HEREDITARY RHABDOID AND NERVE SYSTEM TUMORS PANEL DG-5.2.0 (10 GENES)

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.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ARMC5	100% | 100%	96% | 96%	98% | 98%	{ACTH-independent macronodular adrenal hyperplasia 2}, 615954
CDKN2A	100% | 100%	96% | 96%	99% | 99%	{Melanoma and neural system tumor syndrome}, 155755;{Melanoma, cutaneous malignant, 2}, 155601;{Melanoma-pancreatic cancer syndrome}, 606719
CDKN2B	100% | 100%	96% | 96%	99% | 99%	
DGCR8	96% | 96%	96% | 96%	99% | 99%	
LZTR1	100% | 100%	97% | 97%	99% | 99%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
NF2	100% | 100%	97% | 97%	99% | 99%	Meningioma, NF2-related, somatic, 607174;Schwannomatosis, vestibular, 101000;Schwannomatosis, somatic, 101000
SMARCA4	100% | 100%	96% | 96%	99% | 99%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SMARCB1	96% | 96%	97% | 97%	99% | 99%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCE1	100% | 100%	98% | 98%	99% | 99%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938
SUFU	100% | 100%	96% | 96%	99% | 99%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255