

SHH MEDULLOBLASTOMA GENE PANEL DG 3.3.0 (8 genes)

Releasedate: 13-01-2022

Gene	TWIST covered >10x	TWIST 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 Medulloblastoma, 155255
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 Medulloblastoma, desmoplastic, 155255 Basal cell nevus syndrome, 109400
TP53	91%	91%	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.

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TWIST 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 : January 13th , 2022.

This list is accurate for panel version DG 3.3.0

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