NOONAN SYNDROME/RASOPATHY GENE PANEL DG 2.18 (16 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
BRAF	95,60%	85,10%	100%	100%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
CBL	97,30%	97,10%	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
HRAS	100%	100%	100%	100%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
KRAS	99,50%	96,90%	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%	99,90%	100%	100%	Noonan syndrome 2, 605275 Noonan syndrome 10, 616564

MAP2K1	99,80%	97,10%	100%	100%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	98,50%	95,10%	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	99,90%	99,30%	100%	100%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	99,10%	93,70%	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
SHOC2	99,90%	99,40%	100%	100%	Noonan syndrome-like with loose anagen hair, 607721
SOS1	99,80%	98,40%	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOS2	100%	99,20%	100%	100%	Noonan syndrome 9, 616559
SPRED1	100%	98,90%	100%	100%	Legius syndrome, 611431

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.
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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors