

Patient Name: Jane Doe Date of Birth: 01/01/1990

Sex Assigned at Birth: Female Specimen Type: WHOLE BLOOD

Patient ID: 0123456 Helix ID: Test12345

Collection Date: 2025-01-23

Report Date: 2025-04-29 Provider Name: Client Client

Order Date: 2025-04-29

Note: This report is intended for use by a medical professional. Please discuss any adjustments to your medication with your treating provider.

## **Drug Summary**

Antiemetics	Normal Interaction	Use with Caution	Consider Alternatives	Impact Unknown
Ondansetron (Zofran®)	<b>Ø</b>			
Tropisetron (Novaban®)	<b>Ø</b>			
Antifungals	Normal Interaction	Use with Caution	Consider Alternatives	Impact Unknown
Voriconazole (Vfend®)	<b>Ø</b>			
Antineoplastics	Normal Interaction	Use with Caution	Consider Alternatives	Impact Unknown
Gefitinib (Iressa®)	<b>Ø</b>			
Mercaptopurine (Purixan®)	•			
Tamoxifen (Soltamox®)			•	
Thioguanine (Tabloid®)	<b>Ø</b>			
- Fluoropyramidines	Normal Interaction	Use with Caution	Consider Alternatives	Impact Unknown
Capecitabine (Xeloda®)				?
Fluorouracil (Efudex®, Adrucil®)				?
Tegafur (Teysuno®)				?
mmunosuppressants	Normal Interaction	Use with Caution	Consider Alternatives	Impact Unknown
Azathioprine (Imuran®, Azasan®)	<b>Ø</b>			
Tacrolimus (Prograf®)	<b>Ø</b>			
Proton Pump Inhibitors	Normal Interaction	Use with Caution	Consider Alternatives	Impact Unknown
Dexlansoprazole (Dexilant®)		A		
Lansoprazole (Prevacid®)		<u> </u>		
Omeprazole (Prilosec®)		<u> </u>		
Pantoprazole (Protonix®)		<u> </u>		
Jric Acid Inhibitors	Normal Interaction	Use with Caution	Consider Alternatives	Impact Unknown
Allopurinol (Zyloprim®, Aloprim®)		A		

PFI#9396



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## Legend

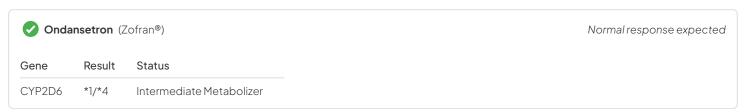
S	YMBOL	IMPLICATION
	<b>•</b>	Major gene-drug interaction, consider different drug
	lack	Major gene-drug interaction, consider reduced or increased dose
		No recommended action
	?	Impact not determined, consider standard dose and alter as needed

## **Gene Summary**

GENE	RESULT	STATUS
ABCG2, rs2231142	C/A	Decreased function
ATIC, rs4673993	T/T	Variant absent
CYP2C19	*1/*1	Normal Metabolizer
CYP2D6	*1/*4	Intermediate Metabolizer
CYP3A5	*3/*3	Poor Metabolizer
DPYD	N/A	Indeterminate
=5, c.1601G>A (p.Arg534Gln)	G/G	Variant absent
HLA-B*57:01	ABSENT	HLA-B: *57:01 negative
MTHFR, c.665C>T (p. Ala222Val)	T/T	Detected, homozygous
NUDT15	*]/*]	Normal Metabolizer
SLC19A1, rs1051266	G/G	rs1051266 G homozygote
ГРМТ	*1/*1	Normal Metabolizer

## **Drug Details**

## **Antiemetics**



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WHOLE BLOOD

Tropis	<b>etron</b> (No	vaban®)
Gene	Result	Status
CYP2D6	*1/*4	Intermediate Metabolizer
	-, -	

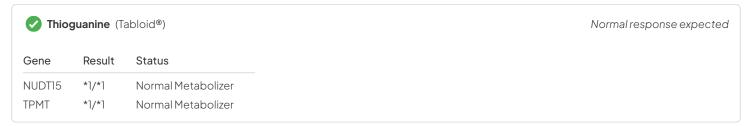
## **Antifungals**

### **Antineoplastics**

Gefitii	<b>nib</b> (Iressa	®)	
Gene	Result	Status	
CYP2D6	*1/*4	Intermediate Metabolizer	

Merc	aptopurin	e (Purixan®)
Gene	Result	Status
NUDT15	*1/*1	Normal Metabolizer
TPMT	*1/*1	Normal Metabolizer

• Tamox	<b>kifen</b> (Solt	amox®)
Gene	Result	Status
OCITO	resure	Status
CYP2D6	*1/*4	Intermediate Metabolizer



### Limited Evidence Drug-Gene Associations

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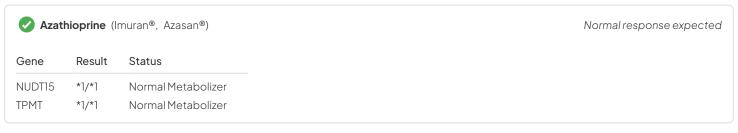
Sex Assigned at Birth: Female Provider Name: Client Client Collection Date: 2025-01-23

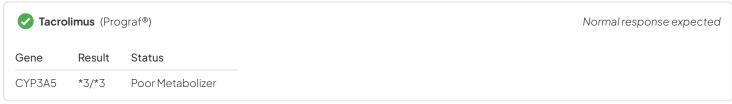
? Pazopanib	(Votrient®)	
ene	Result	Status
		HLA-B: *57:01 negative
TLA-D 37.01	ADSEINI	nla-b. 57.0Thegative

### Fluoropyramidines

? Cap	ecitabine	(Xeloda®)
Gene	Result	Status
DPYD	N/A	Indeterminate
? Fluc	prouracil	(Efudex®, Adrucil®)
Gene	Result	Status
DPYD	N/A	Indeterminate
? Teg	<b>afur</b> (Teys	NUDO®)

#### **Immunosuppressants**





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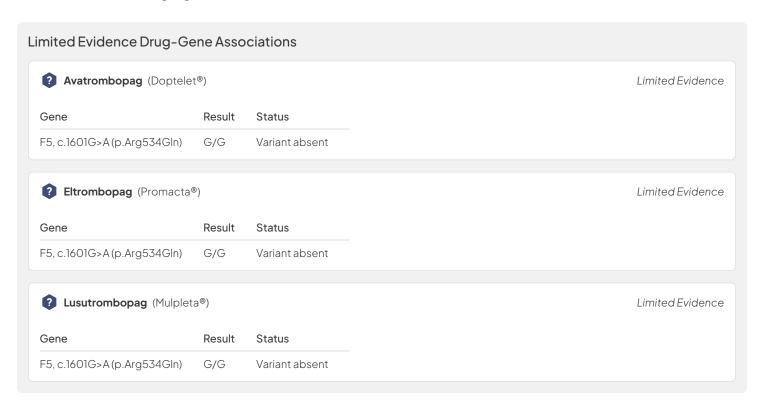
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Collection Date: 2025-01-23

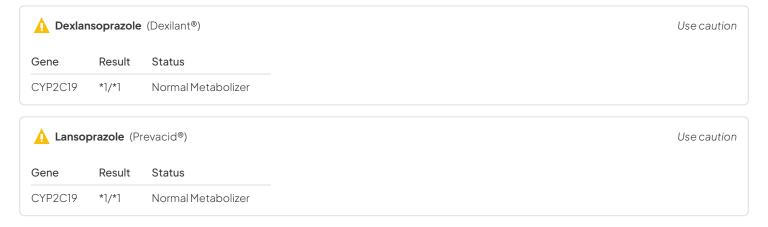
Order Date: 2025-04-29 Report Date: 2025-04-29

Limited Evidence Methotrexate (Rasuvo®, Otrexup®) Gene Result Status ATIC, rs4673993 T/T Variant absent MTHFR, c.665C>T (p. Ala222Val) T/T Detected, homozygous SLC19A1, rs1051266 rs1051266 G homozygote G/G

### Platelet-stimulating agents



#### **Proton Pump Inhibitors**





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⚠ Omep	<b>razole</b> (Pr	ilosec®)
Gene	Result	Status
CYP2C19	*]/*]	Normal Metabolizer
A Pantor	orazole (P	rotonix®)
Gene	Result	Status
CYP2C19	*]/*]	Normal Metabolizer

#### **Uric Acid Inhibitors**

Allopurinol (Zyle	oprim®, A	loprim®)
Gene	Result	Status
ABCG2, rs2231142	C/A	Decreased function

#### **Methods and Limitations**

Data were generated from extracted DNA using the validated Helix Exome+assay by the Helix clinical laboratory. The Exome+assay is based on target enrichment followed by next generation sequencing using paired end reads on an Illumina DNA sequencing system. Star alleles were determined using a proprietary algorithm which performs variant calling and then determines star allele solutions based on a combination of defining SNPs and exon-level copy number.

Metabolizer status was determined based on star allele solutions according to CPIC guidelines, with the following exceptions:

(1) metabolizer status was set as Indeterminate if a novel nonsense or truncating novel mutation was observed within the gene, (2) metabolizer status was set as Indeterminate if the combination of defining SNPs and copy number suggested a novel star allele solution, and (3) if more than two copies of a gene were detected then metabolizer status was set as Indeterminate. Drug/gene considerations were limited to guidelines published by FDA, CPIC, or PharmGKB.

Phasing could not be performed for genotypes, and therefore in some cases the star allele solution could not be disambiguated between two or more equally likely possibilities. In these cases, if the metabolizer status was the same regardless of possible star allele solutions, the more common star allele solution was provided along with the metabolizer status. If the metabolizer status was different for the equally-likely star allele solutions, the star alleles were reported as Unknown and the metabolizer status was considered Indeterminate.

All samples were sequenced and interpreted in Helix's CLIA-certified (#05D2117342) and CAP-accredited (#9382893) laboratory in San Diego, California. These tests have not been cleared or approved by the U.S. Food and Drug Administration.

The reportable range includes the following results: ABCG2: rs2231142; ATIC: rs4673993; CYP2C19: \*1-\*19, \*22-\*26, \*28-\*39; CYP2D6: \*1-\*15, \*4N, \*17-\*65, \*68-\*75, \*81, \*83-\*114; CYP3A5: \*1, \*3, \*6-\*9; DPYD: \*1, \*2A, \*3, \*4, \*6-\*8, \*9B, \*10-\*13, HapB3, rs150036960, rs72549310, rs80081766, rs150385342, rs141462178, rs200562975, rs2297595, rs139834141, rs6670886, rs115232898, rs72549308, rs72549307, rs45589337, rs146356975, rs150437414, rs145112791, rs201018345, rs183385770, rs143154602, rs72549305, rs75017182, rs140602333, rs143815742, rs61622928, rs200064537, rs764666241, rs142512579, rs186169810, rs72975710, rs144395748, rs57918000, rs199549923, rs72549304, rs111858276, rs138391898, rs148994843, rs190951787, rs142619737, rs201615754, rs59086055, rs138616379, rs145773863, rs147601618, rs17376848, rs3918289, rs55971861, rs138545885, rs137999090, rs145548112, rs146529561, rs60511679, rs112766203, rs56005131, rs199634007, rs200687447, rs60139309, rs201035051, rs55674432, rs147545709, rs67376798, rs141044036, rs145529148, rs72547602, rs72547601, rs202144771, rs139459586, rs140114515, rs148799944, rs114096998; F5: c.1601G>A (p.Arg534GIn); HLA-B\*57:01; MTHFR: c.665C>T (p. Ala222Val); NUDT15: \*1-\*20; TPMT: \*1, \*2, \*3A, \*3B, \*3C, \*4-\*44; SLC19A1: rs1051266; Sensitivity may be reduced for the CYP2D6\*13 allele.

Results are based on: Allopurinol, ABCG2 (PGKB1A); Azathioprine, NUDT15, TPMT (FDA Section 1, CPIC A); Capecitabine, DPYD (FDA Section 1, CPIC A); Dexlansoprazole, CYP2C19 (PGKB1A); Fluorouracil, DPYD (FDA Section 1, CPIC A); Gefitinib, CYP2D6 (FDA Section 1); Lansoprazole, CYP2C19 (PGKB 1A; CPIC A); Mercaptopurine, NUDT15, TPMT (FDA Section 1, CPIC A); Omeprazole, CYP2C19 (PGKB 1A; CPIC A); Ondansetron, CYP2D6 (CPIC A); Pantoprazole, CYP2C19 (FDA Section 1; CPIC A); Tacrolimus, CYP3A5 (FDA Section 1); Tamoxifen, CYP2D6 (CPIC A); Tegafur, DPYD (PGKB 1A); Thioguanine, NUDT15, TPMT (FDA Section 1, CPIC A); Tropisetron, CYP2D6 (CPIC A); Voriconazole, CYP2C19 (FDA Section 1, CPIC A).

San Diego, CA 92121

CAP #9382893 PFI #9396

Helix, Inc. 10170 Sorrento Valley Road, Suite 100 CLIA #05D2117342 Laboratory Director: Philip D Cotter, PhD, FACMG, FFSC (RCPA) NYS LD: Kenneth David Becker, PhD, HCLD, CQ, CGMBS



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#### Disclaimer

The interpretations and drug considerations provided by Helix are intended solely for use by a medical professional and do not constitute medical advice by Helix. All treatment decisions and diagnoses remain the full responsibility of the treating provider. Results included in this report are based on the guidelines published by the FDA and CPIC, and do not account for other factors that may impact drug response, such as environment, medical conditions, drug-drug interactions, or additional genetic variants. Helix is not responsible or liable for any errors, omissions, or ambiguities in the interpretation or use of the results of this report. Administration of any medication listed in this report requires careful therapeutic monitoring regardless of the drug considerations outlined in this report. All dates and times displayed are Pacific Time and may vary from the dates and times for Collection, Order and Report for the providers/patients.

#### **Result Notations**

https://www.fda.gov/medical-devices/precision-medicine/table-pharmacogenetic-associations https://cpicpgx.org/guidelines https://www.pharmgkb.org/guidelineAnnotations

### Report Signed By

Kenneth David Becker, PhD, HCLD, CQ, CGMBS

### Helix's Sequence Once, Query Often® Model

When your provider orders a genetic test through Helix, we use our proprietary Sequence Once, Query Often® model to perform whole exome sequencing and analyze the specific genes related to the test. Helix securely stores your whole exome for future clinical use. With your permission, this allows your health care providers to order future medically necessary genetic tests from Helix without needing another sample. Instead, these tests are conducted through digital analysis of your stored genetic information.

To learn more about how Helix protects the privacy and security of your genetic information and learn more about your rights, please visit https://www.helix.com/privacy-and-policy-highlights.

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