SKIN DISORDERS GENE PANEL DG 2.17 (621 genes)

Releasedate: 06-12-2019

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	109.1	100.0%	99.8%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	132.8	100.0%	100.0%	Keratoderma, palmoplantar, punctate type IA, 148600
ABCA12	129.0	99.6%	98.3%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCB6	136.7	100.0%	99.9%	Dyschromatosis universalis hereditaria 3, 615402 Microphthalmia, isolated, with coloboma 7, 614497 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153
ABCC6	116.6	93.7%	93.1%	Pseudoxanthoma elasticum, 264800 Pseudoxanthoma elasticum, forme fruste, 177850 Arterial calcification, generalized, of infancy, 2, 614473
ABCC9	140.4	100.0%	99.9%	Hypertrichotic osteochondrodysplasia, 239850 Atrial fibrillation, familial, 12, 614050 Cardiomyopathy, dilated, 1O, 608569
ABHD5	183.6	100.0%	100.0%	Chanarin-Dorfman syndrome, 275630
ACD	180.3	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
ACTA2	89.6	99.9%	98.6%	Aortic aneurysm, familial thoracic 6, 611788 Multisystemic smooth muscle dysfunction syndrome, 613834 Moyamoya disease 5, 614042
ACTB	92.6	100.0%	99.9%	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACVRL1	125.9	100.0%	98.8%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADA2	88.7	99.8%	97.9%	?Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM10	117.5	94.7%	92.7%	Reticulate acropigmentation of Kitamura, 615537
ADAM17	117.8	99.7%	98.5%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAMTS10	135.4	100.0%	99.9%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	118.3	98.7%	93.9%	Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS2	136.7	100.0%	99.9%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	137.9	100.0%	99.9%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154

ADAMTSL2	126.3	99.1%	96.8%	Geleophysic dysplasia 1, 231050
ADAR	117.2	99.9%	99.4%	Aicardi-Goutieres syndrome 6, 615010 Dyschromatosis symmetrica hereditaria, 127400
AGA	144.3	100.0%	100.0%	Aspartylglucosaminuria, 208400
AGPAT2	180.5	99.7%	97.0%	Lipodystrophy, congenital generalized, type 1, 608594
AIRE	113.3	100.0%	100.0%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKT1	168.9	100.0%	99.8%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500
AKT3	79.2	98.6%	94.0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	101.8	99.4%	95.4%	Porphyria, acute hepatic, 612740
ALAS2	77.2	99.1%	95.5%	Protoporphyria, erythropoietic, X-linked, 300752 Anemia, sideroblastic, 1, 300751
ALDH18A1	116.9	100.0%	99.8%	Cutis laxa, autosomal recessive, type IIIA, 219150 Cutis laxa, autosomal dominant 3, 616603 Spastic paraplegia 9B, autosomal recessive, 616586 Spastic paraplegia 9A, autosomal dominant, 601162
ALDH3A2	116.9	95.3%	94.2%	Sjogren-Larsson syndrome, 270200
ALDOB	140.0	100.0%	99.1%	Fructose intolerance, hereditary, 229600
ALOX12B	137.8	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	137.6	100.0%	99.6%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	168.4	99.9%	99.5%	Hypophosphatasia, adult, 146300 Odontohypophosphatasia, 146300 Hypophosphatasia, childhood, 241510 Hypophosphatasia, infantile, 241500
ALX4	175.4	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMELX	90.7	99.0%	93.8%	Amelogenesis imperfecta, type 1E, 301200
ANKRD11	131.8	99.6%	97.6%	KBG syndrome, 148050
ANOS1	78.3	91.4%	87.3%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	112.9	99.3%	97.3%	GAPO syndrome, 230740
ANTXR2	117.5	99.8%	97.7%	Hyaline fibromatosis syndrome, 228600
AP1S3	110.7	90.5%	90.4%	No OMIM disease ID
AP3B1	108.2	99.4%	95.7%	Hermansky-Pudlak syndrome 2, 608233

APC	143.0	100.0%	99.6%	Desmoid disease, hereditary, 135290 Adenomatous polyposis coli, 175100 Gardner syndrome, 175100 Hepatoblastoma, somatic, 114550 Colorectal cancer, somatic, 114500 Brain tumor-polyposis syndrome 2, 175100 Gastric cancer, somatic, 613659 Adenoma, periampullary, somatic, 0
APCDD1	182.1	100.0%	99.5%	Hypotrichosis 1, 605389
AQP5	136.8	100.0%	99.9%	Palmoplantar keratoderma, Bothnian type, 600231
ARHGAP31	151.5	99.9%	99.1%	Adams-Oliver syndrome 1, 100300
ARID1A	145.9	99.7%	98.9%	Coffin-Siris syndrome 2, 614607
ARID1B	150.6	99.5%	99.3%	Coffin-Siris syndrome 1, 135900
ASIP	166.4	100.0%	99.9%	No OMIM disease ID
ASL	135.7	99.9%	99.2%	Argininosuccinic aciduria, 207900
ASXL1	141.0	100.0%	99.6%	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
ASXL3	141.2	99.8%	99.1%	Bainbridge-Ropers syndrome, 615485
ATIC	114.9	100.0%	99.9%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP2A2	150.4	100.0%	99.9%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	109.2	100.0%	99.5%	Hailey-Hailey disease, 169600
ATP6V0A2	120.5	100.0%	99.6%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP7A	109.4	99.7%	97.2%	Occipital horn syndrome, 304150 Menkes disease, 309400 Spinal muscular atrophy, distal, X-linked 3, 300489
ATR	142.1	99.9%	98.9%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	134.4	100.0%	99.9%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B3GALT6	96.5	87.5%	80.2%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B4GALT7	138.9	100.0%	99.1%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BANF1	52.9	97.3%	86.5%	Nestor-Guillermo progeria syndrome, 614008
BAP1	113.0	85.4%	83.2%	Tumor predisposition syndrome, 614327
BCOR	109.0	99.2%	96.2%	Microphthalmia, syndromic 2, 300166

BCS1L	160.0	100.0%	100.0%	Leigh syndrome, 256000 GRACILE syndrome, 603358 Bjornstad syndrome, 262000 Mitochondrial complex III deficiency, nuclear type 1, 124000
BLM	111.3	99.9%	98.1%	Bloom syndrome, 210900
BLOC1S3	79.6	100.0%	100.0%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	103.0	99.3%	92.1%	?Hermansky-pudlak syndrome 9, 614171
BMS1	79.8	66.8%	65.9%	?Aplasia cutis congenita, nonsyndromic, 107600
BRAF	71.0	91.7%	79.4%	Noonan syndrome 7, 613706 Cardiofaciocutaneous syndrome, 115150 Adenocarcinoma of lung, somatic, 211980 LEOPARD syndrome 3, 613707 Nonsmall cell lung cancer, somatic, 0 Melanoma, malignant, somatic, 0 Colorectal cancer, somatic, 0
BRIP1	122.2	99.9%	98.6%	Fanconi anemia, complementation group J, 609054
BSCL2	112.9	100.0%	99.9%	Lipodystrophy, congenital generalized, type 2, 269700 Silver spastic paraplegia syndrome, 270685 Neuropathy, distal hereditary motor, type VA, 600794 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	135.6	100.0%	99.8%	Biotinidase deficiency, 253260
C1QA	222.4	100.0%	100.0%	C1q deficiency, 613652
C1QB	178.7	100.0%	99.9%	C1q deficiency, 613652
C1QC	209.9	100.0%	100.0%	C1q deficiency, 613652
C2CD3	121.4	95.8%	95.3%	Orofaciodigital syndrome XIV, 615948
C4orf26	207.0	100.0%	100.0%	Amelogenesis imperfecta, type IIA4, 614832
CA2	141.8	100.0%	99.9%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAPN12	116.7	98.8%	94.9%	No OMIM Disease ID
CARD14	136.0	100.0%	99.5%	Pityriasis rubra pilaris, 173200 Psoriasis 2, 602723
CARD9	152.2	100.0%	99.9%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	153.2	99.5%	98.0%	Immunodeficiency 58, 618131
CASP14	89.0	100.0%	99.9%	Ichthyosis, congenital, autosomal recessive 12, 617320
CAST	112.2	99.5%	97.0%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAV1	200.0	100.0%	100.0%	Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721 ?Lipodystrophy, congenital generalized, type 3, 612526

CAVIN1	200.3	100.0%	100.0%	Lipodystrophy, congenital generalized, type 4, 613327
CBL	131.1	97.4%	97.1%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBS	136.4	100.0%	99.3%	Homocystinuria, B6-responsive and nonresponsive types, 236200 Thrombosis, hyperhomocysteinemic, 236200
CCBE1	80.9	99.8%	98.6%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD151	135.4	100.0%	100.0%	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057
CDAN1	123.7	100.0%	99.8%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDH3	148.2	100.0%	99.9%	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 Hypotrichosis, congenital, with juvenile macular dystrophy, 601553
CDK4	102.8	100.0%	99.4%	No OMIM disease ID
CDKN2A	139.4	92.3%	92.3%	No OMIM disease ID
CDSN	141.6	100.0%	100.0%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CELSR1	194.3	99.2%	96.8%	No OMIM Disease ID
CERS3	93.3	100.0%	98.5%	Ichthyosis, congenital, autosomal recessive 9, 615023
CHKB	126.8	100.0%	100.0%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	180.8	100.0%	99.5%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHSY1	134.8	99.8%	99.0%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	124.5	99.7%	98.1%	Cocoon syndrome, 613630
CIB1	134.0	99.8%	97.7%	Epidermodysplasia verruciformis 3, 618267
CKAP2L	154.2	99.9%	98.9%	Filippi syndrome, 272440
CLDN1	129.7	100.0%	100.0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	144.8	100.0%	99.9%	HELIX syndrome, 617671
CNNM4	177.3	100.0%	99.9%	Jalili syndrome, 217080
COL14A1	124.6	99.7%	98.3%	No OMIM Disease ID
COL17A1	110.6	99.6%	97.7%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, localisata variant, 226650
COL1A2	96.3	98.6%	94.6%	Ehlers-Danlos syndrome, cardiac valvular type, 225320 Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821 Osteogenesis imperfecta, type II, 166210 Osteogenesis imperfecta, type IV, 166220 Osteogenesis imperfecta, type III, 259420
COL3A1	99.6	99.4%	97.0%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	146.8	100.0%	99.4%	Ehlers-Danlos syndrome, classic type, 1, 130000

COL5A2	100.3	99.9%	99.3%	Ehlers-Danlos syndrome, classic type, 2, 130010
COL7A1	153.9	99.9%	99.2%	EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, Bart type, 132000 Transient bullous of the newborn, 131705 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 Epidermolysis bullosa, pretibial, 131850 EBD, localisata variant, 0
COX4I2	125.6	100.0%	99.9%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	39.4	62.3%	31.6%	Linear skin defects with multiple congenital anomalies 2, 300887
CPOX	141.1	99.7%	97.8%	Harderoporphyria, 121300 Coproporphyria, 121300
CST6	126.0	100.0%	98.8%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	113.5	100.0%	99.4%	Peeling skin syndrome 4, 607936
CTC1	113.5	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTSA	146.1	100.0%	100.0%	Galactosialidosis, 256540
CTSB	128.0	100.0%	100.0%	No OMIM disease ID
CTSC	119.4	100.0%	100.0%	Periodontitis 1, juvenile, 170650 Papillon-Lefevre syndrome, 245000 Haim-Munk syndrome, 245010
CXCR4	127.9	100.0%	99.9%	WHIM syndrome, 193670 Myelokathexis, isolated, 0
CYLD	110.8	99.8%	97.6%	Cylindromatosis, familial, 132700 Brooke-Spiegler syndrome, 605041 Trichoepithelioma, multiple familial, 1, 601606
CYP26C1	155.8	100.0%	100.0%	Focal facial dermal dysplasia 4, 614974
CYP4F22	124.4	100.0%	99.2%	Ichthyosis, congenital, autosomal recessive 5, 604777
DCAF17	87.5	100.0%	99.2%	Woodhouse-Sakati syndrome, 241080
DCLRE1C	139.2	99.9%	98.0%	Omenn syndrome, 603554 Severe combined immunodeficiency, Athabascan type, 602450
DDB2	154.2	100.0%	98.9%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DHCR7	158.7	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DKC1	93.9	99.7%	98.0%	Dyskeratosis congenita, X-linked, 305000
DLX3	165.9	100.0%	99.8%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510

DLX5	159.7	100.0%	99.8%	?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DOCK6	132.3	99.6%	98.9%	Adams-Oliver syndrome 2, 614219
DOCK8	115.2	100.0%	99.7%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOLK	171.4	100.0%	100.0%	Congenital disorder of glycosylation, type Im, 610768
DSC2	120.1	99.7%	97.3%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	93.8	99.2%	97.2%	?Hypotrichosis and recurrent skin vesicles, 613102
DSE	95.2	99.8%	98.0%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	131.8	99.3%	97.4%	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG3	135.0	100.0%	99.4%	No OMIM Disease ID
DSG4	161.1	99.9%	99.0%	Hypotrichosis 6, 607903
DSP	148.0	100.0%	99.6%	Keratosis palmoplantaris striata II, 612908 Epidermolysis bullosa, lethal acantholytic, 609638 Skin fragility-woolly hair syndrome, 607655 Arrhythmogenic right ventricular dysplasia 8, 607450 Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821
DSPP	83.9	98.8%	95.5%	Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
DST	145.3	99.9%	99.1%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex, autosomal recessive 2, 615425
DTNBP1	118.8	99.8%	97.5%	Hermansky-Pudlak syndrome 7, 614076
DUSP6	184.5	100.0%	100.0%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EBP	68.9	99.8%	96.3%	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
ECM1	169.4	100.0%	99.4%	Urbach-Wiethe disease, 247100
EDA	108.8	96.6%	88.1%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	135.7	100.0%	100.0%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	93.4	99.8%	97.5%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDN3	147.0	100.0%	100.0%	Waardenburg syndrome, type 4B, 613265 Central hypoventilation syndrome, congenital, 209880

EDNRA	153.6	100.0%	99.9%	Mandibulofacial dysostosis with alopecia, 616367
EDNRB	126.2	96.4%	91.5%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EFEMP2	141.3	100.0%	100.0%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	122.4	100.0%	100.0%	Craniofrontonasal dysplasia, 304110
EIF2AK3	134.0	99.5%	96.7%	Wolcott-Rallison syndrome, 226980
ELN	113.2	100.0%	99.6%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	90.9	99.7%	96.6%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	103.3	100.0%	99.6%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENAM	144.6	100.0%	100.0%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENG	137.2	100.0%	99.4%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	128.6	97.9%	92.4%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Cole disease, 615522 Arterial calcification, generalized, of infancy, 1, 208000
EPG5	111.4	99.5%	98.3%	Vici syndrome, 242840
EPS8L3	114.2	99.6%	97.6%	No OMIM Disease ID
ERCC2	139.5	100.0%	99.9%	Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756 Xeroderma pigmentosum, group D, 278730
ERCC3	95.9	99.9%	98.7%	Xeroderma pigmentosum, group B, 610651 Trichothiodystrophy 2, photosensitive, 616390
ERCC4	136.9	100.0%	99.6%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 Fanconi anemia, complementation group Q, 615272 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760
ERCC5	130.9	100.0%	99.4%	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570
ERCC6	161.8	100.0%	100.0%	Cerebrooculofacioskeletal syndrome 1, 214150 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946 UV-sensitive syndrome 1, 600630 De Sanctis-Cacchione syndrome, 278800

ERCC8	79.9	99.0%	89.3%	Cockayne syndrome, type A, 216400 UV-sensitive syndrome 2, 614621
EVC	113.0	96.8%	92.1%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	115.9	99.6%	97.1%	Weyers acrofacial dysostosis, 193530 Ellis-van Creveld syndrome, 225500
EXPH5	167.7	100.0%	100.0%	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028
FAM111B	155.6	99.9%	99.5%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM20A	122.6	99.9%	99.2%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	165.1	100.0%	100.0%	Raine syndrome, 259775
FAM83G	177.1	100.0%	100.0%	No OMIM Disease ID
FAM83H	139.0	100.0%	100.0%	Amelogenesis imperfecta, type IIIA, 130900
FANCA	118.3	100.0%	99.2%	Fanconi anemia, complementation group A, 227650
FANCB	72.8	98.6%	93.0%	Fanconi anemia, complementation group B, 300514
FANCC	104.4	100.0%	99.3%	Fanconi anemia, complementation group C, 227645
FANCD2	116.2	99.2%	96.5%	Fanconi anemia, complementation group D2, 227646
FANCE	127.9	98.0%	91.8%	Fanconi anemia, complementation group E, 600901
FANCF	269.1	100.0%	100.0%	Fanconi anemia, complementation group F, 603467
FANCG	149.5	100.0%	100.0%	Fanconi anemia, complementation group G, 614082
FANCI	136.0	100.0%	98.8%	Fanconi anemia, complementation group I, 609053
FANCL	102.9	99.8%	97.9%	Fanconi anemia, complementation group L, 614083
FANCM	99.4	99.5%	96.6%	Spermatogenic failure 28, 618086 ?Premature ovarian failure 15, 618096
FAT4	195.5	100.0%	100.0%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN5	103.0	91.8%	91.8%	Macular degeneration, age-related, 3, 608895 ?Cutis laxa, autosomal dominant 2, 614434 Neuropathy, hereditary, with or without age-related macular degeneration, 608895 Cutis laxa, autosomal recessive, type IA, 219100
FDPS	64.3	98.1%	92.2%	Porokeratosis 9, multiple types, 616631
FECH	107.9	100.0%	99.6%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	93.0	99.6%	96.7%	Kindler syndrome, 173650
FGF10	118.7	100.0%	99.7%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	130.1	99.9%	98.7%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100

FGF3	160.1	100.0%	100.0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	167.5	100.0%	99.7%	Trichomegaly, 190330
FGF8	141.1	98.8%	90.5%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	131.6	100.0%	99.7%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	118.0	97.7%	97.1%	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FGFR3	157.1	100.0%	99.9%	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482

FH	126.0	95.9%	89.5%	Fumarase deficiency, 606812 Leiomyomatosis and renal cell cancer, 150800
FKBP10	170.1	99.8%	98.3%	Bruck syndrome 1, 259450 Osteogenesis imperfecta, type XI, 610968
FKBP14	77.6	100.0%	99.2%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLCN	165.1	100.0%	100.0%	Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700 Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500
FLG	158.7	100.0%	99.9%	Ichthyosis vulgaris, 146700
FLG2	374.4	100.0%	100.0%	Peeling skin syndrome 6, 618084
FLT4	177.8	99.2%	99.2%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100
FNIP1	154.2	100.0%	99.8%	No OMIM Disease ID
FOXC2	144.2	100.0%	100.0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	110.9	100.0%	100.0%	Bamforth-Lazarus syndrome, 241850
FOXN1	149.7	100.0%	99.8%	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	126.8	99.3%	96.0%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FREM1	112.5	99.8%	98.8%	Manitoba oculotrichoanal syndrome, 248450 Trigonocephaly 2, 614485 Bifid nose with or without anorectal and renal anomalies, 608980
FUCA1	135.9	100.0%	100.0%	Fucosidosis, 230000
FZD6	190.3	100.0%	100.0%	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157
GALNS	118.1	100.0%	99.4%	Mucopolysaccharidosis IVA, 253000
GALNT3	126.0	99.9%	98.8%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GAN	147.5	100.0%	99.6%	Giant axonal neuropathy-1, 256850
GATA2	128.7	100.0%	99.7%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GDF2	157.3	100.0%	100.0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF5	190.2	100.0%	100.0%	?Acromesomelic dysplasia, Hunter-Thompson type, 201250 Symphalangism, proximal, 1B, 615298 Brachydactyly, type A1, C, 615072 Chondrodysplasia, Grebe type, 200700 Brachydactyly, type A2, 112600 Du Pan syndrome, 228900 Brachydactyly, type C, 113100 Multiple synostoses syndrome 2, 610017

GGCX	105.2	100.0%	99.6%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GJA1	162.4	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJB2	151.0	100.0%	100.0%	Deafness, autosomal dominant 3A, 601544 Deafness, autosomal recessive 1A, 220290 Bart-Pumphrey syndrome, 149200 Vohwinkel syndrome, 124500 Keratoderma, palmoplantar, with deafness, 148350 Keratitis-ichthyosis-deafness syndrome, 148210 Hystrix-like ichthyosis with deafness, 602540
GJB3	245.5	100.0%	100.0%	Deafness, autosomal dominant 2B, 612644 Deafness, digenic, GJB2/GJB3, 220290 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, 0 Deafness, autosomal dominant, with peripheral neuropathy, 0
GJB4	270.0	100.0%	100.0%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	146.2	100.0%	100.0%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GJC2	59.7	97.7%	86.5%	Spastic paraplegia 44, autosomal recessive, 613206 Lymphatic malformation 3, 613480 Leukodystrophy, hypomyelinating, 2, 608804
GLA	74.4	99.4%	95.8%	Fabry disease, 301500 Fabry disease, cardiac variant, 301500
GLB1	87.4	99.5%	95.2%	GM1-gangliosidosis, type III, 230650 GM1-gangliosidosis, type I, 230500 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLMN	66.0	99.5%	93.5%	Glomuvenous malformations, 138000
GMPPA	158.4	100.0%	100.0%	Alacrima, achalasia, and mental retardation syndrome, 615510

GNA11	176.2	100.0%	99.8%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GNA14	133.5	100.0%	100.0%	No OMIM Disease ID
GNAQ	53.7	82.9%	66.0%	Sturge-Weber syndrome, somatic, mosaic, 185300 Capillary malformations, congenital, 1, somatic, mosaic, 163000
GNAS	241.4	100.0%	100.0%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ib, 603233 Pseudopseudohypoparathyroidism, 612463 McCune-Albright syndrome, somatic, mosaic, 174800 Osseous heteroplasia, progressive, 166350 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ia, 103580
GORAB	168.3	100.0%	99.1%	Geroderma osteodysplasticum, 231070
GPNMB	154.7	100.0%	100.0%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR143	60.8	91.8%	81.6%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GRHL2	119.8	100.0%	100.0%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
GRHL3	141.9	100.0%	99.9%	Van der Woude syndrome 2, 606713
GSN	123.5	95.6%	93.8%	Amyloidosis, Finnish type, 105120
GTF2E2	81.7	100.0%	99.0%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	82.0	99.8%	97.4%	Trichothiodystrophy 3, photosensitive, 616395
HCCS	90.4	99.6%	96.6%	Linear skin defects with multiple congenital anomalies 1, 309801
HDAC8	110.1	100.0%	99.6%	Cornelia de Lange syndrome 5, 300882
HERC2	99.7	80.6%	76.7%	Mental retardation, autosomal recessive 38, 615516
HLCS	148.0	100.0%	100.0%	Holocarboxylase synthetase deficiency, 253270
HMBS	102.8	100.0%	99.0%	Porphyria, acute intermittent, 176000 Porphyria, acute intermittent, nonerythroid variant, 176000
HMGB3	40.2	84.0%	66.6%	?Microphthalmia, syndromic 13, 300915
HOXC13	197.1	100.0%	100.0%	Ectodermal dysplasia 9, hair/nail type, 614931
HPGD	87.8	100.0%	99.7%	Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HPS1	125.7	100.0%	100.0%	Hermansky-Pudlak syndrome 1, 203300
HPS3	133.9	99.9%	98.2%	Hermansky-Pudlak syndrome 3, 614072

HPS4	135.2	100.0%	100.0%	Hermansky-Pudlak syndrome 4, 614073
HPS5	122.5	99.9%	98.9%	Hermansky-Pudlak syndrome 5, 614074
HPS6	183.5	100.0%	99.2%	Hermansky-Pudlak syndrome 6, 614075
HR	133.4	99.7%	98.0%	Hypotrichosis 4, 146550 Alopecia universalis, 203655 Atrichia with papular lesions, 209500
HRAS	196.0	100.0%	100.0%	Nevus sebaceous or woolly hair nevus, somatic, 162900 Congenital myopathy with excess of muscle spindles, 218040 Bladder cancer, somatic, 109800 Thyroid carcinoma, follicular, somatic, 188470 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Spitz nevus or nevus spilus, somatic, 137550 Costello syndrome, 218040
HTRA1	95.8	98.9%	91.1%	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 CARASIL syndrome, 600142
HYAL1	121.3	100.0%	100.0%	?Mucopolysaccharidosis type IX, 601492
IDUA	169.2	99.3%	96.4%	Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014 Mucopolysaccharidosis Is, 607016
IFT122	126.6	100.0%	99.6%	Cranioectodermal dysplasia 1, 218330
IFT43	119.5	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IKBKG	64.7	90.1%	80.2%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291 Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IL17RA	167.0	100.0%	100.0%	Immunodeficiency 51, 613953
IL17RD	143.4	99.9%	99.3%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RN	145.3	100.0%	99.9%	Interleukin 1 receptor antagonist deficiency, 612852
IL31RA	113.8	99.9%	99.5%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	102.7	100.0%	99.9%	Psoriasis 14, pustular, 614204
INSR	123.5	99.4%	96.1%	Hyperinsulinemic hypoglycemia, familial, 5, 609968 Rabson-Mendenhall syndrome, 262190

				Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Leprechaunism, 246200
IRF4	209.2	100.0%	100.0%	No OMIM Disease ID
IRF6	94.5	99.4%	95.8%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
ITGA3	162.5	99.4%	98.1%	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748
ITGA6	144.0	99.9%	99.3%	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730
ITGB4	166.0	99.4%	98.0%	Epidermolysis bullosa of hands and feet, 131800 Epidermolysis bullosa, junctional, with pyloric atresia, 226730 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
ITGB6	129.5	96.3%	95.1%	Amelogenesis imperfecta, type IH, 616221
JUP	137.0	100.0%	99.9%	Arrhythmogenic right ventricular dysplasia 12, 611528 Naxos disease, 601214
KANK2	180.1	100.0%	100.0%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KAT6B	162.3	99.8%	99.2%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNH1	159.4	98.7%	98.4%	Temple-Baraitser syndrome, 611816 Zimmermann-Laband syndrome 1, 135500
KCNK9	189.3	100.0%	100.0%	Birk-Barel mental retardation dysmorphism syndrome, 612292
KDF1	120.4	100.0%	100.0%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDSR	165.5	100.0%	99.5%	Erythrokeratodermia variabilis et progressiva 4, 617526
KIF11	89.7	97.6%	94.9%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIT	137.7	100.0%	99.7%	Gastrointestinal stromal tumor, familial, 606764 Mastocytosis, cutaneous, 154800 Germ cell tumors, somatic, 273300 Leukemia, acute myeloid, somatic, 601626 Mastocytosis, systemic, somatic, 154800 Piebaldism, 172800
KITLG	82.5	99.8%	96.5%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
KLHL24	174.7	100.0%	100.0%	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294
KLK4	176.5	100.0%	99.8%	Amelogenesis imperfecta, type IIA1, 204700
KLLN	172.4	100.0%	100.0%	Cowden syndrome 4, 615107
KMT2D	150.7	100.0%	99.9%	Kabuki syndrome 1, 147920
KRAS	64.0	99.8%	96.8%	Leukemia, acute myeloid, 601626 Oculoectodermal syndrome, somatic, 600268

				Breast cancer, somatic, 114480 RAS-associated autoimmune leukoproliferative disorder, 614470 Cardiofaciocutaneous syndrome 2, 615278 Arteriovenous malformation of the brain, somatic, 108010 Bladder cancer, somatic, 109800 Pancreatic carcinoma, somatic, 260350 Lung cancer, somatic, 211980 Gastric cancer, somatic, 137215 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Noonan syndrome 3, 609942
KRT1	104.0	100.0%	99.0%	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Palmoplantar keratoderma, nonepidermolytic, 600962 Ichthyosis histrix, Curth-Macklin type, 146590 Epidermolytic hyperkeratosis, 113800
KRT10	137.0	100.0%	99.0%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT13	132.5	100.0%	98.9%	White sponge nevus 2, 615785
KRT14	48.3	91.4%	83.3%	Naegeli-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Dermatopathia pigmentosa reticularis, 125595 Epidermolysis bullosa simplex, Koebner type, 131900 Epidermolysis bullosa simplex, recessive 1, 601001 Epidermolysis bullosa simplex, Weber-Cockayne type, 131800
KRT16	38.7	78.4%	57.3%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200
KRT17	19.4	49.9%	31.2%	Pachyonychia congenita 2, 167210 Steatocystoma multiplex, 184500
KRT2	146.2	100.0%	99.7%	Ichthyosis bullosa of Siemens, 146800
KRT4	130.8	100.0%	99.7%	White sponge nevus 1, 193900
KRT5	121.9	100.0%	99.9%	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 Epidermolysis bullosa simplex-MCR, 609352 Epidermolysis bullosa simplex-MP, 131960 Dowling-Degos disease 1, 179850 Epidermolysis bullosa simplex, Koebner type, 131900

				Epidermolysis bullosa simplex, Dowling-Meara type, 131760 Epidermolysis bullosa simplex, recessive 1, 601001
KRT6A	132.6	98.6%	92.0%	Pachyonychia congenita 3, 615726
KRT6B	132.9	99.7%	95.2%	Pachyonychia congenita 4, 615728
KRT6C	115.2	90.0%	81.1%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	157.5	100.0%	100.0%	?Hypotrichosis 13, 615896
KRT74	154.9	100.0%	99.7%	?Ectodermal dysplasia 7, hair/nail type, 614929 Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981
KRT75	133.3	100.0%	100.0%	No OMIM Disease ID
KRT81	91.4	100.0%	99.3%	Monilethrix, 158000
KRT83	74.3	99.3%	93.0%	Erythrokeratodermia variabilis et progressiva 5, 617756 Monilethrix, 158000
KRT85	112.7	99.1%	96.1%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	95.7	100.0%	99.4%	Monilethrix, 158000
KRT9	71.8	99.7%	97.3%	Palmoplantar keratoderma, epidermolytic, 144200
LAMA3	129.8	100.0%	99.8%	Epidermolysis bullosa, junctional, Herlitz type, 226700 Laryngoonychocutaneous syndrome, 245660 Epidermolysis bullosa, generalized atrophic benign, 226650
LAMB3	125.6	100.0%	99.5%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LAMC2	105.4	99.8%	98.4%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700
LAMTOR2	186.6	100.0%	100.0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LDHA	57.3	97.7%	89.0%	Glycogen storage disease XI, 612933
LDLRAP1	162.0	100.0%	99.9%	Hypercholesterolemia, familial, 4, 603813
LEMD3	127.5	99.8%	97.8%	Osteopoikilosis with or without melorheostosis, 166700 Buschke-Ollendorff syndrome, 166700
LIPH	120.7	100.0%	99.7%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	111.3	100.0%	98.8%	Ichthyosis, congenital, autosomal recessive 8, 613943
LMBRD1	95.5	97.1%	91.2%	Methylmalonic aciduria and homocystinuria, cblF type, 277380
LMNA	118.2	98.3%	93.2%	Muscular dystrophy, congenital, 613205 Lipodystrophy, familial partial, type 2, 151660 Charcot-Marie-Tooth disease, type 2B1, 605588 Cardiomyopathy, dilated, 1A, 115200

				Heart-hand syndrome, Slovenian type, 610140 Hutchinson-Gilford progeria, 176670 Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia, 248370 Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 Malouf syndrome, 212112
LMX1B	163.9	100.0%	99.3%	Nail-patella syndrome, 161200
LONP1	164.8	100.0%	100.0%	CODAS syndrome, 600373
LOR	46.5	100.0%	97.7%	Vohwinkel syndrome with ichthyosis, 604117
LPAR6	101.0	99.8%	97.7%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN2	101.1	100.0%	99.7%	Majeed syndrome, 609628
LRMDA	119.8	99.4%	98.0%	Albinism, oculocutaneous, type VII, 615179
LSS	138.5	100.0%	99.9%	Cataract 44, 616509 Hypotrichosis 14, 618275
LTBP3	166.1	100.0%	100.0%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	167.8	100.0%	99.7%	Cutis laxa, autosomal recessive, type IC, 613177
LYST	135.6	99.3%	97.1%	Chediak-Higashi syndrome, 214500
LYZ	142.4	100.0%	100.0%	Amyloidosis, renal, 105200
MAP2K1	96.7	99.6%	97.1%	Cardiofaciocutaneous syndrome 3, 615279
MAP2K2	139.8	99.3%	95.6%	Cardiofaciocutaneous syndrome 4, 615280
MBTPS2	109.0	99.9%	98.4%	Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918 IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800
MED12	89.4	99.6%	96.5%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MEFV	136.9	99.0%	97.0%	Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MGP	134.2	98.6%	93.2%	Keutel syndrome, 245150
MITF	145.6	100.0%	100.0%	COMMAD syndrome, 617306 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500

MLH1	142.9	100.0%	99.6%	Mismatch repair cancer syndrome, 276300 Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 2, 609310
MLPH	104.1	99.8%	97.9%	Griscelli syndrome, type 3, 609227
MMACHC	214.4	100.0%	100.0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP2	162.9	100.0%	100.0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	91.0	99.9%	97.9%	Amelogenesis imperfecta, type IIA2, 612529
MPLKIP	106.6	100.0%	99.9%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRE11	48.1	97.7%	83.5%	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	110.0	99.5%	95.6%	Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 1, 120435
MSX1	164.1	100.0%	99.4%	Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MTOR	116.6	100.0%	99.2%	Smith-Kingsmore syndrome, 616638 Focal cortical dysplasia, type II, somatic, 607341
MUTYH	165.8	100.0%	100.0%	Gastric cancer, somatic, 613659 Adenomas, multiple colorectal, 608456 Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600
MVD	126.6	99.9%	99.0%	Porokeratosis 7, multiple types, 614714
MVK	130.3	90.5%	90.4%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYH8	119.4	100.0%	99.6%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYO5A	109.9	99.7%	98.5%	Griscelli syndrome, type 1, 214450
NAA10	112.8	100.0%	99.4%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NAGA	131.4	100.0%	100.0%	Kanzaki disease, 609242 Schindler disease, type I, 609241 Schindler disease, type III, 609241
NBAS	138.4	99.9%	99.2%	Infantile liver failure syndrome 2, 616483 Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800
NCSTN	97.9	100.0%	99.7%	Acne inversa, familial, 1, 142690
NDUFB11	110.0	99.2%	96.2%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021

NECTIN1	146.4	100.0%	100.0%	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NECTIN4	132.0	100.0%	100.0%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK11	117.0	99.9%	98.0%	No OMIM Disease ID
NEK9	123.4	99.9%	98.9%	Lethal congenital contracture syndrome 10, 617022 Nevus comedonicus, somatic, 617025 ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262
NF1	105.8	92.5%	89.3%	Watson syndrome, 193520 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210
NFKBIA	148.5	96.7%	90.7%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHP2	135.0	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIPAL4	137.3	100.0%	99.7%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	123.0	98.9%	96.7%	Cornelia de Lange syndrome 1, 122470
NLRP1	126.8	99.5%	97.9%	Palmoplantar carcinoma, multiple self-healing, 615225 Autoinflammation with arthritis and dyskeratosis, 617388
NLRP3	146.0	100.0%	99.9%	Familial cold inflammatory syndrome 1, 120100 Muckle-Wells syndrome, 191900 CINCA syndrome, 607115 Deafness, autosomal dominant 34, with or without inflammation, 617772 Keratoendothelitis fugax hereditaria, 148200
NME1	80.4	100.0%	99.9%	No OMIM Disease ID
NOD2	136.5	100.0%	100.0%	Blau syndrome, 186580
NOP10	124.6	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH1	158.4	99.8%	99.2%	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
NRAS	145.3	100.0%	100.0%	Epidermal nevus, somatic, 162900 Melanocytic nevus syndrome, congenital, somatic, 137550 Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 Colorectal cancer, somatic, 114500 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 Thyroid carcinoma, follicular, somatic, 188470 Neurocutaneous melanosis, somatic, 249400 Noonan syndrome 6, 613224
NSD1	152.6	100.0%	99.8%	Sotos syndrome 1, 117550

NSDHL	133.7	99.9%	98.2%	CHILD syndrome, 308050 CK syndrome, 300831
OCA2	123.6	99.8%	97.4%	Albinism, oculocutaneous, type II, 203200 Albinism, brown oculocutaneous, 203200
ODAM	139.1	100.0%	97.7%	No OMIM Disease ID
OFD1	52.3	85.5%	70.0%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OSMR	131.5	100.0%	99.6%	Amyloidosis, primary localized cutaneous, 1, 105250
PADI3	147.3	100.0%	100.0%	Uncombable hair syndrome, 191480
PAH	128.9	100.0%	100.0%	Phenylketonuria, 261600
PALB2	146.3	100.0%	99.9%	Fanconi anemia, complementation group N, 610832
PAX3	116.1	100.0%	99.9%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX9	262.1	99.8%	99.6%	Tooth agenesis, selective, 3, 604625
PCNA	93.9	100.0%	98.7%	?Ataxia-telangiectasia-like disorder 2, 615919
PDGFB	127.3	100.0%	100.0%	Dermatofibrosarcoma protuberans, 607907 Basal ganglia calcification, idiopathic, 5, 615483 Meningioma, SIS-related, 607174
PDGFRB	138.0	99.8%	98.2%	Kosaki overgrowth syndrome, 616592 Basal ganglia calcification, idiopathic, 4, 615007 Premature aging syndrome, Penttinen type, 601812 Myofibromatosis, infantile, 1, 228550
PEPD	126.5	100.0%	99.9%	Prolidase deficiency, 170100
PERP	180.7	100.0%	100.0%	No OMIM Disease ID
PEX7	108.8	91.3%	91.0%	Peroxisome biogenesis disorder 9B, 614879 Rhizomelic chondrodysplasia punctata, type 1, 215100
PHEX	107.9	99.7%	98.2%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	116.2	100.0%	99.6%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHYH	75.9	100.0%	97.9%	Refsum disease, 266500
PIEZO1	162.6	100.0%	99.7%	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380

PIGA	72.9	93.0%	83.4%	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Paroxysmal nocturnal hemoglobinuria, somatic, 300818
PIGN	103.7	93.6%	90.1%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	129.3	100.0%	100.0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	122.5	100.0%	99.8%	Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Cowden syndrome 5, 615108 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Macrodactyly, somatic, 155500 Keratosis, seborrheic, somatic, 182000 Gastric cancer, somatic, 613659 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Nevus, epidermal, somatic, 162900 CLOVE syndrome, somatic, 612918 Nonsmall cell lung cancer, somatic, 211980
PITX2	186.2	100.0%	99.6%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PKP1	130.1	99.9%	98.7%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLCD1	127.9	100.0%	99.6%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCG2	110.7	100.0%	99.5%	Familial cold autoinflammatory syndrome 3, 614468 Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878
PLEC	165.0	100.0%	100.0%	Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 Epidermolysis bullosa simplex with pyloric atresia, 612138 Epidermolysis bullosa simplex with muscular dystrophy, 226670 ?Epidermolysis bullosa simplex with nail dystrophy, 616487 Epidermolysis bullosa simplex, Ogna type, 131950
PLG	95.7	87.8%	86.7%	Dysplasminogenemia, 217090 Plasminogen deficiency, type I, 217090
PLIN1	107.2	100.0%	99.8%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	141.5	99.9%	97.9%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD3	120.2	100.0%	99.8%	Lysyl hydroxylase 3 deficiency, 612394
PMS2	96.2	83.5%	81.4%	Mismatch repair cancer syndrome, 276300 Colorectal cancer, hereditary nonpolyposis, type 4, 614337
PMVK	130.2	100.0%	99.9%	Porokeratosis 1, multiple types, 175800
PNPLA1	176.0	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 10, 615024

PNPLA2	159.9	100.0%	99.9%	Neutral lipid storage disease with myopathy, 610717
POC1A	120.3	100.0%	100.0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POFUT1	146.2	100.0%	99.2%	Dowling-Degos disease 2, 615327
POGLUT1	100.2	100.0%	99.1%	?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 Dowling-Degos disease 4, 615696
POLD1	137.8	98.3%	94.9%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLH	121.6	100.0%	99.2%	Xeroderma pigmentosum, variant type, 278750
POLR1C	103.3	99.3%	95.4%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	186.7	91.6%	91.6%	Treacher Collins syndrome 2, 613717
POLR3A	119.8	100.0%	99.9%	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 Wiedemann-Rautenstrauch syndrome, 264090
POLR3B	132.0	99.9%	98.3%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381
POMC	165.9	100.0%	100.0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMP	122.5	99.9%	95.8%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
PORCN	119.6	99.9%	98.9%	Focal dermal hypoplasia, 305600
POT1	93.9	100.0%	98.7%	No OMIM disease ID
PPOX	101.7	99.9%	97.6%	Porphyria variegata, 176200
PQBP1	173.3	100.0%	100.0%	Renpenning syndrome, 309500
PRKAR1A	80.5	98.1%	92.8%	Myxoma, intracardiac, 255960 Carney complex, type 1, 160980 Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Adrenocortical tumor, somatic, 0
PSEN1	132.4	100.0%	100.0%	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 ?Acne inversa, familial, 3, 613737 Alzheimer disease, type 3, 607822 Dementia, frontotemporal, 600274 Pick disease, 172700 Cardiomyopathy, dilated, 1U, 613694
PSENEN	98.3	100.0%	100.0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB8	119.3	100.0%	99.3%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSTPIP1	114.3	99.9%	99.3%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416

PTCH1	117.3	100.0%	99.2%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	131.5	99.9%	98.8%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
PTDSS1	116.2	100.0%	100.0%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	125.3	99.7%	95.5%	Prostate cancer, somatic, 176807 Cowden syndrome 1, 158350 Lhermitte-Duclos syndrome, 158350 Macrocephaly/autism syndrome, 605309
PTHLH	138.1	99.1%	91.2%	Brachydactyly, type E2, 613382
PTPN11	80.5	98.8%	91.3%	LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Noonan syndrome 1, 163950 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN14	170.9	99.3%	96.4%	Choanal atresia and lymphedema, 613611
PTPRF	170.4	100.0%	99.9%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PYCR1	105.0	100.0%	99.0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	102.4	100.0%	99.8%	Carpenter syndrome, 201000
RAB27A	123.3	100.0%	99.6%	Griscelli syndrome, type 2, 607624
RAD21	80.8	98.0%	93.5%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RAD50	100.6	96.4%	89.5%	Nijmegen breakage syndrome-like disorder, 613078
RAF1	111.1	100.0%	99.9%	LEOPARD syndrome 2, 611554 Noonan syndrome 5, 611553 Cardiomyopathy, dilated, 1NN, 615916
RAG1	158.7	100.0%	100.0%	Omenn syndrome, 603554 Severe combined immunodeficiency, B cell-negative, 601457 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
RAG2	188.7	100.0%	100.0%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAI1	216.5	100.0%	100.0%	Smith-Magenis syndrome, 182290

RBBP8	117.9	100.0%	99.3%	Jawad syndrome, 251255 Seckel syndrome 2, 606744 Pancreatic carcinoma, somatic, 0
RBM28	132.1	100.0%	100.0%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBP4	149.6	99.8%	96.8%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	72.1	95.8%	87.2%	Adams-Oliver syndrome 3, 614814
RECQL4	181.4	100.0%	100.0%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
RHBDF2	117.3	99.9%	99.0%	Tylosis with esophageal cancer, 148500
RHOA	78.1	81.6%	80.7%	No OMIM Disease ID
RIN2	129.3	100.0%	99.7%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	189.1	100.0%	100.0%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RMRP	NC	NC	NC	Anauxetic dysplasia 1, 607095 Cartilage-hair hypoplasia, 250250 Metaphyseal dysplasia without hypotrichosis, 250460
RNASEH2A	143.0	100.0%	100.0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	98.0	99.8%	96.3%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	314.2	100.0%	100.0%	Aicardi-Goutieres syndrome 3, 610329
RNU4ATAC	NC	NC	NC	Microcephalic osteodysplastic primordial dwarfism, type I, 210710 Roifman syndrome, 616651
ROGDI	141.6	100.0%	99.9%	Kohlschutter-Tonz syndrome, 226750
RPL21	54.3	84.1%	62.5%	Hypotrichosis 12, 615885
RSPO1	115.6	100.0%	100.0%	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
RSPO4	161.1	100.0%	100.0%	Anonychia congenita, 206800
RTEL1	145.6	99.8%	98.2%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
RUNX2	109.5	73.6%	72.3%	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600
SAMD9	161.7	100.0%	100.0%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455

SAMHD1	135.4	100.0%	98.7%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SART3	114.0	99.8%	98.1%	No OMIM Disease ID
SASH1	163.9	99.4%	98.1%	?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 Dyschromatosis universalis hereditaria 1, 127500
SAT1	129.2	100.0%	99.1%	No OMIM Disease ID
SATB2	115.6	99.9%	98.3%	Glass syndrome, 612313
SCN10A	141.5	100.0%	99.5%	Episodic pain syndrome, familial, 2, 615551
SCN11A	125.5	99.3%	97.2%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	128.2	99.0%	97.6%	Small fiber neuropathy, 133020 HSAN2D, autosomal recessive, 243000 Paroxysmal extreme pain disorder, 167400 Epilepsy, generalized, with febrile seizures plus, type 7, 613863 Insensitivity to pain, congenital, 243000 Erythermalgia, primary, 133020 Febrile seizures, familial, 3B, 613863
SDR9C7	183.1	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23B	132.1	99.7%	98.2%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SERPINB7	125.1	100.0%	99.9%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	129.3	95.0%	95.0%	Peeling skin syndrome 5, 617115
SERPING1	101.0	99.6%	97.5%	Angioedema, hereditary, types I and II, 106100 Complement component 4, partial deficiency of, 120790
SERPINH1	216.7	100.0%	99.9%	Osteogenesis imperfecta, type X, 613848
SGPL1	133.4	100.0%	100.0%	Nephrotic syndrome, type 14, 617575
SHOC2	136.8	100.0%	99.4%	Noonan syndrome-like with loose anagen hair, 607721
SKI	149.5	100.0%	99.7%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	150.1	100.0%	99.9%	Trichohepatoenteric syndrome 2, 614602
SLC17A9	155.8	95.8%	95.4%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC24A4	109.6	100.0%	99.9%	Amelogenesis imperfecta, type IIA5, 615887
SLC24A5	102.2	100.0%	99.6%	Albinism, oculocutaneous, type VI, 113750
SLC26A2	203.7	100.0%	99.9%	De la Chapelle dysplasia, 256050 Atelosteogenesis, type II, 256050 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Diastrophic dysplasia, 222600

				Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900
SLC27A4	167.0	100.0%	100.0%	Ichthyosis prematurity syndrome, 608649
SLC29A3	190.1	100.0%	99.7%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A10	167.7	98.0%	97.6%	Arterial tortuosity syndrome, 208050
SLC39A13	158.7	100.0%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A4	130.4	100.0%	99.7%	Acrodermatitis enteropathica, 201100
SLC45A2	119.4	100.0%	99.8%	Albinism, oculocutaneous, type IV, 606574
SLC4A4	114.4	99.8%	97.9%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A19	139.1	100.0%	100.0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC7A7	110.7	100.0%	99.8%	Lysinuric protein intolerance, 222700
SLCO2A1	104.4	100.0%	98.7%	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLURP1	109.0	100.0%	99.6%	Meleda disease, 248300
SLX4	136.8	100.0%	99.9%	Fanconi anemia, complementation group P, 613951
SMAD3	138.0	100.0%	100.0%	Loeys-Dietz syndrome 3, 613795
SMARCA2	109.3	97.3%	96.3%	Nicolaides-Baraitser syndrome, 601358
SMARCA4	163.9	100.0%	99.6%	Coffin-Siris syndrome 4, 614609
SMARCAD1	89.5	99.6%	95.8%	Huriez syndrome, 181600 Basan syndrome, 129200 Adermatoglyphia, 136000
SMARCAL1	119.6	100.0%	99.8%	Schimke immunoosseous dysplasia, 242900
SMARCB1	192.9	100.0%	100.0%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMO	154.2	100.0%	99.5%	Curry-Jones syndrome, somatic mosaic, 601707 Basal cell carcinoma, somatic, 605462
SMOC2	93.7	76.9%	75.9%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	106.3	100.0%	99.1%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	182.5	100.0%	100.0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNRPE	74.7	99.0%	87.8%	Hypotrichosis 11, 615059
SNX10	124.3	96.2%	95.5%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	100.6	99.7%	96.7%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300

SOX10	101.7	100.0%	99.8%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX18	59.1	97.4%	85.4%	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 Hypotrichosis-lymphedema-telangiectasia syndrome, 607823
SOX2	261.8	100.0%	100.0%	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SP7	164.6	100.0%	99.5%	Osteogenesis imperfecta, type XII, 613849
SPINK5	128.1	100.0%	99.2%	Netherton syndrome, 256500
SPINT2	71.9	99.8%	93.0%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPRED1	143.0	99.8%	98.1%	Legius syndrome, 611431
SPRY4	187.2	100.0%	100.0%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRD5A3	149.2	99.9%	98.5%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
ST14	170.8	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL5	104.4	89.3%	85.5%	Salt and pepper developmental regression syndrome, 609056
STAMBP	96.0	99.9%	97.6%	Microcephaly-capillary malformation syndrome, 614261
STAT3	106.9	100.0%	99.4%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	119.7	99.9%	98.8%	Leukemia, acute promyelocytic, somatic, 102578 Growth hormone insensitivity with immunodeficiency, 245590
STIM1	129.2	99.8%	97.1%	Myopathy, tubular aggregate, 1, 160565 Immunodeficiency 10, 612783 Stormorken syndrome, 185070
STK11	142.7	100.0%	100.0%	Testicular tumor, somatic, 273300 Peutz-Jeghers syndrome, 175200 Pancreatic cancer, somatic, 260350 Melanoma, malignant, somatic, 0
STS	81.1	99.6%	96.3%	Ichthyosis, X-linked, 308100
SUFU	141.7	100.0%	100.0%	Basal cell nevus syndrome, 109400 Medulloblastoma, desmoplastic, 155255 Joubert syndrome 32, 617757
SULT2B1	135.3	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	91.7	99.9%	97.6%	Multiple sulfatase deficiency, 272200
TALDO1	158.9	100.0%	99.8%	Transaldolase deficiency, 606003
TAP1	133.9	99.9%	97.7%	Bare lymphocyte syndrome, type I, 604571
TAP2	101.4	99.6%	98.7%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571

TAPBP	130.6	96.6%	96.6%	Bare lymphocyte syndrome, type I, 604571
TAT	119.7	100.0%	99.9%	Tyrosinemia, type II, 276600
TBC1D24	199.9	100.0%	100.0%	Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp, 608105 DOORS syndrome, 220500 Deafness, autosomal dominant 65, 616044 Epileptic encephalopathy, early infantile, 16, 615338 Myoclonic epilepsy, infantile, familial, 605021 Deafness , autosomal recessive 86, 614617
TBX3	109.8	99.9%	98.7%	Ulnar-mammary syndrome, 181450
TCHH	183.8	100.0%	100.0%	?Uncombable hair syndrome 3, 617252
TCIRG1	149.6	99.6%	98.0%	Osteopetrosis, autosomal recessive 1, 259700
TEK	151.2	100.0%	99.8%	Glaucoma 3, primary congenital, E, 617272 Venous malformations, multiple cutaneous and mucosal, 600195
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	141.2	100.0%	99.7%	No OMIM Disease ID
TERT	160.1	99.9%	99.0%	No OMIM disease ID
TFAP2A	124.1	100.0%	99.3%	Branchiooculofacial syndrome, 113620
TGFB2	179.0	100.0%	99.8%	Loeys-Dietz syndrome 4, 614816
TGFBR1	156.6	97.3%	94.3%	Loeys-Dietz syndrome 1, 609192
TGFBR2	169.1	100.0%	100.0%	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
TGM1	153.3	100.0%	100.0%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	144.3	99.9%	98.5%	?Uncombable hair syndrome 2, 617251
TGM5	152.1	100.0%	99.9%	Peeling skin syndrome 2, 609796
TINF2	190.9	100.0%	100.0%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
TMC6	102.1	100.0%	99.8%	Epidermodysplasia verruciformis, 226400
TMC8	148.5	100.0%	99.9%	Epidermodysplasia verruciformis 2, 618231
TMEM165	159.2	99.9%	99.7%	Congenital disorder of glycosylation, type IIk, 614727
TMEM173	104.9	99.6%	94.7%	STING-associated vasculopathy, infantile-onset, 615934
TNFRSF11A	139.5	96.4%	95.6%	Osteolysis, familial expansile, 174810 Osteopetrosis, autosomal recessive 7, 612301
TNFRSF11B	175.0	100.0%	100.0%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF1A	121.5	92.8%	91.4%	Periodic fever, familial, 142680
TNFSF11	133.0	100.0%	100.0%	Osteopetrosis, autosomal recessive 2, 259710

TNXB	119.2	99.8%	97.6%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP63	169.9	100.0%	100.0%	Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285
TPCN2	178.4	96.0%	95.2%	No OMIM disease ID
TREX1	261.9	100.0%	100.0%	Vasculopathy, retinal, with cerebral leukodystrophy, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	132.8	100.0%	100.0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM37	112.8	98.5%	97.4%	Mulibrey nanism, 253250
TRPM4	152.9	100.0%	100.0%	Erythrokeratodermia veriabilis et progressiva 6, 618531 Progressive familial heart block, type IB, 604559
TRPS1	160.6	100.0%	99.9%	Trichorhinophalangeal syndrome, type I, 190350 Trichorhinophalangeal syndrome, type III, 190351
TRPV3	131.6	99.8%	98.6%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome, 614594
TSC1	117.4	99.6%	98.4%	Tuberous sclerosis-1, 191100 Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, 606690
TSC2	155.5	100.0%	100.0%	Tuberous sclerosis-2, 613254 ?Focal cortical dysplasia, type II, somatic, 607341 Lymphangioleiomyomatosis, somatic, 606690
TSPEAR	151.2	100.0%	99.9%	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TTC37	131.5	99.9%	98.9%	Trichohepatoenteric syndrome 1, 222470
TTI2	100.1	100.0%	99.9%	Mental retardation, autosomal recessive 39, 615541
TWIST2	159.1	100.0%	100.0%	Barber-Say syndrome, 209885 Ablepharon-macrostomia syndrome, 200110 Focal facial dermal dysplasia 3, Setleis type, 227260
TYR	153.5	100.0%	100.0%	Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IB, 606952 Albinism, oculocutaneous, type IA, 203100

TYRP1	155.1	100.0%	99.9%	Albinism, oculocutaneous, type III, 203290
UBE2A	122.1	99.6%	97.4%	Mental retardation, X-linked syndromic, Nascimento-type, 300860
UBR1	118.2	99.9%	98.9%	Johanson-Blizzard syndrome, 243800
UROD	139.7	99.6%	96.7%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	104.6	100.0%	99.9%	Porphyria, congenital erythropoietic, 263700
USB1	122.0	99.8%	98.2%	Poikiloderma with neutropenia, 604173
UVSSA	140.1	99.6%	99.5%	UV-sensitive syndrome 3, 614640
VDR	116.5	99.0%	96.4%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	166.9	100.0%	100.0%	Lymphatic malformation 4, 615907
VHL	182.8	100.0%	99.8%	Pheochromocytoma, 171300 Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Hemangioblastoma, cerebellar, somatic, 0
VPS13B	135.9	99.4%	97.8%	Cohen syndrome, 216550
VPS33B	111.7	100.0%	100.0%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085
WAS	75.4	95.3%	84.4%	Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900 Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299
WDR19	125.3	100.0%	99.4%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	137.8	99.5%	98.3%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR72	123.2	96.9%	96.1%	Amelogenesis imperfecta, type IIA3, 613211
WIPF1	95.7	100.0%	99.3%	?Wiskott-Aldrich syndrome 2, 614493
WNT10A	159.7	100.0%	100.0%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	176.5	100.0%	100.0%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
WNT5A	174.9	100.0%	100.0%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	218.7	100.0%	100.0%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820

WRAP53	178.7	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	120.3	100.0%	98.7%	Werner syndrome, 277700
XPA	73.5	99.9%	97.6%	Xeroderma pigmentosum, group A, 278700
XPC	151.8	100.0%	99.9%	Xeroderma pigmentosum, group C, 278720
XYLT1	138.1	100.0%	99.4%	Desbuquois dysplasia 2, 615777
XYLT2	161.8	99.9%	98.7%	Spondyloocular syndrome, 605822
YWHAZ	45.9	81.3%	67.3%	No OMIM Disease ID
ZBTB20	199.5	100.0%	100.0%	Primrose syndrome, 259050
ZMPSTE24	126.2	100.0%	99.7%	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZNF469	180.5	100.0%	100.0%	Brittle cornea syndrome 1, 229200
ZNF592	157.0	100.0%	100.0%	No OMIM Disease ID
ZNF750	197.7	100.0%	99.9%	Seborrhea-like dermatitis with psoriasiform elements, 610227

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors