

Targeted Genes and Methodology Details for SDH Genes Mutation Analysis

The following applies to SDHMP / *SDH* Genes Mutation Analysis, Next-Generation Sequencing. Next-generation sequencing (NGS) is performed to test for the presence of single nucleotide variations, deletions, and insertions in coding regions and intron/exon boundaries of the genes listed. When appropriate, alterations detected are confirmed by an independent reference method, such as Sanger sequencing. Default reportable range offset is +/-2 base pairs around each targeted exon region.

Genomic Build: GRCh37 (hg19) unless otherwise specified

As a result of technical limitations of the assay (including regions of homology, high GC content, and repetitive sequences), there are regions of some genes that cannot be effectively evaluated. Refer to gene regions table below for complete gene coverage information. To verify if a specific region/exon/variant is covered by this assay, contact the laboratory at 800-533-1710.

| Gene | Exon | Chromosome | Genomic Start | Genomic Stop | Reference Transcript |
|------|------|------------|---------------|--------------|-----------------------------|
| SDHA | Ex1 | chr5 | 218469 | 218535 | NM_004168 |
| SDHA | Ex2 | chr5 | 223595 | 223685 | NM_004168 |
| SDHA | Ex3 | chr5 | 224473 | 224638 | NM_004168 |
| SDHA | Ex4 | chr5 | 225532 | 225679 | NM_004168 |
| SDHA | Ex5 | chr5 | 225996 | 226164 | NM_004168 |
| SDHA | Ex6 | chr5 | 228298 | 228450 | NM_004168 |
| SDHA | Ex7 | chr5 | 230989 | 231117 | NM_004168 |
| SDHA | Ex8 | chr5 | 233590 | 233762 | NM_004168 |
| SDHA | Ex9 | chr5 | 235257 | 235456 | NM_004168 |
| SDHA | Ex11 | chr5 | 240471 | 240593 | NM_004168 |
| SDHA | Ex12 | chr5 | 251105 | 251220 | NM_004168 |
| SDHB | Ex1 | chr1 | 17380441 | 17380516 | NM_003000 |
| SDHB | Ex2 | chr1 | 17371254 | 17371385 | NM_003000 |
| SDHB | Ex3 | chr1 | 17359553 | 17359642 | NM_003000 |
| SDHB | Ex4 | chr1 | 17355093 | 17355233 | NM_003000 |
| SDHB | Ex5 | chr1 | 17354242 | 17354362 | NM_003000 |
| SDHB | Ex6 | chr1 | 17350466 | 17350571 | NM_003000 |
| SDHB | Ex7 | chr1 | 17349101 | 17349227 | NM_003000 |
| SDHB | Ex8 | chr1 | 17345374 | 17345455 | NM_003000 |
| SDHC | Ex1 | chr1 | 161284194 | 161284217 | NM_003001 |
| SDHC | Ex2 | chr1 | 161293402 | 161293462 | NM_003001 |
| SDHC | Ex3 | chr1 | 161298184 | 161298289 | NM_003001 |
| SDHC | Ex4 | chr1 | 161310382 | 161310447 | NM_003001 |
| SDHC | Ex5 | chr1 | 161326465 | 161326632 | NM_003001 |
| SDHD | Ex1 | chr11 | 111957630 | 111957685 | NM_003002 |
| SDHD | Ex2 | chr11 | 111958579 | 111958699 | NM_003002 |
| SDHD | Ex3 | chr11 | 111959589 | 111959737 | NM_003002 |
| SDHD | Ex4 | chr11 | 111965527 | 111965696 | NM_003002 |