SHH MEDULLOBLASTOMA GENE PANEL DG 2.18 (9 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
BRCA2	99,80%	98,50%	100%	100%	Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724
ELP1	99,80%	99,00%	100%	100%	Dysautonomia, familial, 223900
GPR161	100%	100%	100%	100%	No OMIM disease ID
PALB2	100%	100%	100%	100%	Fanconi anemia, complementation group N, 610832
PTCH1	99,20%	97,60%	99,90%	99,80%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	99,90%	99,00%	100%	100%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
SMARCB1	100%	100%	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SUFU	100%	100%	100%	100%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 Joubert syndrome 32, 617757
TP53	99,90%	97,70%	91,70%	91,70%	Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Bone marrow failure syndrome 5, 618165 Nasopharyngeal carcinoma, somatic, 607107 Hepatocellular carcinoma, somatic, 114550

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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors