

From Concept to Clinic: Establishing a Pharmacogenomics Clinic to Return Preemptive Test Results

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BACKGROUND

- Pharmacogenomic test results have life-long implications for medication therapy.
- Since 2011, nearly 8,000 patients have undergone preemptive pharmacogenomic testing at St. Jude Children's Research Hospital (St. Jude) through the **PG4KDS** clinical trial (www.stjude.org/pg4kds).
- In a survey conducted among **PG4KDS** participants and families in 2020, 75% of respondents reported that they had not shared their/their child's pharmacogenomic test results with their non-St. Jude providers. Additionally, 89% reported that they had not shared these results with their non-St. Jude pharmacists. The most common reasons for not sharing the results were because patients did not think of it or did not know they had to do so.
- Pharmacogenomic counseling may facilitate the patient's sharing of pharmacogenomic test results to other healthcare providers and promote gene-based prescribing by outside providers.
- A pharmacist-led pharmacogenomics clinic was established at St. Jude to offer individualized gene-based medication counseling and educational resources to patients and families.

PROCESS

- Patients at St. Jude undergo preemptive pharmacogenomic testing via single-gene tests or through the **PG4KDS** clinical trial.
- Results for up to 16 genes are returned and placed in the electronic health record (EHR).
- A pharmacogenomics clinic appointment is scheduled when pharmacogenomic test results are received or at the request of patients or their primary care team (Figure 1).
- The clinical pharmacogenomics team creates personalized summaries to educate patients and their families on the basic principles of pharmacogenomics, the patient's genotype and phenotype results, and provide recommendations for pharmacotherapy (Figure 2).
- Pharmacogenomic test results and the personalized education summaries are posted in the EHR, and viewable in the patient's electronic portal (Figures 2 and 4).
- Patients are seen several times throughout their St. Jude journey to reinforce education of results and the importance of communication to outside healthcare professionals (Figure 5).

RESULTS

Table 1: Clinic Visit Patient Demographics and Characteristics from 11/13/2023 – 1/31/2025 (n=295)

Age, years (median, range)	10 (0.4-24.7)	Pharmacist Time Per Clinic Encounter, minutes (median, range)	25 (10-60)
Gender (Patient Reported)		Number of High-Risk Phenotypes per patient (median, range)	3 (1-8)
Male	157 (53%)		
Female	138 (47%)		
Race		Reason for Visit Initiation	
Asian/Asian Indian	10 (3%)	Patient/Family Request	9 (3%)
Black or African American	88 (30%)	Prescriber Request	6 (2%)
Pacific Islander	1 (1%)	Scheduled by PGx Team	280 (95%)
White	186 (63%)		
Other	10 (3%)		
Primary Service		Patients Receiving Actionable Medications at Time of Counseling (Recommended Therapy Modification)	15
Hematology	15 (5%)	Amitriptyline (monitor for efficacy)	1
Infectious Diseases	15 (5%)	Irinotecan (monitor for neutropenia/diarrhea)	1
Leukemia/Lymphoma	76 (26%)	Mercaptopurine (30% dose reduction)	7
Neuro Oncology	78 (26%)	Omeprazole (dose modification)	1
Radiation Oncology	23 (8%)	Ondansetron (monitor for efficacy)	1
Solid Tumor	75 (26%)	Pantoprazole (dose modification)	2
Transplant and Cellular Therapy	13 (4%)	Voriconazole (monitor serum concentrations)	1
Visit Location		Warfarin (monitor serum concentrations)	
Inpatient	7 (2%)		
Outpatient Clinic	288 (98%)		

RESULTS

My Genetic Profile (Ryker)

This page shows what we know about your genes, including how they might affect the way you respond to certain medications and whether you are predisposed to certain conditions.

Disease-Related Findings

This section contains findings that inform how your genes affect your risk for certain diseases.

No disease-related findings of interest to show.

Medication-Related Findings

This section contains findings that inform how your genes affect your response to different medications.

Do not stop taking any medications without talking to your provider first.

CACNA1S Malignant Hyperthermia Variant Negative	→	CYP2B6 Normal Metabolizer	→
CYP2C19 Rapid Metabolizer	→	CYP2C9 Normal Metabolizer	→
CYP2D6 Normal Metabolizer	→	CYP3A5 Poor Metabolizer	→
DPYD Normal Metabolizer	→	GGPD Normal	→
MT-RNR1 Normal Risk of Aminoglycoside-Induced Hearing Loss	→	NUDT15 Normal Metabolizer	→
RYR1 Malignant Hyperthermia Variant Negative	→	SLCO1B1 Normal Function	→
TPMT Normal Metabolizer	→	UGT1A1 Intermediate Metabolizer	→

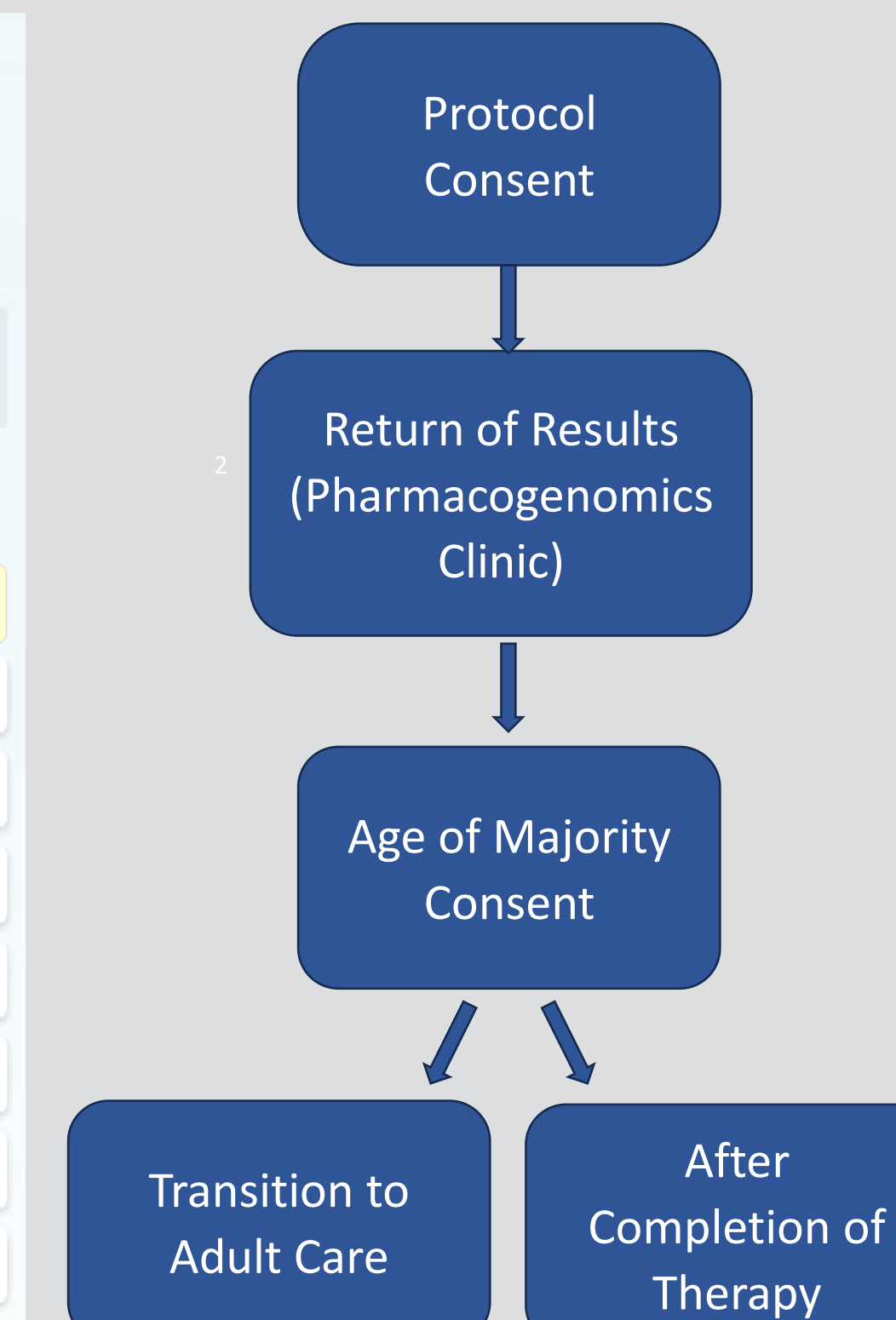


Figure 4: "My Genetics" Section Viewable in the Patient's Online Portal (MyChart)

Figure 5: Pharmacogenomics Points of Contact Throughout Therapy

CONCLUSIONS

- The return of pharmacogenomic test results can be performed within the scope of a pharmacist-led pharmacogenomics clinic.
- Patient education and counseling is an important component in the integration of pharmacogenomics into clinical practice.
- This return-of-results model for pharmacogenomics incorporates a variety of patient education modalities such as personalized education summaries viewable in the patient's online portal, and face-to-face teaching to promote understanding.

REFERENCES

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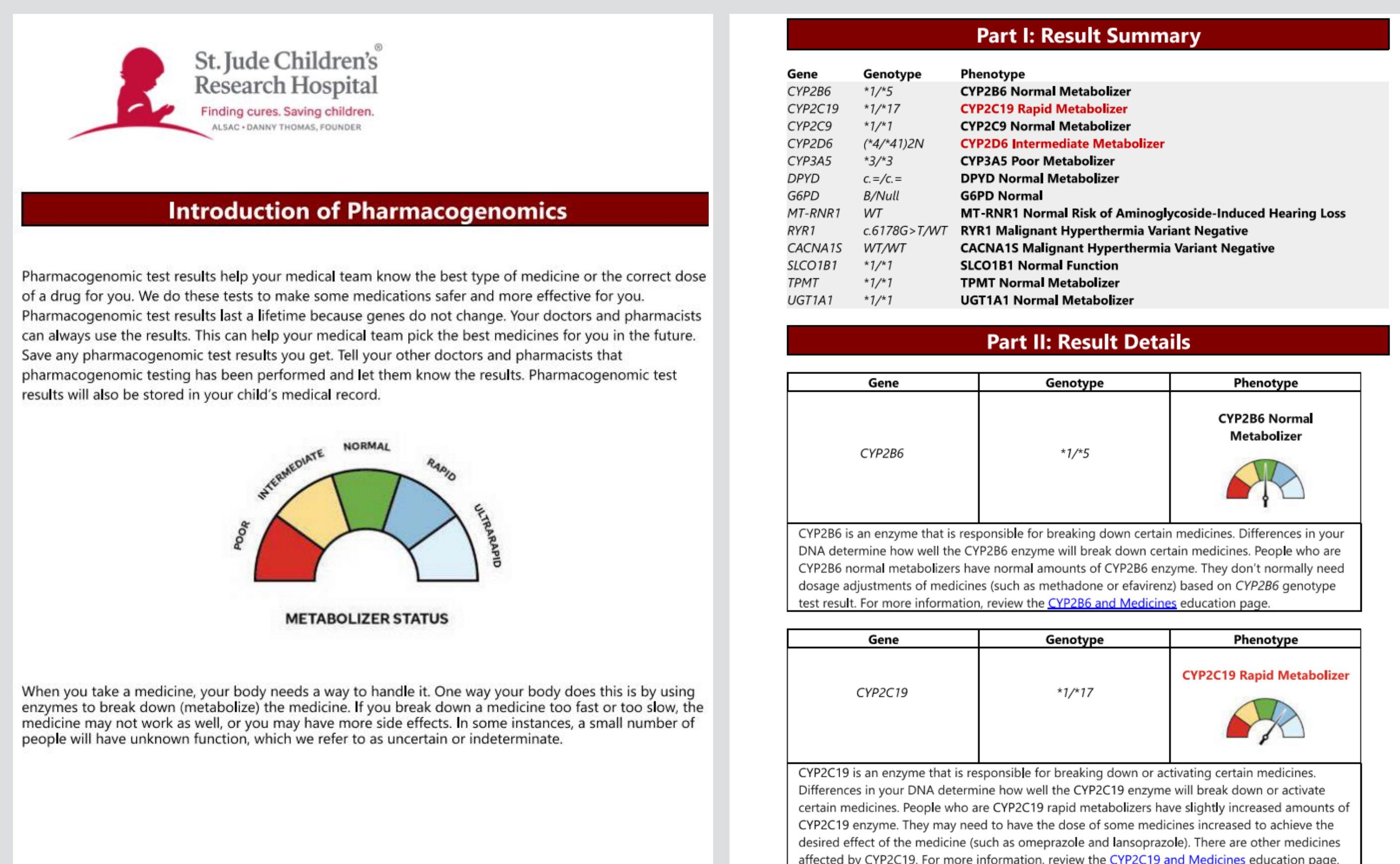
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MORE INFORMATION



SCAN ME

Part I: Result Summary

Gene	Genotype	Phenotype
CYP2B6	*1/*5	CYP2B6 Normal Metabolizer
CYP2C19	*1/*17	CYP2C19 Rapid Metabolizer
CYP2C9	*1/*1	CYP2C9 Normal Metabolizer
CYP2D6	*4/*13N	CYP2D6 Intermediate Metabolizer
CYP3A5	*3/*3	CYP3A5 Poor Metabolizer
DPYD	c.*/c. =	DPYD Normal Metabolizer
GGPD	B/Null	GGPD Normal
MT-RNR1	WT	MT-RNR1 Normal Risk of Aminoglycoside-Induced Hearing Loss
RYR1	c.6178G>TAT	RYR1 Malignant Hyperthermia Variant Negative
CACNA1S	WT/WT	CACNA1S Malignant Hyperthermia Variant Negative
SLCO1B1	*1/*1	SLCO1B1 Normal Function
TPMT	*1/*1	TPMT Normal Metabolizer
UGT1A1	*1/*1	UGT1A1 Normal Metabolizer

Part II: Result Details

Gene	Genotype	Phenotype
CYP2B6	*1/*5	CYP2B6 Normal Metabolizer
CYP2C19	*1/*17	CYP2C19 Rapid Metabolizer

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.

METABOLIZER STATUS

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.

Figure 2: Sample Pharmacogenomics Patient Education Summary

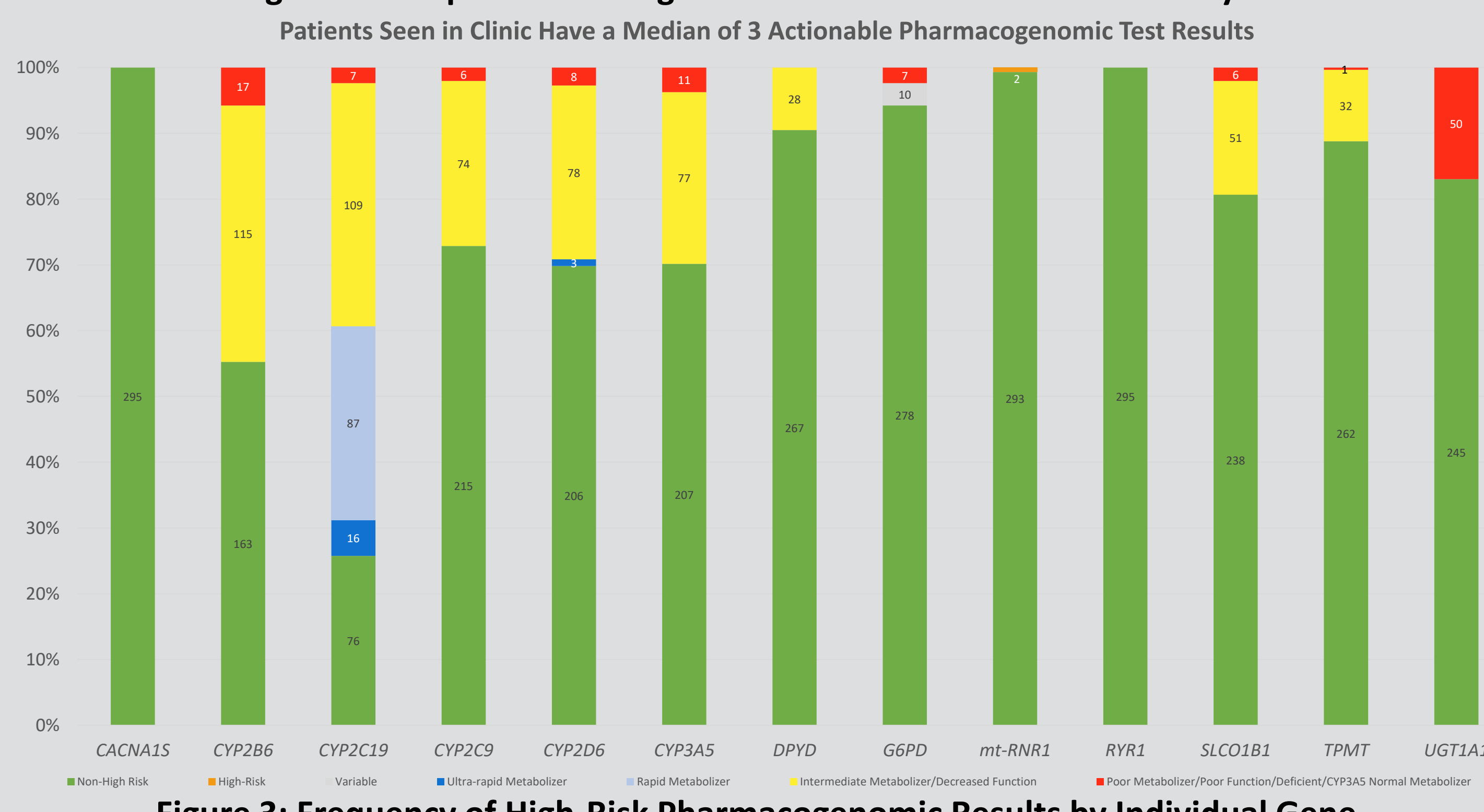


Figure 3: Frequency of High-Risk Pharmacogenomic Results by Individual Gene Returned in the Pharmacogenomics Clinic

Figure 1: Process for Returning Results in the Pharmacogenomics Clinic at St. Jude

