SKIN DISORDERS GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AAAS	108.6	99%	99%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	165.5	100%	100%	Keratoderma, palmoplantar, punctate type IA, 148600
ABCA12	179.2	99%	98%	Ichthyosis, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCB6	141.4	100%	99%	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	121.4	93%	92%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	175	100%	99%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABHD5	256.2	100%	99%	Chanarin-Dorfman syndrome, 275630
ACD	139.5	100%	99%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACTA2	140.7	100%	99%	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	118.8	98%	94%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACVRL1	138.6	100%	98%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAM10	158.7	99%	98%	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590
ADAM17	152.2	99%	95%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAMTS10	124.3	99%	99%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	135.3	91%	88%	Weill-Marchesani-like syndrome, 613195
ADAMTS2	131.5	99%	98%	Ehlers-Danlos syndrome, type VIIC, 225410

ADAMTSL2	124.8	96%	91%	Geleophysic dysplasia 1, 231050
ADAR	136.8	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	175.8	100%	100%	Aspartylglucosaminuria, 208400
AGPAT2	117.9	99%	95%	Lipodystrophy, congenital generalized, type 1, 608594
AIRE	83.4	99%	97%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKT1	182.3	100%	99%	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	97.6	99%	95%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	106.6	100%	98%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	99.8	98%	95%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	143.9	100%	100%	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	153.1	100%	99%	Sjogren-Larsson syndrome, 270200
ALDOB	168.3	99%	98%	Fructose intolerance, 229600
ALOX12B	139.6	99%	98%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	131.6	100%	99%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	164.9	100%	100%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALX4	145.9	99%	94%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMELX	104.2	99%	97%	Amelogenesis imperfecta, type 1E, 301200
ANKRD11	118.5	98%	95%	KBG syndrome, 148050

ANTXR1	144.4	98%	96%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	112.4	99%	96%	Hyaline fibromatosis syndrome, 228600
AP1S3	127.6	90%	90%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	128.1	99%	95%	Hermansky-Pudlak syndrome 2, 608233
APC	186.4	100%	99%	Adenoma, periampullary, somatic 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	190.1	99%	97%	Hypotrichosis 1, 605389
AQP5	123.9	99%	97%	Palmoplantar keratoderma, Bothnian type, 600231
ARHGAP31	141.8	99%	98%	Adams-Oliver syndrome 1, 100300
ARID1A	158.4	95%	91%	Coffin-Siris syndrome 2, 614607
ARID1B	159.9	96%	92%	Coffin-Siris syndrome 1, 135900
ASIP	126.4	100%	99%	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 [Skin/hair/eye pigmentation 9, dark/light hair], 611742
ASL	128.8	100%	98%	Argininosuccinic aciduria, 207900
ASXL1	172.5	99%	98%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	170	99%	99%	Bainbridge-Ropers syndrome, 615485
ATIC	142.5	99%	99%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP2A2	198.5	100%	99%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	156.9	100%	99%	Hailey-Hailey disease, 169600
ATP6V0A2	160.2	99%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	148.2	99%	98%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATR	175.1	99%	98%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564

AXIN2	132.5	100%	99%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B3GALT6	56.4	76%	71%	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B4GALT7	122.7	97%	95%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
BANF1	62.6	95%	86%	Nestor-Guillermo progeria syndrome, 614008
BAP1	153.1	99%	98%	Tumor predisposition syndrome, 614327
BCOR	117.5	99%	97%	Microphthalmia, syndromic 2, 300166
BCS1L	199	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	139.5	99%	97%	Bloom syndrome, 210900
BLOC1S3	56	99%	95%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	111.3	99%	96%	Hermansky-pudlak syndrome 9, 614171
BMS1	91.6	66%	65%	?Aplasia cutis congenita, nonsyndromic, 107600
BRAF	86.4	91%	82%	Adenocarcinoma of lung, somatic, 211980 Cardiofaciocutaneous syndrome, 115150 Colorectal cancer, somatic LEOPARD syndrome 3, 613707 Melanoma, malignant, somatic Nonsmall cell lung cancer, somatic Noonan syndrome 7, 613706
BRIP1	149	99%	98%	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BSCL2	129.8	100%	100%	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	168.1	100%	100%	Biotinidase deficiency, 253260
C10orf11	165.3	99%	99%	Albinism, oculocutaneous, type VII, 615179
C1QA	147.6	100%	98%	C1q deficiency, 613652
C1QB	225.6	100%	100%	C1q deficiency, 613652
C1QC	215.1	100%	99%	C1q deficiency, 613652
C2CD3	155.7	95%	95%	?Orofaciodigital syndrome XIV, 615948

C4orf26	260.2	100%	100%	Amelogenesis imperfecta, type IIA4, 614832
CA2	178.8	99%	97%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAPN12	99.5	97%	92%	No OMIM phenotype Modifying factor in ichthyosis
CARD14	130.2	99%	98%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	129.1	99%	96%	Candidiasis, familial, 2, autosomal recessive, 212050
CASP14	92.3	100%	99%	Ichthyosis, congenital, autosomal recessive 12, 617320
CAST	136.7	98%	95%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAV1	282.6	100%	100%	Pulmonary hypertension, primary, 3, 615343 ?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721
CBL	146	99%	98%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CBS	134.1	98%	94%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCBE1	79.9	99%	94%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD151	154.1	100%	100%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CDAN1	108.7	98%	97%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDH3	164.1	99%	98%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDK4	128.7	100%	100%	{Melanoma, cutaneous malignant, 3}, 609048
CDKN2A	80	93%	92%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDSN	20.4	58%	40%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CECR1	106	99%	98%	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410

CELSR1	200.9	95%	93%	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 (Gonzalez-Garay (2016) Vasc Cell 8,1)
CERS3	143.1	100%	99%	Ichthyosis, congenital, autosomal recessive 9, 615023
CHKB	102.6	99%	97%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	194.6	96%	94%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHSY1	142.1	96%	94%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	146.6	99%	98%	Cocoon syndrome, 613630
CKAP2L	219	99%	97%	Filippi syndrome, 272440
CLDN1	153.7	100%	100%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	196.6	100%	100%	No OMIM phenotype {Ulcerative colitis, susceptibility to} (Saadati (2016) BMC Med Genet 17,26)
CNNM4	212.9	98%	98%	Jalili syndrome, 217080
COL17A1	118.8	98%	96%	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL1A2	118.4	97%	94%	Ehlers-Danlos syndrome, cardiac valvular form, 225320 Ehlers-Danlos syndrome, type VIIB, 130060 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type III, 259420 Osteogenesis imperfecta, type IV, 166220 {Osteoporosis, postmenopausal}, 166710
COL3A1	124.9	98%	93%	Ehlers-Danlos syndrome, type IV, 130050
COL5A1	131.5	98%	95%	Ehlers-Danlos syndrome, classic type, 130000
COL5A2	106.4	99%	97%	Ehlers-Danlos syndrome, classic type, 130000

COL7A1	148.7	99%	98%	EBD inversa, 226600 EBD, Bart type, 132000 EBD, localisata variant 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	123.1	100%	99%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	53.1	83%	56%	Linear skin defects with multiple congenital anomalies, 300887
CPOX	136.6	93%	87%	Coproporphyria, 121300 Harderoporphyria, 121300
CST6	116.4	97%	90%	No OMIM phenotype
CSTA	135.2	100%	99%	Peeling skin syndrome 4, 607936
CTC1	122.4	99%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTSA	153.2	99%	99%	Galactosialidosis, 256540
CTSC	142.5	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000 Periodontitis 1, juvenile, 170650
CXCR4	182.7	100%	99%	Myelokathexis, isolated WHIM syndrome, 193670
CYLD	132.2	99%	95%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP26C1	96.5	99%	98%	Focal facial dermal dysplasia 4, 614974
CYP4F22	134.6	100%	99%	Ichthyosis, congenital, autosomal recessive 5, 604777
DCAF17	125.5	99%	97%	Woodhouse-Sakati syndrome, 241080
DCLRE1C	158.5	98%	95%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDB2	181.7	100%	99%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DHCR7	173	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DKC1	122.5	100%	98%	Dyskeratosis congenita, X-linked, 305000
DLX3	128.9	99%	96%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320

DLX5	122.5	100%	98%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DOCK6	134.7	99%	97%	Adams-Oliver syndrome 2, 614219
DOCK8	148.8	100%	99%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOLK	189.5	100%	99%	Congenital disorder of glycosylation, type Im, 610768
DSC2	158.7	99%	97%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	107.4	98%	94%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	120	99%	98%	?Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	182.9	98%	97%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG3	161.7	99%	98%	No OMIM phenotype
DSG4	228.3	99%	97%	Hypotrichosis 6, 607903
DSP	161.7	100%	99%	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	150.9	99%	99%	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DTNBP1	127.3	99%	97%	Hermansky-Pudlak syndrome 7, 614076
DUSP6	172.7	100%	99%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EBP	89.2	99%	96%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECM1	186	100%	99%	Urbach-Wiethe disease, 247100
EDA	99.3	92%	82%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	151.6	99%	99%	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	105.1	99%	96%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941

EDN3	145.1	100%	99%	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	229.1	100%	100%	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	151.4	98%	93%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EFEMP2	146.1	100%	99%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	144.7	100%	99%	Craniofrontonasal dysplasia, 304110
EIF2AK3	177.3	96%	91%	Wolcott-Rallison syndrome, 226980
ELN	103.3	99%	98%	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
ELOVL4	115.5	100%	99%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
ENAM	154.1	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ENG	148.5	99%	96%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	174.9	94%	88%	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	138.8	99%	98%	Vici syndrome, 242840
ERCC2	143	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	120.8	100%	99%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	165.7	99%	99%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 ?XFE progeroid syndrome, 610965

ERCC5	151.3	100%	99%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	196.3	100%	99%	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	109.4	96%	87%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
EVC	126.1	94%	91%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	137.8	96%	94%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXPH5	229.7	100%	100%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
FAM111B	185.5	100%	99%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM20A	115.2	98%	94%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	118.7	99%	98%	Raine syndrome, 259775
FAM83G	137.4	100%	100%	No OMIM phenotype Palmoplantar keratoderma with leukonychia and abundant curly hair (Maruthappu et al. (2016) ESDR)
FAM83H	93.9	96%	91%	Amelogenesis imperfecta, type III, 130900
FANCA	129.9	99%	98%	Fanconi anemia, complementation group A, 227650
FANCB	84.3	97%	90%	Fanconi anemia, complementation group B, 300514
FANCC	118.8	99%	98%	Fanconi anemia, complementation group C, 227645
FANCD2	156.1	99%	96%	Fanconi anemia, complementation group D2, 227646
FANCE	127.7	88%	85%	Fanconi anemia, complementation group E, 600901
FANCF	179.5	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	158	100%	99%	Fanconi anemia, complementation group G, 614082
FANCI	174.9	99%	98%	Fanconi anemia, complementation group I, 609053
FANCL	105.1	99%	97%	Fanconi anemia, complementation group L, 614083
FANCM	117	99%	96%	No OMIM phenotype

				Fanconi anemia, complementation group M, 614087
FAT4	259.2	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	116.4	91%	91%	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	66.8	98%	92%	Porokeratosis 9, multiple types, 616631
FECH	137.3	100%	99%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FERMT1	109.7	98%	96%	Kindler syndrome,173650
FGF10	176	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	122.5	99%	98%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF3	86.6	96%	87%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	129.1	100%	99%	Trichomegaly, 190330
FGF8	137.4	87%	80%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	165.2	99%	98%	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	155.6	97%	96%	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 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 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	129.1	100%	99%	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	183.5	93%	89%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKBP10	177.3	98%	95%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	96.5	99%	99%	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557

FLCN	176.6	100%	99%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	237.1	100%	99%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	742.9	100%	100%	No OMIM phenotype ?Atopic dermatitis (Margolis (2014) J Invest Dermatol 134,2272)
FLT4	179	99%	98%	Hemangioma, capillary infantile, somatic, 602089 Lymphedema, hereditary, IA, 153100
FNIP1	197.3	99%	99%	No OMIM phenotype Multiple discoid fibromas (Claessens (2013) J Invest Dermatol 133 S136)
FOXC2	64.1	98%	88%	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 Lymphedema-distichiasis syndrome, 153400
FOXE1	37.4	81%	62%	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXN1	127.1	100%	99%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	130.9	98%	92%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FREM1	149.2	99%	99%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FUCA1	151.4	100%	99%	Fucosidosis, 230000
FZD6	260.7	100%	100%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
GALNS	108.4	99%	95%	Mucopolysaccharidosis IVA, 253000
GALNT3	166.7	99%	98%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GAN	199	100%	99%	Giant axonal neuropathy-1, 256850
GATA2	145.4	100%	99%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286
GDF2	185.5	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506

GDF5	179.6	100%	100%	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 ?Acromesomelic dysplasia, Hunter-Thompson type, 201250 {Osteoarthritis-5}, 612400
GGCX	124.5	100%	99%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GJA1	238.2	100%	100%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva, 133200 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	209.4	100%	100%	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	343.4	100%	100%	Deafness, autosomal dominant 2B, 612644 Deafness, autosomal dominant, with peripheral neuropathy Deafness, autosomal recessive Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva, 133200
GJB4	396.3	100%	100%	Erythrokeratodermia variabilis with erythema gyratum repens, 133200

GJB6	214	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	50	88%	71%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GLA	83.5	99%	97%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	97.3	99%	97%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLMN	95.5	99%	94%	Glomuvenous malformations,138000
GMPPA	162.5	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510
GNA11	185.8	100%	99%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNAQ	90.6	89%	75%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	154.4	99%	97%	Acromegaly, somatic, 102200 ACTH-independent macronodular adrenal hyperplasia, 219080 McCune-Albright syndrome, somatic, mosaic 174800 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ia, 103580 Pseudohypoparathyroidism Ib, 603233 Pseudohypoparathyroidism Ic, 612462 Pseudopseudohypoparathyroidism, 612463
GORAB	203.4	99%	97%	Geroderma osteodysplasticum, 231070
GPR143	73.2	90%	83%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GRHL2	152.8	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	162.5	100%	100%	Van der Woude syndrome 2, 606713
GSN	141.9	97%	92%	Amyloidosis, Finnish type, 105120
GTF2H5	152.5	100%	99%	Trichothiodystrophy 3, photosensitive, 616395

HCCS	115.6	99%	98%	Linear skin defects with multiple congenital anomalies 1, 309801
HDAC8	142.5	100%	99%	Cornelia de Lange syndrome 5, 300882
HERC2	120.8	79%	76%	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	182.6	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	117.7	100%	99%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	45.6	90%	74%	?Microphthalmia, syndromic 13, 300915
HOXC13	133.7	98%	93%	Ectodermal dysplasia 9, hair/nail type, 614931
HPS1	136.6	100%	99%	Hermansky-Pudlak syndrome 1, 203300
HPS3	175.7	100%	99%	Hermansky-Pudlak syndrome 3, 614072
HPS4	151.7	100%	99%	Hermansky-Pudlak syndrome 4, 614073
HPS5	160.7	99%	98%	Hermansky-Pudlak syndrome 5, 614074
HPS6	156.4	97%	90%	Hermansky-Pudlak syndrome 6, 614075
HR	110.5	98%	95%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HRAS	204.3	100%	99%	Congenital myopathy with excess of muscle spindles, 218040 Costello syndrome, 218040 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 {Bladder cancer, somatic}, 109800 {Nevus sebaceous or woolly hair nevus, somatic}, 162900 {Spitz nevus or nevus spilus, somatic}, 137550 {Thyroid carcinoma, follicular, somatic}, 188470
HTRA1	116.9	86%	81%	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	104.3	100%	100%	?Mucopolysaccharidosis type IX, 601492
IDUA	120.5	92%	86%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IFT122	165.7	100%	99%	Cranioectodermal dysplasia 1, 218330

IFT43	128	100%	100%	Cranioectodermal dysplasia 3, 614099
IKBKG	57.4	83%	72%	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 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	145.2	99%	97%	Immunodeficiency 51, 613953
IL17RD	146.4	99%	98%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RN	174.7	100%	100%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL31RA	151	100%	99%	Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	94.7	100%	99%	Psoriasis 14, pustular, 614204
INSR	151.6	97%	94%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
IRF4	210.3	100%	99%	[Skin/hair/eye pigmentation, variation in, 8], 611724
IRF6	134.4	99%	98%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
ITGA3	159.6	99%	98%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	174.6	99%	98%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGB4	173.9	98%	95%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	179.8	96%	95%	Amelogenesis imperfecta, type IH, 616221
JUP	160.5	100%	99%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KAL1	102.2	89%	87%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KANK2	171.3	100%	99%	Palmoplantar keratoderma and woolly hair, 616099
KAT6B	194.1	99%	99%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736

KCNH1	195.4	100%	99%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNK9	214.4	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KIF11	98	97%	95%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIT	195.4	100%	99%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800
KITLG	95.5	96%	92%	Deafness, congenital, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KLK4	210	99%	99%	Amelogenesis imperfecta, type IIA1, 204700
KLLN	143.7	100%	100%	Cowden syndrome 4, 615107
KMT2D	158.6	100%	99%	Kabuki syndrome 1, 147920
KRAS	89.6	99%	99%	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	142.2	100%	98%	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	120.2	98%	95%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT13	151.9	99%	98%	White sponge nevus 2, 615785

KRT14	56.7	89%	81%	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	39.1	72%	55%	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	23.6	55%	36%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT2	149	100%	99%	Ichthyosis bullosa of Siemens, 146800
KRT4	138.6	100%	99%	White sponge nevus 1, 193900
KRT5	146.8	100%	99%	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-MP, 131960 Epidermylysis bullosa simplex-MCR, 609352
KRT6A	186.4	95%	88%	Pachyonychia congenita 3, 615726
KRT6B	187.8	97%	90%	Pachyonychia congenita 4, 615728
KRT6C	171.6	88%	80%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	153.2	100%	99%	?Hypotrichosis 13, 615896
KRT74	164	99%	99%	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, autosomal dominant, 194300
KRT75	148.4	100%	100%	{Pseudofolliculitis barbae, susceptibility to}, 612318
KRT81	99.1	99%	95%	Monilethrix, 158000
KRT83	85.7	98%	90%	?Monilethrix, 158000
KRT85	123.2	99%	94%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	108.6	99%	97%	Monilethrix, 158000
KRT9	90.4	97%	95%	Palmoplantar keratoderma, epidermolytic, 144200
LAMA3	165.5	99%	99%	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660

LAMB3	134.9	100%	99%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	137.9	99%	97%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMTOR2	204.7	100%	99%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LDHA	69.6	94%	89%	Glycogen storage disease XI, 612933
LDLRAP1	188	98%	96%	Hypercholesterolemia, familial, autosomal recessive, 603813
LEMD3	116.1	98%	93%	Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950 Osteopoikilosis, 166700
LIPH	154.3	100%	99%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	153.5	99%	98%	Ichthyosis, congenital, autosomal recessive 8, 613943
LMBRD1	96.5	93%	88%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMNA	95.6	97%	90%	Cardiomyopathy, dilated, 1A, 115200 Charcot-Marie-Tooth disease, type 2B1, 605588 Emery-Dreifuss muscular dystrophy 2, AD, 181350 Emery-Dreifuss muscular dystrophy 3, AR, 616516 Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Lipodystrophy, familial partial, 2, 151660 Malouf syndrome, 212112 Mandibuloacral dysplasia, 248370 Muscular dystrophy, congenital, 613205 Muscular dystrophy, limb-girdle, type 1B, 159001 Restrictive dermopathy, lethal, 275210
LMX1B	131.5	99%	96%	Nail-patella syndrome, 161200
LONP1	164.9	98%	97%	CODAS syndrome, 600373
LOR	22.5	91%	56%	Vohwinkel syndrome with ichthyosis, 604117
LPAR6	123.4	100%	98%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN2	124.6	100%	99%	Majeed syndrome, 609628
LTBP3	128.7	98%	97%	Dental anomalies and short stature, 601216
LTBP4	137.6	99%	97%	Cutis laxa, autosomal recessive, type IC, 613177

LYST	171	98%	96%	Chediak-Higashi syndrome, 214500
LYZ	187	100%	100%	Amyloidosis, renal, 105200
MAP2K1	106.2	99%	97%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	122.2	99%	95%	Cardiofaciocutaneous syndrome 4, 615280
MBTPS2	147.5	99%	98%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918
MED12	111.4	98%	94%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEFV	135.7	96%	92%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MGP	161.6	95%	92%	Keutel syndrome, 245150
MITF	173.3	100%	100%	Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MLH1	187.4	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLPH	110.2	99%	96%	Griscelli syndrome, type 3, 609227
MMACHC	227.3	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP2	173.5	100%	99%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	112.5	99%	98%	Amelogenesis imperfecta, type IIA2, 612529
MPLKIP	103.8	99%	91%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRE11A	64.3	97%	89%	Ataxia-telangiectasia-like disorder, 604391
MSH2	137.3	99%	96%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSX1	93.5	98%	94%	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MTOR	152.6	100%	99%	Smith-Kingsmore syndrome, 616638

MUTYH	178	100%	99%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVD	114.6	99%	98%	Porokeratosis 7, multiple types, 614714
MVK	167.2	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYH8	152.7	100%	99%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYO5A	138.4	99%	98%	Griscelli syndrome, type 1, 214450
NAA10	109.4	99%	96%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAGA	159.7	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NBAS	176.4	99%	98%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NCSTN	129.2	100%	99%	Acne inversa, familial, 1, 142690
NDUFB11	108.3	95%	87%	Linear skin defects with multiple congenital anomalies 3, 300952
NEK11	143.9	99%	97%	No OMIM phenotype Pancreatic cancer (Smith (2016) Cancer Lett 370,302)
NF1	146.2	93%	91%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NFKBIA	126	98%	93%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHP2	101.6	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIPAL4	152.5	99%	96%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	142.5	97%	95%	Cornelia de Lange syndrome 1, 122470
NLRP1	144.7	99%	96%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP3	162.9	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900

NME1	114.4	100%	99%	Neuroblastoma, 256700
NOD2	149.9	100%	99%	Blau syndrome, 186580 Yao syndrome, 617321 {Inflammatory bowel disease 1}, 266600 {Psoriatic arthritis, susceptibility to}, 607507
NOP10	159.6	100%	99%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH1	154.9	99%	98%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NRAS	203.3	100%	100%	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 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470
NSD1	181.1	100%	100%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	196.4	100%	99%	CHILD syndrome, 308050 CK syndrome, 300831
OCA2	141.6	99%	97%	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	151.2	99%	95%	No OMIM phenotype
OFD1	59.2	87%	75%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OSMR	173.4	100%	99%	Amyloidosis, primary localized cutaneous, 1, 105250
PAH	186.6	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600

PALB2	180	100%	99%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480 {Pancreatic cancer, susceptibility to, 3}, 613348
PAX3	128.2	100%	99%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX9	266	99%	99%	Tooth agenesis, selective, 3, 604625
PCNA	109	100%	99%	?Ataxia-telangiectasia-like disorder, 615919
PDGFB	110.3	100%	100%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRB	160.1	98%	95%	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	127	99%	98%	Prolidase deficiency, 170100
PEX7	152.7	90%	87%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	138.1	98%	97%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	138	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHYH	90	98%	93%	Refsum disease, 266500
PIEZO1	158.2	99%	98%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphedema, hereditary, III, 616843
PIGA	97	94%	86%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	130.2	99%	94%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	168	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300

PIK3CA	155.4	100%	99%	Breast cancer, somatic, 114480 CLOVE syndrome, somatic, 612918 Colorectal cancer, somatic, 114500 Cowden syndrome 5, 615108 Gastric cancer, somatic, 613659 Hepatocellular carcinoma, somatic, 114550 Keratosis, seborrheic, somatic, 182000 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 Nonsmall cell lung cancer, somatic, 211980 Ovarian cancer, somatic, 167000
PITX2	146.1	99%	98%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PKP1	140	100%	99%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLCD1	122.7	99%	98%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCG2	130.6	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	122.1	99%	99%	Epidermolysis bullosa simplex with muscular dystrophy, 226670 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy, limb-girdle, type 2Q, 613723 ?Epidermolysis bullosa simplex with nail dystrophy, 616487
PLG	138.6	87%	87%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	89.6	98%	90%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	155.4	99%	96%	Ehlers-Danlos syndrome, type VI, 225400
PMS2	109.8	83%	81%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMVK	128.4	100%	99%	Porokeratosis 1, multiple types, 175800
PNPLA1	192.9	99%	99%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	139.3	99%	96%	Neutral lipid storage disease with myopathy, 610717
POC1A	144.7	100%	100%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POFUT1	162.7	100%	99%	Dowling-Degos disease 2, 615327

POGLUT1	150.1	99%	96%	Dowling-Degos disease 4, 615696
POLD1	117.3	95%	92%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLH	170.4	100%	99%	Xeroderma pigmentosum, variant type, 278750
POLR1C	117.5	99%	95%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	193.8	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	153	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	163.8	100%	99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	105.7	100%	99%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMP	167	98%	91%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
PORCN	142.4	100%	99%	Focal dermal hypoplasia, 305600
POT1	125.8	99%	98%	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
PPOX	104.3	99%	97%	Porphyria variegata, 176200
PQBP1	180.7	97%	96%	Renpenning syndrome, 309500
PRKAR1A	104.2	97%	92%	Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, Carney complex, type 1, 160980 Myxoma, intracardiac, 255960 Pigmented nodular adrenocortical disease, primary, 1, 610489
PSEN1	156.6	99%	97%	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	102.7	100%	100%	Acne inversa, familial, 2, 613736
PSMB8	16	61%	25%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	104.7	99%	97%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416

PTCH1	127.7	98%	96%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCH2	131.6	99%	97%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, 155255
PTDSS1	162.9	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	169.6	100%	99%	Bannayan-Riley-Ruvalcaba syndrome, 153480 Cowden syndrome 1, 158350 Endometrial carcinoma, somatic, 608089 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309 Malignant melanoma, somatic, 155600 PTEN hamartoma tumor syndrome Squamous cell carcinoma, head and neck, somatic, 275355 VATER association with macrocephaly and ventriculomegaly, 276950 {Glioma susceptibility 2}, 613028 {Meningioma}, 607174 {Prostate cancer, somatic}, 176807
PTHLH	153.3	99%	98%	Brachydactyly, type E2, 613382 Humoral hypercalcemia of malignancy
PTPN11	105.7	98%	93%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN14	199.8	99%	97%	Choanal atresia and lymphedema, 613611
PTPRF	196.4	100%	100%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTRF	162.4	99%	99%	Lipodystrophy, congenital generalized, type 4, 613327
PVRL1	166.5	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
PVRL4	157.6	100%	99%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
PYCR1	103.5	99%	95%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RAB23	141.3	100%	100%	Carpenter syndrome, 201000
RAB27A	171.6	100%	100%	Griscelli syndrome, type 2, 607624

RAD21	109	99%	97%	Cornelia de Lange syndrome 4, 614701
RAD50	119.9	94%	89%	Nijmegen breakage syndrome-like disorder, 613078
RAF1	144.9	100%	99%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAG1	208.9	100%	100%	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	271.4	100%	100%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	159.2	100%	99%	Smith-Magenis syndrome, 182290
RBBP8	121.9	99%	98%	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744
RBM28	180.5	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBP4	124.5	99%	95%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	94.2	95%	88%	Adams-Oliver syndrome 3, 614814
RECQL4	152.3	99%	98%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RHBDF2	107.7	99%	98%	Tylosis with esophageal cancer, 148500
RIN2	125	100%	99%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	163.1	100%	99%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RMRP	NC	NC	NC	Anauxetic dysplasia, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	157.1	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	128.3	98%	92%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	244.3	99%	98%	Aicardi-Goutieres syndrome 3, 610329
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651

ROGDI	127.1	97%	95%	Kohlschutter-Tonz syndrome, 226750
RPL21	85.4	80%	64%	Hypotrichosis 12, 615885
RSPO1	128.8	100%	100%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO4	130.2	100%	99%	Anonychia congenita, 206800
RTEL1	137.2	99%	97%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RUNX2	111.8	74%	74%	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	226.4	100%	99%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	154.9	99%	98%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SART3	142.3	99%	98%	No OMIM phenotype Disseminated superficial actinic porokeratosis (Zhang (2005) Br J Dermatol 152,658)
SASH1	165.2	99%	97%	No OMIM phenotype Lentiginosis, autosomal dominant (Shellman (2015) J Invest Dermatol 135,3192) Pigmentation defects, palmoplantar keratoderma and skin carcinoma (Courcet (2015) Eur J Hum Genet 23,957)
SAT1	153.9	99%	97%	No OMIM phenotype Keratosis follicularis spinulosa decalvans (Gimelli (2002) Hum Genet 111,235)
SATB2	122.4	99%	94%	Glass syndrome, 612313
SCN10A	187	99%	99%	Episodic pain syndrome, familial, 2, 615551
SCN11A	165	99%	98%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	180.4	98%	97%	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	211.5	100%	100%	No OMIM phenotype Ichtyosis (Shigehara (2016) Hum Mol Genet epub, epub)
SEC23B	169	97%	97%	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SERPINB7	154.7	100%	99%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	167	100%	99%	Peeling skin syndrome 5,617115
SERPINH1	188	100%	99%	?Osteogenesis imperfecta, type X, 613848 {Preterm premature rupture of the membranes, susceptibility to}, 610504
SGPL1	179.7	100%	100%	No OMIM phenotype Charcot-Marie-Tooth disease (Atkinson (2017) Neurology 88,533)
SHOC2	150.5	100%	99%	Noonan-like syndrome with loose anagen hair, 607721
SKI	96.2	98%	96%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	27	81%	57%	Trichohepatoenteric syndrome 2, 614602
SLC17A9	134.6	95%	95%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC24A4	133.3	99%	98%	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	130.5	99%	97%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC26A2	289.4	100%	100%	Achondrogenesis Ib, 600972 Atelosteogenesis II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SLC27A4	184.8	99%	99%	Ichthyosis prematurity syndrome, 608649
SLC29A3	236.3	99%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A10	187.1	100%	99%	Arterial tortuosity syndrome, 208050

SLC39A13	134.6	99%	99%	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350
SLC39A4	93.7	99%	96%	Acrodermatitis enteropathica, 201100
SLC45A2	152.5	99%	99%	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	159.1	99%	99%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A19	177.6	99%	99%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC7A7	114.5	100%	99%	Lysinuric protein intolerance, 222700
SLCO2A1	111.8	99%	98%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLURP1	117	100%	98%	Meleda disease, 248300
SLX4	129.5	100%	99%	Fanconi anemia, complementation group P, 613951
SMAD3	151.7	99%	99%	Loeys-Dietz syndrome 3, 613795
SMARCA2	127.7	97%	96%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	165.4	100%	99%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCAD1	103.7	99%	97%	Adermatoglyphia, 136000
SMARCAL1	153.8	100%	99%	Schimke immunoosseous dysplasia, 242900
SMARCB1	265.7	100%	100%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMO	177.8	98%	94%	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome,somatic mosaic, 601707
SMOC2	135	97%	93%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	128.7	100%	99%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP29	149.1	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNRPE	92.4	99%	97%	Hypotrichosis 11, 615059
SNX10	142	100%	99%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	120.1	98%	95%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300

SOX10	74.6	98%	95%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX18	27.6	78%	58%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	127.1	99%	98%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SP7	173.4	99%	99%	?Osteogenesis imperfecta, type XII, 613849
SPINK5	182.2	99%	98%	Atopy, 147050 Netherton syndrome, 256500
SPINT2	66	99%	91%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPRED1	196.7	99%	96%	Legius syndrome, 611431
SPRY4	146.5	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRD5A3	172	100%	99%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713
ST14	177.8	99%	98%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL5	144.8	95%	94%	Amish infantile epilepsy syndrome, 609056
STAMBP	125	99%	97%	Microcephaly-capillary malformation syndrome, 614261
STAT3	132.1	100%	99%	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	140.5	99%	96%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIM1	144	99%	97%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070
STK11	127.1	99%	97%	Melanoma, malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STS	104	99%	95%	Ichthyosis, X-linked, 308100
SUFU	146	99%	97%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	137.1	99%	94%	Multiple sulfatase deficiency, 272200
TALDO1	153.4	100%	99%	Transaldolase deficiency, 606003

TAP1	13.1	52%	18%	Bare lymphocyte syndrome, type I, 604571
TAP2	12.6	50%	20%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 Wegener-like granulomatosis
TAPBP	23.6	83%	53%	Bare lymphocyte syndrome, type I, 604571
TAT	140.9	100%	100%	Tyrosinemia, type II, 276600
TBC1D24	203.2	100%	99%	Deafness , autosomal recessive 86, 614617 Deafness, autosomal dominant 65, 616044 DOOR syndrome, 220500 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021
TBX3	95.1	99%	97%	Ulnar-mammary syndrome, 181450
TCIRG1	127.8	96%	89%	Osteopetrosis, autosomal recessive 1, 259700
TEK	200.5	100%	100%	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	124.9	99%	97%	No OMIM phenotype Melanoma (Aoude (2015) J Natl Cancer Inst 107)
TERT	150	96%	92%	{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	126.8	100%	99%	Branchiooculofacial syndrome, 113620
TGFB2	203	100%	99%	Loeys-Dietz syndrome 4, 614816
TGFBR1	204.7	94%	93%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	212.4	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGM1	185.6	100%	100%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	184.6	100%	99%	Peeling skin syndrome 2, 609796
TINF2	208.7	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130

TMC6	88.2	99%	97%	Epidermodysplasia verruciformis, 226400
TMC8	128.1	98%	94%	Epidermodysplasia verruciformis, 226400
TMEM165	136.2	99%	98%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	106.3	99%	96%	STING-associated vasculopathy, infantile-onset, 615934
TNFRSF11A	152.6	94%	91%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	258.6	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF1A	96.7	91%	89%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFSF11	177.2	99%	96%	Osteopetrosis, autosomal recessive 2, 259710
TNXB	17.8	58%	32%	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
TP63	226.8	100%	100%	ADULT syndrome, 103285 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 129400 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TPCN2	177.2	99%	96%	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267
TREX1	302.9	100%	100%	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448 Vasculopathy, retinal, with cerebral leukodystrophy, 192315 {Systemic lupus erythematosus, susceptibility to}, 152700
TRIM32	147.3	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRIM37	137.3	100%	99%	Mulibrey nanism, 253250
TRPV3	165.4	100%	99%	Olmsted syndrome, 614594 ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400
TSC1	140.4	99%	97%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	150.1	99%	99%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254

TSPEAR	153.1	100%	99%	Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia (Peled et al. (2016) PLOS Genetics online)
TTC37	159.2	99%	98%	Trichohepatoenteric syndrome 1, 222470
TTI2	109.9	100%	99%	Mental retardation, autosomal recessive 39, 615541
TWIST2	124.4	99%	98%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TYR	205.9	100%	100%	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	200.1	100%	100%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBE2A	110.5	99%	98%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBR1	144.2	99%	98%	Johanson-Blizzard syndrome, 243800
UROD	174	99%	97%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	126.7	100%	100%	Porphyria, congenital erythropoietic, 263700
USB1	157.7	99%	97%	Poikiloderma with neutropenia,604173
UVSSA	138.1	100%	99%	UV-sensitive syndrome 3, 614640
VDR	124.6	99%	96%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
VEGFC	194	100%	99%	Lymphedema, hereditary, ID, 615907
VHL	126	97%	90%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VPS13B	170.3	99%	97%	Cohen syndrome, 216550
VPS33B	142.9	100%	99%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085

WAS	72	89%	80%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDR19	170	100%	99%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	186.3	99%	98%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR72	161.3	99%	98%	Amelogenesis imperfecta, type IIA3, 613211
WIPF1	88.4	100%	99%	?Wiskott-Aldrich syndrome 2, 614493
WNT10A	135.1	99%	99%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	146.6	100%	99%	Split-hand/foot malformation 6, 225300
WNT5A	181.5	100%	99%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	230.2	100%	99%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	175.2	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	149.6	99%	96%	Werner syndrome, 277700
XPA	69.2	97%	89%	Xeroderma pigmentosum, group A, 278700
XPC	176.3	100%	99%	Xeroderma pigmentosum, group C, 278720
XYLT1	144.5	92%	88%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	161.9	98%	96%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
ZBTB20	226.5	100%	100%	Primrose syndrome, 259050
ZMPSTE24	151.9	99%	99%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZNF469	102.9	99%	97%	Brittle cornea syndrome 1, 229200
ZNF592	139.4	100%	99%	Spinocerebellar ataxia, autosomal recessive 5, 251300
ZNF750	144.3	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.

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 : April 14th 2017
This list is accurate for panel version DG 2.9

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors