SHH MEDULLOBLASTOMA GENE PANEL DG 2.17 (8 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
BRCA2	103.2	99.7%	98.8%	Wilms tumor, 194070 Fanconi anemia, complementation group D1, 605724
GPR161	181.5	100.0%	100.0%	No OMIM Disease ID
PALB2	146.3	100.0%	99.9%	Fanconi anemia, complementation group N, 610832
PTCH1	117.3	100.0%	99.2%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	131.5	99.9%	98.8%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
SMARCB1	192.9	100.0%	100.0%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SUFU	141.7	100.0%	100.0%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 Joubert syndrome 32, 617757
TP53	96.8	99.9%	99.0%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.
This list is accurate for panel version DG 2.17

Ad 1. “No OMIM phenotype” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors