

Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
Patient Last Name			Account Address		
Patient First Name Charlotte		Patient Middle Name			
Patient SS#	Patient Phone	Total Volume			
Age (Y/M/D) 52 y.o.	Date of Birth	Sex Female	Fasting		
Patient Address			Additional Information		
			Specimen Type: Peripheral Blood Ethnicity: African American Indication: Possible Family History of poor clopidogrel response		
Date and Time Collected	Date Entered	Date and Time Reported	Physician Name Jane Ferreiro, MD	NPI	Physician ID

Cardio PGx Panel	Tests Ordered
General Comments	
Please send a copy of the final report to the Molecular Science/M1 Training office via Fax at (202) 555-1212	

Clinical test results for Cardio PGx Panel

16 drug response genes tested:

- Warfarin response
- Clopidogrel response
- Metformin response
- Tolbutamide response
- Carvedilol response
- Metoprolol response
- Propafenone response
- Propranolol response
- Timolol response
- Prasugrel response
- Nebivolol Response
- Acenocoumarol response
- Saxagliptin response
- Flecainide response
- Gliclazide response
- Glimepiride response

GENE	TEST RESULTS	EXPLANATION
CYP2C19 (10q23.33)	p.Trp212Ter p.Trp212Ter (also known As *3 *3)	This result confirms risk for Clopidogrel response, as a Poor Metabolizer. This result should be interpreted in the context of clinical presentation, results of other laboratory tests and family history. A PCR/sequencing study has detected two copies of the NM_000769.1(CYP2C19):c.636G>A (p.Trp212Ter) variation. This mutation encoded by a G to A change at nucleotide position 636 in the CYP2C19 mRNA and results in an early termination of the protein at position 212 in the protein. In addition, this individual's result has important implications for other family members. Clinical evaluations should be considered, and genetic counseling is recommended for at-risk individuals.
Benign genetic variant(s) were detected in:		CYP3A4 (7q22.1) <i>(Please contact us for more information, if required.)</i>
No genetic variants were detected in:		CYP2C9 (10q23.33) CYP2D6 (22q13.2) ATM (11q22.3) VKORC1 (16p11.2)

DISCLAIMER:

Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

TEST PROCEDURE:

Buccal swabs are used as the specimen type. Genomic DNA is extracted from buccal swabs using an ABI magnetic particle processor (MagMAX). Purified DNA is then quantified using qPCR. Microarray analysis is then performed using the Illumina Global Screening Array v3 (w/DTC booster) beadchip and the Illumina iScan. CYP2D6 copy number is determined using qPCR. Select SNPs are genotyped using ABI TaqMan assays. All PCR is performed on an ABI QuantStudio 12K Flex (384-well block). PGx reports are generated using the Coriell Life Sciences API interface.

CLINICAL DESCRIPTION

All drugs are metabolized to some extent in the human body. Nearly all of the enzymes and transporters involved in drug metabolism are known to contain genetic polymorphisms across the human population. Some of these polymorphisms are known to contribute to altered function and subsequent altered drug metabolism. Therefore, determination of known polymorphisms contained in each drug-metabolizing enzyme can be predictive of enzyme activity and overall drug metabolism. This allows for the adjustment of dose or selection of an alternate medication if an individual patient's phenotype requires. Pharmacogenomic testing has been proven to be a valuable tool for patients undergoing a wide range of drug therapies.

Check Conditions for which test is offered, Clinical features section, and links for additional information about this condition.

Condition/Phenotype	Identifier
Clopidogrel response	C2674941
Metformin response	C3547187
Tolbutamide response	C4016718
Warfarin response	MIM:122700, C0750384
Acenocoumarol response	CN297587
Carvedilol response	CN077965
Flecainide response	CN298368
Gliclazide response	CN311873
Glimepiride response	CN437678
Metoprolol response	CN077996
Nebivolol Response	CN282565
Prasugrel response	CN239061
Propafenone response	CN078001
Propranolol response	CN078002
Saxagliptin response	CN297809
Timolol response	CN078019