



Helix Pharmacogenomics (PGx) Fluoropyrimidines DPYD Test


Patient Name: Patient-PFPD1	Specimen Type: test	Provider Name: Provider-PFPD1	Report Date: 2025-06-23
Date of Birth: 1990-03-03	Patient ID: MRNPFPD1	Collection Date: 2022-06-23	
Sex Assigned at Birth: F	Helix ID: HelixIDPFPD1	Order Date: 2022-06-23	

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





Results & Interpretations

<div> Capecitabine (Xeloda®)</div>			Normal response expected
Gene	Result	Status	
DPYD	*1/*1	Normal Metabolizer	

<div> Fluorouracil (Efudex®, Adrucil®)</div>			Normal response expected
Gene	Result	Status	
DPYD	*1/*1	Normal Metabolizer	

<div> Tegafur (Tegsuno®)</div>			Normal response expected
Gene	Result	Status	
DPYD	*1/*1	Normal Metabolizer	

Legend

SYMBOL	IMPLICATION
	Major gene-drug interaction, consider different drug
	Major gene-drug interaction, consider reduced or increased dose
	No recommended action
	Impact not determined, consider standard dose and alter as needed



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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:

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.

Results are based on:

Capecitabine, DPYD (FDA Section 1, CPIC A); Fluorouracil, DPYD (FDA Section 1, CPIC A); Tegafur, DPYD (PGKB 1A).

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

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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>.