WES SONIC HEDGEHOG MEDULLOBLASTOMA DG 3.7

Gene	Twist X2 covered >10x	Twist X2 covered >20x	WGS covered >10x	WGS covered >20x	Associated Phenotype description and OMIM disease ID
BRCA2	100.0%	100.0%	100.0%	98.6%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
ELP1	100.0%	100.0%	100.0%	99.6%	Dysautonomia, familial, 223900
GPR161	100.0%	100.0%	100.0%	99.4%	
PALB2	100.0%	100.0%	100.0%	98.6%	Fanconi anemia, complementation group N, 610832
PTCH1	100.0%	100.0%	100.0%	99.4%	Basal cell nevus syndrome 1, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828
SMARCB1	100.0%	100.0%	100.0%	99.9%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SUFU	100.0%	100.0%	100.0%	99.8%	Joubert syndrome 32, 617757 Basal cell nevus syndrome 2, 620343
TP53	94.7%	94.7%	100.0%	99.3%	Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Li-Fraumeni syndrome, 151623 Pancreatic cancer, somatic, 260350 Nasopharyngeal carcinoma, somatic, 607107 Bone marrow failure syndrome 5, 618165

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.
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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 3.7.0.

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