SKIN DISORDERS GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AAAS	87.2	100%	99%	Achalasia-addisonianism-alacrimia syndrome,231550
AAGAB	153.2	97%	92%	Keratoderma palmoplantar punctate type IA,148600
ABCA12	145.7	98%	96%	Ichthyosis, autosomal recessive 4B (harlequin),242500 Ichthyosis, congenital, autosomal recessive 4A,601277
ABCB6	111.6	100%	99%	Dyschromatosis universalis hereditaria 3,615402 Microphthalmia,isolated, with coloboma 7,614497 [Blood group, Langereis system],111600
ABCC6	94.8	93%	91%	Arterial calcification generalized of infancy 2,614473 Pseudoxanthoma elasticum,264800 Pseudoxanthoma elasticum, forme fruste,177850
ABCC9	156.9	100%	99%	Atrial fibrillation familial 12,614050 Cardiomyopathy, dilated, 1O,608569 Hypertrichotic osteochondrodysplasia, 239850
ABHD5	244.7	100%	100%	Chanarin-Dorfman syndrome,275630
ACTA2	150.7	100%	100%	Aortic aneurysm familial thoracic 6,611788 Moyamoya disease 5,614042 Multisystemic smooth muscle dysfunction syndrome,613834
ACVRL1	117.6	100%	99%	Telangiectasia hereditary hemorrhagic type 2,600376
ADAM10	141.1	99%	97%	Reticulate acropigmentation of Kitamura,615537 {Alzheimer disease 18, susceptibility to},615590
ADAM17	125.2	98%	91%	Inflammatory skin and bowel disease neonatal,614328
ADAMTS10	97.1	99%	99%	Weill-Marchesani syndrome 1 recessive,277600
ADAMTS17	112.2	88%	84%	Weill-Marchesani-like syndrome,613195
ADAMTS2	112.9	100%	96%	Ehlers-Danlos syndrome type VIIC,225410
ADAR	107.2	99%	98%	Aicardi-Goutieres syndrome 6,615010 Dyschromatosis symmetrica hereditaria,127400
AGA	134	100%	100%	Aspartylglucosaminuria,208400
AGPAT2	101.5	100%	94%	Lipodystrophy congenital generalized type 1,608594

AIRE	66.7	100%	92%	Autoimmune polyendocrinopathy syndrome type I with or without reversible metaphyseal dysplasia,240300
AKT1	146.7	99%	95%	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	73.2	99%	86%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome,603387
ALAD	95.2	100%	98%	Porphyria acute hepatic,612740 {Lead poisoning, susceptibility to},612740
ALAS2	63.7	97%	91%	Anemia sideroblastic X-linked,300751 Protoporphyria, erythropoietic, X-linked,300752
ALDH18A1	125.3	100%	100%	Cutis laxa autosomal recessive type IIIA,219150
ALDH3A2	133.4	100%	100%	Sjogren-Larsson syndrome,270200
ALDOB	152.7	100%	100%	Fructose intolerance,229600
ALOX12B	124.4	99%	98%	Ichthyosis congenital autosomal recessive 2,242100
ALOXE3	114.8	100%	99%	Ichthyosis congenital autosomal recessive 3,606545
ALPL	134.9	100%	100%	Hypophosphatasia, adult,146300 Hypophosphatasia, childhood,241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia,146300
ALX4	112.6	97%	85%	Frontonasal dysplasia 2,613451 Parietal foramina 2,609597 {Craniosynostosis 5, susceptibility to},615529
AMELX	47.2	100%	90%	Amelogenesis imperfecta, type 1E,301200
ANKRD11	85.4	96%	91%	KBG syndrome,148050
ANTXR1	127.4	97%	96%	GAPO syndrome,230740 {Hemangioma, capillary infantile, susceptibility to},602089
ANTXR2	84.7	99%	88%	Hyaline fibromatosis syndrome,228600
AP3B1	93.9	99%	92%	Hermansky-Pudlak syndrome 2,608233

APC	152.5	99%	97%	Adenoma,periampullary,somatic,175100 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,s
APCDD1	162.2	100%	97%	Hypotrichosis 1,605389
AQP5	106.9	100%	97%	Palmoplantar keratoderma, Bothnian type,600231
ARHGAP31	103.4	99%	98%	Adams-Oliver syndrome 1,100300
ARID1A	135.8	91%	87%	Mental retardation autosomal dominant 14,614607
ARID1B	131.2	92%	84%	Mental retardation,autosomal dominant 12,614562
ASIP	89.4	100%	96%	[Skin/hair/eye pigmentation 9],611742
ASL	88.5	100%	96%	Argininosuccinic aciduria,207900
ASXL1	145	99%	98%	Bohring-Opitz syndrome,605039 Myelodysplastic syndrome,somatic,614286
ASXL3	150.2	98%	97%	Bainbridge-Ropers syndrome ,615485
ATIC	114.3	100%	98%	AICA-ribosiduria due to ATIC deficiency,608688
ATP2A2	166.4	100%	99%	Acrokeratosis verruciformis,101900 Darier disease,124200
ATP2C1	120	99%	98%	Hailey-Hailey disease,169600
ATP6V0A2	141.9	100%	99%	Cutis laxa,autosomal recessive,type IIA,219200 Wrinkly skin syndrome,278250
ATP7A	88.3	99%	93%	Menkes disease,309400 Occipital horn syndrome,304150 Spinal muscular atrophy,distal,X-linked,300489
ATR	137.4	97%	95%	Cutaneous telangiectasia and cancer syndrome familial,614564 Seckel syndrome 1,210600
AXIN2	111.4	100%	99%	Colorectal cancer somatic,114500 Oligodontia-colorectal cancer syndrome,608615
B3GALT6	47.2	70%	64%	Ehlers-Danlos syndrome progeroid type 2,615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1,with or without fractures,271640
B4GALT7	96.5	95%	94%	Ehlers-Danlos syndrome progeroid type 1,130070

BANF1	57.3	99%	91%	Nestor-Guillermo progeria syndrome,614008
BAP1	121.5	98%	97%	Tumor predisposition syndrome,614327
BCOR	71	98%	95%	Microphthalmia syndromic 2,300166
BCS1L	147	100%	100%	Bjornstad syndrome,262000 GRACILE syndrome,603358 Leigh syndrome,256000 Mitochondrial complex III defiency, nuclear type 1,124000
BLM	115.5	98%	92%	Bloom syndrome,210900
BLOC1S3	38.5	99%	84%	Hermansky-Pudlak syndrome 8,614077
BLOC1S6	89.9	99%	84%	Hermansky-pudlak syndrome 9,614171
BMS1	83.4	65%	64%	Aplasia cutis congenita, nonsyndromic,107600
BRAF	68.9	89%	83%	Adenocarcinoma of lung,somatic,211980 Cardiofaciocutaneous syndrome,115150 LEOPARD syndrome 3,613707 Noonan syndrome 7,613706
BRIP1	120.7	99%	97%	Breast cancer early-onset,114480 Fanconi anemia,complementation group J,609054
BSCL2	107.4	100%	98%	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	122.5	100%	99%	Biotinidase deficiency,253260
C10orf11	135.8	99%	99%	Albinism, oculocutaneous type VII,615179
C1QA	106.9	100%	88%	C1q deficiency,613652
C1QB	157.3	100%	100%	C1q deficiency,613652
C1QC	180.5	100%	98%	C1q deficiency,613652
C2CD3	140.4	95%	95%	?Orofaciodigital syndrome XIV,615948
C4orf26	183.7	100%	100%	Amelogenesis imperfecta, type IIA4,614832
CA2	130.8	90%	85%	Osteopetrosis,autosomal recessive 3,with renal tubular acidosis,259730
CARD14	93.1	99%	96%	Pityriasis rubra pilaris,173200 Psoriasis 2,602723
CARD9	100.1	95%	95%	Candidiasis,familial 2,autosomal recessive,212050
CAST	111.8	97%	90%	PLACK syndrome,616295

CAV1	238.9	100%	100%	?Lipodystrophy,congenital generalized,type 3,612526 ?Partial lipodystrophy, congenital cataracts and neurodegeneration syndrome,606721 Pulmonary hypertension, primary, 3,615343
CBL	118.2	98%	96%	Noonan syndrome-like disorder,with or without juvenile meylomonocytic leukemia,613563
CBS	106.9	99%	91%	Homocystinuria B6-responsive and nonresponsive types,236200 Thrombosis,hyperhomocysteinemic,236200
CCBE1	69.9	100%	95%	Hennekam lymphangiectasia-lymphedema syndrome,235510
CD151	122.1	100%	100%	Nephropathy with pretibial epidermolysis bullosa and deafness,609057 [Blood group, Raph],179620
CDAN1	87.3	97%	95%	Dyserythropoietic anemia, congenital, type Ia,224120
CDH3	132.2	100%	97%	Ectodermal dysplasia,ectrodactyly and macular dystrophy,225280 Hypotrichosis, congenital, with juvenile macular dystrophy,601553
CDK4	113.3	100%	97%	{Melanoma, cutaneous malignant, 3},609048
CDKN2A	56.4	93%	91%	Melanoma and neural system tumor syndrome,155755 Pancreatic cancer/melanoma syndrome,606719 {Melanoma,cutaneous malignant, 2},155601
CDSN	16.5	59%	32%	Hypotrichosis 2,146520 Peeling skin syndrome 1,270300
CECR1	95.6	100%	95%	?Sneddon syndrome,182410 Polyarteritis nodosa, childhood-onset,615688
CERS3	118.7	100%	98%	Ichthyosis, congenital, autosomal recessive 9,615023
CHKB	82.4	98%	90%	Muscular dystrophy congenital megaconial type,602541
CHST14	155.8	94%	92%	Ehlers-Danlos syndrome musculocontractural type,601776
CHSY1	121.1	94%	93%	Temtamy preaxial brachydactyly syndrome,605282
CHUK	135.4	99%	95%	Cocoon syndrome,613630
CKAP2L	162.5	98%	95%	Filippi syndrome,272440
CLDN1	138.4	100%	100%	Ichthyosis,leukocyte vacuoles,alopecia and sclerosing cholangitis,607626
CNNM4	174.3	98%	97%	Jalili syndrome,217080
COL17A1	99.3	96%	92%	Epidermolysis bullosa,junctional,non-Herlitz type,226650

COL1A2	96.1	96%	92%	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},16671
COL3A1	100.5	89%	84%	Ehlers-Danlos syndrome,type III,130020 Ehlers-Danlos syndrome, type IV,130050
COL5A1	96.6	95%	92%	Ehlers-Danlos syndrome, classic type I,130000
COL5A2	78.4	99%	96%	Ehlers-Danlos syndrome, classic type I,130000
COL7A1	111.7	99%	95%	EBD inversa,226600 EBD, Bart type,132000 Epidermolysis bullosa dystrophica, AD,131750 Epidermolysis bullosa dystrophica, AR,226600 Epidermolysis bullosa pruriginosa,604129 Epidermolysis bullosa,pretibial,131850 Toenail dystrophy,isolated,607523 Tra
COX4I2	93.5	100%	100%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis,612714
COX7B	41.8	79%	54%	Linear skin defects with multiple congenital anomalies,300887
CPOX	96.6	84%	79%	Coproporphyria,121300 Harderoporphyria,121300
CSTA	99.9	100%	99%	Exfoliative ichthyosis,autosomal recessive,ichthyosis bullosa of Siemens-like,607936
CTC1	93.8	100%	99%	Cerebroretinal microangiopathy with calcifications and cysts,612199
CTSA	126.3	99%	96%	Galactosialidosis,256540
CTSC	128.6	100%	100%	Haim-Munk syndrome,245010 Papillon-Lefevre syndrome,245000 Periodontitis 1, juvenile,170650
CXCR4	195.6	100%	100%	Myelokathexis, isolated WHIM syndrome, 193670
CYLD	116.2	98%	93%	Brooke-Spiegler syndrome,605041 Cylindromatosis,familial,132700 Trichoepithelioma,multiple familial,1,601606

CYP26C1	72.2	100%	95%	Focal facial dermal dysplasia 4,614974
CYP4F22	112.6	100%	99%	Ichthyosis,congenital,autosomal recessive 5,604777
DCAF17	92.2	99%	92%	Woodhouse-Sakati syndrome,241080
DCLRE1C	120.5	95%	92%	Omenn syndrome,603554 Severe combined immunodeficiency, Athabascan type,602450
DDB2	148.5	100%	98%	Xeroderma pigmentosum,group E,DDB-negative subtype,278740
DHCR7	149.9	100%	100%	Smith-Lemli-Opitz syndrome,270400
DKC1	77.4	99%	92%	Dyskeratosis congenita X-linked,305000
DLX3	112.7	100%	96%	Amelogenesis imperfecta,type IV,104510 Trichodontoosseous syndrome,190320
DLX5	126	100%	95%	?Split-hand/foot malformation 1 with sensorineural hearing loss,220600
DOCK6	108.9	99%	95%	Adams-Oliver syndrome 2,614219
DOCK8	129.4	100%	99%	Hyper-IgE recurrent infection syndrome autosomal recessive,243700
DOLK	170.5	100%	100%	Congenital disorder of glycosylation, type Im,610768
DSC2	145.9	97%	95%	Arrhythmogenic right ventricular dysplasia 11Without/with mild palmoplantar keratoderma and woolly hair,610476
DSC3	87.8	91%	81%	?Hypotrichosis and recurrent skin vesicles,613102
DSE	107	100%	96%	?Ehlers-Danlos syndrome, musculocontractural type 2,615539
DSG1	169.4	98%	95%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE,615508 Keratosis palmoplantaris striata I,AD,148700
DSG3	137	100%	98%	No OMIM phenotype
DSG4	214.6	98%	96%	Hypotrichosis 6,607903
DSP	131.6	99%	97%	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 Kerato
DSPP	182.5	100%	98%	Deafness,autosomal dominant 36,with dentinogenesis,605594 Dentin dysplasia,type II,125420 Dentinogenesis imperfecta, Shields type II,125490 Dentinogenesis imperfecta, Shields type III, 125500
DTNBP1	100.5	100%	95%	Hermansky-Pudlak syndrome 7,614076 {Schizophrenia},181500

DUSP6	144.6	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia,615269
EBP	71	97%	89%	Chondrodysplasia punctata X-linked dominant,302960
ECM1	152.7	100%	100%	Urbach-Wiethe disease,247100
EDA	47.4	80%	73%	Ectodermal dysplasia 1,hypohidrotic,X-linked,305100 Tooth agenesis,selective,X-linked 1,313500
EDAR	118.2	100%	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	75.6	91%	91%	Ectodermal dysplasia 11A,hypohidrotic/hair/tooth type, autosomal dominant,614940 Ectodermal dysplasia 11B,hypohidrotic/hair/tooth type, autosomal recessive,614941
EDN3	106.9	100%	95%	Central hypoventilation syndrome congenital,209880 Waardenburg syndrome, type 4B,613265 {Hirshprung disease,susceptibility to,4},613712
EDNRA	206.4	100%	100%	mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to},157300
EDNRB	124.7	95%	89%	ABCD syndrome,600501 Waardenburg syndrome, type 4A,277580 {Hirshprung disease, susceptibility to, 2},600155
EFEMP2	110	100%	100%	Cutis laxa,autosomal recessive,type IB,614437
EFNB1	82.8	100%	98%	Craniofrontonasal dysplasia,304110
EIF2AK3	152.8	97%	89%	Wolcott-Rallison syndrome,226980
ELN	91.5	99%	96%	Cutis laxa AD,123700 Supravalvar aortic stenosis,185500
ELOVL4	77.5	100%	98%	?Spinocerebellar ataxia 34,133190 Ichthyosis,spastic quadriplegia and mental retardation,614457 Stargardt disease 3,600110
ENAM	124.6	100%	100%	Amelogenesis imperfecta type IB,104500 Amelogenesis imperfecta type IC,204650
ENG	122.7	98%	95%	Telangiectasia,hereditary hemorrhagic,type 1,187300

ENPP1	134.4	88%	82%	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	123.6	99%	94%	Vici syndrome,242840
ERCC2	120	100%	99%	Cerebrooculofacioskeletal syndrome 2,610756 Trichothiodystrophy 1, photosensitive,601675 Xeroderma pigmentosum, group D,278730
ERCC3	94.3	100%	99%	Trichothiodystrophy 2, photosensitive,616390 Xeroderma pigmentosum, group B,610651
ERCC4	144.3	100%	99%	Fanconi anemia,complementation group Q,615272 Xeroderma pigmentosum, group F,278760 Xeroderma pigmentosum, type F/Cockayne syndrome,278760 XFE progeroid syndrome,610965
ERCC5	133.9	100%	96%	Xeroderma pigmentosum, group G,278780 Xeroderma pigmentosum, group G/Cockayne syndrome,278780
ERCC6	165.9	100%	100%	Cerebrooculofacioskeletal syndrome 1,214150 Cockayne syndrome, type B,133540 De Sanctis-Cacchione syndrome,278800 UV-sensitive syndrome I,600630 {Lung cancer, susceptibility to},211980 {Macular degeneration, age-related, susceptibility to 5},613761
ERCC8	86.8	90%	83%	Cockayne syndrome type A,216400 UV-sensitive syndrome 2,614621
EVC	95.9	94%	89%	Ellis-van Creveld syndrome,225500 Weyers acrodental dysostosis,193530
EVC2	105.5	95%	91%	Ellis-van Creveld syndrome,225500 Weyers acrodental dysostosis,193530
EXPH5	196.7	100%	100%	Epidermolysis bullosa,nonspecific,autosomal recessive,615028
FAM111B	159.6	100%	100%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis,615704
FAM20A	101.9	95%	91%	Amelogenesis imperfecta,type IG (enamel-renal syndrome),204690
FAM20C	79.6	95%	85%	Raine syndrome,259775
FAM83H	60.9	92%	86%	Amelogenesis imperfecta type 3,130900

FANCA	107	98%	95%	Fanconi anemia complementation group A,227650
FANCB	49.3	89%	75%	Fanconi anemia complementation group B,300514
FANCC	108.3	100%	94%	Fanconi anemia complementation group C,227645
FANCD2	125.5	98%	96%	Fanconi anemia complementation group D2,227646
FANCE	92.2	84%	84%	Fanconi anemia complementation group E,600901
FANCF	120.8	100%	100%	Fanconi anemia complementation group F,603467
FANCG	116.2	100%	98%	Fanconi anemia complementation group G,614082
FANCI	158.1	98%	96%	Fanconi anemia complementation group I,609053
FANCL	73.4	100%	92%	Fanconi anemia complementation group L,614083
FANCM	98.4	98%	94%	Fanconi anemia complementation group M,614087
FAT4	220.5	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 2,616006 Van Maldergem syndrome 2,615546
FBLN5	100.8	91%	89%	Cutis laxa,autosomal dominant 2,614434 Cutis laxa,autosomal recessive,type IA,219100 Macular degeneration,age-related,3,608895
FDPS	58.4	98%	89%	Porokeratosis 9,multiple types,616631
FECH	122.4	100%	100%	Protoporphyria erythropoietic autosomal recessive,177000
FERMT1	97.3	98%	94%	Kindler syndrome,173650
FGF10	140.5	100%	100%	Aplasia of lacrimal and salivary glands,180920 LADD syndrome,149730
FGF23	115.8	100%	97%	Hypophosphatemic rickets,autosomal dominant,193100 Osteomalacia,tumor-induced Tumoral calcinosis,hyperphosphatemic,familial,211900
FGF3	49.5	88%	75%	Deafness,congenital with inner ear agenesis,microtia and microdontia,610706
FGF5	72	92%	87%	trichomegaly,190330
FGF8	90.6	81%	74%	Hypogonadotropic hypogonadism 6 with or without anosmia,612702
FGFR1	135.5	100%	95%	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	133.9	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 Crouzon s
FGFR3	91.3	100%	98%	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 syndr
FH	160.3	90%	87%	Fumarase deficiency,606812 Leiomyomatosis and renal cell cancer,150800
FKBP10	141.6	97%	88%	Bruck syndrome 1,259450 Osteogenesis imperfecta type XI,610968
FKBP14	65.4	100%	100%	Ehlers-Danlos syndrome with progressive kyphoscoliosis myopathy and hearing loss,614557
FLCN	142.8	100%	98%	Birt-Hogg-Dube syndrome,135150 Colorectal cancer,somatic,114500 Pneumothorax,primary spontaneous,173600 Renal carcinoma,chromphobe,somatic,144700
FLG	212.1	100%	99%	Ichthyosis vulgaris,146700 {Dermatitis,atopic,susceptibility to,2},605803
FLG2	756.5	100%	100%	No OMIM phenotype Atopic dermatitis (Margolis (2014) J Invest Dermatol 134,2272)
FLT4	148	99%	95%	Hemangioma,capillary infantile,somatic,602089 Lymphedema,hereditary,IA,153100
FNIP1	150.7	99%	95%	Familial multiple discoid fibromas,190340
FOXC2	42.2	92%	74%	Lymphedema-distichiasis syndrome with/without renal disease and diabetes mellitus,153400
FOXE1	27.1	71%	57%	Bamforth-Lazarus syndrome,241850

FOXN1	90.7	100%	99%	T-cell immunodeficiency congenital alopecia and nail dystrophy,601705
FOXP3	82.4	83%	74%	Immunodysregulation,polyendocrinopathy and enteropathy,X-linked,304790 {Diabetes mellitus,type I,susceptibility to},222100
FREM1	134.9	99%	98%	Bifid nose with or without anorectal and renal anomalies,608980 Manitoba oculotrichoanal syndrome,248450 Trigonocephaly 2,614485
FUCA1	120.8	99%	98%	Fucosidosis,230000
FZD6	181	100%	100%	Nail disorder,nonsyndromic,congenital 10 (claw-shaped nails),614157
GALNS	85.3	99%	93%	Mucopolysaccharidosis IVA,253000
GALNT3	125.7	97%	87%	Tumoral calcinosis,hyperphosphatemic,familial,211900
GAN	179.2	99%	98%	Giant axonal neuropathy-1,256850
GATA2	117.2	100%	98%	Emberger syndrome,614038 Immunodeficiency 21,614172 {Leukemia, acute myeloid, susceptibility to},601626 {Myelodysplastic syndrome, susceptibility to},614286
GDF2	139.3	100%	99%	Telangiectasia, hereditary hemorrhagic, type 5,615506
GDF5	106.2	100%	100%	Acromesomelic dysplasia,Hunter-Thompson type,201250 Brachydactyly,type A1,C,615072 Brachydactyly,type A2,112600 Brachydactyly,type C,113100 Chondrodysplasia,Grebe type,200700 Du Pan syndrome,228900 Multiple synostoses syndrome 2,610017 Symphalangis
GGCX	104.8	99%	96%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency,610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GJA1	205.3	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 dysplas

GJB2	182.1	100%	100%	Bart-Pumphrey syndrome,149200 Deafness,autosomal dominant 3A,601544 Deafness,autosomal recessive 1A,220290 Hystrix-like ichtyosis-deafness syndrome,602540 Keratitis-ichtyosis-deafness syndrome,148210 Keratoderma,palmoplantar,with deafness,148350 Voh
GJB3	300.8	100%	100%	Deafness autosomal dominant 2B,612644 Deafness,digenic,GJB2/GJB3,220290 Erythrokeratodermia variabilis et progressiva,133200
GJB4	363.8	100%	100%	Erythrokeratodermia variabilis with erythema gyratum repens,133200
GJB6	191.1	100%	100%	Deafness,autosomal dominant 3B,612643 Deafness,autosomal recessive 1B,612645 Deafness,digenic GJB2/GJB6,220290 Ectodermal dysplasia 2,Clouston type,129500
GJC2	34.3	81%	62%	Leukodystrophy,hypomyelinating 2,608804 Lymphedema, hereditary,IC,613480 Spastic paraplegia 44,autosomal recessive,613206
GLA	47.8	99%	90%	Fabry disease,301500
GLB1	73.9	98%	90%	GM1-gangliosidosis type I,230500 GM1-gangliosidosis type II,230600 GM1-gangliosidosis type III,230650 Mucopolysaccharidosis type IVB (Morquio),253010
GLMN	81.4	93%	79%	Glomuvenous malformations,138000
GMPPA	124.4	100%	100%	Alacrima, achalasia, and mental retardation syndrome ,615510
GNA11	134.5	100%	100%	Hypocalcemia,autosomal dominant 2,615361 Hypocalciuric hypercalcemia,type II,145981
GNAQ	88.5	82%	58%	Capillary malformations,congenital,1, somatic,mosaic,163000 Sturge-Weber syndrome, somatic, mosaic,185300
GNAS	110.5	95%	94%	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

				Pseudohy
GORAB	166.6	100%	99%	Geroderma osteodysplasticum,231070
GPR143	39.9	90%	67%	Nystagmus 6,congenital,X-linked,300814 Ocular albinism, type I, Nettleship-Falls type,300500
GRHL2	127.4	100%	100%	Deafness,autosomal dominant 28,608641 Ectodermal dysplasia/short stature syndrome,616029
GRHL3	127.8	100%	100%	Van der Woude syndrome 2, 606713
GSN	109	91%	87%	Amyloidosis Finnish type,105120
GTF2H5	160.4	100%	99%	Trichothiodystrophy 3,photosensitive,616395
HCCS	63.8	99%	92%	Linear skin defects with multiple congenital anomalies,309801
HDAC8	90.5	100%	100%	Cornelia de Lange syndrome 5,300882 Wilson-Turner syndrome,309585
HERC2	98.8	77%	72%	Mental retardation, autosomal recessive 38,615516 [Skin/hair/eye pigmentation 1],227220
HLCS	160.7	100%	100%	Holocarboxylase synthetase deficiency,253270
HMBS	90	100%	96%	Porphyria acute intermittent,176000
HMGB3	21.4	66%	54%	?Microphthalmia, syndromic 13,300915
HOXC13	87.2	92%	87%	Ectodermal dysplasia 9 hair/nail type,614931
HPS1	102.9	100%	98%	Hermansky-Pudlak syndrome 1,203300
HPS3	138	100%	95%	Hermansky-Pudlak syndrome 3,614072
HPS4	130.4	100%	98%	Hermansky-Pudlak syndrome 4,614073
HPS5	137.7	99%	98%	Hermansky-Pudlak syndrome 5,614074
HPS6	100.4	100%	92%	Hermansky-Pudlak syndrome 6,614075
HR	76.4	96%	91%	Alopecia universalis,203655 Atrichia with papular lesions,209500 Hypotrichosis 4,146550

HRAS	165.7	100%	98%	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
HTRA1	98.5	78%	75%	CARASIL syndrome,600142 {Macular degeneration,age-related,7},610149 {Macular degeneration,age-related,neovascular type},610149
HYAL1	101	100%	100%	Mucopolysaccharidosis type IX,601492
IDUA	94.1	89%	85%	Mucopolysaccharidosis Ih,607014 Mucopolysaccharidosis Ih/s,607015 Mucopolysaccharidosis Is,607016
IFT122	145.6	100%	99%	Cranioectodermal dysplasia 1,218330
IFT43	110.9	100%	100%	Cranioectodermal dysplasia 3,614099
IKBKG	25.6	68%	55%	Ectodermal dysplasia,hypohidrotic with immune deficiency,300291 Ectodermal dysplasia,anhydrotic,lymphedema and immunodeficiency,300301 Immunodeficiency 33,300636 Immunodeficiency,isolated,300584 Incontinentia pigmenti,308300 Invasive pneumococcal dis
IL17RA	115.9	96%	94%	?Candidiasis,familial 5,autosomal recessive,613953
IL17RD	119.3	99%	96%	Hypogonadotropic hypogonadism 18 with or without anosmia,615267
IL1RN	164.4	100%	100%	Interleukin 1 receptor antagonist deficiency,612852 {Gastric cancer risk after H.pylori infection},137215 {Microvascular complications of diabetes 4},612628
IL31RA	126.8	100%	99%	Amyloidosis,primary localized cutaneous 2,613955
IL36RN	84.2	100%	98%	Psoriasis 14, pustular,614204
INSR	123.7	97%	93%	Diabetes mellitus,insulin-resistant,with acanthosis nigricans,610549 Hyperinsulinemic hypoglycemia,familial,5,609968 Leprechaunism,246200 Rabson-Mendenhall syndrome,262190
IRF4	153.5	99%	97%	Multiple myeloma,254500 [Skin/hair/eye pigmentation, variation in,8],611724

IRF6	121.7	99%	93%	Orofacial cleft 6,608864 Popliteal pterygium syndrome 1,119500 van der Woude syndrome,119300
ITGA3	123.9	99%	96%	Interstitial lung disease, nephrotic syndrome and epidermolysis bullosa, congenital,614748
ITGA6	145.9	99%	98%	Epidermolysis bullosa,junctional, with pyloric stenosis,226730
ITGB4	132.4	96%	93%	Epidermolysis bullosa of hands and feet,131800 Epidermolysis bullosa,junctional,non-Herlitz type,226650 Epidermolysis bullosa,junctional,with pyloric atresia,226730
ITGB6	156.5	95%	91%	Amelogenesis imperfecta, type IH,616221
JUP	134.9	100%	99%	Arrhythmogenic right ventricular dysplasia 12,611528 Naxos disease,601214
KAL1	65.1	89%	85%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1),308700
KANK2	124	99%	99%	palmoplantar keratoderma and woolly hair,616099
KAT6B	167.3	98%	97%	Genitopatellar syndrome,606170 SBBYSS syndrome,603736
KCNH1	185.4	100%	99%	Temple-Baraitser syndrome,611816 Zimmermann-Laband syndrome,135500
KCNK9	164.3	100%	100%	Birk-Barel mental retardation dysmorphism syndrome,612292
KIF11	76.6	97%	95%	Microcephaly with or without chorioretinopathy lymphedema or mental retardation,152950
KIT	153.8	99%	98%	Gastrointestinal stromal tumor,familial,606764 Germ cell tumors,273300 Leukemia,acute myeloid,601626 Mast cell disease,154800 Piebaldism,172800
KITLG	76.3	91%	88%	Hyperpigmentation familial progressive 2,145250 [Skin/hair/eye pigmentation 7],611664
KLK4	180	100%	94%	Amelogenesis imperfecta type IIA1,204700
KLLN	81.4	100%	100%	Cowden syndrome 4,615107
KMT2D	133.1	99%	99%	Kabuki syndrome 1,147920

KRAS	57.8	100%	99%	Bladder cancer,somatic,109800 Breast cancer,somatic,114480 Cardiofaciocutaneous syndrome 2,615278 Gastric cancer,somatic,137215 Lung cancer,somatic,211980 Noonan syndrome 3,609942 Pancreatic carcinoma,somatic,260350 SFM syndrome,somatic mosaic,1632
KRT1	134.3	99%	91%	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 kerat
KRT10	90.8	94%	91%	Epidermolytic hyperkeratosis,113800 Ichthyosis with confetti,609165 Ichthyosis,cyclic,with epidermolytic hyperkeratosis,607602
KRT13	111.4	100%	94%	White sponge nevus 2,615785
KRT14	53.6	86%	78%	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,13
KRT16	28.8	71%	47%	Pachyonychia congenita 1,167200 Palmoplantar keratoderma,nonepidermolytic,focal,613000
KRT17	19.3	44%	34%	Pachyonychia congenita 2,167210 Steatocystoma multiplex,184500
KRT2	121.2	99%	97%	Ichthyosis bullosa of Siemens,146800
KRT4	108.2	100%	98%	White sponge nevus 1,193900
KRT5	109	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 Epiderm

KRT6A	164	90%	85%	Pachyonychia congenita 3,167200
KRT6B	169.4	90%	85%	Pachyonychia congenita Jackson-Lawler type,615726
KRT6C	158.9	89%	82%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse,615735
KRT71	119.5	100%	99%	Hypotrichosis 13,615896
KRT74	126.9	100%	100%	?Ectodermal dysplasia 7, hair/nail type,614929 ?Hypotrichosis 3,613981 Woolly hair, autosomal dominant,194300
KRT75	120.6	100%	100%	{Pseudofolliculitis barbae,susceptibility to},612318
KRT81	73.5	93%	88%	Monilethrix,158000
KRT83	70.6	98%	86%	Monilethrix,158000
KRT85	98.4	98%	92%	Ectodermal dysplasia 4 hair/nail type,602032
KRT86	86.6	100%	87%	Monilethrix,158000
KRT9	71.4	95%	93%	Epidermolytic palmoplantar keratoderma,144200
LAMA3	149.1	99%	99%	Epidermolysis bullosa,generalized atrophic benign,226650 Epidermolysis bullosa,junctional,Herlitz type,226700 Laryngoonychocutaneous syndrome,245660
LAMB3	107.8	100%	99%	Amelogenesis imperfecta,type IA,104530 Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
LAMC2	108.7	100%	97%	Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
LAMTOR2	139	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein,610798
LDHA	51.8	89%	81%	Glycogen storage disease XI,612933
LDLRAP1	136.5	97%	90%	Hypercholesterolemia,familial,autosomal recessive,603813
LEMD3	92	94%	86%	Buschke-Ollendorff syndrome,166700 Melorheostosis with osteopoikilosis,155950 Osteopoikilosis,166700
LIPH	116.4	100%	99%	Hypotrichosis 7,604379 Woolly hair,autosomal recessive 2,with or without hypotrichosis
LIPN	141.2	98%	96%	Ichthyosis,congenital,autosomal recessive 8,613943
LMBRD1	73.2	87%	82%	Methylmalonic aciduria and homocystinuria cblF type,277380

LMNA	72	95%	87%	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 Li
LMX1B	108.5	100%	88%	Nail-patella syndrome,161200
LONP1	134.4	95%	89%	CODAS syndrome,600373
LOR	16.1	82%	32%	Vohwinkel syndrome with ichthyosis,604117
LPAR6	108.2	100%	96%	Hypotrichosis 8,278150 Woolly hair,autosomal recessive 1,with or without hypotrichosis,278150
LPIN2	119.1	100%	99%	Majeed syndrome,609628
LTBP3	99.2	98%	96%	Dental anomalies and short stature,601216
LTBP4	100.8	99%	93%	Cutis laxa autosomal recessive type IC,613177
LYST	129.6	97%	92%	Chediak-Higashi syndrome,214500
LYZ	171.9	100%	100%	Amyloidosis,renal,105200
MAP2K1	89.5	100%	98%	Cardiofaciocutaneous syndrome 3,615279
MAP2K2	103.9	94%	90%	Cardiofaciocutaneous syndrome 4,615280
MBTPS2	84.9	99%	91%	?Olmsted syndrome,X-linked,300918 IFAP syndrome with or without BRESHECK syndrome,308205 Keratosis follicularis spinulosa decalvans,X-linked,308800
MED12	64.3	95%	89%	Lujan-Fryns syndrome,309520 Ohdo syndrome,X-linked,300895 Opitz-Kaveggia syndrome,305450
MEFV	105.6	94%	89%	Familial Mediterranean fever AD,134610 Familial Mediterranean fever AR,249100
MGP	132.8	91%	91%	Keutel syndrome,245150
MITF	134.3	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	162.3	100%	99%	Colorectal cancer,hereditary,nonpolyposis type 2,609310 Mismatch repair cancer syndrome,276300

				Muir-Torre syndrome,158320
MLPH	83.1	97%	94%	Griscelli syndrome type 3,609227
MMACHC	173.1	100%	100%	Methylmalonic aciduria and homocystinuria cblC type,277400
MMP2	148.6	100%	100%	Torg-Winchester syndrome,259600
MMP20	101.1	99%	97%	Amelogenesis imperfecta type IIA2,612529
MPLKIP	76.3	88%	52%	Trichothiodystrophy nonphotosensitive 1,234050
MRE11A	53.1	95%	84%	Ataxia-telangiectasia-like disorder,604391
MSH2	109.1	96%	88%	Colorectal cancer,hereditary,nonpolyposis type 1,120435 Mismatch repair cancer syndrome,276300 Muir-Torre syndrome,158320
MSX1	75.7	94%	87%	Ectodermal dysplasia 3,Witkop type,189500 Orofacial cleft 5,608874 Tooth agenesis,selective,1,with or without orofacial cleft,106600
MTOR	128.5	100%	99%	Smith-Kingsmore syndrome,616638
MUTYH	135	100%	99%	Adenomas,multiple colorectal,608456 Colorectal denomatous polyposis,autosomal recessive,with pilomatricomas,132600 Gastric cancer,somatic,613659
MVD	100.6	100%	99%	Porokeratosis 7,multiple types,614714
MVK	126.3	100%	100%	Hyper-IgD syndrome,260920 Mevalonic aciduria,610377 Porokeratosis 3,disseminated superficial actinic,175900
MYH8	128.8	100%	99%	Carney complex variant,608837 Trismus-pseudocamptodactyly syndrome,158300
MYO5A	122.5	98%	95%	Griscelli syndrome type 1,214450
NAA10	58	100%	91%	N-terminal acetyltransferase deficiency,300855 ?Microphthalmia,syndromic 1,309800
NAGA	147.9	100%	100%	Kanzaki disease,609242 Schindler disease,609241
NBAS	134.1	99%	97%	Short stature,optic nerve atrophy and Pelger-Huet anomaly,614800 Infantile liver failure syndrome 2,616483
NCSTN	111.4	100%	99%	Acne inversa familial 1,142690
NDUFB11	57	92%	69%	Linear skin defects with multiple congenital anomalies 3,300952

NF1	122.7	91%	86%	Neurofibromatosis, type 1,162200
NFKBIA	87.2	96%	89%	Ectodermal dysplasia anhidrotic with T-cell immunodeficiency,612132
NHP2	66.3	100%	99%	Dyskeratosis congenita, autosomal recessive 2,613987
NIPAL4	126.7	100%	93%	Ichthyosis,congenital,autosomal recessive 6,612281
NIPBL	111.4	96%	93%	Cornelia de Lange syndrome 1,122470
NLRP1	111.2	98%	95%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia,615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP3	130.4	100%	100%	CINCA syndrome,607115 Familial cold-induced inflammatory syndrome 1,120100 Muckle-Wells syndrome,191900
NME1	93.2	100%	98%	Neuroblastoma,256700
NOD2	118.2	100%	99%	Blau syndrome,186580 Sarcoidosis,early-onset,609464 {Inflammatory bowel disease 1},266600 {Psoriatic arthritis,susceptibility to},607507
NOP10	159.4	100%	100%	Dyskeratosis congenita, autosomal recessive 1,224230
NOTCH1	121.3	99%	97%	Aortic valve disease,109730 Adams-Oliver syndrome 5,616028
NRAS	179.8	100%	100%	Autoimmune lymphoproliferative syndrome type IV,614470 Colorectal cancer,somatic,114500 Epidermal nevus,somatic,162900 Melanocytic nevus syndrome,congenital,somatic,137550 Neurocutaneous melanosis,somatic,249400 Noonan syndrome 6,613224 Schimmelpenn
NSD1	142.2	100%	100%	Beckwith-Wiedemann syndrome,130650 Leukemia,acute myeloid,601626 Sotos syndrome,117550
NSDHL	119.2	99%	93%	CHILD syndrome,308050 CK syndrome,300831
OCA2	126.1	97%	95%	Albinism brown oculocutaneous,203200 [Skin/hair/eye pigmentation 1],227220
ODAM	119.2	94%	89%	No OMIM phenotype

OFD1	29.6	74%	53%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Orofaciodigital syndrome 1,311200 Simpson-Golabi-Behmel syndrome, type 2,300209
OSMR	135	99%	99%	Amyloidosis primary localized cutaneous 1,105250
PAH	170.6	100%	100%	Phenylketonuria,261600
PALB2	148.7	100%	100%	Fanconi anemia complementation group N,610832 {Breast cancer,susceptibility to},114480 {Pancreatic cancer,susceptibility to 3},613348
PAX3	108.8	100%	99%	Craniofacial-deafness-hand syndrome,122880 Rhabdomyosarcoma 2,alveolar,268220 Waardenburg syndrome,type 1,193500 Waardenburg syndrome,type 3,148820
PAX9	228.6	99%	93%	Tooth agenesis selective 3,604625
PCNA	88.2	100%	100%	Ataxia-telangiectasia-like disorder 2,615919
PDGFB	87	100%	100%	Dermatofibrosarcoma protuberans,607907 Basal ganglia calcification,idiopathic,5,615483 Meningioma, SIS-related,607174
PDGFRB	135.4	98%	96%	Basal ganglia calcification idiopathic 4,615007 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550
PEPD	99.8	100%	97%	Prolidase deficiency,170100
PEX7	121.4	91%	82%	Peroxisome biogenesis disorder 9B,614879 Chondrodysplasia punctata, rhizomelic, type 1,215100
PHEX	78.8	95%	93%	Hypophosphatemic rickets X-linked dominant,307800
PHGDH	119.4	100%	99%	Phosphoglycerate dehydrogenase deficiency,601815 Neu-Laxova syndrome 1,256520
PHYH	87.5	97%	92%	Refsum disease,266500
PIEZO1	125.8	98%	96%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema ,194380
PIGA	56.5	76%	73%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria,somatic,300818
PIGN	116.9	96%	90%	Multiple congenital anomalies-hypotonia-seizures syndrome 1,614080
PIGV	149.2	100%	100%	Hyperphosphatasia with mental retardation syndrome 1,239300

PIK3CA	125.3	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-capillar
PITX2	128.1	96%	93%	Axenfeld-Rieger syndrome type 1,180500 Iridogoniodysgenesis,type 2,137600 Peters anomaly,604229 Ring dermoid of cornea,180550
PKP1	108.1	100%	99%	Ectodermal dysplasia/skin fragility syndrome,604536
PLCD1	101.4	98%	94%	Nail disorder nonsyndromic congenital 3 (leukonychia),151600
PLCG2	116.1	99%	98%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome,614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	87.5	98%	95%	?Epidermolysis bullosa simplex with nail dystrophy,616487 Epidermolysis bullosa simples with muscular dystrophy,226670 Epidermolysis bullosa simplex with pyloric atresia,612138 Epidermolysis bullosa simplex, Ogna type, 131950 Muscular dystrophy,limb-g
PLG	114	87%	87%	Dysplasminogenemia,217090
PLIN1	74.5	92%	81%	Lipodystrophy familial partial type 4,613877
PLOD1	128.3	100%	98%	Ehlers-Danlos syndrome type VI,225400
PMS2	79.5	82%	79%	Colorectal cancer hereditary nonpolyposis type 4,614337 Mismatch repair cancer syndrome,276300
PMVK	108.7	100%	100%	Porokeratosis 1,multiple types,175800
PNPLA1	163.2	99%	97%	Ichthyosis congenital autosomal recessive 10,615024
PNPLA2	104.4	100%	96%	Neutral lipid storage disease with myopathy,610717
POC1A	132.6	100%	100%	Short stature onychodysplasia facial dysmorphism and hypotrichosis,614813
POFUT1	130.1	98%	92%	Dowling-Degos disease 2,615327
POGLUT1	124.6	100%	98%	Dowling-Degos disease 4,615696

POLD1	89	94%	92%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome,615381 {Colorectal cancer,susceptibility to,10},612591
POLH	149	100%	99%	Xeroderma pigmentosum variant type,278750
POLR1C	107.8	97%	94%	Treacher Collins syndrome 3,248390 Leukodystrophy, hypomyelinating,11,616494
POLR1D	171.4	100%	100%	Treacher Collins syndrome 2,613717
POLR3A	140.2	100%	99%	Leukodystrophy hypomyelinating 7 with or without oligodontia and/or hypogonadotropic hypogonadism,607694
POLR3B	143.9	100%	99%	Leukodystrophy hypomyelinating 8 with or without oligodontia and/or hypogonadotropic hypogonadism,614381
POMC	69.2	100%	99%	Obesity adrenal insufficiency and red hair due to POMC deficiency,609734 {Obesity,early-onset,susceptibility to},601665
POMP	138.6	86%	85%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma,601952
PORCN	79.8	100%	96%	Focal dermal hypoplasia,305600
PPOX	92.1	100%	97%	Porphyria variegata,176200
PQBP1	97	97%	94%	Renpenning syndrome,309500
PRKAR1A	90.8	98%	90%	Acrodysostosis 1 with or without hormone resistance,101800 Carney complex,type 1,160980 Myxoma,intracardiac,255960 Pigmented nodular adrenocortical disease,primary,1,610489
PSEN1	140.9	98%	94%	Acne inversa familial 3,613737 Alzheimer disease,type 3,607822 Cardiomyopathy,dilated,1U,613694 Dementia,frontotemporal,600274 Pick disease,172700
PSENEN	83.2	100%	99%	Acne inversa familial 2,613736
PSMB8	13.2	50%	16%	Autoinflammation lipodystrophy and dermatosis syndrome,256040
PSTPIP1	76.9	96%	86%	Pyogenic sterile arthritis pyoderma gangrenosum and acne,604416
PTCH1	116.3	98%	96%	Basal cell carcinoma somatic,605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7,610828

PTCH2	108.1	99%	97%	Basal cell carcinoma somatic,605462 Basal cell nevus syndrome,109400 Medulloblastoma,155255
PTDSS1	130.4	100%	99%	Lenz-Majewski hyperostotic dwarfism,151050
PTEN	142.2	100%	100%	Bannayan-Riley-Ruvalcaba syndrome,153480 Cowden syndrome 1,158350 Endometrial carcinoma,somatic,608089 Macrocephaly/autism syndrome,605309 Malignant melanoma,somatic,155600 Squamous cell carcinoma,head and neck,somatic,275355 Thyroid carcinoma,folli
PTHLH	126.8	99%	83%	Brachydactyly type E2,613382 Humoral hypercalcemia of malignancy
PTPN11	93	96%	89%	LEOPARD syndrome 1,151100 Leukemia,juvenile myelomonocytic,607785 Metachondromatosis,156250 Noonan syndrome 1,163950
PTPN14	161.1	99%	97%	Choanal atresia and lymphedema,613611
PTPRF	169	100%	100%	Breasts and/or nipples, aplasia or hypoplasia of, 2,616001
PTRF	115.2	100%	97%	Lipodystrophy congenital generalized type 4,613327
PVRL1	134.6	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome,225060 Orofacial cleft 7,225060
PVRL4	120	100%	100%	Ectodermal dysplasia-syndactyly syndrome 1,613573
PYCR1	80.4	100%	91%	Cutis laxa autosomal recessive type IIB,612940 Cutis laxa autosomal recessive type IIIB,614438
RAB23	103.9	100%	99%	Carpenter syndrome,201000
RAB27A	177.8	100%	100%	Griscelli syndrome type 2,607624
RAD21	89.1	99%	96%	Cornelia de Lange syndrome 4,614701
RAD50	91.4	93%	86%	Nijmegen breakage syndrome-like disorder,613078
RAF1	118.6	100%	99%	LEOPARD syndrome 2,611554 Cardiomyopathy,dilated,1NN,615916 Noonan syndrome 5,611553

RAG1	203.2	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 ce
RAG2	250.1	100%	99%	Combined cellular and humoral immune defects with granulomas,233650 Omenn syndrome,603554 Severe combined immunodeficiency,B cell-negative,601457
RAI1	113.3	100%	99%	Smith-Magenis syndrome,182290
RBBP8	93.1	100%	95%	Jawad syndrome,251255 Pancreatic carcinoma,somatic Seckel syndrome 2,606744
RBM28	128.8	100%	100%	Alopecia neurologic defects and endocrinopathy syndrome,612079
RBP4	87	93%	91%	Retinol dystrophy iris coloboma and comedogenic acne syndrome,615147 Microphthalmia,isolated,with coloboma 10,616428
RBPJ	75.6	91%	83%	Adams-Oliver syndrome 3,614814
RECQL4	121.3	97%	95%	Baller-Gerold syndrome,218600 RAPADILINO syndrome,266280 Rothmund-Thomson syndrome,268400
RHBDF2	95.5	98%	95%	Tylosis with esophageal cancer,148500
RIN2	110.7	99%	98%	Macrocephaly alopecia cutis laxa and scoliosis,613075
RIPK4	127.7	100%	98%	Popliteal pterygium syndrome 2 lethal type,263650
RMRP				Anauxetic dysplasia,607095 Cartilage-hair hypoplasia,250250 Metaphyseal dysplasia without hypotrichosis,250460
RNASEH2A	124.1	100%	95%	Aicardi-Goutieres syndrome 4,610333
RNASEH2B	106.7	88%	76%	Aicardi-Goutieres syndrome 2,610181
RNASEH2C	175.9	100%	99%	Aicardi-Goutieres syndrome 3,610329
RNU4ATAC				Microcephalic osteodysplastic primordial dwarfism,type I,210710 Roifman syndrome,616651
ROGDI	110.8	96%	94%	Kohlschutter-Tonz syndrome,226750
RPL21	69.2	62%	50%	Hypotrichosis 12,615885
RSPO1	95.6	100%	97%	Palmoplantar hyperkeratosis and true hermaphroditism,610644 Palmoplantar hyperkeratosis with squamous cell skin carcinoma and sex revearsal,610644

RSPO4	92.8	100%	96%	Anonychia congenita,206800
RTEL1	100	98%	92%	Dyskeratosis congenita,autosomal recessive 5,615190 Dyskeratosis congenita,autosomal dominant 4,615190 Pulmonary fibrosis and/or bone marrow failure,telomere-related,616373
RUNX2	94.4	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/without brachydactyly,156510
SAMD9	169.1	100%	96%	Tumoral calcinosis familial normophosphatemic,610455
SAMHD1	124.9	97%	94%	Aicardi-Goutieres syndrome 5,612952 Chilblain lupus 2,614415
SART3	122.1	100%	97%	No OMIM phenotype Disseminated superficial actinic porokeratosis (Zhang (2005) Br J Dermatol 152,658)
SAT1	77.6	93%	92%	Keratosis follicularis spinulosa decalvans,308800
SATB2	98	98%	91%	Cleft palate and mental retardation,119540
SCN10A	164	99%	98%	Epilepsy generalized with febrile seizures plus type 7,613863
SCN11A	131.8	98%	97%	Episodic pain syndrome,familial,3,615552 Neuropathy,hereditary sensory and autonomic, type VII,615548
SCN9A	146.4	96%	94%	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 fi
SEC23B	157.3	97%	96%	Anemia dyserythropoietic congenital type II,224100
SERPINB7	130.6	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598 ,615598
SERPINB8	149.5	100%	100%	No OMIM phenotype
SERPINH1	145.9	100%	100%	Osteogenesis imperfecta type X,613848 {Preterm premature rupture of the membranes, susceptibility to},610504
SHOC2	124.9	100%	99%	Noonan-like syndrome with loose anagen hair,607721
SKI	68.1	96%	95%	Shprintzen-Goldberg syndrome,182212
SKIV2L	21.4	75%	47%	Trichohepatoenteric syndrome 2,614602

SLC17A9	106.2	95%	95%	Porokeratosis, disseminated superficial actinic, 8,616063
SLC24A4	125.6	99%	97%	Ameliogenesis imperfecta, hypomaturation type, IIA5,615887 [Skin/hair/eye pigmentation 6],210750
SLC24A5	119.9	95%	93%	Albinism, oculocutaneous, type VI,113750 [skin/hair/eye pigmentation 4],113750
SLC26A2	223	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	128	100%	97%	Ichthyosis prematurity syndrome,608649
SLC29A3	190.4	99%	99%	Histiocytosis-lymphadenopathy plus syndrome,602782
SLC2A10	138.4	100%	99%	Arterial tortuosity syndrome,208050
SLC39A13	97	100%	94%	Spondylocheirodysplasia Ehlers-Danlos syndrome-like,612350
SLC39A4	67.6	99%	94%	Acrodermatitis enteropathica,201100
SLC45A2	128.1	100%	98%	Oculocutaneous albinism type IV,606574 [skin/hair/eye pigmentation 5],227240
SLC4A4	132.4	100%	99%	Renal tubular acidosis proximal with ocular abnormalities,604278
SLC6A19	147.7	98%	95%	Hartnup disorder,234500 Hyperglycinuria,138500 Iminoglycinuria,digenic,242600
SLC7A7	106.3	100%	100%	Lysinuric protein intolerance,222700
SLCO2A1	94.9	100%	97%	Hypertrophic osteoarthropathy primary autosomal recessive 2,614441
SLURP1	89.6	99%	89%	Meleda disease,248300
SLX4	101.3	100%	99%	Fanconi anemia complementation group P,613951
SMAD3	112.5	99%	97%	Loeys-Dietz syndrome type 3,613795
SMARCA2	109.5	96%	94%	Nicolaides-Baraitser syndrome,601358
SMARCA4	131.9	99%	94%	Mental retardation autosomal dominant 16,614609 {Rhabdoid tumor predisposition syndrome 2},613325
SMARCAD1	83	99%	94%	Adermatoglyphia,136000
SMARCAL1	121.1	100%	98%	Schimke immunoosseous dysplasia,242900
SMARCB1	202.6	100%	100%	Mental retardation autosomal dominant 15,614608 Rhabdoid tumors, somatic,609322

				{Schwannomatosis-1,susceptibility to},162091
SMO	147.3	96%	89%	Basal cell carcinoma, somatic
SMOC2	98.8	97%	92%	Dentin dysplasia type I with microdontia and misshapen teeth,125400
SNAI2	125.1	100%	98%	Piebaldism,172800 Waardenburg syndrome, type 2D,608890
SNAP29	134.3	100%	100%	Cerebral dysgenesis neuropathy, ichtyosis, and palmoplantar keratoderma syndrome,609528
SNRPE	92.7	99%	99%	Hypotrichosis 11,615059
SNX10	95.8	100%	98%	Osteopetrosis autosomal recessive 8,615085
SOS1	87.5	93%	88%	Fibromatosis,gingival,135300 Noonan syndrome 4,610733
SOX10	65.7	92%	88%	PCWH syndrome,609136 Waardenburg syndrome,type 2E,with/without neurological involvement,611584 Waardenburg syndrome,type 4C,613266
SOX18	13.9	56%	44%	Hypotrichosis-lymphedema-telangiectasia syndrome,607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome,137940
SOX2	80.8	99%	96%	Microphthalmia syndromic 3,206900 Optic nerve hypoplasia and abnormalities of the central nervous system,206900
SP7	159.5	100%	100%	Osteogenesis imperfecta type XII,613849
SPINK5	154.4	100%	95%	Atopy,147050 Netherton syndrome,256500
SPINT2	54.1	94%	77%	Diarrhea 3 secretory sodium congenital syndromic,270420
SPRED1	165.6	96%	96%	Legius syndrome,611431
SPRY4	128.5	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia,615266
SRD5A3	140.5	99%	96%	Congenital disorder of glycosylation type Iq,612379 Kahrizi syndrome,612713
ST14	151.4	100%	98%	Ichthyosis with hypotrichosis,610765
ST3GAL5	118.3	94%	93%	Ganglioside GM3 synthase deficiency,609056
STAMBP	118	100%	96%	Microcephaly-capillary malformation syndrome,614261
STAT3	122.2	100%	98%	Hyper-IgE recurrent infection syndrome,147060 Autoimmune disease,multisystem,infantile-onset,615952
STAT5B	116.6	100%	95%	Growth hormone insensitivity with immunodeficiency,245590

				Leukemia,acute promyelocytic,STAT5B/RARA type
STIM1	125.1	99%	95%	Immune dysfunction with T-cell inactivation due to calcium entry defect 2,612783 Myopathy,tubular aggregate,1,160565 Stormorken syndrome,185070
STK11	112.3	100%	97%	Melanoma malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STS	61.2	97%	92%	Ichthyosis X-linked,308100
SUFU	125.3	97%	96%	Medulloblastoma desmoplastic,155255 Basal cell nevus syndrome,109400 {Meningioma,familial,susceptibility to},607174
SUMF1	122.6	91%	86%	Multiple sulfatase deficiency,272200
TALDO1	127.8	100%	100%	Transaldolase deficiency,606003
TAT	117.2	100%	100%	Tyrosinemia type II,276600
TBC1D24	149.2	100%	100%	Epileptic encephalopathy early infantile 16,615338 Deafness, autosomal recessive 86,614617 Deafness, autosomal dominant 65,616044 DOOR syndrome,220500 Myoclonic epilepsy,infantile,familial,605021
TBX3	74.3	99%	94%	Ulnar-mammary syndrome,181450
TCIRG1	107.1	95%	84%	Osteopetrosis autosomal recessive 1,259700
TEK	193	100%	99%	Venous malformations multiple cutaneous and mucosal,600195
TERC				Dyskeratosis congenita,autosomal dominant 1,127550 {Aplastic anemia},614743 {Pulmonary fibrosis,idiopathic,susceptibility to},614743
TERT	119	98%	89%	{Dyskeratosis congenita, autosomal recessive 4},613989 {Dyskeratosis congenita, autosomal dominant 2}, 613979 {Leukemia, acute myeloid},601626 {Melanoma, cutaneous malignant, 9},615134 {Pulmonary fibrosis and/or bone marrow failure, telomere related,
TFAP2A	109.5	100%	98%	Branchiooculofacial syndrome,113620
TGFB2	153	100%	100%	Loeys-Dietz syndrome type 4,614816

TGFBR1	196.2	93%	93%	Loeys-Dietz syndrome type 1A,609192 {Multiple self-healing squamous epithelioma,susceptibility to},132800
TGFBR2	184.5	100%	100%	Colorectal cancer,hereditary,nonpolyposis type 6,614331 Esophageal cancer,somatic,133239 Loeys-Dietz syndrome,type 2,610168
TGM1	157.1	100%	99%	Ichthyosis congenital autosomal recessive 1,242300
TGM5	161.4	100%	99%	Peeling skin syndrome acral type,609796
TINF2	156.3	100%	100%	Dyskeratosis congenita autosomal dominant 3,613990 Revesz syndrome,268130
TMC6	66	97%	92%	Epidermodysplasia verruciformis,226400
TMC8	102.2	96%	88%	Epidermodysplasia verruciformis,226400
TMEM165	107.3	99%	96%	Congenital disorder of glycosylation type IIk,614727
TMEM173	87.1	99%	96%	STING-associated vasculopathy, infantile-onsent (SAVI),615934
TNFRSF11A	127.3	92%	89%	Osteolysis,familial expansile,174810 Osteopetrosis,autosomal recessive 7,612301 {Paget disease of bone 2,early-onset},602080
TNFRSF11B	240.7	100%	100%	Paget disease of bone 5, juvenile-onset,239000
TNFRSF1A	80.4	91%	88%	Periodic fever,familial,142680 {Multiple sclerosis, susceptibility to,5},614810
TNFSF11	172.9	100%	90%	Osteopetrosis,autosomal recessive 2,259710
TNXB	13.3	48%	20%	Ehlers-Danlos syndrome,autosomal dominant,hypermobility type,130020 Vesicoureteral reflux 8,615963
TP63	186.5	100%	100%	ADULT syndrome,103285 Ectrodactyly,ectodermal dysplasia,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	148.4	96%	92%	Skin/hair/eye pigmentation, variation in, 10,612267
TREX1	214.3	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	134.2	100%	100%	?Bardet-Biedl syndrome 11,615988 Muscular dystrophy,limb-girdle,type 2H,254110
TRIM37	112.2	99%	97%	Mulibrey nanism,253250
TRPV3	157.8	100%	99%	?Palmoplantar keratoderma,nonepidermolytic,focal 2,616400 Olmsted syndrome,614594
TSC1	127.2	99%	97%	Focal cortical dysplasia,Taylor balloon cell type,607341 Lymphangioleiomyomatosis,606690 Tuberous sclerosis-1,191100
TSC2	111.2	99%	98%	Lymphangioleiomyomatosis,somatic,606690 Tuberous sclerosis-2,613254
TTC37	127.9	99%	97%	Trichohepatoenteric syndrome 1,222470
TTI2	92.8	99%	97%	Mental retardation, autosomal recessive 39,615541
TWIST2	122.6	99%	91%	Ablepharon-macrostomia syndrome,200110 Barber-Say syndrome,209885 Focal facial dermal dysplasia 3,Setleis type,227260
TYR	189.7	100%	100%	Albinism,oculocutaneous,type IA,203100 Albinism,oculocutaneous,type IB,606952 Waardenburg syndrome/albinism,digenic,103470 [Skin/hair/eye pigmentation 3],601800
TYRP1	190.1	100%	100%	Albinism oculocutaneous type III,203290 [Skin/hair/eye pigmentation,variation in,11(Melanesian blond hair),612271
UBE2A	62.6	96%	91%	Mental retardation,X-linked syndromic,Nascimento-type,300860
UBR1	130.1	99%	95%	Johanson-Blizzard syndrome,243800
UROD	143.9	100%	99%	Porphyria,cutanea tarda,176100 Porphyria,hepatoerythropoietic,176100
UROS	97.3	100%	100%	Porphyria,congenital erythropoietic,263700
USB1	120.7	100%	98%	Poikiloderma with neutropenia,604173
UVSSA	106.4	100%	98%	UV-sensitive syndrome 3,614640
VDR	114.9	97%	94%	Rickets,vitamin D-resistant,type IIA,277440 ?Osteoporosis,involutional,166710
VEGFC	154.9	100%	97%	Lymphedema, hereditary, ID,615907

VHL	91.1	98%	75%	Erythrocytosis,familial,2,263400 Hemangioblastoma,cerebellar,somatic Pheochromocytoma,171300 Renal cell carcinoma,somatic,144700 von Hippel-Lindau syndrome,193300
VPS13B	140	98%	97%	Cohen syndrome,216550
WAS	35.9	81%	62%	Neutropenia,severe congenital,X-linked,300299 Thrombocytopenia,X-linked,313900 Wiskott-Aldrich syndrome,301000
WDR19	133.5	100%	98%	?Cranioectodermal dysplasia 4,614378 ?Short-rib thoracic dysplasia 5 with/without polydactyly,614376 Nephronophthisis 13,614377 Senior-Loken syndrome 8,616307
WDR35	143.9	97%	94%	Cranioectodermal dysplasia 2,613610 Short-rib thoracic dysplasia 7 with/without polydactyly,614091
WDR72	145.6	99%	98%	Amelogenesis imperfecta hypomaturation type IIA3,613211
WIPF1	69.4	100%	97%	Wiskott-Aldrich syndrome 2,614493
WNT10A	90.5	100%	96%	Odontoonychodermal dysplasia,257980 Schopf-Schulz-Passarge syndrome,224750 Tooth agenesis,selective,4,150400
WNT10B	106.1	100%	100%	Split-hand/foot malformation 6,225300
WNT5A	138.5	100%	100%	Robinow syndrome autosomal dominant,180700
WNT7A	198.8	100%	100%	Fuhrmann syndrome,228930 Ulna and fibula,absence of,with severe limb deficiency,276820
WRAP53	127.4	100%	100%	Dyskeratosis congenita autosomal recessive 3,613988
WRN	128.3	97%	94%	Werner syndrome,277700
XPA	46.6	95%	84%	Xeroderma pigmentosum group A,278700
XPC	148.7	100%	100%	Xeroderma pigmentosum group C,278720
XYLT1	124.1	87%	84%	Desbuquois dysplasia 2,615777 {Pseudoxanthoma elasticum,modifier of severity of},264800
XYLT2	130.2	98%	95%	Spondyloocular syndrome,605822 {Pseudoxanthoma elasticum,modifier of severity of},264800
ZBTB20	195.3	100%	100%	Primrose syndrome,259050
ZMPSTE24	125.2	100%	97%	Mandibuloacral dysplasia with type B lipodystrophy,608612

				Restrictive dermopathy,lethal,275210
ZNF469	62.1	98%	94%	Brittle cornea syndrome 1,229200
ZNF592	109.2	99%	98%	Spinocerebellar ataxia,autosomal recessive 5,606937
ZNF750	130.7	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 : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 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