NOONAN SYNDROME / RASOPATHY PANEL DG-5.1.0 (27 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*
BRAF	100%	100%	100%	99.8%	98.2%	Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980
CBL	100%	100%	100%	100%	99.7%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CDC42	100%	100%	100%	100%	99.6%	Takenouchi-Kosaki syndrome, 616737
ERF	100%	100%	100%	100%	97.9%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180

HRAS	100%	100%	100%	99.9%	99.5%	Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040
KRAS	100%	100%	100%	100%	100%	Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;Bladder cancer, somatic, 109800

LZTR1	100%	100%	100%	100%	99.6%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MAP2K1	95.8%	95.8%	100%	100%	99.5%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	92.4%	92.3%	100%	100%	99.5%	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100%	100%	100%	99.9%	99.4%	Noonan syndrome 13, 619087
MRAS	100%	100%	100%	100%	99.7%	Noonan syndrome 11, 618499
NF1	99.4%	99.4%	100%	100%	99.8%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321

NRAS	89.4%	89.4%	100%	100%	99.9%	Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500
PPP1CB	87.8%	87.6%	100%	100%	99.7%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	89.2%	89.2%	100%	100%	99.8%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
RAC1	78.8%	78.8%	100%	100%	99.7%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAF1	96.1%	93.8%	100%	100%	99.7%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RASA2	96.6%	96.6%	100%	100%	99.7%	
RIT1	100%	100%	100%	100%	99.8%	Noonan syndrome 8, 615355

RRAS	93.2%	93.2%	100%	99.9%	98.4%	
RRAS2	95.5%	95.5%	100%	100%	99.5%	Ovarian carcinoma;Noonan syndrome 12, 618624
RREB1	100%	100%	100%	99.9%	98.9%	
SHOC2	100%	100%	100%	100%	99.9%	Noonan syndrome-like with loose anagen hair 1, 607721
SOS1	98.8%	98.6%	100%	100%	99.6%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	95%	95%	100%	100%	99.5%	Noonan syndrome 9, 616559
SPRED1	97.6%	97.6%	100%	100%	100%	Legius syndrome, 611431
SPRED2	100%	100%	100%	100%	99.8%	Noonan syndrome 14, 619745

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