NOONAN SYNDROME/RASOPATHY GENE PANEL DG 3.2.0 (20 genes)

Releasedate: 16-09-2021

Gene	Agilent V5 covered >10x	Agilent V5 covered >20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype Description and OMIM disease ID
BRAF	89,4	77,6	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	97,3	96,9	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
HRAS	100	100	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	99	97,8	100	100	Gastric cancer, somatic, 137215 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	99,9	100	100	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564

Gene					Disease
MAP2K1	99,6	96,1	100	100	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	98,5	95,3	100	100	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100	99,9	100	99,6	Noonan syndrome 13, 619087
MRAS	100	99,3	100	100	Noonan syndrome 11, 618499
NRAS	100	100	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	99,8	98,7	100	100	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	97,7	87,6	100	100	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
RAF1	99,9	99,2	100	100	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RRAS2	95,9	87,4	100	100	Noonan syndrome 12, 618624 Ovarian carcinoma,
RREB1	99,8	99,2	100	100	No OMIM disease ID
SHOC2	99,8	99,6	100	99,9	Noonan syndrome-like with loose anagen hair 1, 607721
SOS1	99,6	97,9	100	99,9	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	99,6	98,7	100	99,9	Noonan syndrome 9, 616559
SPRED1	99,8	98,2	100	100	Legius syndrome, 611431

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 : September 16th , 2021.

This list is accurate for panel version DG 3.2.0

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