ConectGene TESTES GENÉTICOS

Pharmacogenetic Test Results

Medical Director: Dr. Paulo Magno do Bem Filho
CRM ES 13785

Rua Equador, 43 – Bloco 3 – 9º. andar - sala 1011
Porto Atlântico Square Business
20220-410 – Rio de Janeiro (RJ)
55 21 21358716 (Ph)

PATIENT: Doe, Jane (F)
DOB: 1985-01-01
PATIENT ID:

COLLECTED: 10/15/2018
RECEIVED: 11/06/2018
REPORTED: 11/30/2018

SAMPLE TYPE: Buccal
PHYSICIAN: Not Provided
PRACTICE: Not Provided

ACCESSION: LGE-9290880938

QUICK SUMMARY

PROTON PUMP INHIBITORS	RESULTS
Lansoprazole (PREVACID®) Omeprazole (PRILOSEC®) Pantoprazole (PROTONIX®)	✓ Consider label recommended dosage if no contraindication.

IMPORTANT

This Quick Summary provides a brief overview of the predicted response of the patient. This information is based solely on the genotype information and is not based on a complete patient profile. Detection or absence of variants does not replace the need for therapeutic monitoring. Physicians should consider the information contained in the Details section, as well as consider current prescriptions, family history, presenting symptoms, and other factors before making any clinical or therapeutic decisions.

✓ No negative assertions based on genotype.

⚠ Genotype may present increased risk or decreased effectiveness; prescribe with caution.

⊘ Genotype may present increased risk or decreased effectiveness; select alternative drug.

GENE SUMMARY

GENE	GENOTYPE	PHENOTYPE
CYP2C19	*1/*17	⚠ Rapid Metabolizer

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DETAILED INFORMATION

Lansoprazole	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	Consider label recommended dosage of Lansoprazole if no contraindication.		
Omeprazole	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	Consider label recommended dosage of Omeprazole if no contraindication.		
Pantoprazole	CYP2C19 *1/*17	*Rapid metabolizer.*	Evidence ★★★
	Consider label recommended dosage of Pantoprazole if no contraindication.		

KEY FOR VARIANT-DRUG COMBINATION EVIDENCE

★★★ Replicated in multiple studies with statistical significance and strong effect size.
★★ Replicated in multiple studies with and without statistical significance and effect size may be minimal.
★ Not yet replicated or replicated but lacking clear evidence of an association.
Notable information is available and special considerations may be of interest when prescribing for this genotype.
Literature does not indicate additional risks, benefits, or prescription changes to consider for this genotype.

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REPORTED GENOTYPES

This panel performs genotyping analysis on key genes and variant hot-spot regions, focused on analyzing loci documented as altering the effectiveness of drug metabolism. Key genotyping results include the following:

CYP2C19
CYP2C19 *1/*17
rs4244285:G/G Wild
rs4986893:G/G Wild
rs28399504:A/A Wild
rs56337013:C/C Wild
rs72552267:G/G Wild
rs72558186:T/T Wild
rs41291556:T/T Wild
rs17884712:G/G Wild
rs6413438:C/C Wild
rs55640102:A/A Wild
rs12248560:C/T Het

Reported genotype calls are all displayed with respect to the positive DNA strand. Variants indicated as homozygous (Hom) or heterozygous (Het) differ from the GRCh37/hg19 reference sequence (Wild). This report is limited to the following star-alleles: CYP2D6: *1, *2, *3, *3B, *4, *5, *6C, *6, *7, *8, *9, *10, *11, *12, *14A, *14B, *15, *17, *29, *33, *35A, *41, *45A & *46. CYP2C19: *1, *2, *3, *4B, *4, *5, *6, *7, *8, *9, *10, *12 & *17. CYP2C9: *1, *2 & *3. CYP3A5: *1 & *3. CYP3A4: *1, *8, *11, *12, *13, *16, *17 & *22. TPMT: *1, *2, *3A, *3C, *3B & *4. DPYD: *1, *2, *4, *5, *13 & rs67376798A. Any genotype identified as a default star-allele (CYP2D6 *2, CYP2C19 *1, CYP2C9 *1, CYP3A5 *3, CYP3A4 *1, TPMT *1, DPYD *1) indicates the absence only of the other alleles listed and does not imply that other variants in the gene are absent. Full allele deletions and duplications are only analyzed for the CYP2D6 gene. This test does not report polymorphisms other than those specifically listed, and mutations in other genes associated with drug metabolism will not be detected. Rare diagnostic errors may occur if variations occur in primer site locations.

DISCLAIMER

The information presented on this report is provided as supplementary health information. The results presented are intended for use by a physician, pharmacist or other healthcare professional to advise a patient on the use of prescribed medications. This test is not a 510k cleared test, but managed by CMS and FDA under the Clinical Laboratory Improvement Amendment (CLIA) as a LDT. The ordering physician is responsible for the diagnosis and management of disease and decisions based on the data provided. Results are dependent on adequate specimen collection and processing.

METHODOLOGY

Genomic DNA is extracted from dry buccal swabs using magnetic particle processing. DNA from patient samples are amplified with primers specific for CYP2C19 & CYP2C19 using Nested Patch PCR (Varley, et. al.). Positive and negative controls are used with each run. Patient samples, positive, and negative controls are sequenced using a MiSeq (Illumina). Sequences are analyzed using alignment and base call algorithms with Kailos Blue Software for the presence or absence of single nucleotide base changes, insertions and deletions. Results and recommendations are compiled as part of a medical report.

Genetic testing was performed in the Kailos Genetics CLIA facility at 601 Genome Way; Huntsville, Al. 35806. CLIA#: 01D2016114. Medical Director: Ronald McGlennen MD, FCAP, FACMG, ABMG.

This report was reviewed and approved for release by CLIA Lab Manager & Supervisor: Michele R. Erickson-Johnson, PhD, MB (ASCP)^{CM}

REFERENCES

M.V. Relling, T.E. Klein. "CPIC: Clincial Pharmacogenetics Implementation Consortium of the Pharmacogenomics Research Network." Clinical Pharmacology & Therapeutics (2011) 89(3): 464-467.

J.J. Swen, M. Nijenhuis, A. de Boer, L. Grandia, A.H. Maitland-van der Zee, H. Mulder, G.A. Rongen, R.H. van Schaik, T. Schalekamp, D.J. Touw, J. van der Weide, B. Wilffert, V.H. Deneer, H.J. Guchelaar. "Pharmacogenetics: from bench to byte--an update of guidelines." Clinical Pharmacology & Therapeutics (2011) 89(5): 662-673.

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K.E. Varley, R.D. Mitra. "Nested Patch PCR Enables Highly Multiplexed Mutation Discovery in Candidate Genes." Genome Research (2008) 18: 1844-1850.

M. Whirl-Carrillo, E.M. McDonagh, J. M. Hebert, L. Gong, K. Sangkuhl, C.F. Thorn, R.B. Altman and T.E. Klein. "Pharmacogenomics Knowledge for Personalized Medicine" Clinical Pharmacology & Therapeutics (2012) 92(4): 414-417.