Panelinformation Noonans syndrom in-silico

Version: v.1.0

Gen	Transkript*	% täckning > 10x **	% täckning > 20x**	Median täckning **	Associerad fenotyp och OMIM sjukdoms ID	Ärftlighet
PTPN11	NM_002834.3	100	95	58	LEOPARD syndrome, 151100 Noonan syndrome 1, 163950 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785	AD
SOS1	NM_005633.3	95	92	55	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300	AD
RAF1	NM_002880.3	100	100	100	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916	AD
KRAS	NM_033360.3	100	71	43	Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 RAS-associated autoimmune leukoproliferative disorder, 614470	AD
NRAS	NM_002524.4	100	100	61	Noonan syndrome 6, 613224	AD
BRAF	NM_004333.4	98	86	45	LEOPARD syndrome 3, 613707 Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150	AD
RIT1	NM_001256821.1	100	100	70	Noonan syndrome 8, 615355	AD
SOS2	NM_006939.3	97	92	51	Noonan syndrome 9, 616559	AD
LZTR1	NM_006767.3	100	100	166	Schwannomatosis, 615670 Noonan syndrome 10, 616564	AD
MAP2K1	NM_002755.3	100	100	93	Cardiofaciocutaneous syndrome 3, 615279	AD
MAP2K2	NM_030662.3	100	92	106	Cardiofaciocutaneous syndrome 4, 615280	AD
HRAS	NM_005343.3	100	100	210	Costello syndrome, 218040 Congenital myopathy with excess of muscle spindles, 218040	AD

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CBL	NM_005188.3	100	100	94	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	AD
SHOC2	NM_001324337.1	99	95	53	Noonan-like syndrome with loose anagen hair, 607721	AD
PPP1CB	NM_002709.2	100	97	50	Noonan syndrome-like disorder with loose anagen hair 2, 617506	AD
A2ML1	NM_144670.4	100	100	89	Noonan-syndrome like phenotype ? ***	
RASA2	NM_001303246.1	92	84	40	Noonan-syndrome like phenotype ? ***	
RRAS	NM_006270.4	100	100	181	Noonan-syndrome like phenotype ? ***	
SPRED1	NM_152594.2	100	97	58	Legius syndrome, 611431	AD
NF1	NM_001042492.2	94	84	45	Neurofibromatosis, type 1, 162200	AD

* Referenstranskript enligt NCBI Reference Sequences (RefSeq). Detta transkriptnummer används som utgångspunkt för analys av horisontell täckning. Däremot görs varianttolkning från genens hela kodande sekvens, oberoende av isoform.

** Medelvärde av 32 valideringsprov

*** Recent reports have implicated several additional genes associated with a Noonan syndrome-like phenotype in fewer than ten individuals each including RRAS (2 probands) [Flex et al 2014], RASA2 (3 probands) [Chen et al 2014b], A2ML1 (3 probands) [Vissers et al 2015], SOS2 (8 probands) [Cordeddu et al 2015, Yamamoto et al 2015] and LZTR1 (4 probands) [Yamamoto et al 2015].

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