PHARMACOGENOMIC TESTING: A HANDOUT FOR CLINICIANS IN BC

How does PGx testing work?

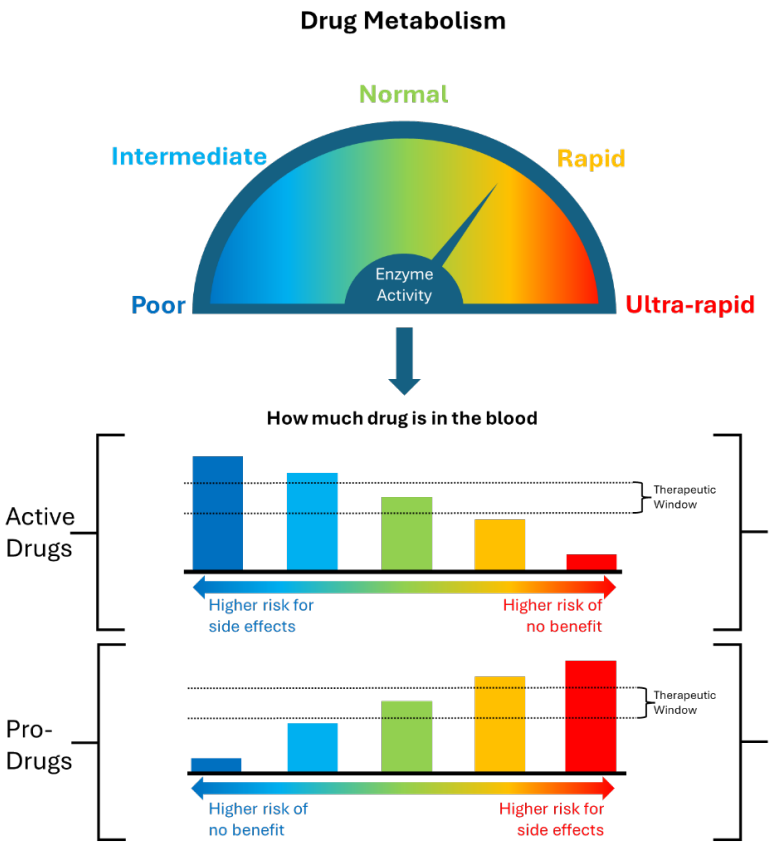
Interindividual differences in medication effectiveness and side effects are partly determined by variations in certain genes that code for enzyme activity. Pharmacogenomic or pharmacogenetic (PGx) testing works by assessing variants in specific genes of interest, which are known to impact that rate at which medications are metabolized. Pro-drugs (such as codeine) become active through metabolization while active drugs (such as sertraline) become inactive through metabolization. The different speeds at which genes tell the body to process medications are referred to as "metabolizer phenotypes." These metabolizer phenotypes represent a range of metabolism speeds, from "poor" (slow) to "normal" (medium) up to "ultrarapid" (fast). PGx tests are drug- and gene-specific, meaning that people do not have just one metabolizer phenotype. Because genes do not change, PGx results can be useful for future prescribing decisions.

UP-TO-DATE GUIDELINES & RESOURCES

Stay up-to-date on the latest guidelines, evidence, and practical information with:

ClinPGx is a comprehensive clinical pharmacogenomics resource integrating all of the PGx projects together
clinpgx.org/

Guidelines:
clinpgx.org/guidelineAnnotations
List of actionable genes:.
clinpgx.org/pgxGenes
List of drugs with PGx guidance:
clinpgx.org/prescribingInfo



Knowing how fast a medication is metabolized gives us information about the concentration of the medication in the blood. For example, a poor metabolizer of escitalopram (an active drug) is at a higher risk of side effects because the medication stays in the body for longer than expected, resulting in a greater serum concentration than intended. Conversely, an ultrarapid metabolizer of sertraline (also an active drug) is at a higher risk of not finding a medication effective because it is excreted quickly, resulting in a lower concentration in the blood than intended.

Figure 1: Drug metabolism and therapeutic effects for active and pro-drugs. In general, drug metabolism speeds vary from very slow (poor) to very fast (ultra-rapid). Depending on the serum concentration in the blood, patients may be at a higher risk of experiencing adverse effects or not seeing a clinical benefit. Figure modified with permission from Dr. Chad Bousman

How do I interpret PGx results & where can I see a sample PGx report?

The Jackson Laboratory has created a Test Report Nomenclature & Terminology Guide that will help with PGx test report interpretation. This resource provides both functional and genetic definitions of common terms, as well as a guide to different types of genotypic results.

Sequence2Script, a free, online, Canadian tool built to help healthcare providers translate PGx results into clinically useful recommendations offers the ability to input specific genetic information and generate specific clinical recommendations.

Who should get PGx testing?

There are three main indications for PGx testing:

- Your patient is starting or considering a drug associated with a PGx guideline.
    - ClinPGx will be your main reference for up-to-date information on drugs with PGx guidelines and label annotations outlined by drug regulatory agencies.
- Your patient is taking a medication with PGx recommendations, and they have not noticed a therapeutic effect despite taking the medication as directed.
- Your patient is experiencing adverse reactions at low doses of a medication associated with PGx recommendations.

What are some considerations of PGx testing that I should keep in mind?

Although PGx has been successfully implemented globally, the research has been primarily focused on European populations. Your patient's ancestral group may be underrepresented in this research and PGx testing could potentially be less informative for them. Sometimes ancestry may be listed as an indication for pharmacogenomic testing on the drug label due to some genetic variants being more common in individuals from specific ancestries. However, genetic variants can occur in people of any ancestry; therefore PGx testing could be useful in people of all ancestries.

- Learn more in *Exploring Pharmacogenomic Testing 2.0* module by the Jackson Laboratory
- Case study: Challenges of ancestry with warfarin dosing is discussed in Johnson *et al.* (2017)

PGx testing is typically not covered by the public health system in Canada and can be costly. Several private health insurance companies offer partial or full coverage of PGx testing, but patients interested in PGx should contact their specific insurance company to inquire if testing is covered under their policy. Some companies' PGx tests are considered deductible medical expenses on federal tax returns but patients should contact the company directly for more information.

- In BC, PGx tests are currently available for abacavir (HIV); allopurinol (gout); and carbamazepine (epilepsy and mental health) and Fluoropyrimidine (5-FU) / Cancer.

The need for a physician referral is company dependent. See the list of companies that provide PGx testing in Canada to learn how you may need to support your patient in the ordering and reviewing process.

For which drugs might I consider PGx testing before prescribing?

Always check CLINPGx for the most up-to-date information. This site combines lists of drug-gene pairs with evidence supporting prescribing recommendations based on their associated metabolizer phenotypes. These resources are periodically updated as new evidence emerges.

- The prescribing info explains how to adjust treatment of certain medications based on the person's genetic information
- Includes info from clinical guidelines, drug label annotations and drug regulatory associations

The table below is a quick reference to some common drug-gene pairs with the highest level of CPIC evidence (A or B) and for which published guidelines exist. **The FDA and Health Canada recommend PGx testing for many drugs beyond the ones listed here. This list is NOT comprehensive and is meant as a quick reference only.** It is up-to-date as of March 2026 and organized alphabetically by drug class.

If you're considering one of these drugs…		**… make sure the PGx test includes this gene**
Drug Class	**Drug (bolded if FDA or Health Canada drug label recommends testing, *italicized* if a prodrug)**	**Gene**
ADHD Medications	Atomoxetine	CYP2D6
Analgesics	*Codeine*, Hydrocodone, *Tramadol*	CYP2D6
Antibiotics	Nitrofurantoin	G6PD
	Peginterferon alfa-2a, Peginterferon alfa-2b	IFNL3
	Amikacin, Gentamicin, Neomycin, Netilmicin, Paromomycin, Streptomycin, Tobramycin	MT-RNR1
Anticoagulants	Warfarin	CYP2C9, CYP4F2, VKORC1
Anticonvulsants	**Carbamazepine**	**HLA-A, HLA-B**
	Oxcarbazepine	**HLA-B**
	Fosphenytoin, Phenytoin	CYP2C9, HLA-B
Antiemetics	Ondansetron	CYP2D6
Antifungals	Voriconazole	CYP2C19
Antimicrobials	Dapsone, Methylene blue, **Primaquine**	**G6PD**
Antineoplastics	*Tamoxifen*	CYP2D6
	Capecitabine, Fluorouracil	**DPYD**
	Rasburicase	**G6PD**
	Azathioprine, Mercaptopurine, Thioguanine	**NUDT15, TPMT**
Antiplatelets	*Clopidogrel*	CYP2C19
Antiretrovirals	**Abacavir**	**HLA-B**
	Atazanavir	UGT1A1
	Efavirenz	CYP2B6
Immunosuppressants	Tacrolimus	CYP3A5
Inhalation Anesthetics	Desflurane, Isoflurane, Methoxyflurane, Sevoflurane	CACNA1S, RYR1
Neuromuscular Blocking Agents	Succinylcholine	CACNA1S, RYR1
NSAIDs	Flurbiprofen, Celecoxib, Ibuprofen, Meloxicam, Prioxicam,	CYP2C9
Proton Pump Inhibitors	Dexlansoprazole, Lansoprazole, Omeprazole, Pantoprazole	CYP2C19

Serotonin Modulators	Vortioxetine	CYP2D6
Selective Serotonin and Norepinephrine Reuptake Inhibitors (SNRIs)	Venlafaxine	CYP2D6
Selective Serotonin Reuptake Inhibitors (SSRIs)	Sertraline	CYP2B6, CYP2C19
	Fluvoxamine, Paroxetine	CYP2D6
	Citalopram, Escitalopram	CYP2C19, CYP2D6
Statins	Rosuvastatin	ABCG2, SLCO1B1
	Atorvastatin, Lovastatin, Pravastatin, Simvastatin	SLCO1B1
	Fluvastatin	SLCO1B1, CYP2C9
Tricyclic Antidepressants (TCAs)	Amitriptyline, Clomipramine, Doxepin, Imipramine, Trimipramine,	CYP2C19, CYP2D6
	Nortriptyline, Desipramine	CYP2D6
Triptans	Rizatriptan, Sumatriptan,	CYP2D6
Xanthine Oxidase Enzyme Inhibitors	Allopurinol	HLA-B

How can I identify a good PGx test?

- **The test should be evidence-based.** This means it should follow gene/variant testing recommendations (as below) and prescribing guidelines, including CPIC, DPWG, and/or FDA.
    - The Association for Molecular Pathology (AMP) has recommendations for the minimum genetic variants in certain genes that should be tested (Tier 1) and an extended panel of genetic variants that are optional (Tier 2). You can view the list here:
- **The test should be performed in a lab certified by the College of Physicians and Surgeons of British Columbia (CPSBC) or an equivalent. CAP, CLIA, or WCDAA accreditation is an additional benefit.** A list of PGx tests available to Canadians notes lab accreditation.
- **The test company should be transparent** about what they test, how they test, and the method they use to translate genetic results into prescribing recommendations. These should be in line with the evidence as noted above.
- **The test company should be supportive.** They should offer or partner with an entity to provide PGx test report/result consultation. This may be a physician, genetic counselor, pharmacist or other professional with sufficient experience in pharmacogenetics (PGx).

This information is based on the 2019 article by Bousman *et al.* in the journal *Clinical Pharmacology & Therapeutics* (resource 2 in the "further reading and viewing" list below).

PGx, privacy, and genetic discrimination – What do I (and my patients) need to know?

Patients should be made aware that The Genetic Non-Discrimination Act (2017) prohibits any person (or company) to collect, use, or disclose one's genetic test results without their consent. Individuals are not obligated to disclose genetic test results to an employer or insurance company, for example.

Patients concerned about the privacy and security of their data should contact the testing company directly to inquire about their storage protocols and safeguards. In general, patients who are worried may wish to choose a company whose laboratory is located (and data are stored) in Canada. Recommendations from the Office of the Privacy Commissioner can be found here

A plain-language summary of the GNDA and how it may apply to your practice is available here:

Where can I find additional resources about PGx?

BC Ministry of Health Resources: For patients and For providers

Further Reading and Viewing:

- (For a list of PGx tests available in Canada) Maruf, A. A., Fan, M., Arnold, P. D., Müller, D. J., Aitchison, K. J., & Bousman, C. A. (2020). Pharmacogenetic Testing Options Relevant to Psychiatry in Canada. Canadian journal of psychiatry. 65(8), 521–530. https://doi.org/10.1177/0706743720904820
- (For a decision tree regarding ordering PGx testing) Bousman, C.A., Zierhut, H. and Müller, D.J. (2019) Navigating the Labyrinth of Pharmacogenetic Testing: A Guide to Test Selection. Clin. Pharmacol. Ther., 106; 309-312. https://doi.org/10.1002/cpt.1432
- (For a quick overview of what PGx is, watch): Pharmacogenomics Info Video produced by The Hospital for Sick Children in Toronto. https://www.youtube.com/watch?v=4532ns1MqsI
- (For an example of what a patient's PGx testing journey might look like, watch): "Pharmacogenetics in Action" by the All of Us Research Program in the USA. https://www.youtube.com/watch?v=oaOSGmhU5-w&ab_channel=AllofUsResearchProgram
- (For more general information about DTC genetic testing in Canada, visit): https://geneticseducation.ca/resources-for-clinicians/genomic-technologies/direct-to-consumer-testing/gecko-on-the-run-9

Where can I get Continuing Medical Education (CME) credits?

Note that the following courses are accredited for CME in the USA. Check with your organization regarding how to claim credits in Canada.

- Jackson Laboratory's "Exploring Pharmacogenomic Testing 2.0"
- National Human Genome Research Institute's "ISCC-PEG Pharmacogenomics Learning Series"

Help us improve this handout! Share your feedback here

This information was updated March 2026. Pharmacogenomic testing is evolving and guidelines should be reviewed regularly. This information is provided for educational purposes only and is not intended as medical advice.