



PGx Panel

Twist Alliance Long Read PGx Panel



- ~99% of adults have an actionable PGx variant (US, UK Biobank studies)
- ~50% of US adults are prescribed a drug for which there are CPIC guidelines
- > 100K adverse drug reactions per year in the US costing >\$30B

Panel design

49 genes - 2 Mb target region
39 full-length genes enable phasing
Includes all 20 genes with CPIC guidelines

Pairs with the Mitochondrial Panel

CYP genes	HLA	Others	
CYP1A2*	<u>HLA-A</u>	ABCB1	HTR2C
CYP2B6*	<u>HLA-B</u>	ABCG2	<u>IFNL3</u>
CYP2C19	HLA-DQA1	ADD1	MTHFR
CYP2C8	HLA-DRB1	ADRA2A	NAGS
CYP2C9		ANKK1	NAT2
CYP2D6		APOL1	<u>NUDT15</u>
CYP3A4		BCHE	OPRD1
CYP3A5		<u>CACNA1S</u>	OPRK1
CYP4F2		<u>CFTR</u>	OPRM1
		COMT	POLG
		CTBP2P2	<u>RYR1</u>
		<u>DPYD</u>	SLC6A4
		DRD2	<u>SLCO1B1</u>
		F2	<u>TPMT</u>
		F5	<u>UGT1A1</u>
		<u>G6PD</u>	UGT2B15
		GBA	<u>VKORC1</u>
		GRIK4	YEATS4

***Bold** denotes full-gene coverage
+Underline denotes inclusion in a CPIC guideline

Ji Y et al. Preemptive Pharmacogenetic Testing: A Comprehensive Analysis of Five Actionable Pharmacogenomic Genes Using Next-Generation DNA Sequencing and a Customized CYP2D6 Genotyping Cascade. *J Mol Diagn* (2015).

Chanfreau-Coffinier C, et al. Projected Prevalence of Actionable Pharmacogenetic Variants and Level A Drugs Prescribed Among US Veterans Health Administration Pharmacy Users. *JAMA Netw Open* (2019).



Panel performance in 24-plex of 2 Mb panel

Example coverage for SLC6A4 in 24-plex

