Panelinformation Noonans syndrom in-silico

Version: v.1.0

Gen	Associerad fenotyp och OMIM sjukdoms ID	Ärftlighet
PTPN11	LEOPARD syndrome, 151100 Noonan syndrome 1, 163950 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785	AD
SOS1	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300	AD
RAF1	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916	AD
KRAS	Noonan syndrome 3, 609942 Cardiofaciocutaneous syndrome 2, 615278 RAS-associated autoimmune leukoproliferative disorder, 614470	AD
NRAS	Noonan syndrome 6, 613224	AD
BRAF	LEOPARD syndrome 3, 613707 Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150	AD
RIT1	Noonan syndrome 8, 615355	AD
SOS2	Noonan syndrome 9, 616559	AD
LZTR1	Schwannomatosis, 615670 Noonan syndrome 10, 616564	AD
MAP2K1	Cardiofaciocutaneous syndrome 3, 615279	AD
MAP2K2	Cardiofaciocutaneous syndrome 4, 615280	AD
HRAS	Costello syndrome, 218040 Congenital myopathy with excess of muscle spindles, 218040	AD
CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563	AD
SHOC2	Noonan-like syndrome with loose anagen hair, 607721	AD
PPP1CB	Noonan syndrome-like disorder with loose anagen hair 2, 617506	AD
A2ML1	Noonan-syndrome like phenotype ? *	
RASA2	Noonan-syndrome like phenotype ? *	
RRAS	Noonan-syndrome like phenotype ? *	
SPRED1	Legius syndrome, 611431	AD
NF1	Neurofibromatosis, type 1, 162200	AD

* 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].