

NOONAN SYNDROME/RASOPATHY GENE PANEL DG 3.1.0 (17 genes)

Releasedate: 23-03-2021

<i>Gene</i>	<i>Agilent V5 covered >10x</i>	<i>Agilent V5 covered >20x</i>	<i>TWIST covered >10x</i>	<i>TWIST covered >20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
BRAF	91	81,1	100	100	Melanoma, malignant, somatic, 155600 Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 114500 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0
CBL	97,3	97,1	100	100	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
HRAS	100	100	100	100	Costello syndrome, 218040 Bladder cancer, somatic, 109800 Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550
KRAS	99,5	96,9	100	100	Oculoectodermal syndrome, somatic, 600268 Leukemia, acute myeloid, somatic, 601626 Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942

LZTR1	100	99,9	100	100	{Schwannomatosis-2, susceptibility to}, 615670 Noonan syndrome 2, 605275 Noonan syndrome 10, 616564
MAP2K1	99,8	97,1	100	100	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	98,5	95,1	100	100	Cardiofaciocutaneous syndrome 4, 615280
NRAS	100	100	100	100	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
PPP1CB	99,9	99,3	100	100	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	99,1	93,7	100	100	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
RAF1	100	100	100	100	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RIT1	100	100	100	100	Noonan syndrome 8, 615355
RREB1	99,9	99,2	100	100	No OMIM disease ID
SHOC2	99,9	99,4	100	100	Noonan syndrome-like with loose anagen hair 1, 607721
SOS1	99,8	98,4	100	100	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100	99,2	100	100	Noonan syndrome 9, 616559
SPRED1	100	98,9	100	100	Legius syndrome, 611431

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.

TWIST is the chemistry used for (in-house) TURBO/RAPID 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 : March 23rd , 2021.

This list is accurate for panel version DG 3.1.0

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