It's in our genes

'Genetics' is a listed risk factor for every condition in every medical textbook. While it has long been recognized as a contributing factor in disease, it is often overlooked in practice, as ethical considerations and a general lack of understanding limit its application in clinical medicine.

However, in recent years there have been many examples of its use that have completely revolutionized clinical management. BRCA1 & BRCA2 are now routinely screened in those with a family history of breast cancer. Biopsy with cytogenetics are now essential to the diagnosis and management of various cancers for targeted chemotherapeutic treatment. Stepping outside of oncology, it has also become important for clinicians to consider genetic screening for alpha-1 antitrypsin deficiency in the young 40-year-old non-smoking patient presenting with COPD. Genetics has also enabled the investigation of potentially curative therapeutics for diseases once considered a death sentence. This includes a phase III trial for potentially curative antisense oligonucleotides targeted at the mutant HTT gene in Huntington's disease.¹

This issue of the UWOMJ presents some of the latest developments and potential applications for genetics in clinical practice. Pickering describes cell-free DNA and its now routine use in prenatal testing, while Johnson describes new potential genetic targets for future Alzheimer's disease therapy. Frederick discusses mitochondrial replacement therapy, which like gene editing has a potential role as a therapeutic strategy, but also poses similar ethical considerations. Finally, Jung & Leu discuss genetics in the context of today's era of big data and its potential advantages as well as drawbacks.

From life-threatening conditions like alpha thalassemia major to basic traits like height and weight, genetics has a role in every aspect of human physiology and pathology. It has been less than 70 years since Watson & Crick's seminal 1953 paper provided the first description of DNA, and the explosion of research and discovery in genetics has now begun to demonstrate the revolutionary role that genetic screening and therapy will have in our understanding of human pathology.²

We hope you enjoy this issue of the UWOMJ as we take on this basic science and translational medicine lens to provide you with a glimpse into the exciting upcoming future for genetics in modern medicine.

Stay safe, and all the best,

Dominic Wang
Editor-in-Chief

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2. Watson JD, Crick FH. Molecular structure of nucleic acids; a structure for deoxyribose nucleic acid. *Nature*. 1953;171(4356):737-8. doi: [10.1038/171737a0](https://doi.org/10.1038/171737a0). PubMed PMID: 13054692.

Cover Art Description – Alexandra Zygowska

The cover art displays several tools and equipment associated with clinicians and clinical practice. Physicians employ various utensils to facilitate the physical examination, investigation, and ultimately the diagnosis and treatment of patients. However, proper training is necessary for the appropriate utilization of these tools and for interpretation of meaningful results. The tools themselves do not make a whole physician. A competent physician uses these tools along with their acquired knowledge and clinical experience to guide patient care.