

SONIC HEDGEHOG MEDULLOBLASTOMA PANEL DG-3.9.0

(2 GENES)

Gene	Twist X2 covered >10x	Twist X2 covered >20x	WGS covered >10x	WGS covered >20x	Associated Phenotype description and OMIM disease ID
PALB2	100.0%	100.0%	100.0%	96.7%	{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832
PTCH1	100.0%	100.0%	100.0%	97.3%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828

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 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 3.9.0