SHH MEDULLOBLASTOMA GENE PANEL DG 3.1.0 (9 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
BRCA2	99,8	98,5	100	100	{Pancreatic cancer 2}, 613347 {Breast cancer, male, susceptibility to}, 114480 {Glioblastoma 3}, 613029 Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724 {Medulloblastoma}, 155255 {Prostate cancer}, 176807 {Breast-ovarian cancer, familial, 2}, 612555
ELP1	99,8	99	100	100	Dysautonomia, familial, 223900
GPR161	100	100	100	100	No OMIM disease ID
PALB2	100	100	100	100	{Pancreatic cancer, susceptibility to, 3}, 613348 Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480
PTCH1	99,2	97,6	99,9	99,8	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	99,9	99	100	100	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
SMARCB1	100	100	100	100	Rhabdoid tumors, somatic, 609322 {Schwannomatosis-1, susceptibility to}, 162091 Coffin-Siris syndrome 3, 614608 {Rhabdoid tumor predisposition syndrome 1}, 609322
SUFU	100	100	100	100	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174 Joubert syndrome 32, 617757
TP53	99,9	97,7	91,7	91,7	{Adrenocortical carcinoma, pediatric}, 202300 {Glioma susceptibility 1}, 137800 {Basal cell carcinoma 7}, 614740

					Bone marrow failure syndrome 5, 618165 {Colorectal cancer}, 114500 Nasopharyngeal carcinoma, somatic, 607107 Breast cancer, somatic, 114480 {Osteosarcoma}, 259500 {Choroid plexus papilloma}, 260500 Li-Fraumeni syndrome, 151623 Hepatocellular carcinoma, somatic, 114550 Pancreatic cancer, somatic, 260350

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors