# PHARMACOGENOMICS (PGX)

• F2

• F5

• G6PD

GRIK4

• HTR2A

• HTR2C

• ITGB3

• MTHFR

NUDT15

OPRD1

• OPRK1

OPRM1

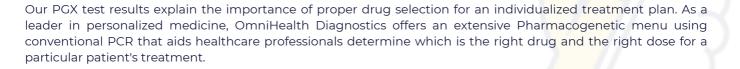
• TPMT

UGTIA1

VKORC1

SLCOIBI

• IFNL3/IFNL4



### **TEST INCLUDES**

GENES

- ABCB1
- ABCG2
- ADC02
  ADRA2A
- ANKK1
- APOE
- COMT
- CYP1A2
- CYP2B6
- CYP2C19
- CYP2C8
- CYP2C9
- CYP2D6
  - CYP3A4
- CYP4F2
- DPYD
- DPYD
- DRD2
- F13A1

## **EXPECTED TURN AROUND TIME**

10-12 Days

## SPECIMEN

OCD-100 Swab

VOLUME

1 OCD-100 Swab

## CONTAINER

OCD-100 Swab



## COLLECTION

- Ensure all proper Personal Protective Equipment measures are taken.
- Ensure that the patient has not had anything to eat, drink, smoke, or chew for 30 minutes prior to sample collection.
- Label the Dry Swab with at least two patient identifiers (i.e. last, first name and DOB).
- Using laboratory gloves (if available) remove the combined swab and tube from its plastic packaging and discard the packaging.
- Open the package and remove collector making sure not to touch the sponge tip. Place the sponge as far back in the mouth as comfortable and rub along the lower gums in a back and forth motion. Gently rub the gums 10 times. If possible, avoid rubbing the teeth.
- Gently repeat rubbing motion on the opposite side of the mouth along the lower gums for an additional 10 times.
- Hold the tube upright to prevent the medium inside the tube from spilling. Unscrew the blue cap from the collection tube without touching the sponge.
- Turn the cap upside down, insert the sponge into the tube and close cap tightly.
- Invert the capped tube and shake vigorously 10 times. NOTE: If sample collection is not completed as instructed, there will NOT be sufficient DNA available for testing.\*
- Once completed, insert the swab into the OCD-100 swab envelope.
- Place the OCD-100 swab into the bio-hazard bag; place all paperwork (test requisition form, medical records, insurance information and demographics) into the bio-hazard bag.

## STORAGE/TRANSPORT INSTRUCTIONS

- Store the sample at room temperature (15°C 25°C) until it is shipped to the lab.
- Ship via UPS Priority Overnight the day of collection, if possible. Samples must be shipped within 3 days of collection. Ship to maintain sample temperature at -20 to 50°C.

## STABILITY

5 Days

## **REJECTION REASONS**

- There are not two patient identifiers that match the requisition form on the OCD-100 tube.
- Swab is returned outside the OCD-100 tube or biohazard bag.
- Anything other than the swab is inside the OCD-100 tube.
- The OCD-100 tube is not fully sealed.
- Swabs other than the OCD-100 specimen collection kit swab is returned.
- Samples are received at the lab later than 30 calendar days after the date of specimen collection.



# PHARMACOGENOMICS (PGX) TEST DETAILS



### CARDIOVASCULAR - 14 Genes

Gene	Therapeutic Class	Drug Examples
ABCG2	Statins	fluvastatin (Lescol®), rosuvastatin (Crestor®)
CYP2C9	Anticoagulants	warfarin (Coumadin®)
	Angiotensin II Receptor Antagonists	losartan (Cozaar®)
	Diuretics	torsemide (Demadex®)
	Statins	fluvastatin (Lescol®)
CYP2C19	Antiplatelets	clopidogrel (Plavix®)
CYP2D6	Antianginal Agents	ranolazine (Ranexa®)
	Antiarrhythmics	flecainide (Tambocor®), propafenone (Rythmol®)
	Beta Blockers	carvedilol (Coreg®), propranolol (Inderal®)
CYP3A4	Statins	atorvastatin (Lipitor®), simvastatin (Zocor®)
CYP4F2	Anticoagulants	warfarin (Coumadin®)
SLCO1B1	Statins	atorvastatin (Lipitor®), rosuvastatin (Crestor®)
VKORC1	Anticoagulants	warfarin (Coumadin®)
Risk Manag	ement	
APOE	Type III Hyperlipoproteinemia	
F2	Thrombosis	
F5	Thrombosis	

### NEUROLOGY - 3 Genes

Gene	Therapeutic Class	Drug Examples
CYP2C9	Anticonvulsants	fosphenytoin (Cerebryx®), phenytoin (Dilantin®)
CYP2C19	Anticonvulsants	brivaracetam (Briviact®), phenobarbital (Luminal®),
		primidone (Mysoline®)
	Benzodiazepines	clobazam (Onfi®)
	Other Neurological Agents	flibanserin (Addyi®)
CYP2D6	Antidementia Agents Other	donepezil (Aricept®), galantamine (Razadyne®)
	Neurological Agents	tetrabenazine (Xenazine®), valbenazine (Ingrezza®)

#### **Risk Management**

UGT1A1 Gilbert Syndrome

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Gene	Therapeutic Class	Drug Examples
COMT	Opioids	morphine (MS Contin®)
CYP1A2	Muscle Relaxants	tizanidine (Zanaflex®)
CYP2B6	Opioids	methadone (Dolophine®)
CYP2C9	NSAIDs	celecoxib (Celebrex®), flurbiprofen (Ansaid®)
CYP2C19	Muscle Relaxants	carisoprodol (Soma®)
CYP2D6	Opioids	codeine, hydrocodone (Vicodin®), oxycodone (Percocet®),
		tramadol (Ultram®)
OPRD1	Opioids	buprenorphine (Butrans®), methadone (Dolophine®)
OPRK1	Opioids	cocaine vaccine (investigational)
OPRM1	Opioids	fentanyl (Actiq®), hydrocodone (Vicodin®)

#### **PSYCHIATRY** - 14 Genes

Gene	Therapeutic Class	Drug Examples
ADRA2A	Anti-ADHD Agents	dexmethylphenidate (Focalin®), methylphenidate (Ritalin®)
ANKK1	Antiaddictives	bupropion (Wellbutrin®)
COMT	Anti-ADHD Agents	amphetamine (Adderall®), methylphenidate (Ritalin®)
CYP1A2	Antipsychotics	clozapine (Clozaril®), olanzapine (Zyprexa®)
CYP2B6	Antiaddictives	bupropion (Wellbutrin®)
CYP2C19	Antidepressants	amitriptyline (Elavil®), citalopram (Celexa®), sertraline
	Benzodiazepines	diazepam (Valium®)
CYP2D6	Antiaddictives	lofexidine (Lucemyra®)
	Anti-ADHD Agents	atomoxetine (Strattera®)
	Antidepressants	amitriptyline (Elavil®), paroxetine (Paxil®), venlafaxine (Effexor®),
	Antipsychotics	vortioxetine (Trintellix®)
DRD2	Antipsychotics	aripiprazole (Abilify®), haloperidol (Haldol®), thioridazine (Mellaril®)
GRIK4	Antidepressants	risperidone (Risperdal®)
HTR2A	Antidepressants	citalopram (Celexa®)
	Antipsychotics	citalopram (Celexa®)
HTR2C	Antipsychotics	clozapine (Clozaril®)
OPRM1	Antiaddictives	clozapine (Clozaril®), olanzapine (Zyprexa®),
		risperidone naltrexone (Vivitrol®)

#### **Risk Management**

ANKK1 Antipsychotic-Induced Hyperprolactinemia, Tardive Dyskinesia, Weight Gain

MTHFR Hyperhomocysteinemia - Depression

## DIABETES - 3 Genes

Thrombosis

Platelet Reactivity

Hyperhomocysteinemia - Thrombosis

F13A1

ITGB3

MTHFR

Gene	Therapeutic Class	Drug Examples
CYP2C8	Meglitinides	repaglinide (Prandin®)
	Thiazolidinediones	pioglitazone (Actos®), rosiglitazone (Avandia®)
CYP2C9	Meglitinides	nateglinide (Starlix®)
SLCO1B1	Meglitinides	nateglinide (Starlix®), repaglinide (Prandin®)

### **GASTROENTEROLOGY** - 9 Genes

Gene	Therapeutic Class	Drug Examples
ABCB1	Antiemetics	granisetron (Sancuso®), ondansetron (Zofran®)
CYP2C9	Antiemetics	dronabinol (Marinol®)
CYP2C19	Proton Pump Inhibitors	lansoprazole (Prevacid®), omeprazole (Prilosec®)
CYP2D6	Antiemetics	dolasetron (Anzemet®), ondansetron (Zofran®)
G6PD	Other Antirheumatic Agents	sulfasalazine (Azulfine®)
MTHFR	Antifolates	methotrexate (Trexall®)
NUDT15	Thiopurines	azathioprine (Imuran®), mercaptopurine (Purinethol®)
TPMT	Thiopurines	azathioprine (Imuran®), mercaptopurine (Purinethol®)

**Risk Management** 

UGT1A1 Gilbert Syndrome

### HEMATOLOGY / ONCOLOGY - 11 Genes

Gene	Therapeutic Class	Drug Examples
CYP2C8	Taxanes	paclitaxel (Taxol®)
CYP2C9	Protein Kinase Inhibitors	erdaftinib (Balversa®)
CYP2D6	Anti-Estrogens	tamoxifen (Nolvadex®)
	Protein Kinase Inhibitors	gefitinib (Iressa®)
DPYD	Fluoropyrimidines	capecitabine (Xeloda®), fluorouracil (Efudex®)
F2	Hemostatic Agents	avatrombopag (Doptelet®), eltrombopag (Promacta®)
F5	Hemostatic Agents	avatrombopag (Doptelet®), eltrombopag (Promacta®)
G6PD	Detoxifying Agents	rasburicase (Elitek®)
	Protein Kinase Inhibitors	dabrafenib (Tafinlar®)
MTHFR	Antifolates	methotrexate (Trexall®)
NUDT15	Thiopurines	azathioprine (Imuran®), mercaptopurine (Purinethol®)
TPMT	Thiopurines	azathioprine (Imuran®), mercaptopurine (Purinethol®)
UGT1A1	Histone Deacetylase Inhibitors	belinostat (Beleodaq®)
	Protein Kinase Inhibitors	erlotinib (Tarceva®), nilotinib (Tasigna®)
	Topoisomerase Inhibitors	irinotecan (Camptosar®), sacituzumab-govitecan (Trodelvy®)

### **INFECTIOUS DISEASE** - 6 Genes

Gene	Therapeutic Class	Drug Examples
CYP2B6	Anti-HIV Agents	efavirenz (Sustiva®)
CYP2C19	Antifungals	voriconazole (Vfend®)
DPYD	Antifungals	flucytosine (Ancobon®)
G6PD	Antibiotics	nitrofurantoin (Macrobid®, sulfamethoxazole hydroxychloroquine
	Antimalarials	(Plaquenil®), quinine
IFNL3/IFNL4	Interferons	peginterferon alfa-2a (Pegasys®), peginterferon alfa-2b (Pegintron®)
UGT1A1	Anti-HIV Agents	atazanavir (Reyataz®)

\*Infectious Disease report minimum should be CYP2C19 and at least one other