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SKIN DISORDERS PANEL[1] DG-5.2.0 (686 GENES)

Gene	Twist X2 covered 10x	Twist X2 covered 20x	srWGS covered 10x	srWGS covered 15x	srWGS covered 20x	Associated Phenotype description and OMIM disease ID
AAAS	n.t.b					Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	n.t.b					Keratoderma, palmoplantar, punctate type IA, 148600
AARS1	n.t.b					Developmental and epileptic encephalopathy 29, 616339;Charcot-Marie-Tooth disease, axonal, type 2N, 613287;?Leukoencephalopathy, hereditary diffuse, with spheroids 2, 619661;Trichothiodystrophy 8, nonphotosensitive, 619691
ABCA12	n.t.b					Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500;Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCB6	n.t.b					Dyschromatosis universalis hereditaria 3, 615402;[Blood group, Langereis system], 111600;Pseudohyperkalemia, familial, 2, due to red cell leak, 609153;Microphthalmia/coloboma 7, 614497

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ABCC6	n.t.b					Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	n.t.b					Cardiomyopathy, dilated, 1O, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719
ABHD5	n.t.b					Chanarin-Dorfman syndrome, 275630
ACD	n.t.b					?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
ACTA2	n.t.b					Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042

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ACTB	n.t.b					Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470
ACVRL1	n.t.b					Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADA2	n.t.b					Sneddon syndrome, 182410;Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM10	n.t.b					{Alzheimer disease 18, susceptibility to}, 615590;Reticulate acropigmentation of Kitamura, 615537
ADAM17	n.t.b					?Inflammatory skin and bowel disease, neonatal, 1, 614328;?Hypotichosis 16, 621490
ADAMTS10	n.t.b					Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	n.t.b					Weill-Marchesani 4 syndrome, recessive, 613195
ADAMTS2	n.t.b					Ehlers-Danlos syndrome, dermatosparaxis type, 225410

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ADAMTS3	n.t.b					Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	n.t.b					Geleophysic dysplasia 1, 231050
ADAR	n.t.b					Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010
AGA	n.t.b					Aspartylglucosaminuria, 208400
AGPAT2	n.t.b					Lipodystrophy, congenital generalized, type 1, 608594
AIRE	n.t.b					Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKT1	n.t.b					Proteus syndrome, somatic mosaic, 176920;Breast cancer, somatic, 114480;Cowden syndrome 6, 615109;Colorectal cancer, somatic, 114500;Ovarian cancer, somatic, 167000
AKT3	n.t.b					Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	n.t.b					Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740
ALAS2	n.t.b					Anemia, sideroblastic, 1, 300751;Protoporphyria, erythropoietic, X-linked, 300752

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ALDH18A1	n.t.b					Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603
ALDH3A2	n.t.b					Sjogren-Larsson syndrome, 270200
ALDOB	n.t.b					Fructose intolerance, hereditary, 229600
ALOX12B	n.t.b					Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	n.t.b					Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	n.t.b					Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300
ALX4	n.t.b					Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMELX	n.t.b					Amelogenesis imperfecta, type 1E, 301200
ANGPT2	n.t.b					Lymphatic malformation 10, 619369
ANKRD11	n.t.b					KBG syndrome, 148050

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ANOS1	n.t.b					Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	n.t.b					GAPO syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089
ANTXR2	n.t.b					Hyaline fibromatosis syndrome, 228600
AP1B1	n.t.b					Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150
AP1S3	n.t.b					{Psoriasis 15, pustular, susceptibility to}, 616106
AP3B1	n.t.b					Hermansky-Pudlak syndrome 2, 608233
APC	n.t.b					Colorectal cancer, somatic, 114500;Brain tumor-polyposis syndrome 2, 175100;Desmoid disease, hereditary, 135290;Adenoma, periampullary, somatic, 175100;Hepatoblastoma, somatic, 114550;Gastric cancer, somatic, 613659;Gastric adenocarcinoma and proximal polyposis of the stomach, 619182;Gardner syndrome, 175100;Adenomatous polyposis coli, 175100
APCDD1	n.t.b					Hypotrichosis 1, 605389
AQP5	n.t.b					Palmoplantar keratoderma, Bothnian type, 600231

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ARHGAP31	n.t.b					Adams-Oliver syndrome 1, 100300
ARID1A	n.t.b					Coffin-Siris syndrome 2, 614607
ARID1B	n.t.b					Coffin-Siris syndrome 1, 135900
ASIP	n.t.b					[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742;[Skin/hair/eye pigmentation 9, dark/light hair], 611742
ASL	n.t.b					Argininosuccinic aciduria, 207900
ASPRV1	n.t.b					Ichthyosis, lamellar, autosomal dominant, 146750
ASXL1	n.t.b					Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
ASXL3	n.t.b					Bainbridge-Ropers syndrome, 615485
ATIC	n.t.b					AICA-ribosiduria due to ATIC deficiency, 608688
ATP2A2	n.t.b					Acrokeratosis verruciformis, 101900;Darier disease, 124200;{Rhabdomyolysis, susceptibility to, 2}, 621236
ATP2C1	n.t.b					Hailey-Hailey disease, 169600
ATP6V0A2	n.t.b					Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200

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ATP7A	n.t.b					Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400
ATR	n.t.b					Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	n.t.b					Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615
B3GALT6	n.t.b					Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465
B4GALT7	n.t.b					Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BANF1	n.t.b					Nestor-Guillermo progeria syndrome, 614008
BAP1	n.t.b					Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661
BCOR	n.t.b					Microphthalmia, syndromic 2, 300166

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BCS1L	n.t.b					GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000
BLM	n.t.b					Bloom syndrome, 210900
BLOC1S3	n.t.b					Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	n.t.b					Hermansky-Pudlak syndrome 9, 614171
BMS1	n.t.b					?Aplasia cutis congenita, nonsyndromic, 107600
BRAF	n.t.b					Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980
BRIP1	n.t.b					Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480

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BSCL2	n.t.b					Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	n.t.b					Biotinidase deficiency, 253260
C1QA	n.t.b					C1q deficiency 1, 613652
C1QB	n.t.b					C1q deficiency 2, 620321
C1QC	n.t.b					C1q deficiency 3, 620322
C2CD3	n.t.b					Orofaciodigital syndrome XIV, 615948
CA2	n.t.b					Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAPN12	n.t.b					
CARD11	n.t.b					B-cell expansion with NFKB and T-cell anergy, 616452;Immunodeficiency 11B with atopic dermatitis, 617638;Immunodeficiency 11A, 615206
CARD14	n.t.b					Psoriasis 2, 602723;Pityriasis rubra pilaris, 173200
CARD9	n.t.b					Immunodeficiency 103, susceptibility to fungal infection, 212050
CARMIL2	n.t.b					Immunodeficiency 58, 618131

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CARS1	n.t.b					Microcephaly, developmental delay, and brittle hair syndrome, 618891
CASP14	n.t.b					Ichthyosis, congenital, autosomal recessive 12, 617320
CAST	n.t.b					Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295
CAV1	n.t.b					Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721
CAVIN1	n.t.b					Lipodystrophy, congenital generalized, type 4, 613327
CAVIN2	n.t.b					
CBL	n.t.b					Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785
CBS	n.t.b					Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200
CCBE1	n.t.b					Hennekam lymphangiectasia-lymphedema syndrome 1, 235510

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CD151	n.t.b					[Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CDAN1	n.t.b					Dyserythropoietic anemia, congenital, type Ia, 224120
CDH3	n.t.b					Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDSN	n.t.b					Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300
CELSR1	n.t.b					Lymphatic malformation 9, 619319;Yellow nail syndrome, 153300
CERS3	n.t.b					Ichthyosis, congenital, autosomal recessive 9, 615023
CFTR	n.t.b					Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}
CHKB	n.t.b					Muscular dystrophy, congenital, megaconial type, 602541
CHST14	n.t.b					Ehlers-Danlos syndrome, musculocontractural type 1, 601776

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CHST8	n.t.b					
CHSY1	n.t.b					Temtamy preaxial brachydactyly syndrome, 605282
CHUK	n.t.b					?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339;?Cocoon syndrome, 613630
CIB1	n.t.b					{Epidermodysplasia verruciformis, susceptibility to, 3}, 618267
CKAP2L	n.t.b					Filippi syndrome, 272440
CLDN1	n.t.b					Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	n.t.b					HELIX syndrome, 617671
CNNM4	n.t.b					Jalili syndrome, 217080
COG6	n.t.b					Shaheen syndrome, 615328;Congenital disorder of glycosylation, type III, 614576
COL14A1	n.t.b					
COL17A1	n.t.b					Epithelial recurrent erosion dystrophy, 122400;Epidermolysis bullosa, junctional 4, intermediate, 619787

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COL1A2	n.t.b					Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210
COL3A1	n.t.b					Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	n.t.b					Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329
COL5A2	n.t.b					Ehlers-Danlos syndrome, classic type, 2, 130010

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COL7A1	n.t.b					Nail disorder, nonsyndromic congenital, 8, 607523;Epidermolysis bullosa dystrophica, Bart type, 132000;Epidermolysis bullosa dystrophica inversa, 226600;Epidermolysis bullosa dystrophica, autosomal recessive, 226600;Epidermolysis bullosa, pretibial, 131850;Epidermolysis bullosa dystrophica, autosomal dominant, 131750;Transient bullous of the newborn, 131705;Epidermolysis bullosa pruriginosa, 604129;Epidermolysis bullosa dystrophica, localisata variant, 226600
COX4I2	n.t.b					Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	n.t.b					Linear skin defects with multiple congenital anomalies 2, 300887
CPOX	n.t.b					Coproporphyria, 121300;Harderoporphyria, 618892
CRIPT	n.t.b					Rothmund-Thomson syndrome, type 3, 615789
CST6	n.t.b					?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	n.t.b					Peeling skin syndrome 4, 607936
CTC1	n.t.b					Cerebroretinal microangiopathy with calcifications and cysts, 612199

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CTSA	n.t.b					Galactosialidosis, 256540;Brain small vessel disease 6 with leukoencephalopathy, 621394
CTSB	n.t.b					Keratolytic winter erythema, 148370
CTSC	n.t.b					Periodontitis 1, aggressive, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000
CTSZ	n.t.b					
CXCR4	n.t.b					WHIM syndrome, 193670
CYLD	n.t.b					Brooke-Spiegler syndrome, 605041;Cylindromatosis, familial, 132700;Trichoepithelioma, multiple familial, 1, 601606;?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132
CYP26C1	n.t.b					Focal facial dermal dysplasia 4, 614974
CYP4F22	n.t.b					Ichthyosis, congenital, autosomal recessive 5, 604777
DCAF17	n.t.b					Woodhouse-Sakati syndrome, 241080
DCLRE1C	n.t.b					Severe combined immunodeficiency, Athabascan type, 602450;Omenn syndrome, 603554
DDB2	n.t.b					Xeroderma pigmentosum, group E, 278740
DHCR7	n.t.b					Smith-Lemli-Opitz syndrome, 270400

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DKC1	n.t.b					?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
DLX3	n.t.b					Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510
DLX5	n.t.b					Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DOCK6	n.t.b					Adams-Oliver syndrome 2, 614219
DOCK8	n.t.b					Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700
DOLK	n.t.b					Congenital disorder of glycosylation, type Im, 610768
DSC2	n.t.b					Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	n.t.b					Hypotrichosis and recurrent skin vesicles, 613102
DSE	n.t.b					Ehlers-Danlos syndrome, musculocontractural type 2, 615539

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DSG1	n.t.b					Keratosis palmoplantaris striata I, AD, 148700;Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG3	n.t.b					?Blistering, acantholytic, of oral and laryngeal mucosa, 619226
DSG4	n.t.b					Hypotrichosis 6, 607903
DSP	n.t.b					Arrhythmogenic right ventricular dysplasia 8, 607450;Epidermolysis bullosa, lethal acantholytic, 609638;Keratosis palmoplantaris striata II, 612908;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676
DSPP	n.t.b					Dentinogenesis imperfecta, Shields type III, 125500;Dentinogenesis imperfecta, Shields type II, 125490;Dentin dysplasia, type II, 125420;Deafness, autosomal dominant 39, with dentinogenesis, 605594

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DST	n.t.b					Congenital myopathy 29 with contractures, 621510;Neuropathy, hereditary sensory and autonomic, type VI, 614653;Lethal congenital contracture syndrome 12, 621511;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425
DTNBP1	n.t.b					Hermansky-Pudlak syndrome 7, 614076
DUSP1	n.t.b					
DUSP6	n.t.b					Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EBP	n.t.b					MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960
ECM1	n.t.b					Urbach-Wiethe disease, 247100
EDA	n.t.b					Tooth agenesis, selective, X-linked 1, 313500;Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	n.t.b					[Hair morphology 1, hair thickness], 612630;Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900

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EDARADD	n.t.b					Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDN3	n.t.b					Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712
EDNRA	n.t.b					{Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367
EDNRB	n.t.b					{Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580
EFEMP2	n.t.b					Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	n.t.b					Craniofrontonasal dysplasia, 304110
EIF2AK3	n.t.b					Wolcott-Rallison syndrome, 226980
ELN	n.t.b					Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500
ELOVL1	n.t.b					Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527

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ELOVL4	n.t.b					Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457
EMP2	n.t.b					Nephrotic syndrome, type 10, 615861
ENAM	n.t.b					Amelogenesis imperfecta, type IC, 204650;Amelogenesis imperfecta, type IB, 104500
ENG	n.t.b					Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	n.t.b					{Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522
EPG5	n.t.b					Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506
EPHB4	n.t.b					Capillary malformation-arteriovenous malformation 2, 618196;Lymphatic malformation 7, 617300
EPHX3	n.t.b					
EPS8L3	n.t.b					?Hypotrichosis 5, 612841

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ERCC2	n.t.b					Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	n.t.b					Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651
ERCC4	n.t.b					Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272
ERCC5	n.t.b					Xeroderma pigmentosum, group G, 278780;Cerebrooculofacioskeletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	n.t.b					UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980

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ERCC8	n.t.b					UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400
ERG	n.t.b					Lymphatic malformation 14, 620602
EVC	n.t.b					Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530
EVC2	n.t.b					Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530
EXPH5	n.t.b					Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028
F13A1	n.t.b					Factor XIIIA deficiency, 613225;{Myocardial infarction, protection against}, 608446;{Venous thrombosis, protection against}, 188050
FAM111B	n.t.b					Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM20A	n.t.b					Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	n.t.b					Raine syndrome, 259775
FAM83G	n.t.b					
FAM83H	n.t.b					

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FANCA	n.t.b					Fanconi anemia, complementation group A, 227650
FANCB	n.t.b					Fanconi anemia, complementation group B, 300514
FANCC	n.t.b					Fanconi anemia, complementation group C, 227645
FANCD2	n.t.b					Fanconi anemia, complementation group D2, 227646
FANCE	n.t.b					Fanconi anemia, complementation group E, 600901
FANCF	n.t.b					Fanconi anemia, complementation group F, 603467
FANCG	n.t.b					Fanconi anemia, complementation group G, 614082
FANCI	n.t.b					Fanconi anemia, complementation group I, 609053
FANCL	n.t.b					Fanconi anemia, complementation group L, 614083
FANCM	n.t.b					Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FAT4	n.t.b					Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006

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FBLN5	n.t.b					Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434
FDFT1	n.t.b					Squalene synthase deficiency, 618156
FDPS	n.t.b					Porokeratosis 9, multiple types, 616631
FECH	n.t.b					Protoporphyria, erythropoietic, 1, 177000
FERMT1	n.t.b					Kindler syndrome, 173650
FGF10	n.t.b					LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920
FGF23	n.t.b					Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	n.t.b					Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	n.t.b					Trichomegaly, 190330
FGF8	n.t.b					Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

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FGFR1	n.t.b					Pfeiffer syndrome, 101600;Hypogonadotropic hypogonadism 2 with or without anosmia, 147950;Jackson-Weiss syndrome, 123150;Hartsfield syndrome, 615465;Trigonocephaly 1, 190440;Osteoglophonic dysplasia, 166250;Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	n.t.b					Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Scaphocephaly and Axenfeld-Rieger anomaly;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;Craniosynostosis, nonspecific;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400

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FGFR3	n.t.b					Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247
FH	n.t.b					Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FKBP10	n.t.b					Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450
FKBP14	n.t.b					Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLCN	n.t.b					Birt-Hogg-Dube syndrome, 135150;Colorectal cancer, somatic, 114500;Pneumothorax, primary spontaneous, 173600;Renal carcinoma, chromophobe, somatic, 144700

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FLG	n.t.b					Ichthyosis vulgaris, 146700;{Dermatitis, atopic, susceptibility to, 2}, 605803
FLG2	n.t.b					Peeling skin syndrome 6, 618084
FLT4	n.t.b					Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780
FNIP1	n.t.b					Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXC2	n.t.b					Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	n.t.b					Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534
FOXN1	n.t.b					T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705;?T-cell immunodeficiency with thymic aplasia, 242700
FOXP3	n.t.b					Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790

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FREM1	n.t.b					Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485
FUCA1	n.t.b					Fucosidosis, 230000
FZD6	n.t.b					Nail disorder, nonsyndromic congenital, 1, 161050
GALNS	n.t.b					Mucopolysaccharidosis IVA, 253000
GALNT3	n.t.b					Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GAN	n.t.b					Giant axonal neuropathy-1, 256850
GATA2	n.t.b					{Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286
GBA1	n.t.b					{Lewy body dementia, susceptibility to}, 127750;Gaucher disease, type II, 230900;Gaucher disease, type IIIC, 231005;Gaucher disease, type III, 231000;Gaucher disease, type I, 230800;Gaucher disease, perinatal lethal, 608013;{Parkinson disease, late-onset, susceptibility to}, 168600

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GDF2	n.t.b					Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF5	n.t.b					Acromesomelic dysplasia 2A, 200700;Acromesomelic dysplasia 2B, 228900;Multiple synostoses syndrome 2, 610017;Symphalangism, proximal, 1B, 615298;Brachydactyly, type A2, 112600;?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072
GGCX	n.t.b					Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GJA1	n.t.b					Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850

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GJB2	n.t.b					Keratoderma, palmoplantar, with deafness, 148350;Deafness, autosomal recessive 1A, 220290;Deafness, autosomal dominant 3A, 601544;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200;Keratitis-ichthyosis-deafness syndrome, 148210;Vohwinkel syndrome, 124500
GJB3	n.t.b					Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratodermia variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644
GJB4	n.t.b					Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	n.t.b					Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290
GJC2	n.t.b					Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804
GLA	n.t.b					Fabry disease, cardiac variant, 301500;Fabry disease, 301500

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GLB1	n.t.b					GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600
GLMN	n.t.b					Glomuvenous malformations, 138000
GMPPA	n.t.b					Alacrima, achalasia, and impaired intellectual development syndrome, 615510
GNA11	n.t.b					Hypocalciuric hypercalcemia, type II, 145981;Hypocalcemia, autosomal dominant 2, 615361
GNA14	n.t.b					
GNAQ	n.t.b					Capillary malformations, congenital, 1, somatic, mosaic, 163000;Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	n.t.b					Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism Ic, 612462;Pseudohypoparathyroidism Ia, 103580;Osseous heteroplasia, progressive, 166350;McCune-Albright syndrome, somatic mosaic, 174800;Pseudohypoparathyroidism Ib, 603233;Pseudopseudohypoparathyroidism, 612463;ACTH-independent macronodular adrenal hyperplasia 1, somatic, 219080

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GORAB	n.t.b					Geroderma osteodysplasticum, 231070
GPNMB	n.t.b					Amyloidosis, primary localized cutaneous, 3, 617920
GPR143	n.t.b					Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814
GREM2	n.t.b					Tooth agenesis, selective, 9, 617275
GRHL2	n.t.b					Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
GRHL3	n.t.b					van der Woude syndrome 2, 606713
GSN	n.t.b					Amyloidosis, Finnish type, 105120
GTF2E2	n.t.b					Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	n.t.b					Trichothiodystrophy 3, photosensitive, 616395
HCCS	n.t.b					Linear skin defects with multiple congenital anomalies 1, 309801
HDAC8	n.t.b					Cornelia de Lange syndrome 5, 300882

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HERC2	n.t.b					Intellectual developmental disorder, autosomal recessive 38, 615516;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220
HLCS	n.t.b					Holocarboxylase synthetase deficiency, 253270
HMBS	n.t.b					Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000
HMCN1	n.t.b					{Macular degeneration, age-related, 1}, 603075
HMGB3	n.t.b					?Microphthalmia, syndromic 13, 300915
HOXC13	n.t.b					Ectodermal dysplasia 9, hair/nail type, 614931
HPGD	n.t.b					?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100
HPS1	n.t.b					Hermansky-Pudlak syndrome 1, 203300
HPS3	n.t.b					Hermansky-Pudlak syndrome 3, 614072
HPS4	n.t.b					Hermansky-Pudlak syndrome 4, 614073

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HPS5	n.t.b					Hermansky-Pudlak syndrome 5, 614074
HPS6	n.t.b					Hermansky-Pudlak syndrome 6, 614075
HR	n.t.b					Atrichia with papular lesions, 209500;Alopecia universalis, 203655
HRAS	n.t.b					Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040
HTRA1	n.t.b					Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy 2, 616779;Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy 2, 600142
HYAL1	n.t.b					Mucopolysaccharidosis type IX, 601492
IDUA	n.t.b					Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014
IFT122	n.t.b					Cranioectodermal dysplasia 1, 218330

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IFT43	n.t.b					?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IKBKB	n.t.b					Immunodeficiency 15B, 615592;Immunodeficiency 15A, 618204
IKBKG	n.t.b					Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081
IL17RA	n.t.b					Immunodeficiency 51, 613953
IL17RD	n.t.b					Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RN	n.t.b					Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852;{Gastric cancer risk after H. pylori infection}, 613659;{Microvascular complications of diabetes 4}, 612628;Interleukin 1 receptor antagonist deficiency, 612852
IL31RA	n.t.b					?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	n.t.b					Psoriasis 14, pustular, 614204

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INSR	n.t.b					Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968
IRF4	n.t.b					[Skin/hair/eye pigmentation, variation in, 8], 611724;Immunodeficiency 131, 621097
IRF6	n.t.b					{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300;Ectodermal dysplasia 19, hypohydrotic/tooth/nail type, 621749
ISG15	n.t.b					Immunodeficiency 38, 616126
ITGA3	n.t.b					Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748
ITGA6	n.t.b					Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817
ITGB4	n.t.b					Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730;Epidermolysis bullosa, junctional 5A, intermediate, 619816
ITGB6	n.t.b					Amelogenesis imperfecta, type IH, 616221

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JUP	n.t.b					Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528
KANK2	n.t.b					Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099
KAT6B	n.t.b					SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KCNH1	n.t.b					Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816
KCNK9	n.t.b					Birk-Barel syndrome, 612292
KCTD1	n.t.b					Scalp-ear-nipple syndrome, 181270;Dental radicular dysplasia, 621434
KDF1	n.t.b					?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDSR	n.t.b					Erythrokeratodermia variabilis et progressiva 4, 617526
KIF11	n.t.b					Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950

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KIT	n.t.b					Gastrointestinal stromal tumor, familial, 606764;Mastocytosis, cutaneous, 154800;Piebaldism, 172800;Germ cell tumors, somatic, 273300;Mastocytosis, systemic, somatic, 154800;Leukemia, acute myeloid, somatic, 601626
KITLG	n.t.b					Hyperpigmentation with or without hypopigmentation, 145250;Waardenburg syndrome, type 2F, 619947;Deafness, autosomal dominant 69, unilateral or asymmetric, 616697;[Skin/hair/eye pigmentation 7, blond/brown hair], 611664
KLF4	n.t.b					
KLHL24	n.t.b					Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294
KLK11	n.t.b					Ichthyosis with erythrokeratoderma, 620507
KLK4	n.t.b					Amelogenesis imperfecta, type IIA1, 204700
KLLN	n.t.b					Cowden syndrome 4, 615107

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KMT2D	n.t.b					Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KRAS	n.t.b					Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;RAS-associated autoimmune leukoproliferative disorder 2, somatic, 621582;Bladder cancer, somatic, 109800

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KRT1	n.t.b					Ichthyosis, annular epidermolytic 2, 620148;Palmoplantar keratoderma, nonepidermolytic, 600962;Epidermolytic hyperkeratosis 1, 113800;Palmoplantar keratoderma, epidermolytic, 2, 620411;Ichthyosis histrix, Curth-Macklin type, 146590;?Keratosis palmoplantaris striata III, 607654
KRT10	n.t.b					Ichthyosis, annular epidermolytic 1, 607602;Epidermolytic hyperkeratosis 2B, autosomal recessive, 620707;Epidermolytic hyperkeratosis 2A, autosomal dominant, 620150;?Ichthyosis histrix, Lambert type, 146600;Ichthyosis with confetti, 609165
KRT13	n.t.b					White sponge nevus 2, 615785
KRT14	n.t.b					Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001;Epidermolysis bullosa simplex 1C, localized, 131800;Dermatopathia pigmentosa reticularis, 125595;Epidermolysis bullosa simplex 1A, generalized severe, 131760;Naegeli-France schetti-Jadassohn syndrome, 161000;Epidermolysis bullosa simplex 1B, generalized intermediate, 131900

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KRT16	n.t.b					Palmoplantar keratoderma, nonepidermolytic, focal, 613000;Pachyonychia congenita 1, 167200
KRT17	n.t.b					Steatocystoma multiplex, 184500;Pachyonychia congenita 2, 167210
KRT2	n.t.b					Ichthyosis bullosa of Siemens, 146800
KRT31	n.t.b					
KRT4	n.t.b					White sponge nevus 1, 193900
KRT5	n.t.b					Epidermolysis bullosa simplex 2A, generalized severe, 619555;Dowling-Degos disease 1, 179850;Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960;Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599;Epidermolysis bullosa simplex 2B, generalized intermediate, 619588;Epidermolysis bullosa simplex 2C, localized, 619594;Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352
KRT6A	n.t.b					Pachyonychia congenita 3, 615726
KRT6B	n.t.b					Pachyonychia congenita 4, 615728
KRT6C	n.t.b					Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735

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KRT71	n.t.b					?Hypotrichosis 13, 615896
KRT74	n.t.b					Woolly hair, autosomal dominant, 194300;?Hypotrichosis 3, 613981;?Ectodermal dysplasia 7, hair/nail type, 614929
KRT75	n.t.b					{Pseudofolliculitis barbae, susceptibility to}, 612318
KRT81	n.t.b					Monilethrix 2, 621169
KRT82	n.t.b					
KRT83	n.t.b					Monilethrix 3, 621170;Erythrokeratodermia variabilis et progressiva 5, 617756
KRT85	n.t.b					Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	n.t.b					Monilethrix 1, 158000
KRT9	n.t.b					Palmoplantar keratoderma, epidermolytic, 1, 144200
LAMA3	n.t.b					Epidermolysis bullosa, junctional 2A, intermediate, 619783;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660;Epidermolysis bullosa, junctional 2B, severe, 619784
LAMB3	n.t.b					Epidermolysis bullosa, junctional 1B, severe, 226700;Epidermolysis bullosa, junctional 1A, intermediate, 226650;Amelogenesis imperfecta, type IA, 104530

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LAMC2	n.t.b					Epidermolysis bullosa, junctional 3B, severe, 619786;Epidermolysis bullosa, junctional 3A, intermediate, 619785
LAMTOR2	n.t.b					?Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LDHA	n.t.b					Glycogen storage disease XI, 612933
LDLRAP1	n.t.b					Hypercholesterolemia, familial, 4, 603813
LEMD3	n.t.b					Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700
LIPH	n.t.b					Hypotrichosis 7, 604379;Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	n.t.b					Ichthyosis, congenital, autosomal recessive 8, 613943
LMBRD1	n.t.b					Methylmalonic aciduria and homocystinuria, cblF type, 277380

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LMNA	n.t.b					Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112
LMX1B	n.t.b					Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200
LONP1	n.t.b					CODAS syndrome, 600373
LORICRIN	n.t.b					Vohwinkel syndrome with ichthyosis, 604117
LPAR6	n.t.b					Hypotrichosis 8, 278150;Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN2	n.t.b					Majeed syndrome, 609628
LRMDA	n.t.b					Albinism, oculocutaneous, type VII, 615179

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LRP6	n.t.b					Osteopetrosis, autosomal dominant 4, 621449;{Coronary artery disease, autosomal dominant, 2}, 610947;Tooth agenesis, selective, 7, 616724;Exudative vitreoretinopathy 8, 621268
LSS	n.t.b					Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840
LTBP3	n.t.b					Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809
LTBP4	n.t.b					Cutis laxa, autosomal recessive, type IC, 613177
LTV1	n.t.b					Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199
LYST	n.t.b					Chediak-Higashi syndrome, 214500
LYZ	n.t.b					Amyloidosis, hereditary systemic 5, 620658
MAGT1	n.t.b					Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853;Congenital disorder of glycosylation, type Icc, 301031
MAP2K1	n.t.b					Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	n.t.b					Cardiofaciocutaneous syndrome 4, 615280

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MARS1	n.t.b					Spastic paraplegia 70, autosomal recessive, 620323;Interstitial lung and liver disease, 615486;?Trichothiodystrophy 9, nonphotosensitive, 619692;Charcot-Marie-Tooth disease, axonal, type 2U, 616280
MBTPS2	n.t.b					Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;IFAP syndrome with or without BRESHECK syndrome, 308205;?Olmsted syndrome, X-linked, 300918
MDFIC	n.t.b					Lymphatic malformation 12, 620014
MED12	n.t.b					Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MEFV	n.t.b					Neutrophilic dermatosis, acute febrile, 608068;Familial Mediterranean fever, AR, 249100;Familial Mediterranean fever, AD, 134610
MGP	n.t.b					Keutel syndrome, 245150

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MITF	n.t.b					Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MLH1	n.t.b					Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MLPH	n.t.b					Griscelli syndrome, type 3, 609227
MMACHC	n.t.b					Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP1	n.t.b					
MMP14	n.t.b					Winchester syndrome, 277950
MMP2	n.t.b					Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	n.t.b					Amelogenesis imperfecta, type IIA2, 612529
MPDU1	n.t.b					Congenital disorder of glycosylation, type If, 609180
MPLKIP	n.t.b					Trichothiodystrophy 4, nonphotosensitive, 234050
MRE11	n.t.b					Ataxia-telangiectasia-like disorder 1, 604391
MSH2	n.t.b					Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096

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MSX1	n.t.b					Tooth agenesis, selective, 1, with or without orofacial cleft, 106600;Ectodermal dysplasia 3, Witkop type, 189500;Orofacial cleft 5, 608874
MTOR	n.t.b					Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638
MUTYH	n.t.b					Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659
MVD	n.t.b					Porokeratosis 7, multiple types, 614714
MVK	n.t.b					Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377
MYH8	n.t.b					Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300
MYO5A	n.t.b					Griscelli syndrome, type 1, 214450
NAA10	n.t.b					Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855
NAGA	n.t.b					Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241

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NBAS	n.t.b					Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483;?Acrofrontofacionasal dysostosis 3, 621629
NCF1	n.t.b					Chronic granulomatous disease 1, autosomal recessive, 233700
NCSTN	n.t.b					Acne inversa, familial, 1, 142690
NDUFB11	n.t.b					Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021
NECTIN1	n.t.b					Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060
NECTIN4	n.t.b					Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK11	n.t.b					
NEK9	n.t.b					?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022

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NF1	n.t.b					Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321
NFKBIA	n.t.b					Ectodermal dysplasia and immunodeficiency 2, 612132
NHP2	n.t.b					Dyskeratosis congenita, autosomal recessive 2, 613987
NIPAL4	n.t.b					Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	n.t.b					Cornelia de Lange syndrome 1, 122470
NKPD1	n.t.b					
NLRP1	n.t.b					{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579;?Respiratory papillomatosis, juvenile recurrent, congenital, 618803;Autoinflammation with arthritis and dyskeratosis, 617388;Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	n.t.b					Familial cold autoinflammatory syndrome 2, 611762

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NLRP3	n.t.b					CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendotheliitis fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900
NME1	n.t.b					
NOD2	n.t.b					Blau syndrome, 186580;{Yao syndrome}, 617321;{Inflammatory bowel disease 1, Crohn disease}, 266600
NOP10	n.t.b					?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230
NOTCH1	n.t.b					Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730

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NRAS	n.t.b					Noonan syndrome 6, 613224;RAS-associated autoimmune lymphoproliferative syndrome type 1, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500
NSD1	n.t.b					Sotos syndrome, 117550
NSDHL	n.t.b					CK syndrome, 300831;CHILD syndrome, 308050
OCA2	n.t.b					[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;Albinism, brown oculocutaneous, 203200;Albinism, oculocutaneous, type II, 203200
ODAM	n.t.b					
ODAPH	n.t.b					Amelogenesis imperfecta, type IIA4, 614832

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OFD1	n.t.b					Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
OSMR	n.t.b					Amyloidosis, primary localized cutaneous, 1, 105250
PADI3	n.t.b					Uncombable hair syndrome, 191480
PAH	n.t.b					[Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600
PALB2	n.t.b					{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832
PAX3	n.t.b					Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PAX9	n.t.b					Tooth agenesis, selective, 3, 604625
PCNA	n.t.b					?Ataxia-telangiectasia-like disorder 2, 615919
PDGFB	n.t.b					Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483

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PDGFRB	n.t.b					Premature aging syndrome, Penttinen type, 601812;?Ocular pterygium-digital keloid dysplasia syndrome, 621091;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007
PEPD	n.t.b					Prolidase deficiency, 170100
PER3	n.t.b					?Advanced sleep phase syndrome, familial, 3, 616882
PERP	n.t.b					Erythrokeratodermia variabilis et progressiva 7, 619209;Olmsted syndrome 2, 619208
PEX7	n.t.b					Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879
PHEX	n.t.b					Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	n.t.b					Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PHYH	n.t.b					Refsum disease, 266500

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PIEZO1	n.t.b					[ER blood group system], 620207;Lymphatic malformation 6, 616843;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIGA	n.t.b					Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGL	n.t.b					CHIME syndrome, 280000
PIGN	n.t.b					Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	n.t.b					Hyperphosphatasia with impaired intellectual development syndrome 1, 239300

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PIK3CA	n.t.b					Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Nevus, epidermal, somatic mosaic, 162900;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108
PIK3R1	n.t.b					Immunodeficiency 36, 616005;SHORT syndrome, 269880;Agammaglobulinemia 7, autosomal recessive, 615214
PIK3R2	n.t.b					Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387

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PITX2	n.t.b					Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PKP1	n.t.b					Ectodermal dysplasia/skin fragility syndrome, 604536
PLCD1	n.t.b					Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCG2	n.t.b					Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468
PLEC	n.t.b					?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487;Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670;Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138;Epidermolysis bullosa simplex 5A, Ogna type, 131950;Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723
PLG	n.t.b					Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090
PLIN1	n.t.b					Lipodystrophy, familial partial, type 4, 613877

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PLOD1	n.t.b					Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD3	n.t.b					BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394
PMS2	n.t.b					Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
PMVK	n.t.b					Porokeratosis 1, multiple types, 175800
PNPLA1	n.t.b					Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	n.t.b					Neutral lipid storage disease with myopathy, 610717
PNPT1	n.t.b					Spinocerebellar ataxia 25, 608703;Deafness, autosomal recessive 70, with or without adult-onset neurodegeneration, 614934;Combined oxidative phosphorylation deficiency 13, 614932
POC1A	n.t.b					Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POFUT1	n.t.b					Dowling-Degos disease 2, 615327
POGLUT1	n.t.b					Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232

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POLA1	n.t.b					Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220;Van Esch-O'Driscoll syndrome, 301030
POLD1	n.t.b					Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLH	n.t.b					Xeroderma pigmentosum, variant type, 278750
POLR1C	n.t.b					Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	n.t.b					Treacher Collins syndrome 2, 613717
POLR3A	n.t.b					Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	n.t.b					Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742

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POMC	n.t.b					{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMP	n.t.b					Proteasome-associated autoinflammatory syndrome 2, 618048;Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
PORCN	n.t.b					Focal dermal hypoplasia, 305600
POT1	n.t.b					Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367
PPOX	n.t.b					Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200
PQBP1	n.t.b					Renpenning syndrome, 309500
PRDM10	n.t.b					?Birt-Hogg-Dube syndrome 2, 620459
PRDM12	n.t.b					Neuropathy, hereditary sensory and autonomic, type VIII, 616488

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PRKAR1A	n.t.b					Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Adrenocortical tumor, somatic;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960
PSAT1	n.t.b					Neu-Laxova syndrome 2, 616038;Phosphoserine aminotransferase deficiency, 610992
PSEN1	n.t.b					Pick disease, 172700;Dementia, frontotemporal, 600274;?Acne inversa, familial, 3, 613737;?Cardiomyopathy, dilated, 1U, 613694;Alzheimer disease, type 3, with or without spastic paraparesis, 607822
PSENEN	n.t.b					Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB8	n.t.b					Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSPH	n.t.b					Phosphoserine phosphatase deficiency, 614023
PSTPIP1	n.t.b					Autoinflammatory syndrome with cytopenia, hyperzincemia, and hypercalprotectinemia, 601979;Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416

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PTCH1	n.t.b					Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828
PTCH2	n.t.b					Medulloblastoma, somatic, 155255;Basal cell carcinoma, somatic, 605462
PTDSS1	n.t.b					Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	n.t.b					{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
PTHLH	n.t.b					Brachydactyly, type E2, 613382
PTPN11	n.t.b					Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN14	n.t.b					Choanal atresia and lymphedema, 613611
PTPRF	n.t.b					?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PYCR1	n.t.b					Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940

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RAB23	n.t.b					Carpenter syndrome 1, 201000
RAB27A	n.t.b					Griscelli syndrome, type 2, 607624
RAD21	n.t.b					Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RAD50	n.t.b					Nijmegen breakage syndrome-like disorder, 613078
RAF1	n.t.b					Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554
RAG1	n.t.b					Omenn syndrome, 603554;Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889
RAG2	n.t.b					Severe combined immunodeficiency, B cell-negative, 601457;Combined cellular and humoral immune defects with granulomas, 233650;Omenn syndrome, 603554
RAI1	n.t.b					Smith-Magenis syndrome, 182290

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RASA1	n.t.b					Capillary malformation-arteriovenous malformation 1, 608354;Basal cell carcinoma, somatic, 605462
RBBP8	n.t.b					Seckel syndrome 2, 606744;Jawad syndrome, 251255;Pancreatic carcinoma, somatic
RBM28	n.t.b					?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBP4	n.t.b					Microphthalmia/coloboma 10, 616428;Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	n.t.b					Adams-Oliver syndrome 3, 614814
RECQL4	n.t.b					Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
RHBDF2	n.t.b					Tylosis with esophageal cancer, 148500
RHOA	n.t.b					Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RIN2	n.t.b					Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075

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RIPK4	n.t.b					CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RIT1	n.t.b					Noonan syndrome 8, 615355
RMRP	n.t.b					Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250
RNASEH2A	n.t.b					Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	n.t.b					Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	n.t.b					Aicardi-Goutieres syndrome 3, 610329
RNF113A	n.t.b					Trichothiodystrophy 5, nonphotosensitive, 300953
RNU4ATAC	n.t.b					Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710
ROGDI	n.t.b					Kohlschutter-Tonz syndrome, 226750
RPL10A	n.t.b					
RPL21	n.t.b					Hypotrichosis 12, 615885
RSPO1	n.t.b					Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644

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RSPO4	n.t.b					Anonychia congenita, 206800
RTEL1	n.t.b					Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373
RUNX2	n.t.b					Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510;Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600;Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600;Cleidocranial dysplasia, 119600
S100A3	n.t.b					
SAMD9	n.t.b					Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SAMHD1	n.t.b					?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952
SART3	n.t.b					
SASH1	n.t.b					Dyschromatosis universalis hereditaria 1, 127500;?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373

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SAT1	n.t.b					Systemic lupus erythematosus 19, 301172
SATB2	n.t.b					Glass syndrome, 612313
SCN10A	n.t.b					Episodic pain syndrome, familial, 2, 615551
SCN11A	n.t.b					Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	n.t.b					Erythermalgia, primary, 133020;Insensitivity to pain, congenital, 243000;Small fiber neuropathy, 133020;Paroxysmal extreme pain disorder, 167400;Neuropathy, hereditary sensory and autonomic, type IID, 243000
SDR9C7	n.t.b					Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23B	n.t.b					?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100
SERPINA12	n.t.b					
SERPINA3	n.t.b					Alpha-1-antichymotrypsin deficiency;Cerebrovascular disease, occlusive
SERPINB7	n.t.b					Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	n.t.b					Peeling skin syndrome 5, 617115

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SERPING1	n.t.b					Angioedema, hereditary, 1 and 2, 106100;Complement component 4, partial deficiency of, 120790
SERPINH1	n.t.b					{Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848
SGPL1	n.t.b					RENI syndrome, 617575
SHOC2	n.t.b					Noonan syndrome-like with loose anagen hair 1, 607721
SKI	n.t.b					Shprintzen-Goldberg syndrome, 182212
SKIC2	n.t.b					Trichohepatoenteric syndrome 2, 614602
SKIC3	n.t.b					Trichohepatoenteric syndrome 1, 222470
SLC17A9	n.t.b					Porokeratosis 8, disseminated superficial actinic type, 616063
SLC24A4	n.t.b					[Skin/hair/eye pigmentation 6, blond/brown hair], 210750;Amelogenesis imperfecta, type IIA5, 615887;[Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC24A5	n.t.b					[Skin/hair/eye pigmentation 4, fair/dark skin], 113750;Albinism, oculocutaneous, type VI, 113750

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SLC26A2	n.t.b					Epiphyseal dysplasia, multiple, 4, 226900;Diastrophic dysplasia, 222600;Diastrophic dysplasia, broad bone-platyspondylic variant, 222600;Achondrogenesis IB, 600972;Atelosteogenesis, type II, 256050
SLC27A4	n.t.b					Ichthyosis prematurity syndrome, 608649
SLC29A3	n.t.b					Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A10	n.t.b					Arterial tortuosity syndrome, 208050
SLC39A13	n.t.b					Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A4	n.t.b					Acrodermatitis enteropathica, 201100
SLC45A2	n.t.b					[Skin/hair/eye pigmentation 5, dark/light eyes], 227240;[Skin/hair/eye pigmentation 5, black/nonblack hair], 227240;Albinism, oculocutaneous, type IV, 606574;[Skin/hair/eye pigmentation 5, dark/fair skin], 227240
SLC4A4	n.t.b					Proximal renal tubular acidosis-ocular anomaly syndrome, 604278;Progressive macrocephaly-brain edema disorder, 621716
SLC6A19	n.t.b					Hartnup disorder, 234500

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SLC7A7	n.t.b					Lysinuric protein intolerance, 222700
SLCO2A1	n.t.b					Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441
SLURP1	n.t.b					Meleda disease, 248300
SLX4	n.t.b					Fanconi anemia, complementation group P, 613951
SMAD3	n.t.b					Loeys-Dietz syndrome 3, 613795
SMARCA2	n.t.b					Nicolaides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	n.t.b					Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SMARCAD1	n.t.b					Basan syndrome, 129200;Huriez syndrome, 181600;Adermatoglyphia, 136000
SMARCAL1	n.t.b					Schimke immunoosseous dysplasia, 242900
SMARCB1	n.t.b					Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322

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SMO	n.t.b					Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	n.t.b					Dentin dysplasia, type IA, 125400
SNAI2	n.t.b					
SNAP29	n.t.b					Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNRPE	n.t.b					Hypotrichosis 11, 615059
SNX10	n.t.b					Osteopetrosis, autosomal recessive 8, 615085
SOS1	n.t.b					Noonan syndrome 4, 610733;Fibromatosis, gingival, 1, 135300
SOX10	n.t.b					Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX18	n.t.b					Hypotrichosis-lymphedema-telangiectasia syndrome, 607823;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	n.t.b					Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900

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SP7	n.t.b					Osteogenesis imperfecta, type XII, 613849
SPINK5	n.t.b					Netherton syndrome, 256500
SPINT2	n.t.b					Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPRED1	n.t.b					Legius syndrome, 611431
SPRY4	n.t.b					Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRD5A3	n.t.b					Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379
SREBF1	n.t.b					Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016;Mucoepithelial dysplasia, hereditary, 158310
ST14	n.t.b					Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL5	n.t.b					Salt and pepper developmental regression syndrome, 609056
STAMBP	n.t.b					Microcephaly-capillary malformation syndrome, 614261
STAT3	n.t.b					Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952

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STAT5B	n.t.b					Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985
STIM1	n.t.b					Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783
STING1	n.t.b					STING-associated vasculopathy, infantile-onset, 615934
STK11	n.t.b					Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300
STK4	n.t.b					T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STS	n.t.b					Ichthyosis, X-linked, 308100
SUFU	n.t.b					{Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255
SULT2B1	n.t.b					Ichthyosis, congenital, autosomal recessive 14, 617571

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SUMF1	n.t.b					Multiple sulfatase deficiency, 272200
TAB3	n.t.b					
TALDO1	n.t.b					Transaldolase deficiency, 606003
TAP1	n.t.b					MHC class I deficiency 1, 604571
TAP2	n.t.b					MHC class I deficiency 2, 620813
TAPBP	n.t.b					?MHC class I deficiency 3, 620814
TARS1	n.t.b					Trichothiodystrophy 7, nonphotosensitive, 618546
TAT	n.t.b					Tyrosinemia, type II, 276600
TBC1D24	n.t.b					Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500
TBX3	n.t.b					Ulnar-mammary syndrome, 181450
TCHH	n.t.b					?Uncombable hair syndrome 3, 617252
TCIRG1	n.t.b					Osteopetrosis, autosomal recessive 1, 259700

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TEK	n.t.b					Venous malformations, multiple cutaneous and mucosal, 600195;Glaucoma 3, primary congenital, E, 617272
TERC	n.t.b					Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	n.t.b					
TERT	n.t.b					Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626
TFAP2A	n.t.b					Branchiooculofacial syndrome, 113620
TFE3	n.t.b					Intellectual developmental disorder, X-linked syndromic, with pigmentary mosaicism and coarse facies, 301066;Renal cell carcinoma, papillary, 1, 300854
TGFB2	n.t.b					Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631

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TGFBR1	n.t.b					{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	n.t.b					Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239
TGM1	n.t.b					Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	n.t.b					?Uncombable hair syndrome 2, 617251
TGM5	n.t.b					Peeling skin syndrome 2, 609796
TINF2	n.t.b					Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
TMC6	n.t.b					{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400
TMC8	n.t.b					{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231
TMEM165	n.t.b					Congenital disorder of glycosylation, type IIk, 614727
TNFRSF11A	n.t.b					Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810
TNFRSF11B	n.t.b					Paget disease of bone 5, juvenile-onset, 239000

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TNFRSF1A	n.t.b					{Multiple sclerosis, susceptibility to, 5}, 614810;Periodic fever, familial, 142680
TNFSF11	n.t.b					Osteopetrosis, autosomal recessive 2, 259710
TNXB	n.t.b					Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963
TP63	n.t.b					Premature ovarian failure 21, 620311;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292;Hay-Wells syndrome, 106260;Split-hand/foot malformation 4, 605289;Orofacial cleft 8, 618149;Rapp-Hodgkin syndrome, 129400;ADULT syndrome, 103285;Limb-mammary syndrome, 603543
TPCN2	n.t.b					[Skin/hair/eye pigmentation 10, blond/brown hair], 612267
TREX1	n.t.b					Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448

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TRIM32	n.t.b					?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM37	n.t.b					Mulibrey nanism, 253250
TRPM4	n.t.b					Progressive familial heart block, type IB, 604559;Erythrokeratodermia variabilis et progressiva 6, 618531
TRPS1	n.t.b					Trichorhinophalangeal syndrome, type III, 190351;Trichorhinophalangeal syndrome, type I, 190350
TRPV3	n.t.b					?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594
TSC1	n.t.b					Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangioleiomyomatosis, 606690
TSC2	n.t.b					Lymphangioleiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254
TSPEAR	n.t.b					Tooth agenesis, selective, 10, 620173;?Deafness, autosomal recessive 98, 614861;Ectodermal dysplasia 14, hypohidrotic/hair/tooth/nail type, 618180

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TTI2	n.t.b					Intellectual developmental disorder, autosomal recessive 39, 615541
TUFT1	n.t.b					Woolly hair-skin fragility syndrome, 620415
TWIST2	n.t.b					Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885;Focal facial dermal dysplasia 3, Setleis type, 227260
TYR	n.t.b					[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800;[Skin/hair/eye pigmentation 3, blue/green eyes], 601800;{Melanoma, cutaneous malignant, susceptibility to, 8}, 601800;Albinism, oculocutaneous, type IB, 606952;Albinism, oculocutaneous, type IA, 203100
TYRP1	n.t.b					[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290
UBA2	n.t.b					ACCES syndrome, 619959
UBE2A	n.t.b					Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860
UBR1	n.t.b					Johanson-Blizzard syndrome, 243800
UGCG	n.t.b					

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UROD	n.t.b					Porphyria, hepatoerythropoietic, 176100;Porphyria cutanea tarda, 176100
UROS	n.t.b					Porphyria, congenital erythropoietic, 263700
USB1	n.t.b					Poikiloderma with neutropenia, 604173
UVSSA	n.t.b					UV-sensitive syndrome 3, 614640
VANGL2	n.t.b					Neural tube defects, 182940
VDR	n.t.b					Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	n.t.b					Lymphatic malformation 4, 615907
VHL	n.t.b					Hemangioblastoma, cerebellar, somatic;Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300
VIPAS39	n.t.b					Arthrogryposis, renal dysfunction, and cholestasis 2, 613404
VPS13B	n.t.b					Cohen syndrome, 216550
VPS33B	n.t.b					Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085

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WAS	n.t.b					Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900
WDR19	n.t.b					Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867
WDR35	n.t.b					Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WDR72	n.t.b					Amelogenesis imperfecta, type IIA3, 613211
WIPF1	n.t.b					Wiskott-Aldrich syndrome 2, 614493
WNT10A	n.t.b					Schopf-Schulz-Passarge syndrome, 224750;Tooth agenesis, selective, 4, 150400;Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980
WNT10B	n.t.b					Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300
WNT5A	n.t.b					Robinow syndrome, autosomal dominant 1, 180700

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WNT7A	n.t.b					Fuhrmann syndrome, 228930;Ulna and fibula, absence of, with severe limb deficiency, 276820;?Santos syndrome, 613005
WRAP53	n.t.b					Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	n.t.b					Werner syndrome, 277700
XPA	n.t.b					Xeroderma pigmentosum, group A, 278700
XPC	n.t.b					Xeroderma pigmentosum, group C, 278720
XYLT1	n.t.b					Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
XYLT2	n.t.b					{Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822
YWHAZ	n.t.b					Popov-Chang syndrome, 618428
ZBTB20	n.t.b					Primrose syndrome, 259050
ZMPSTE24	n.t.b					Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZNF469	n.t.b					Brittle cornea syndrome 1, 229200
ZNF592	n.t.b					
ZNF750	n.t.b					?Seborrhea-like dermatitis with psoriasiform elements, 610227

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Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors