RPRD DIAGNOSTICS
— Right Patient | Right Drug —

SPECIFICATIONS

Cytochrome P450 family 2 subfamily C type 19 enzyme (CYP2C19) Genotyping

CYP2C19 encodes the cytochrome P450 family 2 subfamily C type 19 enzyme. CYP2C19 is estimated to be responsible for the metabolism of 5-10% of the medications that are currently in clinical use. Clinical Pharmacogenetics Implementation Consortium (CPIC) has curated several clinical recommendations to guide the therapy of clopidogrel, proton pump inhibitors (PPIs), voriconazole, and selective serotonin reuptake inhibitors (SSRIs) and tricyclic antidepressants respectively, using the pharmacogenetic information of CYP2C19.

Alleles Detected

Normal function allele: *1
Decreased function allele: *9, *10
No function allele: *2, *3, *4A, *4B, *5, *6, *7, *8, *35
Increased function allele: *17

Phenotype Categories (Possible Results)

- **Normal Metabolizer**: Result indicates the presence of two copies of a normal function allele in the CYP2C19 gene.

  Diplotype: *1/*1

- **Intermediate Metabolizer**: Result indicates the presence of one copy of a normal function or increased function allele and one copy of a no function allele in the CYP2C19 gene. Based on the genotype result this patient is predicted to be a CYP2C19 intermediate metabolizer. This patient is at risk for adverse reactions to medications that are metabolized by CYP2C19. Dose adjustments or alternative therapeutic agents may be necessary.

  Diplotypes: *1/*2; *1/*3; *1/*4A; *1/*4B or *4A/*17; *1/*5; *1/*6; *1/*7; *1/*8; *2/*17; *3/*17; *4B/*17; *5/*17; *6/*17; *7/*17; *8/*17; *17/*35

- **Possible Intermediate Metabolizer**: Result indicates the presence of normal, increased or decreased function alleles in the CYP2C19 gene. Based on the genotype result this patient's phenotype may fall into the range of a normal to intermediate metabolizer. This patient is at possible risk for adverse reactions to medications that are metabolized by CYP2C19. Dose adjustments or alternative therapeutic agents may be necessary.

  Diplotypes: *1/*9; *1/*10; *9/*9; *9/*10; *9/*17 *10/*10; *10/*17

- **Poor Metabolizer:** Result indicates the presence of two copies of a no function allele in the CYP2C19 gene. Based on the genotype result this patient is predicted to be a CYP2C19 poor metabolizer. This patient is at a high risk for adverse reactions to medications that are metabolized by CYP2C19.

  Diplotypes: *2/*2; *2/*3; *2/*4A; *2/*4B; *2/*5; *2/*6; *2/*7; *2/*8; *2/*35; *3/*3; *3/*4A; *3/*4B; *3/*5; *3/*6; *3/*7; *3/*8; *3/*35;*4A/*4A; *4A/*5; *4A/*6; *4A/*7; *4A/*8; *4A/*35; *4A/*4B; *4B/*4B; *4B/*5; *4B/*6; *4B/*7; *4B/*8; *4B/*35; *5/*5; *5/*6; *5/*7; *5/*8; *5/*35; *6/*6; *6/*7; *6/*8; *6/35; *7/*7; *7/*8; *7/*35; *8/*8; *8/*35; *35/*35

- **Likely Poor Metabolizer**: Result indicates the presence of two copies of a no function allele in the CYP2C19 gene. Based on the genotype result this patient's phenotype may fall into the range of an intermediate to poor metabolizer. This patient is at possible high risk for adverse reactions to medications that are metabolized by CYP2C19. Dose adjustments or alternative therapy may be necessary.

  *Diplotypes***:** *2/*9; *2/*10; *3/*9; *3/*10; *4A/*9; *4A/*10; *4B/*9; *4B/*10; *5/*9; *5/*10; *6/*9; *6/*10; *7/*9; *7/*10; *8/*9; *8/*10; *9/*35

Limitations

The presence of other CYP2C19 variants that are not detected in this assay may influence drug metabolism. Additional variants that are not detected but located proximally to variants interrogated may affect the assay's ability to detect the above listed alleles.

Please consider all relevant patient information (including but not limited to additional medications, sex, weight, etc.) when making any dosing decisions.

Methodology

Real-time polymerase chain reaction with fluorescence detection.

For More Information

RPRD Diagnostics, LLC
1225 Discovery Parkway Ste 260
Wauwatosa, WI 53226
(414) 316-3097
info@rprdx.com
www.rprdx.com

References

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9. PharmGKB, a pharmacogenomics knowledge resource, see https://www.pharmgkb.org/