NOONAN SYNDROME AND RASOPATHY GENE PANEL DG 3.5.0 (24 genes)

Releasedate: 05-12-2022

Gene	*TWIST X2 covered >10x*	*TWIST X2 covered >20x*	*Associated Phenotype description and OMIM disease ID*
BRAF	100%	100%	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%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CDC42	100%	100%	Takenouchi-Kosaki syndrome, 616737
HRAS	100%	100%	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%	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%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAP2K1	100%	100%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100%	100%	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100%	100%	Noonan syndrome 13, 619087
MRAS	100%	100%	Noonan syndrome 11, 618499
NRAS	100%	100%	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%	100%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	100%	100%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAC1	100%	100%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAF1	100%	100%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RIT1	100%	100%	Noonan syndrome 8, 615355
RRAS	100%	100%	No OMIM disease ID
RRAS2	100%	100%	Noonan syndrome 12, 618624 Ovarian carcinoma,
RREB1	100%	100%	No OMIM disease ID
SHOC2	100%	100%	Noonan syndrome-like with loose anagen hair 1, 607721
SOS1	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100%	100%	Noonan syndrome 9, 616559
SPRED1	100%	100%	Legius syndrome, 611431
SPRED2	100%	100%	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.
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TWIST X2 is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 28th , 2022.
This list is accurate for panel version DG 3.5.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors