SKIN DISORDERS GENE PANEL DG 2.7/DG 2.8

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AAAS	108	100%	99%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	166.9	99%	97%	Keratoderma, palmoplantar, punctate type IA, 148600
ABCA12	163	99%	97%	Ichthyosis, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCB6	136.3	99%	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	113.8	93%	92%	Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	177.1	100%	99%	Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia, 239850
ABHD5	265.7	99%	99%	Chanarin-Dorfman syndrome, 275630
ACTA2	166.5	100%	99%	Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042 Multisystemic smooth muscle dysfunction syndrome, 613834
ACTB	134.1	98%	93%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACVRL1	138	99%	98%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADAM10	154.1	99%	97%	Reticulate acropigmentation of Kitamura, 615537 {Alzheimer disease 18, susceptibility to}, 615590
ADAM17	140.9	97%	93%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAMTS10	118.7	99%	99%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	134.9	90%	87%	Weill-Marchesani-like syndrome, 613195
ADAMTS2	136.3	99%	98%	Ehlers-Danlos syndrome, type VIIC, 225410
ADAR	131.7	100%	99%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400

AGA	154.1	100%	100%	Aspartylglucosaminuria, 208400
AGPAT2	120.1	98%	92%	Lipodystrophy, congenital generalized, type 1, 608594
AIRE	83.3	99%	93%	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	83.7	97%	89%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	115	100%	99%	Porphyria, acute hepatic, 612740 {Lead poisoning, susceptibility to}, 612740
ALAS2	107.4	99%	96%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	143	100%	99%	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	157.4	100%	99%	Sjogren-Larsson syndrome, 270200
ALDOB	174.3	99%	98%	Fructose intolerance, 229600
ALOX12B	148.4	99%	99%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	145.8	100%	99%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	163.1	100%	100%	Hypophosphatasia, adult, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia, 146300
ALX4	132.6	96%	89%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMELX	95.2	99%	96%	Amelogenesis imperfecta, type 1E, 301200
ANKRD11	107.9	97%	93%	KBG syndrome, 148050
ANTXR1	141.9	98%	96%	GAPO syndrome, 230740 {Hemangioma, capillary infantile, susceptibility to}, 602089

ANTXR2	97.9	97%	91%	Hyaline fibromatosis syndrome, 228600
AP1S3	120	90%	90%	{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	111.5	97%	91%	Hermansky-Pudlak syndrome 2, 608233
APC	175.4	99%	98%	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	183.5	99%	97%	Hypotrichosis 1, 605389
AQP5	116.5	99%	96%	Palmoplantar keratoderma, Bothnian type, 600231
ARHGAP31	128.3	99%	98%	Adams-Oliver syndrome 1, 100300
ARID1A	155.1	95%	90%	Coffin-Siris syndrome 2, 614607
ARID1B	157	95%	90%	Coffin-Siris syndrome 1, 135900
ASIP	114.2	100%	99%	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 [Skin/hair/eye pigmentation 9, dark/light hair], 611742
ASL	118.4	99%	98%	Argininosuccinic aciduria, 207900
ASXL1	168.4	99%	98%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	178.5	99%	99%	Bainbridge-Ropers syndrome, 615485
ATIC	134.7	99%	98%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP2A2	197.2	100%	99%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	135.5	99%	98%	Hailey-Hailey disease, 169600
ATP6V0A2	159.8	100%	99%	Cutis laxa, autosomal recessive, type IIA, 219200 Wrinkly skin syndrome, 278250
ATP7A	157.3	99%	97%	Menkes disease, 309400 Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489
ATR	160.3	98%	96%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	133.3	100%	99%	Colorectal cancer, somatic, 114500

				Oligodontia-colorectal cancer syndrome, 608615
B3GALT6	54	76%	71%	Ehlers-Danlos syndrome, progeroid type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B4GALT7	116.3	96%	95%	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070
BANF1	72.4	99%	96%	Nestor-Guillermo progeria syndrome, 614008
BAP1	154.2	99%	98%	Tumor predisposition syndrome, 614327
BCOR	131.4	99%	97%	Microphthalmia, syndromic 2, 300166
BCS1L	184.4	100%	100%	Bjornstad syndrome, 262000 GRACILE syndrome, 603358 Leigh syndrome, 256000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	139.6	98%	95%	Bloom syndrome, 210900
BLOC1S3	51.7	97%	90%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	106.6	98%	92%	Hermansky-pudlak syndrome 9, 614171
BMS1	94.2	66%	64%	?Aplasia cutis congenita, nonsyndromic, 107600
BRAF	77	89%	79%	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	137.1	99%	96%	Breast cancer, early-onset, 114480 Fanconi anemia, complementation group J, 609054
BSCL2	126.4	100%	99%	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	163.8	100%	99%	Biotinidase deficiency, 253260
C10orf11	167.4	99%	99%	Albinism, oculocutaneous, type VII, 615179
C1QA	130.2	98%	95%	C1q deficiency, 613652
C1QB	195	99%	99%	C1q deficiency, 613652
C1QC	232.3	100%	99%	C1q deficiency, 613652
C2CD3	163.2	95%	95%	?Orofaciodigital syndrome XIV, 615948

C4orf26	226.3	100%	100%	Amelogenesis imperfecta, type IIA4, 614832
CA2	166.3	98%	93%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAPN12	92.7	96%	91%	No OMIM phenotype Modifying factor in ichthyosis
CARD14	120.7	99%	98%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	126.3	97%	96%	Candidiasis, familial, 2, autosomal recessive, 212050
CAST	124.5	97%	93%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAV1	269.1	100%	100%	Pulmonary hypertension, primary, 3, 615343 ?Lipodystrophy, congenital generalized, type 3, 612526 ?Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721
CBL	145.2	99%	98%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia,607785
CBS	127.1	97%	92%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCBE1	81.9	98%	92%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD151	150.2	100%	100%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 [Blood group, Raph], 179620
CDAN1	113.1	98%	96%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDH3	155.9	99%	97%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDK4	137	100%	99%	{Melanoma, cutaneous malignant, 3}, 609048
CDKN2A	77.7	92%	90%	Melanoma and neural system tumor syndrome, 155755 Orolaryngeal cancer, multiple, Pancreatic cancer/melanoma syndrome, 606719 {Melanoma, cutaneous malignant, 2}, 155601
CDSN	25.5	63%	46%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CECR1	111.6	99%	98%	Polyarteritis nodosa, childhood-onset, 615688 ?Sneddon syndrome, 182410
CERS3	139.7	100%	99%	Ichthyosis, congenital, autosomal recessive 9, 615023
CHKB	101.2	99%	96%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	190.5	96%	94%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHSY1	148.3	96%	94%	Temtamy preaxial brachydactyly syndrome, 605282

CHUK	144.9	99%	96%	Cocoon syndrome, 613630
CKAP2L	195.2	98%	95%	Filippi syndrome, 272440
CLDN1	156.7	100%	100%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CNNM4	211.2	98%	98%	Jalili syndrome, 217080
COL17A1	123.3	98%	95%	Epidermolysis bullosa, junctional, localisata variant, 226650 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epithelial recurrent erosion dystrophy, 122400
COL1A2	111.6	96%	93%	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	115.8	95%	88%	Ehlers-Danlos syndrome, type IV, 130050
COL5A1	125.6	97%	95%	Ehlers-Danlos syndrome, classic type, 130000
COL5A2	93.4	99%	96%	Ehlers-Danlos syndrome, classic type, 130000
COL7A1	139.7	99%	97%	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	107	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	60.7	76%	49%	Linear skin defects with multiple congenital anomalies, 300887
CPOX	124.8	91%	85%	Coproporphyria, 121300 Harderoporphyria, 121300
CSTA	119.4	100%	98%	Peeling skin syndrome 4, 607936
CTC1	118.9	99%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTSA	149.8	99%	99%	Galactosialidosis, 256540
CTSC	148.6	100%	100%	Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000

				Periodontitis 1, juvenile, 170650
CXCR4	210.4	100%	99%	Myelokathexis, isolated WHIM syndrome, 193670
CYLD	135.1	97%	93%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606
CYP26C1	89.4	99%	97%	Focal facial dermal dysplasia 4, 614974
CYP4F22	137.9	99%	99%	Ichthyosis, congenital, autosomal recessive 5, 604777
DCAF17	110.1	98%	92%	Woodhouse-Sakati syndrome, 241080
DCLRE1C	144.2	97%	94%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDB2	173.8	100%	99%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DHCR7	176.8	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DKC1	138.1	99%	98%	Dyskeratosis congenita, X-linked, 305000
DLX3	137.4	100%	99%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DLX5	158.6	100%	97%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DOCK6	133.6	99%	97%	Adams-Oliver syndrome 2, 614219
DOCK8	151.8	100%	99%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOLK	201.8	99%	99%	Congenital disorder of glycosylation, type Im, 610768
DSC2	154.4	98%	94%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	105.7	96%	87%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	117.9	99%	98%	?Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	195.3	98%	96%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 Keratosis palmoplantaris striata I, AD, 148700
DSG3	159.4	99%	97%	No OMIM phenotype
DSG4	241	98%	96%	Hypotrichosis 6, 607903
DSP	161.9	99%	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	191	100%	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	119.9	99%	96%	Hermansky-Pudlak syndrome 7, 614076
DUSP6	173.3	100%	99%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EBP	101.3	99%	96%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECM1	178.8	100%	99%	Urbach-Wiethe disease, 247100
EDA	101	90%	79%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	145.9	99%	98%	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	98.6	97%	92%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN3	138.2	100%	98%	Central hypoventilation syndrome, congenital, 209880 Waardenburg syndrome, type 4B, 613265 {Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	226.9	99%	99%	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EDNRB	143.9	95%	91%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580 {Hirschsprung disease, susceptibility to, 2}, 600155
EFEMP2	139.9	100%	100%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	142.4	100%	99%	Craniofrontonasal dysplasia, 304110
EIF2AK3	169.7	96%	91%	Wolcott-Rallison syndrome, 226980
ELN	111.9	99%	98%	Cutis laxa, AD, 123700 Supravalvar aortic stenosis, 185500
ELOVL4	104.8	99%	97%	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 Stargardt disease 3, 600110 ?Spinocerebellar ataxia 34, 133190
ENAM	152.1	100%	99%	Amelogenesis imperfecta, type IB, 104500

				Amelogenesis imperfecta, type IC, 204650
ENG	146.3	98%	96%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	155.5	91%	83%	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	144.7	99%	97%	Vici syndrome, 242840
ERCC2	143.5	100%	99%	Cerebrooculofacioskeletal syndrome 2, 610756 Trichothiodystrophy 1, photosensitive, 601675 Xeroderma pigmentosum, group D, 278730
ERCC3	117.6	99%	98%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	168.4	99%	98%	Fanconi anemia, complementation group Q, 615272 Xeroderma pigmentosum, group F, 278760 Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 ?XFE progeroid syndrome, 610965
ERCC5	161.6	99%	99%	Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G, 278780 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	192.3	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	95.1	93%	81%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
EVC	117.3	93%	90%	Ellis-van Creveld syndrome, 225500 Weyers acrodental dysostosis, 193530
EVC2	125.8	96%	92%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXPH5	225.8	100%	99%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
FAM111B	165.3	100%	100%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704

FAM20A	118.6	97%	92%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	102.3	99%	96%	Raine syndrome, 259775
FAM83G	134.3	99%	99%	No OMIM phenotype Palmoplantar keratoderma with leukonychia and abundant curly hair (Maruthappu et al. (2016) ESDR)
FAM83H	83.2	95%	89%	Amelogenesis imperfecta, type III, 130900
FANCA	130.4	99%	98%	Fanconi anemia, complementation group A, 227650
FANCB	78.3	95%	86%	Fanconi anemia, complementation group B, 300514
FANCC	123.2	98%	96%	Fanconi anemia, complementation group C, 227645
FANCD2	142.7	98%	95%	Fanconi anemia, complementation group D2, 227646
FANCE	117	88%	85%	Fanconi anemia, complementation group E, 600901
FANCF	158.8	100%	100%	Fanconi anemia, complementation group F, 603467
FANCG	149.5	99%	99%	Fanconi anemia, complementation group G, 614082
FANCI	182	99%	97%	Fanconi anemia, complementation group I, 609053
FANCL	86	98%	93%	Fanconi anemia, complementation group L, 614083
FANCM	108.9	97%	92%	No OMIM phenotype Fanconi anemia, complementation group M, 614087
FAT4	245.8	100%	99%	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 Van Maldergem syndrome 2, 615546
FBLN5	124.6	91%	90%	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	75.8	98%	94%	Porokeratosis 9, multiple types, 616631
FECH	142.4	99%	99%	Protoporphyria, erythropoietic, autosomal recessive, 177000
FERMT1	111.7	98%	95%	Kindler syndrome,173650
FGF10	150.1	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	129.9	99%	98%	Hypophosphatemic rickets, autosomal dominant, 193100 Osteomalacia, tumor-induced Tumoral calcinosis, hyperphosphatemic, familial, 211900
FGF3	80.8	95%	83%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	112	99%	96%	Trichomegaly, 190330
FGF8	127.4	85%	79%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	161.5	99%	97%	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	156.4	96%	95%	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	120.2	99%	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	175.4	92%	88%	Fumarase deficiency, 606812

				Leiomyomatosis and renal cell cancer, 150800
FKBP10	174.6	98%	95%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	81.3	99%	98%	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557
FLCN	172.1	99%	98%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	245.1	100%	99%	Ichthyosis vulgaris, 146700 {Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	798.1	100%	99%	No OMIM phenotype ?Atopic dermatitis (Margolis (2014) J Invest Dermatol 134,2272)
FLT4	181.2	99%	98%	Hemangioma, capillary infantile, somatic, 602089 Lymphedema, hereditary, IA, 153100
FNIP1	174.2	99%	97%	No OMIM phenotype
FOXC2	54.5	96%	85%	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 Lymphedema-distichiasis syndrome, 153400
FOXE1	31.4	76%	60%	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FOXN1	110.9	99%	98%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	136.8	98%	91%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 {Diabetes mellitus, type I, susceptibility to}, 222100
FREM1	156.4	99%	98%	Bifid nose with or without anorectal and renal anomalies, 608980 Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485
FUCA1	156.2	99%	99%	Fucosidosis, 230000
FZD6	225.6	100%	100%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
GALNS	105.4	99%	94%	Mucopolysaccharidosis IVA, 253000
GALNT3	144.6	98%	94%	Tumoral calcinosis, hyperphosphatemic, familial, 211900
GAN	210.8	99%	99%	Giant axonal neuropathy-1, 256850
GATA2	145.6	100%	99%	Emberger syndrome, 614038 Immunodeficiency 21, 614172 {Leukemia, acute myeloid, susceptibility to}, 601626 {Myelodysplastic syndrome, susceptibility to}, 614286

GDF2	170.3	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF5	156.9	99%	99%	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	99%	98%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GJA1	218.1	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	239.7	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	357.6	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	420	100%	100%	Erythrokeratodermia variabilis with erythema gyratum repens, 133200
GJB6	226.9	100%	100%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645

				Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	52.2	85%	68%	Leukodystrophy, hypomyelinating, 2, 608804 Lymphedema, hereditary, IC, 613480 Spastic paraplegia 44, autosomal recessive, 613206
GLA	87.1	99%	97%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	93.9	99%	95%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type II, 230600 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010
GLMN	80.8	95%	86%	Glomuvenous malformations,138000
GMPPA	148.3	100%	100%	Alacrima, achalasia, and mental retardation syndrome, 615510
GNA11	173.1	100%	99%	Hypocalcemia, autosomal dominant 2, 615361 Hypocalciuric hypercalcemia, type II, 145981
GNAQ	96.6	88%	74%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	140.1	98%	96%	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	197.1	100%	99%	Geroderma osteodysplasticum, 231070
GPR143	77.7	91%	85%	Nystagmus 6, congenital, X-linked, 300814 Ocular albinism, type I, Nettleship-Falls type, 300500
GRHL2	151.5	100%	99%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	160	100%	100%	Van der Woude syndrome 2, 606713
GSN	131.2	97%	93%	Amyloidosis, Finnish type, 105120
GTF2H5	149.4	100%	99%	Trichothiodystrophy 3, photosensitive, 616395
HCCS	123.6	100%	98%	Linear skin defects with multiple congenital anomalies 1, 309801

HDAC8	165.1	100%	99%	Cornelia de Lange syndrome 5, 300882
HERC2	117.3	79%	75%	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	193.1	100%	100%	Holocarboxylase synthetase deficiency, 253270
HMBS	117.7	100%	99%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	43.1	91%	75%	?Microphthalmia, syndromic 13, 300915
HOXC13	111.7	95%	90%	Ectodermal dysplasia 9, hair/nail type, 614931
HPS1	130.1	99%	99%	Hermansky-Pudlak syndrome 1, 203300
HPS3	159.5	99%	97%	Hermansky-Pudlak syndrome 3, 614072
HPS4	159.7	100%	99%	Hermansky-Pudlak syndrome 4, 614073
HPS5	155	99%	98%	Hermansky-Pudlak syndrome 5, 614074
HPS6	140.9	97%	89%	Hermansky-Pudlak syndrome 6, 614075
HR	104.2	97%	94%	Alopecia universalis, 203655 Atrichia with papular lesions, 209500 Hypotrichosis 4, 146550
HRAS	195.3	99%	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	113	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	125	100%	100%	?Mucopolysaccharidosis type IX, 601492
IDUA	116.9	91%	85%	Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Is, 607016
IFT122	164.4	100%	99%	Cranioectodermal dysplasia 1, 218330

IFT43	130.6	100%	100%	Cranioectodermal dysplasia 3, 614099
IKBKG	56.2	84%	70%	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	136.4	99%	96%	?Candidiasis, familial, 5, autosomal recessive, 613953
IL17RD	144.8	99%	97%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RN	186.9	100%	99%	Interleukin 1 receptor antagonist deficiency, 612852 {Gastric cancer risk after H. pylori infection}, 137215 {Microvascular complications of diabetes 4}, 612628
IL31RA	150.8	100%	99%	Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	106.9	100%	99%	Psoriasis 14, pustular, 614204
INSR	146.5	96%	94%	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968 Leprechaunism, 246200 Rabson-Mendenhall syndrome, 262190
IRF4	198.7	99%	99%	[Skin/hair/eye pigmentation, variation in, 8], 611724
IRF6	136	99%	97%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
ITGA3	154.5	99%	97%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	165.8	99%	98%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGB4	162.4	97%	94%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, with pyloric atresia, 226730
ITGB6	169.4	95%	94%	Amelogenesis imperfecta, type IH, 616221
JUP	159	99%	98%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KAL1	106.4	89%	87%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
KANK2	159.1	99%	99%	Palmoplantar keratoderma and woolly hair, 616099
KAT6B	202.7	99%	98%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736

KCNH1	198.9	100%	99%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNK9	195.6	100%	100%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KIF11	91.9	96%	93%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIT	175	100%	99%	Gastrointestinal stromal tumor, familial, 606764 Germ cell tumors, 273300 Leukemia, acute myeloid, 601626 Mast cell disease, 154800 Piebaldism, 172800
KITLG	89.7	94%	90%	Deafness, congenital, unilateral or asymmetric, 616697 Hyperpigmentation with or without hypopigmentation, 145250 [Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KLK4	210.9	99%	98%	Amelogenesis imperfecta, type IIA1, 204700
KLLN	121.9	100%	99%	Cowden syndrome 4, 615107
KMT2D	162.1	99%	99%	Kabuki syndrome 1, 147920
KRAS	72.1	99%	96%	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	151.5	99%	96%	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	117.5	97%	93%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT13	145.9	99%	98%	White sponge nevus 2, 615785

KRT14	60.4	89%	82%	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	38.2	73%	54%	Pachyonychia congenita 1, 167200 Palmoplantar keratoderma, nonepidermolytic, focal, 613000
KRT17	23.9	56%	35%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT2	143.9	99%	99%	Ichthyosis bullosa of Siemens, 146800
KRT4	130.9	100%	99%	White sponge nevus 1, 193900
KRT5	137.9	99%	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	189.5	93%	87%	Pachyonychia congenita 3, 615726
KRT6B	190.3	94%	89%	Pachyonychia congenita 4, 615728
KRT6C	174.7	88%	80%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	154	100%	99%	?Hypotrichosis 13, 615896
KRT74	156.6	100%	99%	?Ectodermal dysplasia 7, hair/nail type, 614929 ?Hypotrichosis 3, 613981 Woolly hair, autosomal dominant, 194300
KRT75	145.1	100%	99%	{Pseudofolliculitis barbae, susceptibility to}, 612318
KRT81	98.3	98%	94%	Monilethrix, 158000
KRT83	84.5	98%	90%	?Monilethrix, 158000
KRT85	112.3	98%	93%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	103.4	99%	94%	Monilethrix, 158000
KRT9	85.2	97%	94%	Palmoplantar keratoderma, epidermolytic, 144200
LAMA3	173.5	99%	99%	Epidermolysis bullosa, generalized atrophic benign, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700

				Laryngoonychocutaneous syndrome, 245660
LAMB3	131.6	100%	99%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	131.4	99%	98%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMTOR2	175.9	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LDHA	60.1	92%	86%	Glycogen storage disease XI, 612933
LDLRAP1	168.7	99%	96%	Hypercholesterolemia, familial, autosomal recessive, 603813
LEMD3	111.7	95%	90%	Buschke-Ollendorff syndrome, 166700 Melorheostosis with osteopoikilosis, 155950 Osteopoikilosis, 166700
LIPH	148.3	100%	99%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	155.4	99%	96%	Ichthyosis, congenital, autosomal recessive 8, 613943
LMBRD1	81.3	89%	81%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMNA	90.4	96%	89%	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	130.3	99%	95%	Nail-patella syndrome, 161200
LONP1	168.1	97%	95%	CODAS syndrome, 600373
LOR	21.7	88%	54%	Vohwinkel syndrome with ichthyosis, 604117
LPAR6	115.9	99%	98%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN2	129.6	99%	99%	Majeed syndrome, 609628

LTBP3	126.4	98%	96%	Dental anomalies and short stature, 601216
LTBP4	125.8	98%	95%	Cutis laxa, autosomal recessive, type IC, 613177
LYST	151.3	97%	94%	Chediak-Higashi syndrome, 214500
LYZ	192.2	100%	100%	Amyloidosis, renal, 105200
MAP2K1	107.3	99%	95%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	118.5	97%	92%	Cardiofaciocutaneous syndrome 4, 615280
MBTPS2	142.6	99%	97%	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 ?Olmsted syndrome, X-linked, 300918
MED12	116.5	98%	95%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEFV	131.1	95%	91%	Familial Mediterranean fever, AD, 134610 Familial Mediterranean fever, AR, 249100
MGP	164.5	92%	91%	Keutel syndrome, 245150
MITF	163	99%	99%	Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MLH1	188.5	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MLPH	104.8	99%	96%	Griscelli syndrome, type 3, 609227
MMACHC	205.3	100%	100%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP2	172.4	99%	99%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	117.5	99%	99%	Amelogenesis imperfecta, type IIA2, 612529
MPLKIP	87.3	94%	81%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRE11A	57.6	95%	85%	Ataxia-telangiectasia-like disorder, 604391
MSH2	122	98%	92%	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320
MSX1	88.1	97%	91%	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600

MTOR	152.8	100%	99%	Smith-Kingsmore syndrome, 616638
MUTYH	164.2	99%	99%	Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 Gastric cancer, somatic, 613659
MVD	116.4	100%	99%	Porokeratosis 7, multiple types, 614714
MVK	146.1	100%	99%	Hyper-IgD syndrome, 260920 Mevalonic aciduria, 610377 Porokeratosis 3, multiple types, 175900
MYH8	148.6	99%	99%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYO5A	142.9	99%	97%	Griscelli syndrome, type 1, 214450
NAA10	111	99%	96%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAGA	162.6	100%	100%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NBAS	159.5	99%	97%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NCSTN	127.9	100%	99%	Acne inversa, familial, 1, 142690
NDUFB11	101.4	94%	84%	Linear skin defects with multiple congenital anomalies 3, 300952
NF1	140.5	93%	89%	Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Watson syndrome, 193520
NFKBIA	117.7	98%	94%	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132
NHP2	87.3	99%	98%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIPAL4	151.5	99%	94%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	129.3	96%	94%	Cornelia de Lange syndrome 1, 122470
NLRP1	137	99%	96%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia, 615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP3	153.2	100%	99%	CINCA syndrome, 607115 Familial cold-induced inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900

NME1	109.2	99%	98%	Neuroblastoma, 256700
NOD2	148.9	100%	99%	Blau syndrome, 186580 Sarcoidosis, early-onset, 609464 {Inflammatory bowel disease 1}, 266600 {Psoriatic arthritis, susceptibility to}, 607507
NOP10	189.4	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH1	150.2	99%	98%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NRAS	205.7	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	172.3	100%	99%	Beckwith-Wiedemann syndrome, 130650 Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
NSDHL	205.2	99%	99%	CHILD syndrome, 308050 CK syndrome, 300831
OCA2	147.7	98%	96%	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	130.1	96%	88%	No OMIM phenotype
OFD1	56.1	84%	71%	Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424
OSMR	159.9	100%	99%	Amyloidosis, primary localized cutaneous, 1, 105250
PAH	191.3	100%	100%	Phenylketonuria, 261600 [Hyperphenylalaninemia, non-PKU mild], 261600
PALB2	180.1	100%	99%	Fanconi anemia, complementation group N, 610832 {Breast cancer, susceptibility to}, 114480

				{Pancreatic cancer, susceptibility to, 3}, 613348
PAX3	129.7	99%	99%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX9	254.3	99%	98%	Tooth agenesis, selective, 3, 604625
PCNA	102.9	99%	98%	?Ataxia-telangiectasia-like disorder, 615919
PDGFB	107.8	100%	99%	Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907 Meningioma, SIS-related, 607174
PDGFRB	166.4	99%	97%	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	121.4	99%	98%	Prolidase deficiency, 170100
PEX7	138.5	89%	85%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	145.5	98%	96%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	138.6	100%	99%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHYH	86.5	98%	92%	Refsum disease, 266500
PIEZO1	156.3	99%	97%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 Lymphedema, hereditary, III, 616843
PIGA	102.1	92%	84%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	128.6	95%	89%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	171.1	100%	100%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	136.5	99%	98%	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	155.7	99%	97%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Peters anomaly, 604229 Ring dermoid of cornea, 180550
PKP1	135.1	99%	98%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLCD1	124.1	99%	97%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCG2	133.6	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	114.2	99%	98%	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	133.1	87%	87%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	92.4	97%	89%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	149.8	100%	99%	Ehlers-Danlos syndrome, type VI, 225400
PMS2	95.9	83%	80%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome, 276300
PMVK	124	100%	99%	Porokeratosis 1, multiple types, 175800
PNPLA1	194.6	99%	98%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	127.6	99%	97%	Neutral lipid storage disease with myopathy, 610717
POC1A	150.5	100%	99%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POFUT1	154.1	99%	97%	Dowling-Degos disease 2, 615327
POGLUT1	146.8	99%	95%	Dowling-Degos disease 4, 615696
POLD1	108.5	94%	91%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 {Colorectal cancer, susceptibility to, 10}, 612591
POLH	171.7	100%	99%	Xeroderma pigmentosum, variant type, 278750
POLR1C	124.8	99%	95%	Leukodystrophy, hypomyelinating, 11, 616494

				Treacher Collins syndrome 3, 248390
POLR1D	196.1	100%	100%	Treacher Collins syndrome 2, 613717
POLR3A	162.2	100%	99%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	168.9	99%	98%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	101.8	100%	99%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 {Obesity, early-onset, susceptibility to}, 601665
POMP	146.6	91%	87%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
PORCN	145.4	100%	99%	Focal dermal hypoplasia, 305600
PPOX	108.1	99%	97%	Porphyria variegata, 176200
PQBP1	174.3	97%	96%	Renpenning syndrome, 309500
PRKAR1A	102.8	97%	91%	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	161.6	99%	98%	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	93.7	99%	99%	Acne inversa, familial, 2, 613736
PSMB8	16.1	57%	25%	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040
PSTPIP1	99.8	99%	95%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	138.6	98%	96%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7, 610828
PTCH2	130.2	99%	97%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, 155255
PTDSS1	151.7	100%	100%	Lenz-Majewski hyperostotic dwarfism, 151050

PTEN	152.4	99%	98%	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	146.8	98%	89%	Brachydactyly, type E2, 613382 Humoral hypercalcemia of malignancy
PTPN11	101.2	96%	90%	LEOPARD syndrome 1, 151100 Leukemia, juvenile myelomonocytic, somatic, 607785 Metachondromatosis, 156250 Noonan syndrome 1, 163950
PTPN14	199.6	99%	98%	Choanal atresia and lymphedema, 613611
PTPRF	194.8	100%	100%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PTRF	142.9	99%	98%	Lipodystrophy, congenital generalized, type 4, 613327
PVRL1	163	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
PVRL4	146.8	100%	99%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
PYCR1	105.4	99%	94%	Cutis laxa, autosomal recessive, type IIB, 612940 Cutis laxa, autosomal recessive, type IIIB, 614438
RAB23	121.6	99%	98%	Carpenter syndrome, 201000
RAB27A	178	100%	98%	Griscelli syndrome, type 2, 607624
RAD21	97.7	99%	96%	Cornelia de Lange syndrome 4, 614701
RAD50	106.1	92%	85%	Nijmegen breakage syndrome-like disorder, 613078
RAF1	138	100%	99%	Cardiomyopathy, dilated, 1NN, 615916 LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553
RAG1	230.7	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	258.8	100%	99%	Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457
RAI1	142.2	100%	99%	Smith-Magenis syndrome, 182290
RBBP8	112.7	99%	95%	Jawad syndrome, 251255 Pancreatic carcinoma, somatic Seckel syndrome 2, 606744
RBM28	160.7	100%	100%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBP4	120.7	98%	96%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	86.4	93%	85%	Adams-Oliver syndrome 3, 614814
RECQL4	150	98%	97%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RHBDF2	113.1	99%	98%	Tylosis with esophageal cancer, 148500
RIN2	122.2	99%	99%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	157.6	99%	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	149.3	100%	99%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	125.1	94%	84%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	207.6	99%	97%	Aicardi-Goutieres syndrome 3, 610329
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROGDI	134	97%	95%	Kohlschutter-Tonz syndrome, 226750
RPL21	75	72%	57%	Hypotrichosis 12, 615885
RSPO1	123.9	100%	99%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO4	118.5	100%	98%	Anonychia congenita, 206800
RTEL1	127.4	99%	96%	Dyskeratosis congenita, autosomal dominant 4, 615190

				Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
RUNX2	106.2	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	183.8	99%	98%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SAMHD1	149.9	99%	98%	Aicardi-Goutieres syndrome 5, 612952 ?Chilblain lupus 2, 614415
SART3	142.7	99%	98%	No OMIM phenotype Disseminated superficial actinic porokeratosis (Zhang (2005) Br J Dermatol 152,658)
SASH1	151.8	98%	96%	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	162.6	99%	98%	No OMIM phenotype Keratosis follicularis spinulosa decalvans (Gimelli (2002) Hum Genet 111,235)
SATB2	120.1	98%	93%	Glass syndrome, 612313
SCN10A	194.2	99%	99%	Episodic pain syndrome, familial, 2, 615551
SCN11A	155	99%	97%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	160.5	98%	96%	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
SEC23B	185.2	97%	96%	Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SERPINB7	147.9	99%	99%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	167.2	100%	99%	Peeling skin syndrome 5,617115
SERPINH1	171	100%	100%	?Osteogenesis imperfecta, type X, 613848

				{Preterm premature rupture of the membranes, susceptibility to}, 610504
SHOC2	148.9	99%	98%	Noonan-like syndrome with loose anagen hair, 607721
SKI	90.1	98%	95%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	26.9	80%	57%	Trichohepatoenteric syndrome 2, 614602
SLC17A9	130.3	95%	95%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC24A4	143	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	123	98%	94%	Albinism, oculocutaneous, type VI, 113750 [Skin/hair/eye pigmentation 4, fair/dark skin], 113750
SLC26A2	266	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	166.2	99%	99%	Ichthyosis prematurity syndrome, 608649
SLC29A3	226.4	99%	99%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A10	171.2	100%	99%	Arterial tortuosity syndrome, 208050
SLC39A13	124.2	99%	97%	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350
SLC39A4	88.4	99%	96%	Acrodermatitis enteropathica, 201100
SLC45A2	146.2	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	150.3	99%	98%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A19	173.8	99%	99%	Hartnup disorder, 234500 Hyperglycinuria, 138500 Iminoglycinuria, digenic, 242600
SLC7A7	127.2	100%	100%	Lysinuric protein intolerance, 222700
SLCO2A1	114.9	99%	99%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLURP1	112.5	99%	94%	Meleda disease, 248300
SLX4	127.4	100%	99%	Fanconi anemia, complementation group P, 613951
SMAD3	142.2	99%	98%	Loeys-Dietz syndrome 3, 613795

SMARCA2	131.1	96%	94%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	156.7	99%	98%	Coffin-Siris syndrome 4, 614609 {Rhabdoid tumor predisposition syndrome 2}, 613325
SMARCAD1	88.2	98%	92%	Adermatoglyphia, 136000
SMARCAL1	148.2	100%	99%	Schimke immunoosseous dysplasia, 242900
SMARCB1	248.9	100%	100%	Coffin-Siris syndrome 3, 614608 Rhabdoid tumors, somatic, 609322 {Rhabdoid predisposition syndrome 1}, 609322 {Schwannomatosis-1, susceptibility to}, 162091
SMO	169.5	97%	92%	Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome,somatic mosaic, 601707
SMOC2	119.4	97%	92%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	151.2	100%	99%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SNAP29	160.1	100%	100%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNRPE	100.5	99%	94%	Hypotrichosis 11, 615059
SNX10	117.9	100%	99%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	106.9	96%	90%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOX10	80.4	97%	93%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX18	27.1	83%	59%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	111.4	99%	97%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SP7	182.3	100%	100%	?Osteogenesis imperfecta, type XII, 613849
SPINK5	170.1	99%	96%	Atopy, 147050 Netherton syndrome, 256500
SPINT2	68.3	98%	90%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPRED1	187.2	98%	96%	Legius syndrome, 611431
SPRY4	153	99%	99%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRD5A3	166.9	99%	99%	Congenital disorder of glycosylation, type Iq, 612379 Kahrizi syndrome, 612713

ST14	171.8	99%	98%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL5	138.4	95%	94%	Amish infantile epilepsy syndrome, 609056
STAMBP	130.1	99%	96%	Microcephaly-capillary malformation syndrome, 614261
STAT3	141	99%	98%	Autoimmune disease, multisystem, infantile-onset, 615952 Hyper-IgE recurrent infection syndrome, 147060
STAT5B	141.7	99%	96%	Growth hormone insensitivity with immunodeficiency, 245590 Leukemia, acute promyelocytic, somatic, 102578
STIM1	143.1	99%	96%	Immunodeficiency 10, 612783 Myopathy, tubular aggregate, 1 160565 Stormorken syndrome, 185070
STK11	130.1	99%	94%	Melanoma, malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STS	111.8	99%	96%	Ichthyosis, X-linked, 308100
SUFU	149.5	99%	97%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 {Meningioma, familial, susceptibility to}, 607174
SUMF1	138.4	97%	92%	Multiple sulfatase deficiency, 272200
TALDO1	143.2	100%	99%	Transaldolase deficiency, 606003
TAT	141.9	100%	100%	Tyrosinemia, type II, 276600
TBC1D24	178.8	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	97.9	99%	96%	Ulnar-mammary syndrome, 181450
TCIRG1	125.6	95%	88%	Osteopetrosis, autosomal recessive 1, 259700
TEK	214.7	100%	99%	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
TERT	148.3	96%	91%	{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	138	100%	99%	Branchiooculofacial syndrome, 113620
TGFB2	182.2	100%	99%	Loeys-Dietz syndrome 4, 614816
TGFBR1	213.8	95%	93%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	215.3	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGM1	187.8	100%	100%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM5	187.8	100%	99%	Peeling skin syndrome 2, 609796
TINF2	196.8	100%	99%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMC6	88	99%	96%	Epidermodysplasia verruciformis, 226400
TMC8	119	97%	92%	Epidermodysplasia verruciformis, 226400
TMEM165	122.2	98%	96%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	102.7	99%	96%	STING-associated vasculopathy, infantile-onset, 615934
TNFRSF11A	144.8	93%	91%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301 {Paget disease of bone 2, early-onset}, 602080
TNFRSF11B	274.1	100%	100%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF1A	104.1	91%	88%	Periodic fever, familial, 142680 {Multiple sclerosis, susceptibility to, 5}, 614810
TNFSF11	184.1	99%	94%	Osteopetrosis, autosomal recessive 2, 259710
TNXB	17.8	59%	32%	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408 Vesicoureteral reflux 8, 615963
TP63	217.6	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	178.6	99%	97%	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267
TREX1	272.2	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	152.9	100%	100%	Muscular dystrophy, limb-girdle, type 2H, 254110 ?Bardet-Biedl syndrome 11, 615988
TRIM37	131.2	99%	98%	Mulibrey nanism, 253250
TRPV3	184.2	100%	99%	Olmsted syndrome, 614594 ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400
TSC1	149.6	99%	98%	Lymphangioleiomyomatosis, 606690 Tuberous sclerosis-1, 191100
TSC2	144.5	99%	98%	Lymphangioleiomyomatosis, somatic, 606690 Tuberous sclerosis-2, 613254
TSPEAR	154.4	100%	99%	Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia (Peled et al. (2016) PLOS Genetics online)
TTC37	139	99%	97%	Trichohepatoenteric syndrome 1, 222470
TTI2	114.2	99%	98%	Mental retardation, autosomal recessive 39, 615541
TWIST2	134	99%	96%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TYR	205.6	100%	99%	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	209.7	100%	100%	Albinism, oculocutaneous, type III, 203290 [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271
UBE2A	115.3	99%	95%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBR1	144.4	98%	95%	Johanson-Blizzard syndrome, 243800
UROD	178.2	100%	99%	Porphyria cutanea tarda, 176100 Porphyria, hepatoerythropoietic, 176100
UROS	119.9	100%	100%	Porphyria, congenital erythropoietic, 263700

USB1	144.5	99%	97%	Poikiloderma with neutropenia,604173
UVSSA	134.4	99%	99%	UV-sensitive syndrome 3, 614640
VDR	134.4	99%	96%	Rickets, vitamin D-resistant, type IIA, 277440 ?Osteoporosis, involutional, 166710
VEGFC	181	99%	99%	Lymphedema, hereditary, ID, 615907
VHL	120.5	95%	88%	Erythrocytosis, familial, 2, 263400 Hemangioblastoma, cerebellar, somatic Pheochromocytoma, 171300 Renal cell carcinoma, somatic, 144700 von Hippel-Lindau syndrome, 193300
VPS13B	156.8	98%	96%	Cohen syndrome, 216550
WAS	68.7	87%	78%	Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, 313900 Thrombocytopenia, X-linked, intermittent, 313900 Wiskott-Aldrich syndrome, 301000
WDR19	153.5	99%	97%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	167.2	98%	96%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR72	161.7	99%	97%	Amelogenesis imperfecta, type IIA3, 613211
WIPF1	94	99%	98%	?Wiskott-Aldrich syndrome 2, 614493
WNT10A	114.7	99%	98%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	142.7	100%	99%	Split-hand/foot malformation 6, 225300
WNT5A	175.2	100%	100%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	226.4	100%	100%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	164.3	100%	100%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	143.3	97%	93%	Werner syndrome, 277700
XPA	58.3	95%	84%	Xeroderma pigmentosum, group A, 278700
XPC	163.9	100%	99%	Xeroderma pigmentosum, group C, 278720

XYLT1	148.3	93%	88%	Desbuquois dysplasia 2, 615777 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	152.3	98%	95%	Spondyloocular syndrome, 605822 {Pseudoxanthoma elasticum, modifier of severity of}, 264800
ZBTB20	248.6	100%	100%	Primrose syndrome, 259050
ZMPSTE24	134.4	99%	98%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZNF469	91.3	99%	96%	Brittle cornea syndrome 1, 229200
ZNF592	140.6	100%	99%	Spinocerebellar ataxia, autosomal recessive 5, 251300
ZNF750	161.7	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 : October 1st, 2016.
This list is accurate for panel versions DG 2.7 and DG 2.8 From DG 2.7 to DG 2.8 no changes were made to the content of the gene panels.

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