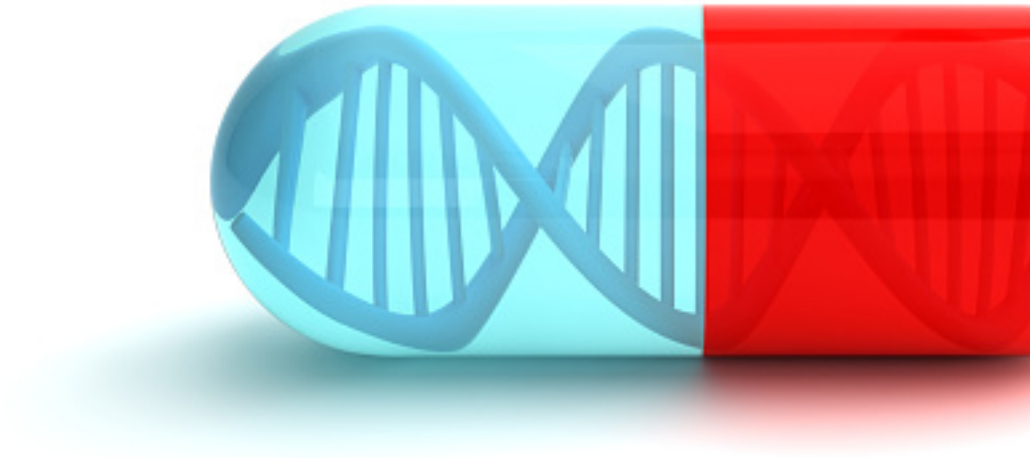
Pharmacogenetic Passport

Pharmacogenetic guidelines and clinical annotations connected to influential DNA changes

STE0097



Contents

0.1 Patient haplotypes

Gene	Phylogenetic method[1]	Set method[2]
VKORC1	H2 / H7	H2 / *2
CYPC191	*1 / *3	*1 / *18

[1]Method using phylogenetic trees to find closest 'relative' to patient alleles. Trees can be seen in Appendix A.

[2]Method looking for overlap of haplotype non-reference variants and patient non-reference variants. Reference variants are filtered out of both sets prior to comparison. Top 5 hits can be seen in Appendix 2.

0.2 Drug-gene connections

Drug	Gene	Guideline	Annotations[3]
Warfarin	VKORC1	Yes	*Level 1-2*: 15 *Level 3-4*: 13
Warfarin	CYPC191	Yes	*Level 1-2*: 15 *Level 3-4*: 13

[3]Level 1A and 1B clinical annotations meet the highest levels of criteria and are manually curated by PharmGKB. Level 1A annotations contain a variant-drug combination in a CPIC or medical society endorsed PGx guideline, or, implemented at a PGRN site, or, in another major health system. Level 1B annotations contain a variant-drug combination where the preponderance of evidence shows an association. The association must be replicated in more than one cohort with significant p-values, and, preferably with a strong effect size. Lower levels (3-4) are less significant and may only be based on a single study or case report, which may be performed in vitro.(PHARMGKB)

0.3 Haplotype Guidelines

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0.4 Clinical Annotations

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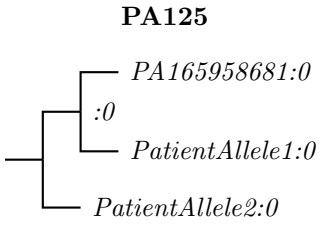
0.5 Appendix 1: Haplotype trees



0.6 Appendix 2: Top 3 haplotypes

	Allele 1	Allele 2
CYP2D6	*1	*1
	*2	*2
	*3	*3

0.7 Appendix 3: Low level annotations

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