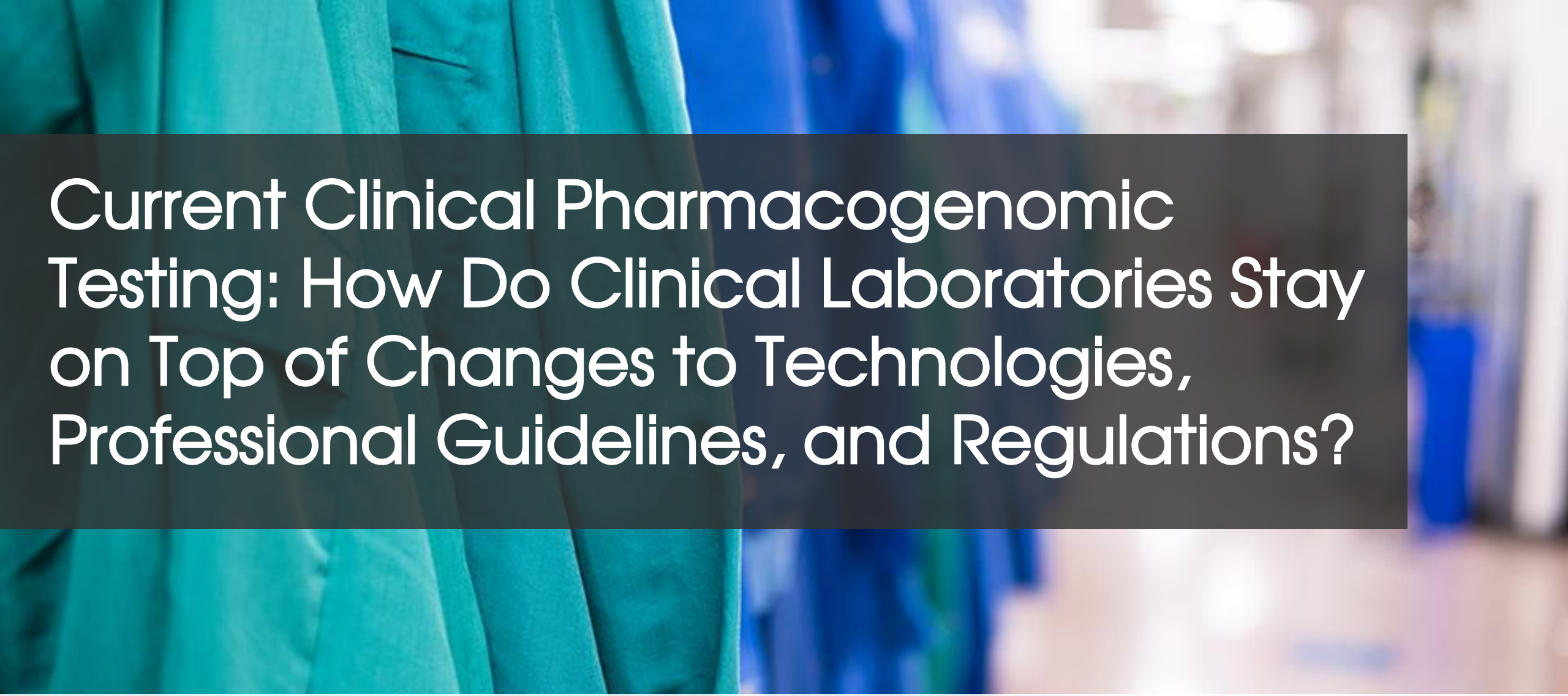


Current Clinical Pharmacogenomic Testing: How Do Clinical Laboratories Stay on Top of Changes to Technologies, Professional Guidelines, and Regulations?

Yuan Ji, PhD, MBA, FACMG
Associate Professor, University of Utah
Section Chief and Medical Director, ARUP Laboratories

JUNE 2024

Learning Objectives

1. Elucidate key distinctions between clinical pharmacogenomic (PGx) testing and other molecular genetic tests

2. Discuss latest technological advancements driving continual improvement in clinical PGx testing

3. Describe the present state of professional guidelines developed to tackle unanswered clinical questions within the realm of clinical PGx testing

4. Expound on the challenges and opportunities emerging from the ever-evolving regulatory landscape shaping laboratory medicine, with a specific focus on the clinical implementation of PGx

OUTLINE

Clinical pharmacogenomics: an overview

Technological advancements

Updates on professional guidelines and standardization efforts

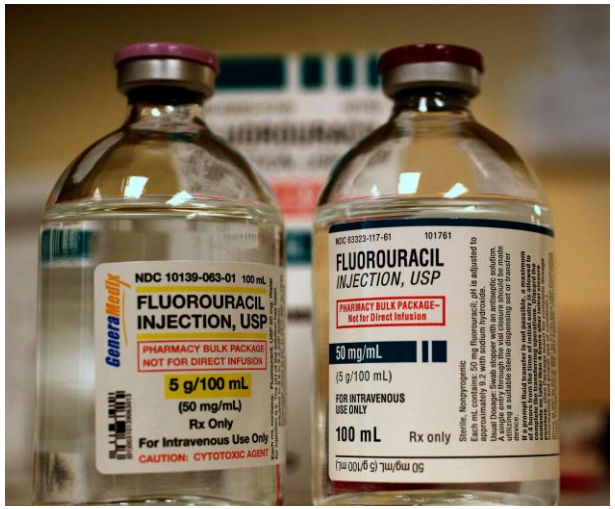
Ever changing regulatory landscape

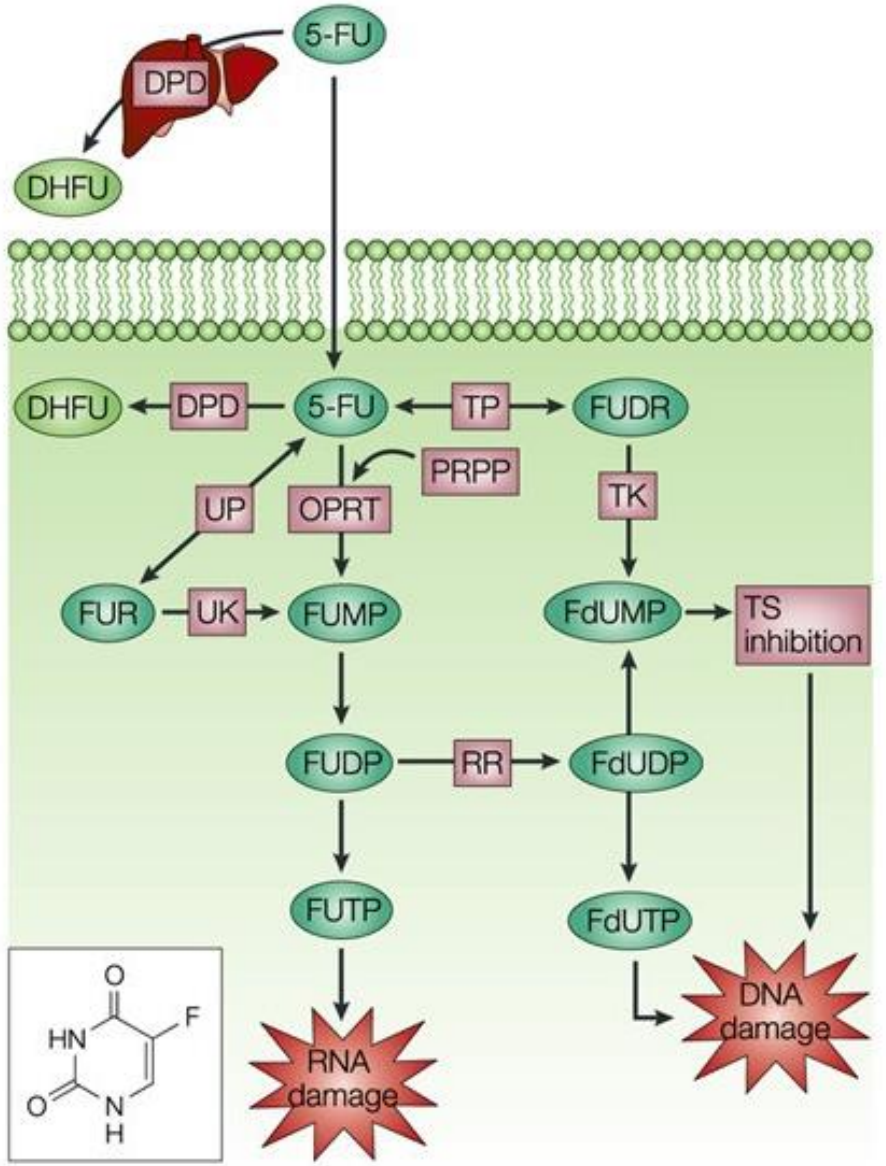
Clinical pharmacogenomics: an overview

Patient Story – Dr. Anil Kapoor

https://www.cbc.ca/news/canada/toronto/cancer-drug-5fu-genetic-variant-testing-1.7039145

DPYD Variants and Fluorouracil (5-FU)





Nature Reviews | Cancer

https://www.nature.com/articles/nrc1074

FDA approves safety labeling changes regarding DPD deficiency for fluorouracil injection products

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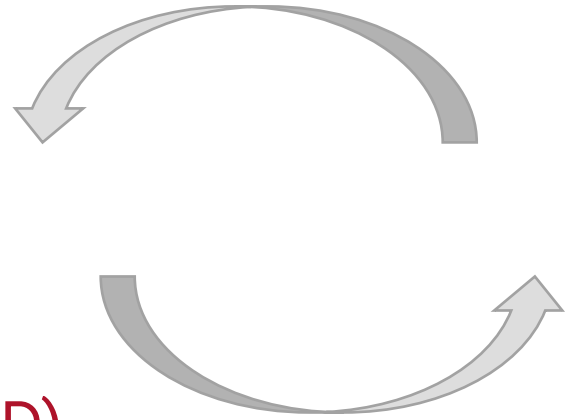
On March 21, 2024, the Food and Drug Administration approved safety labeling changes for fluorouracil injection products. This effort was a collaboration between FDA's Office of Generic Drugs and the Oncology Center of Excellence (OCE).

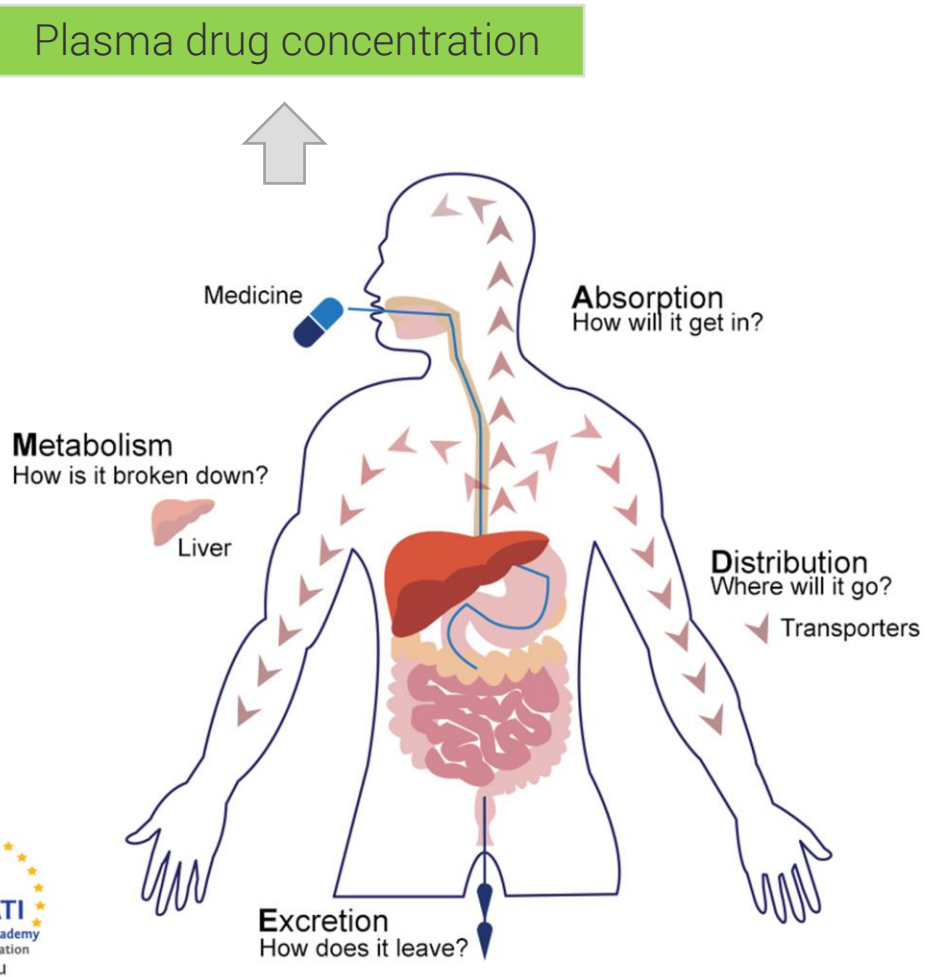
Content current as of:
03/21/2024

Variations in Drug Response

What is a pharmacogenomic test?

Pharmacokinetics (PK)
"What the body does to a drug"





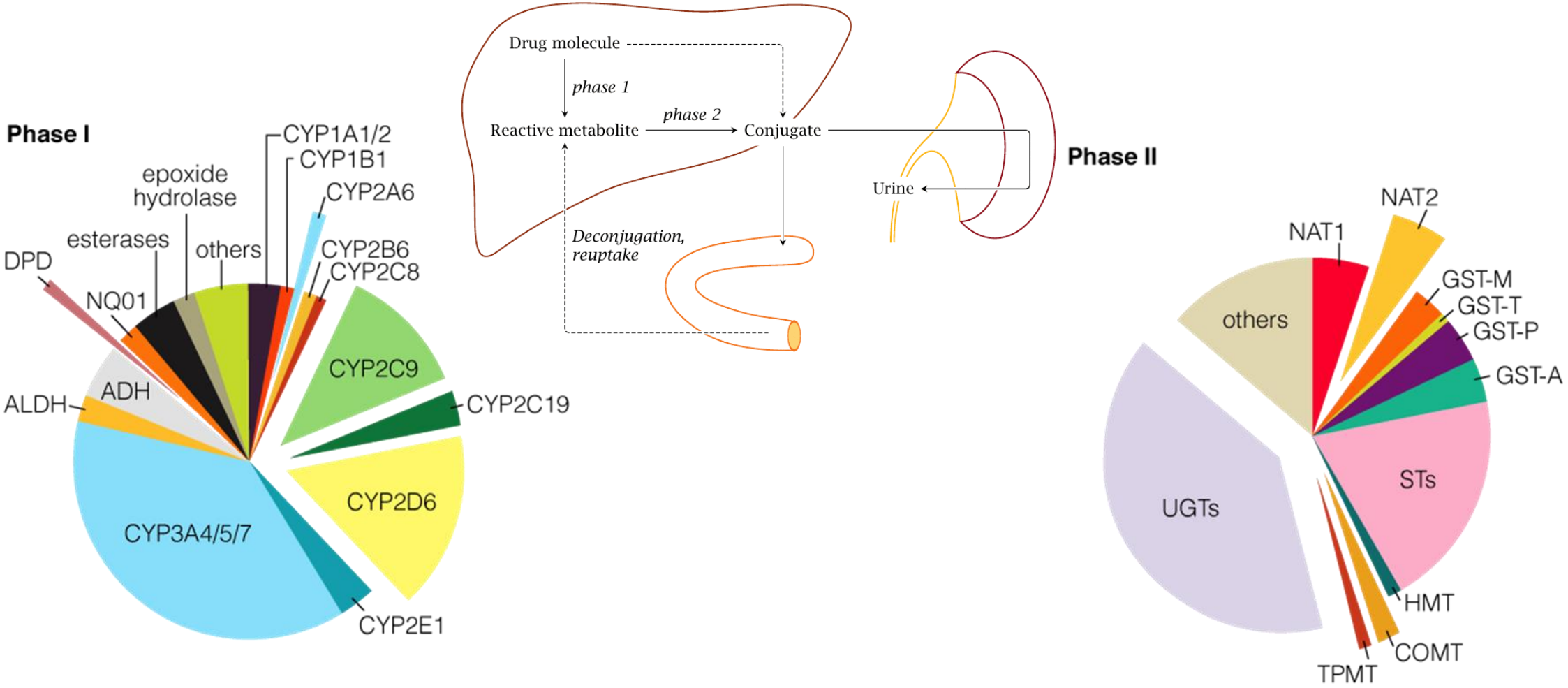
Pharmacodynamics (PD)
"What a drug does to the body"

- Target organ/tissue
- Receptor, protein
- Signaling pathway
- Physiologic effects
- Mechanism of action

EUPATI
European Patients' Academy on Therapeutic Innovation
www.eupati.eu

Dose response

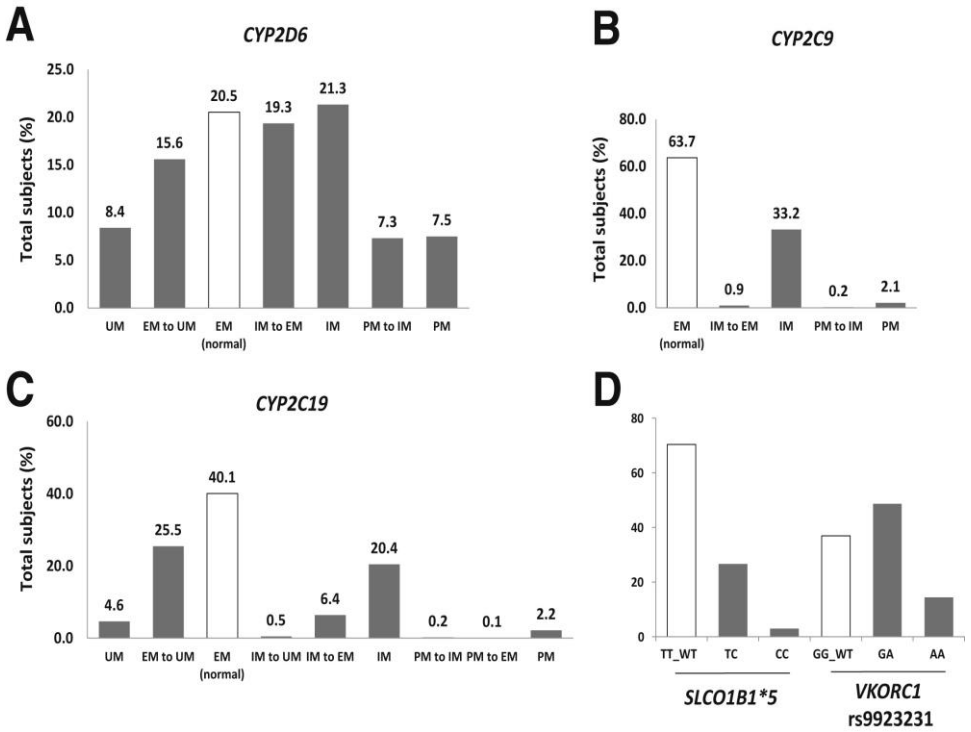
Drug Metabolism and Pharmacogenes



http://www.cyprotex.com/admepk/polymorphic-and-non-cyp-mediated-metabolism

How common are pharmacogenetic variants?

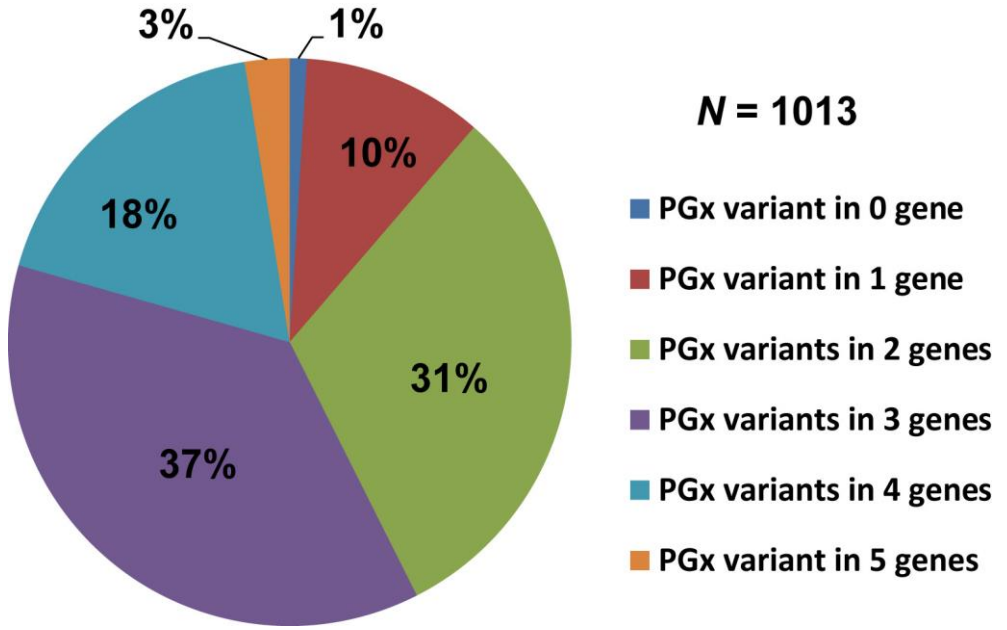
Distribution of drug metabolizer phenotypes



Ji et al. JMD, volume 18, issue 3, p438-445, may 2016

Patients with actional PGx variants

(in *CYP2D6, CYP2C9, CYP2C19, SLCO1B1*5,* and *VKORC1*)



Characters of Clinical PGx Tests

As molecular genetic tests

- Testing platforms and instruments
- General regulatory requirements and laboratory practices (CAP Mol checklist, NY)
- Testing personnels and qualifications
- Laboratory director training pathways

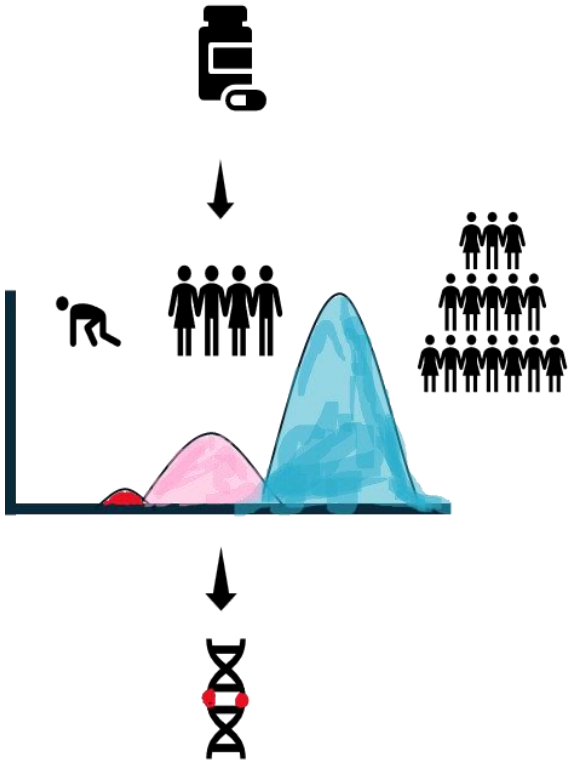
Unique to PGx tests

- Targets: pharmacogenes and variants (from rare to common)
- Inheritance pattern: co-dominant inheritance
- Primary testing strategy: targeted genotyping for common PGx alleles
- Sequencing-based PGx tests are emerging
- Nomenclatures: standard vs. legacy star(*) alleles
- Additional considerations (e.g., TAT, specimen requirements, LDT development, reporting, result consultation)
- Gene-specific considerations (e.g., *CYP2D6*, *HLA* alleles)
- Considerations around clinical implementation
- Additional requirements for reimbursements

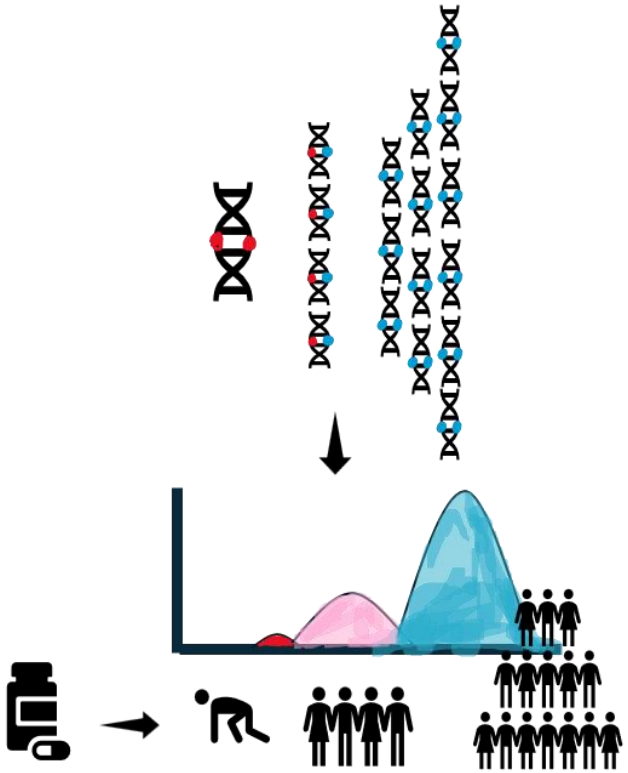
Technological advancements

From targeted genotyping to sequencing-based approaches

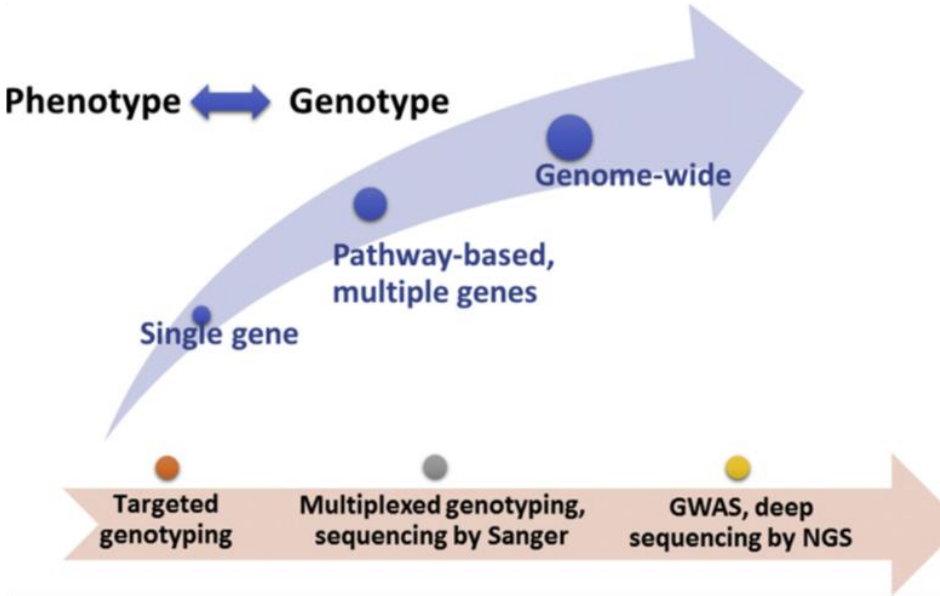
Pharmacogenetics Discovery



Phenotype to Genotype



Genotype to Phenotype



Targeted Testing vs. Sequencing

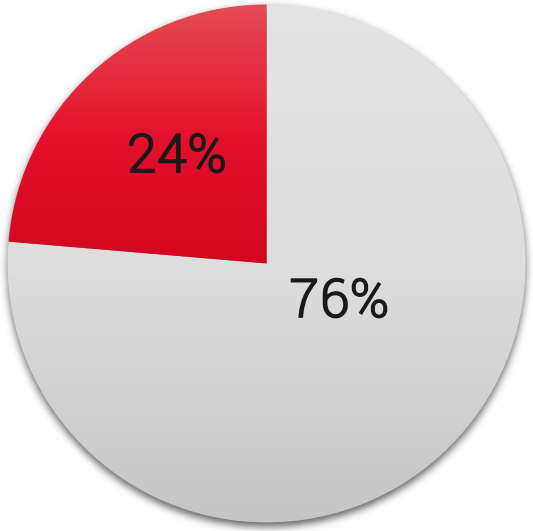
Targeted Genotyping Assays

- More commonly used
- Strong clinical evidence with or without clinical guidelines
- Technically feasible
- Can be cost-effective
- Rare variants or ethnicity-specific variants can be missed
- Causal variants may not be detected
- Limitations in detecting complex genomic variations beyond SNPs (e.g., CNVs, hybrid alleles in *CYP2D6*)
- *Inability to phase
- Variability among laboratories

Sequencing (Sanger, NGS etc.)

- Ability to detect both common and rare/novel variants
- Might be cost saving by shortened testing cycle and when multiple genes interrogated
- Classification of rare/novel variants can be challenging
- Assembling rare variants using star (*) allele nomenclature can be challenging
- *Inability to phase (Sanger, and short-read NGS)
- Challenges with complex genomic regions, e.g., *CYP2D6* and *HLA* alleles
- Bioinformatic tools, i.e., star (*) allele callers, are published and become available through instrument vendor(s) and performance varies depends on the genes (*CYP2D6* vs. other genes) and require validation prior to clinical use

NGS for PGx Assays



Targeted genotyping Sequencing

12/185 tests including del/dup analysis

- Most pharmacogenes are technically feasible by NGS
- Potential cost and time-saving when interrogating multiple genes or at genome-wide scales
- More variant types can be detected
- Many commonly tested PGx targets are common SNPs, and require modified filter setting to detect common sequence variants
- Interpretations and reporting of novel rare variants and alleles can be challenging. There is currently no professional guideline for rare PGx variant classification and interpretation.
- The function of a pharmacogene and end point clinical drug metabolizer phenotype prediction can also be substrate-specific,
- VUSs in pharmacogenes should not be recommended
- Star (*) allele calling and diplotype assembling requires bioinformatic tools
- Calling important PGx risk *HLA* alleles (*HLA-B*57:01*, *HLA-B*58:01*, *HLA-B*15:02* and *HLA-A*31:01*) can be challenging with short-read NGS platforms and requires special informatic tools/callers
- Targeted callers need to be verified prior to clinical use using DNA samples with known or orthogonally confirmed genotypes

WES/WGS for PGx Profiling

Clinical indications

- For patients experiencing acute symptoms or sudden death when an adverse drug event is suspected
- Can be reported as secondary findings when WES/WGS performed
- Not currently included in the ACMG SF v3.2 list except *RYR1* and *CACNA1S* (for malignant hyperthermia)
- Can be used as reflex test following a targeted PGx genotyping test

Considerations

- Laboratories should define regions of interest against the "must-to-test" PGx targets
- Laboratories should establish policies on reporting rare variants (and variant types) in pharmacogenes
- Laboratories should publish limitations on PGx-related genes to avoid false assurance
- Polypharmacy can complicate the phenotype-driven analysis process
- Can be costly to perform and interpret, and challenging for reimbursement if using as a standalone PGx test

Considerations for NGS-based PGx Tests

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APPLIED LABORATORY MEDICINE

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Volume 9, Issue 1
January 2024

JOURNAL ARTICLE

Interrogating Pharmacogenetics Using Next-Generation Sequencing Get access >

Yuan Ji ✉, Sherin Shaaban

The Journal of Applied Laboratory Medicine, Volume 9, Issue 1, January 2024, Pages 50–60, https://doi.org/10.1093/jalm/jfad097

Published: 03 January 2024 **Article history** ▾

Test Design

- Testing format, i.e., single-gene, multi-gene panel, WES/WGS
- Laboratory developed procedure (LDP) or commercial product
- Content selection, i.e., pharmacogenes and alleles (tiers 1 and 2 alleles, or more?)
- Overall clinical sensitivity
- Test (technical) indications, specimen collection, turn-around-time (TAT)

Testing and Reporting

- Technical feasibility for important pharmacogenes and variants, i.e., *CYP2D6*, HLA loci, region of interest (ROI)
- Bioinformatic pipeline designed to call both common and rare PGx variants
- Star (*) alleles caller, diplotype, and drug metabolizer status prediction
- Rare/novel variant classification
- Handling copy number variation (CNV) results for pharmacogenes
- WES/WGS for PGx profiling, primary or secondary findings?
- Reportable range, i.e., what to include in the reports?

Clinical Implementation

- User-friendly PGx reports
- Availability of drug-gene pair-based clinical dosing guidelines for implementation
- Availability of pre- and post-testing counseling service
- Coverage and reimbursement policies
- Continuing monitoring patient's medication used based on preemptive PGx results, i.e., by electronic medical record (EMR) alert
- Longitudinal follow-up studies of test outcomes such as data on healthcare cost saving based on PGx results

https://academic.oup.com/jalm/article-abstract/9/1/50/7502984?redirectedFrom=fulltext&login=false

Updates on professional guidelines and standardization efforts

Professional Guidelines for Clinical PGx Implementation

- CAP MOL checklist
- CPIC guidelines
- ACMG Technical Standard (Tayeh et al. Genet Med. 2022)
- AMP PGx Working Group “Tiers 1 and 2” Allele Recommendations
- Other resources: PharmGKB, PharmVar, FDA Table of PGx Biomarkers in Drug Labeling and Table of PGx Associations, etc.

AMP Clinical PGx Working Group Recommendations

Goals

- To develop recommendations on a minimum set of variants for clinical PGx tests
- Can be used by the clinical PGx testing community as a reference for test development

Members

- Subject matter experts from the clinical PGx testing community
- Participating organizations: CAP, CPIC, PharmGKB, PharmVar, DPWG, ESPT, and ACMG

Documents

- *CYP2C19* Pratt VM, et al. *JMD*, 2018;20:269-276
- *CYP2C9* Pratt VM, et al. *JMD*, 2019;21:746-755
- Warfarin-Related Genes Pratt VM, et al. *JMD*, 2020;22:847-859
- *CYP2D6* Pratt VM, et al. *JMD*, 2021; 23:1047-1064
- *TPMT/NUDT15* Pratt VM, et al. *JMD*, 2022; 24:1079-1088
- *CYP3A4/CYP3A5* Pratt VM, et al. *JMD*, 2023; 25:619-629
- *DPYD* Pratt VM, et al. *JMD*, May 2024 *in press*

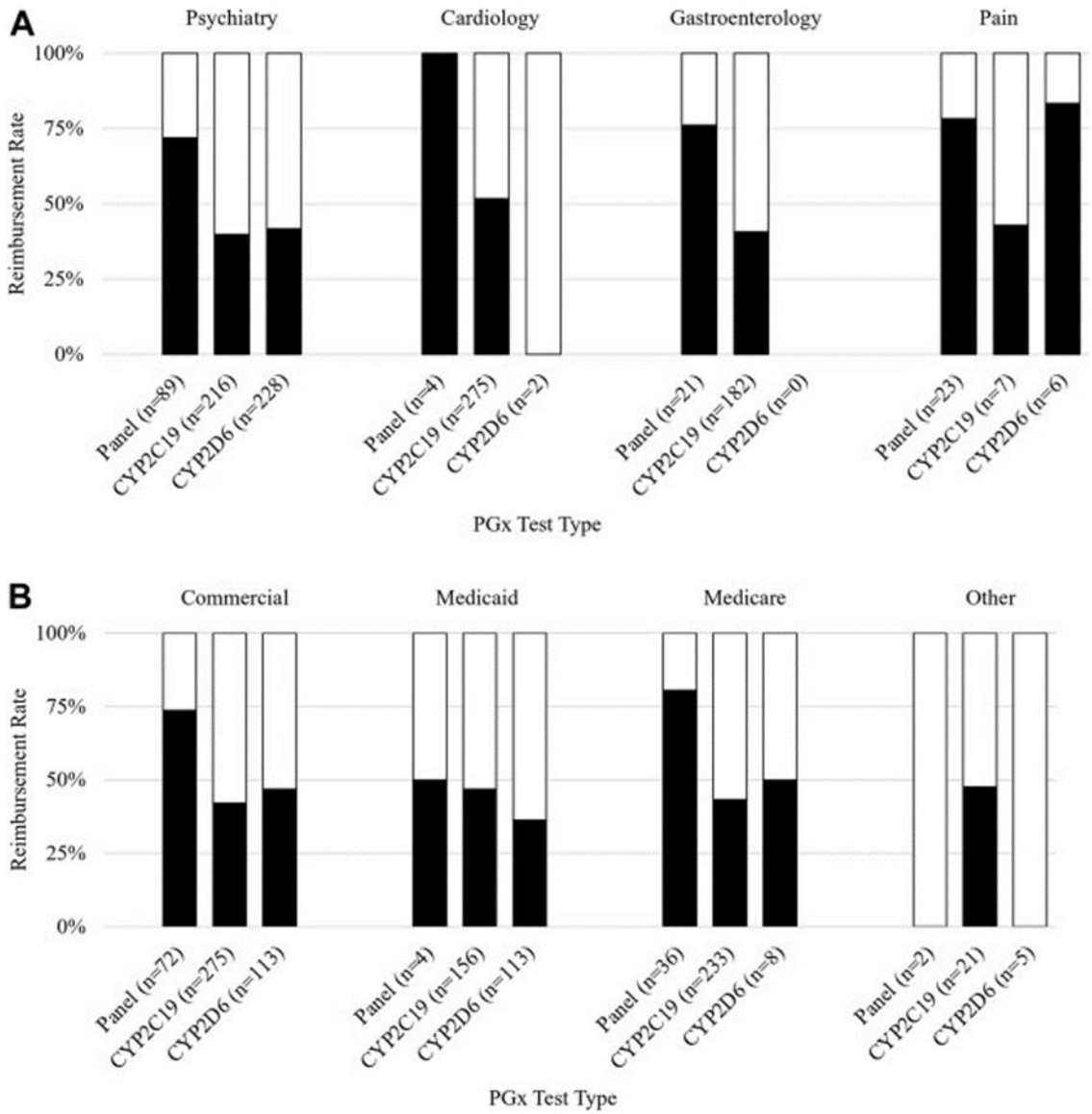
Additional Clinical Questions

- Allele recommendations for more genes
- Clinical validity and clinical utility of testing pharmacogenes
- PGx variant classifications (SNVs and CNVs) guidelines
- PGx LDT good practice (pre-analytical, analytical, and post-analytical)
- Points to consider when using NGS platforms for PGx profiling including NGS panels, whole exome, and genome sequencing
- Communicating and consultation on PGx results
- Considerations and standardizations of clinical implementations
- Training and competency of laboratory directors conducting PGx tests
- Others?

Reimbursement and regulatory updates

Reimbursement

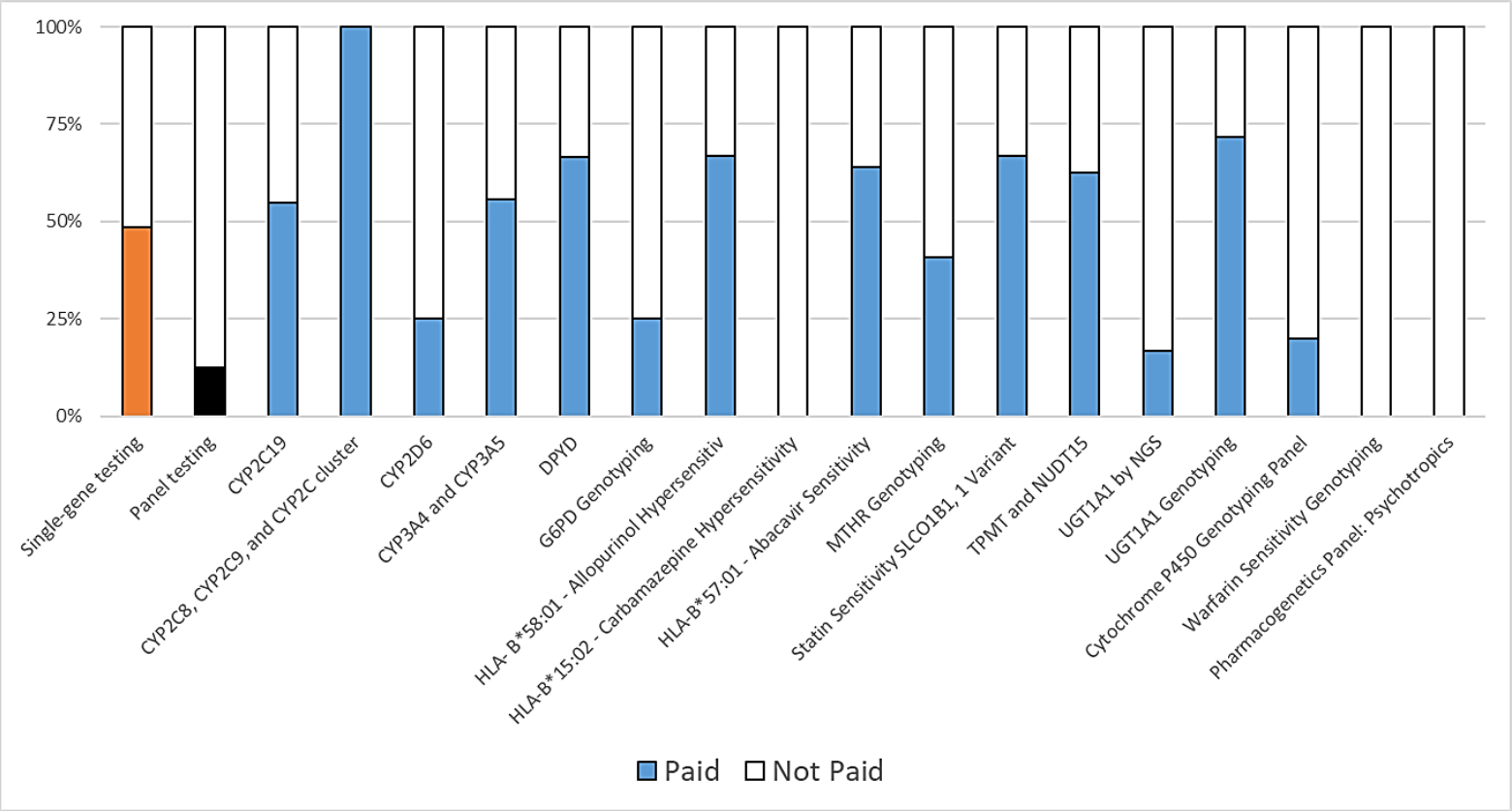
- A barrier for clinical implementation and equitable access
- 2-year data of reimbursement rate for 1,039 outpatient claims: 46%, with 36-48% across payers



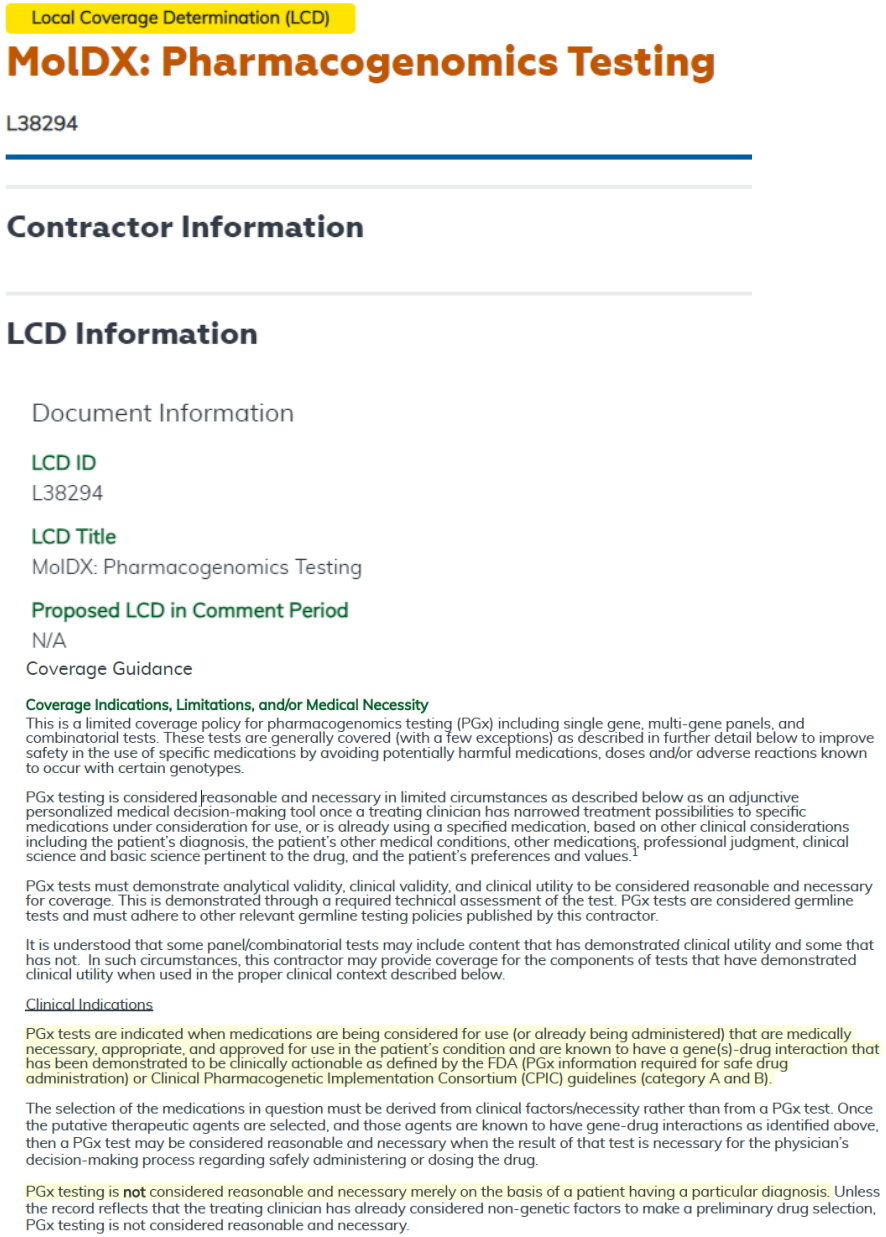
Lemke et al, Front Pharmacol. 2023; 14: 1179364.

PGx Third-Party Reimbursement Rate

The overall reimbursement rate for 2,023 PGx tests was 48% with 49% for single-gene PGx tests and 13% for PGx panel testing between 2021-2023



Data credited to ARUP MA team, 2023



Local Coverage Determination (LCD)

MolDX: Pharmacogenomics Testing

L38294

Contractor Information

LCD Information

Document Information

LCD ID
L38294

LCD Title
MolDX: Pharmacogenomics Testing

Proposed LCD in Comment Period
N/A

Coverage Guidance

Coverage Indications, Limitations, and/or Medical Necessity

This is a limited coverage policy for pharmacogenomics testing (PGx) including single gene, multi-gene panels, and combinatorial tests. These tests are generally covered (with a few exceptions) as described in further detail below to improve safety in the use of specific medications by avoiding potentially harmful medications, doses and/or adverse reactions known to occur with certain genotypes.

PGx testing is considered reasonable and necessary in limited circumstances as described below as an adjunctive personalized medical decision-making tool once a treating clinician has narrowed treatment possibilities to specific medications under consideration for use, or is already using a specified medication, based on other clinical considerations including the patient's diagnosis, the patient's other medical conditions, other medications, professional judgment, clinical science and basic science pertinent to the drug, and the patient's preferences and values.[1]

PGx tests must demonstrate analytical validity, clinical validity, and clinical utility to be considered reasonable and necessary for coverage. This is demonstrated through a required technical assessment of the test. PGx tests are considered germline tests and must adhere to other relevant germline testing policies published by this contractor.

It is understood that some panel/combinatorial tests may include content that has demonstrated clinical utility and some that has not. In such circumstances, this contractor may provide coverage for the components of tests that have demonstrated clinical utility when used in the proper clinical context described below.

Clinical Indications

PGx tests are indicated when medications are being considered for use (or already being administered) that are medically necessary, appropriate, and approved for use in the patient's condition and are known to have a gene(s)-drug interaction that has been demonstrated to be clinically actionable as defined by the FDA (PGx information required for safe drug administration) or Clinical Pharmacogenetic Implementation Consortium (CPIC) guidelines (category A and B).

The selection of the medications in question must be derived from clinical factors/necessity rather than from a PGx test. Once the putative therapeutic agents are selected, and those agents are known to have gene-drug interactions as identified above, then a PGx test may be considered reasonable and necessary when the result of that test is necessary for the physician's decision-making process regarding safely administering or dosing the drug.

PGx testing is **not** considered reasonable and necessary merely on the basis of a patient having a particular diagnosis. Unless the record reflects that the treating clinician has already considered non-genetic factors to make a preliminary drug selection, PGx testing is not considered reasonable and necessary.

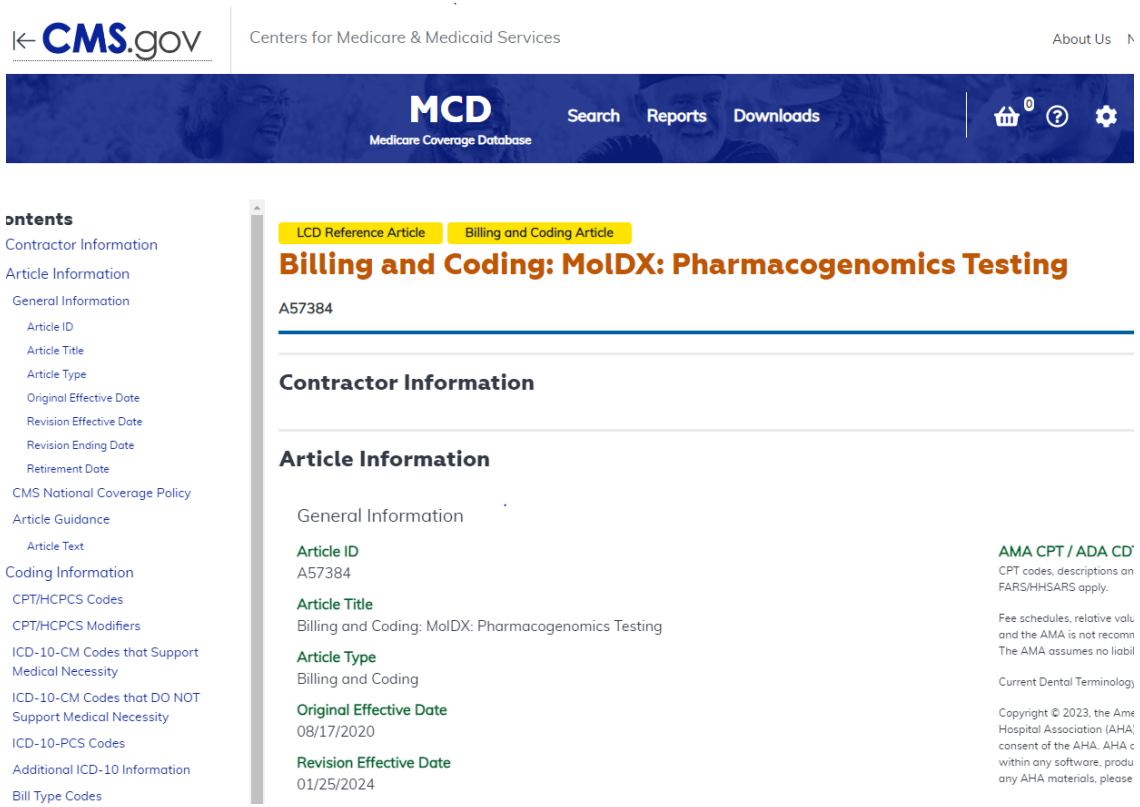
Specific Coverage Guidelines

- Clinical indications and definitions
- Coverage information
- Test components that are not reasonable and necessary
- Technical requirements (for clinical laboratories performing the test)
- Specific documentation requirements
- Noncovered indications
  - PGx testing is not covered when a treating clinician is not considering treatment with a medication that has an actionable drug-gene interaction, or when the use of the medication with a drug-gene interaction is not reasonable and necessary.

https://www.cms.gov/medicare-coverage-database/

Updates on Billing and Coding

- Records of the drugs under consideration for use or in use by the ordering physician that necessitate the use of ordered test are required (updated on Jan 25, 2024)
- Only one test may be performed per date of service; the test should be the most likely to identify the necessary alleles/variants for drug/drugs in question. This applies to both single gene tests and multigene panels.
- Covered multigene panels with a specific intended use such as major depressive disorder (MDD) or neuropsychiatric must include relevant ICD-10 codes (provided)
- Table 1: CPT coding for gene/drug associations from CPIC and FDA sources
- Table 2: Relevant therapeutic gene/drug associations from CPIC and FDA sources
- 35 CPT codes in Group 1 includes both multi-gene panels and single-gene tests



https://www.cms.gov/medicare-coverage-database/

United Healthcare Z-Code Requirements

Scope of requirement: for certain genetic tests including clinical PGx tests

Z-Codes: a five-character alphanumeric code assigned to molecular diagnostic tests by Palmetto GBA's MolDX program. Z-codes are not replacing the CPT® codes but are used in conjunction with the CPT® codes on lab claims

UHC policy will initially cover 133 CPT® codes and 104 PLA codes, but will likely expand to molecular infectious disease tests

UHC currently covers 27.3 million members who use commercial healthcare issuance plan

Clinical labs: must register with the DEX Diagnostic Exchange, and must register each unique test performed in-house in the DEX System with required information for review as Technical Assessment (with assignment of the Z-code and CPT to the test)

First announced official effective date of August 1, 2023; updated date: from April 1, 2024, to June 1, 2024, for phase 1 planned rollout

Send-out labs billing UHC for their tests: need to request the performing lab to share the Z-codes assigned

https://www.palmettogba.com/palmetto/moldxv2.nsf

https://www.uhcprovider.com/en/resource-library/news/2024/ensure-molecular-tests-have-z-code.html

FDA Final Rule on LDTs


FDA

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FDA NEWS RELEASE

FDA Proposes Rule Aimed at Helping to Ensure Safety and Effectiveness of Laboratory Developed Tests

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For Immediate Release:

September 29, 2023

ARUP Resources for FDA's Final Rule on LDTs

https://www.aruplab.com/fda-ldt-final-rule


FDA NEWS RELEASE

FDA Takes Action Aimed at Helping to Ensure the Safety and Effectiveness of Laboratory Developed Tests

Share Post Linkedin Email Print

For Immediate Release: April 29, 2024

https://www.fda.gov/news-events/press-announcements/fda-takes-action-aimed-helping-ensure-safety-and-effectiveness-laboratory-developed-tests

FDA and PGx

The FDA Warns Against the Use of Many Genetic Tests with Unapproved Claims to Predict Patient Response to Specific Medications: FDA Safety Communication

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April 4, 2019 UPDATE: Following issuance of the safety communication, the FDA has taken additional actions. Please see the FDA

FDA STATEMENT

FDA Announces Collaborative Review of Scientific Evidence to Support Associations Between Genetic Information and Specific Medications

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FDA U.S. FOOD & DRUG ADMINISTRATION

Home / Drugs / Science and Research | Drugs / Table of Pharmacogenomic Biomarkers in Drug Labeling

Table of Pharmacogenomic Biomarkers in Drug Labeling

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WARNING LETTER

Inova Genomics Laboratory

MARCS-CMS 577422 – APRIL 04, 2019

FDA has not created a legal "carve-out" for LDTs such that they are not required to comply with the requirements under the Act that otherwise would apply. FDA has never established such an exemption. As a matter of practice, FDA, however, has exercised enforcement discretion for LDTs, which means that FDA has generally not enforced the premarket review and other FDA legal requirements that do apply to LDTs. Although FDA has generally exercised enforcement discretion for LDTs, the Agency always retains discretion to take action when appropriate, such as when it is appropriate to address significant public health concerns.

Based on the above, FDA has determined that the MediMap tests are adulterated under section 501(f)(1)(B) of the Act, 21 U.S.C. § 351(f)(1)(B), because your firm does not have an approved application for premarket approval (PMA) in effect pursuant to section 515(a) of the Act, 21 U.S.C. § 360e(a), or an approved application for an investigational device exemption under section 520(g) of the Act, 21 U.S.C. § 360j(g). The MediMap tests are also misbranded under section 502(o) of the Act, 21 U.S.C. § 352(o), because your firm did not notify the Agency of its intent to introduce the devices into commercial distribution, as required by section 510(k) of the Act, 21 U.S.C. § 360(k). For a device requiring premarket approval, the notification required by section 510(k) is deemed satisfied when a PMA is pending before the Agency. (See 21 CFR 807.81(b)). Information that may be helpful in preparing a premarket submission is available at

Table of Pharmacogenetic Associations

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Precision Medicine

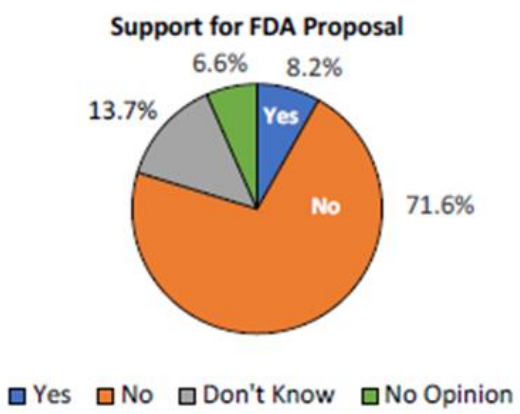
Table of Pharmacogenetic Associations

Pharmacogenetic tests, along with other information about patients and their disease or condition, can play an important role in drug therapy. When a health care provider is considering prescribing a drug, knowledge of a patient's genotype may be used to aid in determining a therapeutic strategy, determining an appropriate dosage, or assessing the likelihood of benefit or toxicity.

LDT Customer Survey

- Led by Dr. Jonathan Genzen, CMO, ARUP Laboratories

a



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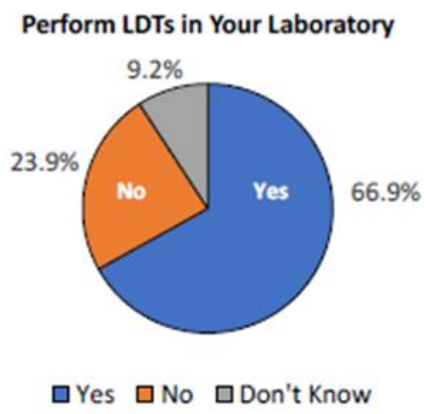

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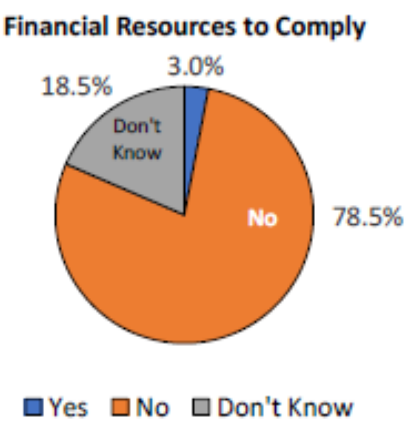

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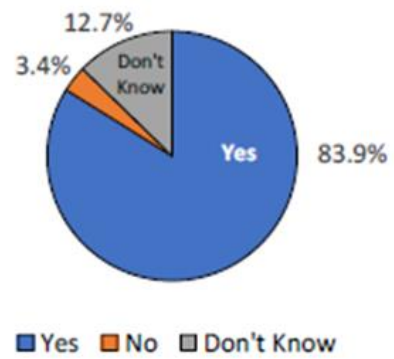

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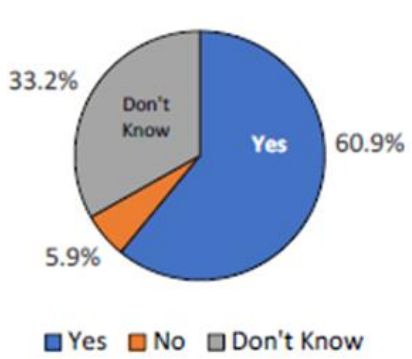

Respondents: N=532, 45 US states

https://www.medrxiv.org/content/10.1101/2024.02.28.24303459v2

FDA Cleared or Approved PGx Tests

Drug Metabolizing enzymes	xTAG CYP2D6 Kit v3	Luminex Molecular Diagnostics, Inc.	K130189, K093420
Drug Metabolizing enzymes	xTAG CYP2D6 Kit v3	Luminex Molecular Diagnostics, Inc.	K130189, K131565
Drug Metabolizing enzymes	Spartan RX CYP2C19 Test System	Akonni Biosystems Inc.	K183530
Drug Metabolizing enzymes	TruDiagnosis System	Akonni Biosystems Inc	K183530
Drug Metabolizing enzymes	Verigene CYP2C 19 Nucleic Acid Test	Nanosphere, Inc.	K120466
Drug Metabolizing enzymes	INFINITI CYP2C19 Assay	AutoGenomics, Inc.	K101683
Drug Metabolizing enzymes	Invader UGT1A1 Molecular Assay	Third Wave Technologies Inc.	K051824
Drug Metabolizing enzymes	Roche AmpliChip CYP450 microarray	Roche Molecular Systems, Inc.	K043576, K042259
Drug Metabolizing enzymes	eSensor Warfarin Sensitivity Saliva Test	GenMark Diagnostics	K110786
Drug Metabolizing enzymes	eQ-PCR LC Warfarin Genotyping kit	TrimGen Corporation	K073071
Drug Metabolizing enzymes	eSensor Warfarin Sensitivity Test and XT-8 Instrument	Osmetech Molecular Diagnostics	K073720
Drug Metabolizing enzymes	Gentris Rapid Genotyping Assay - CYP2C9 & VKORC1	ParagonDx, LLC	K071867
Drug Metabolizing enzymes	INFINITI 2C9 & VKORC1 Assay for Warfarin	AutoGenomics, Inc.	K073014
Drug Metabolizing enzymes	Verigene Warfarin Metabolism Nucleic Acid Test and Verigene System	Nanosphere, Inc.	K070804
Pharmacogenetic Reports	23ANDME PERSONAL GENOME SERVICE (PGS) PHARMACOGENETIC REPORTS	23andMe	DEN180028, DEN180028, K193492, K221885

https://www.fda.gov/medical-devices/in-vitro-diagnostics/nucleic-acid-based-tests

AUG 18, 2020 - NEWS

FDA Clearance for 23andMe Drug Interaction Report

This week, the U.S. Food and Drug Administration (FDA) granted 23andMe a 510(k) clearance for a pharmacogenetics report on two medications, clopidogrel, prescribed for certain heart conditions, and citalopram, which is prescribed for depression.

CYP2C19

The decision this week modifies the labeling of the previously authorized CYP2C19 Drug Metabolism report which was granted FDA authorization in 2018. The new 510(k) clearance for the pharmacogenetics report for CYP2C19 modifies the labeling to remove the need for confirmatory testing and allows 23andMe to report interpretive drug information for two medications.

"This impactful pharmacogenetics information can now be delivered without the need for confirmatory testing, a testament to the clinical validity of 23andMe results," said Kathy Hibbs, 23andMe Chief Legal, and Regulatory Officer. "23andMe remains the only company with direct-to-consumer pharmacogenetic reports cleared by the FDA. Now that we have pioneered a regulatory path, we believe all companies marketing

https://blog.23andme.com/articles/pharmacogenetics-report

Potential Patient Impacts

- Decrease patient's access to quality clinical PGx tests
- Added regulatory burdens and laboratory logistic cost will likely to be distributed among laboratories, patients, healthcare system, payers etc.
- Inflexibility to assay modifications (platform/technological advancements, specimen types, new clinical evidences, professional guidelines updates etc.)
- Discourage innovation, esoteric testing, and testing for patients with rare diseases

Summary

- Clinical PGx tests are a group of unique genetic tests that can be beneficial to many
- Professional guidelines are being developed to provide guidance and resources for clinical PGx testing and implementation
- Updates on clinical PGx test coverage policies provide further clarification and requirements on coverage
- Clinical indication or necessity is the key for payment
- Preemptive PGx testing is not currently supported health insurance coverage policies
- Regulatory: FDA's new rule on LDTs will likely reshape the clinical laboratory industry and will further compound the existing challenges for patients' access to clinical PGx tests

Patient story

YJ

Yuan 100%

East Asian 99.9%

- **Chinese** 99.1%
  - **Northern Chinese & Tibetan** 91.7%
    Shandong, Mainland China (northern provinces)
    +9 regions
  - **Southern Chinese & Taiwanese** NEW 7.4%
    Jiangsu, Mainland China (southern provinces)
    Kaohsiung City, Taiwan
    +11 regions
- **Korean** 0.8%

Trace Ancestry 0.1%

We detected traces of the following populations in your DNA. Read more about trace ancestry in the FAQ.

- **Broadly European** 0.1%

People with your genetic result are predicted to be **CYP2C19 intermediate metabolizers** and may process some medications slightly slower than normal. However, since many factors impact how medications are processed, the variant detected may have no noticeable effects on how you process medications.

1 variant detected
Predicted CYP2C19 intermediate metabolizer

VARIANT(S) DETECTED	OVERALL FUNCTIONAL EFFECT
*2 (one copy)	Decreased enzyme function

Predicted CYP2C19 intermediate metabolizer

People who are predicted to be CYP2C19 intermediate metabolizers may process some medications slightly slower than normal, but most medications won't be affected.

Depending on the medication, being a CYP2C19 intermediate metabolizer may lead to higher or lower than normal medication levels in the body, or have no noticeable effects.

CYP2C19 Metabolism

Acknowledgement

ARUP team

- Dr. Sherin Shaaban: Medical Director for pharmacogenomics
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- PGx R&D Scientists
- Technologists, ARUP Molecular Genetics and Fragment Analysis Labs
- Valerie Collier: Palmetto technical assessment expert

External collaborators

- Drs. Vicky Pratt, Karen Weck, and AMP PGx Working Group
- Dr. Ann Moyer and CAP PGx Best Practice Working Group
- CLSI MM29 Project Team
- PGx education project team: Drs. Reynold Ly at Nationwide Children's, and Marwan Tayeh, Indiana University of School of Medicine

Additional Resources

LabMind - An Interview With Dr. Yuan Ji:

Bringing Pharmacogenomics Into Mainstream Clinical Practice hosted by Brian R. Jackson, MD, MS

https://arup.utah.edu/education/shortTopics.php#podcasts

https://youtu.be/zwUAdFwfvG0?si=mH3K0YYList4oadq

https://open.spotify.com/episode/4B5CWOHtRAed8M4gzAnh51

- ARUP Consult® Germline Pharmacogenetics https://arupconsult.com/content/germline-pharmacogenetics
- ARUP Resources on the FDA's Final Rule on LDTs https://www.aruplab.com/fda-ldt-final-rule

- AMP Clinical Practice Guidelines and Reports including AMP PGx Working Group documents https://www.amp.org/clinical-practice/practice-guidelines/
- AMP Best Practice for Clinical Pharmacogenomic Testing https://www.amp.org/AMP/assets/File/position-statements/2019/Best_Practices_for_PGx_9_4_2019.pdf?pass=83
- Drug Interaction Flockhart TableTM https://medicine.iu.edu/internal-medicine/specialties/clinical-pharmacology/drug-interaction-flockhart-table
- PharmGKB and Clinical Guideline Annotations https://www.pharmgkb.org/guidelineAnnotations
- Clinical Pharmacogenetics Implementation Consortium (CPIC) https://cpicpgx.org/
- Pharmacogene Variation Consortium (PharmVar) https://www.pharmvar.org/
- Genetic Testing Reference Materials Coordination Program (GeT-RM) https://www.cdc.gov/labquality/get-rm
- FDA Table of Pharmacogenetic Associations https://www.fda.gov/medical-devices/precision-medicine/table-pharmacogenetic-associations
- FDA Table of Pharmacogenomic Biomarkers in Drug Labeling https://www.fda.gov/drugs/science-and-research-drugs/table-pharmacogenomic-biomarkers-drug-labeling
- ClinGen Pharmacogenomics (PGx) Working Group https://clinicalgenome.org/working-groups/pharmacogenomics/
- Pharmacogenomics Global Research Network (PGRN) https://www.pgrn.org/
- FDA LDT rulemaking https://www.fda.gov/medical-devices/in-vitro-diagnostics/laboratory-developed-tests

Thank You!

Questions or Comments?

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ARUP is a nonprofit enterprise of the University of Utah and its Department of Pathology.