LYMPHATIC ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.2.0 (57 GENES)

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.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ABCC9	95% | 95%	98% | 98%	100% | 100%	Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ADAMTS3	100% | 100%	97% | 97%	100% | 100%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ALG8	78% | 78%	95% | 95%	99% | 99%	Congenital disorder of glycosylation, type Ih, 608104;Polycystic liver disease 3 with or without kidney cysts, 617874
ANGPT2	100% | 100%	97% | 96%	99% | 99%	Lymphatic malformation 10, 619369
ARAF	97% | 97%	98% | 98%	100% | 100%	
BRAF	100% | 100%	95% | 95%	99% | 99%	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%	97% | 97%	99% | 99%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CCBE1	100% | 100%	97% | 97%	99% | 99%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD55	93% | 93%	96% | 96%	99% | 99%	[Blood group Cromer], 613793;Complement hyperactivation, angiopathic thrombosis, and protein-losing enteropathy, 226300
CDC42	100% | 100%	98% | 98%	99% | 99%	Takenouchi-Kosaki syndrome, 616737
CELSR1	100% | 100%	95% | 95%	98% | 98%	Lymphatic malformation 9, 619319;Yellow nail syndrome, 153300
DGAT1	100% | 100%	96% | 96%	98% | 98%	Diarrhea 7, protein-losing enteropathy type, 615863
EPHB4	95% | 95%	95% | 95%	97% | 97%	Capillary malformation-arteriovenous malformation 2, 618196;Lymphatic malformation 7, 617300
ERF	100% | 100%	95% | 95%	98% | 98%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180

ERG	100% | 100%	97% | 97%	99% | 99%	Lymphatic malformation 14, 620602
FAT4	100% | 100%	97% | 97%	99% | 99%	Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FLT4	97% | 96%	95% | 95%	98% | 98%	Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780
FOXC2	100% | 100%	93% | 93%	99% | 99%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
GATA2	86% | 86%	96% | 96%	98% | 98%	{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286
GJA1	100% | 100%	97% | 97%	99% | 99%	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	95% | 95%	94% | 94%	99% | 99%	Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GLA	91% | 91%	99% | 99%	100% | 100%	Fabry disease, cardiac variant, 301500;Fabry disease, 301500
HRAS	100% | 100%	96% | 96%	98% | 98%	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%	98% | 98%	99% | 99%	Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950
KRAS	100% | 100%	98% | 98%	99% | 99%	Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;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;RAS-associated autoimmune leukoproliferative disorder 2, somatic, 621582;Bladder cancer, somatic, 109800
LZTR1	100% | 100%	97% | 97%	99% | 99%	Noonan syndrome 2, 605275;Noonan syndrome 10, 616564;{Schwannomatosis-2, susceptibility to}, 615670
MAP2K1	96% | 96%	97% | 97%	99% | 99%	Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	92% | 92%	94% | 94%	97% | 97%	Cardiofaciocutaneous syndrome 4, 615280
MAPK1	100% | 100%	98% | 98%	99% | 99%	Noonan syndrome 13, 619087
MDFIC	100% | 100%	97% | 97%	99% | 99%	Lymphatic malformation 12, 620014

MPI	100% | 100%	97% | 97%	99% | 99%	Congenital disorder of glycosylation, type Ib, 602579
MRAS	100% | 100%	97% | 97%	99% | 99%	Noonan syndrome 11, 618499
NF1	99% | 99%	98% | 98%	100% | 100%	Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NRAS	91% | 91%	97% | 97%	100% | 100%	Noonan syndrome 6, 613224;RAS-associated autoimmune lymphoproliferative syndrome type 1, 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	99% | 99%	94% | 94%	95% | 90%	[ER blood group system], 620207;Lymphatic malformation 6, 616843;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIK3CA	100% | 100%	98% | 98%	99% | 99%	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;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Nevus, epidermal, somatic mosaic, 162900;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108
PLVAP	95% | 95%	96% | 96%	98% | 98%	Diarrhea 10, protein-losing enteropathy type, 618183
PMM2	95% | 95%	97% | 97%	99% | 99%	Congenital disorder of glycosylation, type Ia, 212065
PPP1CB	89% | 89%	97% | 97%	99% | 99%	Noonan syndrome-like disorder with loose anagen hair 2, 617506
PTPN11	89% | 89%	98% | 98%	99% | 99%	Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN14	100% | 100%	96% | 96%	99% | 99%	Choanal atresia and lymphedema, 613611
RAC1	79% | 79%	94% | 94%	99% | 99%	Intellectual developmental disorder, autosomal dominant 48, 617751
RAF1	93% | 93%	97% | 97%	99% | 99%	Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RASA1	100% | 100%	98% | 98%	99% | 99%	Capillary malformation-arteriovenous malformation 1, 608354;Basal cell carcinoma, somatic, 605462
RIT1	100% | 100%	97% | 97%	99% | 99%	Noonan syndrome 8, 615355
RRAS	94% | 94%	94% | 94%	98% | 98%	
RRAS2	97% | 97%	97% | 97%	99% | 98%	Ovarian carcinoma;Noonan syndrome 12, 618624
RREB1	100% | 100%	95% | 95%	99% | 99%	

SHOC2	100% | 100%	97% | 97%	100% | 100%	Noonan syndrome-like with loose anagen hair 1, 607721
SOS1	98% | 98%	97% | 97%	99% | 99%	Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOS2	95% | 95%	96% | 96%	99% | 99%	Noonan syndrome 9, 616559
SOX18	98% | 98%	95% | 95%	97% | 96%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SPRED1	98% | 98%	97% | 97%	99% | 99%	Legius syndrome, 611431
SPRED2	100% | 100%	97% | 97%	99% | 99%	Noonan syndrome 14, 619745
THSD1	100% | 100%	97% | 97%	99% | 99%	?Aneurysm, intracranial berry, 12, 618734;Lymphatic malformation 13, 620244
TIE1	100% | 100%	96% | 96%	99% | 99%	Lymphatic malformation 11, 619401
VEGFC	100% | 100%	97% | 97%	100% | 100%	Lymphatic malformation 4, 615907