Pharmacogenomic Testing for *TPMT* and *NUDT15* with Thiopurine Use

Genetic Testing Recommendations

PROVINCIAL GENETICS PROGRAM

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Ontario Health

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Introduction

Pharmacogenomics at Ontario Health

Ontario Health is building a comprehensive and integrated system for clinical genetic services across the province. Through collaboration with health system partners, the Provincial Genetics Program (PGP) provides evidence-based guidance for genetic diagnostic testing and counselling services in key areas. PGP Expert Groups work with health care professionals, laboratory scientists, administrators, and patient and care partner advisors to establish standardized practices for collecting, using, and reporting clinical genetics data. These efforts ensure that people in Ontario can access high-quality genetic testing services when needed. The PGP and Provincial Genetics Advisory Committee (PGAC) identified pharmacogenetics, or the study of how genetic variability can impact a person's response to a drug, as a priority domain for development in Ontario. This resulted in the formation of a Pharmacogenomics (PGx) Working Group in 2022. This working group produced and published a recommendations report for the implementation of PGx testing in Ontario. This led to the formation of the Pharmacogenomics Expert Group in April 2024. The role of the Expert Group is to develop evidence-based guidance for the provision of PGx testing.

Specific drug-gene pairs may be submitted by clinicians, researchers, industry representatives, or other members of the public to the PGx Expert Group to be considered for guidance generation. Submissions may be sent via email to the PGP along with basic supporting evidence and any relevant published literature. Topics are assessed using Intake Criteria including stipulations on the level of evidence and clinical actionability as well as the availability of peer-reviewed dosing recommendations (see Appendix A). The PGx Expert Group selected this drug-gene pair based on recommendations by the PGx Working Group and based on the aforementioned Intake Criteria. *TPMT* has nine clinical annotations at Level 1A[1] and *NUDT15* has four clinical annotations at Level 1A on ClinPGx (formerly The Pharmacogenomics Knowledgebase [PharmGKB]). Clinical guidance is available from the Clinical Pharmacogenomics Implementation Consortium (CPIC), the Food and Drug Administration (FDA), Health Canada, the Dutch Pharmacogenetics Working Group (DPWG), the Réseau National de Pharmacogénétique (RNPGx) in France, and the International Network of Agencies for Health Technology Assessment (INAHTA).

This report provides recommendations for who is eligible for *TPMT* and *NUDT15* genotypic testing prior to initiating thiopurine therapy, what the minimum test components should be, the clinical actionability following test results, and best practices for reporting. The intended audience for this guideline includes clinical pharmacologists, gastroenterologists, oncologists, pharmacists, and other clinicians who may prescribe thiopurines or manage patients who have been prescribed thiopurines.

Thiopurines and *TPMT* and *NUDT15*

Thiopurine medications are purine antimetabolites with immunosuppressive effects. They may be prescribed for the treatment of acute lymphoblastic leukemia (ALL), autoimmune disorders such as Crohn's disease, vasculitis, rheumatoid arthritis, or for organ transplant recipients.[1,2] Thiopurines

[1] Level 1A clinical annotations on ClinPGx describe variant-drug combinations that have variant-specific prescribing guidance available in a current clinical guideline annotation or an FDA-approved drug label annotation and prescribing guidance is available for specific variants. These variant-drug combinations are considered to have very strong evidence.

include mercaptopurine and its prodrug azathioprine, as well as the less frequently prescribed thioguanine. Thiopurines were one of the first medications that were studied for pharmacogenomic associations, in part due to unexpectedly severe adverse effects observed in some patients.[3,4] Subsequently, they were one of the first to have PGx information added to their drug label[5] and to have guidelines produced by CPIC.[6] Internationally, several countries offer publicly funded testing for *TPMT* and/or *NUDT15* for thiopurine use including Australia, England, France, Japan, Switzerland, and potentially others. Some health insurance companies and health systems in the United States of America also offer this testing.

Thiopurine methyltransferase (TPMT) catabolizes thiopurines and TPMT activity is determined through a monogenic autosomal codominant inheritance pattern. Variant alleles of *TPMT* are associated with low enzyme activity which is then associated with enhanced pharmacologic effects of thiopurine medications. There are three single nucleotide polymorphisms (SNPs) which are associated with unstable proteins and increased degradation of TPMT proteins. Over 90% of low TPMT activity phenotypes are explained by these common inactivating alleles. Individuals with two loss-of-function *TPMT* alleles are at 100% risk of potentially fatal myelosuppression when indication-specific doses of thiopurines are prescribed, due to an increased buildup of cytotoxic 6-thioguanine nucleotides (TGNs). Patients with only one loss-of-function *TPMT* allele are at an increased risk of myelosuppression, although response varies.[7] Over the past decade, it has been recognized that nudix hydrolase-15 (NUDT15) is another enzyme involved in the metabolism of thiopurines where the presence of loss-of-function genetic variations more commonly observed in certain non-European populations results in severe myelosuppression.[7]

Testing

Genetic testing for variations in the *TPMT* and *NUDT15* genes is recommended for all patients prior to initiating thiopurine therapy or when a thiopurine medication is being considered as a treatment option. Pre-emptive testing is preferred; however, in certain cases if testing was not conducted prior to therapy initiation, then testing is still recommended for patients who are not tolerating the medication (i.e., myelosuppression), particularly with regards to dosing. The target turnaround time for testing is 14 days. Repeat testing is not recommended unless the provincial test has been expanded to include more variants and there is a clear clinical need.

The frequencies of loss-of-function alleles in *TPMT* versus *NUDT15* vary based on ancestry. Loss-of-function alleles in *TPMT* are more commonly found in individuals with African or European ancestry, while loss-of-function alleles in *NUDT15* are more commonly found in those with Asian ancestry or with Indigenous ancestry from southern regions of North America or Central and South America. In East Asians specifically, about 1 in 50 people carry poor metabolizer status for *NUDT15*, higher than the rate of the same status for *TPMT* in those of European descent. It is recommended, however, to test the same variants in all eligible patients for both *TPMT* and *NUDT15* as opposed to attempting to determine ancestry and tailor variants respectively. It is important to note that targeted genotype testing alone cannot account for all potentially deleterious effects of the presence of rare or previously undiscovered variants.

This guidance document is intended to be used once a clinical decision to order genetic testing has been made. It is not meant to guide a clinician in the choice between phenotypic or genotypic testing (i.e., TPMT enzyme activity testing vs. genotype testing for *TPMT*). While complementary phenotype laboratory tests can be helpful adjuncts to genotyping tests, this decision should be made using clinical judgement and/or professional guidelines and may vary on a case-by-case basis.

Clinicians who prescribe thiopurines were identified and consulted on current practices and ideal reporting. These clinicians provided perspectives from specialties including pediatric hematology oncology, pediatric and adult gastroenterology, and pediatric rheumatology. Since this medication is not routinely initiated in the primary care setting, family medicine practitioners were consulted via the PGP's Primary Care Genetics Reference Table as per the standard PGP review process.

Variant Curation

Minimum testing recommendations were formed based on experts' clinical experience, current practices in Ontario, and recommendations from the recent consensus paper on *TPMT* and *NUDT15* genotyping from CPIC, DPWG, the Association for Molecular Pathology (AMP), College of American Pathologists, European Society for Pharmacogenomics and Personalized Therapy, and PharmGKB (now ClinPGx).[8] A variant curation sub-group was formed within the PGx Expert Group and included members with notable expertise in *TPMT* and *NUDT15* genotyping. Testing recommendations were formed by the sub-group and then presented back to the PGx Expert Group for review and adoption. These recommendations will be evaluated and updated by the PGx Expert Group as new evidence emerges in the field.

Online and published PGx resources often include both common and rare variants that have varying levels of evidence and/or frequency in certain populations. For an Ontario context, the following factors were the foundation of discussions around whether a particular variant should be included or excluded for provincial testing:

- The potential toxicity associated with a complete or partial loss-of-function gene variant.
- The clinical actionability or level of evidence used to inform dosing associated with the variant.

Testing offered in Ontario currently includes all the Tier 1 variants outlined in the consensus paper from Pratt *et al*. since these variants have strong evidence, clear clinical actionability, and are most common in the general population. The variant curation sub-group unanimously voted to include these variants in provincially funded testing. This decision was then unanimously supported by the full PGx Expert Group as well. The Tier 1 alleles to be tested in Ontario include *TPMT*2, TPMT*3A, TPMT*3B, TPMT*3C*, and *NUDT15*3* (see Appendix B).

It is important to note that by testing only Tier 1 alleles, the *TPMT* and *NUDT15* genotype-predicted phenotypes for a given individual will be based on a genotyping test that interrogates only the most common and already-proven sites of functional variation. It is always possible that a new, previously undiscovered site of variation may confer loss-of-function in an individual. This could thus lead to the rare possibility of a non-functional allele being erroneously reported as wild-type (i.e., normal metabolizer status).

Testing for additional genes or variants beyond the Tier 1 alleles mentioned here can be considered by individual institutions, however this would not be part of the provincial test. Any additions should have clear clinical actionability and readily accessible support for result interpretation (e.g., consult with clinical pharmacology).

Clinical Actionability

Thiopurines are prescribed for a variety of indications, both malignant and non-malignant. The dosing recommendations that follow below are derived from the CPIC guidelines originally produced in 2011[6], updated in 2018[7] with minor updates[9] in 2020 and 2024, and then updated again in 2025[10]. These provide baseline guidance on clinical actionability for *TPMT* and *NUDT15* variations. Prior and future updates to any CPIC guidelines can be found on the CPIC website (cpicpgx.org) by searching for the associated drug-gene pair(s). Managing physicians should follow disease- or specialty-specific guidance if available, particularly in pediatric settings.

Pre-emptive testing for *TPMT* and *NUDT15* variation helps prescribers tailor starting doses to better predict the patient's ability to metabolize thiopurines and thereby reduce the likelihood of acute myelosuppression, a significant benefit to at-risk patients. Clinical outcomes when dosed based on *TPMT* and *NUDT15* genotype testing have not been shown to be compromised in ALL and inflammatory bowel disease patients.[11–15] It should be noted that there are no other phenotypic traits associated with variations in *TPMT* or *NUDT15* alleles beyond those associated with the use of thiopurine therapy.[7,16]

Prescribing Recommendations

The evidence informing CPIC recommendations for *TPMT* and *NUDT15* is noted to come from clinical studies focused on a few common diseases; however, thiopurines can be prescribed for a broader set of indications. Since the functional impact of the known genotype/phenotype is well established, particularly as it relates to genotype-dependent pharmacokinetic profile, recommendations can be extrapolated to all conditions where thiopurines are prescribed.[7]

Individuals who do not carry any loss-of-function genetic variations in *TPMT* or *NUDT15* alleles are ascribed as "normal metabolizers" and have an NM status. Standard dosing for mercaptopurine, azathioprine, and thioguanine is recommended, based on the disease-specific guidelines and/or patient presentation. Nevertheless, in the absence of toxicity, clinicians should wait to reach steady state before making any dose adjustments and continue to monitor the individual for any undesired pharmacologic effects.

The metabolizer status of a patient helps predict their risk for experiencing thiopurine-related leukopenia, neutropenia, or thrombocytopenia. Normal Metabolizer (NM) status means the patient has a normal risk level and no known loss-of-function genetic variations are observed in the interrogated alleles for *TPMT* or *NUDT15*. Intermediate Metabolizer (IM) or possible intermediate metabolizer status means the patient has inherited only one loss-of-function *TPMT* or *NUDT15* allele and has an increased risk level of these adverse effects with around 30-60% of patients being unable to tolerate conventional doses of mercaptopurine or azathioprine.[12,17,18] Individuals with two loss-of-function *TPMT* alleles are at 100% risk of potentially fatal myelosuppression, due to an increased buildup of toxic TGNs and are categorized as Poor Metabolizer (PM) status.

TPMT and *NUDT15* are independent genes and the likelihood of having variations in both genes is based on the population frequencies for each gene. There have been reports of compound intermediate/poor metabolizers though and in such cases, there was a trend for lower thiopurine tolerance when compared to individuals with variations in only one of the genes. In these cases, clinicians should consult the relevant *TPMT* and *NUDT15* recommendations and when uncertain regarding the functional effects, the starting dose should be on the lower level of metabolism.

GUIDING PRINCIPLES FOR DOSING

1) If a more specific guideline is available (e.g., for a specific population, specialty, or disease) which accounts for *TPMT* and *NUDT15* status and applies to a patient, then the most specific guidance should be used by the clinician. The guidelines in this document are generalized and are meant to help support clinicians in navigating dose adjustments in the absence of specific guidelines or treatment protocols.
2) For the most up-to-date guidelines, please refer to the CPIC website at cpicpgx.org and search for the relevant drug-gene pair.
3) All patients should be monitored for adverse reactions regardless of genetic results.

The recommendations below should be assumed to be relevant for both *TPMT* and *NUDT15* results and for the use of azathioprine, mercaptopurine, or thioguanine unless otherwise stated.

THIOPURINE DOSING RECOMMENDATIONS BY *TPMT* & *NUDT15* GENOTYPE-PREDICTED PHENOTYPE

Normal Metabolizer (NM) status for both *TPMT* **and** *NUDT15*:

- Proceed with the standard dose

Intermediate Metabolizer (IM) status for either *TPMT* **or** *NUDT15*:

- Reduce the starting dose based on disease-specific guidance if available, or as per the general CPIC guidelines (i.e., 30-80% of standard dose for azathioprine or mercaptopurine; 50-80% of standard dose for thioguanine).

Intermediate Metabolizer (IM) status for both *TPMT* **and** *NUDT15*:

- Reduce the starting dose based on disease-specific guidance if available, or as per the general CPIC guidelines (i.e., 20-50% of standard dose). Take the starting dose (e.g., mg/kg, mg/m^2) into consideration based on protocol or guideline-specific recommendations when determining if/how to reduce the dose for this category of IM status individuals.

Poor Metabolizer (PM) status for *TPMT* **and/or** *NUDT15*:

- Avoid thiopurine therapy if possible
- If not possible to avoid (e.g., some malignant conditions), significantly reduce the starting dose based on the associated clinical protocol, disease-specific guidelines, or CPIC guidance (i.e., 10% of standard dose and thrice weekly dosing instead of daily)

For more detailed recommendations, please consult the most recent CPIC guideline on thiopurine use where dose recommendations are organized by gene, phenotype, and medication with example doses provided. CPIC guidelines are accessible on their website (cpicpgx.org/guidelines/). Refer to specialty-specific practice guidelines if available for more specific dosing recommendations. An interactive clinical annotation of the CPIC guidelines by genotype is accessible on ClinPGx.

Reporting

PGx results follow a patient for life and may be used by clinicians with various backgrounds such as physicians or pharmacists, across various specialties, and in both acute and community-based settings. It is thus imperative that these PGx results are communicated in a clear, concise, and standardized manner that accounts for all of these variables. Recommendations for reporting of *TPMT* and *NUDT15* testing results are included below.

Lab Report Content

- Genotype and phenotype should be clearly reported at the beginning of the report.
- The only clinical actionability steps indicated should be those associated with the specific result.
- Date of the report must be clearly indicated along with a note that interpretation of the results could change over time and testing may need to be updated in the future.
- Optional background information may be included, especially if various clinicians may access the results, but this should not precede the results or interpretation sections.
- The possibility for drug interactions should be mentioned along with a reminder to consult the product monograph, regardless of genetic results.
- References for any recommendations given within the report should be clearly listed and additional resources may be linked or listed as well.
- Clinical actionability directives need to indicate that actions may differ based on speciality, especially between malignant and non-malignant contexts. Reports should also acknowledge that some specialties may follow alternative practice guidelines that are specific to that specialty.

Lab Report Format

- Visual cues should be used to indicate abnormal results whenever possible (e.g., bold text, underlined text, alternate colours).
- Reports should start with patient details, followed directly by results and interpretation sections. Additional resources or background information may be last and/or included on a second page.

Beyond the structure and content of the report itself, accessibility of results is also critical. Results may be integrated into electronic health record (EHR) management systems through flags or alerts which follow the patient for life and should also be searchable. PGx results should be available for managing clinicians to view in a provincial electronic health record or clinical viewer, supported by the Ontario Laboratories Information System (OLIS), and should ideally be searchable. Labs providing testing for *TPMT* and *NUDT15* should aim to follow standard procedures for how results are communicated, especially for urgent results (e.g., paper report, phone call, etc.). Education will be a critical component of implementation of this testing and will directly support how reporting is received and utilized. Strong awareness of the availability of the test, when and how to order the testing, and where to find further resources or support when results are received are all needed.

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Appendices

Appendix A: Intake Criteria

A submission must meet at least one of the following levels of evidence:

- Level of evidence is 1A or 1B on ClinPGx
- Level of evidence is 2A on ClinPGx (Tier 1 of Very Important Pharmacogene list)

  AND

- Dosing recommendations are available from a consensus source (e.g., CPIC)

Potential reasons for exclusion:

- Submission is a panel of drug-gene pairs (currently too broad of scope)
- Level of evidence is 2B or lower, or not curated (ClinPGx)
- No dosing or clinical actionability recommendations are available

Appendix B: Variants Tested in Ontario

***TPMT* AND *NUDT15* VARIANTS TESTED IN ONTARIO:**

Allele	rsID	HGVS genomic nomenclature (GRCh38)	HGVS cDNA nomenclature	HGVS protein nomenclature
TPMT*2	rs1800462	NC_000006.12: g.18143724C>G	NM_000367.5: c.238G>C	NP_000358.1: p.(Ala80Pro)
TPMT*3A	rs1800460, rs1142345	NC_000006.12: g.18138997C>T, NC_000006.12: g.18130687T>C	NM_000367.5: c.460G>A, NM_000367.5: c.719A>G	NP_000358.1: p.(Ala154Thr), NP_000358.1: p.(Tyr240Cys)
TPMT*3B	rs1800460	NC_000006.12: g.18138997C>T	NM_000367.5: c.460G>A	NP_000358.1: p.(Ala154Thr)
TPMT*3C	rs1142345	NC_000006.12: g.18130687T>C	NM_000367.5: c.719A>G	NP_000358.1: p.(Tyr240Cys)
NUDT15*3	rs116855232	NC_000013.11: g.48045719C>T	NM_018283.4: c.415C>T	NP_060753.1: p.(Arg139Cys)

Appendix C: Abbreviations

ALL	Acute lymphoblastic leukemia
AMP	Association for Molecular Pathology
cDNA	Complementary DNA (deoxyribonucleic acid)
CPIC	Clinical Pharmacogenetics Implementation Consortium
DPWG	Dutch Pharmacogenetics Working Group
FDA	Food and Drug Administration
HGVS	Human Genome Variation Society
IM	Intermediate metabolizer
INAHTA	International Network of Agencies for Health Technology Assessment
NM	Normal metabolizer
NUDT15	Nudix hydrolase 15
OLIS	Ontario Laboratories Information System
PGAC	Provincial Genetics Advisory Committee
PGP	Provincial Genetics Program
PGx	Pharmacogenomics / Pharmacogenetics
PharmGKB	Pharmacogenomics Knowledgebase (now ClinPGx)
PM	Poor metabolizer
RNPGx	Réseau National de Pharmacogénétique (French National Network of Pharmacogenetics)
rsID	Reference SNP cluster ID
SNP	Single nucleotide polymorphism
TGN	6-thioguanine nucleotide
TPMT	Thiopurine methyltransferase