WES NOONAN SYNDROME / RASOPATHY DG 3.7

Gene	Twist X2 covered >10x	Twist X2 covered >20x	WGS covered >10x	WGS covered >20x	Associated Phenotype description and OMIM disease ID
BRAF	100.0%	100.0%	100.0%	99.5%	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	100.0%	100.0%	100.0%	99.7%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CDC42	100.0%	100.0%	100.0%	99.4%	Takenouchi-Kosaki syndrome, 616737
HRAS	100.0%	100.0%	100.0%	99.7%	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	100.0%	100.0%	100.0%	99.8%	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	100.0%	100.0%	100.0%	99.8%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAP2K1	100.0%	100.0%	100.0%	99.2%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100.0%	100.0%	100.0%	99.8%	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100.0%	100.0%	100.0%	99.3%	Noonan syndrome 13, 619087
MRAS	100.0%	100.0%	100.0%	99.7%	Noonan syndrome 11, 618499

NF1	100.0%	100.0%	100.0%	99.3%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NRAS	100.0%	100.0%	100.0%	99.8%	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	100.0%	100.0%	99.9%	99.0%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	100.0%	100.0%	100.0%	98.9%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAC1	100.0%	100.0%	100.0%	99.4%	Intellectual developmental disorder, autosomal dominant 48, 617751

RAF1	100.0%	100.0%	100.0%	99.7%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RIT1	100.0%	100.0%	100.0%	99.8%	Noonan syndrome 8, 615355
RRAS	100.0%	99.8%	100.0%	98.6%	
RRAS2	100.0%	100.0%	100.0%	97.6%	Noonan syndrome 12, 618624 Ovarian carcinoma,
RREB1	100.0%	100.0%	100.0%	99.9%	
SHOC2	100.0%	100.0%	100.0%	98.7%	Noonan syndrome-like with loose anagen hair 1, 607721
SOS1	100.0%	100.0%	100.0%	99.0%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100.0%	100.0%	100.0%	99.0%	Noonan syndrome 9, 616559
SPRED1	100.0%	100.0%	100.0%	99.6%	Legius syndrome, 611431
SPRED2	100.0%	100.0%	100.0%	99.8%	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.
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 3.7.0.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors