Seraseq®

Pharmacogenomics DNA Mix

Package Insert

NAME AND INTENDED USE

The Seraseq® Pharmacogenomics DNA Mix is a reference material intended for use in the development, validation, and evaluation of routine performance of Next Generation Sequencing (NGS) (and other amplified nucleic acid-based methods) assays that identify inherited (germline) variants in gene alleles associated with adverse reactions to certain medications or treatments. This reference material is suitable for use by clinical laboratories, research institutions, and diagnostic assay developers to ensure consistent and reliable results across different sequencing runs and laboratory conditions.

The Seraseq Pharmacogenomics DNA Mix is not intended for use in patient diagnosis, treatment, or in any therapeutic procedures. It is intended for use by trained laboratory personnel proficient in NGS technologies and familiar with proper laboratory practices and quality control procedures.
For Research Use Only (RUO). Not for use in diagnostic procedures.

REAGENT PROVIDED

Seraseq Pharmacogenomics DNA Mix is a mixture of synthetic DNA constructs and genomic DNA extracted from the human cell line GM24385. It contains 79 clinically relevant biosynthetic DNA variants from 13 different genes (not including those present in the GM24385 background) (Table 2). The product is formulated to simulate a heterozygous state for each mutation at a 50% variant allele frequency (VAF) measured by droplet digital PCR and confirmed by NGS.

Table 1. Seraseq Pharmacogenomics DNA Mix

Material No.	Product	Format
0750-9503	Seraseq® Pharmacogenomics DNA Mix	1x 600 ng

One (1) vial, 600 ng per vial, at a nominal concentration of 30 ng/ µL is provided. Refer to the batch-specific Technical Product Report for exact concentration. Manufactured in the USA.

WARNINGS AND PRECAUTIONS

Safety and Handling Precautions

Handle Seraseq Pharmacogenomics DNA Mix and all materials derived from human blood products as though it is capable of transmitting infectious agents. Use Centers for Disease Control and Prevention (CDC) recommended universal precautions for handling reference materials and human specimens[1]. Do not pipette by mouth; do not smoke, eat, or drink in areas where specimens are being handled. Clean any spillage by immediately wiping up with 0.5% sodium hypochlorite solution. Avoid contamination of the product when opening and closing the vials. Dispose of all specimens and materials appropriately.

STORAGE INSTRUCTIONS

Store Seraseq Pharmacogenomics DNA Mix frozen between -30°C to -10°C. Once opened, a vial can be thawed and re-frozen up to five (5) times. Sub-aliquoting of the product into low DNA binding tubes may be advisable to limit the number of freeze/thaw cycles to five (5) or less.

INDICATIONS OF REAGENT INSTABILITY OR DETERIORATION

Seraseq Pharmacogenomics DNA Mix should appear as a clear liquid. Alterations in this appearance may indicate instability or deterioration of the product and vials should be discarded.

PROCEDURE

Materials Required but not Provided

Refer to instructions supplied by manufacturers of the test kits to be used.

Instructions for Use

Allow the product vial to come to room temperature before use. Mix by vortexing to ensure a homogeneous solution and spin briefly. Seraseq Pharmacogenomics DNA Mix should be integrated into library preparation after the DNA isolation step; no further purification or DNA isolation is needed. If a DNA shearing step is part of the workflow, the reference material should be sheared and go through the target selection and library preparation in parallel with test specimens. Refer to standard assay procedures in order to determine the amount of material to use.

EXPECTED RESULTS & INTERPRETATION OF RESULTS

Table 2 indicates each of the mutations represented in the Seraseq Pharmacogenomics DNA Mix. While the presence and frequency of each variant in this product was confirmed during manufacture using digital PCR assays and NGS, there may be differences in observed allele frequencies due to assay characteristics.

NOTE: The Seraseq Pharmacogenomics DNA Mix does not have assigned values for allele frequencies of the variants present. Furthermore, specific detection of variants and variant allele frequencies within the product will vary among different assays, different procedures, different lot numbers, and different laboratories. *Each laboratory must establish an assay-specific expected value and acceptance range for each variant and lot of the Mutation Mix prior to its routine use*. When results for the product are outside of the established acceptance range, it may indicate unsatisfactory test performance. Possible sources of error include: deterioration of test kit reagents, operator error, faulty performance of equipment, contamination of reagents or change in bioinformatics pipeline parameters.

LIMITATIONS OF THE PROCEDURE

SERASEQ PHARMACOGENOMICS DNA MIX MUST NOT BE SUBSTITUTED FOR THE MANDATORY POSITIVE AND NEGATIVE CONTROL REAGENTS PROVIDED WITH MANUFACTURED TEST KITS.

TEST PROCEDURES and *INTERPRETATION OF RESULTS* provided by manufacturers of test kits must be followed closely. Deviations from procedures recommended by test kit manufacturers may produce unreliable results. Data are provided for informational purposes. LGC Clinical Diagnostics does not claim that others can duplicate test results exactly. Note that based on your particular assay and analysis parameters, a different fetal fraction value may be calculated. Seraseq Pharmacogenomics DNA Mix is not a calibrator and should not be used for assay calibration. Adverse shipping and storage conditions or use of outdated products may produce erroneous results.

REFERENCES

1. Siegel JD, Rhinehart E, Jackson M, Chiarello L, and the Healthcare Infection Control Practices Advisory Committee, 2007 Guideline for Isolation Precautions: Preventing Transmission of Infectious Agents in Healthcare Settings.

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15047-01 August 2025

Table 2. List of Mutations

Gene	Nucleotide Change	Protein Change	Star Allele	dbSNP NIH
CYP2B6	c.983T>C	Ile328Thr (I328T)	*18	rs28399499
	c.785A>G	K262R	*4 (*6, 87, *13)	rs2279343
	c.516G>T	Q172H, splice defect	*9 (*6)	rs3745274
CYP2C cluster	g.96405502G>A	N/A	N/A	rs12777823
CYP2C19	c.1A>G	M1V	*4B	rs28399504
	c.332-23A>G	Splice defect	*35	rs12769205
	c.358T>C	W120R	*8	rs41291556
	c.395G>A	R132Q	*6	rs72552267
	c.431G>A	R144H	*9	rs17884712
	c.636G>A	W212X	*3	rs4986893
	c.680C>T	P227L	*10	rs6413438
	c.681G>A	Splice defect	*2	rs4244285
	c.-806C>T	expression	*17	rs12248560
	c.819+2T>A	Splice defect	*7	rs72558186
	c.1297C>T	R433W	*5	rs56337013
CYP2C9	c.269T>C	L90P	*13	rs72558187
	c.430C>T	R144C	*2	rs1799853
	c.449G>A	R150H	*8	rs7900194
	c.485C>A	S162X	*15	rs72558190
	c.818delA	K273fs	*6	rs9332131
	c.1003C>T	R335W	*11	rs28371685
	c.1075A>C	I359L	*3	rs1057910
	c.1080C>G	D360E	*5	rs28371686
	c.1465C>T	P489S	*12	rs9332239
CYP2D6	c.100C>T	P34S	*10	rs1065852
	c.124G>A	G42R	*12	rs5030862
	c.137dup	L47fs	*15	rs774671100
	c.320C>T	T107I	*17	rs28371706
	c.358T>A	F120I	*49	rs1135822
	c.406G>A	V136M	*29 (*107,*149)	rs61736512
	c.454del	W152fs	*6	rs5030655
	c.505G>T	G169X	*8	rs5030865
	c.505G>A	G169*	*14	rs5030865
	c.506-1G>A	splice defect	*4	rs3892097
	c.514_522dup	174_175insFRP	*40	rs72549356
	c.775del	R259fs	*3	rs35742686
	c.805dup	R269fs	*21	rs72549352
	c.841_843del	K281del	*9	rs5030656
	c.886C>T	R296C	*2, *8,*11, *12,*14, *17	rs16947

Gene	Nucleotide Change	Protein Change	Star Allele	dbSNP NIH
	c.971A>C	H324P	*7	rs5030867
	c.975G>A	splice defect	*59	rs79292917
	c.985+39G>A	splice defect	*41	:rs28371725
	c.1012G>A	V338M	*29	rs59421388
	c.1030C>T	R344X	*56	rs72549347
	c.1088_1089dup	Q364fs	*42	rs72549346
	c.1319G>A	R440H	*31	rs267608319
	c.1457G>C	S486T	*2	rs1135840
CYP3A4	c.522-191C>T	splice defect	*22	rs35599367
	c.1461dup	P487fs	*20	rs67666821
CYP3A5	c.219-237A>G	splice defect	*3	rs776746
	c.624G>A	splice defect	*6	rs10264272
	c.1035_1036insT	T346fs	*7	rs41303343
CYP4F2	c.1297G>A	V433M	*3	rs2108622
DPYD	c.1156G>T	p.E386*	*12	rs78060119
	c.1236G>A	p.E412E	HapB3	rs56038477
	c.1314T>G	p.F438L	N/A	rs186169810
	c.1601G>A	p.S534N	*4	rs1801158
	c.1627A>G	p.I543V	*5	rs1801159
	c.1679T>G	p.I560S	*13	rs55886062
	c.1775G>A	p.R592Q	N/A	rs138616379
	c.1898delC	p.P633fs	3*	rs72549303
	c.1905+1G>A	p.IVS14	*2A	rs3918290
	c.2846A>T	p.D949V	N/A	rs67376798
	c.2872A>G	p.K958E	N/A	rs141044036
NUDT15	c.415C>T	R139C	*3	rs116855232
	c.416G>A	R139H	*4	rs147390019
	c.50delGAGTCG	del17_18GV	*9	rs746071566
	c.80_81insCGGG	C28fs	*14	rs777311140
TPMT	c.2T>G	M1W	*29	rs267607275
	c.95dup	W33fs	*42	rs759836180
	c.238G>C	A80P	*2	rs1800462
	c.395G>A	C132T	*11	rs72552738
	c.460G>A	A154T	*3A	rs1800460
	c.719A>G	Y240S	*3C	rs1142345
UGT1A1	c.862-6800AT[8]	Splice defect	*28	rs3064744
	c.862-6536G>A	G71R	*6	rs4148323
VKORC1	c.-1639G>A	N/A	N/A	rs9923231
	c.106G>T	D36Y	N/A	rs61742245
	c.196G>A	V66M	N/A	rs72547529

NOTE: Above list does not include variants present in the GM24385 background. Target variant allele frequency is 50%. Substitution refers to a single nucleotide variant; Indel is defined as a deletion/insertion less than 10 base pairs.

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