LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DG-4.2.0 (57 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ABCC9	96%	96%	100%	100%	99.3%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ADAMTS3	100%	100%	100%	100%	99.4%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ALG8	78.5%	77.5%	100%	99.9%	98.9%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ANGPT2	100%	100%	100%	100%	99.3%	Lymphatic malformation 10, 619369

ARAF	100%	99.3%	98.4%	85.5%	65.5%	
BRAF	100%	100%	99.9%	99.1%	97.1%	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	100%	100%	100%	99.9%	98.9%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CCBE1	100%	100%	100%	100%	99.3%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD55	96.1%	92.5%	100%	100%	99.3%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300

CDC42	100%	100%	100%	100%	99.5%	Takenouchi-Kosaki syndrome, 616737
CELSR1	100%	99.7%	100%	99.8%	98.3%	Lymphatic malformation 9, 619319
DGAT1	100%	100%	100%	100%	99%	Diarrhea 7, protein-losing enteropathy type, 615863
EPHB4	100%	100%	100%	99.9%	98.9%	Capillary malformation-arteriovenous malformation 2, 618196;Lymphatic malformation 7, 617300
ERF	100%	100%	100%	99.6%	96%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180
ERG	100%	100%	100%	100%	99.4%	Lymphatic malformation 14, 620602
FAT4	100%	100%	100%	99.9%	99.2%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FLT4	100%	100%	100%	99.9%	98.8%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780

FOXC2	100%	100%	100%	99.6%	96.7%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
GATA2	85.7%	85.7%	100%	100%	99.2%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286
GJA1	100%	100%	100%	100%	98.9%	Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850

GJC2	99.7%	95.8%	100%	100%	98%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GLA	91.4%	91.3%	98.9%	87.4%	67.5%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
HRAS	100%	100%	100%	99.9%	99.3%	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

KIF11	100%	100%	100%	100%	99.7%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950
KRAS	100%	100%	100%	100%	99.6%	Gastric cancer, somatic, 613659;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%	100%	100%	100%	99.2%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MAP2K1	95.8%	95.8%	100%	100%	99.2%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100%	100%	100%	99.9%	98.4%	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100%	100%	100%	100%	99.4%	Noonan syndrome 13, 619087
MDFIC	100%	100%	100%	99.9%	98.9%	Lymphatic malformation 12, 620014
MPI	100%	100%	100%	100%	99.8%	Congenital disorder of glycosylation, type Ib, 602579
MRAS	100%	100%	100%	100%	99.5%	Noonan syndrome 11, 618499

NF1	99.4%	99.4%	100%	99.9%	99.3%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NRAS	100%	100%	100%	99.9%	99.2%	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

PIEZO1	100%	100%	100%	99.9%	98.7%	[ER blood group system], 620207;Lymphatic malformation 6, 616843;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380

PIK3CA	100%	100%	100%	100%	99.5%	Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Nevus, epidermal, somatic, 162900;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108

PLVAP	100%	100%	100%	99.9%	98.5%	Diarrhea 10, protein-losing enteropathy type, 618183
PMM2	100%	100%	100%	100%	99.1%	Congenital disorder of glycosylation, type Ia, 212065
PPP1CB	87.9%	87.3%	100%	99.9%	99.2%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	89.4%	89.2%	100%	100%	99.5%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN14	100%	100%	100%	99.9%	99.2%	Choanal atresia and lymphedema, 613611
RAC1	86.4%	86.4%	100%	100%	99%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAF1	96.6%	93.7%	100%	100%	99.3%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554

RASA1	100%	100%	100%	100%	99.5%	Capillary malformation-arteriovenous malformation 1, 608354;Basal cell carcinoma, somatic, 605462
RIT1	100%	100%	100%	100%	99.3%	Noonan syndrome 8, 615355
RRAS	100%	99.7%	100%	99.6%	96.9%	
RRAS2	100%	100%	100%	100%	99.5%	Ovarian carcinoma;Noonan syndrome 12, 618624
RREB1	100%	100%	100%	100%	98.4%	
SHOC2	100%	100%	100%	100%	99.4%	Noonan syndrome-like with loose anagen hair 1, 607721
SOS1	98.8%	98.5%	100%	100%	99.4%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	100%	100%	100%	99.9%	99%	Noonan syndrome 9, 616559
SOX18	99.2%	96.3%	100%	99.8%	94.9%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940

SPRED1	100%	100%	100%	100%	99.7%	Legius syndrome, 611431
SPRED2	100%	100%	100%	99.8%	98.9%	Noonan syndrome 14, 619745
THSD1	100%	100%	100%	100%	99.7%	?Aneurysm, intracranial berry, 12, 618734;Lymphatic malformation 13, 620244
TIE1	100%	100%	100%	99.9%	99%	Lymphatic malformation 11, 619401
VEGFC	100%	100%	100%	100%	98.7%	Lymphatic malformation 4, 615907

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.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors