

# Genomic Medicine: Clinical Primer for Primary Care Physicians

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The Singapore National Precision Medicine initiative is a whole-of-government 10-year initiative aiming to generate precision medicine data of up to one million individuals, integrating genomic, lifestyle, health, social, and environmental data. Beyond technologies, routine adoption of precision medicine in clinical practice requires social, ethical, legal, and regulatory barriers to be addressed.<sup>1</sup> At present, Precision Health Research, Singapore (PRECISE) has awarded five Clinical Implementation Pilots (CIPs) in: Breast Cancer, Hereditary & Familial Cancers, Familial Hypercholesterolaemia, Primary Glomerular Diseases, and Pharmacogenomics. These CIPs will seek to embed clinical application of genetic/genomic tests to diagnose, manage, and/or treat a particular patient cohort(s) and/or population for their specific conditions or disease phenotypes.<sup>3</sup> This aligns with the Ministry of Health's Healthier SG strategy, leading to better patient outcomes through early detection, accurate diagnosis, and targeted treatments.

Precision medicine aims to revolutionise and transform healthcare for groups and individuals through early disease detection, refining diagnoses and tailoring treatments. Analysis of large-scale genomic–phenotypic databases is a critical enabler of precision medicine. Although Asia is home to 60 percent of the world's population, many Asian ancestries are under-represented in existing databases, leading to missed opportunities for new discoveries, particularly for diseases most relevant for these populations.

Unlike personal clinical information, genomic information is “shared” with blood relatives (i.e., can be common to members of family) – in a genealogical network. The implications of this for individual consent, and authorisation of access to personal data, has led to an amendment of many countries worldwide to support guidelines for the disclosure of a patient's genetic information to at-risk relatives without consent.<sup>4</sup>

Genomics is defined as “the study of genes, their function and their interaction with all the other genes in the genome and the environment”, and genomic medicine is defined as medicine that “uses genomic information and technologies (e.g., DNA sequencing) to determine an individual's risk, predisposition, diagnosis and prognosis, and the selection and prioritisation of therapeutic options” (e.g., pharmacogenetic testing prior to administration of certain medications).<sup>5</sup>

This issue of the *Singapore Family Physician* aims to provide family physicians with a clinical primer on genomic medicine.

In Unit 1, Ms Jeanette Yuen and A/Prof Joanne Ngeow explore the fundamental principles of clinical genetics and the ethical, legal, and social implications associated with clinical genetic testing.

In Unit 2, Dr Jeremy Hoe, Ms Sharon Pek, and A/Prof Subramaniam Tavintharan elaborate on the importance of early detection and intervention of familial hypercholesterolemia (FH) through genetic testing and cascade screening of families of individuals with FH.

In Unit 3, Dr Rebecca Caesar and A/Prof Joanne Ngeow discuss the need to practise preventative oncology such as germline genetic testing for patients and their families with suspected hereditary cancer syndromes.

In Unit 4, Dr Koh Chee Teck, Ms Ng Jun Li, and A/Prof Ng Kar Hui provide an overview of the role of genetics in glomerular diseases and chronic kidney disease.

In Unit 5, Ms Chua Hui Min and Dr Elaine Lo write about the role of pharmacogenomics in optimising pharmacotherapy and how this is relevant for general practitioners.

In Unit 6, Drs Li Jingmei, Ho Peh Joo, Wong Fuh Yong, and A/Prof Mikael Hartman provide a timely primer for primary care practitioners on breast cancer risk counselling and evaluation.

In Unit 7, Dr Rose Fok and A/Prof Goh Lee Gan discuss the role primary care physicians can play in co-managing patients with genetic conditions.

In this issue, A/Prof Goh Lee Gan has selected ten current readings on topics related to genomic medicine ranging from paediatric conditions to cardiovascular and gastrointestinal issues, and the pitfalls associated with genetic testing.

## REFERENCES

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