HEREDITARY RHABDOID AND NERVE SYSTEM TUMORS PANEL DG-5.1.0 (10 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ARMC5	100%	100%	100%	100%	99.4%	{ACTH-independent macronodular adrenal hyperplasia 2}, 615954
CDKN2A	100%	100%	100%	99.8%	98.5%	{Melanoma and neural system tumor syndrome}, 155755;{Melanoma, cutaneous malignant, 2}, 155601;{Melanoma-pancreatic cancer syndrome}, 606719
CDKN2B	100%	100%	100%	100%	99.5%	
DGCR8	95.6%	95.6%	100%	100%	99.3%	
LZTR1	100%	100%	100%	100%	99.6%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
NF2	100%	100%	100%	100%	99.5%	Meningioma, NF2-related, somatic, 607174;Schwannomatosis, vestibular, 101000;Schwannomatosis, somatic, 101000
SMARCA4	100%	100%	100%	99.9%	99.3%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792

SMARCB1	95.2%	95.2%	100%	100%	99.7%	Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322
SMARCE1	100%	100%	100%	100%	99.8%	{Meningioma, familial, susceptibility to}, 607174;Coffin-Siris syndrome 5, 616938
SUFU	100%	100%	100%	100%	99.6%	{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255

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 mapped against GRCh38.
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 mapped against GRCh38.
srWGS 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 covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS 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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors