



# Genomics in Action:

Integrating Pharmacogenomics  
into Specialty Pharmacy Workflow

St. Jude Children's Research Hospital

Memphis, TN

Encore Presentation: 2025 Annual Meeting & Expo Highlights

# Meet the Presenters



**Tiffany Nason, PharmD**

Manager-Specialty Pharmacy



**Timothy Howze, PharmD**

Senior Clinical Staff Pharmacist



# Disclaimer

This presentation is for educational purposes only and does not constitute medical advice. Information provided is based on current knowledge and clinical consideration but may change over time. Always consult official sources.



# Disclosure

We have no relevant financial conflicts of interest in relation to this activity to disclose



# Learning Objectives

- Describe the factors that are used to define specialty medication
- Describe how a specialty program in collaboration with pharmacogenomic expertise can ensure patients receive the most effective therapies for complex, high risk conditions
- Discuss three specialty medications that have phenotypic implications and when to intervene if genomic information is available for patients





# Defining Specialty Medications

# Specialty Medication



## Complex

Higher complexity; comprehensive patient care, clinical managements and product support



## Serious Conditions

Drug is utilized to treat serious, life threatening, and/or catastrophic conditions



## Administration Routes

Oral, injection and/or infusion—typically administered within an infusion center



## Special Handling

May have special administration, storage and delivery requirements



## Access Restrictions

Special access conditions by the manufacturer

# Specialty Medications

- High-cost prescription drugs
- **No single universal definition**
- Often defined by **payers, providers, manufacturers, and pharmacies** differently



# Other Key Definitions

Specialty Pharmacy (SP): Dispenses and manages medications for complex, chronic, or rare conditions.

Genomics: Study of genes and their functions.

- Pharmacogenomics (PGx): The study of genetic differences that influence drug response.

Clinical goal: To tailor medication therapy using a patient's genetic profile, improving outcomes and reducing adverse effects.



# Specialty Medications with PGx Association

## Testing Required

- Abacavir
- Capecitabine - ERBB2
- Cetuximab
- Dabrafenib
- Eliglustat
- Erdafitinib – FGFR3
- Gefitinib - EGFR
- Ivacaftor (+ combination products)
- Lapatinib
- Nilotinib - ABL1, BCR
- Pegloticase
- Siponimod
- Tetrabenazine
- Trametinib

## Testing Recommended

- Capecitabine - DPYD
- Mercaptopurine

## Actionable PGx

- Abrocitinib
- Amifampridine
- Amikacin
- Avatrombopag - F2, F5, PROC, PROS1, SERPINC1
- Belzutifan
- Deutetrabenazine
- Eltrombopag
- Erdafitinib - CYP2C9
- Gefitinib - CYP2D6
- Pitolisant
- Tobramycin
- Valbenazine

## Informative PGx

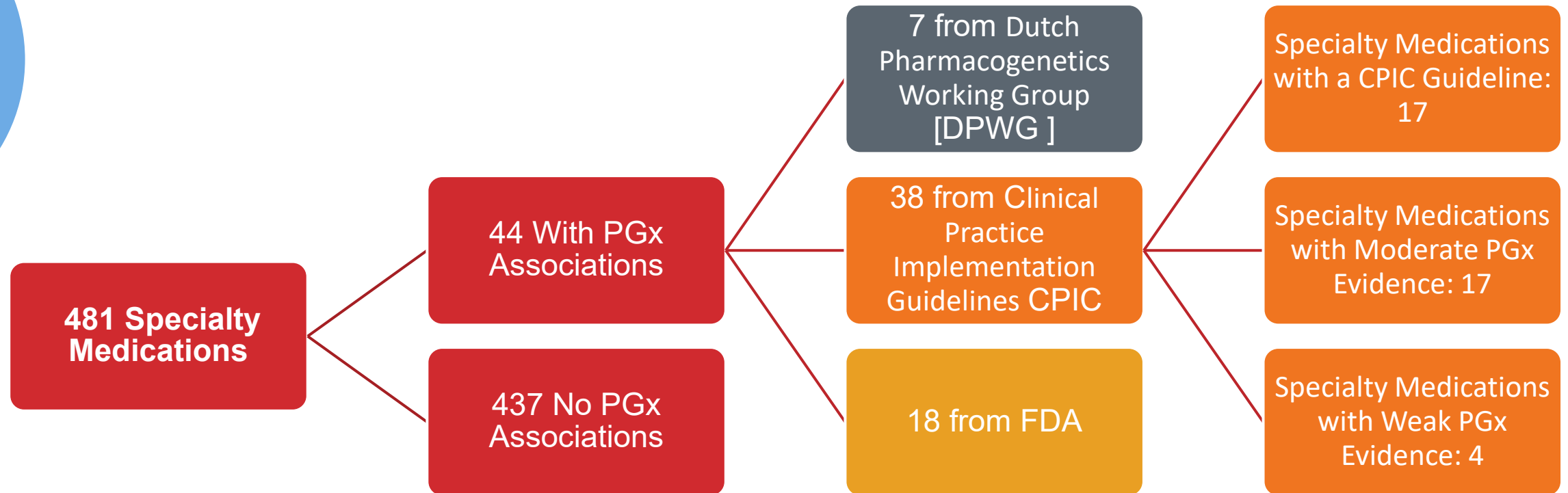
- Avatrombopag - CYP2C9
- Nilotinib - UGT1A1
- Pazopanib
- Peginterferon alfa-2a

## Criteria Not Met

- Carglumic acid
- Rituximab
- Trametinib - HRAS, KRAS, NRAS, G6PD
- Velaglucerase alfa



# Results: Roughly 10% of Specialty Medications Have PGx Associations



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Genes-drugs. CPIC. <https://cpicpgx.org/genes-drugs/>

Clinical Guideline Annotations. PharmGKB. <https://www.pharmgkb.org/guidelineAnnotations>

FDA. Table of pharmacogenetic associations. <https://www.fda.gov/medical-devices/precision-medicine/table-pharmacogenetic-associations>



# PGx Relevance Beyond Specialty Medications

Drug category	Patients, n (%)	Age when initiated, median (range)	Genes (drugs) with CPIC guidelines
Serotonin 5-HT <sub>3</sub> receptor antagonists: ondansetron <sup>†</sup> , granisetron <sup>‡</sup> , palonosetron <sup>‡</sup> , dolasetron <sup>‡</sup>	693 (72.4)	18 (0–86)	CYP2D6 (ondansetron)
Opioids: codeine <sup>†</sup> , hydrocodone <sup>†</sup> , tramadol <sup>†</sup> , hydromorphone <sup>‡</sup> , morphine <sup>‡</sup> , oxycodone <sup>‡</sup> , fentanyl <sup>‡</sup>	814 (85.1)	14 (0–86)	CYP2D6 (codeine, hydrocodone, tramadol)
Proton pump inhibitors: dexlansoprazole <sup>†</sup> , esomeprazole <sup>†</sup> , lansoprazole <sup>†</sup> , omeprazole <sup>†</sup> , pantoprazole <sup>†</sup> , rabeprazole <sup>†</sup>	324 (33.9)	25 (0–85)	CYP2C19 (dexlansoprazole, esomeprazole, lansoprazole, omeprazole, pantoprazole, rabeprazole)
NSAIDs: aspirin <sup>‡</sup> , ibuprofen <sup>†</sup> , meloxicam <sup>†</sup> , naproxen <sup>‡</sup>	774 (80.9)	13 (0–85)	CYP2C9 (ibuprofen, naproxen)
Selective serotonin reuptake inhibitors: citalopram <sup>†</sup> , escitalopram <sup>†</sup> , fluvoxamine <sup>†</sup> , paroxetine <sup>†</sup> , sertraline <sup>†</sup>	121 (12.6)	28 (8–93)	CYP2C19 (citalopram, escitalopram, sertraline), CYP2D6 (fluvoxamine, paroxetine)
Tricyclic antidepressants: amitriptyline <sup>†</sup> , clomipramine <sup>†</sup> , desipramine <sup>†</sup> , doxepin <sup>†</sup> , imipramine <sup>†</sup> , nortriptyline <sup>†</sup> , trimipramine <sup>†</sup>	71 (7.4)	27 (4–78)	CYP2D6 (amitriptyline, nortriptyline, desipramine, trimipramine), CYP2C19 (amitriptyline, clomipramine, doxepin, imipramine, trimipramine)
Anticonvulsants: carbamazepine <sup>†</sup> , oxcarbazepine <sup>†</sup>	20 (2.1)	23 (2–69)	HLA-A (carbamazepine), HLA-B (oxcarbazepine)
Allopurinol <sup>†</sup>	17 (1.8)	56 (11–74)	HLA-B
Clopidogrel <sup>†</sup>	25 (2.6)	65 (21–77)	CYP2C19
Voriconazole <sup>†</sup>	10 (1.0)	8 (1–72)	CYP2C19
Tacrolimus <sup>†</sup>	13 (1.4)	36 (7–71)	CYP3A5
Warfarin <sup>†</sup>	60 (6.3)	38.5 (3–85)	CYP2C9
Phenytoin <sup>†</sup>	11 (1.1)	8 (0–33)	CYP2C9

<sup>†</sup>Drugs with CPIC guideline recommendations for dosing adjustment based on genotype.  
<sup>‡</sup>Alternative therapeutics to drugs with CPIC guidelines.  
 CPIC: Clinical Pharmacogenetics Implementation Consortium; Non-steroidal anti-inflammatory drugs.

- PGx testing includes common medications such as codeine, statins, and warfarin—providing added value for specialty patients.

- Literature review of hematology patients (2015–2021) within Indiana University Health System.



# St. Jude Clinical Specialties

- **Pediatric Oncology – Leukemia, lymphoma, solid tumors**
- **Hematology (SP) – Sickle cell disease, thalassemia**
- **Neuro-Oncology & Neurology – Brain/spinal tumors, neurogenetic disorders**
- **Infectious Diseases (SP) – HIV, immunocompromised care**
- **Immunology – Rare immune deficiencies**
- **Genetic Syndromes – Hereditary cancer predispositions**
- **Transplant & Cellular Therapy – Stem cell, CAR-T therapy**
- **Radiology & Imaging – Diagnostic and treatment planning**
- **Pathology – Molecular, genetic diagnostics**
- **Psychology – Neuropsych, behavioral health**





Comprehensive Patient Care  
*should include*  
Pharmacogenomics when  
available

# Comprehensive Care Includes Pharmacogenomics: Literature Review

Principi, N.; Petropulacos, K.; Esposito, S. Impact of Pharmacogenomics in Clinical Practice. *Pharmaceuticals* 2023, 16, 1596.

Review of trials across various drug classes (e.g., clopidogrel, warfarin, thiopurines, antidepressants) shows that pharmacogenomics-guided therapy often offers meaningful benefits, with fewer adverse effects and better efficacy.

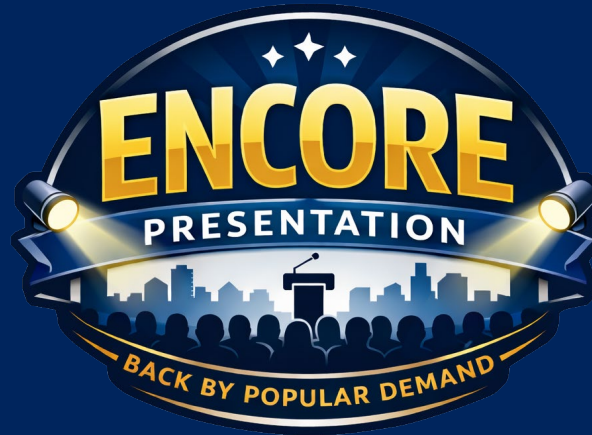
Cicali EJ, Lemke L, Al Alshaykh H, et al. How to Implement a Pharmacogenetics Service at your Institution. *J Am Coll Clin Pharm.* 2022 Nov;5(11):1161-1175.

Guidance from early adopters highlights that pharmacogenetics usage can improve outcomes, reduce adverse effects, and lower healthcare spending, with successful implementation requiring multi-disciplinary coordination.

Jennifer K. Hockings, Amy L. Pasternak, Angelika L. Erwin, et al. PG: An evolving clinical tool for precision medicine. *Cleveland Clinic Journal of Medicine* Feb 2020, 87 (2) 91-99.

A review in the *Cleveland Clinic Journal of Medicine* explains that pharmacogenomics can guide drug selection and dosing to improve treatment outcomes, reduce adverse events (including life-threatening ones), and be cost-effective—stressing that many patients carry genetic variants impacting drug response.





# Pharmacogenomics Introduction

# History of PGx at St. Jude (Oncology)

TPMT Testing for  
Select Patients  
(Leukemia Diagnosis)

1991

2005

UGT1A1 Testing for  
Select Patients

2006

2007

CYP2D6 Testing for  
Select Patients

Clinical  
Pharmacogenetic  
Implementation  
Consortium (CPIC)  
Established

2009

Institution-wide  
Testing  
Initiation of PG4KDS  
(Multi-gene, Panel  
Genotyping)

2011

First CPIC Guideline  
Published

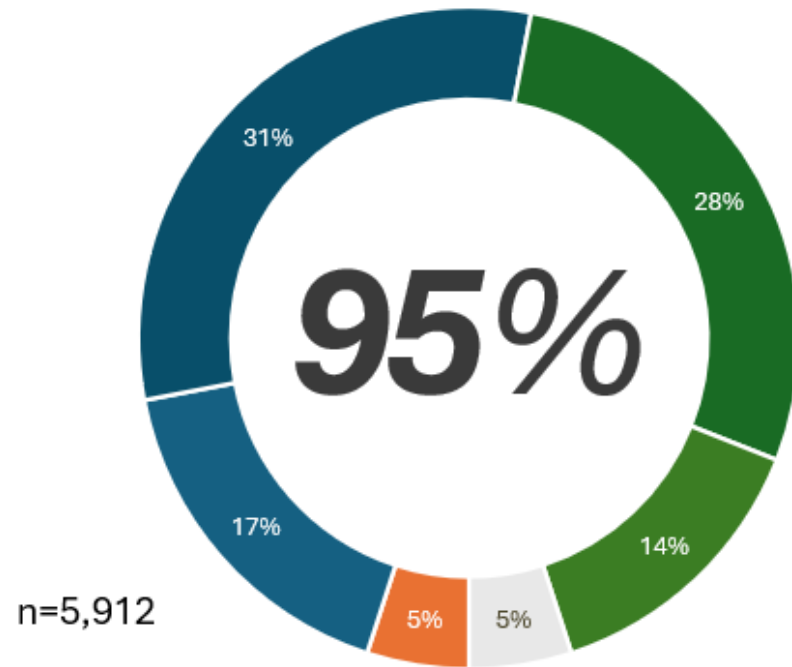
2012

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Adapted from Crews KR, et al. *Am J Health Syst Pharm.* 2011;68:143.

# Frequency of Actionable Pharmacogenomic Test Results at St. Jude



of patients genotyped on the **PG4KDS** protocol had at least one high-risk pharmacogenomic in their health record

■ Zero high-risk results

■ 1 high-risk result

■ 2 high-risk results

■ 3 high-risk results

■ 4 high-risk results

■ 5 or more high-risk results

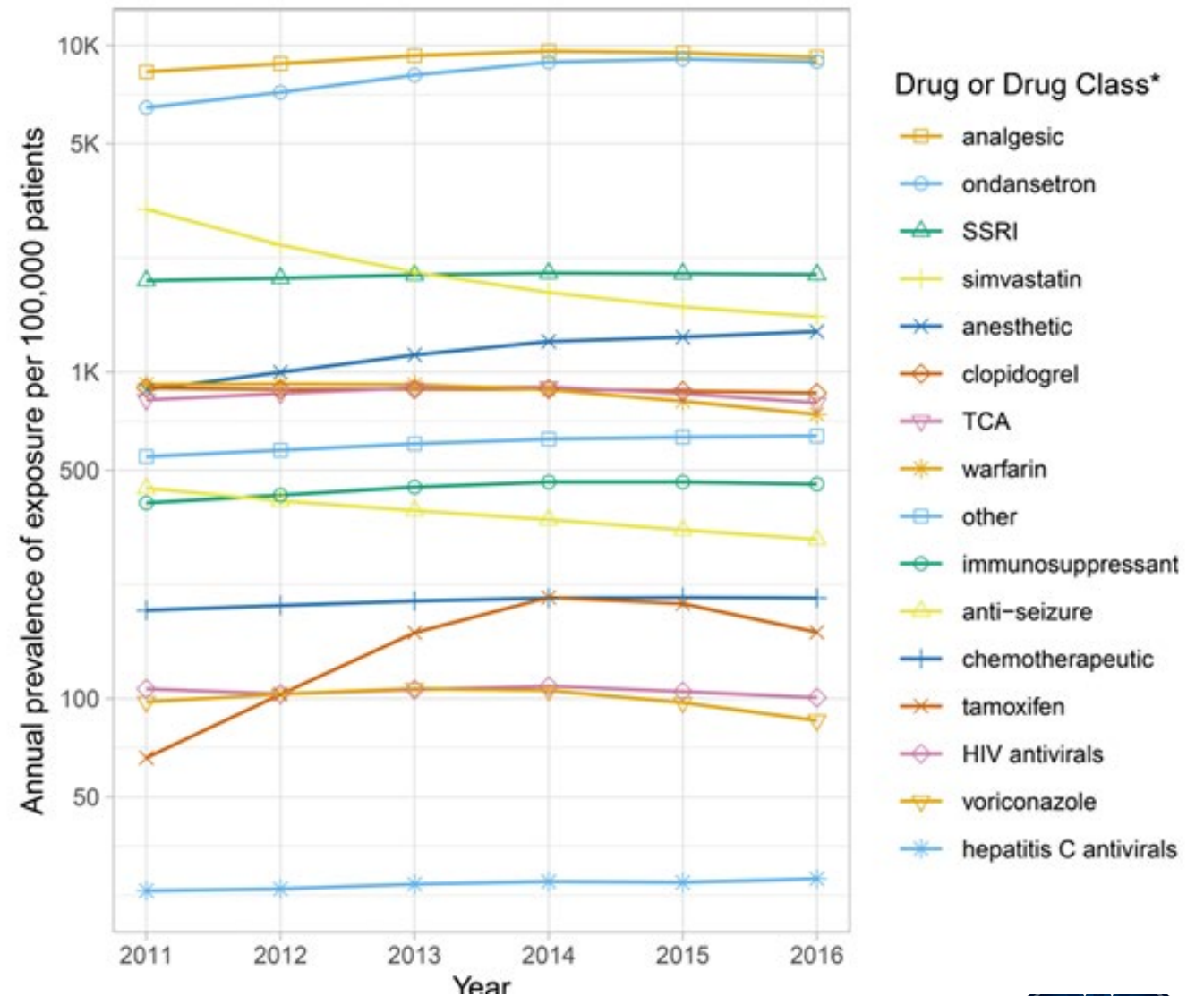
Haidar CE, et al. *Annu Rev Genomics Hum Genet.* 2022;23:449.

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# 11 US Health Systems:

Most common medications/  
classes that  
have PGx  
action-ables



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Confidential <https://pubmed.ncbi.nlm.nih.gov/33428770/>.



Permission to use by Hicks JK.



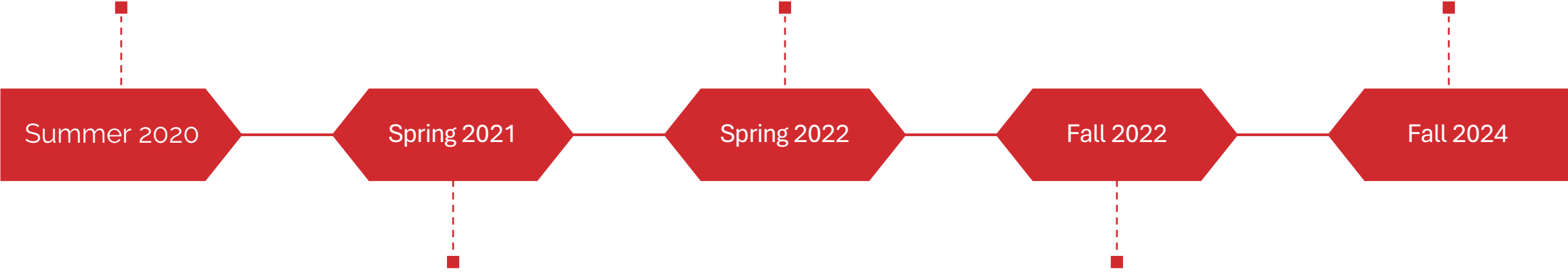
# Implementing Pharmacogenomics in Specialty Pharmacy Practice

# St. Jude Specialty Pharmacy (SP)

SP established at St. Jude with ID patients on antiretrovirals

ACHC accreditation until present. Added hematology patients.

PGx collaboration with specialty went live



URAC accreditation until present

Institution change to Epic EHR & Compass Rose for SP



# Specialty Pharmacy's Role in PGx Enrollment and Clinical Use

## Specialty Pharmacy Technicians

- Review charts for availability of pharmacogenomic test results
- If no genetic information known and meet inclusion criteria, notate patient as candidate to approach
- Document as task in Specialty Pharmacy Patient Management System

## Specialty Pharmacy Pharmacists

- Screen and refer new patients.
- Review medication list at each assessment and compare to problem list for any medications of concern
- Review PowerBI tool for phenotypic medications that may need intervention
- Document Intervention in EHR\* template note /Specialty Pharmacy Patient Management System



# Pharmacist Initial Assessment Example

## Step 1:

### Pharmacogenomics:

Patient .

## Step 2:

### Pharmacogenomics:

Patient has not had pharmacogenomics testing. See "Assess PG4KDS enrollment" checklist task for further information.

### Pharmacogenomics:

Patient has had pharmacogenomics testing. Upon review of medication list, patient .

## Step 3:

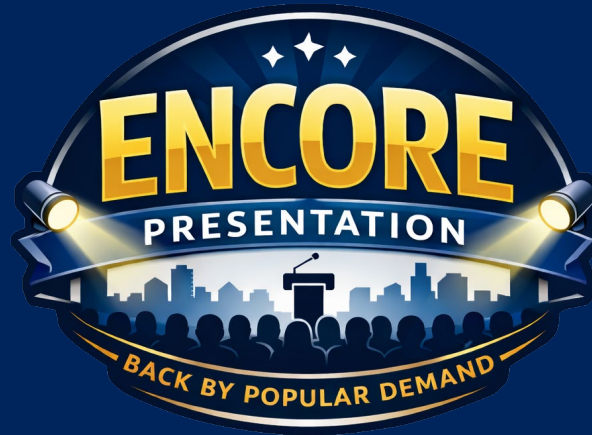
### Pharmacogenomics:

Patient has had pharmacogenomics testing. Upon review of medication list, patient is not currently taking medications (including specialty medications) affected by the pharmacogenomic test result listed in patient's problem list.


### Pharmacogenomics:

Patient has had pharmacogenomics testing. Upon review of medication list, patient is currently taking medications (including specialty medications) affected by the pharmacogenomic test result listed in patient's problem list. Phenotype:  / Medication(s):  Pharmacogenomics pharmacy team follow up  required.





# PGx Review of Specialty Patients at Health System Based Specialty Pharmacy (October 2024 - April 2025)



# PGx Inclusion Criteria

Prescribed a  
specialty  
medication

Opted in to  
specialty  
pharmacy  
services

Provided  
written consent

Patients must  
be 18 years or  
older, or

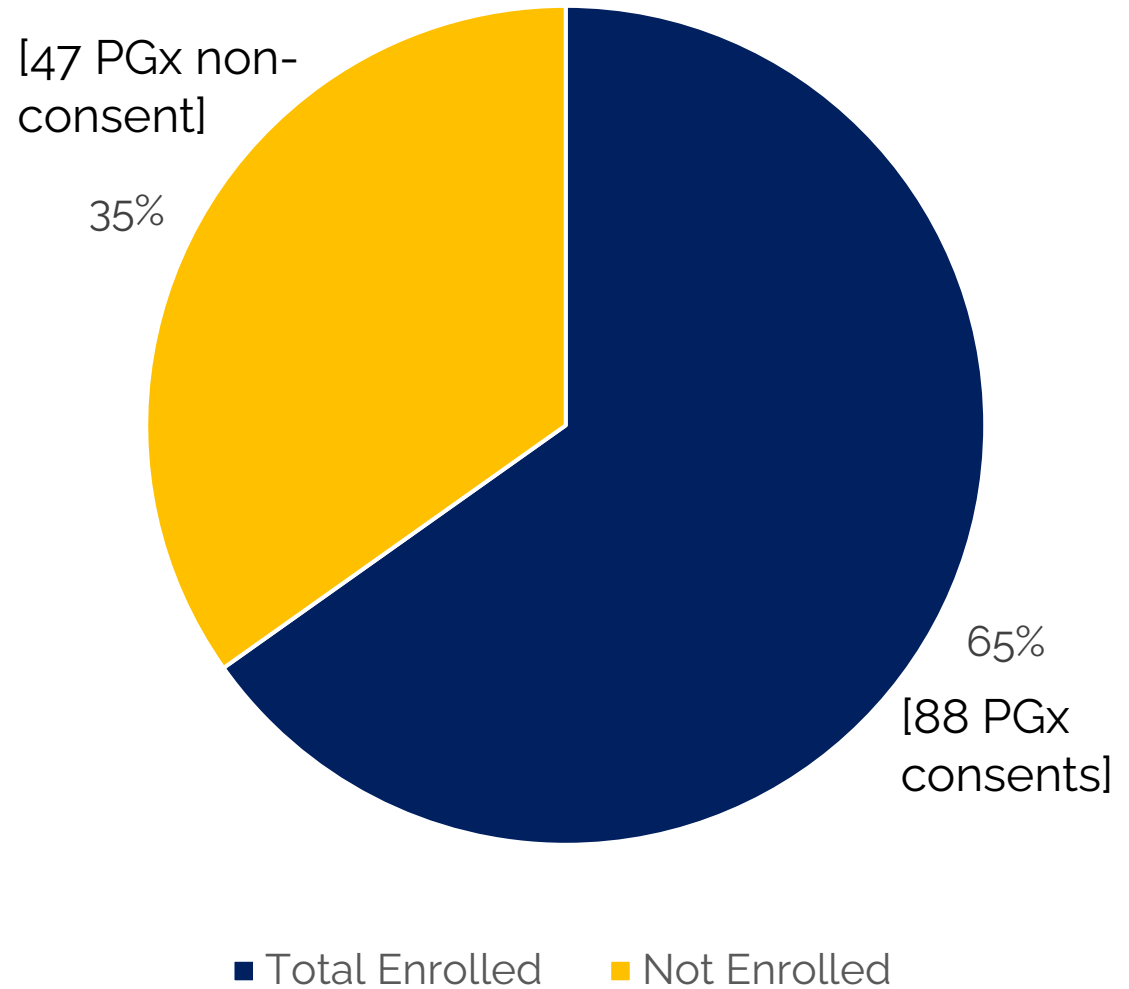
Under 18 with  
parental  
consent

U.S. Citizen

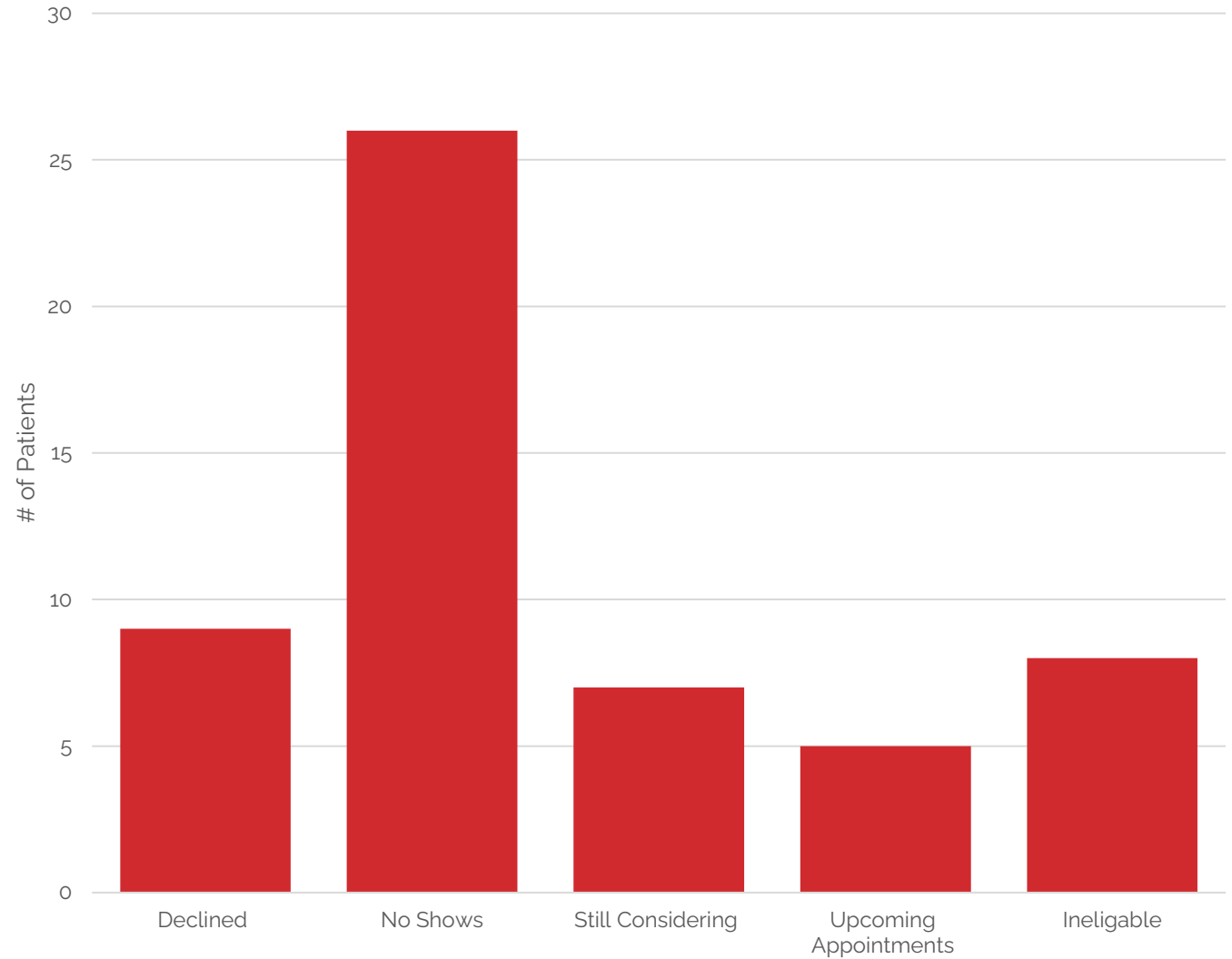


# PGx Test Consents: Impact of Specialty Pharmacy Identification

- Total eligible to be approached: 135 ID and Heme SP patients.

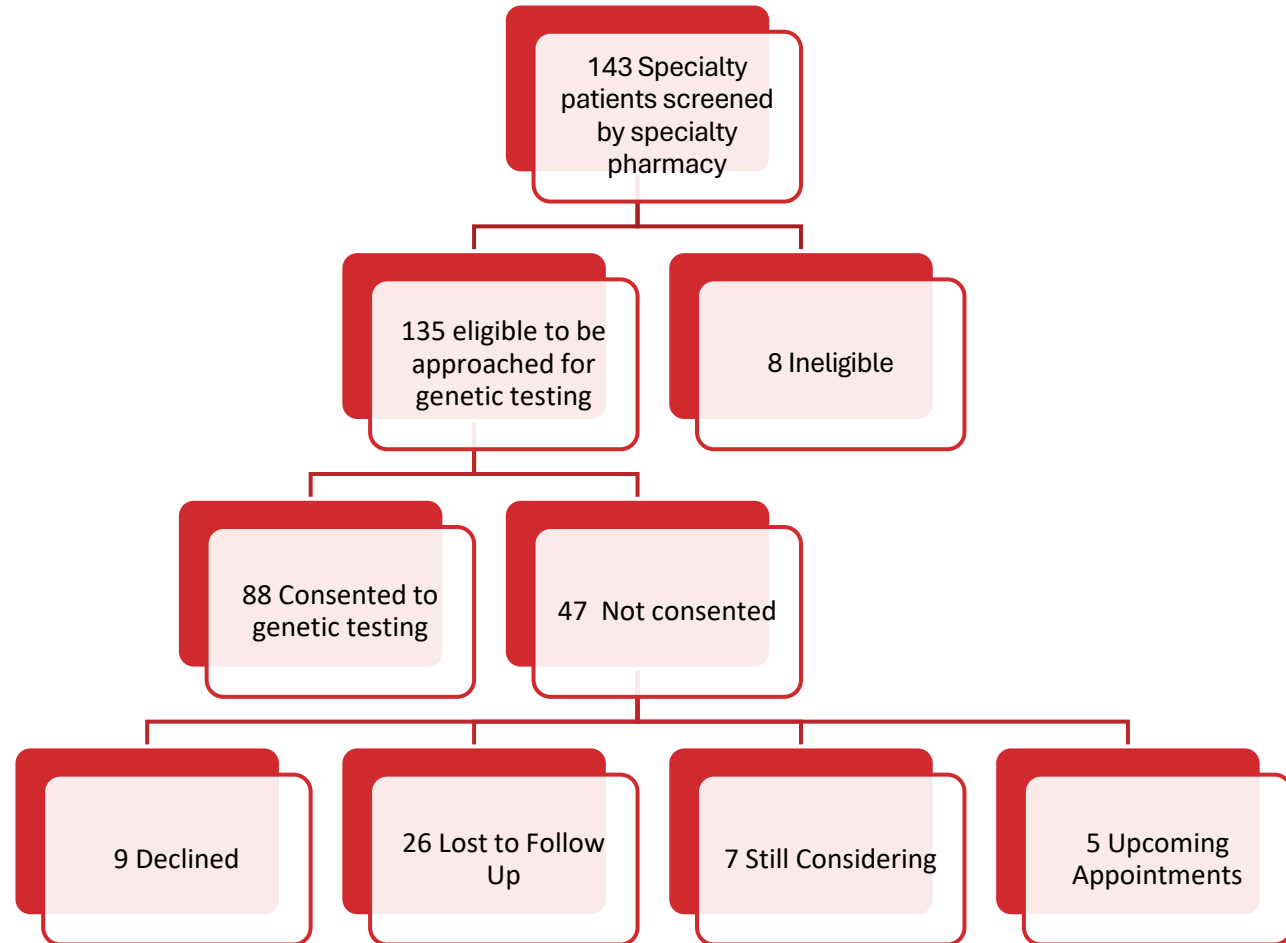


# Analysis of Specialty Pharmacy Patients Who Have Not Consented To PGx Testing



# PGx Test Consents: Impact of Specialty Pharmacy Identification

- PGx Enrollment Review (October 2024 - April 2025)
- 135 original specialty patients assessed beginning in October. New patients continue to be added.
- Total Specialty Population around 160 . Roughly only 15% had PGx testing prior to specialty team screening



# Patient Case

RH is a 17 yo M with PMH significant for HgbSS disease, asthma, enuresis, bicuspid aortic valve, aortic root dilation, restless legs, silent cerebral infarcts, Vitamin D deficiency, chronic headache, and chronic pain

RH is enrolled in Specialty Pharmacy: Active on deferasirox (Jadenu) & hydroxyurea (Siklos).

RH and caregiver consented to pharmacogenomics

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# Patient Case

Full medication list: albuterol HFA, amitriptyline, Symbicort, cyclobenzaprine, cyproheptadine, famotidine, Flonase, gabapentin, ibuprofen, oxycodone, propranolol, Sudafed, Senokot, Ubrelyv, Vit D3 gummies

You are the Hematology Specialty Pharmacist and will see RH in clinic today for his 6-month reassessment

Let's review his recently resulted PGx labs together.



# EPIC and Compass Rose Utilization

The screenshot displays the 'Rx SnapShot' interface in EPIC. The top navigation bar includes 'Patient', 'Med Profile', 'Rx Management', 'Allergies/Contraindications', 'SnapShot', 'Chart Review', and 'Sale History'. Below this, the 'Rx SnapShot' section is active, showing a list of genomic indicators under the heading 'Genomic Indicators'. The list includes various drug-related markers such as CACNA1S, CYP2B6, CYP2C19, CYP2C9, CYP2D6, CYP3A5, DPYD, G6PD, MT-RNR1, RYR1, SLCO1B1, TPMT, and UGT1A1, each with a 'Results Report' link and a dropdown arrow. The interface also features 'Expand All' and 'Collapse All' options.

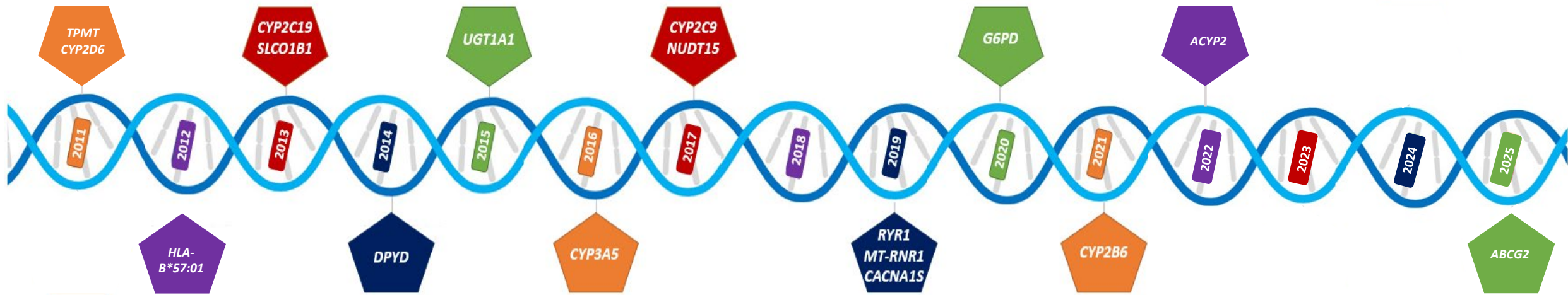
Drug	Expand All	Collapse All
CACNA15 Malignant Hyperthermia Variant Negative <a href="#">Results Report</a>	⌵	⌵
CYP2B6 Intermediate Metabolizer <a href="#">Results Report</a>	⌵	⌵
CYP2C19 Rapid Metabolizer <a href="#">Results Report</a>	⌵	⌵
CYP2C9 Normal Metabolizer <a href="#">Results Report</a>	⌵	⌵
CYP2D6 Normal Metabolizer <a href="#">Results Report</a>	⌵	⌵
CYP3A5 Intermediate Metabolizer <a href="#">Results Report</a>	⌵	⌵
DPYD Normal Metabolizer <a href="#">Results Report</a>	⌵	⌵
G6PD Normal <a href="#">Results Report</a>	⌵	⌵
MT-RNR1 Normal Risk of Aminoglycoside-Induced Hearing Loss <a href="#">Results Report</a>	⌵	⌵
RYR1 Malignant Hyperthermia Variant Negative <a href="#">Results Report</a>	⌵	⌵
SLCO1B1 Increased Function <a href="#">Results Report</a>	⌵	⌵
TPMT Intermediate Metabolizer <a href="#">Results Report</a>	⌵	⌵
UGT1A1 Intermediate Metabolizer <a href="#">Results Report</a>	⌵	⌵

## PGx Workflow Overview:

- Rx SnapShot: Identify patient's current medications
- Genomic Indicators: Review relevant pharmacogenomic markers
- Results Report: Analyze PGx test results and clinical implications
- Interventions Recorded via I-Vents: Document pharmacist actions and recommendations



# 17 Gene Test Results Returned in EHR; Used Daily to Tailor Pharmacotherapy of 76 Medications



EHR: Electronic Health Record

Permission to use by Haidar CE.

# EPIC and Compass Rose Utilization

## CYP2B6 Intermediate Metabolizer Results Report

Updated 3/22/2025 by Genomics, Background

### CYP2B6 Intermediate Metabolizer

#### Reference Links

[CYP2B6 guideline \(CPIC\)](#) | [CYP2B6 implemented gene \(St. Jude\)](#) |  
[St. Jude Pharmacogenomic Prescribing Recommendations](#) |

## CYP2C19 Rapid Metabolizer Results Report

Updated 3/22/2025 by Genomics, Background

### CYP2C19 Rapid Metabolizer

#### Reference Links

[CYP2C19 guidelines \(CPIC\)](#) | [CYP2C19 implemented gene \(St. Jude\)](#) |  
[St. Jude Pharmacogenomic Prescribing Recommendations](#) |  
<https://pharmsci.stjude.org/PGENΔKids/Alert/Formularv> |

## PGx Workflow Overview:

- Rx SnapShot: Identify patient's current medications
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- Results Report: Analyze PGx test results and clinical implications
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# EPIC and Compass Rose Utilization

Rx SnapShot ⚠ This patient has outside data that is only available in Hyperspace.

← Rx SnapShot Today's Visit Roadmap (Tx Plan 1) Roadmap (Tx Plan 2) Roadmap (Tx Plan 3) Variant Results Report Pharmacogenomics Results Report Specialty Pharmacy Overview More Rx SnapShot

## Results ⚠ CYP2C19 PG4KDS GENOTYPE (Order 14788455)

**⚠ CYP2C19 PG4KDS GENOTYPE: 25K-055RP005** Order: 14788455 - Reflex for Order 14487542

Collected 2/24/2025 5:13 PM Status: Final result Dx: Hemoglobin SS disease without crisis ...  
Test Result Released: Yes (not seen)

0 Result Notes

**Component**  
Ref Range & Units (hover)  
CYP2C19 PG4KDS Consult  
\*1/\*17

\*\*\*PHARMACOGENOMICS CONSULT FOR\*\*\*  
\*CYP2C19 GENOTYPE\*

Sample for CYP2C19 Genotype Obtained: 20250227152331  
PG4KDS CYP2C19 Genotype Result: \*1/\*17  
CYP2C19 Phenotype Assignment: CYP2C19 Rapid Metabolizer

This result signifies that the patient has one copy of a normal function allele (\*1) and one copy of an increased function allele (\*17). Based on the genotype result, this patient is predicted to be a rapid metabolizer of CYP2C19 substrates. This means that the patient may be at a high risk for an adverse or poor response to medications that are metabolized by CYP2C19 (such as amitriptyline). To avoid an untoward drug response, dose adjustments or alternative therapeutic agents may be necessary for medications metabolized by CYP2C19. For more information about specific medications metabolized by CYP2C19, please go to [www.stjude.org/pg4kds](http://www.stjude.org/pg4kds).

Understanding the PGx Results Report  
→ Red Exclamation Point = Abnormal Result Identified  
→ Triggers Pharmacogenomics Consult



# EPIC and Compass Rose Utilization

Search for problem  [+ Add](#) [DxReference](#) Show:  Past Problems

Diagnosis	Notes	Priority	Change Dx	Resolved	U
Chronic headache	<a href="#">Edit Overview</a>	Medium	▲	✕	
Vaso-occlusive pain due to sickle cell disease (HCC)	<a href="#">Create Overview</a>	Medium	▲	✕	
Respiratory					
Moderate persistent asthma	<a href="#">Create Overview</a>	Medium	▲	✕	
Circulatory					
Bicuspid aortic valve	<a href="#">Create Overview</a>	Medium	▲	✕	
Genitourinary					
Enuresis	<a href="#">Create Overview</a>	Medium	▲	✕	
Priapism due to sickle cell disease	<a href="#">Edit Overview</a>	Medium	▲	✕	
Musculoskeletal					
Unspecified fracture of left toe(s), initial encounter for closed fracture	<a href="#">Edit Overview</a>	Medium	▲	✕	
Hematologic					
Sickle cell disease, type SS (HCC)	<a href="#">Edit Overview</a>	Medium	▲	✕	
Hemoglobin SS disease with crisis and other complication (HCC)	<a href="#">Create Overview</a>	Medium	▲	✕	
Other					
CYP3A5 intermediate metabolizer (HCC)	<a href="#">Create Overview</a>	Unprioritized	▲	✕	
CYP2C19 rapid metabolizer (HCC)	<a href="#">Create Overview</a>	Unprioritized	▲	✕	
CYP2B6 intermediate metabolizer (HCC)	<a href="#">Create Overview</a>	Unprioritized	▲	✕	
UGT1A1 intermediate metabolizer (HCC)	<a href="#">Create Overview</a>	Unprioritized	▲	✕	
TPMT intermediate metabolizer (HCC)	<a href="#">Create Overview</a>	Unprioritized	▲	✕	

[Mark as Reviewed](#) Last Reviewed by Darla Jane Pickett, PA on 4/1/2025 at 1:20 PM

**Understanding the PGx**  
**→ Include Result in Problem list for quick access by all providers**



# Mapping CYP Enzyme to Drug Response Using Power BI

Click here to clear all filters

## Step 1: Select Gene

- Select all
- ABCG2
- ACYP2
- CACNA1S
- CYP2B6
- CYP2C19
- CYP2C9
- CYP2D6
- CYP3A5
- DPYD
- G6PD
- HLA-B\*57:01
- MT-RNR1
- NUDT15
- RYR1
- SLCO1B1
- TPMT
- UGT1A1

## Step 2: Select Phenotype

- Select all
- CYP2C19 Intermediate Metabolizer
- CYP2C19 Poor Metabolizer
- CYP2C19 Rapid Metabolizer
- CYP2C19 Ultra-rapid Metabolizer

Gene Name	Problem List Entry/Phenotype	High-Risk Medication
CYP2C19	CYP2C19 Rapid Metabolizer	Amitriptyline
CYP2C19	CYP2C19 Rapid Metabolizer	Clomipramine
CYP2C19	CYP2C19 Rapid Metabolizer	Doxepin
CYP2C19	CYP2C19 Rapid Metabolizer	Imipramine
CYP2C19	CYP2C19 Rapid Metabolizer	Lansoprazole
CYP2C19	CYP2C19 Rapid Metabolizer	Omeprazole
CYP2C19	CYP2C19 Rapid Metabolizer	Pantoprazole
CYP2C19	CYP2C19 Rapid Metabolizer	Trimipramine



# Patient Case: W AMB Custom Decision Support (CDS)

Patient Med Profile Rx Management Allergies/Contraindications SnapShot Chart Review Sale History

New Rx:

Profile only  Transfer  Advanced view

Scan Rx Use Last Scan  Schedule Fill: 7/7/2025

Provider Prescribed Pharmacy to Dispense

References: [Lexi-Comp](#) [Dosing for CYP2D6 genotype](#)

- ⓘ Prior prescription: [amitriptyline \(Elavil\) 25 MG tablet](#)
- ⓘ Associated diagnoses: *Chronic nonintractable headache, unspecified headache type*
- ⓘ The prescription was changed from how it was originally entered.

Written: 7/1/2025 Expires: 7/1/2026 Serial #:

Medication: Prescription Validation for Hendrix, Rashawn Anthony

Dose: **amitriptyline 25 MG tablet (Elavil)**  
Route: Pharmacogenomic Alert - CYP2D6 Normal Metabolizer - CYP2C19 Rapid Metabolizer / Amitriptyline  
Frequency: Based on the genotype result, this patient is predicted to be a CYP2D6 normal metabolizer and a CYP2C19 rapid metabolizer. If amitriptyline is prescribed to this patient, decreased effectiveness is likely. Please consult a clinical pharmacist for assistance with selecting an alternative agent, alternative dosing, or therapeutic drug monitoring. Review the Pharmacogenomics Results Report, or click Amitriptyline Pharmacogenomics Formulary Page CYP2D6 implemented gene (St. Jude) or CYP2C19 implemented gene (St. Jude) for more information.

End after:

Dispense:

Authorized:

Supervised:

Comments:

Add flags:

Do you want to accept these prescriptions anyway?

amitriptyline 25 MG tablet (Elavil) 60 tablet (20 days) **First fill**  Dispense at: St. Jude Outpatient Pharmacy  
Pending Fill \$?.?? Rx: 20372403 Ready: 7/7/2025 08:30



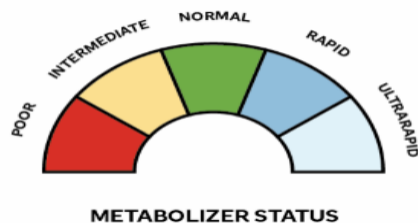
# Patient Example: Patient Friendly Language

## Notes Report



### Introduction of Pharmacogenomics

Pharmacogenomic test results help your medical team know the best type of medicine or the correct dose of a drug for you. We do these tests to make some medications safer and more effective for you. Pharmacogenomic test results last a lifetime because genes do not change. Your doctors and pharmacists can always use the results. This can help your medical team pick the best medicines for you in the future. Save any pharmacogenomic test results you get. Tell your other doctors and pharmacists that pharmacogenomic testing has been performed and let them know the results. Pharmacogenomic test results will also be stored in your child's medical record.



When you take a medicine, your body needs a way to handle it. One way your body does this is by using enzymes to break down (metabolize) the medicine. If you break down a medicine too fast or too slow, the medicine may not work as well, or you may have more side effects. In some instances, a small number of people will have unknown function, which we refer to as uncertain or indeterminate.

### Part I: Result Summary

Gene	Genotype	Phenotype
CYP2B6	*1/*6	<b>CYP2B6 Intermediate Metabolizer</b>
CYP2C19	*1/*17	<b>CYP2C19 Rapid Metabolizer</b>
CYP2C9	*1/*1	<b>CYP2C9 Normal Metabolizer</b>
CYP2D6	(*2/*17)2N	<b>CYP2D6 Normal Metabolizer</b>
CYP3A5	*1/*3	<b>CYP3A5 Intermediate Metabolizer</b>
DPYD	c./c.=	<b>DPYD Normal Metabolizer</b>
G6PD	B/Null	<b>G6PD Normal</b>
MT-RNR1	WT	<b>MT-RNR1 Normal Risk of Aminoglycoside-Induced Hearing Loss</b>






# Patient Example: Patient Friendly Language

[Patient](#) | [Med Profile](#) | [Rx Management](#) | [Allergies/Contraindications](#) | [SnapShot](#) | [Chart Review](#) | [Sale History](#) | [Notes Report](#)

## Notes Report

← ↻ 🧑 🖨 📄 🖼 🔗

Part II: Result Details

Gene	Genotype	Phenotype
CYP2B6	*1/*6	<b>CYP2B6 Intermediate Metabolizer</b> 
<p>CYP2B6 is an enzyme that is responsible for breaking down certain medicines. Differences in your DNA determine how well the CYP2B6 enzyme will break down certain medicines. People who are CYP2B6 intermediate metabolizers have decreased amounts of CYP2B6 enzyme. They may need to have the dose of some medicines adjusted to prevent side effects (such as efavirenz). There may be other medicines affected by CYP2B6. For more information, review the <a href="#">CYP2B6 and Medicines</a> education page.</p>		
Gene	Genotype	Phenotype
CYP2C19	*1/*17	<b>CYP2C19 Rapid Metabolizer</b> 
<p>CYP2C19 is an enzyme that is responsible for breaking down or activating certain medicines. Differences in your DNA determine how well the CYP2C19 enzyme will break down or activate certain medicines. People who are CYP2C19 rapid metabolizers have slightly increased amounts of CYP2C19 enzyme. They may need to have the dose of some medicines increased to achieve the desired effect of the medicine (such as omeprazole and lansoprazole). There are other medicines affected by CYP2C19. For more information, review the <a href="#">CYP2C19 and Medicines</a> education page.</p>		
Gene	Genotype	Phenotype
CYP2C9	*1/*1	<b>CYP2C9 Normal Metabolizer</b> 

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# Pharmacogenomics Patient Education



Conditions ▾

Treatments, Tests, and Procedures ▾

Medical Care ▾

Emotional Support and Daily Life ▾

## Pharmacogenomics (Pharmacogenetics)

[Home](#) > [Treatments, Tests, and Procedures](#) > [Tests](#) > Pharmacogenomics (Pharmacogenetics)

### What is pharmacogenomics?

Pharmacogenomics is the study of how a person's [genes](#) <sup>Ⓞ</sup> affect the way they respond to a medicine. This helps doctors know the best type of medicine or the correct dose of a drug to use for a person. Pharmacogenomics is also called **pharmacogenetics**.

For example, knowing about your child's genes can tell the doctor:

- Which medicine is likely to work best for your child's illness
- The best dose of a medicine for your child

### Key Points

- Pharmacogenomics, also called pharmacogenetics, is the study of how a person's genes affect the way they respond to a medicine.
- Pharmacogenomic testing helps doctors know the best type or dose of medicine to use for a person.
- Your child may have a blood test to identify genes that affect medications.
- Pharmacogenomic test results last a lifetime. They may be used now or in the future to pick the best medicines for your child.
- Share the results of pharmacogenomic tests with your other doctors and pharmacists.





# Underutilization of Pharmacogenomics in Specialty Pharmacy and Next Steps

# Steps To Increase Utilization

Does your specialty pharmacy have access to EHR?

If YES, incorporate custom decision support (CDS) to link pharmacogenomics labs to the specialty or non-specialty medication of interest (Slides 31-32)

If NO, standardize the technician or pharmacist workflow to include documentation of genomics OR

If NO, request part of referral documentation from provider if known.

Specialty Pharmacy asks about allergies.....why would we not ask for genomics information??

# Steps To Increase Utilization

Include documentation into the technician or pharmacist note templates (Slide 21)

Create patient and family literature that helps explain the value of PGx and how it can impact treatment outcomes (Slides 33-34)

Create a tool to support pharmacist education and learning for genomics that links phenotype with medications of interest. (Slides 29-30)



# Next Steps

Expand the process to Non-specialty hematology patients and Oncology Specialty Patients at St. Jude

Encourage both health system based and non health system based specialty pharmacies to incorporate genomics into conversations with all patients (for specialty and non-specialty medication evaluations)

Support greater access to genomics opportunities for patients

Use the data and literature for value-based care agreements with PBMs





# Barriers & Limitations

# Barriers to PGx Implementation within Specialty Pharmacy

- **Cost**
  - Approx. **\$500** for panel tests
  - **Insurance reimbursement** varies by payer. effective 7/13/2025, pharmacogenomic testing is now accessible to ALL Medicare beneficiaries
- **Infrastructure**
  - Often **outsourced testing** with longer turnaround times
  - Requires **specialized equipment** and blood collection capabilities
- **Knowledge Gaps**
  - Limited PGx education in pharmacy and medical training
  - Focus often limited to **CYP enzymes**
  - PGx concepts not consistently integrated into curricula



# Barriers to PGx Implementation within Specialty Pharmacy

- **Privacy Concerns**
  - Risk of unauthorized access or data breaches
  - Fears of genetic discrimination by insurers or employers
- **Provider Acceptance & Confidence**
  - Clinicians may be **uncertain how to interpret** or act on PGx data
  - Limited **experience or confidence** in using PGx for decision-making
  - Concerns about **liability** or making changes to standard treatment
- **Incomplete Access to Medication History**
  - Fragmented prescribing across providers limits visibility into potential gene-drug interactions
  - Poor integration of PGx data with med records impairs decision-making and continuity of care



# Take Home Points



Significance of Genetic Testing



Impact of the Specialty Pharmacy



Implementation into other Pharmacies

# References

- 1. Bigler R. Pharmacogenomics for precision specialty pharmacy care. *Pharmacy Today*. 2019;11(6).
- 2. Crews KR, Gaedigk A, Dunnenberger HM, et al. Clinical Pharmacogenetics Implementation Consortium (CPIC) guidelines for cytochrome P450 2D6 genotype and codeine therapy: 2011 update. *Am J Health Syst Pharm*. 2011;68(16):143-150.
- 3. Gammal RS, Cavallari LH, Lam Y. Clinical Pharmacogenomics. In: DiPiro JT, Yee GC, Haines ST, Nolin TD, Ellingrod VL, Posey L, eds. *DiPiro's Pharmacotherapy: A Pathophysiologic Approach*. 12th ed. McGraw Hill; 2023. Accessed May 14, 2025. <https://accesspharmacy-mhmedical-com.ezproxy.uthsc.edu/content.aspx?bookid=3097&sectionid=267224711>
- 4. Hoffman JM, Haidar CE, Wilkinson MR, et al. PG4KDS: a model for the clinical implementation of pre-emptive pharmacogenetics. *Am J Med Genet C Semin Med Genet*. 2014;166C(1):45-55. doi:10.1002/ajmg.c.31391
- 5. Pharmacogenomics for precision specialty pharmacy care. *Pharmacy Times*. Published October 22, 2019. Accessed May 19, 2025. <https://www.pharmacytimes.com/view/pharmacogenomics-for-precision-specialty-pharmacy-care>
- 6. St. Jude Children's Research Hospital. PG4KDS: Clinical Implementation of Pharmacogenetics. Published June 2020. Accessed May 19, 2025. <https://www.stjude.org/care-treatment/clinical-trials/pg4kds-pharmacogenetics.html>



# References

- [Clinical Pharmacogenomics Guidelines – CPIC](#)
- [Pharmacogenomics Program - St. Jude](#)
- [Pharmacogenomics \(Pharmacogenetics\) - Together by St. Jude](#): patient educational resources about pharmacogenomics
- [DPWG: Dutch Pharmacogenetics Working Group](#)



# Thank You

Tiffany Nason PharmD [tiffany.nason@stjude.org]

Timothy Howze PharmD [timothy.howze@stjude.org]

<https://www.stjude.org/research/departments/pharmacy-pharmaceutical-sciences.html>

<https://www.stjude.org/care-treatment/treatment/clinical-initiatives/meds-for-me.html>

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# Appendix Slides

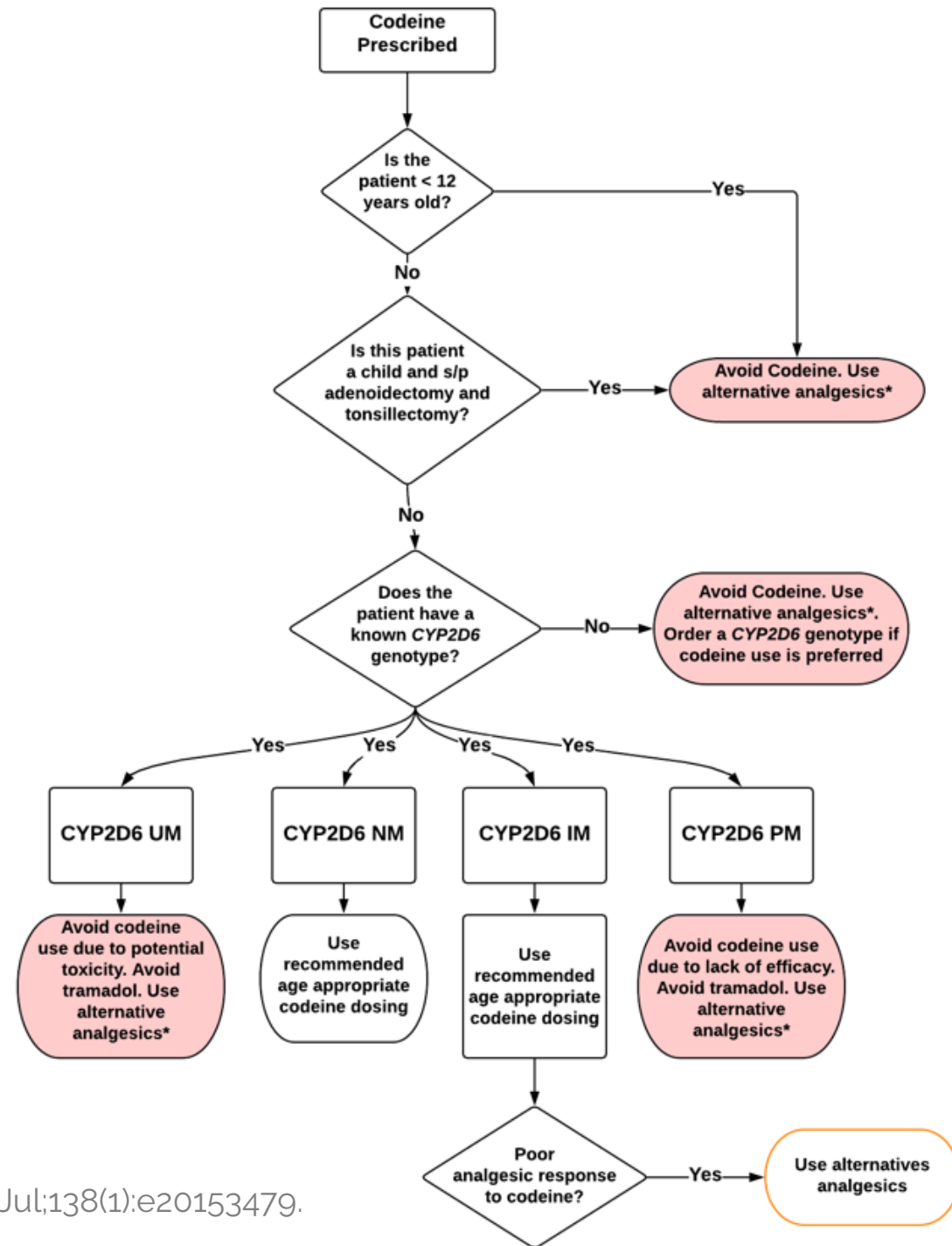
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Confidential



# Codeine Prescribing Algorithm

- Implemented in 2014
- Patients only receive codeine if they have a non-high-risk CYP2D6 phenotype
- Alternative analgesics are recommended otherwise





# No patient with a high-risk CYP2D6 phenotype newly prescribed codeine

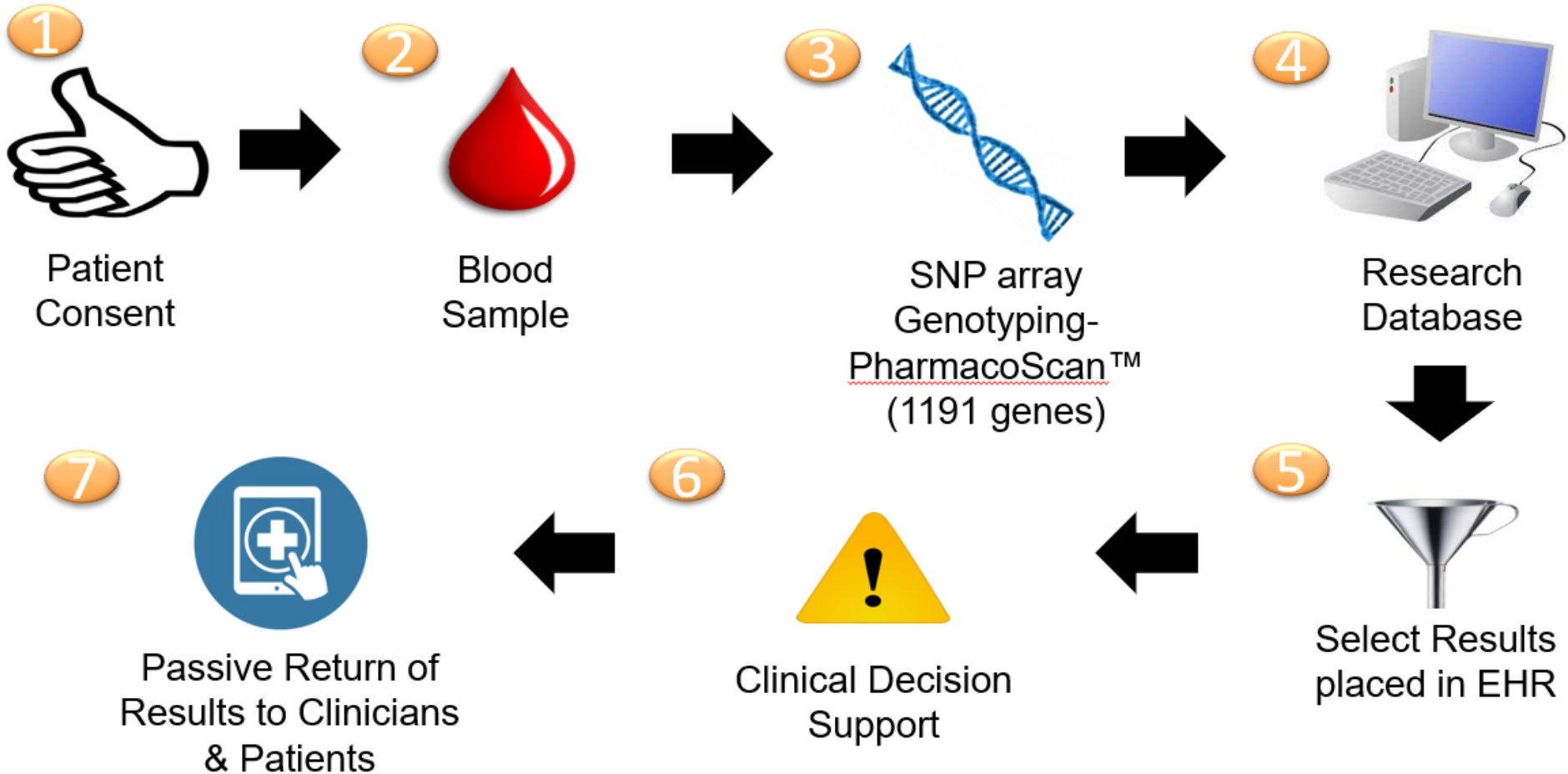
Codeine Prescribing Patterns Among Patients with Sickle Cell Disease and Known CYP2D6 Phenotypes

CYP2D6 phenotype	<i>n</i>	Patients for Whom a Codeine-Containing Analgesic Was Dispensed, <i>n</i> (%)
High-risk phenotypes		
Ultra-rapid metabolizer	19	0
Possible ultra-rapid metabolizer	25	1 <sup>a</sup> (4)
Poor metabolizer	9	0
Non-high-risk phenotypes		
Extensive metabolizer	488	161 (33)
Intermediate metabolizer	42	8 (19)
Possible intermediate metabolizer	13	4 (31)
Unknown risk		
Indeterminate	25	1 (4)

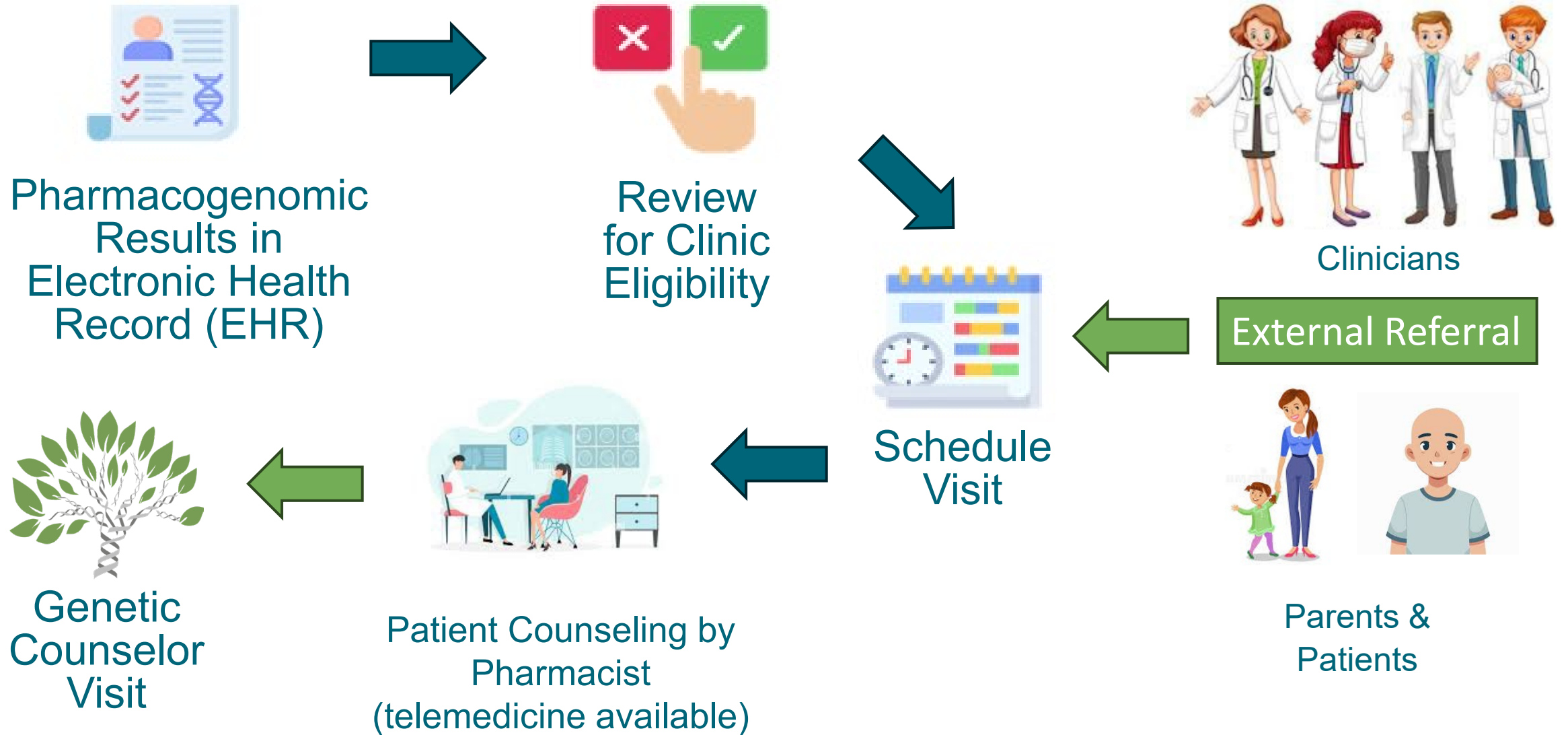
<sup>a</sup> Patient had a documented history of tolerating codeine well in the past.

# Pharmacogenomic Testing Process

No formal interaction with patients once results received



# Pharmacogenomics Clinic Workflow



# 17 Genes and 76 Drugs Implemented

- **HLA-B\*57:01 (1%)**
  - Abacavir-2012
- **CYP2D6 (17%)**
  - Codeine-2011
  - Tramadol-2012
  - Oxycodone-2013
  - Amitriptyline, nortriptyline-2014
  - Clomipramine, imipramine, trimipramine, doxepin-2016
  - Fluoxetine, paroxetine-2012
  - Ondansetron-2013
- **CYP2C19 (62%)**
  - Clopidogrel-2103
  - Amitriptyline-2014
  - Clomipramine, imipramine, trimipramine, doxepin-2016
  - Voriconazole-2015
  - Omeprazole, pantoprazole, lansoprazole-2018
  - Citalopram, escitalopram, sertraline-2022
- **CACNA1S/RYR1 (0.3%)**
  - Enflurane, methoxyflurane, desflurane, halothane, isoflurane, sevoflurane-2019
  - Succinylcholine-2019
- **CYP3A5 (41%)**
  - Tacrolimus-2015
- **ACYP2 (8%)**
  - Cisplatin-2022
- **ABCG2 (8%)**
  - Rosuvastatin-2025
- **CYP2B6 (5%)**
  - Methadone-2021
  - Efavirenz-2021
- **SLCO1B1 (13%)**
  - Simvastatin -2013
- **TPMT/NUDT15 (11%)**
  - Mercaptopurine, thioguanine-2011/2017
  - Azathioprine -2011/2017
- **DPYD (0.4%)**
  - Fluorouracil, capecitabine-2014
- **UGT1A1 (28%)**
  - Atazanavir-2015
- **CYP2C9 (32%)**
  - Celecoxib-2017
  - Meloxicam, ibuprofen-2020
- **mt-RNR1 (0.2%)**
  - Amikacin, gentamicin, tobramycin-2019
- **G6PD (2%)**
  - Dapsone, methylene blue, nitrofurantoin, phenazopyridine, primaquine, rasburicase-2020
  - Toluidine blue, pegloticase, tafenoquine-2022

**Presentation Title: Genomics in Action: Integrating Pharmacogenomics into Specialty Pharmacy Workflow**

**Presenter Name: Tiffany Nason and Timothy Howze**

**Learning Objectives:**

**At the completion of this activity, the participant will be able to:**

- Describe the factors that are used to define specialty medication
- Describe how a specialty program in collaboration with pharmacogenomic expertise can ensure patients receive the most effective therapies for complex, high risk conditions
- Discuss three specialty medications that have phenotypic implications and when to intervene if genomic information is available for patients

Test Question	Corresponding Learning objective	Corresponding text or slide number with answer	Rationale for correct/incorrect answers
<p>Which of the following factors is not typically used to define a specialty medication?</p> <ul style="list-style-type: none"> <li>A. Complex administration routes (oral, injection, infusion)</li> <li>B. Treatment of serious, catastrophic conditions</li> <li><b>C. Available as generic formulations</b></li> <li>D. Special handling, storage and cold chain requirements</li> <li>E. Special access conditions by the manufacturer.</li> </ul>	<p>Describe the factors that are used to define specialty medication.</p>	<p>Slide 6 &amp; 7</p>	<p>"Defining Specialty Medications" shows the key factors including complex conditions, administration routes, special handling, and access restrictions. Specialty medications are typically brand-name, high-cost drugs.</p>
<p>In the St. Jude specialty pharmacy PGx implementation (October 2024 - April 2025), what percentage of eligible patients consented to pharmacogenomic testing?</p> <ul style="list-style-type: none"> <li>A) 55%</li> <li><b>B) 65%</b></li> </ul>	<p>Describe how a specialty program in collaboration with pharmacogenomic expertise can ensure effective therapies</p>	<p>Slide 25</p>	<p>"PGx Test Consents: Impact of Specialty Pharmacy Identification" shows 88 consents out of 135 eligible patients (65%).</p>

<p>C) 75% D) 85% E) 95%</p>			
<p>Which of the following represents a key role of specialty pharmacy technicians in PGx implementation according to the model described? A) Interpreting PGx test results for clinical decision-making B) Providing genetic counseling to patients <b>C) Reviewing charts for availability of pharmacogenomic test results</b> D) Prescribing alternative medications based on PGx results E) Conducting patient education sessions about genomics</p>	<p>Describe how a specialty program in collaboration with pharmacogenomic expertise can ensure effective therapies</p>	<p>Slide 20</p>	<p>"Specialty Pharmacy's Role in PGx Enrollment and Clinical Use" specifically lists this as a technician responsibility.</p>
<p>According to the presentation, which of the following is a significant barrier to PGx implementation in specialty pharmacy? A) Lack of FDA-approved PGx tests <b>B) Limited PGx education in pharmacy and medical training</b> C) Inability to collect blood samples D) Lack of specialty medications with PGx associations E) Patient unwillingness to provide genetic information</p>	<p>Describe how a specialty program in collaboration with pharmacogenomic expertise can ensure effective therapies</p>	<p>Slide 46</p>	<p>"Barriers to PGx Implementation within Specialty Pharmacy" lists "Knowledge Gaps" including limited PGx education in pharmacy and medical training.</p>
<p>In the patient case example, RH is a 17-year-old male with sickle cell disease taking specialty medications deferasirox and hydroxyurea. His medication list</p>	<p>Discuss specialty medications that have phenotypic implications and when to intervene if genomic information is available</p>	<p>Slide 34 and 35</p>	<p>PowerBI tool or pharmacy resources can provide guidance on medication/phenotypic implications similar to the</p>

<p>also includes amitriptyline. Which pharmacogenomic marker would be MOST relevant for considerations of his amitriptyline therapy?</p> <p>A) TPMT <b>B) CYP2C19</b> C) UGT1A1 D) HLA-B*57:01 E) DPYD</p>			<p>Cyp2c19 example with amitriptyline</p>
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