

PHARMACOGENOMICS TESTING

Why Medication Management Matters

Phi Life Sciences



About Phi Life Sciences

Phi Life Sciences is a Molecular Diagnostic and Laboratory Service company.

Our goal is to commercialize novel technologies and develop products that empower physicians to deliver precision treatments and diagnoses by incorporating molecular tests that will improve patients' health.





Charleston, SC Location

645 Meeting Street, Suite 3
Charleston, SC 29403
CLIA#: 42D2114196
CAP#: 8079143





Rockville, MD Location

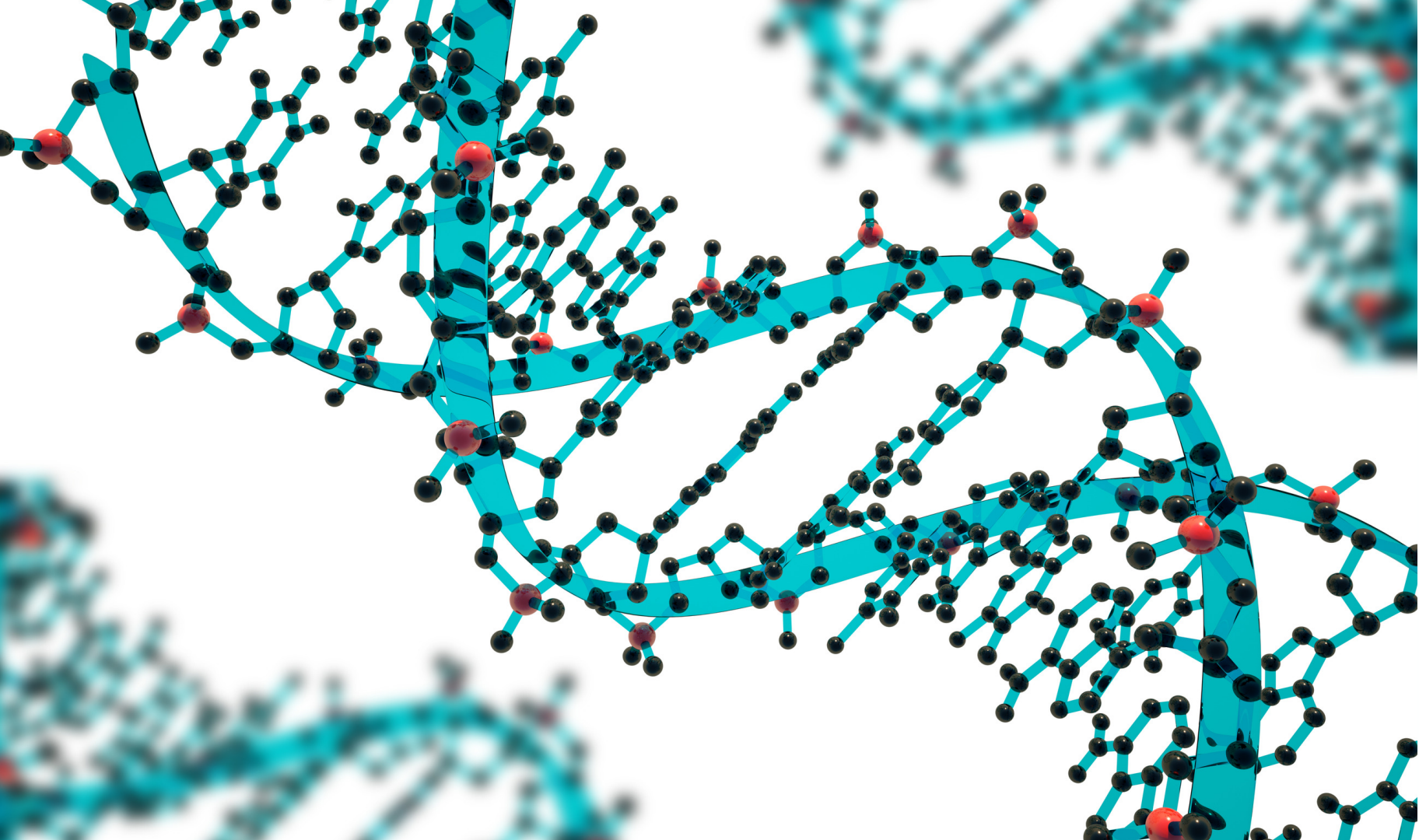
9430 Key West Ave #130
Rockville, MD 20850
CLIA #: 21D2158103

WHAT IS PHARMACOGENETICS?

Pharmacogenetics is the clinical testing of the genetic variations (i.e. SNPs) that give rise to differing responses to drugs.

Genetic differences in metabolic pathways can affect an individual's response to drugs in terms of therapeutic effect as well as adverse events.





Genes and SNPs

All enzymes (like the CYP enzymes) are constructed by DNA “blueprints” called genes. Genes commonly have DNA code variations called “Single Nucleotide Polymorphism” or “SNPs” (pronounced “snips”).

SNPs can be neutral, meaning they have no effect on the enzyme “blueprint” or they can have small to profound effects on the enzyme “blueprint” depending on the change.

Types of Metabolizers

POOR METABOLIZER

There are two variants present. Typically one is on the maternal-derived chromosome and a second variant on the paired paternal-derived chromosome. The alleles on both chromosomes produce "loss of function" enzymes.

INTERMEDIATE METABOLIZER

Having one gene variant which produces an enzyme with a significant loss of enzyme function.

or

Having one or two gene variants which produce only a modest loss of enzyme function.

or

Having one significant loss of function variant and one modest loss of function variant.

NORMAL METABOLIZER

No variant alleles present on either of the patient's paired chromosomes. Enzyme activity is normal.

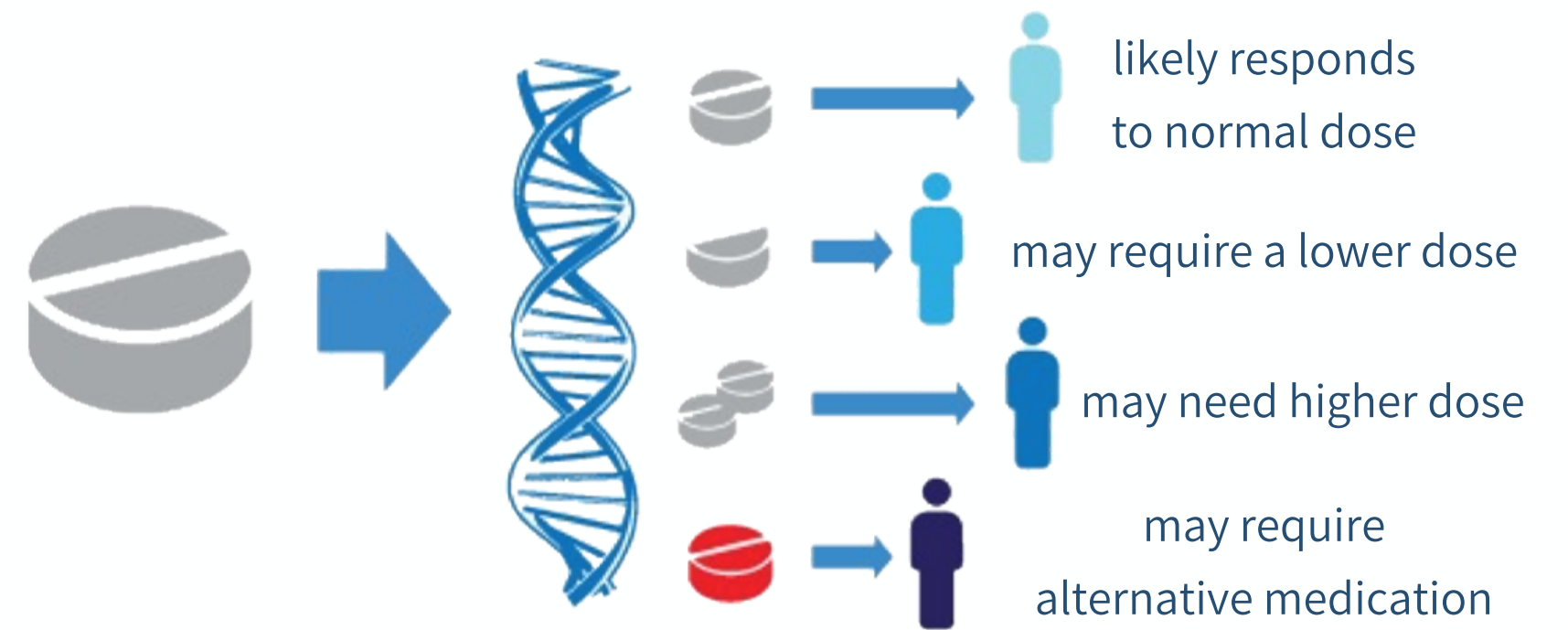
ULTRA RAPID OR RAPID METABOLIZER

Having one or two enzymes with significant gain of enzyme function. If there are two variants present, this will often cause more rapid metabolism.

Enzyme Function



Personalized Medicine



The one-size-fits-all approach to prescribing may not work for all patients.

Personalized prescribing helps you select the right drug and the right dose.



How is Pharmacogenetics Testing Used?

- To minimize adverse drug reactions (ADRs)
- To select the best medication the first time around
- To adjust the dose based on the genetic information, versus trial and error dosing

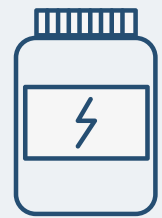


WHO NEEDS PHARMACOGENETIC TESTING?

Anyone...

- Taking medications
- Long Term Medication usage
- Not getting full benefit from medication(s)
- Taking warfarin (Coumadin), clopidogrel (Plavix) or blood thinners or beta-blockers
- SSRI or Psychotropic medication(s)
- Statins
- With cancer, psychiatric symptoms or other conditions where “trial and error” prescribing carries high risk
- With unexplained symptoms

Phi Life Sciences' PGx Tests



General Panel



Cardiology Panel



Psychiatry Panel



Pain Management

List of Genes Tested



- ABCG2 421C>A
- ANKK1/DRD2 DRD2:Taq1A
- Apolipoprotein E ϵ 2, ϵ 4, (ϵ 3 is reference)
- COMT Val158Met
- CYP1A2 *1F
- CYP2B6 *6, *9
- CYP2C19 *2, *3, *4, *4B, *5, *6, *7, *8, *9, *10, *17
- CYP2C9 *2, *3
- CYP2D6 *2, *3, *4, *4M, *6, *7, *8, *9, *10, *12, *17, *29, *35, *41, *5 (gene deletion), XN (gene duplication)
- CYP3A4 *1B, *22
- CYP3A5 *3, *3C, *6, *7
- DPYD rs1801265, rs1801267, rs3918290, rs55886062, rs67376798
- DRD2 -241A>G
- Factor II 20210G>A
- Factor V Leiden 1691G>A
- HTR2A -1438G>A, rs7997012
- HTR2C -759C>T, 114138144C>G
- ITGB3 176T>C
- LPA rs3798220, rs10455872
- MTHFR 1298A>C, 677C>T
- OPRM1 A118G
- SLCO1B1 521T>C
- TPMT *3A, *3B, *3C
- VKORC1 -1639G>A



MEDICATION THERAPY MANAGEMENT

Genetic testing will identify the individual's ability to metabolize medication via the P-450 enzyme for medication efficacy.



How Often are Prescriptions Written?

Two-thirds of patient visits result in a prescription.

Specialist Providers write 2.3 prescriptions per visit.

Three **BILLION** outpatient prescriptions are written a year.

89.2% of Medicare patients take a prescription medications every day.

REDUCING AND PREVENTING ADVERSE DRUG EVENTS TO DECREASE HOSPITAL COSTS

Adverse drug events (ADEs) result in more than 770,000 injuries and deaths each year and **cost up to \$5.6 million per hospital**, depending on size. Many ADE injuries and resulting hospital costs can be reduced if hospitals make changes to their systems for preventing and detecting ADEs.





Why Learn About Adverse Drug Reactions?



- There are over **2,000,000** serious adverse drug reactions every year. (2,3,4)
- There are **100,000** deaths due to adverse drug reactions yearly. (2,3,4)
- Approximately **10%** of hospital admissions are due to adverse drug reactions. (2,3,4)
- There are about **350,000** nursing home patients that have adverse drug reactions every year. (2,3,4)

Adverse Reactions and Drug Interactions



- Adverse drug reactions increase exponentially when taking **more than four prescription medications**. (7,8,9)
- 46.1% of Medicare patients take more than five prescriptions every day. (7,8,9)
- 53.6% Medicare patients take prescription medications written by more than two or more providers. (7,8,9)
- Drug interactions are preventable and account for 3-5% of total preventable in-hospital adverse drug reactions. (10,11)
- Adverse events due to drug interactions are one of the important contributors to the number of emergency room visits and hospital admissions seen. (10,11)
- Adverse drug reactions cause **1 in 5 hospital patients to experience an injury or death**. (4,5,6)
- Hospital patients with adverse drug reactions stay in the hospital **twice as long, cost twice as much and have a twice as greater risk for mortality**. (4,5,6)



\$136 Billion

The cost of adverse drug reactions nationwide is \$136,000,000,000 every year. (4,5,6)

The total cost of adverse drug reactions is larger than diabetic and cardiovascular care. (4,5,6)

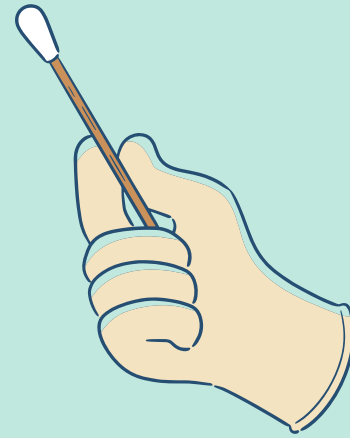
Clinical Summary

- Access to the latest molecular technology
- Most comprehensive panels, designed with clinically significant variants
- 48-72 Hour turn-around-time after receipt to the laboratory
- Insurance verification
- Easy to interpret reporting
- Unmatched Clinical Support – MD, PharmD, PhD, & Nursing
- Our HIPAA Compliant Portal: cloud based and easy to use





Collection Directions



1. Use one puritan double swab per patient.
2. Place barcode with patient DOB on the swab tube
3. Label the Requisition with the matching barcode and information as in #2. **If the swab or requisition is missing information, then the test cannot be run.**
4. Open the swab package at the handle end (NOT the collection tip end) and carefully peel the package away to within about an inch of the end. Uncap the dry transport tube and remove the swab from the package. Do not touch the collection swab from the package. Do not touch the collection tip of the swab.
5. Do not allow the tip of the swab to touch anything other than the inside of the patient's mouth and the inside of the packaging.
6. While holding the handle end of the swab, insert the tips along the inside of the cheek. Rotate the collection tips along the inside of the cheeks for 60 seconds. Pushing on the outside of the patient's cheek while rotating the swab will firmly press it between the gum and cheek and will enhance **cell collection**.
7. Hold the swab while it air dries for several seconds. **Do NOT blow on the swab.**
8. Break off top of Dry Transport tube and place swab into the tube then place it into Specimen Bag with Patient's requisition and documents.



Accepted Insurance Carriers



Federal Carriers:

- Medicare (Palmetto and Novitas MAC)
- Medicaid (ID, MT, OH, OR, SC, GA, CO)
- RR Medicare

Medicare Advantage Plans:

- United Healthcare
- Aetna
- Cigna
- Humana
- And most others with insurance verification

Commercial Insurance Carriers:

- Most commercial carriers accepted with insurance verification
- No BCBS

PHI LIFE SCIENCES REQUISITION FORM - PGX		CLIA# 21D2158103 9430 KEY WEST AVE., #130 ROCKVILLE, MD 20850 CLIA# 42D2114196 645 MEETING ST., SUITE 3, CHARLESTON, SC 29403	
		AFFIX BAR CODE HERE	
PATIENT INFORMATION			
Last Name		First Name	
Street Address		City	
Phone Number		State	
Race		Sex	
		☐ Male ☐ Female ☐ Other	
FACILITY INFORMATION			
Facility Name		Phone	
Address		City	
		State	
		Zip Code	
INSURANCE INFORMATION			
Insurance Carrier		Carrier Phone Number	
Carrier Address		City	
		State	
		Zip Code	
☐ Workers Comp/Auto/LOC ☐ Self-Pay ☐ Insurance (See Below)			
Member ID		RX Group	
		RX BIN	
		PCN	
PHYSICIAN INFORMATION			
Name		Phone Number	
		NPI Number	
		DEA Number	
Address		City	
		State	
		Zip Code	
SPECIMEN INFORMATION			
Collection Date (MM/DD/YYYY)		Certification of Specimen Handler REQUIRED	
Please make sure all applicable paperwork is filled out and signed			
PAYMENT OPTIONS			
☐ Medicare Advantage/Medicare Replacement Plan ☐ Medicare ☐ Medicaid ☐ TriCare ☐ Commercial ☐ HSA			
COMPREHENSIVE		CARDIO & THROMBOSIS	
☐ GENERAL GENE PANEL ABCG2, ANK11/DRD2, COMT, CYP1A2, CYP2A6, CYP2C8, CYP2C9, CYP2C19, CYP2D6, CYP2E1, CYP3A4, CYP3A5, CYP3A6, CYP3A7, CYP3A8, CYP3A9, CYP3A10, CYP3A11, CYP3A12, CYP3A13, CYP3A14, CYP3A15, CYP3A16, CYP3A17, CYP3A18, CYP3A19, CYP3A20, CYP3A21, CYP3A22, CYP3A23, CYP3A24, CYP3A25, CYP3A26, CYP3A27, CYP3A28, CYP3A29, CYP3A30, CYP3A31, CYP3A32, CYP3A33, CYP3A34, CYP3A35, CYP3A36, CYP3A37, CYP3A38, CYP3A39, CYP3A40, CYP3A41, CYP3A42, CYP3A43, CYP3A44, CYP3A45, CYP3A46, CYP3A47, CYP3A48, CYP3A49, CYP3A50, CYP3A51, CYP3A52, CYP3A53, CYP3A54, CYP3A55, CYP3A56, CYP3A57, CYP3A58, CYP3A59, CYP3A60, CYP3A61, CYP3A62, CYP3A63, CYP3A64, CYP3A65, CYP3A66, CYP3A67, CYP3A68, CYP3A69, CYP3A70, CYP3A71, CYP3A72, CYP3A73, CYP3A74, CYP3A75, CYP3A76, CYP3A77, CYP3A78, CYP3A79, CYP3A80, CYP3A81, CYP3A82, CYP3A83, CYP3A84, CYP3A85, CYP3A86, CYP3A87, CYP3A88, CYP3A89, CYP3A90, CYP3A91, CYP3A92, CYP3A93, CYP3A94, CYP3A95, CYP3A96, CYP3A97, CYP3A98, CYP3A99, CYP3A100, CYP3A101, CYP3A102, CYP3A103, CYP3A104, CYP3A105, CYP3A106, CYP3A107, CYP3A108, CYP3A109, CYP3A110, CYP3A111, CYP3A112, CYP3A113, CYP3A114, CYP3A115, CYP3A116, CYP3A117, CYP3A118, CYP3A119, CYP3A120, CYP3A121, CYP3A122, CYP3A123, CYP3A124, CYP3A125, CYP3A126, CYP3A127, CYP3A128, CYP3A129, CYP3A130, CYP3A131, CYP3A132, CYP3A133, CYP3A134, CYP3A135, CYP3A136, CYP3A137, CYP3A138, CYP3A139, CYP3A140, CYP3A141, CYP3A142, CYP3A143, CYP3A144, CYP3A145, CYP3A146, CYP3A147, CYP3A148, CYP3A149, CYP3A150, CYP3A151, CYP3A152, CYP3A153, CYP3A154, CYP3A155, CYP3A156, CYP3A157, CYP3A158, CYP3A159, CYP3A160, CYP3A161, CYP3A162, CYP3A163, CYP3A164, CYP3A165, CYP3A166, CYP3A167, CYP3A168, CYP3A169, CYP3A170, CYP3A171, CYP3A172, CYP3A173, CYP3A174, CYP3A175, CYP3A176, CYP3A177, CYP3A178, CYP3A179, CYP3A180, CYP3A181, CYP3A182, CYP3A183, CYP3A184, CYP3A185, CYP3A186, CYP3A187, CYP3A188, CYP3A189, CYP3A190, CYP3A191, CYP3A192, CYP3A193, CYP3A194, CYP3A195, CYP3A196, CYP3A197, CYP3A198, CYP3A199, CYP3A200, CYP3A201, CYP3A202, CYP3A203, CYP3A204, CYP3A205, CYP3A206, CYP3A207, CYP3A208, CYP3A209, CYP3A210, CYP3A211, CYP3A212, CYP3A213, CYP3A214, CYP3A215, CYP3A216, CYP3A217, CYP3A218, CYP3A219, CYP3A220, CYP3A221, CYP3A222, CYP3A223, CYP3A224, CYP3A225, CYP3A226, CYP3A227, CYP3A228, CYP3A229, CYP3A230, CYP3A231, CYP3A232, CYP3A233, CYP3A234, CYP3A235, CYP3A236, CYP3A237, CYP3A238, CYP3A239, CYP3A240, CYP3A241, CYP3A242, CYP3A243, CYP3A244, CYP3A245, CYP3A246, CYP3A247, CYP3A248, CYP3A249, CYP3A250, CYP3A251, CYP3A252, CYP3A253, CYP3A254, CYP3A255, CYP3A256, CYP3A257, CYP3A258, CYP3A259, CYP3A260, CYP3A261, CYP3A262, CYP3A263, CYP3A264, CYP3A265, CYP3A266, CYP3A267, CYP3A268, CYP3A269, CYP3A270, CYP3A271, CYP3A272, CYP3A273, CYP3A274, CYP3A275, CYP3A276, CYP3A277, CYP3A278, CYP3A279, CYP3A280, CYP3A281, CYP3A282, CYP3A283, CYP3A284, CYP3A285, CYP3A286, CYP3A287, CYP3A288, CYP3A289, CYP3A290, CYP3A291, CYP3A292, CYP3A293, CYP3A294, CYP3A295, CYP3A296, CYP3A297, CYP3A298, CYP3A299, CYP3A300, CYP3A301, CYP3A302, CYP3A303, CYP3A304, CYP3A305, CYP3A306, CYP3A307, CYP3A308, CYP3A309, CYP3A310, CYP3A311, CYP3A312, CYP3A313, CYP3A314, CYP3A315, CYP3A316, CYP3A317, CYP3A318, CYP3A319, CYP3A320, CYP3A321, CYP3A322, CYP3A323, CYP3A324, CYP3A325, CYP3A326, CYP3A327, CYP3A328, CYP3A329, CYP3A330, CYP3A331, CYP3A332, CYP3A333, CYP3A334, CYP3A335, CYP3A336, CYP3A337, CYP3A338, CYP3A339			

General PGx Results



PATIENT INFORMATION	SPECIMEN DETAILS	ORDERED BY
NAME: Patient, Patient ACC #: PHI Sample DOB: 01/01/2001 SEX: Female	SPECIMEN TYPE: Buccal Swab COLLECTION DATE: 7/27/2017 RECEIVED DATE: 7/28/2017 REPORT DATE: 8/2/2017	Doctor, Doctor

General Gene Panel

Test Details

Gene	Genotype	Phenotype	Clinical Consequences
ABCG2	421C>A C/C	Normal Function	Consistent with a normal ABCG2 transporter function. The patient's risk for statin-induced adverse events is normal.
ANKK1/DRD2	DRD2:Taq1A G/G	Unaltered DRD2 function	Consistent with a normal dopamine receptor D2 function.
Apolipoprotein E	ε3/ε3	Normal APOE function	Not associated with type III hyperlipoproteinemia - No increased risk of cardiovascular disease
COMT	Val158Met A/G	Intermediate COMT Activity	Consistent with a reduced catechol O-methyltransferase (COMT) function.
CYP1A2	*1F/*1F	Normal Metabolizer - Higher Inducibility	Consistent with a typical CYP1A2 activity in absence of inducing substances. Rapid Metabolism occurs in presence of inducers such as barbiturates, cruciferous vegetables, carbamazepine, rifampin and smoking.
CYP2B6	*1/*1	Normal Metabolizer	Consistent with a typical CYP2B6 activity. This test did not identify risks for side effects or loss of efficacy with drug substrates.
CYP2C19	*1/*2	Intermediate Metabolizer	Consistent with a moderate deficiency in CYP2C19 activity. Potential risk for side effects or loss of efficacy with drug substrates.
CYP2C9	*1/*2	Intermediate Metabolizer	Consistent with a moderate deficiency in CYP2C9 activity. Potential risk for side effects or loss of efficacy with drug substrates.
CYP2D6	*4/*41	Intermediate Metabolizer	Consistent with moderate to substantial deficiency in CYP2D6 activity. Increased risk for side effects or loss of efficacy with drug substrates.
CYP3A4	*1/*1B	Normal Metabolizer	Consistent with a typical CYP3A4 activity. Caution is advised when prescribing narrow therapeutic index drugs. Alternative drugs or dose adjustment may be required if CYP3A inhibitors or inducers are co-prescribed.
CYP3A5	*3/*3	Poor Metabolizer	Consistent with a poor CYP3A5 activity. This phenotype is the most common in the general population. Caution is advised when prescribing narrow therapeutic index drugs. Alternative drugs or dose adjustment may be required if CYP3A inhibitors or inducers are co-prescribed.
DPYD	*1/*1	Normal Metabolizer	Consistent with a typical DPD activity and a typical risk of side effects with conventional doses of fluoropyridines.
DRD2	-241A>G T/C	Heterozygous for rs1799978 C Allele	Associated with a favorable response to Risperidone.
Factor II Factor V Leiden	20210G>A GG 1691G>A GG	No Increased Risk of Thrombosis	The patient's risk of thrombosis is not increased (average risk of clotting is about 1 in 1000 for anyone in a year). However, because this test cannot find all of the inherited reasons for abnormal clotting, other factors may affect this risk assessment.
HTR2A	-1438G>A C/T	Heterozygous for the T Allele (rs6311)	The patient carries one copy of the variant allele at rs6311 which may be associated with greater serotonin 2A receptor gene expression.
HTR2A	rs7997012 A/G	Heterozygous for the A allele (rs7997012)	Reduced response to citalopram and escitalopram
HTR2C	2565G>C G/C	Heterozygous for the C allele (rs1414334)	This genotype is associated with risperidone- and clozapine-induced metabolic syndrome.
HTR2C	-759C>T C/T	Heterozygous for the C allele (rs3813929)	Consistent with altered satiety signaling mediated by the serotonin receptor 2C (HTR2C). Increased incidence of metabolic side effects (weight gain, hyperglycemia, hyperlipidemia) with atypical antipsychotic medications.

ITGB3	176T>C T/T	Normal Platelet Reactivity	The patient does not carry the 176T>C mutation of the integrin β3 gene. Unless other genetic or circumstantial risk factors are present, he is not expected to have an increased platelet reactivity.
LPA	rs10455872 A/A rs3798220 T/T	No increased risk of cardiovascular disease	The patient is a non carrier of the risk alleles in LPA gene for both the variants (rs3798220 and rs10455872).
MTHFR	677C>T CT	Reduced MTHFR Activity	The patient carries one MTHFR C677T mutation (heterozygous) and the patient's MTHFR activity is reduced slightly. This is not associated with an increased risk of hyperhomocysteinemia.
MTHFR	1298A>C AC 677C>T CT	No Increased Risk of Hyperhomocysteinemia	The patient's reduced MTHFR activity is not a risk factor for hyperhomocysteinemia. Unless other risk factors are present, the patient is not expected to have an increased risk for venous thromboembolism (VTE).
OPRM1	A118G A/A	Normal OPRM1 Function	Consistent with a normal OPRM1 receptor signaling efficiency induced by exogenous opioids. This is associated with a good analgesia following standard opioid doses and a poor response to naltrexone.
SLCO1B1	521T>C T/T	Normal Function	Consistent with a typical SLCO1B1 transporter function. The patient's risk for statin-induced myopathy is not increased.
TPMT	*1/*1	Normal Metabolizer	Consistent with a typical TPMT activity and a typical risk of side effects with conventional doses of thiopurines.
VKORC1	-1639G>A G/G	Low Warfarin Sensitivity	VKORC1 is the site of action of warfarin. The patient may require an increase in warfarin dose.

Alleles Tested: **ABCG2** 421C>A; **ANKK1/DRD2** DRD2:Taq1A; **Apolipoprotein E** ε2, ε4, (ε3 is reference); **COMT** Val158Met; **CYP1A2** *1F; **CYP2B6** *6, *9; **CYP2C19** *2, *3, *4, *4B, *5, *6, *7, *8, *9, *10, *17; **CYP2C9** *2, *3; **CYP2D6** *2, *3, *4, *4M, *6, *7, *8, *9, *10, *12, *17, *29, *35, *41, *5 (gene deletion), XN (gene duplication); **CYP3A4** *1B, *22; **CYP3A5** *3, *3C, *6, *7; **DPYD** *2A, *9A, *9B, rs67376798 A, *13; **DRD2** -241A>G; **Factor II** 20210G>A; **Factor V Leiden** 1691G>A; **HTR2A** -1438G>A, rs7997012; **HTR2C** -759C>T, 2565G>C; **ITGB3** 176T>C; **LPA** rs3798220, rs10455872; **MTHFR** 1298A>C, 677C>T; **OPRM1** A118G; **SLCO1B1** 521T>C; **TPMT** *3A, *3B, *3C; **VKORC1** -1639G>A

Psychiatry PGx Results



PATIENT INFORMATION	SPECIMEN DETAILS	ORDERED BY
NAME: Patient, Patient	SPECIMEN TYPE: Buccal Swab	Doctor, Doctor
ACC #: PHI Sample	COLLECTION DATE: 7/27/2017	
DOB: 01/01/2001	RECEIVED DATE: 7/28/2017	
SEX: Female	REPORT DATE: 8/2/2017	

Psych Gene Panel

Test Details

Gene	Genotype	Phenotype	Clinical Consequences
ANKK1/DRD2	DRD2:Taq1A G/G	Unaltered DRD2 function	Consistent with a normal dopamine receptor D2 function.
COMT	Val158Met A/G	Intermediate COMT Activity	Consistent with a reduced catechol O-methyltransferase (COMT) function.
CYP1A2	*1F/*1F	Normal Metabolizer - Higher Inducibility	Consistent with a typical CYP1A2 activity in absence of inducing substances. Rapid Metabolism occurs in presence of inducers such as barbiturates, cruciferous vegetables, carbamazepine, rifampin and smoking.
CYP2B6	*1/*1	Normal Metabolizer	Consistent with a typical CYP2B6 activity. This test did not identify risks for side effects or loss of efficacy with drug substrates.
CYP2C19	*1/*2	Intermediate Metabolizer	Consistent with a moderate deficiency in CYP2C19 activity. Potential risk for side effects or loss of efficacy with drug substrates.
CYP2C9	*1/*2	Intermediate Metabolizer	Consistent with a moderate deficiency in CYP2C9 activity. Potential risk for side effects or loss of efficacy with drug substrates.
CYP2D6	*4/*41	Intermediate Metabolizer	Consistent with moderate to substantial deficiency in CYP2D6 activity. Increased risk for side effects or loss of efficacy with drug substrates.
CYP3A4	*1/*1B	Normal Metabolizer	Consistent with a typical CYP3A4 activity. Caution is advised when prescribing narrow therapeutic index drugs. Alternative drugs or dose adjustment may be required if CYP3A inhibitors or inducers are co-prescribed.
CYP3A5	*3/*3	Poor Metabolizer	Consistent with a poor CYP3A5 activity. This phenotype is the most common in the general population. Caution is advised when prescribing narrow therapeutic index drugs. Alternative drugs or dose adjustment may be required if CYP3A inhibitors or inducers are co-prescribed.
DRD2	-241A>G T/C	Heterozygous for rs1799978 C Allele	Associated with a favorable response to Risperidone.
HTR2A	-1438G>A C/T	Heterozygous for the T Allele (rs6311)	The patient carries one copy of the variant allele at rs6311 which may be associated with greater serotonin 2A receptor gene expression.
HTR2A	rs7997012 A/G	Heterozygous for the A allele (rs7997012)	Reduced response to citalopram and escitalopram
HTR2C	2565G>C G/C	Heterozygous for the C allele (rs1414334)	This genotype is associated with risperidone- and dozapine-induced metabolic syndrome.
HTR2C	-759C>T C/T	Heterozygous for the C allele (rs3813929)	Consistent with altered satiety signaling mediated by the serotonin receptor 2C (HTR2C). Increased incidence of metabolic side effects (weight gain, hyperglycemia, hyperlipidemia) with atypical antipsychotic medications.
MTHFR	677C>T CT	Reduced MTHFR Activity	The patient carries one MTHFR C677T mutation (heterozygous) and the patient's MTHFR activity is reduced slightly. This is not associated with an increased risk of hyperhomocysteinemia.
VKORC1	-1639G>A G/G	Low Warfarin Sensitivity	VKORC1 is the site of action of warfarin. The patient may require an increase in warfarin dose.

Alleles Tested: ANKK1/DRD2 DRD2:Taq1A; COMT Val158Met; CYP1A2 *1F; CYP2B6 *6, *9; CYP2C19 *2, *3, *4, *4B, *5, *6, *7, *8, *9, *10, *17; CYP2C9 *2, *3; CYP2D6 *2, *3, *4, *4M, *6, *7, *8, *9, *10, *12, *17, *29, *35, *41, *5 (gene deletion), XN (gene duplication); CYP3A4 *1B, *22; CYP3A5 *3, *3C, *6, *7; DRD2 - 241A>G; HTR2A -1438G>A, rs7997012; HTR2C -759C>T, 2565G>C; MTHFR 1298A>C, 677C>T; VKORC1 -1639G>A

Cardiology PGx Results



PATIENT INFORMATION	SPECIMEN DETAILS	ORDERED BY
NAME: Patient, Patient ACC #: PHI Sample DOB: 01/01/2001 SEX: Female	SPECIMEN TYPE: Buccal Swab COLLECTION DATE: 7/27/2017 RECEIVED DATE: 7/28/2017 REPORT DATE: 8/2/2017	Doctor, Doctor

Cardiac Gene Panel

Test Details

Gene	Genotype	Phenotype	Clinical Consequences
ABCG2	421C>A C/C	Normal Function	Consistent with a normal ABCG2 transporter function. The patient's risk for statin-induced adverse events is normal.
Apolipoprotein E	ε3/ε3	Normal APOE function	Not associated with type III hyperlipoproteinemia - No increased risk of cardiovascular disease
COMT	Val158Met A/G	Intermediate COMT Activity	Consistent with a reduced catechol O-methyltransferase (COMT) function.
CYP1A2	*1F/*1F	Normal Metabolizer - Higher Inducibility	Consistent with a typical CYP1A2 activity in absence of inducing substances. Rapid Metabolism occurs in presence of inducers such as barbiturates, cruciferous vegetables, carbamazepine, rifampin and smoking.
CYP2C19	*1/*2	Intermediate Metabolizer	Consistent with a moderate deficiency in CYP2C19 activity. Potential risk for side effects or loss of efficacy with drug substrates.
CYP2C9	*1/*2	Intermediate Metabolizer	Consistent with a moderate deficiency in CYP2C9 activity. Potential risk for side effects or loss of efficacy with drug substrates.
CYP2D6	*4/*41	Intermediate Metabolizer	Consistent with moderate to substantial deficiency in CYP2D6 activity. Increased risk for side effects or loss of efficacy with drug substrates.
CYP3A4	*1/*1B	Normal Metabolizer	Consistent with a typical CYP3A4 activity. Caution is advised when prescribing narrow therapeutic index drugs. Alternative drugs or dose adjustment may be required if CYP3A inhibitors or inducers are co-prescribed.
CYP3A5	*3/*3	Poor Metabolizer	Consistent with a poor CYP3A5 activity. This phenotype is the most common in the general population. Caution is advised when prescribing narrow therapeutic index drugs. Alternative drugs or dose adjustment may be required if CYP3A inhibitors or inducers are co-prescribed.
Factor II Factor V Leiden	20210G>A GG 1691G>A GG	No Increased Risk of Thrombosis	The patient's risk of thrombosis is not increased (average risk of clotting is about 1 in 1000 for anyone in a year). However, because this test cannot find all of the inherited reasons for abnormal clotting, other factors may affect this risk assessment.
ITGB3	176T>C T/T	Normal Platelet Reactivity	The patient does not carry the 176T>C mutation of the integrin β3 gene. Unless other genetic or circumstantial risk factors are present, he is not expected to have an increased platelet reactivity.
LPA	rs10455872 A/A rs3798220 T/T	No increased risk of cardiovascular disease	The patient is a non carrier of the risk alleles in LPA gene for both the variants (rs3798220 and rs10455872).
MTHFR	1298A>C AC 677C>T CT	No Increased Risk of Hyperhomocysteinemia	The patient's reduced MTHFR activity is not a risk factor for hyperhomocysteinemia. Unless other risk factors are present, the patient is not expected to have an increased risk for venous thromboembolism (VTE).
SLCO1B1	521T>C T/T	Normal Function	Consistent with a typical SLCO1B1 transporter function. The patient's risk for statin-induced myopathy is not increased.
VKORC1	-1639G>A G/G	Low Warfarin Sensitivity	VKORC1 is the site of action of warfarin. The patient may require an increase in warfarin dose.

Alleles Tested: ABCG2 421C>A; Apolipoprotein E ε2, ε4, (ε3 is reference); COMT Val158Met; CYP1A2 *1F; CYP2C19 *2, *3, *4, *4B, *5, *6, *7, *8, *9, *10, *17; CYP2C9 *2, *3; CYP2D6 *2, *3, *4, *4M, *6, *7, *8, *9, *10, *12, *17, *29, *35, *41, *5 (gene deletion), XN (gene duplication); CYP3A4 *1B, *22; CYP3A5 *3, *3C, *6, *7; Factor II 20210G>A; Factor V Leiden 1691G>A; ITGB3 176T>C; LPA rs3798220, rs10455872; MTHFR 1298A>C, 677C>T; SLCO1B1 521T>C; VKORC1 - 1639G>A

Pain Management PGx Results



PATIENT INFORMATION	SPECIMEN DETAILS	ORDERED BY
NAME: Patient, Patient	SPECIMEN TYPE: Buccal Swab	
ACC #: PHI Sample	COLLECTION DATE: 7/27/2017	Doctor, Doctor
DOB: 01/01/2001	RECEIVED DATE: 7/28/2017	
SEX: Female	REPORT DATE: 8/2/2017	

Pain Management Panel

Test Details

Gene	Genotype	Phenotype	Clinical Consequences
COMT	Val158Met A/G	Intermediate COMT Activity	Consistent with a reduced catechol O-methyltransferase (COMT) function.
CYP1A2	*1F/*1F	Normal Metabolizer - Higher Inducibility	Consistent with a typical CYP1A2 activity in absence of inducing substances. Rapid Metabolism occurs in presence of inducers such as barbiturates, cruciferous vegetables, carbamazepine, rifampin and smoking.
CYP2B6	*1/*1	Normal Metabolizer	Consistent with a typical CYP2B6 activity. This test did not identify risks for side effects or loss of efficacy with drug substrates.
CYP2C19	*1/*2	Intermediate Metabolizer	Consistent with a moderate deficiency in CYP2C19 activity. Potential risk for side effects or loss of efficacy with drug substrates.
CYP2C9	*1/*2	Intermediate Metabolizer	Consistent with a moderate deficiency in CYP2C9 activity. Potential risk for side effects or loss of efficacy with drug substrates.
CYP2D6	*4/*41	Intermediate Metabolizer	Consistent with moderate to substantial deficiency in CYP2D6 activity. Increased risk for side effects or loss of efficacy with drug substrates.
CYP3A4	*1/*1B	Normal Metabolizer	Consistent with a typical CYP3A4 activity. Caution is advised when prescribing narrow therapeutic index drugs. Alternative drugs or dose adjustment may be required if CYP3A inhibitors or inducers are co-prescribed.
CYP3A5	*3/*3	Poor Metabolizer	Consistent with a poor CYP3A5 activity. This phenotype is the most common in the general population. Caution is advised when prescribing narrow therapeutic index drugs. Alternative drugs or dose adjustment may be required if CYP3A inhibitors or inducers are co-prescribed.
Factor II Factor V Leiden	20210G>A GG 1691G>A GG	No Increased Risk of Thrombosis	The patient's risk of thrombosis is not increased (average risk of clotting is about 1 in 1000 for anyone in a year). However, because this test cannot find all of the inherited reasons for abnormal clotting, other factors may affect this risk assessment.
OPRM1	A118G A/A	Normal OPRM1 Function	Consistent with a normal OPRM1 receptor signaling efficiency induced by exogenous opioids. This is associated with a good analgesia following standard opioid doses and a poor response to naltrexone.
VKORC1	-1639G>A G/G	Low Warfarin Sensitivity	VKORC1 is the site of action of warfarin. The patient may require an increase in warfarin dose.

Alleles Tested: COMT Val158Met; CYP1A2 *1F; CYP2B6 *6, *9; CYP2C19 *2, *3, *4, *4B, *5, *6, *7, *8, *9, *10, *17; CYP2C9 *2, *3; CYP2D6 *2, *3, *4, *4M, *6, *7, *8, *9, *10, *12, *17, *29, *35, *41, *5 (gene deletion), XN (gene duplication); CYP3A4 *1B, *22; CYP3A5 *3, *3C, *6, *7; Factor II 20210G>A; Factor V Leiden 1691G>A; MTHFR 1298A>C, 677C>T; OPRM1 A118G; VKORC1 -1639G>A

Methodology

Caution: It is important to note that the combination of genetic metabolizer and non-genetic factors, such as, but not limited to: body size, age, inhibition caused by interference of drugs or food, liver function, and kidney function produce the overall response to a given drug dose.

Methods: This specimen was analyzed for gene mutations by real-time PCR (TaqMan SNP Genotyping, Thermo Fisher) developed by Phi Life Sciences, LLC. These assays were validated following the 1988 CLIA standards. Performance characteristics were validated by Phi Life Sciences Laboratory with analytical specificity and sensitivity of >99% for detection of the variants above.

Lab Disclaimer: The FDA has neither cleared nor approved these assays, nor is FDA pre-market review required. These tests are used for clinical purposes and should not be regarded as investigational or for research only. Diagnosis and treatment decisions are the sole responsibility of the practitioner and does not replace the need for clinical and therapeutic drug monitoring. Hence, the interpretation and commentary are provided to the practitioner for educational purposes only and should not be taken as diagnostic or treatment recommendations. For further assistance with interpretation of these results or referral for genetic counseling, please contact Phi Life Sciences' Clinical Support at 888-576-5445.

Testing Performed By: Phi Life Sciences, LLC, CLIA No. 42D2114196, CLIA No. 21D2158103, CAP 807914301, 645 Meeting St, Suite 3, Charleston, SC 29403.

Laboratory Director: Edwin Naylor, PhD

Disclaimer: The information presented on this report is provided as general educational health information. The content is not intended to be a substitute for professional medical advice, diagnosis, or treatment. Only a physician, pharmacist or other healthcare professional should advise a patient on the use of the medications prescribed. The pharmacogenetic assay involves use of reporting software and genotype-phenotype associations performed by Translational Software (www.translationalsoftware.com). The software has not been evaluated by the Food and Drug Administration. The software, and the report generated by the software, is not intended to diagnose, treat, cure, or prevent any disease. A qualified designee within the lab uses Translational Software to generate and subsequently review the report. The pharmacogenetic report is one of multiple pieces of information that clinicians should consider in guiding their therapeutic choice for each patient. It remains the responsibility of the health-care provider to determine the best course of treatment for a patient. Adherence to dose guidelines does not necessarily assure a successful medical outcome.

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