SKIN DISORDERS GENE PANEL DGD09072015

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
AAAS	108.9	100%	100%	Achalasia-addisonianism-alacrimia syndrome,231550
AAGAB	133.3	100%	98%	Keratoderma palmoplantar punctate type IA,148600
ABCA12	115.7	100%	99%	Ichthyosis, autosomal recessive 4B (harlequin),242500 Ichthyosis, congenital, autosomal recessive 4A,601277
ABCB6	133.9	100%	100%	Dyschromatosis universalis hereditaria 3,615402 Microphthalmia,isolated, with coloboma 7,614497 [Blood group, Langereis system],111600
ABCC6	56.2	72%	68%	Arterial calcification generalized of infancy 2,614473 Pseudoxanthoma elasticum,264800 Pseudoxanthoma elasticum, forme fruste,177850
ABCC9	120.2	100%	97%	Atrial fibrillation familial 12,614050 Cardiomyopathy, dilated, 1O,608569 Hypertrichotic osteochondrodysplasia, 239850
ABHD5	129.1	100%	95%	Chanarin-Dorfman syndrome,275630
ACTA2	92.6	100%	100%	Aortic aneurysm familial thoracic 6,611788 Moyamoya disease 5,614042 Multisystemic smooth muscle dysfunction syndrome,613834
ACVRL1	56.7	98%	85%	Telangiectasia hereditary hemorrhagic type 2,600376
ADAM10	137.8	100%	100%	Reticulate acropigmentation of Kitamura,615537 {Alzheimer disease 18, susceptibility to},615590
ADAM17	129.1	100%	98%	Inflammatory skin and bowel disease neonatal,614328
ADAMTS10	77	98%	91%	Weill-Marchesani syndrome 1 recessive,277600
ADAMTS17	83.2	93%	79%	Weill-Marchesani-like syndrome,613195
ADAMTS2	108	94%	92%	Ehlers-Danlos syndrome type VIIC,225410
ADAR	148.1	99%	98%	Aicardi-Goutieres syndrome 6,615010 Dyschromatosis symmetrica hereditaria,127400
AGA	140.5	100%	97%	Aspartylglucosaminuria,208400

AGPAT2	58.6	99%	92%	Lipodystrophy congenital generalized type 1,608594
AIRE	82.8	99%	93%	Autoimmune polyendocrinopathy syndrome type I with or without reversible metaphyseal dysplasia,240300
AKT1	129	97%	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	117.4	100%	100%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome,603387
ALAD	92.5	99%	95%	Porphyria acute hepatic,612740 {Lead poisoning, susceptibility to},612740
ALAS2	96.7	94%	88%	Anemia sideroblastic X-linked,300751 Protoporphyria, erythropoietic, X-linked,300752
ALDH18A1	102.8	97%	92%	Cutis laxa autosomal recessive type IIIA,219150
ALDH3A2	105.3	100%	100%	Sjogren-Larsson syndrome,270200
ALDOB	126	100%	98%	Fructose intolerance,229600
ALOX12B	116.1	100%	100%	Ichthyosis congenital autosomal recessive 2,242100
ALOXE3	100.6	100%	100%	Ichthyosis congenital autosomal recessive 3,606545
ALPL	96.4	100%	100%	Hypophosphatasia, adult,146300 Hypophosphatasia, childhood,241510 Hypophosphatasia, infantile, 241500 Odontohypophosphatasia,146300
ALX4	77.7	100%	93%	Frontonasal dysplasia 2,613451 Parietal foramina 2,609597 {Craniosynostosis 5, susceptibility to},615529
AMELX	131.2	100%	100%	Amelogenesis imperfecta, type 1E,301200
ANKRD11	123.4	89%	87%	KBG syndrome,148050
ANTXR1	93.4	97%	92%	GAPO syndrome,230740 {Hemangioma, capillary infantile, susceptibility to},602089
ANTXR2	122.9	100%	97%	Hyaline fibromatosis syndrome,228600
AP3B1	124	100%	100%	Hermansky-Pudlak syndrome 2,608233

APC	161.2	100%	100%	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,somatic,114550
APCDD1	138.9	100%	100%	Hypotrichosis 1,605389
AQP5	103.2	100%	99%	Palmoplantar keratoderma, Bothnian type,600231
ARHGAP31	152.3	100%	99%	Adams-Oliver syndrome 1,100300
ARID1A	110.7	97%	94%	Mental retardation autosomal dominant 14,614607
ARID1B	116.1	99%	95%	Mental retardation,autosomal dominant 12,614562
ASIP	71.7	74%	71%	[Skin/hair/eye pigmentation 9],611742
ASL	88.9	97%	93%	Argininosuccinic aciduria,207900
ASXL1	169.6	98%	97%	Bohring-Opitz syndrome,605039 Myelodysplastic syndrome,somatic,614286
ASXL3	174.4	99%	99%	Bainbridge-Ropers syndrome ,615485
ATIC	118.4	100%	96%	AICA-ribosiduria due to ATIC deficiency,608688
ATP2A2	133.1	100%	100%	Acrokeratosis verruciformis,101900 Darier disease,124200
ATP2C1	128.2	100%	99%	Hailey-Hailey disease,169600
ATP6V0A2	115.9	100%	100%	Cutis laxa,autosomal recessive,type IIA,219200 Wrinkly skin syndrome,278250
ATP7A	143.9	100%	100%	Menkes disease,309400 Occipital horn syndrome,304150 Spinal muscular atrophy,distal,X-linked,300489
ATR	131.2	99%	99%	Cutaneous telangiectasia and cancer syndrome familial,614564 Seckel syndrome 1,210600
AXIN2	111.3	98%	90%	Colorectal cancer somatic,114500 Oligodontia-colorectal cancer syndrome,608615
B3GALT6	57.2	77%	75%	Ehlers-Danlos syndrome progeroid type 2,615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1,with or without fractures,271640
B4GALT7	89.3	100%	95%	Ehlers-Danlos syndrome progeroid type 1,130070

BANF1	50.2	55%	54%	Nestor-Guillermo progeria syndrome,614008
BAP1	102.6	100%	99%	Tumor predisposition syndrome,614327
BCOR	137.4	100%	99%	Microphthalmia syndromic 2,300166
BCS1L	144.5	100%	100%	Bjornstad syndrome,262000 GRACILE syndrome,603358 Leigh syndrome,256000 Mitochondrial complex III defiency, nuclear type 1,124000
BLM	131.8	99%	98%	Bloom syndrome,210900
BLOC1S3	16.6	63%	35%	Hermansky-Pudlak syndrome 8,614077
BLOC1S6	128.8	89%	82%	Hermansky-pudlak syndrome 9,614171
BMS1	52.7	37%	37%	Aplasia cutis congenita, nonsyndromic,107600
BRAF	82.1	100%	97%	Adenocarcinoma of lung,somatic,211980 Cardiofaciocutaneous syndrome,115150 LEOPARD syndrome 3,613707 Noonan syndrome 7,613706
BRIP1	141.4	100%	100%	Breast cancer early-onset,114480 Fanconi anemia,complementation group J,609054
BSCL2	117.1	100%	100%	Encephalopathy, progressive, with or without lipodystrophy,615924 Lipodystrophy congenital generalized type 2,269700 Neuropathy, distal hereditary motor, type VA,600794 Silver spastic paraplegia syndrome,270685
BTD	159.2	100%	100%	Biotinidase deficiency,253260
C10orf11	93.5	99%	99%	Albinism, oculocutaneous type VII,615179
C1QA	132.7	94%	89%	C1q deficiency,613652
C1QB	104.6	95%	88%	C1q deficiency,613652
C1QC	140.6	92%	70%	C1q deficiency,613652
C2CD3	122.7	95%	95%	?Orofaciodigital syndrome XIV,615948
C4orf26	156.6	100%	100%	Amelogenesis imperfecta, type IIA4,614832
CA2	164.4	100%	100%	Osteopetrosis,autosomal recessive 3,with renal tubular acidosis,259730
CARD14	66.8	96%	90%	Pityriasis rubra pilaris,173200 Psoriasis 2,602723
CARD9	62.8	100%	97%	Candidiasis,familial 2,autosomal recessive,212050
CAST	109.1	97%	97%	PLACK syndrome,616295

CAV1	139.7	100%	100%	?Lipodystrophy,congenital generalized,type 3,612526 ?Partial lipodystrophy, congenital cataracts and neurodegeneration syndrome,606721 Pulmonary hypertension, primary, 3,615343
CBL	142.8	100%	100%	Noonan syndrome-like disorder,with or without juvenile meylomonocytic leukemia,613563
CBS	81.4	95%	84%	Homocystinuria B6-responsive and nonresponsive types,236200 Thrombosis,hyperhomocysteinemic,236200
CCBE1	96.4	96%	89%	Hennekam lymphangiectasia-lymphedema syndrome,235510
CD151	96.8	100%	100%	Nephropathy with pretibial epidermolysis bullosa and deafness,609057 [Blood group, Raph],179620
CDAN1	95.8	100%	98%	Dyserythropoietic anemia, congenital, type Ia,224120
CDH3	101.4	99%	95%	Ectodermal dysplasia,ectrodactyly and macular dystrophy,225280 Hypotrichosis, congenital, with juvenile macular dystrophy,601553
CDK4	142.8	97%	90%	{Melanoma, cutaneous malignant, 3},609048
CDKN2A	93.3	93%	93%	Melanoma and neural system tumor syndrome,155755 Pancreatic cancer/melanoma syndrome,606719 {Melanoma,cutaneous malignant, 2},155601
CDSN	12.6	59%	19%	Hypotrichosis 2,146520 Peeling skin syndrome 1,270300
CECR1	103.9	97%	95%	?Sneddon syndrome,182410 Polyarteritis nodosa, childhood-onset,615688
CERS3	98.2	100%	99%	Ichthyosis, congenital, autosomal recessive 9,615023
CHKB	91.6	92%	89%	Muscular dystrophy congenital megaconial type,602541
CHST14	125.3	100%	96%	Ehlers-Danlos syndrome musculocontractural type,601776
CHSY1	167.9	95%	92%	Temtamy preaxial brachydactyly syndrome,605282
CHUK	107	100%	97%	Cocoon syndrome,613630
CKAP2L	157	100%	100%	Filippi syndrome,272440
CLDN1	116.1	100%	99%	Ichthyosis,leukocyte vacuoles,alopecia and sclerosing cholangitis,607626
CNNM4	179.8	98%	97%	Jalili syndrome,217080

COL17A1	94.3	99%	93%	Epidermolysis bullosa,junctional,non-Herlitz type,226650
COL1A2	105.1	98%	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},166710
COL3A1	72.5	97%	94%	Ehlers-Danlos syndrome,type III,130020 Ehlers-Danlos syndrome, type IV,130050
COL5A1	107.9	98%	97%	Ehlers-Danlos syndrome, classic type I,130000
COL5A2	94	98%	94%	Ehlers-Danlos syndrome, classic type I,130000
COL7A1	117.3	100%	99%	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 Transient bullous of the newborn,131705
COX4I2	58.4	97%	87%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis,612714
COX7B	72.7	99%	98%	Linear skin defects with multiple congenital anomalies,300887
CPOX	80.3	100%	95%	Coproporphyria,121300 Harderoporphyria,121300
CSTA	117.9	100%	100%	Exfoliative ichthyosis,autosomal recessive,ichthyosis bullosa of Siemens-like,607936
CTC1	114.8	100%	99%	Cerebroretinal microangiopathy with calcifications and cysts,612199
CTSA	110.3	100%	100%	Galactosialidosis,256540
CTSC	101.3	100%	98%	Haim-Munk syndrome,245010 Papillon-Lefevre syndrome,245000 Periodontitis 1, juvenile,170650
CXCR4	239.8	100%	100%	Myelokathexis, isolated WHIM syndrome, 193670
CYLD	129.8	100%	100%	Brooke-Spiegler syndrome,605041 Cylindromatosis,familial,132700 Trichoepithelioma,multiple familial,1,601606

CYP26C1	53.8	97%	81%	Focal facial dermal dysplasia 4,614974
CYP4F22	112.9	99%	97%	Ichthyosis,congenital,autosomal recessive 5,604777
DCAF17	109.1	100%	97%	Woodhouse-Sakati syndrome,241080
DCLRE1C	116.6	90%	90%	Omenn syndrome,603554 Severe combined immunodeficiency, Athabascan type,602450
DDB2	106	100%	98%	Xeroderma pigmentosum,group E,DDB-negative subtype,278740
DHCR7	127.4	100%	99%	Smith-Lemli-Opitz syndrome,270400
DKC1	107.1	100%	100%	Dyskeratosis congenita X-linked,305000
DLX3	71.3	100%	96%	Amelogenesis imperfecta,type IV,104510 Trichodontoosseous syndrome,190320
DLX5	112.1	91%	87%	?Split-hand/foot malformation 1 with sensorineural hearing loss,220600
DOCK6	94.8	99%	96%	Adams-Oliver syndrome 2,614219
DOCK8	97.6	100%	98%	Hyper-IgE recurrent infection syndrome autosomal recessive,243700
DOLK	169	100%	100%	Congenital disorder of glycosylation, type Im,610768
DSC2	111.4	100%	99%	Arrhythmogenic right ventricular dysplasia 11Without/with mild palmoplantar keratoderma and woolly hair,610476
DSC3	110.5	99%	98%	?Hypotrichosis and recurrent skin vesicles,613102
DSE	112.4	95%	84%	?Ehlers-Danlos syndrome, musculocontractural type 2,615539
DSG1	164.4	100%	100%	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis and hyper IgE,615508 Keratosis palmoplantaris striata I,AD,148700
DSG3	165.9	100%	99%	No OMIM phenotype
DSG4	133	100%	99%	Hypotrichosis 6,607903
DSP	147.5	100%	98%	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	178.3	98%	96%	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	122.4	100%	100%	Hermansky-Pudlak syndrome 7,614076 {Schizophrenia},181500
DUSP6	162.6	100%	98%	Hypogonadotropic hypogonadism 19 with or without anosmia,615269
EBP	92.2	99%	98%	Chondrodysplasia punctata X-linked dominant,302960
ECM1	138.5	100%	99%	Urbach-Wiethe disease,247100
EDA	92.9	99%	98%	Ectodermal dysplasia 1,hypohidrotic,X-linked,305100 Tooth agenesis,selective,X-linked 1,313500
EDAR	89.6	100%	100%	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	134.3	99%	95%	Ectodermal dysplasia 11A,hypohidrotic/hair/tooth type, autosomal dominant,614940 Ectodermal dysplasia 11B,hypohidrotic/hair/tooth type, autosomal recessive,614941
EDN3	110.3	100%	100%	Central hypoventilation syndrome congenital,209880 Waardenburg syndrome, type 4B,613265 {Hirshprung disease,susceptibility to,4},613712
EDNRA	137.7	100%	100%	mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to},157300
EDNRB	156.5	100%	98%	ABCD syndrome,600501 Waardenburg syndrome, type 4A,277580 {Hirshprung disease, susceptibility to, 2},600155
EFEMP2	117.1	100%	100%	Cutis laxa,autosomal recessive,type IB,614437
EFNB1	109.9	100%	100%	Craniofrontonasal dysplasia,304110
EIF2AK3	125.8	92%	91%	Wolcott-Rallison syndrome,226980
ELN	75.7	100%	98%	Cutis laxa AD,123700 Supravalvar aortic stenosis,185500
ELOVL4	111.1	100%	100%	?Spinocerebellar ataxia 34,133190 Ichthyosis,spastic quadriplegia and mental retardation,614457 Stargardt disease 3,600110
ENAM	144	100%	100%	Amelogenesis imperfecta type IB,104500 Amelogenesis imperfecta type IC,204650

ENG	77.1	97%	90%	Telangiectasia,hereditary hemorrhagic,type 1,187300
ENPP1	122.8	95%	92%	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	97.6	100%	99%	Vici syndrome,242840
ERCC2	98.3	100%	96%	Cerebrooculofacioskeletal syndrome 2,610756 Trichothiodystrophy 1, photosensitive,601675 Xeroderma pigmentosum, group D,278730
ERCC3	133.7	100%	99%	Trichothiodystrophy 2, photosensitive,616390 Xeroderma pigmentosum, group B,610651
ERCC4	160.9	98%	95%	Fanconi anemia,complementation group Q,615272 Xeroderma pigmentosum, group F,278760 Xeroderma pigmentosum, type F/Cockayne syndrome,278760 XFE progeroid syndrome,610965
ERCC5	134.7	97%	96%	Xeroderma pigmentosum, group G,278780 Xeroderma pigmentosum, group G/Cockayne syndrome,278780
ERCC6	164.8	99%	98%	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	103.5	100%	100%	Cockayne syndrome type A,216400 UV-sensitive syndrome 2,614621
EVC	85.9	90%	88%	Ellis-van Creveld syndrome,225500 Weyers acrodental dysostosis,193530
EVC2	119.4	93%	92%	Ellis-van Creveld syndrome,225500 Weyers acrodental dysostosis,193530
EXPH5	172.5	100%	99%	Epidermolysis bullosa,nonspecific,autosomal recessive,615028
FAM111B	207.9	100%	98%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis,615704
FAM20A	76.7	99%	88%	Amelogenesis imperfecta,type IG (enamel-renal syndrome),204690
FAM20C	80.3	90%	85%	Raine syndrome,259775

FAM83H	76.7	99%	95%	Amelogenesis imperfecta type 3,130900
FANCA	101.3	99%	97%	Fanconi anemia complementation group A,227650
FANCB	149.6	100%	98%	Fanconi anemia complementation group B,300514
FANCC	89.8	99%	95%	Fanconi anemia complementation group C,227645
FANCD2	113.3	89%	86%	Fanconi anemia complementation group D2,227646
FANCE	97.2	96%	85%	Fanconi anemia complementation group E,600901
FANCF	136.6	100%	100%	Fanconi anemia complementation group F,603467
FANCG	143.3	100%	99%	Fanconi anemia complementation group G,614082
FANCI	143.3	100%	99%	Fanconi anemia complementation group I,609053
FANCL	104	100%	100%	Fanconi anemia complementation group L,614083
FANCM	130.6	100%	99%	Fanconi anemia complementation group M,614087
FAT4	183.1	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 2,616006 Van Maldergem syndrome 2,615546
FBLN5	84.9	91%	91%	Cutis laxa,autosomal dominant 2,614434 Cutis laxa,autosomal recessive,type IA,219100 Macular degeneration,age-related,3,608895
FECH	113.3	100%	100%	Protoporphyria erythropoietic autosomal recessive,177000
FERMT1	115.8	100%	98%	Kindler syndrome,173650
FGF10	126.9	100%	100%	Aplasia of lacrimal and salivary glands,180920 LADD syndrome,149730
FGF23	77.4	98%	92%	Hypophosphatemic rickets,autosomal dominant,193100 Osteomalacia,tumor-induced Tumoral calcinosis,hyperphosphatemic,familial,211900
FGF3	73.2	100%	91%	Deafness,congenital with inner ear agenesis,microtia and microdontia,610706
FGF5	190.9	100%	100%	trichomegaly,190330
FGF8	52.9	90%	62%	Hypogonadotropic hypogonadism 6 with or without anosmia,612702
FGFR1	132	100%	99%	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	128.4	97%	97%	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 syndrome,123500 Gastric cancer,somatic,613659 Jackson-Weiss syndrome,123150 LADD syndrome,149730 Pfeiffer syndrome,101600 Saethre-Chotzen syndrome,101400 Scaphocephaly, maxillary retrusion, and mental retardation,609579
FGFR3	73.2	94%	90%	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	99.9	97%	91%	Fumarase deficiency,606812 Leiomyomatosis and renal cell cancer,150800
FKBP10	88	100%	100%	Bruck syndrome 1,259450 Osteogenesis imperfecta type XI,610968

FKBP14	144.5	100%	100%	Ehlers-Danlos syndrome with progressive kyphoscoliosis myopathy and hearing loss,614557
FLCN	120.2	100%	97%	Birt-Hogg-Dube syndrome,135150 Colorectal cancer,somatic,114500 Pneumothorax,primary spontaneous,173600 Renal carcinoma,chromphobe,somatic,144700
FLG	55.2	100%	88%	Ichthyosis vulgaris,146700 {Dermatitis,atopic,susceptibility to,2},605803
FLG2	275.4	100%	100%	No OMIM phenotype
FLT4	101.5	98%	95%	Hemangioma,capillary infantile,somatic,602089 Lymphedema,hereditary,IA,153100
FNIP1	169	99%	98%	Familial multiple discoid fibromas,190340
FOXC2	84.3	99%	89%	Lymphedema-distichiasis syndrome with/without renal disease and diabetes mellitus,153400
FOXE1	38.3	100%	79%	Bamforth-Lazarus syndrome,241850
FOXN1	132.1	99%	98%	T-cell immunodeficiency congenital alopecia and nail dystrophy,601705
FOXP3	102.3	100%	98%	Immunodysregulation,polyendocrinopathy and enteropathy,X-linked,304790 {Diabetes mellitus,type I,susceptibility to},222100
FREM1	125.1	100%	99%	Bifid nose with or without anorectal and renal anomalies,608980 Manitoba oculotrichoanal syndrome,248450 Trigonocephaly 2,614485
FUCA1	85.1	100%	99%	Fucosidosis,230000
FZD6	163.9	100%	100%	Nail disorder,nonsyndromic,congenital 10 (claw-shaped nails),614157
GALNS	78.6	93%	93%	Mucopolysaccharidosis IVA,253000
GALNT3	123.7	100%	99%	Tumoral calcinosis,hyperphosphatemic,familial,211900
GAN	151.6	100%	98%	Giant axonal neuropathy-1,256850
GATA2	110.8	99%	93%	Emberger syndrome,614038 Immunodeficiency 21,614172 {Leukemia, acute myeloid, susceptibility to},601626 {Myelodysplastic syndrome, susceptibility to},614286
GDF2	153	100%	100%	Telangiectasia, hereditary hemorrhagic, type 5,615506

GDF5	103.7	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 Symphalangism,proximal, 1B,615298 {Osteoarthritis 5},612400
GGCX	107.3	100%	98%	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency,610842 Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450
GJA1	62.6	90%	78%	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	190	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 Vohwinkel syndrome,124500
GJB3	126.3	100%	100%	Deafness autosomal dominant 2B,612644 Deafness,digenic,GJB2/GJB3,220290 Erythrokeratodermia variabilis et progressiva,133200
GJB4	171.7	100%	100%	Erythrokeratodermia variabilis with erythema gyratum repens,133200
GJB6	176.8	100%	100%	Deafness,autosomal dominant 3B,612643 Deafness,autosomal recessive 1B,612645 Deafness,digenic GJB2/GJB6,220290 Ectodermal dysplasia 2,Clouston type,129500

GJC2	44.2	89%	81%	Leukodystrophy,hypomyelinating 2,608804 Lymphedema, hereditary,IC,613480 Spastic paraplegia 44,autosomal recessive,613206
GLA	102.7	100%	99%	Fabry disease,301500
GLB1	84.9	100%	96%	GM1-gangliosidosis type I,230500 GM1-gangliosidosis type II,230600 GM1-gangliosidosis type III,230650 Mucopolysaccharidosis type IVB (Morquio),253010
GLMN	105.4	100%	99%	Glomuvenous malformations,138000
GMPPA	152.1	100%	100%	Alacrima, achalasia, and mental retardation syndrome ,615510
GNAQ	79.3	100%	94%	Capillary malformations,congenital,1, somatic,mosaic,163000 Sturge-Weber syndrome, somatic, mosaic,185300
GNAS	123.6	99%	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	168.6	100%	100%	Geroderma osteodysplasticum,231070
GPR143	58.7	89%	81%	Nystagmus 6,congenital,X-linked,300814 Ocular albinism, type I, Nettleship-Falls type,300500
GRHL2	119.6	100%	100%	Deafness,autosomal dominant 28,608641 Ectodermal dysplasia/short stature syndrome,616029
GRHL3	117.5	99%	98%	Van der Woude syndrome 2, 606713
GSN	77.1	99%	89%	Amyloidosis Finnish type,105120
GTF2H5	121.4	100%	100%	Trichothiodystrophy 3,photosensitive,616395
HCCS	131.2	100%	99%	Linear skin defects with multiple congenital anomalies,309801
HDAC8	111.2	100%	99%	Cornelia de Lange syndrome 5,300882 Wilson-Turner syndrome,309585
HERC2	71.7	62%	60%	Mental retardation, autosomal recessive 38,615516 [Skin/hair/eye pigmentation 1],227220
HLCS	153.9	100%	100%	Holocarboxylase synthetase deficiency,253270

HMBS	112	100%	98%	Porphyria acute intermittent,176000
HMGB3	39.4	90%	63%	?Microphthalmia, syndromic 13,300915
HOXC13	64.6	100%	98%	Ectodermal dysplasia 9 hair/nail type,614931
HPS1	83	100%	94%	Hermansky-Pudlak syndrome 1,203300
HPS3	133.2	100%	97%	Hermansky-Pudlak syndrome 3,614072
HPS4	121.8	99%	98%	Hermansky-Pudlak syndrome 4,614073
HPS5	110.6	96%	96%	Hermansky-Pudlak syndrome 5,614074
HPS6	95.1	92%	81%	Hermansky-Pudlak syndrome 6,614075
HR	86.6	99%	93%	Alopecia universalis,203655 Atrichia with papular lesions,209500 Hypotrichosis 4,146550
HRAS	101.3	100%	100%	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	73.8	82%	77%	CARASIL syndrome,600142 {Macular degeneration,age-related,7},610149 {Macular degeneration,age-related,neovascular type},610149
HYAL1	95.8	100%	95%	Mucopolysaccharidosis type IX,601492
IDUA	89.5	93%	85%	Mucopolysaccharidosis Ih,607014 Mucopolysaccharidosis Ih/s,607015 Mucopolysaccharidosis Is,607016
IFT122	96.2	96%	95%	Cranioectodermal dysplasia 1,218330
IFT43	103.1	100%	100%	Cranioectodermal dysplasia 3,614099
IKBKG	30.9	26%	26%	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 disease,recurrent isolated,2,300640
IL17RA	96.8	98%	89%	?Candidiasis,familial 5,autosomal recessive,613953

IL17RD	113.2	100%	96%	Hypogonadotropic hypogonadism 18 with or without anosmia,615267
IL1RN	126.4	100%	100%	Interleukin 1 receptor antagonist deficiency,612852 {Gastric cancer risk after H.pylori infection},137215 {Microvascular complications of diabetes 4},612628
IL31RA	147.1	100%	97%	Amyloidosis,primary localized cutaneous 2,613955
IL36RN	109.7	100%	100%	Psoriasis 14, pustular,614204
INSR	133.4	97%	96%	Diabetes mellitus,insulin-resistant,with acanthosis nigricans,610549 Hyperinsulinemic hypoglycemia,familial,5,609968 Leprechaunism,246200 Rabson-Mendenhall syndrome,262190
IRF4	142.1	100%	100%	Multiple myeloma,254500 [Skin/hair/eye pigmentation, variation in,8],611724
IRF6	106.5	97%	93%	Orofacial cleft 6,608864 Popliteal pterygium syndrome 1,119500 van der Woude syndrome,119300
ITGA3	123	99%	92%	Interstitial lung disease, nephrotic syndrome and epidermolysis bullosa, congenital,614748
ITGA6	145.6	100%	99%	Epidermolysis bullosa,junctional, with pyloric stenosis,226730
ITGB4	89	98%	93%	Epidermolysis bullosa of hands and feet,131800 Epidermolysis bullosa,junctional,non-Herlitz type,226650 Epidermolysis bullosa,junctional,with pyloric atresia,226730
ITGB6	128.4	96%	95%	Amelogenesis imperfecta, type IH,616221
JUP	64.7	85%	76%	Arrhythmogenic right ventricular dysplasia 12,611528 Naxos disease,601214
KAL1	99.3	97%	92%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1),308700
KANK2	110.6	100%	100%	palmoplantar keratoderma and woolly hair,616099
KAT6B	163.2	100%	99%	Genitopatellar syndrome,606170 SBBYSS syndrome,603736
KCNH1	119	100%	99%	Temple-Baraitser syndrome,611816 Zimmermann-Laband syndrome,135500
KCNK9	128.2	100%	100%	Birk-Barel mental retardation dysmorphism syndrome,612292
KIF11	102.1	100%	98%	Microcephaly with or without chorioretinopathy lymphedema or mental retardation,152950

KIT	127.7	100%	98%	Gastrointestinal stromal tumor,familial,606764 Germ cell tumors,273300 Leukemia,acute myeloid,601626 Mast cell disease,154800 Piebaldism,172800
KITLG	76.8	100%	98%	Hyperpigmentation familial progressive 2,145250 [Skin/hair/eye pigmentation 7],611664
KLK4	161.4	100%	100%	Amelogenesis imperfecta type IIA1,204700
KLLN	104.9	100%	100%	Cowden syndrome 4,615107
KMT2D	115.1	99%	98%	Kabuki syndrome 1,147920
KRAS	71.2	97%	89%	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,163200
KRT1	113.1	100%	100%	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	113.4	87%	85%	Epidermolytic hyperkeratosis,113800 Ichthyosis with confetti,609165 Ichthyosis,cyclic,with epidermolytic hyperkeratosis,607602
KRT13	110	100%	100%	White sponge nevus 2,615785
KRT14	29	73%	51%	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	8	33%	10%	Pachyonychia congenita 1,167200 Palmoplantar keratoderma,nonepidermolytic,focal,613000
KRT17	10	39%	10%	Pachyonychia congenita 2,167210 Steatocystoma multiplex,184500
KRT2	123.7	100%	97%	Ichthyosis bullosa of Siemens,146800
KRT4	90.1	100%	99%	White sponge nevus 1,193900
KRT5	78.9	100%	95%	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 Epidermolysis bullosa simplex-MCR,609352
KRT6A	33.6	65%	46%	Pachyonychia congenita 3,167200
KRT6B	37.7	72%	44%	Pachyonychia congenita Jackson-Lawler type,615726
KRT6C	26	51%	41%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse,615735
KRT71	114.4	99%	92%	Hypotrichosis 13,615896
KRT74	89.1	90%	83%	?Ectodermal dysplasia 7, hair/nail type,614929 ?Hypotrichosis 3,613981 Woolly hair, autosomal dominant,194300
KRT75	107.9	100%	98%	{Pseudofolliculitis barbae,susceptibility to},612318
KRT81	21.6	61%	43%	Monilethrix,158000
KRT83	28.4	62%	48%	Monilethrix,158000
KRT85	42.4	86%	64%	Ectodermal dysplasia 4 hair/nail type,602032
KRT86	30.1	67%	56%	Monilethrix,158000
KRT9	131.6	96%	93%	Epidermolytic palmoplantar keratoderma,144200
LAMA3	113.1	99%	99%	Epidermolysis bullosa,generalized atrophic benign,226650 Epidermolysis bullosa,junctional,Herlitz type,226700 Laryngoonychocutaneous syndrome,245660
LAMB3	86.9	98%	96%	Amelogenesis imperfecta,type IA,104530 Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650
LAMC2	115.1	100%	99%	Epidermolysis bullosa,junctional,Herlitz type,226700 Epidermolysis bullosa,junctional,non-Herlitz type,226650

LAMTOR2	83.9	100%	100%	Immunodeficiency due to defect in MAPBP-interacting protein,610798
LDHA	51	70%	58%	Glycogen storage disease XI,612933
LDLRAP1	101.6	95%	87%	Hypercholesterolemia,familial,autosomal recessive,603813
LEMD3	119.3	100%	98%	Buschke-Ollendorff syndrome,166700 Melorheostosis with osteopoikilosis,155950 Osteopoikilosis,166700
LIPH	142.5	100%	100%	Hypotrichosis 7,604379 Woolly hair,autosomal recessive 2,with or without hypotrichosis
LIPN	127.2	100%	100%	Ichthyosis,congenital,autosomal recessive 8,613943
LMBRD1	120.7	100%	100%	Methylmalonic aciduria and homocystinuria cblF type,277380
LMNA	78.1	97%	90%	Cardiomyopathy dilated 1A,115200 Charcot-Marie-Tooth disease,type 2B1,605588 Emery-Dreifuss muscular dystrophy 2, AD,181350 Emery-Dreifuss muscular dystrophy 3,AR,616516 Heart-hand syndrome,Slovenian type,610140 Hutchinson-Gilford progeria,176670 Lipodystrophy,familial,partial,2,151660 Malouf syndrome,212112 Mandibuloacral dysplasia,248370 Muscular dystrophy,congenital,613205 Muscular dystrophy,limb-girdle,type 1B,159001 Restrictive dermopathy,lethal,275210
LMX1B	97.3	100%	96%	Nail-patella syndrome,161200
LONP1	88.4	97%	91%	CODAS syndrome,600373
LOR	33.4	95%	78%	Vohwinkel syndrome with ichthyosis,604117
LPAR6	144.8	100%	100%	Hypotrichosis 8,278150 Woolly hair,autosomal recessive 1,with or without hypotrichosis,278150
LPIN2	84.6	100%	98%	Majeed syndrome,609628
LTBP3	77.8	100%	94%	Dental anomalies and short stature,601216
LTBP4	90.1	98%	88%	Cutis laxa autosomal recessive type IC,613177
LYST	135.2	99%	97%	Chediak-Higashi syndrome,214500
LYZ	109.7	100%	100%	Amyloidosis,renal,105200
MAP2K1	102.4	96%	91%	Cardiofaciocutaneous syndrome 3,615279
MAP2K2	113.8	99%	92%	Cardiofaciocutaneous syndrome 4,615280

MBTPS2	155.3	100%	100%	?Olmsted syndrome,X-linked,300918 IFAP syndrome with or without BRESHECK syndrome,308205 Keratosis follicularis spinulosa decalvans,X-linked,308800
MED12	144.1	98%	94%	Lujan-Fryns syndrome,309520 Ohdo syndrome,X-linked,300895 Opitz-Kaveggia syndrome,305450
MEFV	113.7	96%	96%	Familial Mediterranean fever AD,134610 Familial Mediterranean fever AR,249100
MGP	84.7	100%	98%	Keutel syndrome,245150
MITF	140.9	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	114.6	100%	98%	Colorectal cancer,hereditary,nonpolyposis type 2,609310 Mismatch repair cancer syndrome,276300 Muir-Torre syndrome,158320
MLPH	87	95%	88%	Griscelli syndrome type 3,609227
MMACHC	197.5	100%	100%	Methylmalonic aciduria and homocystinuria cblC type,277400
MMP2	114	100%	99%	Torg-Winchester syndrome,259600
MMP20	113.6	100%	100%	Amelogenesis imperfecta type IIA2,612529
MPLKIP	67.6	100%	100%	Trichothiodystrophy nonphotosensitive 1,234050
MRE11A	105.2	99%	99%	Ataxia-telangiectasia-like disorder,604391
MSH2	114.6	99%	97%	Colorectal cancer,hereditary,nonpolyposis type 1,120435 Mismatch repair cancer syndrome,276300 Muir-Torre syndrome,158320
MSX1	47.6	85%	78%	Ectodermal dysplasia 3,Witkop type,189500 Orofacial cleft 5,608874 Tooth agenesis,selective,1,with or without orofacial cleft,106600
MUTYH	124.9	100%	100%	Adenomas,multiple colorectal,608456 Colorectal denomatous polyposis,autosomal recessive,with pilomatricomas,132600 Gastric cancer,somatic,613659
MVK	93.8	100%	98%	Hyper-IgD syndrome,260920 Mevalonic aciduria,610377 Porokeratosis 3,disseminated superficial actinic,175900

MYH8	116.4	98%	90%	Carney complex variant,608837 Trismus-pseudocamptodactyly syndrome,158300
MYO5A	106.4	99%	98%	Griscelli syndrome type 1,214450
NAA10	120.4	97%	97%	N-terminal acetyltransferase deficiency,300855 ?Microphthalmia,syndromic 1,309800
NAGA	82.2	100%	95%	Kanzaki disease,609242 Schindler disease,609241
NBAS	116	100%	100%	Short stature,optic nerve atrophy and Pelger-Huet anomaly,614800 Infantile liver failure syndrome 2,616483
NCSTN	94.7	96%	92%	Acne inversa familial 1,142690
NDUFB11	103.6	99%	98%	Linear skin defects with multiple congenital anomalies 3,300952
NF1	91.1	83%	81%	Neurofibromatosis, type 1,162200
NFKBIA	106.9	100%	100%	Ectodermal dysplasia anhidrotic with T-cell immunodeficiency,612132
NHP2	62.6	100%	97%	Dyskeratosis congenita, autosomal recessive 2,613987
NIPAL4	124	99%	95%	Ichthyosis,congenital,autosomal recessive 6,612281
NIPBL	131	98%	98%	Cornelia de Lange syndrome 1,122470
NLRP1	120.7	99%	96%	?Corneal intraepithelial dyskeratosis and ectodermal dysplasia,615225 {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579
NLRP3	131	100%	99%	CINCA syndrome,607115 Familial cold-induced inflammatory syndrome 1,120100 Muckle-Wells syndrome,191900
NME1	150.2	100%	100%	Neuroblastoma,256700
NOD2	104.3	100%	98%	Blau syndrome,186580 Sarcoidosis,early-onset,609464 {Inflammatory bowel disease 1},266600 {Psoriatic arthritis,susceptibility to},607507
NOP10	178.9	100%	100%	Dyskeratosis congenita, autosomal recessive 1,224230
NOTCH1	74.3	97%	90%	Aortic valve disease,109730 Adams-Oliver syndrome 5,616028

NRAS	140.8	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 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic,163200 Thyroid carcinoma,follicular,somatic,188470
NSD1	136.5	100%	99%	Beckwith-Wiedemann syndrome,130650 Leukemia,acute myeloid,601626 Sotos syndrome,117550
NSDHL	104.7	100%	98%	CHILD syndrome,308050 CK syndrome,300831
OCA2	104.3	100%	99%	Albinism brown oculocutaneous,203200 [Skin/hair/eye pigmentation 1],227220
ODAM	123	100%	100%	No OMIM phenotype
OFD1	82.5	95%	91%	?Retinitis pigmentosa 23,300424 Joubert syndrome 10,300804 Orofaciodigital syndrome 1,311200 Simpson-Golabi-Behmel syndrome, type 2,300209
OSMR	151.5	100%	100%	Amyloidosis primary localized cutaneous 1,105250
PAH	96.2	96%	95%	Phenylketonuria,261600
PALB2	148.8	100%	99%	Fanconi anemia complementation group N,610832 {Breast cancer,susceptibility to},114480 {Pancreatic cancer,susceptibility to 3},613348
PAX3	127.7	100%	99%	Craniofacial-deafness-hand syndrome,122880 Rhabdomyosarcoma 2,alveolar,268220 Waardenburg syndrome,type 1,193500 Waardenburg syndrome,type 3,148820
PAX9	218.5	99%	99%	Tooth agenesis selective 3,604625
PCNA	108.1	100%	100%	Ataxia-telangiectasia-like disorder 2,615919
PDGFB	71.7	100%	100%	Dermatofibrosarcoma protuberans,607907 Basal ganglia calcification,idiopathic,5,615483 Meningioma, SIS-related,607174

PDGFRB	95	100%	97%	Basal ganglia calcification idiopathic 4,615007 Myeloproliferative disorder with eosinophilia, 131440 Myofibromatosis, infantile, 1, 228550
PEPD	77.7	95%	92%	Prolidase deficiency,170100
PEX7	109.7	84%	81%	Peroxisome biogenesis disorder 9B,614879 Chondrodysplasia punctata, rhizomelic, type 1,215100
PHEX	138.2	98%	98%	Hypophosphatemic rickets X-linked dominant,307800
PHGDH	100.9	100%	99%	Phosphoglycerate dehydrogenase deficiency,601815 Neu-Laxova syndrome 1,256520
PHYH	100.9	100%	100%	Refsum disease,266500
PIEZO1	98.4	98%	94%	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema ,194380
PIGA	151.7	100%	99%	Multiple congenital anomalies-hypotonia-seizures syndrome 2,300868 Paroxysmal nocturnal hemoglobinuria,somatic,300818
PIGN	116.5	100%	100%	Multiple congenital anomalies-hypotonia-seizures syndrome 1,614080
PIGV	219	100%	100%	Hyperphosphatasia with mental retardation syndrome 1,239300
PIK3CA	136.7	94%	92%	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 malformation-polymicrogyria syndrome, somatic,602501 Nevus,epidermal,somatic,162900 Nonsmall cell lung cancer,somatic,211980 Ovarian cancer,somatic,167000
PITX2	124.5	96%	93%	Axenfeld-Rieger syndrome type 1,180500 Iridogoniodysgenesis,type 2,137600 Peters anomaly,604229 Ring dermoid of cornea,180550
PKP1	86.6	98%	90%	Ectodermal dysplasia/skin fragility syndrome,604536

PLCD1	113.9	100%	93%	Nail disorder nonsyndromic congenital 3 (leukonychia),151600
PLCG2	113.1	100%	99%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome,614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	105.6	98%	96%	?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-girdle,type 2Q,613723
PLG	72.4	75%	72%	Dysplasminogenemia,217090
PLIN1	52.4	85%	75%	Lipodystrophy familial partial type 4,613877
PLOD1	85.4	100%	97%	Ehlers-Danlos syndrome type VI,225400
PMS2	77.8	56%	56%	Colorectal cancer hereditary nonpolyposis type 4,614337 Mismatch repair cancer syndrome,276300
PNPLA1	140.4	100%	99%	Ichthyosis congenital autosomal recessive 10,615024
PNPLA2	89.3	100%	96%	Neutral lipid storage disease with myopathy,610717
POC1A	118.4	100%	95%	Short stature onychodysplasia facial dysmorphism and hypotrichosis,614813
POFUT1	128.6	100%	96%	Dowling-Degos disease 2,615327
POGLUT1	123.7	100%	97%	Dowling-Degos disease 4,615696
POLD1	81.5	94%	92%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome,615381 {Colorectal cancer,susceptibility to,10},612591
POLH	152.6	97%	96%	Xeroderma pigmentosum variant type,278750
POLR1C	124.4	90%	87%	Treacher Collins syndrome 3,248390 Leukodystrophy, hypomyelinating,11,616494
POLR1D	177.3	100%	100%	Treacher Collins syndrome 2,613717
POLR3A	100.6	100%	94%	Leukodystrophy hypomyelinating 7 with or without oligodontia and/or hypogonadotropic hypogonadism,607694
POLR3B	120	100%	99%	Leukodystrophy hypomyelinating 8 with or without oligodontia and/or hypogonadotropic hypogonadism,614381
POMC	53.5	80%	66%	Obesity adrenal insufficiency and red hair due to POMC deficiency,609734 {Obesity,early-onset,susceptibility to},601665
POMP	197.6	100%	100%	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma,601952
PORCN	122.4	94%	92%	Focal dermal hypoplasia,305600
PPOX	117.9	100%	97%	Porphyria variegata,176200
PQBP1	154.9	100%	99%	Renpenning syndrome,309500

PRKAR1A	118	96%	91%	Acrodysostosis 1 with or without hormone resistance,101800 Carney complex,type 1,160980 Myxoma,intracardiac,255960 Pigmented nodular adrenocortical disease,primary,1,610489
PSEN1	110.7	100%	96%	Acne inversa familial 3,613737 Alzheimer disease,type 3,607822 Cardiomyopathy,dilated,1U,613694 Dementia,frontotemporal,600274 Pick disease,172700
PSENEN	132.2	100%	99%	Acne inversa familial 2,613736
PSMB8	11.2	48%	10%	Autoinflammation lipodystrophy and dermatosis syndrome,256040
PSTPIP1	62.3	99%	91%	Pyogenic sterile arthritis pyoderma gangrenosum and acne,604416
PTCH1	89.5	97%	94%	Basal cell carcinoma somatic,605462 Basal cell nevus syndrome, 109400 Holoprosencephaly-7,610828
PTCH2	91	98%	96%	Basal cell carcinoma somatic,605462 Basal cell nevus syndrome,109400 Medulloblastoma,155255
PTDSS1	128.3	100%	100%	Lenz-Majewski hyperostotic dwarfism,151050
PTEN	136	95%	94%	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,follicular,somatic,188470 VATER association with macrocephaly and ventriculomegaly,276950 {Glioma susceptibility 2},613028 {Meningioma},607174 {Prostate cancer,somatic},176807
PTHLH	144.9	100%	100%	Brachydactyly type E2,613382 Humoral hypercalcemia of malignancy

PTPN11	55.5	88%	72%	LEOPARD syndrome 1,151100 Leukemia,juvenile myelomonocytic,607785 Metachondromatosis,156250 Noonan syndrome 1,163950
PTPN14	129.6	100%	98%	Choanal atresia and lymphedema,613611
PTPRF	110.7	100%	99%	Breasts and/or nipples, aplasia or hypoplasia of, 2,616001
PTRF	138.1	100%	100%	Lipodystrophy congenital generalized type 4,613327
PVRL1	87	99%	98%	Cleft lip/palate-ectodermal dysplasia syndrome,225060 Orofacial cleft 7,225060
PVRL4	112.7	100%	99%	Ectodermal dysplasia-syndactyly syndrome 1,613573
PYCR1	92.8	100%	99%	Cutis laxa autosomal recessive type IIB,612940 Cutis laxa autosomal recessive type IIIB,614438
RAB23	156.8	100%	100%	Carpenter syndrome,201000
RAB27A	136.9	100%	100%	Griscelli syndrome type 2,607624
RAD21	106	100%	96%	Cornelia de Lange syndrome 4,614701
RAD50	123.9	100%	100%	Nijmegen breakage syndrome-like disorder,613078
RAF1	97.4	100%	100%	LEOPARD syndrome 2,611554 Cardiomyopathy,dilated,1NN,615916 Noonan syndrome 5,611553
RAG1	161	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	231.7	100%	100%	Combined cellular and humoral immune defects with granulomas,233650 Omenn syndrome,603554 Severe combined immunodeficiency,B cell-negative,601457
RAI1	144.1	99%	98%	Smith-Magenis syndrome,182290
RBBP8	136.9	100%	100%	Jawad syndrome,251255 Pancreatic carcinoma,somatic Seckel syndrome 2,606744
RBM28	123.7	100%	99%	Alopecia neurologic defects and endocrinopathy syndrome,612079
RBP4	82.8	96%	85%	Retinol dystrophy iris coloboma and comedogenic acne syndrome,615147 Microphthalmia,isolated,with coloboma 10,616428

RBPJ	78	98%	95%	Adams-Oliver syndrome 3,614814
RECQL4	103.3	97%	95%	Baller-Gerold syndrome,218600 RAPADILINO syndrome,266280 Rothmund-Thomson syndrome,268400
RHBDF2	70.3	98%	92%	Tylosis with esophageal cancer,148500
RIN2	120.5	99%	97%	Macrocephaly alopecia cutis laxa and scoliosis,613075
RIPK4	104.4	99%	96%	Popliteal pterygium syndrome 2 lethal type,263650
RNASEH2A	102.6	100%	92%	Aicardi-Goutieres syndrome 4,610333
RNASEH2B	110.6	99%	93%	Aicardi-Goutieres syndrome 2,610181
RNASEH2C	160.2	100%	100%	Aicardi-Goutieres syndrome 3,610329
ROGDI	107.4	95%	95%	Kohlschutter-Tonz syndrome,226750
RPL21	57.5	99%	84%	Hypotrichosis 12,615885
RSPO1	47.3	92%	87%	Palmoplantar hyperkeratosis and true hermaphroditism,610644 Palmoplantar hyperkeratosis with squamous cell skin carcinoma and sex revearsal,610644
RSPO4	87.9	100%	100%	Anonychia congenita,206800
RTEL1	84.8	100%	90%	Dyskeratosis congenita,autosomal recessive 5,615190 Dyskeratosis congenita,autosomal dominant 4,615190 Pulmonary fibrosis and/or bone marrow failure,telomere-related,616373
RUNX2	101.2	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/without brachydactyly,156510
SAMD9	227.9	100%	100%	Tumoral calcinosis familial normophosphatemic,610455
SAMHD1	136	100%	98%	Aicardi-Goutieres syndrome 5,612952 Chilblain lupus 2,614415
SART3	101.1	100%	98%	No OMIM phenotype
SAT1	149.1	100%	100%	Keratosis follicularis spinulosa decalvans,308800
SATB2	118.8	100%	97%	Cleft palate and mental retardation,119540
SCN10A	139.9	99%	98%	Epilepsy generalized with febrile seizures plus type 7,613863
SCN11A	134.2	100%	99%	Episodic pain syndrome,familial,3,615552 Neuropathy,hereditary sensory and autonomic, type VII,615548

SCN9A	123.2	100%	100%	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	125	100%	100%	Anemia dyserythropoietic congenital type II,224100
SERPINB7	123.4	100%	100%	Palmoplantar keratoderma, Nagashima type, 615598 ,615598
SERPINH1	135.9	100%	100%	Osteogenesis imperfecta type X,613848 {Preterm premature rupture of the membranes, susceptibility to},610504
SHOC2	129.4	100%	97%	Noonan-like syndrome with loose anagen hair,607721
SKI	60.5	82%	77%	Shprintzen-Goldberg syndrome,182212
SKIV2L	21.9	78%	48%	Trichohepatoenteric syndrome 2,614602
SLC17A9	99.3	99%	92%	Porokeratosis, disseminated superficial actinic, 8,616063
SLC24A4	120.7	99%	96%	Ameliogenesis imperfecta, hypomaturation type, IIA5,615887 [Skin/hair/eye pigmentation 6],210750
SLC24A5	122.6	100%	99%	Albinism, oculocutaneous, type VI,113750 [skin/hair/eye pigmentation 4],113750
SLC26A2	156.8	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	90.8	87%	83%	Ichthyosis prematurity syndrome,608649
SLC29A3	168.5	100%	99%	Histiocytosis-lymphadenopathy plus syndrome,602782
SLC2A10	101.8	100%	98%	Arterial tortuosity syndrome,208050
SLC39A13	124.6	100%	98%	Spondylocheirodysplasia Ehlers-Danlos syndrome-like,612350
SLC39A4	79.9	100%	98%	Acrodermatitis enteropathica,201100
SLC45A2	130.7	99%	99%	Oculocutaneous albinism type IV,606574 [skin/hair/eye pigmentation 5],227240
SLC4A4	122.3	100%	100%	Renal tubular acidosis proximal with ocular abnormalities,604278

SLC6A19	98.2	100%	95%	Hartnup disorder,234500 Hyperglycinuria,138500 Iminoglycinuria,digenic,242600
SLC7A7	109	100%	99%	Lysinuric protein intolerance,222700
SLCO2A1	84.3	100%	96%	Hypertrophic osteoarthropathy primary autosomal recessive 2,614441
SLURP1	37.6	99%	87%	Meleda disease,248300
SLX4	144.8	99%	95%	Fanconi anemia complementation group P,613951
SMAD3	84.3	88%	85%	Loeys-Dietz syndrome type 3,613795
SMARCA2	94	96%	92%	Nicolaides-Baraitser syndrome,601358
SMARCA4	93.3	98%	94%	Mental retardation autosomal dominant 16,614609 {Rhabdoid tumor predisposition syndrome 2},613325
SMARCAD1	134.3	100%	100%	Adermatoglyphia,136000
SMARCAL1	131.8	99%	97%	Schimke immunoosseous dysplasia,242900
SMARCB1	144	100%	100%	Mental retardation autosomal dominant 15,614608 Rhabdoid tumors, somatic,609322 {Schwannomatosis-1,susceptibility to},162091
SMO	117.2	99%	96%	Basal cell carcinoma, somatic
SMOC2	88.6	95%	90%	Dentin dysplasia type I with microdontia and misshapen teeth,125400
SNAI2	88.7	100%	100%	Piebaldism,172800 Waardenburg syndrome, type 2D,608890
SNAP29	117.1	100%	100%	Cerebral dysgenesis neuropathy, ichtyosis, and palmoplantar keratoderma syndrome,609528
SNRPE	60.8	79%	79%	Hypotrichosis 11,615059
SNX10	106.1	100%	100%	Osteopetrosis autosomal recessive 8,615085
SOS1	126.7	100%	99%	Fibromatosis,gingival,135300 Noonan syndrome 4,610733
SOX10	72.4	100%	100%	PCWH syndrome,609136 Waardenburg syndrome,type 2E,with/without neurological involvement,611584 Waardenburg syndrome,type 4C,613266
SOX18	16.1	66%	38%	Hypotrichosis-lymphedema-telangiectasia syndrome,607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome,137940
SOX2	144.4	100%	99%	Microphthalmia syndromic 3,206900 Optic nerve hypoplasia and abnormalities of the central nervous system,206900
SP7	89.1	100%	100%	Osteogenesis imperfecta type XII,613849

SPINK5	113.6	100%	99%	Atopy,147050 Netherton syndrome,256500
SPINT2	63	89%	60%	Diarrhea 3 secretory sodium congenital syndromic,270420
SPRED1	147.3	100%	100%	Legius syndrome,611431
SPRY4	100.3	100%	100%	Hypogonadotropic hypogonadism 17 with or without anosmia,615266
SRD5A3	132.2	100%	97%	Congenital disorder of glycosylation type Iq,612379 Kahrizi syndrome,612713
ST14	86.6	99%	92%	Ichthyosis with hypotrichosis,610765
ST3GAL5	104.9	94%	92%	Ganglioside GM3 synthase deficiency,609056
STAMBP	133.6	100%	99%	Microcephaly-capillary malformation syndrome,614261
STAT3	94.5	99%	98%	Hyper-IgE recurrent infection syndrome,147060 Autoimmune disease,multisystem,infantile-onset,615952
STAT5B	78.6	79%	74%	Growth hormone insensitivity with immunodeficiency,245590 Leukemia,acute promyelocytic,STAT5B/RARA type
STIM1	99.3	100%	94%	Immune dysfunction with T-cell inactivation due to calcium entry defect 2,612783 Myopathy,tubular aggregate,1,160565 Stormorken syndrome,185070
STK11	77.9	99%	96%	Melanoma malignant, somatic Pancreatic cancer, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STS	160.9	100%	100%	Ichthyosis X-linked,308100
SUFU	108.8	97%	91%	Medulloblastoma desmoplastic,155255 Basal cell nevus syndrome,109400 {Meningioma,familial,susceptibility to},607174
SUMF1	84.2	100%	95%	Multiple sulfatase deficiency,272200
TALDO1	100	100%	100%	Transaldolase deficiency,606003
TAT	107.8	100%	100%	Tyrosinemia type II,276600
TBC1D24	110.7	100%	99%	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	77.4	96%	91%	Ulnar-mammary syndrome,181450

TCIRG1	78.9	90%	81%	Osteopetrosis autosomal recessive 1,259700
TEK	123.2	99%	99%	Venous malformations multiple cutaneous and mucosal,600195
TERT	118.2	100%	99%	{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, 1},614742
TFAP2A	73.2	98%	87%	Branchiooculofacial syndrome,113620
TGFB2	128.4	100%	97%	Loeys-Dietz syndrome type 4,614816
TGFBR1	135.2	93%	93%	Loeys-Dietz syndrome type 1A,609192 {Multiple self-healing squamous epithelioma,susceptibility to},132800
TGFBR2	110.9	100%	99%	Colorectal cancer,hereditary,nonpolyposis type 6,614331 Esophageal cancer,somatic,133239 Loeys-Dietz syndrome,type 2,610168
TGM1	124.9	100%	98%	Ichthyosis congenital autosomal recessive 1,242300
TGM5	118.9	100%	100%	Peeling skin syndrome acral type,609796
TINF2	209.7	100%	100%	Dyskeratosis congenita autosomal dominant 3,613990 Revesz syndrome,268130
TMC6	66.6	99%	95%	Epidermodysplasia verruciformis,226400
TMC8	85.6	98%	93%	Epidermodysplasia verruciformis,226400
TMEM165	89	100%	100%	Congenital disorder of glycosylation type IIk,614727
TMEM173	71.5	99%	96%	STING-associated vasculopathy, infantile-onsent (SAVI),615934
TNFRSF11A	105.5	94%	91%	Osteolysis,familial expansile,174810 Osteopetrosis,autosomal recessive 7,612301 {Paget disease of bone 2,early-onset},602080
TNFRSF11B	185.2	100%	100%	Paget disease of bone 5, juvenile-onset,239000
TNFRSF1A	71.3	93%	88%	Periodic fever,familial,142680 {Multiple sclerosis, susceptibility to,5},614810
TNFSF11	147.7	100%	100%	Osteopetrosis,autosomal recessive 2,259710
TNXB	11.9	47%	21%	Ehlers-Danlos syndrome,autosomal dominant,hypermobility type,130020 Vesicoureteral reflux 8,615963

TP63	144.7	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	98.9	100%	97%	Skin/hair/eye pigmentation, variation in, 10,612267
TREX1	154	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	118	100%	100%	?Bardet-Biedl syndrome 11,615988 Muscular dystrophy,limb-girdle,type 2H,254110
TRIM37	118.7	100%	97%	Mulibrey nanism,253250
TRPV3	106	98%	94%	?Palmoplantar keratoderma,nonepidermolytic,focal 2,616400 Olmsted syndrome,614594
TSC1	106.4	99%	97%	Focal cortical dysplasia,Taylor balloon cell type,607341 Lymphangioleiomyomatosis,606690 Tuberous sclerosis-1,191100
TSC2	92.2	99%	96%	Lymphangioleiomyomatosis,somatic,606690 Tuberous sclerosis-2,613254
TTC37	128.8	100%	100%	Trichohepatoenteric syndrome 1,222470
TTI2	107.9	100%	100%	Mental retardation, autosomal recessive 39,615541
TWIST2	74.9	100%	91%	Ablepharon-macrostomia syndrome,200110 Barber-Say syndrome,209885 Focal facial dermal dysplasia 3,Setleis type,227260
TYR	138.7	74%	74%	Albinism,oculocutaneous,type IA,203100 Albinism,oculocutaneous,type IB,606952 Waardenburg syndrome/albinism,digenic,103470 [Skin/hair/eye pigmentation 3],601800
TYRP1	127.3	100%	99%	Albinism oculocutaneous type III,203290 [Skin/hair/eye pigmentation,variation in,11(Melanesian blond hair),612271
UBE2A	107.6	100%	100%	Mental retardation,X-linked syndromic,Nascimento-type,300860

UBR1	117.2	100%	100%	Johanson-Blizzard syndrome,243800
UROD	93.8	99%	93%	Porphyria,cutanea tarda,176100 Porphyria,hepatoerythropoietic,176100
UROS	91.9	97%	91%	Porphyria,congenital erythropoietic,263700
USB1	60.8	92%	84%	Poikiloderma with neutropenia,604173
UVSSA	71.1	100%	95%	UV-sensitive syndrome 3,614640
VDR	92.3	100%	100%	Rickets,vitamin D-resistant,type IIA,277440 ?Osteoporosis,involutional,166710
VEGFC	115	99%	97%	Lymphedema, hereditary, ID,615907
VHL	118.5	100%	100%	Erythrocytosis,familial,2,263400 Hemangioblastoma,cerebellar,somatic Pheochromocytoma,171300 Renal cell carcinoma,somatic,144700 von Hippel-Lindau syndrome,193300
VPS13B	124	99%	98%	Cohen syndrome,216550
WAS	66.1	100%	94%	Neutropenia,severe congenital,X-linked,300299 Thrombocytopenia,X-linked,313900 Wiskott-Aldrich syndrome,301000
WDR19	140.6	100%	100%	?Cranioectodermal dysplasia 4,614378 ?Short-rib thoracic dysplasia 5 with/without polydactyly,614376 Nephronophthisis 13,614377 Senior-Loken syndrome 8,616307
WDR35	131.6	100%	99%	Cranioectodermal dysplasia 2,613610 Short-rib thoracic dysplasia 7 with/without polydactyly,614091
WDR72	129.4	100%	100%	Amelogenesis imperfecta hypomaturation type IIA3,613211
WIPF1	107.1	100%	95%	Wiskott-Aldrich syndrome 2,614493
WNT10A	73.1	98%	81%	Odontoonychodermal dysplasia,257980 Schopf-Schulz-Passarge syndrome,224750 Tooth agenesis,selective,4,150400
WNT10B	104.7	99%	95%	Split-hand/foot malformation 6,225300
WNT5A	117.8	100%	99%	Robinow syndrome autosomal dominant,180700
WNT7A	129.4	100%	100%	Fuhrmann syndrome,228930 Ulna and fibula,absence of,with severe limb deficiency,276820
WRAP53	149	100%	99%	Dyskeratosis congenita autosomal recessive 3,613988

WRN	153.4	100%	99%	Werner syndrome,277700
XPA	94.3	100%	96%	Xeroderma pigmentosum group A,278700
XPC	123.3	98%	97%	Xeroderma pigmentosum group C,278720
XYLT1	124.2	91%	85%	Desbuquois dysplasia 2,615777 {Pseudoxanthoma elasticum,modifier of severity of},264800
XYLT2	85.4	95%	90%	Spondyloocular syndrome,605822 {Pseudoxanthoma elasticum,modifier of severity of},264800
ZBTB20	132.9	100%	100%	Primrose syndrome,259050
ZMPSTE24	171	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy,608612 Restrictive dermopathy,lethal,275210
ZNF469	95	100%	99%	Brittle cornea syndrome 1,229200
ZNF592	124.5	92%	91%	Spinocerebellar ataxia,autosomal recessive 5,606937
ZNF750	122	100%	99%	Seborrhea-like dermatitis with psoriasiform elements,610227

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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
OMIM release used for OMIM disease identifiers and descriptions : June 30th, 2015
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors