

Certificate of Analysis

Product Description: Seraseq® Inherited Cancer DNA Mix v2

Material Number: 0730-0069 Batch Number: 10711165

Material Description: Mixture of purified DNA constructs of variants associated with inherited cancers and genomic DNA from GM24385 cells

Fill Volume: 20 µL

Date of Manufacture: 29 AUG 2024 Expiration Date: 03 JUL 2028

Storage: -20 °C

Concentration (Invitrogen™ Qubit™ dsDNA BR Assay): Nominal value: 25 ng/µL
Average measured value: 30.2 ng/µL

dPCR Test Method: Bio-Rad QX200™ Droplet Digital PCR (ddPCR™) System

NGS Assay Test Method: Axen Hereditary Cancer Panel v2

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| Gene | Nucleotide change | Protein change | AF% dPCR | AF% NGS |
|--------|---|----------------------|----------|---------|
| APC | c.4348C>T | p.R1450* | 44.9 | 42.86 |
| APC | c.4666dup | p.T1556fs | 45.4 | 43.97 |
| ATM | c.640del | p.S214fs | 43.13 | 42.39 |
| AXIN2 | c.1994dup | p.N666fs | 46.6 | 44.13 |
| BAP1 | c.1433_1449dup | p.P484fs | 42.87 | 35.38 |
| BARD1 | c.1600_1634delinsGCG | p.T546fs | 46.77 | 42.65 |
| BMPR1A | c.1131_1153dup | p.V385fs | 46.0 | 33.33 |
| BRCA1 | c.3481_3491del | p.E1161fs | 42.43 | 28.81 |
| BRCA2 | c.1813dup | p.I605fs | 42.27 | 23.16 |
| BRCA2 | c.8954-8_9136del | Deletion | 41.47 | ND |
| BRCA2 | c.9342_9343insAluY | N/A | 42.83 | ND |
| BRIP1 | c.2392C>T | p.R798* | 40.97 | 44.23 |
| CDH1 | c.2037_2061dup | p.C688delinsHLRGQRV* | 42.67 | 38.89 |
| CDK4 | c.70C>T | p.R24C | 43.70 | 41.32 |
| CDKN1B | c.59_77dup | p.S27fs | 42.77 | NT |
| CDKN2A | c.9_32dup | p.A4_P11dup | 42.27 | 28.55 |
| CHEK2 | c.1100del | p.T367fs | 42.93 | 44.79 |
| DICER1 | c.682_724dup | p.V242fs | 42.23 | 21.43 |
| EPCAM | c.859-1462_*1999del | N/A | 44.8 | ND |
| FH | c.37_92del | p.P13fs | 45.6 | 24.71 |
| FLCN | c.1285dup | p.H429fs | 43.13 | 23.44 |
| HOXB13 | c.844_845del | p.A282fs | 43.6 | 36.50 |
| KIT | c.1676T>C | p.V559A | 41.9 | NT |
| MAX | c.183_195del | p.Q62fs | 42.20 | 39.84 |
| MEN1 | c.1382_1404dup | p.E469fs | 41.97 | 36.62 |
| MET | c.3281A>G | p.H1094R | 46.13 | 58.33 |
| MITF | c.773_785dup | p.D263fs | 41.43 | 46.55 |
| MLH1 | c.1852_1854del | p.K618del | 42.73 | 50.00 |
| MSH2 | c.942+3A>T | N/A | 42.43 | ND |
| MSH2 | c.-125-9509096_1277-3165inv (Boland Inversion) | N/A | 43.58 | NT |
| MSH6 | c.3261dup | p.F1088fs | 41.50 | 22.81 |
| MUTYH | c.549_576del | p.L184fs | 45.50 | 18.14 |
| NBN | c.667_668insTTTATATTTTTATTATATAAAAATAATAAAGA AAACATTTATATTTTATAATATAAAAATATTATATTATATAT AAAATATAATAAAGAAAACATTATATA | p.K223delinsYIFII* | 40.13 | 14.58 |
| NF1 | c.2410-110_2850+65delinsAAAA | N/A | 42.30 | ND |
| NF1 | c.4600C>T | p.R1534* | 46.68 | 44.09 |

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|---------|---------------------------------------|-----------------|----------|---------|
| NTHL1 | c.417_436dup | p.L146fs | 41.13 | 32.03 |
| PALB2 | c.3114-1_3201+2del | N/A | 42.93 | ND |
| PDGFRA | c.2537A>T | p.D846V | 42.23 | NT |
| PMS2 | c.2243_2246del | p.K748fs | 43.50 | 41.67 |
| PMS2 | c.861_864del | p.R287fs | 44.27 | 40.00 |
| POLD1 | c.1433G>A | p.S478N | 42.30 | 35.72 |
| POLE | c.1270C>G | p.L424V | 44.80 | 33.03 |
| PTCH1 | c.202-16_227del | N/A | 44.63 | 27.67 |
| PTEN | c.750_751del | p.C250fs | 42.0 | 33.33 |
| RAD50 | c.326_329del | p.T109fs | 44.30 | 46.84 |
| RAD51C | c.706-2A>G | N/A | 43.27 | 53.42 |
| RAD51D | c.694_715delinsTGAGAGCTGAAGACCCTGGCCT | p.R232* | 43.77 | 42.07 |
| RET | c.2753T>C | p.M918T | 44.93 | 59.63 |
| SDHA | c.1785delinsCTTCTGGCGCGCATGCCAGG | p.E595fs | 43.07 | 57.52 |
| SDHAF2 | c.267del | p.F89fs | 43.43 | 44.57 |
| SDHB | c.42_43insCACTCTCCTTGAGGCGCCGGTTGCCG | p.A15delinsHSP* | 45.80 | 32.69 |
| SDHC | c.250_251del | p.L84fs | 41.47 | 41.46 |
| SDHD | c.383_386dup | p.L129fs | 43.97 | 46.67 |
| SMAD4 | c.1349_1376dup | p.A460fs | 43.33 | 51.72 |
| SMARCA4 | c.917_941del | p.Q306fs | 43.80 | 36.54 |
| STK11 | c.291-10_922del | N/A | 42.0 | ND |
| TMEM127 | c.24_48del | p.L9fs | 50.13 | 46.81 |
| TP53 | c.524G>A | p.R175H | 43.80 | 38.87 |
| TSC1 | c.850_881delinsGCTTTCCTCATCGTT | p.R284fs | 43.07 | 32.03 |
| TSC2 | c.2640-1G>A | N/A | 40.23 | 30.84 |
| VHL | c.481C>T | p.R161* | 43.77 | 41.03 |

N/A= not applicable

NT = not tested; panel does not cover variant.

ND= not detected, test does cover it, but the variant wasn't reported

Approval:


10 SEP 2024
 Prepared By _____ Date _____


13 Sep 2024
 QA Verified By _____ Date _____