Seraseq® Pharmacogenomics DNA Mix

Assay validation and daily-run QC material for testing various hereditary alleles of pharmacogenes involved in differential sensitivity to therapeutics

INTRODUCTION

Pharmacogenomics is revolutionizing the way medications are prescribed, enabling the development of personalized treatment plans tailored to an individual's genetic makeup. Reference materials are critical in pharmacogenomics, as they provide standardized benchmarks for validating and ensuring the accuracy of genetic tests.

Seraseq® reference materials help laboratories and researchers verify that their assays can accurately detect genetic variations that affect drug metabolism and response. Without reliable reference materials, the risk of inaccurate test results increases, potentially leading to improper treatment decisions. This is why high-quality reference materials are essential for advancing personalized medicine and ensuring patient safety in clinical settings.

The Seraseq® Pharmacogenomics DNA Mix provides a standardized means to evaluate and validate the performance of genetic tests. It offers:

- An invaluable alternative to hard-to-source clinical samples, providing labs with consistent, reliable, and high-quality controls to simulate real-world genomic complexities, including structural variations and mutations in challenging regions.
- Robust quality control, enabling consistent and reliable assay results.
- Regulatory compliance and accreditation requirements by demonstrating test accuracy and reliability.

By integrating this high-quality reference material into their workflows, clinical laboratories can ensure the robustness of their assays and improve confidence in their genetic test results.

FEATURES

- Targeted for Pharmacogenomics Genetic Testing: Contains variants in 13 genes linked to adverse reaction to specific pharmacological therapies.
- Comprehensive Variant Coverage: Includes 79 clinically-relevant mutations, all classified as pathogenic or likely pathogenic according to AMP recommendations, including a mix of AMP Tier 1 and hard-to-source Tier 2 variants.
- Expertly Designed: Crafted to mimic challenging genomic regions, providing a robust tool for clinical laboratories working on next-generation sequencing (NGS) assays for inherited pharmacological sensitivity.
- High Variant Diversity: Features a wide variety of mutations, including single nucleotide variants (SNVs), indels, and duplications.
- Single-Vial Convenience: Simplify workflows with a highly multiplexed reference material packaged in a single vial, reducing costs and saving time while boosting QC robustness.

HIGHLIGHTS

High-quality third-party QC material in mutation mix format to develop, validate, monitor, and troubleshoot your assay.

Convenient highly multiplexed material in a single vial format to save time, cost, and increase QC robustness.

Designed to support compliance with regulatory standards like CAP and CLIA ensures your lab meets performance, quality, and reporting requirements effortlessly.

ORDERING INFORMATION

Material #	Product	Concentration*	Fill Size	Total Mass
0750-9503	Seraseq® Pharmacogenomics DNA Mix	30 ng/µL	1 vial x 20µL	600 ng

* Reported concentration values are based on the Qubit dsDNA BR Assay.

Not for In Vitro Diagnostic Use. Research Use Only.

MUTATIONS PRESENT IN THE SERASEQ® PHARMACOGENOMICS DNA

Gene	Nucleotide change	Type of nucleotide alteration	Molecular Consequence	AMP Tier	Star allele	Clinical Significance NIH - PharmGKB (CPIC & DPWG*)
CYP2B6	c.983T>C	SNV	Missense variant	—	*18	Drug response - No function
	c.785A>G	SNV	Missense variant	—	*4 (*6, 87, *13)	Drug response - Increased function
	c.516G>T	SNV	Missense variant	—	*9 (*6)	Drug response - Decreased function
CYP2C cluster	g.96405502G>A	SNV	Missense variant	2	N/A	Drug response
CYP2C19	c.-806C>T	SNP	Missense initiator codon variant - Synonymous	1	*17	Drug response - No function
	c.1A>G	SNV	Intron variant	2	*4B	Drug response - No function
	c.332-23A>G	SNV	Missense variant	2	*35	Drug response - No function
	c.358T>C	SNV	Missense variant	2	*8	Drug response - No function
	c.395G>A	SNV	Missense variant	2	*6	Drug response - Decreased function
	c.431G>A	SNP	Nonsense. Stop Codon	2	*9	Drug response - No function
	c.636G>A	SNV	Missense variant	1	*3	Drug response - Decreased function
	c.680C>T	SNV	Synonymous variant	2	*10	Drug response - No function
	c.681G>A	SNV	2KB Upstream Variant	1	*2	Drug response - Increased function
	c.819+2T>A	SNV	Splice donor	2	*7	Drug response - No function
	c.1297C>T	SNV	Missense variant	2	*5	Drug response - No function
CYP2C9	c.269T>C	SNV	Missense variant	2	*13	N/A - DPWG: Decreased function; CPIC: No function
	c.430C>T	SNV	Synonymous variant	1	*2	No classification - Decreased function
	c.449G>A	SNV	Missense variant	1	*8	Benign/Likely benign; Drug response - Decreased function
	c.485C>A	SNV	Missense. Stop gained	2	*15	N/A - No function
	c.818delA	Indel	Frameshift variant	1	*6	Benign/Likely benign; drug response; other
	c.1003C>T	SNV	Missense variant	1	*11	Drug Response - Decreased function
	c.1075A>C	SNV	Missense in exon 7	1	*3	Likely benign; Drug response - DPWG: Decreased function; CPIC: No function

MUTATIONS PRESENT IN THE SERASEQ® PHARMACOGENOMICS DNA continued

Gene	Nucleotide change	Type of nucleotide alteration	Molecular Consequence	AMP Tier	Star allele	Clinical Significance NIH - PharmGKB (CPIC & DPWG*)
CYP2C9	c.1080C>G	SNV	Missense variant	1	*5	Benign/Likely benign; drug response; other - Decreased function
	c.1465C>T	SNV	Missense variant	2	*12	N/A - Decreased function
CYP2D6	c.100C>T	SNV	Missense variant	1	*10	Likely benign; drug response - Decreased function
	c.124G>A	SNV	Missense variant	2	*12	Likely benign - No function
	c.137dup	Duplication	Frameshift	2	*15	Likely benign - No function
	c.320C>T	SNV	Missense variant	1	*17	Likely benign; other - No function
	c.358T>A	SNV	Missense variant	2	*49	Drug Response - Decreased function
	c.406G>A	SNV	Intronic, missense	1	*29 (*107, *149)	N/A - Decreased function
	c.454del	Deletion	Frameshift	1	*6	Benign/Likely benign; drug response - No function
	c.505G>T	SNV	Nonsense	2	*8	Likely benign - No function
	c.505G>A	SNV	Nonsense. Intronic	2	*14	Likely benign - Decreased function
	c.506-1G>A	SNV	Splice acceptor	1	*4	Likely benign; drug response; other - No function
	c.514_522dup	Indel	In frame Insertion	2	*40	Not Available - No function
	c.775del	Indel	Frameshift	1	*3	Likely benign; other - No function
	c.805dup	Indel	Frameshift	2	*21	N/A - No function
	c.841_843del	Indel	In frame Insertion	1	*9	Likely benign - Decreased function
	c.886C>T	SNV	Missense variant	1	*17	Decreased function
	c.971A>C	SNV	Missense variant	2	*7	Likely benign - No function
	c.975G>A	SNV	Synonymous	2	*59	N/A - Decreased function
	c.985+39G>A	SNV	Intron variant	1	*41	Likely benign; drug response - Decreased function
	c.1012G>A	SNV	Missense variant	1	*29	Likely benign - Decreased function
	c.1030C>T	SNP	Stop Codon	2	*56	N/A - No function
	c.1088_1089dup	Indel	Frameshift	2	*42	N/A - No function
	c.1319G>A	SNV	Missense variant	2	*31	Drug Response - No function
	c.1457G>C	SNV	No sequence alteration	1	*2	Benign/Likely benign - Normal function

MUTATIONS PRESENT IN THE SERASEQ® PHARMACOGENOMICS DNA continued

Gene	Nucleotide change	Type of nucleotide alteration	Molecular Consequence	AMP Tier	Star allele	Clinical Significance NIH - PharmGKB (CPIC & DPWG*)
CYP3A4	c.522-191C>T	SNV	Intronic polymorphism	1	*22	N/A - Decreased function
	c.1461dup	Duplication	frameshift	2	*20	Likely benign - No function
CYP3A5	c.219-237A>G	SNV	Intron , Splice Acceptor. 3 Flanking	1	*3	Association; drug response; risk factor - No function
	c.624G>A	SNV	Synonymous Variant	1	*6	N/A - No function
	c.1035dup	Insertion	Frameshift	1	*7	N/A - No function
CYP4F2	c.1297G>A	SNP	Missense variant	2	*3	Drug response
DPYD	c.1156G>T	SNV	Nonsense - Exon 11	—	*12	Drug Response - No function
	c.1236G>A	SNV	Synonymous - Exon 11	1	HapB3	Drug Response - Decreased function
	c.1314T>G	SNV	Missense - Exon 11	2	N/A	Drug Response - Decreased function
	c.1601G>A	SNV	Missense - Exon 14	—	*4	Drug Response - Normal function
	c.1627A>G	SNV	Missense - Exon 12	—	*5	Drug Response - Normal function
	c.1679T>G	SNV	Missense - Exon 13	1	*13	Drug Response - No function
	c.1775G>A	SNV	Missense - Exon 14	—	N/A	Uncertain significance - No function
	c.1898delC	Indel	Frameshift - Exon 14	—	*3	Drug Response - No function
	c.1905+1G>A	SNV	Splice donor causing Exon 14 skipping	1	*2A	Drug Response - No function
	c.2846A>T	SNV	Missense - Exon 22	1	N/A	Drug Response - Decreased function
	c.2872A>G	SNV	Missense - Exon 22	—	N/A	Uncertain significance - No function
NUDT15	c.415C>T	SNV	Missense	1	*3	Drug Response - No function
	c.416G>A	SNV	Missense	2	*4	Drug Response - DPWG: Decreased function; CPIC: Uncertain function
	c.50delGAGTCG	In frame insertion	Microsatellite	2	*9	Drug Response - DPWG: Decreased function; CPIC: No function
	c.80_81insCGGG	Indel	Frameshift	2	*14	-
TPMT	c.2T>G	SNV	Initiator Codon Variant	—	N/A	N/A - No function
	c.95dup	Indel	Frameshift	2	*42	Drug response - N/A
	c.238G>C	SNV	Missense	1	*2	Drug Response - No function

MUTATIONS PRESENT IN THE SERASEQ® PHARMACOGENOMICS DNA continued

Gene	Nucleotide change	Type of nucleotide alteration	Molecular Consequence	AMP Tier	Star allele	Clinical Significance NIH - PharmGKB (CPIC & DPWG*)
TPMT	c.395G>A	SNV	Missense	2	*11	N/A - No function
	c.460G>A	SNV	Missense	1	*3A	Drug Response - No function
	c.719A>G	SNV	Missense	1	*3C	Drug Response - No function
UGT1A1	c.862-6800AT[8]	Insertion	Microsatellite	—	*28	Drug response
	c.862-6536G>A	SNV	Missense intronic variant	—	*6	Drug response
VKORC1	c.-1639G>A	SNP	2KB Upstream. 5 Flanking	1	N/A	Drug Response - Higher coumarin sensitivity
	c.106G>T	SNV	Missense	2	N/A	Drug Response
	c.196G>A	SNV	Missense Intronic	2	N/A	Drug response

ABOUT US

SeraCare offers a comprehensive portfolio of reference materials for oncology and reproductive health, designed and manufactured to meet the precision demanded by NGS assays. The portfolio includes high quality ground-truth RNA, ctDNA and genomic DNA-based reference materials that are NGS platform agnostic for tumor profiling, immuno-oncology, liquid biopsy, NIPT and germline cancer assay workflows. **For more information visit seracare.com**

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