SKIN DISORDERS GENE PANEL DG 2.18 (622 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
AAAS	100%	99,90%	100%	100%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	100%	99,20%	100%	100%	Keratoderma, palmoplantar, punctate type IA, 148600
ABCA12	99,50%	98,70%	100%	100%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCB6	100%	99,80%	100%	100%	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153
ABCC6	93,60%	92,40%	100%	100%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC9	100%	99,90%	100%	100%	Hypertrichotic osteochondrodysplasia, 239850 ?Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABHD5	100%	100%	100%	100%	Chanarin-Dorfman syndrome, 275630
ACD	100%	99,90%	100%	100%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACTA2	100%	99,00%	100%	100%	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTB	99,70%	96,10%	100%	100%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACVRL1	100%	98,90%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADA2	100%	99,00%	100%	100%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM10	94,80%	93,90%	100%	100%	Reticulate acropigmentation of Kitamura, 615537
ADAM17	99,90%	99,00%	100%	100%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAMTS10	99,90%	98,50%	100%	100%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	92,80%	89,00%	97,60%	95,80%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS2	99,00%	96,60%	98,00%	97,80%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100%	100%	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154

ADAMTSL2	97,10%	93,30%	99,80%	99,40%	Geleophysic dysplasia 1, 231050
ADAR	100%	99,80%	100%	100%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	100%	100%	100%	100%	Aspartylglucosaminuria, 208400
AGPAT2	99,60%	96,10%	100%	100%	Lipodystrophy, congenital generalized, type 1, 608594
AIRE	100%	99,80%	100%	100%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKT1	100%	99,50%	100%	100%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500
AKT3	98,70%	94,50%	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	99,30%	94,10%	100%	100%	Porphyria, acute hepatic, 612740
ALAS2	98,90%	94,90%	100%	100%	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
ALDH18A1	100%	99,90%	100%	100%	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH3A2	95,30%	94,60%	100%	100%	Sjogren-Larsson syndrome, 270200
ALDOB	100%	99,10%	100%	100%	Fructose intolerance, hereditary, 229600
ALOX12B	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	100%	99,50%	100%	100%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	100%	100%	100%	100%	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALX4	100%	99,30%	100%	100%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMELX	99,90%	96,80%	100%	100%	Amelogenesis imperfecta, type 1E, 301200
ANKRD11	97,50%	94,80%	100%	100%	KBG syndrome, 148050
ANOS1	89,80%	88,90%	99,90%	99,40%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	99,70%	97,90%	100%	100%	GAPO syndrome, 230740
ANTXR2	100%	98,20%	100%	100%	Hyaline fibromatosis syndrome, 228600
AP1B1	100%	99,50%	100%	100%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1S3	90,40%	90,10%	90,50%	90,50%	No OMIM disease ID
AP3B1	99,20%	95,80%	100%	100%	Hermansky-Pudlak syndrome 2, 608233

APC	100%	99,70%	100%	100%	Desmoid disease, hereditary, 135290 Adenomatous polyposis coli, 175100 Gardner syndrome, 175100 Hepatoblastoma, somatic, 114550 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Gastric cancer, somatic, 613659 Adenoma, periampullary, somatic, 0
APCDD1	100%	99,80%	100%	100%	Hypotrichosis 1, 605389
AQP5	100%	97,00%	100%	100%	Palmoplantar keratoderma, Bothnian type, 600231
ARHGAP31	99,90%	98,80%	100%	100%	Adams-Oliver syndrome 1, 100300
ARID1A	98,10%	96,40%	100%	100%	Coffin-Siris syndrome 2, 614607
ARID1B	99,50%	98,60%	99,90%	99,20%	Coffin-Siris syndrome 1, 135900
ASIP	100%	100%	100%	100%	No OMIM disease ID
ASL	100%	99,60%	100%	100%	Argininosuccinic aciduria, 207900
ASXL1	100%	99,50%	99,90%	99,90%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	99,90%	99,70%	100%	100%	Bainbridge-Ropers syndrome, 615485
ATIC	99,90%	99,30%	100%	100%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP2A2	100%	100%	100%	100%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	100%	99,60%	100%	100%	Hailey-Hailey disease, 169600
ATP6V0A2	100%	99,50%	100%	100%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP7A	99,70%	97,50%	100%	100%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATR	99,90%	99,40%	100%	100%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	100%	99,90%	100%	99,90%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B3GALT6	75,70%	69,70%	89,80%	81,60%	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B4GALT7	99,80%	97,40%	99,90%	98,60%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BANF1	98,30%	86,60%	100%	100%	Nestor-Guillermo progeria syndrome, 614008
BAP1	84,40%	83,00%	100%	100%	Tumor predisposition syndrome, 614327
BCOR	99,60%	97,40%	100%	99,90%	Microphthalmia, syndromic 2, 300166

BCS1L	100%	100%	100%	100%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	99,80%	98,30%	100%	100%	Bloom syndrome, 210900
BLOC1S3	98,50%	81,30%	100%	100%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	99,90%	97,10%	94,90%	94,90%	?Hermansky-pudlak syndrome 9, 614171
BMS1	66,70%	66,40%	100%	100%	?Aplasia cutis congenita, nonsyndromic, 107600
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
BRIP1	99,90%	99,00%	100%	100%	Fanconi anemia, complementation group J, 609054
BSCL2	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	100%	99,90%	100%	100%	Biotinidase deficiency, 253260
C1QA	100%	100%	100%	100%	C1q deficiency, 613652
C1QB	100%	100%	100%	100%	C1q deficiency, 613652
C1QC	100%	99,20%	100%	100%	C1q deficiency, 613652
C2CD3	95,80%	95,60%	95,90%	95,90%	Orofaciodigital syndrome XIV, 615948
C4orf26	100%	100%	100%	100%	Amelogenesis imperfecta, type IIA4, 614832
CA2	100%	100%	100%	100%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAPN12	94,00%	88,60%	100%	100%	No OMIM disease ID
CARD14	100%	99,10%	100%	100%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	99,90%	98,40%	100%	100%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	96,30%	94,50%	99,70%	98,20%	Immunodeficiency 58, 618131
CASP14	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 12, 617320
CAST	98,30%	95,40%	100%	100%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAV1	100%	100%	100%	100%	Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526
CAVIN1	100%	100%	100%	100%	Lipodystrophy, congenital generalized, type 4, 613327

CBL	97,30%	97,10%	100%	100%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBS	99,80%	98,30%	100%	100%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCBE1	99,80%	98,80%	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD151	100%	100%	100%	100%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057
CDAN1	100%	99,60%	100%	100%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDH3	100%	99,50%	100%	100%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDK4	100%	99,70%	100%	100%	No OMIM disease ID
CDKN2A	92,30%	92,10%	100%	100%	No OMIM disease ID
CDSN	100%	100%	100%	100%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CELSR1	94,70%	92,80%	99,60%	98,80%	No OMIM disease ID
CERS3	99,90%	98,90%	100%	100%	Ichthyosis, congenital, autosomal recessive 9, 615023
CHKB	100%	99,70%	100%	100%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	99,90%	98,90%	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHSY1	97,20%	95,70%	99,70%	98,00%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	100%	99,10%	100%	100%	Cocoon syndrome, 613630
CIB1	97,30%	93,60%	100%	100%	Epidermodysplasia verruciformis 3, 618267
CKAP2L	99,70%	98,60%	100%	100%	Filippi syndrome, 272440
CLDN1	100%	100%	100%	100%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100%	100%	100%	100%	HELIX syndrome, 617671
CNNM4	99,80%	98,90%	99,70%	98,80%	Jalili syndrome, 217080
COL14A1	100%	99,40%	100%	100%	No OMIM disease ID
COL17A1	98,70%	96,80%	100%	100%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, localisata variant, 226650
COL1A2	99,40%	97,00%	100%	100%	Ehlers-Danlos syndrome, cardiac valvular type, 225320 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL3A1	99,60%	97,60%	100%	100%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	98,80%	98,00%	100%	99,90%	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100%	99,50%	100%	100%	Ehlers-Danlos syndrome, classic type, 2, 130010

COL7A1	99,90%	99,10%	100%	100%	EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, Bart type, 132000 Transient bullous of the newborn, 131705 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 EBD, localisata variant, 0
COX4I2	100%	100%	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	77,40%	48,80%	100%	100%	Linear skin defects with multiple congenital anomalies 2, 300887
CPOX	99,90%	95,40%	100%	100%	Harderoporphyria, 121300 Coproporphyria, 121300
CST6	98,20%	92,50%	100%	100%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	100%	99,80%	100%	100%	Peeling skin syndrome 4, 607936
CTC1	100%	99,60%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTSA	100%	100%	100%	100%	Galactosialidosis, 256540
CTSB	100%	100%	100%	100%	No OMIM disease ID
CTSC	100%	100%	100%	100%	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CXCR4	100%	100%	100%	100%	WHIM syndrome, 193670 Myelokathexis, isolated, 0
CYLD	99,80%	98,00%	100%	100%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
CYP26C1	99,70%	97,10%	100%	99,80%	Focal facial dermal dysplasia 4, 614974
CYP4F22	100%	99,40%	100%	100%	Ichthyosis, congenital, autosomal recessive 5, 604777
DCAF17	98,90%	93,30%	100%	100%	Woodhouse-Sakati syndrome, 241080
DCLRE1C	100%	99,40%	100%	100%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDB2	99,60%	97,50%	100%	100%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DHCR7	100%	100%	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DKC1	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
DLX3	99,90%	98,40%	100%	100%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX5	100%	99,90%	100%	100%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DOCK6	99,30%	98,90%	100%	100%	Adams-Oliver syndrome 2, 614219

DOCK8	100%	99,60%	100%	100%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOLK	100%	100%	100%	100%	Congenital disorder of glycosylation, type Im, 610768
DSC2	99,80%	98,40%	100%	100%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	99,50%	96,80%	100%	100%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	99,00%	96,10%	100%	100%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	99,30%	97,50%	100%	100%	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG3	99,90%	99,40%	100%	100%	No OMIM disease ID
DSG4	100%	99,20%	100%	100%	Hypotrichosis 6, 607903
DSP	100%	99,60%	100%	100%	Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655 Arrhythmogenic right ventricular dysplasia 8, 607450 Epidermolysis bullosa, lethal acantholytic, 609638 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821
DSPP	96,80%	86,10%	100%	100%	Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	99,90%	99,40%	100%	100%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DTNBP1	99,80%	98,70%	100%	100%	Hermansky-Pudlak syndrome 7, 614076
DUSP6	100%	100%	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EBP	99,70%	95,80%	100%	100%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECM1	100%	99,60%	100%	100%	Urbach-Wiethe disease, 247100
EDA	98,10%	91,60%	100%	99,90%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	100%	99,90%	100%	100%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	99,90%	98,80%	100%	100%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDN3	100%	99,90%	100%	100%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880
EDNRA	100%	100%	100%	100%	Mandibulofacial dysostosis with alopecia, 616367

EDNRB	98,00%	93,80%	100%	100%	Waardenburg syndrome, type 4A, 277580 ABCD syndrome, 600501
EFEMP2	100%	100%	100%	100%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	100%	100%	100%	100%	Craniofrontonasal dysplasia, 304110
EIF2AK3	97,20%	94,50%	100%	100%	Wolcott-Rallison syndrome, 226980
ELN	99,80%	97,80%	100%	100%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	99,80%	97,60%	100%	100%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100%	99,50%	100%	100%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENAM	100%	100%	100%	100%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENG	99,60%	96,00%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	96,40%	91,20%	98,70%	97,80%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000
EPG5	99,50%	98,50%	100%	100%	Vici syndrome, 242840
EPS8L3	98,90%	97,30%	100%	100%	?Hypotrichosis 5, 612841
ERCC2	100%	99,70%	100%	100%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	100%	99,40%	100%	100%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC4	100%	99,90%	100%	100%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC5	100%	99,60%	100%	100%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	100%	100%	100%	100%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800
ERCC8	99,50%	95,80%	100%	100%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621

EVC	93,90%	88,60%	96,90%	94,80%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	97,70%	96,10%	100%	100%	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EXPH5	100%	100%	100%	100%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
FAM111B	100%	99,90%	100%	100%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM20A	99,60%	94,70%	100%	100%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	100%	100%	100%	99,80%	Raine syndrome, 259775
FAM83G	100%	100%	100%	100%	No OMIM disease ID
FAM83H	99,00%	95,00%	100%	100%	Amelogenesis imperfecta, type IIIA, 130900
FANCA	100%	99,40%	100%	100%	Fanconi anemia, complementation group A, 227650
FANCB	98,60%	94,10%	100%	100%	Fanconi anemia, complementation group B, 300514
FANCC	99,90%	99,30%	100%	100%	Fanconi anemia, complementation group C, 227645
FANCD2	99,50%	97,50%	98,80%	98,80%	Fanconi anemia, complementation group D2, 227646
FANCE	89,80%	85,10%	100%	99,90%	Fanconi anemia, complementation group E, 600901
FANCF	100%	100%	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	100%	99,90%	100%	100%	Fanconi anemia, complementation group G, 614082
FANCI	99,90%	99,20%	100%	100%	Fanconi anemia, complementation group I, 609053
FANCL	100%	98,60%	100%	100%	Fanconi anemia, complementation group L, 614083
FANCM	99,60%	97,30%	100%	100%	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
FAT4	100%	100%	100%	100%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN5	91,80%	91,80%	91,80%	91,80%	Macular degeneration, age-related, 3, 608895 ?Cutis laxa, autosomal dominant 2, 614434 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 Cutis laxa, autosomal recessive, type IA, 219100
FDPS	99,10%	93,50%	100%	100%	Porokeratosis 9, multiple types, 616631
FECH	100%	100%	100%	100%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	99,90%	97,90%	100%	100%	Kindler syndrome, 173650
FGF10	100%	99,80%	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	99,60%	97,50%	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	99,80%	95,10%	100%	100%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	100%	100%	100%	100%	Trichomegaly, 190330
FGF8	98,20%	88,90%	100%	99,60%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

FGFR1	100%	99,90%	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 HEARTsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	97,70%	97,10%	100%	100%	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FGFR3	99,80%	97,70%	100%	99,80%	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482
FH	92,10%	88,30%	100%	100%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800

FKBP10	98,80%	97,20%	100%	100%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	100%	99,90%	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLCN	100%	100%	100%	100%	Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500
FLG	100%	99,90%	100%	100%	Ichthyosis vulgaris, 146700
FLG2	100%	100%	99,90%	99,90%	Peeling skin syndrome 6, 618084
FLT4	99,20%	98,30%	100%	100%	Congenital heart defects, multiple types, 7, 618780 Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FNIP1	100%	99,80%	100%	100%	No OMIM disease ID
FOXC2	100%	96,70%	100%	99,80%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	96,90%	78,50%	99,90%	99,10%	Bamforth-Lazarus syndrome, 241850
FOXN1	100%	99,60%	100%	100%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	99,20%	95,50%	100%	100%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FREM1	99,90%	99,10%	100%	100%	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FUCA1	100%	99,90%	100%	100%	Fucosidosis, 230000
FZD6	100%	100%	100%	100%	Nail disorder, nonsyndromic congenital, 1, 161050
GALNS	100%	99,80%	100%	100%	Mucopolysaccharidosis IVA, 253000
GALNT3	99,80%	99,00%	100%	100%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GAN	100%	99,60%	100%	100%	Giant axonal neuropathy-1, 256850
GATA2	100%	98,30%	100%	100%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GDF2	100%	100%	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF5	100%	100%	100%	100%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A1, C, 615072 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A2, 112600 Du Pan syndrome, 228900 Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017

GGCX	100%	99,90%	100%	100%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GJA1	100%	100%	100%	100%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJB2	100%	100%	100%	100%	Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Bart-Pumphrey syndrome, 149200 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540
GJB3	100%	100%	100%	100%	Deafness, autosomal dominant 2B, 612644 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, 0 Deafness, autosomal dominant, with peripheral neuropathy, 0
GJB4	100%	100%	100%	100%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	100%	100%	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	78,20%	58,70%	96,90%	91,40%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GLA	99,80%	96,60%	100%	100%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	99,90%	97,40%	100%	100%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLMN	99,30%	94,70%	100%	100%	Glomuvenous malformations, 138000
GMPPA	100%	100%	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510

GNA11	99,90%	95,00%	100%	100%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GNA14	100%	100%	100%	100%	No OMIM disease ID
GNAQ	84,30%	74,90%	100%	100%	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000
GNAS	100%	99,90%	100%	99,90%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ib, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580
GORAB	100%	99,10%	100%	100%	Geroderma osteodysplasticum, 231070
GPNMB	100%	100%	100%	100%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR143	85,80%	76,40%	99,80%	97,90%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GRHL2	100%	100%	100%	100%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	100%	100%	100%	100%	Van der Woude syndrome 2, 606713
GSN	95,80%	93,50%	99,90%	99,30%	Amyloidosis, Finnish type, 105120
GTF2E2	100%	99,80%	100%	100%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	100%	99,60%	100%	100%	Trichothiodystrophy 3, photosensitive, 616395
HCCS	99,80%	97,60%	100%	100%	Linear skin defects with multiple congenital anomalies 1, 309801
HDAC8	100%	99,80%	100%	100%	Cornelia de Lange syndrome 5, 300882
HERC2	79,90%	77,20%	100%	100%	Mental retardation, autosomal recessive 38, 615516
HLCS	100%	100%	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	99,90%	99,40%	100%	100%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	78,60%	67,20%	100%	100%	?Microphthalmia, syndromic 13, 300915
HOXC13	100%	99,90%	100%	100%	Ectodermal dysplasia 9, hair/nail type, 614931
HPGD	100%	98,90%	100%	100%	Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HPS1	100%	100%	100%	100%	Hermansky-Pudlak syndrome 1, 203300
HPS3	99,70%	97,50%	100%	100%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100%	100%	100%	100%	Hermansky-Pudlak syndrome 4, 614073

HPS5	100%	99,70%	100%	100%	Hermansky-Pudlak syndrome 5, 614074
HPS6	97,10%	88,90%	100%	100%	Hermansky-Pudlak syndrome 6, 614075
HR	98,50%	95,60%	100%	100%	Hypotrichosis 4, 146550 Alopecia universalis, 203655 Atrichia with papular lesions, 209500
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
HTRA1	83,90%	80,00%	96,50%	92,10%	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 CARASIL syndrome, 600142
HYAL1	100%	100%	100%	100%	?Mucopolysaccharidosis type IX, 601492
IDUA	93,70%	86,80%	100%	100%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IFT122	100%	99,60%	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT43	100%	100%	100%	100%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IKBKG	84,10%	77,20%	100%	100%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IL17RA	100%	99,40%	100%	100%	Immunodeficiency 51, 613953
IL17RD	99,90%	99,10%	100%	100%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RN	100%	100%	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852
IL31RA	99,90%	99,90%	100%	100%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	100%	100%	100%	100%	Psoriasis 14, pustular, 614204
INSR	97,80%	94,70%	99,90%	99,20%	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200

MRE11	98,90%	93,30%	100%	100%	Ataxia-telangiectasia-like disorder 1, 604391
IRF4	100%	100%	100%	100%	No OMIM disease ID
IRF6	99,60%	95,90%	100%	100%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
ITGA3	99,50%	97,40%	100%	100%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	99,90%	98,90%	100%	100%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGB4	98,40%	96,20%	100%	100%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
ITGB6	97,20%	95,80%	100%	100%	Amelogenesis imperfecta, type IH, 616221
JUP	100%	99,50%	100%	100%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KANK2	100%	100%	100%	100%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KAT6B	99,90%	99,00%	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNH1	98,70%	98,70%	98,70%	98,70%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNK9	100%	100%	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KDF1	100%	99,80%	100%	100%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDSR	100%	99,50%	100%	100%	Erythrokeratodermia variabilis et progressiva 4, 617526
KIF11	97,60%	94,80%	100%	100%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIT	100%	99,60%	100%	100%	Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, somatic, 601626 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KITLG	100%	98,50%	100%	100%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
KLHL24	100%	100%	100%	100%	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KLK4	100%	100%	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KLLN	100%	100%	100%	100%	Cowden syndrome 4, 615107
KMT2D	100%	99,40%	100%	100%	Kabuki syndrome 1, 147920
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
KRT1	98,70%	95,60%	100%	100%	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962 Ichthyosis histrix, Curth-Macklin type, 146590 Epidermolytic hyperkeratosis, 113800
KRT10	100%	99,30%	100%	100%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT13	100%	100%	100%	100%	White sponge nevus 2, 615785
KRT14	89,00%	81,90%	100%	100%	Naegeli-Franceschetti-Jadassohn syndrome, 161000 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800
KRT16	74,20%	56,50%	100%	100%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200
KRT17	39,80%	22,80%	100%	100%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT2	100%	99,80%	100%	100%	Ichthyosis bullosa of Siemens, 146800
KRT4	100%	99,70%	100%	100%	White sponge nevus 1, 193900
KRT5	100%	100%	100%	100%	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, recessive 1, 601001

KRT6A	92,30%	87,70%	100%	100%	Pachyonychia congenita 3, 615726
KRT6B	93,60%	88,60%	100%	100%	Pachyonychia congenita 4, 615728
KRT6C	88,30%	81,30%	99,90%	99,80%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	100%	100%	100%	100%	?Hypotrichosis 13, 615896
KRT74	100%	100%	100%	100%	?Ectodermal dysplasia 7, hair/nail type, 614929 Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981
KRT75	100%	100%	100%	100%	No OMIM disease ID
KRT81	99,20%	94,20%	100%	100%	Monilethrix, 158000
KRT83	96,60%	84,40%	100%	100%	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000
KRT85	99,00%	93,60%	100%	100%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	99,70%	96,30%	100%	100%	Monilethrix, 158000
KRT9	99,20%	95,00%	100%	100%	Palmoplantar keratoderma, epidermolytic, 144200
LAMA3	100%	99,70%	100%	100%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660 Epidermolysis bullosa, generalized atrophic benign, 226650
LAMB3	100%	99,60%	100%	100%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	99,80%	98,00%	100%	100%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700
LAMTOR2	100%	99,70%	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LDHA	95,00%	91,70%	100%	100%	Glycogen storage disease XI, 612933
LDLRAP1	98,80%	94,20%	100%	100%	Hypercholesterolemia, familial, 4, 603813
LEMD3	99,90%	98,70%	100%	100%	Osteopoikilosis with or without melorheostosis, 166700 Buschke-Ollendorff syndrome, 166700
LIPH	100%	99,80%	100%	100%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	100%	98,90%	100%	100%	Ichthyosis, congenital, autosomal recessive 8, 613943
LMBRD1	98,50%	93,90%	100%	100%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMNA	97,40%	91,90%	100%	100%	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210

					Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMX1B	99,60%	96,30%	100%	100%	Nail-patella syndrome, 161200
LONP1	100%	99,80%	100%	100%	CODAS syndrome, 600373
LOR	99,00%	80,80%	100%	100%	Vohwinkel syndrome with ichthyosis, 604117
LPAR6	99,60%	97,80%	100%	100%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN2	100%	100%	100%	100%	Majeed syndrome, 609628
LRMDA	96,80%	95,60%	99,60%	99,60%	Albinism, oculocutaneous, type VII, 615179
LSS	100%	99,90%	100%	100%	Alopecia-mental retardation syndrome 4, 618840 Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP3	99,60%	98,10%	100%	100%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	99,90%	97,50%	100%	100%	Cutis laxa, autosomal recessive, type IC, 613177
LYST	99,60%	98,30%	100%	100%	Chediak-Higashi syndrome, 214500
LYZ	100%	100%	100%	100%	Amyloidosis, renal, 105200
MAP2K1	99,80%	97,10%	100%	100%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	98,50%	95,10%	100%	100%	Cardiofaciocutaneous syndrome 4, 615280
MBTPS2	100%	99,00%	100%	100%	Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MED12	99,80%	96,70%	100%	100%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MEFV	99,90%	98,60%	96,40%	96,40%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MGP	98,70%	95,10%	100%	100%	Keutel syndrome, 245150
MITF	100%	99,90%	100%	100%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MLH1	100%	99,90%	100%	100%	Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 2, 609310

MLPH	100%	98,80%	100%	100%	Griscelli syndrome, type 3, 609227
MMACHC	100%	100%	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP2	100%	100%	100%	100%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100%	100%	100%	100%	Amelogenesis imperfecta, type IIA2, 612529
MPLKIP	100%	99,40%	100%	100%	Trichothiodystrophy 4, nonphotosensitive, 234050
MSH2	99,80%	97,70%	100%	100%	Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 1, 120435
MSX1	96,90%	89,30%	100%	100%	Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MTOR	100%	99,50%	100%	100%	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MUTYH	100%	100%	100%	100%	Gastric cancer, somatic, 613659 Adenomas, multiple colorectal, 608456
MVD	99,90%	98,30%	100%	100%	Porokeratosis 7, multiple types, 614714
MVK	90,90%	90,50%	90,50%	90,50%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYH8	100%	99,60%	100%	100%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYO5A	99,80%	98,90%	100%	100%	Griscelli syndrome, type 1, 214450
NAA10	99,70%	98,50%	99,90%	99,90%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAGA	100%	100%	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NBAS	100%	99,60%	100%	100%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NCSTN	100%	99,80%	100%	100%	Acne inversa, familial, 1, 142690
NDUFB11	99,50%	96,50%	100%	99,50%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NECTIN1	100%	99,90%	100%	100%	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NECTIN4	100%	100%	100%	100%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK11	99,90%	98,80%	100%	100%	No OMIM disease ID

NEK9	100%	99,60%	100%	100%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NF1	92,60%	90,20%	100%	100%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210
NFKBIA	95,20%	88,00%	100%	100%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHP2	100%	100%	100%	100%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIPAL4	100%	99,10%	100%	100%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	98,90%	97,00%	100%	100%	Cornelia de Lange syndrome 1, 122470
NLRP1	99,60%	98,00%	100%	100%	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388 ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803
NLRP3	100%	99,90%	100%	100%	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NME1	100%	100%	100%	100%	No OMIM disease ID
NOD2	100%	99,90%	100%	100%	Blau syndrome, 186580
NOP10	100%	99,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH1	99,20%	97,20%	100%	100%	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
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
NSD1	100%	99,90%	100%	100%	Sotos syndrome 1, 117550
NSDHL	100%	98,70%	100%	100%	CHILD syndrome, 308050 CK syndrome, 300831
OCA2	99,90%	98,70%	100%	100%	Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200

ODAM	99,80%	98,70%	100%	100%	No OMIM disease ID
OFD1	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OSMR	100%	99,70%	100%	100%	Amyloidosis, primary localized cutaneous, 1, 105250
PADI3	100%	100%	100%	100%	Uncombable hair syndrome, 191480
PAH	100%	100%	100%	100%	Phenylketonuria, 261600
PALB2	100%	100%	100%	100%	Fanconi anemia, complementation group N, 610832
PAX3	100%	99,90%	100%	100%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX9	99,70%	99,60%	100%	100%	Tooth agenesis, selective, 3, 604625
PCNA	100%	98,40%	100%	100%	?Ataxia-telangiectasia-like disorder 2, 615919
PDGFB	100%	99,30%	100%	100%	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174
PDGFRB	99,20%	97,50%	100%	100%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PEPD	100%	98,80%	100%	100%	Prolidase deficiency, 170100
PERP	100%	100%	100%	100%	No OMIM disease ID
PEX7	87,80%	80,70%	91,30%	91,30%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	100%	99,60%	99,90%	99,20%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	99,90%	98,80%	100%	100%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHYH	100%	99,60%	100%	100%	Refsum disease, 266500
TALDO1	100%	97,90%	100%	100%	Transaldolase deficiency, 606003
TAP1	100%	99,20%	100%	100%	Bare lymphocyte syndrome, type I, 604571
TAP2	99,90%	99,30%	100%	100%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,50%	95,50%	96,60%	96,60%	Bare lymphocyte syndrome, type I, 604571
TAT	100%	100%	100%	100%	Tyrosinemia, type II, 276600
PIEZO1	99,90%	98,80%	100%	100%	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380

PIGA	93,80%	86,70%	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	93,80%	91,50%	98,80%	98,80%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	100%	100%	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	100%	99,80%	100%	100%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Macrodactyly, somatic, 155500 Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
PITX2	99,90%	97,70%	100%	100%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PKP1	100%	99,10%	100%	100%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLCD1	99,90%	97,80%	100%	100%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCG2	100%	99,80%	100%	100%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLEC	100%	99,80%	100%	100%	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex with muscular dystrophy, 226670 ?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950
PLG	87,80%	87,50%	100%	100%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	99,60%	94,90%	100%	99,50%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	100%	98,40%	100%	100%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD3	99,80%	98,00%	100%	100%	Lysyl hydroxylase 3 deficiency, 612394
PMS2	84,30%	82,80%	100%	100%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
PMVK	100%	100%	100%	100%	Porokeratosis 1, multiple types, 175800
PNPLA1	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 10, 615024

PNPLA2	99,70%	96,10%	100%	100%	Neutral lipid storage disease with myopathy, 610717
POC1A	100%	100%	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POFUT1	100%	99,00%	100%	100%	Dowling-Degos disease 2, 615327
POGLUT1	99,40%	94,60%	100%	100%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POLD1	98,50%	95,20%	100%	100%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLH	100%	99,60%	100%	100%	Xeroderma pigmentosum, variant type, 278750
POLR1C	99,30%	95,50%	90,70%	90,70%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,60%	91,60%	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	100%	99,70%	100%	100%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	99,90%	98,60%	100%	100%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	100%	100%	100%	100%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMP	100%	99,10%	100%	100%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
PORCN	100%	99,10%	100%	100%	Focal dermal hypoplasia, 305600
POT1	99,90%	99,00%	100%	100%	No OMIM disease ID
PPOX	99,70%	96,80%	100%	100%	Porphyria variegata, 176200
PQBP1	100%	100%	100%	100%	Renpenning syndrome, 309500
PRKAR1A	99,30%	93,50%	100%	100%	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0
PSEN1	100%	100%	100%	100%	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 ?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694
PSENEN	100%	100%	100%	100%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB8	99,90%	98,50%	100%	100%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSTPIP1	100%	99,10%	100%	99,90%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416

PTCH1	99,20%	97,60%	99,90%	99,80%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	99,90%	99,00%	100%	100%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTDSS1	100%	100%	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	99,50%	97,00%	100%	100%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
PTHLH	99,70%	98,40%	100%	100%	Brachydactyly, type E2, 613382
PTPN11	99,10%	93,70%	100%	100%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN14	99,70%	97,40%	100%	100%	Choanal atresia and lymphedema, 613611
PTPRF	100%	99,70%	100%	100%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PYCR1	99,90%	97,70%	100%	100%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	100%	99,50%	100%	100%	Carpenter syndrome, 201000
RAB27A	100%	100%	100%	100%	Griscelli syndrome, type 2, 607624
RAD21	99,20%	96,60%	100%	100%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	97,50%	91,60%	100%	100%	Nijmegen breakage syndrome-like disorder, 613078
RAF1	100%	100%	100%	100%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RAG1	100%	100%	100%	100%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 Combined cellular and humoral immune defects with granulomas, 233650
RAG2	100%	100%	100%	100%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAI1	100%	100%	100%	100%	Smith-Magenis syndrome, 182290

RBBP8	100%	99,70%	100%	100%	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBM28	100%	100%	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBP4	99,90%	97,70%	100%	100%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	98,40%	92,80%	100%	100%	Adams-Oliver syndrome 3, 614814
RECQL4	99,80%	98,10%	100%	99,90%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
RHBDF2	99,90%	98,60%	100%	100%	Tylosis with esophageal cancer, 148500
RHOA	81,20%	80,70%	80,70%	80,70%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RIN2	100%	100%	100%	100%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RMRP	NC	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RIPK4	100%	99,90%	100%	100%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RNASEH2A	100%	100%	100%	100%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	96,00%	92,50%	100%	99,80%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100%	99,50%	100%	100%	Aicardi-Goutieres syndrome 3, 610329
RNU4ATAC	NC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROGDI	98,40%	95,20%	99,90%	99,10%	Kohlschutter-Tonz syndrome, 226750
RPL21	88,80%	71,70%	100%	100%	Hypotrichosis 12, 615885
RSPO1	100%	99,90%	100%	100%	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
RSPO4	100%	100%	100%	100%	Anonychia congenita, 206800
RTEL1	99,50%	96,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RUNX2	72,20%	72,20%	100%	100%	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600
SAMD9	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455

SAMHD1	100%	99,60%	100%	100%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SART3	99,60%	98,60%	100%	100%	No OMIM disease ID
SASH1	99,90%	98,70%	100%	100%	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 Dyschromatosis universalis hereditaria 1, 127500
SAT1	99,90%	98,50%	100%	99,90%	No OMIM disease ID
SATB2	99,70%	97,40%	100%	100%	Glass syndrome, 612313
SCN10A	100%	99,70%	100%	100%	Episodic pain syndrome, familial, 2, 615551
SCN11A	99,80%	98,30%	100%	100%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	99,30%	97,80%	100%	100%	Small fiber neuropathy, 133020 HSAN2D, autosomal recessive, 243000 Paroxysmal extreme pain disorder, 167400 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863
SDR9C7	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23B	99,90%	99,30%	100%	100%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SERPINB7	100%	99,90%	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	95,00%	95,00%	100%	100%	Peeling skin syndrome 5, 617115
SERPING1	99,70%	97,50%	100%	100%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	100%	98,30%	100%	100%	Osteogenesis imperfecta, type X, 613848
SGPL1	100%	100%	100%	100%	Nephrotic syndrome, type 14, 617575
SHOC2	99,90%	99,40%	100%	100%	Noonan syndrome-like with loose anagen hair, 607721
SKI	99,30%	94,90%	100%	99,40%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	100%	99,80%	100%	100%	Trichohepatoenteric syndrome 2, 614602
SLC17A9	96,30%	95,40%	100%	100%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC24A4	100%	99,80%	100%	100%	Amelogenesis imperfecta, type IIA5, 615887
SLC24A5	99,90%	99,10%	100%	100%	Albinism, oculocutaneous, type VI, 113750
SLC26A2	100%	100%	100%	100%	De la Chapelle dysplasia, 256050 Atelosteogenesis, type II, 256050 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Diastrophic dysplasia, 222600 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900

SLC27A4	100%	99,80%	100%	100%	Ichthyosis prematurity syndrome, 608649
SLC29A3	100%	99,60%	100%	100%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A10	97,70%	97,70%	100%	100%	Arterial tortuosity syndrome, 208050
SLC39A13	99,80%	98,20%	100%	100%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A4	99,50%	95,50%	100%	100%	Acrodermatitis enteropathica, 201100
SLC45A2	100%	99,90%	100%	100%	Albinism, oculocutaneous, type IV, 606574
SLC4A4	99,80%	99,20%	100%	100%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A19	100%	100%	100%	100%	Iminoglycinuria, digenic, 242600 HEARTnup disorder, 234500 Hyperglycinuria, 138500
SLC7A7	100%	99,90%	100%	100%	Lysinuric protein intolerance, 222700
SLCO2A1	100%	99,40%	100%	100%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLURP1	100%	99,30%	100%	100%	Meleda disease, 248300
SLX4	100%	99,80%	100%	100%	Fanconi anemia, complementation group P, 613951
SMAD3	99,90%	99,00%	100%	100%	Loeys-Dietz syndrome 3, 613795
SMARCA2	96,70%	96,20%	97,40%	96,80%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	99,90%	99,00%	100%	100%	Coffin-Siris syndrome 4, 614609
SMARCAD1	99,30%	95,80%	100%	100%	Huriez syndrome, 181600 Basan syndrome, 129200 Adermatoglyphia, 136000
SMARCAL1	100%	99,90%	100%	100%	Schimke immunoosseous dysplasia, 242900
SMARCB1	100%	100%	100%	100%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMO	97,80%	94,70%	100%	100%	Curry-Jones syndrome, somatic mosaic, 601707 Basal cell carcinoma, somatic, 605462
SMOC2	77,00%	76,70%	100%	100%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	100%	99,10%	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	100%	100%	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNRPE	99,50%	92,60%	100%	100%	Hypotrichosis 11, 615059
SNX10	96,20%	95,70%	100%	99,60%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	99,80%	98,40%	100%	100%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOX10	99,90%	97,90%	100%	100%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266

SOX18	70,70%	55,20%	96,10%	92,60%	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SOX2	100%	100%	100%	100%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SP7	100%	99,80%	100%	100%	Osteogenesis imperfecta, type XII, 613849
SPINK5	99,90%	99,50%	100%	100%	Netherton syndrome, 256500
SPINT2	98,50%	83,80%	100%	100%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPRED1	100%	98,90%	100%	100%	Legius syndrome, 611431
SPRY4	100%	100%	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRD5A3	99,90%	99,10%	100%	100%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
ST14	99,90%	98,60%	100%	100%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL5	85,00%	84,20%	98,70%	98,40%	Salt and pepper developmental regression syndrome, 609056
STAMBP	100%	99,40%	100%	100%	Microcephaly-capillary malformation syndrome, 614261
STAT3	100%	99,80%	100%	100%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	100%	98,50%	100%	100%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
STIM1	99,80%	98,00%	100%	100%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STK11	100%	99,30%	100%	100%	Testicular tumor, somatic, 273300 Peutz-Jeghers syndrome, 175200 Pancreatic cancer, somatic, 260350 Melanoma, malignant, somatic, 0
STS	99,70%	98,10%	100%	99,90%	Ichthyosis, X-linked, 308100
SUFU	100%	100%	100%	100%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 Joubert syndrome 32, 617757
SULT2B1	100%	100%	100%	100%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	97,50%	90,80%	100%	100%	Multiple sulfatase deficiency, 272200
TBC1D24	100%	100%	100%	100%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TBX3	99,20%	96,80%	100%	100%	Ulnar-mammary syndrome, 181450

TCHH	100%	98,80%	100%	100%	?Uncombable hair syndrome 3, 617252
TCIRG1	97,60%	90,10%	100%	100%	Osteopetrosis, autosomal recessive 1, 259700
TEK	100%	100%	100%	100%	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	99,90%	97,30%	100%	100%	No OMIM disease ID
TERT	96,20%	94,50%	100%	100%	No OMIM disease ID
TFAP2A	99,40%	94,30%	100%	100%	Branchiooculofacial syndrome, 113620
TGFB2	100%	100%	100%	100%	Loeys-Dietz syndrome 4, 614816
TGFBR1	93,70%	93,60%	99,00%	96,30%	Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	100%	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
TGM1	100%	99,90%	100%	100%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	100%	99,70%	100%	100%	?Uncombable hair syndrome 2, 617251
TGM5	100%	99,70%	100%	100%	Peeling skin syndrome 2, 609796
TINF2	100%	100%	100%	100%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TMC6	100%	99,30%	100%	100%	Epidermodysplasia verruciformis, 226400
TMC8	100%	98,70%	100%	100%	Epidermodysplasia verruciformis 2, 618231
TMEM165	100%	100%	100%	100%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	99,70%	95,30%	100%	100%	STING-associated vasculopathy, infantile-onset, 615934
TNFRSF11A	94,60%	93,30%	99,20%	98,00%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	100%	100%	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF1A	90,60%	87,60%	92,80%	92,80%	Periodic fever, familial, 142680
TNFSF11	100%	99,90%	100%	100%	Osteopetrosis, autosomal recessive 2, 259710
TNXB	99,60%	95,10%	100%	100%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP63	100%	100%	100%	100%	Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285
TPCN2	95,10%	92,40%	100%	100%	No OMIM disease ID

TREX1	100%	100%	100%	100%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	100%	100%	100%	100%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM37	98,60%	98,10%	98,70%	98,70%	Mulibrey nanism, 253250
TRPM4	100%	99,50%	100%	100%	Erythrokeratodermia veriabilis et progressiva 6, 618531 Progressive familial heart block, type IB, 604559
TRPS1	100%	99,90%	100%	100%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	99,80%	98,50%	97,10%	97,10%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TSC1	99,80%	98,70%	100%	100%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	100%	99,60%	100%	100%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSPEAR	100%	99,20%	97,90%	97,90%	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TTC37	100%	99,30%	100%	100%	Trichohepatoenteric syndrome 1, 222470
TTI2	100%	100%	100%	100%	Mental retardation, autosomal recessive 39, 615541
TWIST2	100%	100%	100%	100%	Barber-Say syndrome, 209885 Ablepharon-macrostomia syndrome, 200110 Focal facial dermal dysplasia 3, Setleis type, 227260
TYR	100%	100%	100%	100%	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 Albinism, oculocutaneous, type IA, 203100
TYRP1	100%	99,80%	100%	100%	Albinism, oculocutaneous, type III, 203290
UBE2A	99,70%	96,00%	100%	99,70%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBR1	99,90%	99,10%	98,00%	98,00%	Johanson-Blizzard syndrome, 243800
UROD	98,90%	96,10%	100%	100%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100%	99,90%	100%	100%	Porphyria, congenital erythropoietic, 263700
USB1	100%	99,40%	100%	100%	Poikiloderma with neutropenia, 604173
UVSSA	99,30%	98,80%	99,40%	99,30%	UV-sensitive syndrome 3, 614640
VDR	99,90%	99,00%	100%	100%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	100%	100%	100%	100%	Lymphatic malformation 4, 615907

VHL	96,30%	91,40%	100%	100%	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
VPS13B	99,50%	98,20%	99,50%	99,40%	Cohen syndrome, 216550
VPS33B	100%	100%	100%	100%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WAS	95,90%	85,30%	100%	99,80%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WDR19	100%	99,40%	100%	100%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	99,80%	98,90%	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR72	96,80%	96,40%	96,90%	96,90%	Amelogenesis imperfecta, type IIA3, 613211
WIPF1	100%	99,90%	100%	100%	?Wiskott-Aldrich syndrome 2, 614493
WNT10A	100%	99,40%	100%	100%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100%	99,40%	100%	100%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT5A	100%	100%	100%	100%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	100%	100%	100%	100%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	100%	100%	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	99,90%	98,80%	100%	100%	Werner syndrome, 277700
XPA	99,60%	95,60%	100%	100%	Xeroderma pigmentosum, group A, 278700
XPC	100%	100%	100%	100%	Xeroderma pigmentosum, group C, 278720
XYLT1	97,40%	89,60%	98,10%	94,80%	Desbuquois dysplasia 2, 615777
XYLT2	100%	98,30%	96,70%	96,70%	Spondyloocular syndrome, 605822
YWHAZ	81,20%	71,70%	100%	100%	No OMIM disease ID
ZBTB20	100%	100%	100%	100%	PriINTELLECTUAL DISABILITYose syndrome, 259050
ZMPSTE24	100%	99,90%	100%	100%	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZNF469	100%	100%	100%	100%	Brittle cornea syndrome 1, 229200

ZNF592	100%	99,60%	100%	100%	No OMIM disease ID
ZNF750	100%	100%	100%	100%	Seborrhea-like dermatitis with psoriasiform elements, 610227

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