

# Technical Product Report

*For Research Use Only; Not for use in Diagnostic Procedures*

Product Description: Seraseq® Carrier Screening DNA Mix  
Material Number: 0730-0569 Batch Number: 10709655  
Material Description: A genomic DNA mutation mix containing deletions, insertions, and single nucleotide variants blended with GM24385 DNA as reference wild-type background  
Date of Manufacture: 25 JUN 2024 Expiration Date: 25 JUN 2028  
Fill Volume: 20 µL  
Nominal Concentration: 30 ng/µL  
Concentration (Qubit dsDNA BR Assay): 35.1 ng/µL  
dPCR Test Method: Bio-Rad QX200 Droplet Digital™ System  
NGS Assay Test Method: Whole genome sequencing using an S4 flow cell and Illumina DNA PCR Free Tagmentation Kit sequenced on Illumina NovaSeq 6000. 300 ng of DNA mix was used as input. The average read depth was 31.1X.

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Gene	Nucleotide Change	Protein Change	% Allele Frequency (dPCR)	% Allele Frequency (NGS)
ABCC8	c.3989-9G>A	Splice Variant	44.50	35.14
ACADM	c.136_158dup	p.Phe55fs	44.27	69.77
ACADVL	c.848T>C	p.Val283Ala	47.37	38.60
AGXT	c.823_824dup	p.Ser275fs	44.43	38.24
ASL	c.532G>A	p.Val178Met	42.63	40.00
ASPA	c.437_449del	p.Ser146fs	44.10	52.46
ATP7B	c.2009_2015del	p.Ile669_Tyr670insTer	44.77	34.04
BBS2	c.256_278dup	p.Val94fs	44.00	47.83
BCKDHB	c.93_103del	p.Ala32fs	44.30	32.43
BLM	c.839_888del	p.Glu279_Leu280insTer	44.07	32.50
CBS	c.833T>C	p.Ile278Thr	43.27	57.69
CFTR	c.1521_1523del	p.Phe508del	45.77	46.55
CFTR	c.1624G>T	p.Gly542Ter	41.50	38.81
CFTR	c.1973_1985delinsAGAAA	p.Arg658fs	42.67	45.55
CLRN1	c.300T>G	p.Tyr100Ter	45.10	57.41
CPT2	c.338C>T	p.Ser113Leu	46.30	42.55
CYP21A2	c.332_339del	p.Gly111fs	44.97	29.73
DHCR7	c.964-1G>T	Splice Variant	45.80	46.67
DLD	c.685G>T	p.Gly229Cys	45.63	43.33
DMD	c.9568C>T	p.Arg3190Ter	49.67	48.28
ELP1	c.2204+6T>C	Splice Variant	46.53	48.28
ERCC2	c.854_866del	p.Tyr285fs	44.40	40.74
FAH	c.1062+5G>A	Splice Variant	45.63	29.82
FANCC	c.456+4A>T	Splice Variant	45.23	50.77
FKTN	c.1167dup	p.Phe390fs	43.23	43.18
FMR1	c.52-1_52delinsTA	Splice Variant	50.90	NC
G6PC	c.379_380dup	p.Tyr128fs	44.33	45.83
GAA	c.-32-13T>G	Splice Variant	46.03	45.45
GALT	c.-119_-116del	Splice Variant	47.73	42.00
GBA1	c.1265_1319del	p.Leu422fs	47.83	ND
GJB2	c.35del	p.Gly12fs	46.73	37.78
HBA1	c.283_300+3dup	Splice Variant	47.07	52.63
HBA2	c.427T>C	p.Ter143Gln	46.67	44.00
HBB	c.20A>T	p.Glu7Val	47.47	37.50

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HEXA	c.1274_1277dup	p.Tyr427fs	46.60	42.11
IDUA	c.1525-11_1536del	Splice Variant	46.57	54.55
MCOLN1	c.406-2A>G	Splice Variant	47.80	25.93
MMACHC	c.271dup	p.Arg91fs	46.27	42.00
NEB	c.3255+1G>A	Splice Variant	46.20	53.85
NPHS1	c.3250dup	p.Val1084fs	45.47	37.14
OTC	c.72_77+18del	Splice Variant	54.97	NC
PAH	c.707-12_711del	Splice Variant	51.93	29.82
PCDH15	c.733C>T	p.Arg245Ter	47.93	34.88
PKHD1	c.3463_3464dup	p.Gln1155fs	46.80	32.50
PMM2	c.422G>A	p.Arg141His	46.50	43.14
SLC26A4	c.365dup	p.Ile124fs	46.27	37.50
SMN1	c.399_402del	p.Glu134fs	45.87	26.32
SMN1	c.770_780dup	p.Gly261fs	47.50	29.17
SMN1	c.835-21_*3+17del	Splice Variant	40.01	ND
SMN1	c.*3+80T>G	Splice Variant	40.85	42.86
SMN1	c.*239G>A	Splice Variant	40.85	ND
SMPD1	c.996del	p.Phe333fs	46.03	25.58
TMEM216	c.228dup	p.Gly77fs	45.87	34.38
XPC	c.1643_1644del	p.Val548fs	46.07	46.67

ND = not detected

NC = detected but no VAF call

Approval:

*Alpha Higgs*

Prepared By

27 JUN 2024

Date