## NOONAN SYNDROME / RASOPATHY PANEL DG-4.3.0 (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%	99.7%	98.4%	96%	Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocuta neous syndrome, 115150;Adenocarcinom a of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980
CBL	100%	100%	100%	99.9%	98.9%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785

CDC42	100%	100%	100%	100%	99.7%	Takenouchi-Kosaki syndrome, 616737
ERF	100%	100%	100%	99.1%	94.2%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180
HRAS	100%	100%	100%	100%	98.4%	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;Schimmelpenni ng-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040

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KRAS	100%	100%	100%	99.9%	99.3%	Gastric cancer,
						somatic,
						613659;Oculoectoderm
						al 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;Schimmelpenni
						ng-Feuerstein-Mims
						syndrome, somatic
						mosaic,
						163200;Cardiofaciocuta
						neous syndrome 2,
						615278;Bladder cancer,
						somatic, 109800
LZTR1	100%	100%	100%	99.8%	98.5%	Noonan syndrome 2,
L_ 11X1	10070	10070	10070	33.070	30.070	605275;Noonan
						syndrome 10,
						616564;{Schwannomat
						osis-2, susceptibility to},
						615670

MAP2K1	95.8%	95.8%	100%	100%	99.4%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100%	100%	100%	99.5%	97%	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100%	100%	100%	99.8%	98.9%	Noonan syndrome 13, 619087
MRAS	100%	100%	100%	100%	99%	Noonan syndrome 11, 618499
NF1	99.4%	99.4%	100%	99.9%	99.5%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromato sis, familial spinal, 162210;Neurofibromato sis, type 1, 162200;Neurofibromato sis-Noonan syndrome, 601321

NRAS	100%	100%	100%	100%	99.1%	Noonan syndrome 6, 613224;?RAS- associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic,
						162900;Schimmelpenni ng-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneou s melanosis, somatic, 249400;Colorectal cancer, somatic, 114500
PPP1CB	88%	87.4%	100%	100%	99.2%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	90.1%	89.2%	100%	99.9%	99.4%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondrom atosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785

RAC1	86.4%	86.4%	100%	100%	98.7%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAF1	97.6%	94.2%	100%	100%	99.4%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RASA2	100%	100%	100%	99.9%	99.2%	
RIT1	100%	100%	100%	99.9%	98.4%	Noonan syndrome 8, 615355
RRAS	100%	99.7%	100%	99.3%	94.9%	
RRAS2	100%	100%	100%	99.9%	99.2%	Ovarian carcinoma;Noonan syndrome 12, 618624
RREB1	100%	100%	100%	99.8%	97.6%	
SHOC2	100%	100%	100%	100%	99.2%	Noonan syndrome-like with loose anagen hair 1, 607721
SOS1	98.8%	98.6%	100%	100%	99.5%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	100%	100%	100%	99.9%	99%	Noonan syndrome 9, 616559
SPRED1	100%	100%	100%	100%	99.3%	Legius syndrome, 611431
SPRED2	100%	100%	100%	99.8%	98.5%	Noonan syndrome 14, 619745

Gene symbols used follow HGCN 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 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors