

SONIC HEDGEHOG MEDULLOBLASTOMA PANEL DG-4.4.0 (8 GENES)

| Gene | Twist X2 covered 10x | Twist X2 covered 20x | srWGS covered 10x | srWGS covered 15x | srWGS covered 20x | Associated Phenotype description and OMIM disease ID |
|--------|----------------------|----------------------|-------------------|-------------------|-------------------|---|
| BRCA2 | 100% | 100% | 100% | 100% | 99.8% | Fanconi anemia, complementation group D1, 605724;{Glioblastoma 3}, 613029;{Medulloblastoma}, 155255;{Prostate cancer}, 176807;{Breast-ovarian cancer, familial, 2}, 612555;{Breast cancer, male, susceptibility to}, 114480;{Pancreatic cancer 2}, 613347;Wilms tumor, 194070 |
| ELP1 | 100% | 100% | 100% | 100% | 99.6% | {Medulloblastoma}, 155255;Dysautonomia, familial, 223900 |
| GPR161 | 100% | 100% | 100% | 100% | 99.6% | {Medulloblastoma predisposition syndrome}, 155255 |
| PALB2 | 100% | 100% | 100% | 100% | 99.6% | {Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832 |
| PTCH1 | 100% | 100% | 100% | 99.9% | 99.1% | Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828 |

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|---------|-------|-------|------|-------|-------|--|
| SMARCB1 | 100% | 100% | 100% | 100% | 99.6% | Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322 |
| SUFU | 100% | 100% | 100% | 99.9% | 98.8% | {Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255 |
| TP53 | 94.7% | 94.7% | 100% | 100% | 99% | {Basal cell carcinoma 7}, 614740;{Adrenocortical carcinoma, pediatric}, 202300;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Li-Fraumeni syndrome, 151623;Pancreatic cancer, somatic, 260350;Nasopharyngeal carcinoma, somatic, 607107;{Osteosarcoma}, 259500;{Choroid plexus papilloma}, 260500;{Colorectal cancer}, 114500;{Glioma susceptibility 1}, 137800;Bone marrow failure syndrome 5, 618165 |

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. *Nucleic Acids Res.* 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.

srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.

srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.

srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.

non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.

This list is accurate for panel version DG 4.4.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors