PHARMACOGENETICS

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Pharmacogenetics Introduction

The tools and information in this guide are provided as a basic introduction and reference to clinical pharmacogenetics and is not an all-inclusive guide. For a more in-depth understanding of the content, please consider taking a pharmacogenetics/pharmacogenomics certificate course. This information is not to be used as a substitute for professional training and judgment. Use of this information indicates acknowledgment that neither PSW nor its contributing authors will be responsible for any loss or injury, including death, sustained in connection with or as the result of using this information. When making judgments regarding specific medications, pharmacists should consult the complete information available in the product prescribing information or other published literature as appropriate. PSW is under no obligation to update information contained herein.

Key Terms and Abbreviations

PGx – Shorthand for pharmacogenetics/pharmacogenomics.

Actionable – Institution/user-specific criteria that defines which genes/results would be used to change/modify a patient's drug therapy. This Toolkit only considers the following criteria as actionable: (1) a drug-gene pair for which there is a CPIC guideline, (2) on the FDA's Table of Pharmacogenetic Associations Sections 1 or 2, or (3) the potential risk of harm to a patient if the results are not considered (e.g. Factor V and estrogen medications).

Informative – Institution/user-specific criteria that defines which genes/results would NOT be used to change/modify a patient's drug therapy. This Toolkit considers informative results as anything not actionable (as defined above) or a given drug/gene pair for which CPIC recommends "No Recommendation." Note, this information changes constantly, so always check ClinPGx/resources for updates to the recommendations.

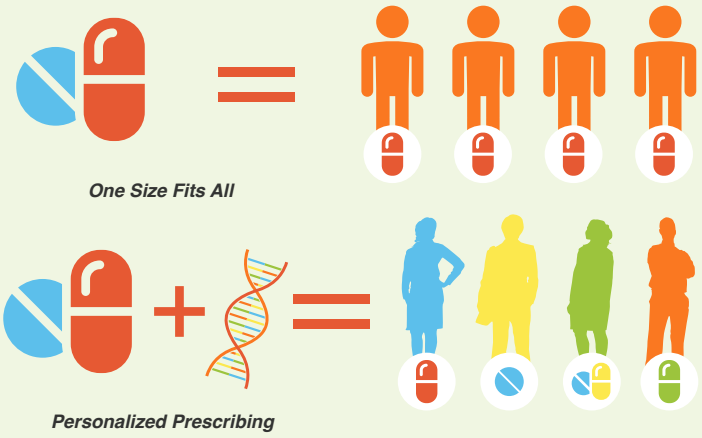
Gene – A specific stretch of DNA that usually serves as a template for a protein (e.g., enzyme) product.

Single Nucleotide Polymorphism (SNP) – A change in the DNA at a single location.

Pharmacogenetics (PGx)

Pharmacogenetics is the study of how a person's DNA may impact their response (efficacy or safety) to a specific medication. Pharmacogenetics usually only considers one drug-gene pair, whereas pharmacogenomics often includes multiple genes, however these two terms are often used interchangeably.

Figure 1. Proteins Impacted by Pharmacogenetics



One Size Fits All

Personalized Prescribing

Goals and Importance of Pharmacogenomics (PGx):

- One size does not fit all when it comes to medicines.
- PGx testing is a medication safety initiative that avoids potentially unsafe therapies and decreases adverse effects.
- PGx testing may also improve efficacy to certain medications and helps minimize therapeutic failures.
- PGx provides individualized medication recommendations based upon a patient's genomic variants by pharmacogenetic testing.
- According to the American Society of Pharmacovigilance, "Forty-six million adverse drug reactions (ADRs) are reported each year, leading to 1.3 million emergency room visits."[1]
- Adverse drug events rank as the third most common cause of death in the US.[1]

Allele – One of two or more versions of a gene.

Haplotype – Same as allele, but each allele contains one or more SNPs that tend to be inherited together in a given pattern.

Genotype – A given individual's collection of alleles for a given gene; in humans (which are diploid), there are usually two copies that describe the genotype.

Diplotype – Same as genotype, but each allele has a collection of SNPs that tend to be inherited together (rather, haplotype).

Phenotype – An individual's observable traits (e.g., eye or hair color) which are usually a result of their genotype.

Metabolizer Phenotypes – An individual's pharmacogenetic phenotype (specific to one gene) that has an effect when exposed to that drug. For example: Ultrarapid Metabolizer (UM), Rapid Metabolizer (RM), Normal (Extensive) Metabolizer (NM), Intermediate Metabolizer (IM), Poor Metabolizer (PM).

Activity Score (AS) – A quantitative method of assigning metabolizer phenotype for a given gene: a numerical value is assigned to a specific allele (e.g., value of 0 = no function, 0.25-0.5 = decreased function, and 1.0 = normal function) for a given gene, and adding the two alleles' values determines a patient's Activity Score; the metabolizer phenotypes are defined by a range of activity scores. Note, this is used for several (but not all) of the drug metabolizing enzymes. For example, CYP2C9*1/*3 genotype: *1 allele = 1.0, *3 allele = 0, CYP2C9 AS = (1.0+0=) 1, which correlates to the CYP2C9 Intermediate Metabolizer phenotype (AS Range of 1.0-1.5) as noted in CPIC Guidelines.

Phenoconversion – Presence of an enzyme inhibitor or inducer that causes the patient to convert from their expected Metabolizer phenotype to a different phenotype (e.g., fluoxetine usually causes phenoconversion from a CYP2D6 Normal to an Intermediate or Poor Metabolizer). Synonymous with phenocopy.

Actionability of Pharmacogenomics[2,3]

Current evidence reviews from Clinical Pharmacogenetics Implementation Consortium (CPIC) and the Food & Drug Administration (FDA) help determine if changes in the prescribing is necessary for drug:gene pairs. This evidence is continually reviewed, and additional medications may be added as more information is available.

CPIC Level A: Genetic information used to change prescribing of affected drug

CPIC Level B: Genetic information could be used to change prescribing of the affected drug because alternative therapies or dosing are extremely likely to be as effective and as safe as non-genetically based dosing

FDA Table of Pharmacogenomic Associations Section 1: Data supports therapeutic management recommendations

Table 1. Select Drug - Pharmacogene Table[4]
This is not an all-inclusive list but rather an example of the most common pharmacogenes and associated drugs with CPIC Level A evidence.

Category	Medications
CYP2C19	amitriptyline, citalopram, clomipramine, clopidogrel, dexlansoprazole, doxepin, escitalopram, esomeprazole, fluoxetine, fluvoxamine, imipramine, lansoprazole, omeprazole, pantoprazole, paroxetine, rabeprazole, sertraline, trimipramine, venlafaxine, voriconazole, vortioxetine
CYP2D6	amitriptyline, atomoxetine, citalopram, codeine, clomipramine, desipramine, doxepin, escitalopram, fluoxetine, fluvoxamine, hydrocodone, imipramine, metoprolol, nortriptyline, ondansetron, paroxetine, propranolol, sertraline, tamoxifen, trimipramine, tramadol, venlafaxine, vortioxetine
CYP2C9	celecoxib, flurbiprofen, fluvastatin, fosphenytoin, ibuprofen, lornoxicam, meloxicam, phenytoin, piroxicam, tenoxicam, warfarin
HLA-B	atorvastatin, fluvastatin, lovastatin, pitavastatin, pravastatin, rosuvastatin, simvastatin
TPMT	azathioprine, mercaptopurine, thioguanine
DPYD	capecitabine, fluorouracil
CYP3A5	tacrolimus
SLCO1B1	Atorvastatin, Fluvastatin, Lovastatin, Pitavastatin, Pravastatin, Rosuvastatin, Simvastatin
UGT1A1	irinotecan, atazanavir

Body Systems Impacted by Pharmacogenetics

Table 2. Proteins Impacted by Pharmacogenetics

Proteins	Impact
Drug Metabolizing Enzymes (DMEs)	Responsible for drug metabolism (e.g., CYP2D6, CYP3A4, UGT1A1, etc.)
Transport Proteins	Responsible for absorption, distribution, elimination of medications (e.g., P-glycoprotein, SLCO1B1, etc.)
Drug Receptors/ Targets	Responsible for drug binding and action at therapeutic target (e.g., BCR-ABL fusion protein, HER2 receptor, etc.)
Immune-Related Targets	Responsible for susceptibility to certain adverse reactions (e.g., HLA-A, HLA-B, etc.)

It is important to note that PGx can impact either or both of pharmacodynamics (PD, how a drug affects the body) and pharmacokinetics (PK, how the body breaks down a drug).

Common Pharmacogenetic Nomenclature

Table 3. Common Pharmacogenetic Nomenclature

Gene Nomenclature Involving Star (*) Alleles:	
Example: CYP2C19*1 • CYP = gene for Cytochrome P450 Superfamily • 2 = Family • C = Subfamily • 19 = Isoenzyme • *1 = Allele (Haplotype) Variant	*For example, to describe a given patient:* • CYP2C19 *1 = allele (haplotype) • CYP2C19 *3 = allele (haplotype) • CYP2C19 *1/*3 = genotype (diplotype)

Gene Nomenclature Involving Single Nucleotide Polymorphisms (SNPs):	
Example: VKORC1 c.-1693 G>A • VKORC1 = gene for Vitamin K epoxide Reductase • c.-1693 = cDNA location of the SNP (note: this is sometimes omitted in a given context) • G = the reference or "normal" nucleotide at this position • > = denotes nucleotide change • A = the new variant nucleotide	*For example, to describe a given patient:* • VKORC1 G = allele (in this case, this is the reference allele) • VKORC1 A = allele (in this case, this is the variant allele) • VKORC1 G/A = genotype (the patient is heterozygous at this position)

Pre-Pharmacogenetic Testing Considerations

1. **PGx Test Selection:** Select an appropriate pharmacogenomics test based upon considerations of your patient population, potential medication use, cost, and other pertinent factors.
2. **Benefits/Reasons for Testing:**
   a. To help the care team make safer choices about medicines based upon the patient's DNA (e.g., avoid side effects and help with optimal dosing).
   b, Pharmacogenetic test results do not change over time because one's DNA does not change. The clinical interpretation of those results may be updated as scientific knowledge expands.
   c. This type of testing can not only help guide medication selection, but sometimes can also explain past medication failures, which can be reassuring for patients.

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3. **Limitations of Testing:**
   a. Pharmacogenetic testing does not explain all adverse drug reactions or all medication non-response. Other factors (like kidney function, overall health, cigarette smoke, etc.) can also affect how a person responds to medications.
   b. The utility of pharmacogenetic testing may be limited in the setting of a liver transplant, bone marrow transplant, or recent blood transfusion (i.e., within 6 weeks).
   c. Not all Pharmacogenetic tests can be used to adjust medication therapy. Before using a patient's direct-to-consumer (DTC) PGx results for treatments, clinicians should confirm the results in a CLIA-certified lab if necessary and make sure the data are FDA-approved for therapeutic use. Patients should be informed about the value of confirmatory testing and expert interpretations, as well as advised not to alter their prescriptions based on DTC results.[7]

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4. **Protection of Results:** The Genetic Information Non-discrimination Act (GINA) prevents discrimination based on genetic results for health insurance and employment. Sometimes there are additional state laws that further protect patient results. Test results are also generally subject to HIPAA, privacy and confidentiality protections.[8]
5. **Incidental/Secondary Findings:** While uncommon, some pharmacogenes may predict genetic disease risk (e.g., DPYD, UGT1A1, etc.). Referral to a genetic counselor may be appropriate.
6. **Appropriate Use of PGx Results:** PGx results are one piece of a larger puzzle in determining the best treatment options. There may be times when not following the PGx recommendation is the best treatment option for the patient. Additionally, many testing companies test for genes with limited guidance, and thus may not be actionable for clinical recommendations. Results should always be considered in the setting of other factors, such as drug-drug interactions, renal and hepatic function and other conditions.
7. **Cost of Testing:** Pharmacogenetic testing cost and insurance coverage varies substantially; patients should be encouraged to check with their insurance and the testing company prior to getting tested. While insurance reimbursement for PGx testing is improving, often it is not covered by insurance companies.

Clinical Pharmacogenetic Resources

Table 4. Clinical Pharmacogenetic Resource

Resource	Details
Clinical	• **Website**: www.clinpgx.org • **Summary**: This resource integrates Pharmacogenomics Knowledge Base (PharmGKB), Clinical Pharmacogenetics Implementation Consortium (CPIC) and Pharmacogenomics Clinical Annotation Tool (PharmCAT) into one resource • **How to Use:** Access all resources from one central location with ongoing enhancements aimed at further increasing integration, including CPIC guidelines, PharmGKB data, and PharmCAT functionalities.
The Clinical Pharmacogenetics Implementation Consortium (CPIC)	• **Website**: www.cpicpgx.org • **Summary**: An international consortium of over 500 members creates, curates, and posts freely available, peer-reviewed, evidence-based, and detailed gene/drug clinical practice guidelines. CPIC guidelines are indexed in PubMed as clinical guidelines, endorsed by ASHP and ASCPT, and adhere to the Institute of Medicine's Standards for Developing Trustworthy Clinical Practice Guidelines. • **How to Use:** Access the PDF CPIC Guideline publications and supplemental information including allele definition and frequency tables. View upcoming drug-gene pairs under consideration for guideline development, as well as other PGx resources.
The FDA Table of Pharmacogenetic Associations	• **Website:** www.fda.gov • **Summary:** The FDA created tables based upon evaluation of current data, and believes there is sufficient scientific evidence to suggest that individuals with certain pharmacogenetic findings (when taking specific medications) may experience altered metabolism or differing therapeutic effects. There are three sections of Pharmacogenetic Associations for which the Data Support Therapeutic Management Recommendations (Section 1), the Data Indicate a Potential Impact on Safety or Response (Section 2), and the Data Demonstrate a Potential Impact on Pharmacokinetic Properties Only (Section 3). • **How to Use:** Using the search function, users are able to quickly identify drugs or genes for which some pharmacogenetic recommendation(s) exist.
The Genetic Testing Registry (GTR)	• **Website:** www.ncbi.nlm.nih.gov/gtr • **Summary:** This resource provides a central repository for voluntary submission of available genetic testing offerings. • **How to Use:** Search for drugs or genes of interest and find which clinical laboratories test for the item of interest. Learn about the test's purpose, methodology, validity, utility and laboratory contacts and credentials.
OncoKB	• **Website:** www.oncokb.org • **Summary:** OncoKB is a comprehensive precision oncology knowledge base that provides information on the effects of specific gene alterations on cancer treatment. It aggregates data from various sources, including clinical trials, drug approvals, and guidelines, to inform clinicians and researchers about actionable mutations and their corresponding therapies. • **How to Use:** Users can search by genes, alteration, cancer type, drug, or genomic variant to explore clinical evidence for targeted therapies.
PharmVar	• **Website:** www.pharmvar.org • **Summary:** The Pharmacogene Variation Consortium is a central repository for pharmacogenes focusing on haplotype structure and allelic variation. This information is useful for translating between various nomenclature representations of PGx. • **How to Use:** Use the Genes tab to identify a PGx gene (= pharmacogene) of interest. Learn what SNP(s) define a given diplotype. Translate between different reference sequences for a given allele.
Pharmacogenetics Phenoconversion Calculator – University of Florida	• **Website:** www.precisionmedicine.ufhealth.org • **Summary:** The PROP™ Pharmacogenetics Calculator is intended to help clinicians integrate a standardized method of assessing CYP2D6 phenoconversion into practice when a CYP2D6 genotype is available. The CYP2D6 drug metabolizing enzyme is susceptible to inhibition by concomitant drugs, which can lead to a clinical phenotype that is different from the genotype-based phenotype, a process referred to as phenoconversion. • **How to Use:** Use this calculator to enter a patient's CYP2D6 genotype and choose any interacting medications and the patient's activity score and clinical phenotype will be displayed. This calculator uses updated recommendations for translating CYP2D6 genotype to phenotype from the Clinical Pharmacogenetics Implementation Consortium (CPIC) and includes drugs classified as strong or moderate CYP2D6 inhibitors by the Food and Drug Administration.

Implementation of Pharmacogenetics

There are many barriers to implementation of pharmacogenetics, however many settings have successfully addressed these barriers. Please see below for pharmacogenomics stakeholders and roles, as well as suggested readings.

Table 5. Recommended Stakeholders and Collaborators

Stakeholder/ Collaborator	Description
Clinician Champion (MD, DO, NP, PA, etc.)	Help garner support for initiative, as well as financial support. Sometimes lead pharmacogenetic initiatives.
Informatics Personnel	Help with integration of pharmacogenetic results into electronic medical record (EMR), which is the most high-leverage method of ensuring consistent pharmacogenetic recommendations.
Pharmacists	Help provide interpretation, education and consultation of pharmacogenetic results. Can help design implementation and lead pharmacogenetic initiatives..
Genetic Counselors	Help with laboratory selection considerations, educational/consultation initiatives, guidance on incidental/secondary findings, program design/consideration advisement.
Research Scientists	Often pharmacogenetic implementation starts as a research initiative, thus these individuals often have a wealth of experience and considerations that greatly assist with implementation
Laboratory Personnel	Help with laboratory and testing considerations, as well as reporting of results.

Pharmacogenetic Research Initiatives and Collaboratives

- **All Of Us** – This NIH funded research is an effort to collect and study data from one million plus people living in the United States to advance areas of precision medicine, including pharmacogenetics. Learn more at www.joinallofus.org
- **eMERGE PGx** – This partnership of the electronic MEdical Records and GEnomics Network and the Pharmacogenomics Research Network advanced pharmacogenetic implementation by by providing funding and guidance for early PGx implementation efforts with many healthcare institutions across the United States. Learn more at www.genome.gov/Funded-Programs-Projects/Electronic-Medical-Records-and-Genomics-Network-eMERGE
- **IGNITE** – The Implementing GeNomics In practice also advanced pharmacogenetic implementation by providing funding and guidance for early PGx implementation efforts with many healthcare institutions across the US. Learn more at www.genome.gov/Funded-Programs-Projects/Implementing-Genomics-in-Practice-IGNITE-Pragmatic-Clinical-Trials-Network

Precision Oncology[5,6]

Precision oncology is a branch of pharmacogenomics that focuses on somatic and germline alterations within a tumor. Somatic alterations identify genomic changes within the tumor tissue and are not passed to offspring. Germline alterations are inherited variations that are present in every cell and can be passed to offspring.

Table 6. Somatic and Germline Alterations

	Somatic Alterations	Germline Alterations
Biological Origin	Acquired in tumor cells during a person's lifetime	Inherited from parent; present at birth
Clinical Use	Assist with therapy selection, prognosis, and resistance patterns of tumor	Reveals hereditary cancer syndromes that may influence surgery, systemic therapy, and surveillance intensity
Examples of Genes that can be Altered	ALK, BRAF V600, BRCA1, BRCA2, EGFR, HER2, KRAS, MET, PIK3CA, RET, ROS1	APC, ATM, BRCA1, BRCA2, Lynch Syndrome (MMR genes), PTEN, STK11, TP53
When to Test?	**Indication based:** Breast, Colorectal, Non-small cell lung cancer, Pancreatic, Prostate **Other considerations:** Any advanced or metastatic malignancy	**Indication based:** Breast, Ovarian, Pancreatic, Prostate **Other considerations:** Younger age, multiple primaries, or family history raises concern regardless of tumor site

Key Provider and Patient Consultations

Top 3 Key Elements of Informed Consent prior to PGx Testing[9]:

1. General Test Description
   a. Patients should be made aware of the nature of PGx testing and the ways in which genes can affect medication response, side effects, and dosing.
2. Goal of the Test
   a. Describe the purpose of PGx testing and highlight how the results can guide drug selection or alternative therapies to increase the safety of medications.
3. Summarize findings and actions requested
   a. Talk about anticipated benefits including selection of the safest medication or dose, preventing adverse effects, and providing lifetime utility of results.

Instead of:	Try:
Precision Medicine	Treatment plan designed specifically for you
Pharmacogenomics	Drugs and your genes, Medicine and your DNA
Metabolize	Break down, Process, Remove from your body
Mutation	Variant, Variation, Difference
Rapid or Ultrarapid Metabolizer	Faster than expected
Wild-Type or "Normal"	Expected, Most common
Poor Metabolizer	Slower than expected
Actionable	Impacts your health/safety, Recommended changes

Mnemonic for Pre-test Counseling	Mnemonic for Post-test Counseling
P - Purpose and benefit of PGx testing G - Genetic concepts X - Example of a PGx concept D - Drawbacks of PGx testing R - Risks and concerns U - Understand the patient's perspective G - Game plan and Process S - Sharing results	O - Overview of PGx results P - PGx interactions T - Take account of gene-drug-drug interactions I - Initial drug change considerations M - Manage genes with future, familial, and disease-risk implications I - Interpretation and applicaiton updates over time Z - Zero in on patient comprehension ED - Education and Sharing results and supplemental information

Suggested Implementation Readings

- Arwood et al. “Implementing pharmacogenomics at your institution: establishment and overcoming implementation challenges.” *Clin Transl Sci*. 2016;9(5):233-245. PMID 27214750
- Hall BT et al. Implementing pharmacogenomics clinical decision support: a comprehensive tutorial on how to Integrate the Epic genomics module. *Clin Pharmacol Ther.* 2025;117(6):1670-1681. doi:10.1002/cpt.3599
- Cicali EJ et al. How to implement a pharmacogenetics service at your institution. *J Am Call Clin Pharm*. 2022;5:1161-1175. doi:10.1002/jac5.1699
- Catherine B Oliver, Dhara Naik, Mark Kirkikis, Mary Ghaffari, Lance J Oyen, Karen C Thomas, Implementation of pharmacogenomics: Experience at Ochsner Health, *American Journal of Health-System* Pharmacy, Volume 82, Issue 6, 15 March 2025, 341–349, https://doi.org/10.1093/ajhp/zxae295
- Uber R et al. Pharmacogenomics implementation and multidisciplinary genomics collaboration: Real-world experience from Geisinger, *American Journal of Health-System Pharmacy*, Volume 79, Issue 13, 1 July 2022, Pages 1038–1041, https://doi.org/10.1093/ajhp/zxac065
- Mitaly S et al. Pharmacists’ role in supporting the return of over 10,000 preemptive pharmacogenomics results: The Mayo Clinic experience, *American Journal of Health-System Pharmacy*, Volume 80, Issue 23, 1 December 2023, 1733–1742, https://doi.org/10.1093/ajhp/zxad159
- Duarte et al. “Multisite investigation of strategies for the clinical implementation of pre-emptive pharmacogenetic testing” *Genet Med*. 2021:23(12):2335-2341. PMID 34282303
- Dunnenberger H et al. “Implementation of a multidisciplinary pharmacogenomics clinic in a community health system” *Am J Health Syst Pharm*. 2016:73(23):1956-1966. PMID 27864203
- Gammal R et al. “Documenting pharmacogenomics test results in electronic health records: practical considerations for primary care teams” *J Pers Med*. 2021:11(12):1296. PMID 34945768
- Nicholson et al. “Considerations when applying pharmacogenomics to your practice” *Mayo Clin Proc.* 2021;96(1):218-230. PMID 33308868
- Vo T et al. “Pharmacogenomics Implementation: Considerations for Selecting a Reference Laboratory” *Pharmacotherapy.* 2017;37(9):1014-1022. PMID 28699700

References

1. Rodziewicz TL, Houseman B, Vaqar S, et al. Medical Error Reduction and Prevention. [Updated 2024 Feb 12]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK499956/
2. CPIC Level Definitions for CPIC Genes/Drugs. Accessed January 2026. https://cpicpgx.org/prioritization/#flowchart
3. FDA Table of Pharmacogenetic Associations. Last Updated October 26, 2022. Accessed January 2026. https://www.fda.gov/medical-devices/precision-medicine/table-pharmacogenetic-associations#section1
4. Relling MV, Klein TE. CPIC: Clinical Pharmacogenetics Implementation Consortium of the Pharmacogenomics Research Network. *Clin Pharmacol Ther*. 2011;89(3):464-467. doi:10.1038/clpt.2010.279
5. Bailey MH, Tokheim C, Porta-Pardo E, Sengupta S, Bertrand D, Weerasinghe A, Colaprico A, Wendl MC, Kim J, Reardon B, Ng PK, Jeong KJ, Cao S, Wang Z, Gao J, Gao Q, Wang F, Liu EM, Mularoni L, Rubio-Perez C, Nagarajan N, Cortés-Ciriano I, Zhou DC, Liang WW, Hess JM, Yellapantula VD, Tamborero D, Gonzalez-Perez A, Suphavilai C, Ko JY, Khurana E, Park PJ, Van Allen EM, Liang H; MC3 Working Group; Cancer Genome Atlas Research Network; Lawrence MS, Godzik A, Lopez-Bigas N, Stuart J, Wheeler D, Getz G, Chen K, Lazar AJ, Mills GB, Karchin R, Ding L. Comprehensive Characterization of Cancer Driver Genes and Mutations. Cell. 2018 Apr 5;173(2):371-385.e18. doi: 10.1016/j.cell.2018.02.060. Erratum in: Cell. 2018 Aug 9;174(4):1034-1035. doi: 10.1016/j.cell.2018.07.034. PMID: 29625053; PMCID: PMC6029450.
6. Garutti M, Foffano L, Mazzeo R, Michelotti A, Da Ros L, Viel A, Miolo G, Zambelli A, Puglisi F. Hereditary Cancer Syndromes: A Comprehensive Review with a Visual Tool. Genes (Basel). 2023 Apr 30;14(5):1025. doi: 10.3390/genes14051025. PMID: 37239385; PMCID: PMC10218093.
7. Donnelly RS, Ayoubieh H, Kudron EL, et al. When Patients Present with Direct-to-Consumer Pharmacogenomic Test Results. *Mayo Clin Proc.* 2025;100(2):197-203. doi:10.1016/j.mayocp.2024.10.016
8. Genetic information discrimination. U.S. Equal Employment Opportunity Commission. Accessed October 31, 2025. https://www.eeoc.gov/genetic-information-discrimination.
9. Pereira L, Haidar CE, Haga SB, et al. Assessment of the current status of real-world pharmacogenomic testing: informed consent, patient education, and related practices. *Front Pharmacol*. 2024;15:1355412. Published 2024 Feb 8. doi:10.3389/fphar.2024.1355412
10. Ho TT, Bell G, Gammal RS, Gregornik D, Wake DT, Dunnenberger HM. A clinician’s guide for counseling patients on results of a multigene pharmacogenomic panel. *Am J Health Syst Pharm.* 2022;79(19):1634-1644. doi:10.1093/ajhp/zxac189

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