

SHH MEDULLOBLASTOMA

GENE PANEL DG 3.6.0 (8 GENES)

Releasedate: 05-04-2023

Gene	TWIST X2 covered >10x	TWIST X2 covered >20x	Associated Phenotype description and OMIM disease ID
BRCA2	100%	100%	Fanconi anemia, complementation group D1, 605724 Wilms tumor, 194070
ELP1	100%	100%	Dysautonomia, familial, 223900
GPR161	100%	100%	No OMIM disease ID
PALB2	100%	100%	Fanconi anemia, complementation group N, 610832
PTCH1	100%	100%	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
SMARCB1	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SUFU	100%	100%	Joubert syndrome 32, 617757 Basal cell nevus syndrome, 109400
TP53	94%	94%	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 HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

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 is the chemistry used for 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 17th, 2023

This list is accurate for panel version DG 3.6.0

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