Genetics and genomics: a frontier for clinicians

Welcome to the *SMJ's* special issue on genetics and genomics.

The 21st century bears witness to remarkable advancements in genetics and genomics, a field that is evolving at an ever accelerating and breakneck pace over the recent years. Many will attribute the kick-off to when Watson and Crick cracked the genetic code,[1] marking the start of this century's genomic revolution.[2] Undoubtedly, the Human Genome Project, which announced the completion of its first working draft in 2000, was only an early landmark in the journey.[3] The costly endeavour, which mapped the human genome for the first time, has given an introduction to the concept of personalised (precision) medicine.[4,5] Today, next-generation whole genome sequencing is routinely available to those who seek it, and at only a fraction of the cost and time compared to the first genome completed in 2000. Nations have entered the community with their population flagship precision medicine programmes,[6] all built upon next-generation genomic sequencing. Ongoing transformative technological development promises to take us further into the future, and the convergence (or translation) between research and clinical practice is an urgent imperative.

Despite inroads with genomic technology and discoveries, clinical genetics is not a new specialist field. There are robust training programmes for clinicians who choose to specialise in this domain.[7,8] What the new and more accessible technology has afforded is for multiple disease disciplines and specialists now to consider holistically how genetics (and genomics) play a role in the patients they care for. Besides the clarion call for precision medicine,[9] practising clinicians cannot afford to ignore genetics and genomics. Many of their patients will be seeking genomic answers to their health and conditions.

Therefore, with this issue, we hope to raise attention to genetics and genomics, first in the space of rare diseases, where new genomic technology has already started changing practice: prenatal and paediatric genetic diseases,[10,11] inherited eye conditions,[12] cancer genetics,[13] and cardiac and muscular disorders.[14,15] Next, ethical, legal and practice implications are highlighted.[16-18] Case examples in a cancer genetic practice illustrate the necessary considerations.[16] 'Omics in the microbiome' is a topical companion, especially with growing attention regarding the way it affects health systemically.[19] The piece on genomics data science aims to help clinicians understand how it may be possible to navigate large genomic datasets.[20] Finally, we have used gene therapy as a window into the future in this series.[10,12,19]

The future of genomic medicine is already very much at our doorstep. Many areas have not been covered in the current collection. These include: What lies beyond the practice of precision medicine? How can we incorporate other 'omic' technologies, apart from genomes and microbiomes? How can we involve artificial intelligence and machine learning tools? How can disease specialities share practice workflow? How are genomic data kept safe in the hospital? How can societies ensure equitable representation and genomic access across ethnicities, gender and creed? These are complex issues requiring resources for evaluating health policies at the national and international levels, and they go hand in hand as we venture to use genetics and genomes in our clinical practice.

'*This is not the end. It is not even the beginning of the end. But it is, perhaps, the end of the beginning'.*

— Winston Churchill, The Lord Mayor's luncheon, Mansion House, November 1942

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REFERENCES

1. Watson JD, Crick FH. Molecular structure of nucleic acids; a structure for deoxyribose nucleic acid. Nature 1953;171:737-8.
2. Pang T. The impact of genomics on global health. Am J Public Health 2002;92:1077-9.
3. Lander ES, Linton LM, Birren B, Nusbaum C, Zody MC, Baldwin J, *et al*. Initial sequencing and analysis of the human genome. Nature 2001;409:860-921.
4. Langreth R, Waldholz M. Genetic mapping ushers in new era of profitable personal medicines. The Wall Street Journal 1999. Available from: https://www.wsj.com/articles/SB924225073307249185. [Last assessed on 08 Dec 2021].
5. Collins FS, Varmus H. A new initiative on precision medicine. N Engl J Med 2015;372:793-5.
6. Chung BHY, Chau JFT, Wong GK. Rare versus common diseases: A false dichotomy in precision medicine. NPJ Genom Med 2021;6:19.
7. Joint Royal College of Physicians Training Board. Clinical genetics. Available from: https://www.jrcptb.org.uk/specialties/clinical-genetics. [Last assessed on 08 Dec 2021].
8. American Board of Medical Specialties. Medical genetics and genomics. Available from: https://www.abms.org/board/american-board-of-medical-genetics-and-genomics/. [Last assessed on 08 Dec 2021].
9. Precision Health Research, Singapore (PRECISE). Available from: https://www.npm.sg/. [Last assessed on 08 Dec 2021].
10. Koh AL, Jamuar SS. Therapeutics in paediatric genetic diseases: current and future landscape. Singapore Med J 2023;64:7-16.
11. Lim KMX, Mahyuddin AP, Gosavi AT, Choolani M. Genetics in prenatal diagnosis. Singapore Med J 2023;64:27-36.
12. Chan HW, Oh J, Leroy B. Therapeutic landscape for inherited ocular diseases: current and emerging therapies. Singapore Med J 2023;64:17-26.
13. Chiang J, Shaw T, Ngeow J. Understanding cancer predisposition in

Singapore: what's next? Singapore Med J 2023;64:37-44.
14. Zheng Y, Sia CH, Wong RCC, Foo RSY, Lin W, Singh D, *et al*. Hereditary transthyretin amyloid cardiomyopathy. Singapore Med J 2023;64:74-80.
15. Quak ZX, Tan SML, Tan KB, Lin W, Chai P, Ng KWP. A manifesting female carrier of Duchenne muscular dystrophy: importance of genetics for the dystrophinopathies. Singapore Med J 2023;64:81-7.
16. Shaw T, Fok R, Courtney E, Li ST, Chiang J, Ngeow J. Missed diagnosis or misdiagnosis: common pitfalls in genetic testing. Singapore Med J 2023;64:67-73.
17. Chin HL, Goh DL. Pitfalls in clinical genetics. Singapore Med J 2023;64:53-8.
18. Lin J, Ngiam KY. How data science and AI-based technologies impact genomics. Singapore Med J 2023;64:59-66.
19. Kwa WT, Sundarajoo S, Toh KY, Lee J. Application of emerging technologies for gut microbiome research. Singapore Med J 2023;64:45-52.
20. Loh M, Chambers JC. Polygenic risk scores for complex diseases: where are we now? Singapore Med J 2023;64:88-9.

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