy of Sciences Malaysia; 2020. Available from: https://issuu.com/asmpub/docs/precision_medicine_report.

4. Malaysian Genome and Vaccine Institute. MyGenom Project: National Human Genome Sequencing Initiative. Kajang: National Institutes of Biotechnology Malaysia (NIBM), MOSTI; 2023.
5. The Petri Dish. UNESCO appoints local expert to lead in genetic research on thalassaemia. The Petri Dish. 2023. Available from: https://thepetridish.my/2023/02/06/unesco-appoints-local-expert-to-lead-in-genetic-research-on-thalassaemia/.
6. Shafie AA, Supian A, Ahmad Hassali MA, et al. Rare disease in Malaysia: Challenges and solutions. PLoS One 2020; 15(4): e0230850.
7. Brlek P, Škaro V, Hrvatin N, et al. Advances in Precision Oncology: From Molecular Profiling to Regulatory-Approved Targeted Therapies. Cancers (Basel) 2025; 17(21): 3500.
8. Aziz A, Farizan NH, Patar MNAA, et al. Molecular genetics and neurogenetics in central nervous system tumours: clinical implications for precision medicine in Malaysia. Malays J Med Sci 2024; 31(2): 45-58.
9. Principi N, Petropulacos K, Esposito S. Impact of pharmacogenomics in clinical practice. Pharmaceuticals (Basel) 2023; 16(11): 1596.
10. Phanthunane C, Pongcharoen S, Pannarunothai S, et al. Precision medicine in Asia enhanced by next-generation sequencing: Implications for Thailand through a scoping review and interview study. Clin Transl Sci 2024; 17: e13868.
11. Halim-Fikri H, Etemad A, Abdul Latif AZ, et al. The first Malay database toward the ethnic-specific target molecular variation. BMC Res Notes 2015; 8: 176.
12. Azizova M, Ochilova G, Azimova K, et al. Bridging pharmacogenomics and medical genetics education to prepare students for personalized medicine. Genetics and Molecular Research 2026; 25(1): gmr24124.
13. Zee TW, Abdul Aziz MFB, Wei PC. Ethical challenges of conducting and reviewing human genomics research in Malaysia: An exploratory study. Dev World Bioeth 2024; 24(4): 331-41.