# A SELECTION OF TEN READINGS ON TOPICS RELATED TO GENOMIC EDUCATION 101 FOR PRIMARY CARE

**Some are available as free full text, some require payment**

**Selection of readings made by A/Prof Goh Lee Gan**

## READING 1 – APPROACHES TO GENETIC TESTING AND COUNSELLING IN NEUROFIBROMATOSIS- AND SCHWANNOMATOSIS-ASSOCIATED TUMOURS

**Goetsch Weisman A,[1,2] Weiss McQuaid S,[1,2,3] Radtke HB,[4,5] Stoll J,[6] Brown B,[7] Gomes A.[7] Neurofibromatosis- and schwannomatosis- associated tumours: Approaches to genetic testing and counselling considerations. Am J Med Genet A. 2023 Jul 24. PMID: 37485904.**

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ABSTRACT

Neurofibromatosis (NF) and schwannomatosis (SWN) are genetic conditions characterised by the risk of developing nervous system tumours. Recently revised diagnostic criteria include the addition of genetic testing to confirm a pathogenic variant, as well as to detect the presence of mosaicism. Therefore, the use and interpretation of both germline and tumour-based testing have increasing importance in the diagnostic approach, treatment decisions, and risk stratification of these conditions.

This focused review discusses approaches to genetic testing of NF- and SWN-related tumour types, which are somewhat rare and perhaps lesser known to non-specialised clinicians.

These include gastrointestinal stromal tumours, breast cancer, plexiform neurofibromas with or without transformation to malignant peripheral nerve sheath tumours, gliomas, and schwannomas, and emphasises the need for inclusion of genetic providers in patient care and appropriate pre- and post-test education, genetic counselling, and focused evaluation by a medical geneticist or other healthcare provider familiar with clinical manifestations of these disorders.

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## READING 2 – INTRODUCTORY TUTORIAL ON CARDIOVASCULAR PHARMACOGENETICS FOR HEALTHCARE PROVIDERS

**Oni-Orisan A,[#,1] Tuteja S,[#,2] Hoffecker G,[2] Smith DM,[3,4] Castrichini M,[5] Crews KR,[6] Murphy WA,[7] Nguyen NHK,[8] Huang Y,[8] Lteif C,[8] Cavallari LH,[8] Duarte JD,[8] Friede KA,[9] Tantisira,[10] Aminkeng F,[11,12] Voora D,[13] Whirl-Carrillo M,[14] Luzum JA[#,15,16]; Pharmacogenomics Global Research Network (PGRN) Publications Committee. An Introductory Tutorial on Cardiovascular Pharmacogenetics for Healthcare Providers. Clin Pharmacol Ther. 2023 Aug;114(2):275-287. PMID: 37303270**

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ABSTRACT

Pharmacogenetics can improve clinical outcomes by reducing adverse drug effects and enhancing therapeutic efficacy for commonly used drugs that treat a wide range of cardiovascular diseases. One of the major barriers to the clinical implementation of cardiovascular pharmacogenetics is limited education on this field for current healthcare providers and students. The abundance of pharmacogenetic literature underscores its promise, but it can also be challenging to learn such a wealth of information. Moreover, current clinical recommendations for cardiovascular pharmacogenetics can be confusing because they are outdated, incomplete, or inconsistent. A myriad of misconceptions about the promise and feasibility of cardiovascular pharmacogenetics among healthcare providers also has halted clinical implementation.

Therefore, the main goal of this tutorial is to provide introductory education on the use of cardiovascular pharmacogenetics in clinical practice. The target audience is any healthcare provider (or student) with patients that use or have indications for cardiovascular drugs.

This tutorial is organised into the following six steps: (1) understand basic concepts in pharmacogenetics; (2) gain foundational knowledge of cardiovascular pharmacogenetics; (3) learn the different organisations that release cardiovascular pharmacogenetic guidelines and recommendations; (4) know the current cardiovascular drugs/drug classes to focus on clinically and the supporting evidence; (5) discuss an example patient case of cardiovascular pharmacogenetics; and (6) develop an appreciation for emerging areas in cardiovascular pharmacogenetics.

Ultimately, improved education among healthcare providers on cardiovascular pharmacogenetics will lead to a greater understanding for its potential in improving outcomes for a leading cause of morbidity and mortality.

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## READING 3 – PROTEOMICS IN ADULT AND PEDIATRIC IMFLAMMATORY BOWEL DISEASES

**Fabian O,[1,2] Sticova E,[1,6] Drastich P,[3] Bajer L,[3,4] Harant K,[5] Daskova N,[7] Cahova M,[7] Modos I,[8] Tichanek F.[8] A Current State of Proteomics in Adult and Pediatric Inflammatory Bowel Diseases: A Systematic Search and Review. Int J Mol Sci. 2023 May 27;24(11):9386.PMID: 37298338**

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ABSTRACT
Inflammatory bowel diseases (IBD) are systemic immune-mediated conditions with predilection for the gastrointestinal tract and include Crohn's disease and ulcerative colitis. Despite the advances in the fields of basic and applied research, the aetiopathogenesis remains largely unknown.

As a result, only one-third of patients achieve endoscopic remission. A substantial portion of patients also develop severe clinical complications or neoplasia. The need for novel biomarkers that can enhance diagnostic accuracy, more precisely reflect disease activity, and predict a complicated disease course thus remains high. Genomic and transcriptomic studies contribute substantially to our understanding of the immunopathological pathways involved in disease initiation and progression. However, eventual genomic alterations do not necessarily translate into the final clinical picture.

Proteomics may represent a missing link between the genome, transcriptome, and phenotypical presentation of the disease. Based on the analysis of a large spectrum of proteins in tissues, it seems to be a promising method for the identification of new biomarkers.

This systematic search and review summarise the current state of proteomics in human IBD. It comments on the utility of proteomics in research, describes the basic proteomic techniques, and provides an up-to-date overview of available studies in both adult and paediatric IBD.

## READING 4 – PRADER-WILLI AND ANGELMAN SYNDROMES

**Ma VK,[1,2] Egense AS,[1,2] Shankar SP,[1,2,5] Toth JN,[3] Mao R,[3,4] Fulmer ML.[3,4] Prader-Willi and Angelman Syndromes: Mechanisms and Management. Appl Clin Genet. 2023 Apr 6;16:41-52. PMID: 37051256.**

**doi: 10.2147/TACG.S372708. PMID: 37051256. Free full text.**

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ABSTRACT
Prader-Willi syndrome (PWS) and Angelman syndrome (AS) are genetic imprinting disorders resulting from absent or reduced expression of paternal or maternal genes in chromosome 15q11q13 region, respectively.

The most common aetiology is deletion of the maternal or paternal 15q11q13 region. Methylation is the first line for molecular diagnostic testing; MS-MLPA is the most sensitive test. The molecular subtype of PWS/AS provides more accurate recurrence risk information for parents and for the individual affected with the condition. Management should include a multidisciplinary team by various medical subspecialists and therapists. Developmental and behavioural management of PWS and AS in infancy and early childhood includes early intervention services and individualised education programmes for school-aged children. Here, we compare and discuss the mechanisms, pathophysiology, clinical features, and management of the two imprinting disorders, PWS and AS.

## READING 5 – COMMON PITFALLS IN GENETIC TESTING

**Shaw T,[1] Fok R,[1] Courtney E,[1] Li ST,[1] Chiang J,[2] Ngeow J.[3] Missed diagnosis or misdiagnosis: Common pitfalls in genetic testing. Singapore Med J. 2023 Jan;64(1):67-73.PMID: 36722519**

**doi: 10.4103/singaporemedj.SMJ-2021-467. PMID: 36722519. Free full text.**

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ABSTRACT

Genetic testing has the power to identify individuals with increased predisposition to disease, allowing individuals the opportunity to make informed management, treatment, and reproductive decisions. As genomic medicine continues to be integrated into aspects of everyday patient care and the indications for genetic testing continue to expand, genetic services are increasingly being offered by non-genetic clinicians.

The current complexities of genetic testing highlight the need to support and ensure non-genetic professionals are adequately equipped with the knowledge and skills to provide services.

We describe a series of misdiagnosed/mismanaged cases, highlighting the common pitfalls in genetic testing to identify the knowledge gaps and where education and support is needed. We highlight that education focusing on differential diagnoses, test selection, and result interpretation is needed. Collaboration and communication between genetic and non-genetic clinicians and integration of genetic counsellors into different medical settings are important.

This will minimise the risks and maximise the benefits of genetic testing, ensuring adverse outcomes are mitigated.

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## READING 6 – UPDATED EPIDEMIOLOGY OF GASTROINTESTINAL CANCERS IN EAST ASIA

**Huang J,[1,2] Lucero-Prisno DE 3rd,[3] Zhang L,[4,5,6] Xu W,[7] Wong SH,[8,9,10] Ng SC,[8,9,11] Wong MCS.[12,13,14,15] Updated epidemiology of gastrointestinal cancers in East Asia. Nat Rev Gastroenterol Hepatol. 2023 May;20(5):271-287.PMID: 36631716.**

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ABSTRACT
Globally, gastrointestinal cancers represent more than one-fourth of all cancer incidence and one-third of cancer-related mortality. Although there has been much progress in screening colorectal cancer, the prognosis of other gastrointestinal cancers tends to be poor. The highest burden of gastrointestinal cancers, including stomach, liver, oesophageal, and gallbladder cancers, was observed in regions in East Asia. The increasing burden of gastrointestinal cancers in East Asian regions is related to population growth, ageing, and the westernisation of lifestyle habits in this region. Furthermore, the rising incidence of young-onset colorectal cancer is an emerging trend in East Asia.

This Review provides a comprehensive and updated summary of the epidemiology of gastrointestinal cancers in East Asia, with emphasis on comparing their epidemiology in East Asia with that in Western regions, and highlights the major risk factors and implications for prevention.

Overall, to optimally reduce the disease burden incurred by gastrointestinal cancers in East Asian regions, a concerted effort will be needed to modify unhealthy lifestyles, promote vaccination against the hepatitis virus, control Helicobacter pylori, liver fluke, and hepatitis virus infections, increase the uptake rate of colorectal cancer screening, enhance detection of early cancers and their precursors, and improve cancer survivorship through an organised rehabilitation programme.

## READING 7 – IMPLEMENTING A PHARMACOGENETIC SERVICE

**Nguyen K,[1] Cicali EJ,[1,2] Lemke L,[1,2] Al Alshaykh H,[1,2] Cavallari LH,[1,2] Wiisanen K.[1,2] How to Implement a Pharmacogenetics Service at your Institution. J Am Coll Clin Pharm. 2022 Nov;5(11):1161-1175. PMID: 36589694.**

**doi: 10.1002/jac5.1699. PMID: 36589694. Payment required.**

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ABSTRACT
The vast majority of patients possess one or more pharmacogenetic variants that can influence optimal medication use. When pharmacogenetic data are used to guide drug choice and dosing, evidence points to improved disease outcomes, fewer adverse effects, and lower healthcare spending. Although its science is well established, clinical use of pharmacogenetic data to guide drug therapy is still in its infancy.

Pharmacogenetics essentially involves the intersection of an individual's genetic data with their medications, which makes pharmacists uniquely qualified to provide clinical support and education in this field. In fact, most pharmacogenetics implementations, to date, have been led by pharmacists as leaders or members of a multidisciplinary team or as individual practitioners.

A successful large-scale pharmacogenetics implementation requires coordination and synergy among administrators, clinicians, informatics teams, laboratories, and patients. Because clinical implementation of pharmacogenetics is in its early stages, there is an urgent need for guidance and dissemination of shared experiences to provide a framework for clinicians. Many early adopters of pharmacogenetics have explored various strategies among diverse practice settings.

This article relies on the experiences of early adopters to provide guidance for critical steps along the pathway to implementation, including strategies to engage stakeholders; evaluate pharmacogenetic evidence; coordinate laboratory testing, results interpretation, and their integration into the electronic health record; identify reimbursement avenues; educate providers and patients; and maintain a successful programme.

Learning from early adopters' published experiences and strategies can allow clinicians leading a new pharmacogenetics implementation to avoid pitfalls and adapt and apply lessons learnt by others to their own practice.

## READING 8 – MODERN NEWBORN SCREENING IN TAIWAN

**Chien YH,[1] Hwu WL.[2] The modern face of newborn screening. Pediatr Neonatol. 2023 Feb;64 Suppl 1:S22-S29. PMID: 36481189.**

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ABSTRACT
Newborn screening (NBS) has been used for years to identify newborns with severe but treatable conditions.

Following the initial setup for a total coverage of newborns in 1990s, Taiwan's NBS system was later optimised to ensure the timely return of results in infants with abnormal results.

Advances in techniques such as Tandem mass spectrometry enable the screening in a multiplex format and increase the conditions that are screened. Furthermore, advances in therapies, such as enzyme replacement therapy, stem cell transplantation, and gene therapy, significantly increase the need for newborn screening.

Advances in genomics and biomarkers discovery have improved the test accuracy with the assistance of second-tier tests, and NBS has the potential to be the first-tier test in the future. Therefore, the challenge facing NBS now is the knowledge gap, including the evidence of long-term clinical benefits in large cohorts, especially in conditions with new therapies, phenotypic variations, the corresponding management of certain screened diseases, and cost-effectiveness of extended NBS programmes.

A short-term and a long-term follow-up programme should be implemented to better collate those outcomes especially in the genomic era. Ethical and psychosocial issues are also encountered frequently.

Essential education and better informed consent should be considered fundamental alongside these new tests in future NBS.

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## READING 9 – PHARMACOGENOMICS IN STROKE AND CARDIOVASCULAR DISEASE

**Ross S,[1] Paré G,[1,3,4,5] Krebs K,[2] Milani L.[2] Pharmacogenomics in Stroke and Cardiovascular Disease: State of the Art. Stroke. 2023 Jan;54(1):270-278. PMID: 36325912**

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ABSTRACT
There is considerable interindividual variability in the response to antiplatelet and anticoagulant therapies, and this variation may be attributable to genetic variants. There has been an increased understanding of the genetic architecture of stroke and cardiovascular disease, which has been driven by advancements in genomic technologies, and this has raised the possibility of more targeted pharmaceutical treatments.

Pharmacogenetics promises to use a patient's genetic profile to treat those who are more likely to benefit from a particular intervention by selecting the best possible therapy. Although there are numerous studies indicating strong evidence for the effect of specific genotypes on the outcomes of vascular drugs, the adoption of pharmacogenetic testing in clinical practice has been slow.

This resistance may stem from occasional conflicting findings among pharmacogenetic studies, a lack of stroke-specific randomised controlled trials to test the effectiveness of genetically-guided therapies, and the practical and cost-effective implementation of genetic testing within the clinic.

Thus, this review provides an overview of the genetic variants that influence the individual responses to aspirin, clopidogrel, warfarin, and statins, and the different methods for pharmacogenetic testing and guidelines for clinical implementation for stroke patients.

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## READING 10 – PHARMACOGENETICS IN PRACTICE

**Luzum JA,[1] Petry N,[2,3] Taylor AK,[4] Van Driest SL,[5] Dunnenberger HM,[6] Cavallari LH.[7] Moving Pharmacogenetics Into Practice: It's All About the Evidence! Clin Pharmacol Ther. 2021 Sep;110(3):649-661.PMID: 34101169.**

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ABSTRACT

The evidence for pharmacogenetics has grown rapidly in recent decades. However, the strength of evidence required for the clinical implementation of pharmacogenetics is highly debated. Therefore, the purpose of this review is to summarise different perspectives on the evidence required for the clinical implementation of pharmacogenetics.

First, we present two patient cases that demonstrate how knowledge of pharmacogenetic evidence affected their care. Then we summarise resources that curate pharmacogenetic evidence, types of evidence (with an emphasis on randomised controlled trials [RCT]) and their limitations, and different perspectives from implementers, clinicians, and patients. We compare pharmacogenetics to a historical example (i.e., the evidence required for the clinical implementation of pharmacokinetics/therapeutic drug monitoring), and we provide future perspectives on the evidence for pharmacogenetic panels and the need for more education in addition to evidence.

Although there are differences in the interpretation of pharmacogenetic evidence across resources, efforts for standardisation are underway. Survey data illustrate the value of pharmacogenetic testing from the patient perspective, with their providers seen as key to ensuring maximum benefit from test results. However, clinicians and practice guidelines from medical societies often rely on RCT data to guide treatment decisions, which are not always feasible or ethical in pharmacogenetics. Thus, recognition of other types of evidence to support pharmacogenetic implementation is needed.

Among pharmacogenetic implementers, consistent evidence of pharmacogenetic associations is deemed most critical. Ultimately, moving pharmacogenetics into practice will require consideration of multiple stakeholder perspectives, staying particularly attuned to the voice of the ultimate stakeholder – the patient.