SKIN DISORDERS GENE PANEL DG 2.16 (619 genes)

Releasedate: 07-06-2019

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated phenotype description and OMIM disease ID
AAAS	102,3	100.0%	99.6%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	134,7	99.9%	99.8%	Keratoderma, palmoplantar, punctate type IA, 148600
ABCA12	129,3	99.6%	98.4%	Ichthyosis, congenital, autosomal recessive 4A, 601277 Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500
ABCB6	127,9	100.0%	99.9%	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 [Blood group, Langereis system], 111600
ABCC6	109,1	93.6%	92.8%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	142,6	100.0%	99.7%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABHD5	180,9	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ACD	159,6	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACTA2	87,3	99.9%	98.6%	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	80,5	100.0%	99.7%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACVRL1	113,6	100.0%	98.4%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADA2	83,5	99.9%	97.6%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM10	121,4	94.8%	93.4%	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590

ADAM17	119	99.8%	98.6%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAMTS10	122,8	100.0%	99.8%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	109,2	97.6%	92.3%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS2	126,3	100.0%	99.6%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	136,6	100.0%	99.8%	?Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	115,9	99.0%	96.3%	Geleophysic dysplasia 1, 231050
ADAR	109,2	99.9%	99.3%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	142,7	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGPAT2	162,6	99.1%	94.8%	Lipodystrophy, congenital generalized, type 1, 608594
AIRE	102,3	100.0%	99.9%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKT1	152,6	100.0%	99.4%	Breast cancer, somatic, 114480 Colorectal cancer, somatic, 114500 Cowden syndrome 6, 615109 Ovarian cancer, somatic, 167000 Proteus syndrome, somatic, 176920 {Schizophrenia, susceptibility to}, 181500
AKT3	82,3	99.2%	94.2%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	94,9	99.5%	94.7%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	74,7	98.9%	94.7%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	113,7	100.0%	99.8%	Cutis laxa, autosomal dominant 3, 616603 Cutis laxa, autosomal recessive, type IIIA, 219150 Spastic paraplegia 9A, autosomal dominant, 601162 Spastic paraplegia 9B, autosomal recessive, 616586
ALDH3A2	113,5	95.3%	94.3%	Sjogren-Larsson syndrome, 270200
ALDOB	135,3	100.0%	99.3%	Fructose intolerance, hereditary, 229600
ALOX12B	125,6	100.0%	99.8%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	124,7	100.0%	99.4%	Ichthyosis, congenital, autosomal recessive 3, 606545

ALPL	154,8	100.0%	99.7%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALX4	157,1	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMELX	85,7	98.9%	93.5%	Amelogenesis imperfecta, type 1E, 301200
ANKRD11	119,6	99.2%	97.1%	KBG syndrome, 148050
ANOS1	76,7	91.7%	88.0%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	108,3	99.0%	96.9%	GAPO syndrome, 230740 {?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	119,3	99.9%	98.6%	Hyaline fibromatosis syndrome, 228600
AP1S3	110,7	90.5%	90.4%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	112,1	99.5%	96.5%	Hermansky-Pudlak syndrome 2, 608233
APC	141,4	99.9%	99.6%	Adenoma, periampullary, somatic, 0 Adenomatous polyposis coli, 175100 Brain tumor-polyposis syndrome 2, 175100 Colorectal cancer, somatic, 114500 Desmoid disease, hereditary, 135290 Gardner syndrome, 175100 Gastric cancer, somatic, 613659 Hepatoblastoma, somatic, 114550
APCDD1	171,4	100.0%	99.3%	Hypotrichosis 1, 605389
AQP5	122,4	100.0%	99.5%	Palmoplantar keratoderma, Bothnian type, 600231
ARHGAP31	141,4	99.8%	98.7%	Adams-Oliver syndrome 1, 100300
ARID1A	134,4	99.4%	98.4%	Coffin-Siris syndrome 2, 614607
ARID1B	139,6	99.5%	99.2%	Coffin-Siris syndrome 1, 135900
ASIP	150	100.0%	100.0%	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 [Skin/hair/eye pigmentation 9, dark/light hair], 611742
ASL	123,6	100.0%	98.5%	Argininosuccinic aciduria, 207900
ASXL1	132,4	100.0%	99.5%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	138,1	99.7%	99.1%	Bainbridge-Ropers syndrome, 615485

ATIC	113,9	100.0%	99.7%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP2A2	143,2	100.0%	99.8%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	111,6	99.9%	99.3%	Hailey-Hailey disease, 169600
ATP6V0A2	117,4	99.9%	99.0%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	111,2	99.5%	96.7%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATR	144,6	99.8%	98.6%	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 Seckel syndrome 1, 210600
AXIN2	124,2	100.0%	99.9%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B3GALT6	81,7	82.6%	77.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B4GALT7	123,9	99.8%	98.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BANF1	51,1	96.6%	84.1%	Nestor-Guillermo progeria syndrome, 614008
BAP1	104,8	85.0%	82.9%	Tumor predisposition syndrome, 614327
BCOR	102,7	98.8%	95.3%	Microphthalmia, syndromic 2, 300166
BCS1L	147,9	100.0%	100.0%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	111	99.6%	98.0%	Bloom syndrome, 210900
BLOC1S3	67,4	100.0%	99.9%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	101,1	99.2%	95.1%	?Hermansky-pudlak syndrome 9, 614171
BMS1	76,4	66.8%	65.6%	?Aplasia cutis congenita, nonsyndromic, 107600
BRAF	72,5	92.4%	80.2%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic, 0 LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic, 0 Nonsmall cell lung cancer, somatic, 0 Noonan syndrome 7, 613706

BRIP1	125,8	99.7%	98.8%	Fanconi anemia, complementation group J, 609054 {Breast cancer, early-onset, susceptibility to}, 114480
BSCL2	105,2	100.0%	100.0%	Encephalopathy, progressive, with or without lipodystrophy, 615924 Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VA, 600794 Silver spastic paraplegia syndrome, 270685
BTD	126,6	99.9%	99.7%	Biotinidase deficiency, 253260
C1QA	196,1	100.0%	100.0%	C1q deficiency, 613652
C1QB	161,5	100.0%	100.0%	C1q deficiency, 613652
C1QC	187	100.0%	99.6%	C1q deficiency, 613652
C2CD3	116,9	95.8%	95.2%	Orofaciodigital syndrome XIV, 615948
C4orf26	NC	NC	NC	Amelogenesis imperfecta, type IIA4, 614832
CA2	137,4	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAPN12	105,4	98.1%	93.2%	No OMIM phenotype Modifying factor in ichthyosis
CARD14	124,5	100.0%	99.2%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	133,2	100.0%	99.5%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	137,3	98.4%	96.6%	Immunodeficiency 58, 618131
CASP14	85,8	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 12, 617320
CAST	112,4	99.8%	97.1%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAV1	189,3	100.0%	100.0%	?Lipodystrophy, congenital generalized, type 3, 612526 Lipodystrophy, familial partial, type 7, 606721 Pulmonary hypertension, primary, 3, 615343
CAVIN1	174,1	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CBL	126	97.3%	97.0%	?Juvenile myelomonocytic leukemia, 607785 Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563
CBS	123,3	99.9%	99.0%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCBE1	75,3	99.8%	99.1%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510

CD151	122,2	100.0%	100.0%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CDAN1	112,4	100.0%	99.6%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDH3	140,5	100.0%	99.8%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDK4	100	100.0%	99.1%	{Melanoma, cutaneous malignant, 3}, 609048
CDKN2A	121,7	92.3%	92.3%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, 0 Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDSN	131	100.0%	100.0%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CELSR1	173,9	98.4%	96.0%	No OMIM phenotype Congenital heart defects (Qiao (2016) Clin Sci (Lond)) Craniorachischisis (Robinson (2012) Hum Mutat 33,440) Neural tube defects (Qiao (2016) Clin Sci (Lond)) Spina bifida (Lei (2014) PLoS One 9,e92207) Lymphoedema (Gonzal
CERS3	95,2	99.8%	98.2%	Ichthyosis, congenital, autosomal recessive 9, 615023
CHKB	115,4	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	160,6	99.9%	98.9%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHSY1	125,9	99.3%	97.9%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	126,5	99.8%	99.2%	Cocoon syndrome, 613630
CIB1	122,3	99.3%	96.3%	Epidermodysplasia verruciformis 3, 618267
CKAP2L	156	99.9%	98.9%	Filippi syndrome, 272440
CLDN1	122,5	100.0%	100.0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	138,2	100.0%	100.0%	HELIX syndrome, 617671
CNNM4	161,6	100.0%	99.5%	Jalili syndrome, 217080
COL14A1	123,6	99.9%	98.7%	No OMIM phenotype Keratoderma, palmoplantar, punctate (Guo (2012) J Med Genet 49,563)

COL17A1	104,5	99.3%	96.9%	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL1A2	93,3	98.5%	94.6%	Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Ehlers-Danlos syndrome, cardiac valvular type, 225320 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL3A1	99,2	99.3%	96.8%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	136,4	99.9%	98.9%	Ehlers-Danlos syndrome, classic type, 1, 130000
COL5A2	100,2	99.9%	99.4%	Ehlers-Danlos syndrome, classic type, 2, 130010
COL7A1	139,7	99.8%	98.9%	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant, 0 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa dystrophica, AR, 226600 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 Toenail dystrophy, isolated, 607523 Transient bullous of the newborn, 131705
COX4I2	116,5	100.0%	99.6%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	38,6	62.3%	33.6%	Linear skin defects with multiple congenital anomalies 2, 300887
CPOX	134,1	99.5%	97.2%	Coproporphyria, 121300 Harderoporphyria, 121300
CST6	116,8	100.0%	98.2%	No OMIM phenotype Epilepsy, progressive myoclonus (Lalioti (1997) Am J Hum Genet 60,342) Unverricht-Lundborg disease (Canafoglia (2012) Epilepsia 53,2120)
CSTA	110,9	99.9%	99.1%	Peeling skin syndrome 4, 607936
CTC1	105,5	100.0%	99.3%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTSA	132,9	100.0%	99.9%	Galactosialidosis, 256540
CTSB	120,8	100.0%	100.0%	Keratolytic winter erythema, 148370

CTSC	116,2	100.0%	100.0%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CXCR4	122,8	100.0%	100.0%	Myelokathexis, isolated, 0 WHIM syndrome, 193670
CYLD	109,2	99.7%	97.8%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP26C1	133	100.0%	99.9%	Focal facial dermal dysplasia 4, 614974
CYP4F22	115,3	100.0%	98.8%	Ichthyosis, congenital, autosomal recessive 5, 604777
DCAF17	90,4	99.9%	97.9%	Woodhouse-Sakati syndrome, 241080
DCLRE1C	138,9	99.9%	97.2%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDB2	147,3	99.8%	98.4%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DHCR7	144,9	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DKC1	91,2	99.8%	97.7%	Dyskeratosis congenita, X-linked, 305000
DLX3	146,7	100.0%	99.0%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DLX5	145,3	100.0%	99.8%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DOCK6	121,5	99.6%	98.6%	Adams-Oliver syndrome 2, 614219
DOCK8	112,1	100.0%	99.6%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOLK	157,2	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DSC2	123,7	99.6%	97.2%	Arrhythmogenic right ventricular dysplasia 11, 610476 Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476
DSC3	95,6	99.3%	97.3%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	89,7	99.7%	97.2%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	131	99.4%	97.7%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG3	133,9	100.0%	99.4%	No OMIM phenotype

DSG4	158,3	99.8%	99.0%	Hypotrichosis 6, 607903
DSP	140,6	100.0%	99.6%	Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 Epidermolysis bullosa, lethal acantholytic, 609638 Keratosis palmoplantaris striata II, 612908 Skin fragility-woolly hair syndrome, 607655
DSPP	79	98.4%	93.8%	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	144,8	99.9%	99.2%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DTNBP1	113,4	99.8%	97.9%	Hermansky-Pudlak syndrome 7, 614076
DUSP6	164,1	100.0%	100.0%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EBP	63,2	99.5%	95.2%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECM1	158,6	99.9%	99.0%	Urbach-Wiethe disease, 247100
EDA	102	95.6%	85.7%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	126,6	100.0%	100.0%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	89,8	99.7%	98.3%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN3	135,2	100.0%	100.0%	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	150,7	100.0%	99.8%	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300

EDNRB	120,9	96.9%	92.5%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EFEMP2	129,4	100.0%	100.0%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	116,7	100.0%	99.9%	Craniofrontonasal dysplasia, 304110
EIF2AK3	134,2	99.5%	96.3%	Wolcott-Rallison syndrome, 226980
ELN	103,1	100.0%	98.9%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	85,9	99.8%	97.5%	No OMIM phenotype
ELOVL4	104,4	99.9%	99.1%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110
ENAM	139,5	100.0%	100.0%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	128,4	99.9%	98.7%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	129,2	97.5%	93.3%	Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522 Hypophosphatemic rickets, autosomal recessive, 2, 613312 {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 {Obesity, susceptibility to}, 601665
EPG5	110,3	99.3%	97.9%	Vici syndrome, 242840
EPS8L3	104,6	99.4%	97.1%	No OMIM phenotype Marie Unna hereditary hypotrichosis (Zhang (2012) J Med Genet 49,727)
ERCC2	128	100.0%	99.8%	?Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	92	99.9%	98.4%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	132	100.0%	99.8%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965
ERCC5	126,3	99.9%	99.5%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780

ERCC6	158,2	100.0%	99.9%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 De Sanctis-Cacchione syndrome, 278800 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 {Lung cancer, susceptibility to}, 211980 {Macular degeneration, age-related, susceptibility to, 5}, 613761
ERCC8	82,8	98.9%	90.0%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
EVC	106,3	95.9%	92.4%	?Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EVC2	110,2	99.4%	96.3%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXPH5	163,6	100.0%	99.9%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
FAM111B	157,9	99.9%	99.6%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM20A	111,1	100.0%	99.4%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	145,2	100.0%	100.0%	Raine syndrome, 259775
FAM83G	156,8	100.0%	100.0%	No OMIM phenotype Palmoplantar keratoderma with leukonychia and abundant curly hair (Maruthappu et al. (2016) ESDR)
FAM83H	120,2	100.0%	99.9%	Amelogenesis imperfecta, type IIIA, 130900
FANCA	112,4	99.9%	98.9%	Fanconi anemia, complementation group A, 227650
FANCB	76,4	98.6%	93.2%	Fanconi anemia, complementation group B, 300514
FANCC	100,8	99.7%	99.2%	Fanconi anemia, complementation group C, 227645
FANCD2	115,6	99.1%	96.6%	Fanconi anemia, complementation group D2, 227646
FANCE	118,2	96.6%	89.9%	Fanconi anemia, complementation group E, 600901
FANCF	244,4	100.0%	100.0%	Fanconi anemia, complementation group F, 603467
FANCG	140,7	100.0%	99.8%	Fanconi anemia, complementation group G, 614082
FANCI	136,2	99.9%	98.9%	Fanconi anemia, complementation group I, 609053
FANCL	105,8	99.7%	98.0%	Fanconi anemia, complementation group L, 614083
FANCM	100,6	99.3%	97.1%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086

FAT4	190,3	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	96,6	91.8%	91.5%	Cutis laxa, autosomal dominant 2, 614434 Cutis laxa, autosomal recessive, type IA, 219100 Macular degeneration, age-related, 3, 608895 Neuropathy, hereditary, with or without age-related macular degeneration, 608895
FDPS	58,4	97.9%	91.6%	Porokeratosis 9, multiple types, 616631
FECH	104	100.0%	99.7%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	90,8	99.6%	96.6%	Kindler syndrome, 173650
FGF10	120,5	100.0%	99.6%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	122,3	99.7%	97.7%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced, 0 Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993
FGF3	139,5	100.0%	100.0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	155,5	99.9%	99.5%	Trichomegaly, 190330
FGF8	130	97.9%	86.8%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	122,6	100.0%	99.6%	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440

FGFR2	113,1	97.7%	96.8%	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific, 0 Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	138,5	100.0%	99.6%	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FH	128	95.0%	88.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKBP10	157,5	99.5%	97.3%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	80,8	99.8%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLCN	152,3	100.0%	100.0%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700

FLG	147,1	100.0%	99.9%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	352,3	99.9%	99.9%	Peeling skin syndrome 6, 618084
FLT4	160,3	99.2%	99.1%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FNIP1	157,8	100.0%	99.8%	No OMIM phenotype Multiple discoid fibromas (Claessens (2013) J Invest Dermatol 133 S136)
FOXC2	122,3	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	87,5	100.0%	99.7%	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXN1	133	100.0%	99.5%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	115,6	99.1%	94.8%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FREM1	110,7	99.8%	98.4%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FUCA1	125,9	100.0%	99.9%	Fucosidosis, 230000
FZD6	186,4	100.0%	100.0%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
GALNS	108,3	100.0%	99.3%	Mucopolysaccharidosis IVA, 253000
GALNT3	125,8	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GAN	142,2	99.9%	99.4%	Giant axonal neuropathy-1, 256850
GATA2	115	100.0%	99.0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GDF2	142,4	100.0%	100.0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506

GDF5	169,6	100.0%	100.0%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Brachydactyly, type A1, C, 615072 Brachydactyly, type A2, 112600 Brachydactyly, type C, 113100 Chondrodysplasia, Grebe type, 200700 Du Pan syndrome, 228900 Multiple synostoses syndrome 2, 610017 Symphalangism, proximal, 1B, 615298 {Osteoarthritis-5}, 612400
GGCX	101,2	100.0%	99.4%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GJA1	156,2	100.0%	100.0%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJB2	141,4	100.0%	100.0%	Bart-Pumphrey syndrome, 149200 Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Hystrix-like ichthyosis with deafness, 602540 Keratitis-ichthyosis-deafness syndrome, 148210 Keratoderma, palmoplantar, with deafness, 148350 Vohwinkel syndrome, 124500
GJB3	228,5	100.0%	100.0%	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy, 0 Deafness, autosomal recessive, 0 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200
GJB4	246,1	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	140,9	100.0%	100.0%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500

GJC2	45,3	92.6%	75.4%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphatic malformation 3, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GLA	73,6	99.5%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	82,6	99.7%	95.4%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLMN	70,2	99.0%	95.0%	Glomuvenous malformations, 138000
GMPPA	147,2	100.0%	99.8%	Alacrima, achalasia, and mental retardation syndrome, 615510
GNA11	162,4	100.0%	99.5%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNA14	128,8	100.0%	100.0%	No OMIM phenotype
GNAQ	52,8	81.0%	64.3%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	211,3	100.0%	100.0%	ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GORAB	165,7	100.0%	98.9%	Geroderma osteodysplasticum, 231070
GPR143	59,5	91.0%	79.1%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GRHL2	116,8	100.0%	100.0%	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	133,2	100.0%	99.8%	Van der Woude syndrome 2, 606713
GSN	115,5	95.6%	93.5%	Amyloidosis, Finnish type, 105120
GTF2E2	85,2	100.0%	98.2%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	81,8	99.9%	95.9%	Trichothiodystrophy 3, photosensitive, 616395

HCCS	92,4	99.2%	95.2%	Linear skin defects with multiple congenital anomalies 1, 309801
HDAC8	108,1	100.0%	99.2%	Cornelia de Lange syndrome 5, 300882
HERC2	95,1	80.0%	76.1%	Mental retardation, autosomal recessive 38, 615516 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HLCS	142,3	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMBS	97,3	100.0%	98.4%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	37,1	81.1%	62.4%	?Microphthalmia, syndromic 13, 300915
HOXC13	172,8	100.0%	100.0%	Ectodermal dysplasia 9, hair/nail type, 614931
HPGD	90,6	99.9%	98.9%	Cranioosteoarthropathy, 259100 Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100
HPS1	115,8	100.0%	99.9%	Hermansky-Pudlak syndrome 1, 203300
HPS3	132,7	99.9%	98.8%	Hermansky-Pudlak syndrome 3, 614072
HPS4	128,1	100.0%	99.9%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122,8	99.9%	98.7%	Hermansky-Pudlak syndrome 5, 614074
HPS6	164,6	99.9%	97.8%	Hermansky-Pudlak syndrome 6, 614075
HR	117,4	99.6%	97.3%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HRAS	182,3	100.0%	100.0%	Bladder cancer, somatic, 109800 Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Nevus sebaceous or woolly hair nevus, somatic, 162900 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Thyroid carcinoma, follicular, somatic, 188470
HTRA1	89	95.2%	87.0%	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 {Macular degeneration, age-related, 7}, 610149 {Macular degeneration, age-related, neovascular type}, 610149
HYAL1	110,7	100.0%	100.0%	?Mucopolysaccharidosis type IX, 601492

IDUA	148,1	98.9%	94.6%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IFT122	120,5	99.9%	99.0%	Cranioectodermal dysplasia 1, 218330
IFT43	112,4	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IKBKG	60,1	88.1%	78.8%	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IL17RA	149,1	100.0%	100.0%	Immunodeficiency 51, 613953
IL17RD	134	99.9%	99.0%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RN	139,3	100.0%	99.7%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL31RA	109,5	99.9%	99.6%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	92,8	100.0%	100.0%	Psoriasis 14, pustular, 614204
INSR	116,4	99.0%	95.1%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
IRF4	196,4	100.0%	99.9%	[Skin/hair/eye pigmentation, variation in, 8], 611724
IRF6	90,3	99.4%	95.0%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
ITGA3	150,1	99.7%	98.0%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	138,9	99.9%	99.0%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGB4	152,1	99.2%	97.4%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730

ITGB6	127,5	96.7%	95.0%	Amelogenesis imperfecta, type IH, 616221
JUP	124,5	100.0%	99.8%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KANK2	163,4	100.0%	99.9%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KAT6B	155,7	99.9%	99.1%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KCNH1	148,4	98.7%	98.3%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNK9	171,2	100.0%	100.0%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KDF1	110,3	100.0%	99.9%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDSR	158,2	99.9%	99.5%	Erythrokeratodermia variabilis et progressiva 4, 617526
KIF11	92,1	97.8%	94.5%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIT	136,2	100.0%	99.6%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, 601626 Mastocytosis, cutaneous, 154800 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KITLG	83,3	99.6%	97.2%	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KLHL24	172,7	100.0%	100.0%	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KLK4	164,3	100.0%	100.0%	Amelogenesis imperfecta, type IIA1, 204700
KLLN	152,3	100.0%	100.0%	Cowden syndrome 4, 615107
KMT2D	136,2	100.0%	99.7%	Kabuki syndrome 1, 147920

KRAS	67,2	99.4%	97.3%	Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Breast cancer, somatic, 114480 Cardiofaciocutaneous syndrome 2, 615278 Gastric cancer, somatic, 137215 Leukemia, acute myeloid, 601626 Lung cancer, somatic, 211980 Noonan syndrome 3, 609942 Pancreatic carcinoma, somatic, 260350 RAS-associated autoimmune leukoproliferative disorder, 614470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200
KRT1	98	99.9%	98.8%	Epidermolytic hyperkeratosis, 113800 Ichthyosis histrix, Curth-Macklin type, 146590 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962
KRT10	128,8	99.9%	98.6%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT13	120,3	100.0%	99.3%	White sponge nevus 2, 615785
KRT14	42,6	89.0%	80.0%	Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Naegeli-Franceschetti-Jadassohn syndrome, 161000
KRT16	34,9	75.2%	53.2%	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	17,6	46.8%	28.0%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT2	134,6	100.0%	99.5%	Ichthyosis bullosa of Siemens, 146800
KRT4	121,3	100.0%	99.5%	White sponge nevus 1, 193900

KRT5	110,6	100.0%	100.0%	Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960
KRT6A	121,7	96.9%	89.2%	Pachyonychia congenita 3, 615726
KRT6B	118,2	98.2%	91.8%	Pachyonychia congenita 4, 615728
KRT6C	105,5	88.6%	79.8%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	143,1	100.0%	99.9%	?Hypotrichosis 13, 615896
KRT74	138,6	100.0%	99.6%	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, autosomal dominant, 194300
KRT75	120,3	100.0%	100.0%	{Pseudofolliculitis barbae, susceptibility to}, 612318
KRT81	83,7	99.9%	97.8%	Monilethrix, 158000
KRT83	67,6	98.6%	90.0%	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000
KRT85	101,3	99.0%	95.2%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	85,3	100.0%	97.7%	Monilethrix, 158000
KRT9	68,1	99.4%	96.3%	Palmoplantar keratoderma, epidermolytic, 144200
LAMA3	125,2	99.9%	99.6%	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660
LAMB3	116,9	100.0%	99.4%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	100,5	99.7%	98.3%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMTOR2	172,2	100.0%	100.0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LDHA	55,6	96.6%	88.0%	Glycogen storage disease XI, 612933
LDLRAP1	149	99.9%	99.1%	Hypercholesterolemia, familial, autosomal recessive, 603813

LEMD3	122,5	99.8%	98.4%	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700
LIPH	120,2	100.0%	99.7%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	113,9	100.0%	99.1%	Ichthyosis, congenital, autosomal recessive 8, 613943
LMBRD1	100,1	98.9%	94.1%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMNA	104,7	97.7%	91.9%	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, type 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Restrictive dermopathy, lethal, 275210
LMX1B	146,6	99.9%	98.5%	Nail-patella syndrome, 161200
LONP1	148	100.0%	100.0%	CODAS syndrome, 600373
LOR	39,8	100.0%	93.8%	Vohwinkel syndrome with ichthyosis, 604117
LPAR6	100,3	99.8%	98.4%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN2	97,8	100.0%	99.6%	Majeed syndrome, 609628
LRMDA	114,9	99.4%	97.6%	Albinism, oculocutaneous, type VII, 615179
LSS	127,6	100.0%	99.7%	Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP3	147,5	100.0%	99.6%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	148	100.0%	99.4%	Cutis laxa, autosomal recessive, type IC, 613177
LYST	136,3	99.4%	97.8%	Chediak-Higashi syndrome, 214500
LYZ	143	100.0%	100.0%	Amyloidosis, renal, 105200
MAP2K1	92,3	99.5%	96.3%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	124,2	98.5%	94.1%	Cardiofaciocutaneous syndrome 4, 615280

MBTPS2	111,2	99.9%	98.6%	?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014
MED12	85,1	99.5%	95.5%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEFV	126,8	98.6%	96.5%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MGP	134,2	98.7%	94.6%	Keutel syndrome, 245150
MITF	141,1	100.0%	99.8%	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MLH1	139,2	99.9%	99.3%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLPH	97,4	99.7%	97.2%	Griscelli syndrome, type 3, 609227
MMACHC	196	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP2	154,2	100.0%	100.0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	90,8	99.8%	97.6%	Amelogenesis imperfecta, type IIA2, 612529
MPLKIP	104,3	100.0%	99.9%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRE11	49,7	97.3%	86.0%	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	111,7	99.4%	96.4%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSX1	143,3	99.9%	98.6%	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MTOR	112	99.9%	99.1%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MUTYH	152	100.0%	100.0%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659

MVD	113	99.9%	98.4%	Porokeratosis 7, multiple types, 614714
MVK	121,4	91.0%	90.5%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYH8	115,4	100.0%	99.4%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYO5A	109	99.7%	98.6%	Griscelli syndrome, type 1, 214450
NAA10	105	100.0%	98.8%	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NAGA	121,7	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NBAS	138,5	99.9%	99.1%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NCSTN	92,7	100.0%	99.6%	Acne inversa, familial, 1, 142690
NDUFB11	103,3	98.6%	95.0%	?Mitochondrial complex I deficiency, nuclear type 30, 301021 Linear skin defects with multiple congenital anomalies 3, 300952
NECTIN1	134	100.0%	99.9%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NECTIN4	121,6	100.0%	99.9%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK11	116,1	99.9%	98.5%	No OMIM phenotype Pancreatic cancer (Smith (2016) Cancer Lett 370,302)
NEK9	118,9	99.8%	98.2%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025
NF1	106,2	92.5%	89.4%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NFKBIA	134,6	95.3%	89.4%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHP2	121,9	100.0%	99.2%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIPAL4	126,7	100.0%	99.3%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	124,9	98.8%	96.9%	Cornelia de Lange syndrome 1, 122470

NLRP1	117,7	99.5%	97.6%	Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP3	134,6	100.0%	99.9%	CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Muckle-Wells syndrome, 191900
NME1	77,1	100.0%	99.9%	Neuroblastoma, 256700
NOD2	125,3	100.0%	99.9%	Blau syndrome, 186580 {Inflammatory bowel disease 1, Crohn disease}, 266600 {Psoriatic arthritis, susceptibility to}, 607507 {Yao syndrome}, 617321
NOP10	120,5	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH1	141,8	99.8%	98.9%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NRAS	145,5	100.0%	100.0%	?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Colorectal cancer, somatic, 114500 Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Thyroid carcinoma, follicular, somatic, 188470
NSD1	147	100.0%	99.8%	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	125,8	99.7%	97.1%	CHILD syndrome, 308050 CK syndrome, 300831
OCA2	116,8	99.7%	97.7%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200 [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
ODAM	142,4	99.9%	98.4%	No OMIM phenotype

OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OSMR	131,7	100.0%	99.5%	Amyloidosis, primary localized cutaneous, 1, 105250
PADI3	139	100.0%	100.0%	Uncombable hair syndrome, 191480
PAH	126,4	100.0%	100.0%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PALB2	143,5	100.0%	99.9%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PAX3	106,9	100.0%	99.7%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX9	236,1	99.8%	99.6%	Tooth agenesis, selective, 3, 604625
PCNA	92	100.0%	98.2%	?Ataxia-telangiectasia-like disorder 2, 615919
PDGFB	115,4	100.0%	100.0%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRB	126,6	99.7%	98.0%	Basal ganglia calcification, idiopathic, 4, 615007 Kosaki overgrowth syndrome, 616592 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550 Premature aging syndrome, Penttinen type, 601812
PEPD	117,4	100.0%	99.6%	Prolidase deficiency, 170100
PERP	166,2	100.0%	100.0%	No OMIM phenotype
PEX7	111	91.2%	89.3%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	107,9	99.8%	98.6%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	106,6	100.0%	99.3%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHYH	74	99.9%	96.9%	Refsum disease, 266500

PIEZO1	144,5	100.0%	99.5%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphatic malformation 6, 616843
PIGA	70,9	92.9%	84.0%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	106,3	93.6%	91.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	124,4	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	127,7	100.0%	99.8%	Breast cancer, somatic, 114480 CLAPO syndrome, somatic, 613089 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Macrodactyly, somatic, 155500 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PITX2	164,8	100.0%	99.5%	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PKP1	120,8	99.8%	98.4%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLCD1	116,1	100.0%	99.3%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCG2	105,8	100.0%	99.3%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	144,1	100.0%	100.0%	?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723

PLG	93,4	87.8%	86.8%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	93,1	100.0%	99.3%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	131,9	99.8%	97.3%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD3	109,7	100.0%	99.9%	Lysyl hydroxylase 3 deficiency, 612394
PMS2	94,7	83.4%	81.0%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMVK	118,7	100.0%	99.9%	Porokeratosis 1, multiple types, 175800
PNPLA1	164,8	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	142,7	100.0%	99.8%	Neutral lipid storage disease with myopathy, 610717
POC1A	112,9	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POFUT1	134,6	99.9%	99.4%	Dowling-Degos disease 2, 615327
POGLUT1	101,2	100.0%	98.7%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POLD1	124,5	98.0%	93.9%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLH	116,2	99.9%	98.6%	Xeroderma pigmentosum, variant type, 278750
POLR1C	98,3	98.9%	94.9%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	183,1	91.6%	91.6%	Treacher Collins syndrome 2, 613717
POLR3A	116,8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	129,8	99.7%	98.2%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	148,2	100.0%	100.0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMP	124,6	99.9%	97.6%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 Proteasome-associated autoinflammatory syndrome 2, 618048
PORCN	111,2	99.9%	98.8%	Focal dermal hypoplasia, 305600

POT1	97,7	99.9%	98.5%	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
PPOX	95,2	99.8%	97.5%	Porphyria variegata, 176200
PQBP1	163,5	100.0%	100.0%	Renpenning syndrome, 309500
PRKAR1A	79,4	98.6%	92.6%	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0 Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PSEN1	131,5	100.0%	100.0%	?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Cardiomyopathy, dilated, 1U, 613694 Dementia, frontotemporal, 600274 Pick disease, 172700
PSENEN	90,1	100.0%	100.0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB8	113,5	100.0%	98.8%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSTPIP1	103,8	99.9%	98.5%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	110,2	99.9%	98.4%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	120,3	99.9%	98.7%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTDSS1	112	100.0%	99.9%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	129,7	99.6%	97.0%	Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Prostate cancer, somatic, 176807 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174
PTHLH	127,2	98.4%	90.3%	Brachydactyly, type E2, 613382

PTPN11	78,3	98.6%	90.7%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN14	159,2	99.3%	96.8%	Choanal atresia and lymphedema, 613611
PTPRF	154,8	100.0%	99.9%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PYCR1	96	99.7%	97.4%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RAB23	107,4	100.0%	99.2%	Carpenter syndrome, 201000
RAB27A	126,1	100.0%	99.8%	Griscelli syndrome, type 2, 607624
RAD21	83	97.8%	93.4%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	102	97.5%	91.1%	Nijmegen breakage syndrome-like disorder, 613078
RAF1	108,3	100.0%	99.9%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAG1	150,9	100.0%	100.0%	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 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAG2	186,2	100.0%	100.0%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	194,4	100.0%	100.0%	Smith-Magenis syndrome, 182290
RBBP8	120,6	99.9%	99.3%	Jawad syndrome, 251255 Pancreatic carcinoma, somatic, 0 Seckel syndrome 2, 606744
RBM28	130,1	100.0%	99.9%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBP4	137,9	99.2%	95.8%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	70,7	96.3%	87.0%	Adams-Oliver syndrome 3, 614814

RECQL4	159,9	100.0%	99.8%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RHBDF2	105,1	99.9%	98.9%	Tylosis with esophageal cancer, 148500
RHOA	75,9	81.8%	80.7%	No OMIM phenotype
RIN2	119,5	100.0%	99.6%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	167,5	100.0%	100.0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	129,8	100.0%	99.7%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	100,8	98.9%	95.2%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	281,7	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROGDI	127,6	100.0%	99.4%	Kohlschutter-Tonz syndrome, 226750
RPL21	54,9	84.6%	64.0%	Hypotrichosis 12, 615885
RSPO1	103,8	100.0%	99.9%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO4	144,4	100.0%	100.0%	Anonychia congenita, 206800
RTEL1	131,1	99.7%	97.7%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RUNX2	102,8	73.4%	72.2%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
SAMD9	163,9	100.0%	99.9%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	133,4	99.8%	98.5%	?Chilblain lupus 2, 614415

				Aicardi-Goutieres syndrome 5, 612952
SART3	109,4	99.6%	97.8%	No OMIM phenotype Disseminated superficial actinic porokeratosis (Zhang (2005) Br J Dermatol 152,658)
SASH1	152,1	99.3%	97.8%	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 Dyschromatosis universalis hereditaria 1, 127500
SAT1	122,5	100.0%	98.9%	No OMIM phenotype Keratosis follicularis spinulosa decalvans (Gimelli (2002) Hum Genet 111,235)
SATB2	107,4	99.8%	97.7%	Glass syndrome, 612313
SCN10A	133,3	100.0%	99.4%	Episodic pain syndrome, familial, 2, 615551
SCN11A	122,1	99.3%	97.1%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	128,4	99.1%	97.7%	Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863 HSAN2D, autosomal recessive, 243000 Insensitivity to pain, congenital, 243000 Paroxysmal extreme pain disorder, 167400 Small fiber neuropathy, 133020 {Dravet syndrome, modifier of}, 607208
SDR9C7	168	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23B	131	99.8%	99.0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SERPINB7	124,3	100.0%	99.6%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	125,8	95.0%	95.0%	Peeling skin syndrome 5, 617115
SERPING1	96,7	99.5%	96.7%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	195,8	100.0%	99.6%	Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SGPL1	132,3	100.0%	100.0%	Nephrotic syndrome, type 14, 617575
SHOC2	139,6	99.9%	99.4%	Noonan-like syndrome with loose anagen hair, 607721
SKI	132,9	100.0%	99.3%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	138,4	100.0%	99.7%	Trichohepatoenteric syndrome 2, 614602
SLC17A9	140,1	95.8%	95.4%	Porokeratosis 8, disseminated superficial actinic type, 616063

SLC24A4	103,5	100.0%	99.8%	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	104,1	99.9%	99.3%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC26A2	205,1	100.0%	99.9%	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC27A4	150,9	100.0%	100.0%	Ichthyosis prematurity syndrome, 608649
SLC29A3	173,3	100.0%	99.5%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A10	152,6	98.0%	97.5%	Arterial tortuosity syndrome, 208050
SLC39A13	145,1	100.0%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A4	114,2	100.0%	99.0%	Acrodermatitis enteropathica, 201100
SLC45A2	115,2	100.0%	99.8%	Albinism, oculocutaneous, type IV, 606574 [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 [Skin/hair/eye pigmentation 5, dark/light eyes], 227240
SLC4A4	113,9	99.8%	98.3%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A19	129,3	100.0%	100.0%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC7A7	105,5	100.0%	99.6%	Lysinuric protein intolerance, 222700
SLCO2A1	97,7	99.9%	98.2%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLURP1	100,1	100.0%	99.4%	Meleda disease, 248300
SLX4	124,2	100.0%	99.7%	Fanconi anemia, complementation group P, 613951
SMAD3	126,7	100.0%	99.8%	Loeys-Dietz syndrome 3, 613795
SMARCA2	105,9	96.8%	95.9%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	150,9	100.0%	99.4%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325

SMARCAD1	93,5	99.5%	96.7%	Adermatoglyphia, 136000 Basan syndrome, 129200 Huriez syndrome, 181600
SMARCAL1	113,2	100.0%	99.6%	Schimke immunoosseous dysplasia, 242900
SMARCB1	179,1	100.0%	99.9%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid tumor predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMO	140,4	99.9%	98.3%	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	88,7	77.0%	75.7%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	102,7	99.9%	99.1%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP29	168,4	100.0%	100.0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNRPE	73	98.1%	89.9%	Hypotrichosis 11, 615059
SNX10	131,4	96.2%	95.7%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	102	99.6%	97.4%	?Fibromatosis, gingival, 1, 135300 Noonan syndrome 4, 610733
SOX10	88,2	100.0%	99.1%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX18	50	91.5%	76.2%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	230	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SP7	148,4	100.0%	99.3%	Osteogenesis imperfecta, type XII, 613849
SPINK5	128	99.9%	99.5%	Netherton syndrome, 256500
SPINT2	68,8	99.7%	90.0%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPRED1	146,5	99.8%	98.8%	Legius syndrome, 611431
SPRY4	164,5	100.0%	99.6%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266

SRD5A3	139,9	99.8%	98.3%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST14	154,8	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL5	101,8	89.0%	84.9%	Salt and pepper developmental regression syndrome, 609056
STAMBP	93,7	99.8%	97.9%	Microcephaly-capillary malformation syndrome, 614261
STAT3	103,2	100.0%	99.0%	Autoimmune disease, multisystem, infantile-onset, 1, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	114,1	99.8%	97.8%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIM1	120,7	99.8%	96.8%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070
STK11	131	100.0%	100.0%	Melanoma, malignant, somatic, 0 Pancreatic cancer, somatic, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STS	78,7	99.3%	95.2%	Ichthyosis, X-linked, 308100
SUFU	132,8	100.0%	99.9%	Basal cell nevus syndrome, 109400 Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SULT2B1	124,7	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	89,7	99.7%	96.8%	Multiple sulfatase deficiency, 272200
TALDO1	148,2	100.0%	99.6%	Transaldolase deficiency, 606003
TAP1	117,7	99.9%	97.3%	Bare lymphocyte syndrome, type I, 604571
TAP2	93	99.6%	98.4%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	116,6	96.6%	96.5%	Bare lymphocyte syndrome, type I, 604571
TAT	115	100.0%	100.0%	Tyrosinemia, type II, 276600
TBC1D24	177,7	100.0%	100.0%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOORS syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBX3	100,7	99.8%	98.2%	Ulnar-mammary syndrome, 181450

TCHH	162,8	100.0%	99.9%	?Uncombable hair syndrome 3, 617252
TCIRG1	131,4	99.2%	96.6%	Osteopetrosis, autosomal recessive 1, 259700
TEK	148	100.0%	99.7%	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERF2IP	128,4	100.0%	99.6%	No OMIM phenotype Melanoma (Aoude (2015) J Natl Cancer Inst 107) Chronic lymphocytic leukaemia (Speedy (2016) Blood 128,2319)
TERT	144,1	99.7%	97.6%	{Dyskeratosis congenita, autosomal dominant 2}, 613989 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742
TFAP2A	112,7	99.8%	98.0%	Branchiooculofacial syndrome, 113620
TGFB2	173,2	99.9%	99.0%	Loeys-Dietz syndrome 4, 614816
TGFBR1	156,4	95.4%	93.8%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	156,8	100.0%	100.0%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGM1	141	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	137,1	99.9%	98.4%	?Uncombable hair syndrome 2, 617251
TGM5	144,8	100.0%	99.9%	Peeling skin syndrome 2, 609796
TINF2	177,1	100.0%	100.0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMC6	91,1	100.0%	99.7%	Epidermodysplasia verruciformis, 226400
TMC8	133	100.0%	99.7%	Epidermodysplasia verruciformis 2, 618231
TMEM165	148,2	100.0%	99.8%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	95,3	99.1%	94.0%	STING-associated vasculopathy, infantile-onset, 615934
TNFRSF11A	131	96.1%	95.2%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080

TNFRSF11B	172,4	100.0%	100.0%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF1A	106,8	92.5%	89.7%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFSF11	129,8	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710
TNXB	105,6	99.5%	95.8%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP63	162,8	100.0%	100.0%	ADULT syndrome, 103285 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TPCN2	163,7	95.9%	94.4%	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267
TREX1	233,4	100.0%	100.0%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM32	123	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM37	113,8	98.5%	97.4%	Mulibrey nanism, 253250
TRPS1	154	100.0%	99.9%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	122,8	99.9%	98.5%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TSC1	112,5	99.6%	98.2%	Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	140,5	100.0%	99.9%	?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSPEAR	139,3	100.0%	99.8%	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TTC37	135,1	99.9%	99.2%	Trichohepatoenteric syndrome 1, 222470

TTI2	96,2	100.0%	99.9%	Mental retardation, autosomal recessive 39, 615541
TWIST2	132,6	100.0%	100.0%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TYR	147,9	100.0%	99.9%	Albinism, oculocutaneous, type IA, 203100 Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 [Skin/hair/eye pigmentation 3, blue/green eyes], 601800 [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 {Melanoma, cutaneous malignant, susceptibility to, 8}, 601800
TYRP1	152,3	100.0%	100.0%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBE2A	117,9	99.9%	96.4%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBR1	119,9	99.8%	99.0%	Johanson-Blizzard syndrome, 243800
UROD	130,8	98.9%	95.6%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	103,8	100.0%	99.7%	Porphyria, congenital erythropoietic, 263700
USB1	118,2	99.8%	97.2%	Poikiloderma with neutropenia, 604173
UVSSA	122,6	99.2%	98.9%	UV-sensitive syndrome 3, 614640
VDR	108,8	99.1%	96.0%	?Osteoporosis, involutional, 166710 Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	161,7	100.0%	100.0%	Lymphatic malformation 4, 615907
VHL	169,6	100.0%	98.3%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic, 0 Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VPS13B	134,5	99.3%	98.0%	Cohen syndrome, 216550
VPS33B	107,2	100.0%	99.9%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WAS	70,4	94.2%	83.6%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000

WDR19	126,8	100.0%	99.2%	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR35	141,8	99.7%	98.4%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR72	123,8	96.8%	96.1%	Amelogenesis imperfecta, type IIA3, 613211
WIPF1	89,1	100.0%	99.1%	?Wiskott-Aldrich syndrome 2, 614493
WNT10A	141,8	100.0%	99.9%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	157	100.0%	100.0%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT5A	159	100.0%	100.0%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	195,8	100.0%	100.0%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	162,8	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	124,8	99.7%	98.8%	Werner syndrome, 277700
XPA	74,7	99.7%	98.2%	Xeroderma pigmentosum, group A, 278700
XPC	143,5	100.0%	99.8%	Xeroderma pigmentosum, group C, 278720
XYLT1	128,1	99.9%	98.2%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	147,5	99.7%	98.1%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
YWHAZ	45,9	79.5%	66.9%	No OMIM phenotype
ZBTB20	180,2	100.0%	100.0%	Primrose syndrome, 259050
ZMPSTE24	128,7	100.0%	99.6%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZNF469	157,6	100.0%	100.0%	Brittle cornea syndrome 1, 229200
ZNF592	142,8	100.0%	100.0%	Spinocerebellar ataxia, autosomal recessive 5, 251300
ZNF750	176,9	100.0%	99.9%	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.
Median Coverage describes the average number of reads seen across 50 exomes.

% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors