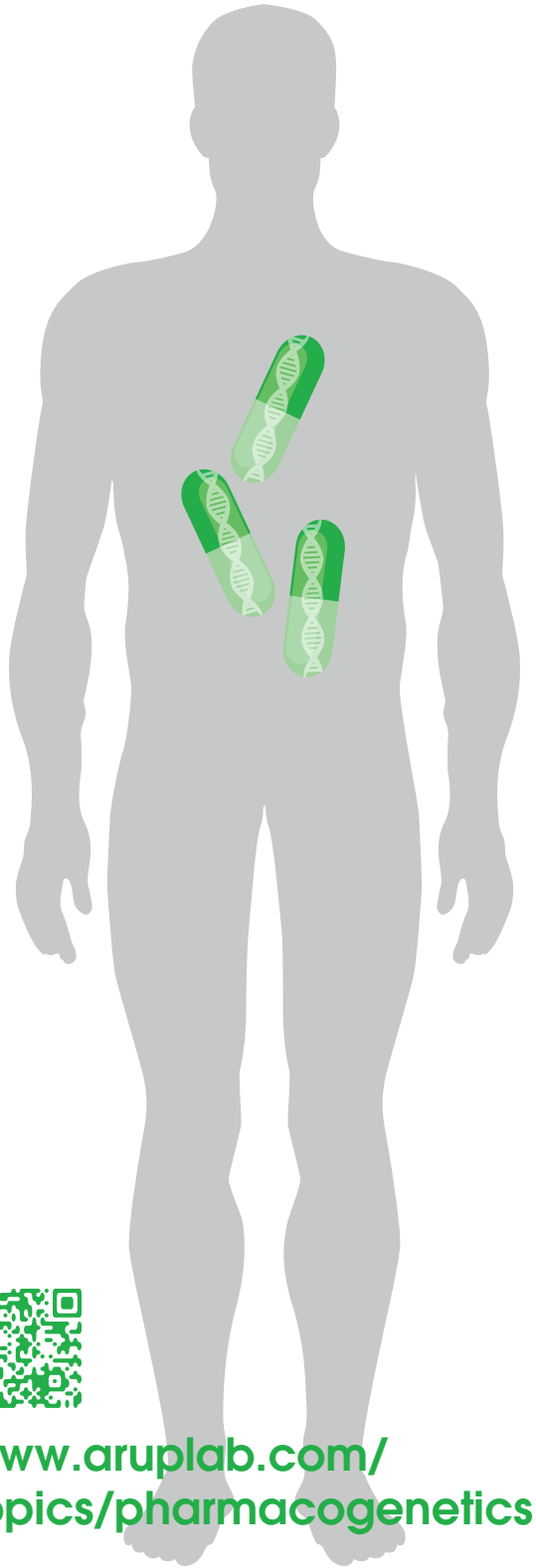

www.aruplab.com/
topics/pharmacogenetics

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1. Agency for Healthcare Research and Quality. Reducing and preventing adverse drug events to decrease hospital costs. archive.ahrq.gov/research/findings/factsheets/errors-safety/aderia/ade.html (accessed August 25, 2016).

ARUP LABORATORIES

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keyword: Phamacogenetics

A nonprofit enterprise of the University of Utah and its Department of Pathology

BD-CS-041, Rev 0, September 2016

Pharmacogenetics

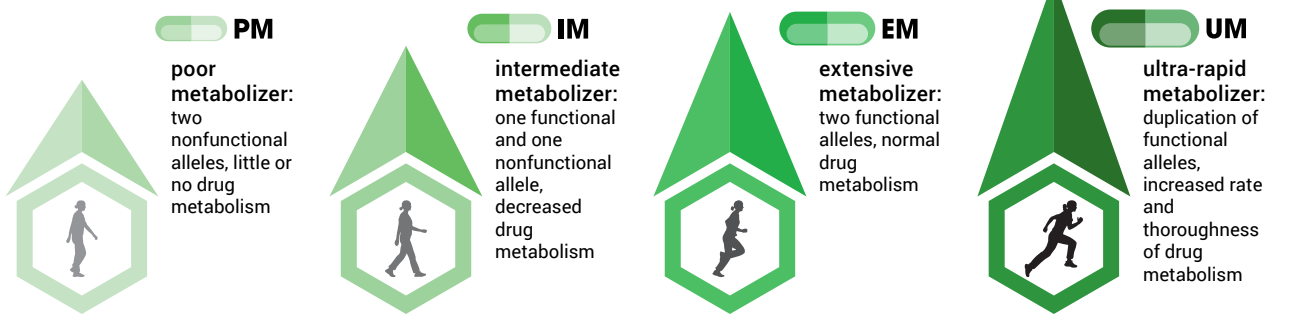
testing at ARUP Laboratories

ARUP LABORATORIES

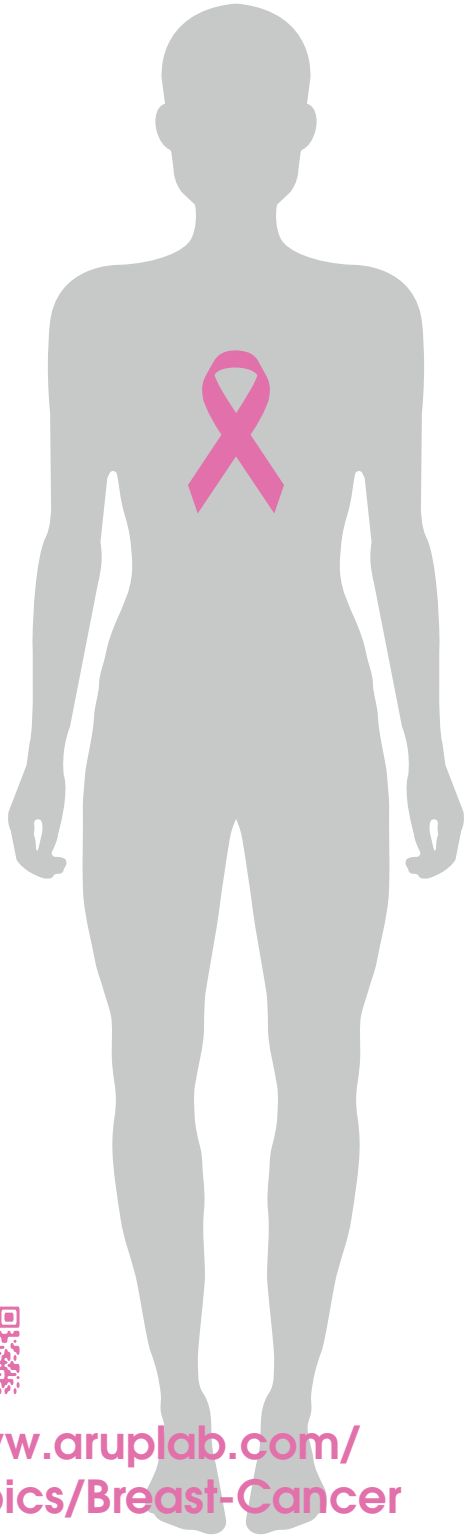
According to the U.S. Department of Health & Human Services, more than **770,000 injuries and deaths due to drug reactions occur each year in the United States. These events may cost a hospital up to $5.6 million each year**. This number does not include the adverse drug events that cause hospital admissions, malpractice and litigation costs, or the costs associated with patient injuries.[1] Pharmacogentic testing provides an opportunity to lower this number.

Pharmacogenetic testing can help save lives and money.

- Prevents many adverse drug events before they occur.
- Reduces pharmacy costs by optimizing dosage.
- Decreases number of patient hospitalizations.
- Improves patient compliance with their drug therapies.
- Becomes a part of a patient's medical record, so it can be referenced in future medical situations when medication metabolized by the same pathway is prescribed.



ARUP TEST CODE AND NAME		ORDERING RECOMMENDATIONS
2013098	Cytochrome P450 Genotype Panel	Assesses genetic risk of abnormal drug metabolism for drugs metabolized by *CYP2D6*, *CYP2C9*, *CYP2C19*, and *CYP3A5*; may aid in drug selection and dose planning for many drugs. This test includes an enhanced report that provides the genotype of the patient and the result interpretation signed off by a medical directors. It also provides a medication summary, which indicates the medications the clinician should avoid with the patient, the medications that could work with some dosage adjustment, and the drugs that are most likely to work according to the genotype. The report is created in conjunction with Coriel Life Sciences; all results are cited.
0051232	Cytochrome P450 2D6 (*CYP2D6*), 14 Variants and Gene Duplication	Assesses genetic risk of abnormal drug metabolism for drugs metabolized by *CYP2D6*.
2012766	Cytochrome P450 2C9 (*CYP2C9*), 2 Variants	Assesses genetic risk of abnormal drug metabolism for drugs metabolized by *CYP2C9*.
2012769	Cytochrome P450 2C19 (*CYP2C19*), 9 Variants	Assesses genetic risk of abnormal drug metabolism for drugs metabolized by *CYP2C19*.
2012740	Cytochrome P450 3A5 Genotyping (*CYP3A5*), 2 Variants	Assesses genetic risk of abnormal drug metabolism for drugs metabolized by *CYP3A5*. All individual *CYP* tests are included in the panel.
2012166	Dihydropyrimidine Dehydrogenase (*DPYD*), 3 Variants	Predicts the risk of dose-related toxicity to 5-Fluorouracil (5-FU), which is the most frequently used chemotherapeutic drug for the treatment of many types of cancer.
2007228	5-Fluorouracil (5-FU) Toxicity and Chemotherapeutic Response, 5 Mutations	Predicts the risk of dose-related toxicity to 5-FU. This test includes the *DPYD* allele and the *TYMS* allele.
2012233	Thiopurine Methyltransferase (*TPMT*) Genotyping, 4 Variants	Assesses genetic risk for severe myelosuppression with standard dosing of thiopurine drugs.
2012772	Warfarin Sensitivity (*CYP2C9* and *VKORC1*), 3 Variants	Identifies individuals with inherited variants that affect metabolism and/or sensitivity to Warfarin.
0080135	Glucose-6-Phosphate Dehydrogenase	Preferred initial screening test for G6PD deficiency.
0051684	Glucose-6-Phosphate Dehydrogenase (*G6PD*) 2 Mutations	Preferred genetic test for individuals of African descent. Detects the single most common pathogenic *G6PD* mutation (the A- allele) in individuals of African descent.
2007163	Glucose-6-Phosphate Dehydrogenase Deficiency (*G6PD*) Sequencing	Preferred genetic test for individuals of high-risk ethnic backgrounds other than those of African descent.
2002429	HLA-B*57:01 for Abacavir Sensitivity	Standard of care prior to treatment with abacavir therapy in HIV-positive patients. Reactions to this drug can get worse with each dose and can be fatal.
2012049	HLA-B*15:02 Genotyping, Carbamazepine Hypersensitivity	Identifies patients who may be at risk for Stevens-Johnson syndrome or toxic epidermal necrolysis prior to treatment with carbamazepine. The symptoms include skin rash, hives, sores in the mouth, blistering or peeling of the skin, and erosion of the mucosal membranes in the respiratory and gastrointestinal tract. Recommended for patients not currently taking carbamazepine.
2008767	Opioid Receptor, Mu 1 (*OPRM1*) Genotyping, 1 Variant	Used for pretheraputic identification of individuals who may require higher or lower doses of opioid drugs to achieve adequate pain control or those who may respond better to naltrexone for the treatment of alcohol and/or opioid dependency.
2008426	Statin Sensitivity (*SLCO1B1*), 1 Variant	Identifies individuals at increased risk for statin-related muscle toxicity.
0051332	UDP Glucuronosyltransferase 1A1 (*UGT1A1*) Genotyping	Useful in dosage planning for individuals who will receive high-dose irinotecan, have a history of irinotecan sensitivity, or experience neutropenia while receiving irinotecan.
2004680	Interleukin 28 B (IL28B) Associated Variants, 2 SNPs	Predict response to peginterferon (PEG-IFNa) and ribavirin (RBV) therapy for chronic HCV-1 infection.



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topics/Breast-Cancer

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2. Breast Cancer Reserach Foundation. Breast cancer in women: know the subtype. www.bcrfcure.org/sites/default/files/blog/breastcancer_lf.png (accessed on August 5, 2015).

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keyword: BREAST CANCER

A nonprofit enterprise of the University of Utah and its Department of Pathology

BD-CS-019, Rev 2, October 2016

Breast Cancer

testing at ARUP Laboratories

ARUP LABORATORIES

One in eight women will be diagnosed with breast cancer in her lifetime.

Proper testing will assist physicians in diagnosing and treating current cancer, predicting reoccurrence, and determining hereditary status in their patients.

Breast Cancer Subtypes

It is important to know the subtype of breast cancer in women for guiding treatment and predicting survival.

About 5% to 10% of all breast cancers are thought to be hereditary.[1] If hereditary breast cancer is suspected, *BRCA1* and *BRCA2* testing is recommended.

Breast Cancer Testing

DIAGNOSTIC, PROGNOSTIC AND PREDICTIVE MARKER TESTING
Circulating Tumor Cell Count **(0093399)**
Cytokeratin 8,18 Low Molecular Weight (CAM 5.2) by Immunohistochemistry **(2003493)**
DNA Cell Cycle Analysis—Ploidy and S-Phase **(0095155)**
E-Cadherin by Immunohistochemistry **(2003869)**
ERBB2 (*HER2*) (HercepTest) by Immunohistochemistry **(2007332)**
ERBB2 (*HER2/neu*) (HercepTest) by Immunohistochemistry, Tissue with Reflex to FISH if 2+ **(0049178)**
ERBB2 (*HER2/neu*) (HercepTest) with Interpretation by Immunohistochemistry, Tissue **(0049174)**
ERBB2 (*HER2/neu*) Gene Amplification by FISH, Tissue **(2008603)**
Estrogen/Progesterone Receptor with Interpretation by Immunohistochemistry **(0049210)**
Estrogen Receptor (ER) by Immunohistochemistry **(2004516)**
HER2/neu Quantitative by ELISA **(2004672)**
Keratin 903 (K903) High Molecular Weight by Immunohistochemistry **(2003978)**
p53 with Interpretation by Immunohistochemistry **(0049250)**
PAX8 by Immunohistochemistry **(2010787)**
PIK3CA Mutation Detection **(2004510)**
Progesterone Receptor (PR) by Immunohistochemistry **(2004525)**

HEREDITARY CANCER TESTING
Ashkenazi Jewish (*BRCA1* and *BRCA2*), 3 Mutations **(2011958)**
Breast and Ovarian Hereditary Cancer Syndrome (*BRCA1* and *BRCA2*) Sequencing and Deletion/Duplication **(2011949)**
Breast and Ovarian Hereditary Cancer Syndrome (*BRCA1* and *BRCA2*) Deletion/Duplication **(2011915)**
Breast and Ovarian Hereditary Cancer Syndrome (*BRCA1* and *BRCA2*) Sequencing **(2011954)**
Breast and Ovarian Hereditary Cancer Panel, Sequencing and Deletion/Duplication, 20 Genes **(2012026)**
Cancer Panel, Hereditary, Deletion/Duplication, 46 Genes **(2010757)**
Cancer Panel, Hereditary, Sequencing and Deletion/Duplication, 47 Genes **(2012032)**
Familial Mutation, Targeted Sequencing **(2001961)**
PHARMACOGENETIC TESTING
Cytochrome P450 2D6 (*CYP2D6*), 14 Variants and Gene Duplication **(0051232)**
Cytochrome P450 2C9, *CYP2C9*, 2 Variants **(2012766)**
Cytochrome P450 3A5 Genotyping, *CYP3A5*, 2 Variants **(2012740)**
Cytochrome P450 Genotype Panel **(2013098)**
Opioid Receptor, Mu (*OPRM1*) Genotype, 1 Variant **(2008767)**
RISK OF RECURRENCE TESTING
Prosigna Breast Cancer Prognostic Gene Signature **(2010248)**

Hormone Receptor HR+/HER2-

Typically treated with hormone receptor blockade

Diagnostic testing

Prognostic and predictive testing

Pharmacogenetic testing

Risk of recurrence

70%
of all breast cancer cases

HR-/HER2-

Hormone receptor blockade and anti-HER2 targeted therapy are not effective

Diagnostic testing

Prognostic and predictive testing

Pharmacogenetic

Hereditary testing

15%
of all breast cancer cases

HR+/HER2+ HR-/HER2+

Treated with anti-HER2 targeted therapy

If HR+, also treated with hormone receptor blockade

Diagnostic testing

Prognostic and predictive testing

Pharmocogenetic testing

15%
of all breast cancer cases