NOONAN SYNDROME/RASOPATHY GENE PANEL DG 3.3.0 (23 genes)

Releasedate: 13-01-2022

Gene	TWIST covered >10x	TWIST 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%				
MAP2K1 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 6, 613224 PRAS-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 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 PPP1CB 100% 100% Noonan syndrome, congenital, somatic, 249400 Colorectal cancer, somatic, 114500 Noonan syndrome, 1, 163950 LEOPARD syndrome 1, 163950 LEOPARD syndrome 1, 163950 LEOPARD syndrome 1, 163950 LEOPARD syndrome 1, 163950 LEOPARD syndrome 2, 611553 LEOPARD syndrome 2, 611553 LEOPARD syndrome 2, 611553 RRAS 100% Noonan syndrome 8, 613355 S RRAS 100% Noonan syndrome 8, 61354 Noonan syndrome 2, 613555 RRAS 100% Noonan syndrome 2, 618624 Noonan syndrome 3, 618624 Noonan syndrome 4, 610733 Noonan syndrome 4, 61073	LZTR1	100%	100%	Noonan syndrome 2, 605275
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	SPRED2	100%	100%	No OMIM disease ID

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. 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 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 : January 13th , 2022.

This list is accurate for panel version DG 3.3.0

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