Illumina, Inc.
5200 Illumina Way
San Diego, CA 92122 USA
tel 858.202.4500
fax 858.202.4766
www.illumina.com

Pharmacogenomics Screen

Description

Test Indication

The Pharmacogenomics Screen is included as part of the offerings for the TruGenome™ Predisposition Screen and the TruGenome Undiagnosed Disease Test, and is not offered as a standalone test. The Pharmacogenomics Screen is intended to provide information to physicians regarding well-established relationships between genetic variants and drug response, but is not intended to determine whether a drug should or should not be administered, or what dosage to prescribe. The genomic information provided in the offering may help physicians make informed management decisions regarding drug administration, but final decisions are at the discretion of the ordering physician. The analysis and interpretation is designed to detect and report on 34 genomic positions, encompassing 11 genes and 16 drugs (as established using the Clinical Pharmacogenetics Implementation Consortium (CPIC) Database or FDA Guidelines). Please see Table 1 below for a summary of genes, drugs, and the number of positions covered in the panel. The pharmacogenomics report is included as a separate document from the TruGenome Predisposition Screen and Undiagnosed Disease Test clinical reports.

Table1: Summary of TruGenome Pharmacogenomics Screen Test

Drug	Gene	Positions Tested
Ivacaftor	CFTR	10
Rebetron	IFNL3	1
Fluoropyrimidines	DPYD	3
Clopidogrel	CYP2C19	8
Azathioprine, mercaptopurine, thioguanine	TPMT	4
Warfarin	CYP2C9, VKORC1	3 (2 CYP2C9 & 1 VKORC1)
Simvastatin	SLCO1B1	1
Abacavir	HLA-B (HCP5)	1
Amitriptyline and nortriptyline	CYP2C19	3
Boceprevir, telaprevir	IFNL3	1
Valproic Acid	POLG	2
Eltrombopag	F5	1

Test Method

Whole-genome sequencing is performed for this test utilizing Illumina Sequencing-By-Synthesis (SBS) chemistry and paired-end read technology. Alignment and variant identification is performed with NCBI Human Genome Reference build 37.1. Only the 34 selected genomic positions are interpreted and reported for this offering. Resources including the CPIC guidelines, FDA recommendations, and peer-reviewed literature are used in the interpretation of these pharmacogenomic findings.

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Test Specifications

Refer to Intended Use Statement documents for TruGenome Predisposition Screen and TruGenome Undiagnosed Disease Test.

Deliverables for Pharamacogenomics Screen

A separate pharmacogenomics report is included for individuals ordering the TruGenome Predisposition Screen and Undiagnosed Disease Test. This report includes detailed information and references regarding the drugs, genes and genomic positions interpreted for the test.

Criteria for variant classification

Interpretation and classification of variants based on patient genotype and/or diplotype are based on recommendations provided through the CPIC guidelines, FDA recommendations, and the peer-reviewed scientific literature.

Limitations

Refer to Intended Use Statement documents for TruGenome Predisposition Screen and TruGenome Undiagnosed Disease Test.

Contact

Please contact the lab with any questions you may have regarding test selection, ordering, sample submission and results interpretation and implications.

Phone: 858.736.8080

Fax: 858.255.5285

EveryGenome@Illumina.com