SKIN DISORDERS GENE PANEL DG 3.4.0 (630 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	100,0%	100,0%	Achalasia-addisonianism-alacrimia syndrome, 231550
AAGAB	100,0%	100,0%	Keratoderma, palmoplantar, punctate type IA, 148600
ABCA12	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 Ichthyosis, congenital, autosomal recessive 4A, 601277
ABCB6	100,0%	100,0%	Microphthalmia, isolated, with coloboma 7, 614497 Dyschromatosis universalis hereditaria 3, 615402 Pseudohyperkalemia, familial, 2, due to red cell leak, 609153
ABCC6	100,0%	100,0%	Pseudoxanthoma elasticum, 264800 Arterial calcification, generalized, of infancy, 2, 614473 Pseudoxanthoma elasticum, forme fruste, 177850
ABCC9	100,0%	100,0%	Cardiomyopathy, dilated, 1O, 608569 Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 ?Atrial fibrillation, familial, 12, 614050 Intellectual disability and myopathy syndrome, 619719
ABHD5	100,0%	100,0%	Chanarin-Dorfman syndrome, 275630
ACD	100,0%	100,0%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
ACTA2	100,0%	100,0%	Multisystemic smooth muscle dysfunction syndrome, 613834 Aortic aneurysm, familial thoracic 6, 611788 Moyamoya disease 5, 614042
ACTB	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACVRL1	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 2, 600376
ADA2	100,0%	100,0%	Sneddon syndrome, 182410 Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688
ADAM10	100,0%	100,0%	Reticulate acropigmentation of Kitamura, 615537
ADAM17	100,0%	100,0%	?Inflammatory skin and bowel disease, neonatal, 1, 614328
ADAMTS10	100,0%	100,0%	Weill-Marchesani syndrome 1, recessive, 277600
ADAMTS17	100,0%	99,8%	Weill-Marchesani 4 syndrome, recessive, 613195

ADAMTS2	98,1%	98,1%	Ehlers-Danlos syndrome, dermatosparaxis type, 225410
ADAMTS3	100,0%	100,0%	Hennekam lymphangiectasia-lymphedema syndrome 3, 618154
ADAMTSL2	99,9%	99,7%	Geleophysic dysplasia 1, 231050
ADAR	100,0%	100,0%	Dyschromatosis symmetrica hereditaria, 127400 Aicardi-Goutieres syndrome 6, 615010
AGA	100,0%	100,0%	Aspartylglucosaminuria, 208400
AGPAT2	100,0%	100,0%	Lipodystrophy, congenital generalized, type 1, 608594
AIRE	100,0%	100,0%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKT1	100,0%	100,0%	Breast cancer, somatic, 114480 Cowden syndrome 6, 615109 Colorectal cancer, somatic, 114500 Proteus syndrome, somatic, 176920 Ovarian cancer, somatic, 167000
AKT3	100,0%	100,0%	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937
ALAD	100,0%	100,0%	Porphyria, acute hepatic, 612740
ALAS2	100,0%	100,0%	Anemia, sideroblastic, 1, 300751 Protoporphyria, erythropoietic, X-linked, 300752
ALDH18A1	100,0%	100,0%	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	93,2%	93,2%	Sjogren-Larsson syndrome, 270200
ALDOB	100,0%	100,0%	Fructose intolerance, hereditary, 229600
ALOX12B	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 2, 242100
ALOXE3	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 3, 606545
ALPL	100,0%	100,0%	Odontohypophosphatasia, 146300 Hypophosphatasia, infantile, 241500 Hypophosphatasia, childhood, 241510 Hypophosphatasia, adult, 146300
ALX4	100,0%	100,0%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMELX	100,0%	100,0%	Amelogenesis imperfecta, type 1E, 301200
ANKRD11	100,0%	100,0%	KBG syndrome, 148050
ANOS1	100,0%	100,0%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
ANTXR1	100,0%	100,0%	GAPO syndrome, 230740
ANTXR2	100,0%	100,0%	Hyaline fibromatosis syndrome, 228600
AP1B1	100,0%	100,0%	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150

AP1S3	90,5%	90,5%	No OMIM Disease ID
AP3B1	100,0%	100,0%	Hermansky-Pudlak syndrome 2, 608233
APC	100,0%	100,0%	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	100,0%	100,0%	Hypotrichosis 1, 605389
AQP5	100,0%	100,0%	Palmoplantar keratoderma, Bothnian type, 600231
ARHGAP31	100,0%	100,0%	Adams-Oliver syndrome 1, 100300
ARID1A	100,0%	100,0%	Coffin-Siris syndrome 2, 614607
ARID1B	98,6%	98,3%	Coffin-Siris syndrome 1, 135900
ASIP	100,0%	100,0%	No OMIM Disease ID
ASL	100,0%	100,0%	Argininosuccinic aciduria, 207900
ASPRV1	100,0%	100,0%	Ichthyosis, lamellar, autosomal dominant, 146750
ASXL1	99,9%	99,9%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
ASXL3	100,0%	100,0%	Bainbridge-Ropers syndrome, 615485
ATIC	100,0%	100,0%	AICA-ribosiduria due to ATIC deficiency, 608688
ATP2A2	100,0%	100,0%	Acrokeratosis verruciformis, 101900 Darier disease, 124200
ATP2C1	100,0%	100,0%	Hailey-Hailey disease, 169600
ATP6V0A2	100,0%	100,0%	Wrinkly skin syndrome, 278250 Cutis laxa, autosomal recessive, type IIA, 219200
ATP7A	100,0%	100,0%	Occipital horn syndrome, 304150 Spinal muscular atrophy, distal, X-linked 3, 300489 Menkes disease, 309400
ATR	100,0%	100,0%	Seckel syndrome 1, 210600 ?Cutaneous telangiectasia and cancer syndrome, familial, 614564
AXIN2	100,0%	100,0%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
B3GALT6	99,8%	98,8%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465

B4GALT7	100,0%	100,0%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
BANF1	100,0%	100,0%	Nestor-Guillermo progeria syndrome, 614008
BAP1	100,0%	100,0%	Tumor predisposition syndrome, 614327 Kury-Isidor syndrome, 619762
BCOR	100,0%	100,0%	Microphthalmia, syndromic 2, 300166
BCS1L	100,0%	100,0%	GRACILE syndrome, 603358 Mitochondrial complex III deficiency, nuclear type 1, 124000 Bjornstad syndrome, 262000
BLM	100,0%	100,0%	Bloom syndrome, 210900
BLOC1S3	100,0%	100,0%	Hermansky-Pudlak syndrome 8, 614077
BLOC1S6	100,0%	100,0%	?Hermansky-Pudlak syndrome 9, 614171
BMS1	100,0%	100,0%	?Aplasia cutis congenita, nonsyndromic, 107600
BRAF	100,0%	100,0%	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	100,0%	100,0%	Fanconi anemia, complementation group J, 609054
BSCL2	100,0%	100,0%	Lipodystrophy, congenital generalized, type 2, 269700 Neuropathy, distal hereditary motor, type VC, 619112 Silver spastic paraplegia syndrome, 270685 Encephalopathy, progressive, with or without lipodystrophy, 615924
BTD	83,1%	83,1%	Biotinidase deficiency, 253260
C1QA	100,0%	100,0%	C1q deficiency, 613652
C1QB	100,0%	100,0%	C1q deficiency, 613652
C1QC	100,0%	100,0%	C1q deficiency, 613652
C2CD3	95,9%	95,9%	Orofaciodigital syndrome XIV, 615948
CA2	100,0%	100,0%	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730
CAPN12	100,0%	100,0%	No OMIM Disease ID
CARD14	100,0%	100,0%	Psoriasis 2, 602723 Pityriasis rubra pilaris, 173200
CARD9	100,0%	100,0%	Candidiasis, familial, 2, autosomal recessive, 212050
CARMIL2	100,0%	100,0%	Immunodeficiency 58, 618131
CASP14	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 12, 617320
CAST	100,0%	100,0%	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295

CAV1	100,0%	100,0%	?Lipodystrophy, congenital generalized, type 3, 612526 Pulmonary hypertension, primary, 3, 615343 Lipodystrophy, familial partial, type 7, 606721
CAVIN1	100,0%	100,0%	Lipodystrophy, congenital generalized, type 4, 613327
CBL	100,0%	100,0%	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 ?Juvenile myelomonocytic leukemia, 607785
CBS	100,0%	100,0%	Thrombosis, hyperhomocysteinemic, 236200 Homocystinuria, B6-responsive and nonresponsive types, 236200
CCBE1	100,0%	100,0%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CD151	100,0%	100,0%	Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057
CDAN1	100,0%	100,0%	Dyserythropoietic anemia, congenital, type Ia, 224120
CDH3	100,0%	100,0%	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280
CDK4	100,0%	100,0%	No OMIM Disease ID
CDKN2A	100,0%	100,0%	No OMIM Disease ID
CDSN	100,0%	100,0%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CELSR1	99,9%	99,9%	Lymphatic malformation 9, 619319
CERS3	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 9, 615023
CHKB	100,0%	100,0%	Muscular dystrophy, congenital, megaconial type, 602541
CHST14	100,0%	100,0%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CHSY1	100,0%	99,9%	Temtamy preaxial brachydactyly syndrome, 605282
CHUK	100,0%	100,0%	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 ?Cocoon syndrome, 613630
CIB1	100,0%	100,0%	Epidermodysplasia verruciformis 3, 618267
CKAP2L	100,0%	100,0%	Filippi syndrome, 272440
CLDN1	100,0%	100,0%	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626
CLDN10	100,0%	100,0%	HELIX syndrome, 617671
CNNM4	100,0%	100,0%	Jalili syndrome, 217080
COL14A1	100,0%	100,0%	No OMIM Disease ID
COL17A1	100,0%	100,0%	Epithelial recurrent erosion dystrophy, 122400 Epidermolysis bullosa, junctional 4, intermediate, 619787
COL1A2	100,0%	100,0%	Osteogenesis imperfecta, type III, 259420 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	100,0%	100,0%	Ehlers-Danlos syndrome, vascular type, 130050 Polymicrogyria with or without vascular-type EDS, 618343
COL5A1	100,0%	100,0%	Ehlers-Danlos syndrome, classic type, 1, 130000 Fibromuscular dysplasia, multifocal, 619329
COL5A2	100,0%	100,0%	Ehlers-Danlos syndrome, classic type, 2, 130010
COL7A1	100,0%	100,0%	Epidermolysis bullosa, pretibial, 131850 Transient bullous of the newborn, 131705 EBD, Bart type, 132000 Epidermolysis bullosa dystrophica, AD, 131750 Epidermolysis bullosa pruriginosa, 604129 EBD inversa, 226600 Epidermolysis bullosa dystrophica, AR, 226600 Toenail dystrophy, isolated, 607523 EBD, localisata variant,
COX4I2	100,0%	100,0%	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714
COX7B	100,0%	100,0%	Linear skin defects with multiple congenital anomalies 2, 300887
CPOX	100,0%	100,0%	Coproporphyria, 121300 Harderoporphyria, 618892
CST6	100,0%	100,0%	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535
CSTA	100,0%	100,0%	Peeling skin syndrome 4, 607936
CTC1	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
CTSA	100,0%	100,0%	Galactosialidosis, 256540
CTSB	100,0%	100,0%	No OMIM Disease ID
CTSC	100,0%	100,0%	Periodontitis 1, juvenile, 170650 Haim-Munk syndrome, 245010 Papillon-Lefevre syndrome, 245000
CXCR4	100,0%	100,0%	WHIM syndrome 1, 193670 Myelokathexis, isolated, 193670
CYLD	100,0%	100,0%	Brooke-Spiegler syndrome, 605041 Cylindromatosis, familial, 132700 Trichoepithelioma, multiple familial, 1, 601606 ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132
CYP26C1	100,0%	100,0%	Focal facial dermal dysplasia 4, 614974
CYP4F22	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 5, 604777
DCAF17	100,0%	100,0%	Woodhouse-Sakati syndrome, 241080

DCLRE1C	100,0%	100,0%	Severe combined immunodeficiency, Athabascan type, 602450 Omenn syndrome, 603554
DDB2	100,0%	100,0%	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740
DHCR7	100,0%	100,0%	Smith-Lemli-Opitz syndrome, 270400
DKC1	100,0%	100,0%	Dyskeratosis congenita, X-linked, 305000
DLX3	100,0%	100,0%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX5	100,0%	100,0%	Split-hand/foot malformation 1, 183600 ?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600
DOCK6	100,0%	100,0%	Adams-Oliver syndrome 2, 614219
DOCK8	100,0%	100,0%	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700
DOLK	100,0%	100,0%	Congenital disorder of glycosylation, type Im, 610768
DSC2	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 Arrhythmogenic right ventricular dysplasia 11, 610476
DSC3	100,0%	100,0%	Hypotrichosis and recurrent skin vesicles, 613102
DSE	100,0%	100,0%	Ehlers-Danlos syndrome, musculocontractural type 2, 615539
DSG1	100,0%	100,0%	Keratosis palmoplantaris striata I, AD, 148700 Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508
DSG3	100,0%	100,0%	Blistering, acantholytic, of oral and laryngeal mucosa, 619226
DSG4	100,0%	100,0%	Hypotrichosis 6, 607903
DSP	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia 8, 607450 Skin fragility-woolly hair syndrome, 607655 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	100,0%	100,0%	Dentinogenesis imperfecta, Shields type III, 125500 Dentinogenesis imperfecta, Shields type II, 125490 Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594
DST	95,6%	95,6%	?Neuropathy, hereditary sensory and autonomic, type VI, 614653 Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425
DTNBP1	100,0%	100,0%	Hermansky-Pudlak syndrome 7, 614076
DUSP6	100,0%	100,0%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
EBP	100,0%	100,0%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
ECM1	100,0%	100,0%	Urbach-Wiethe disease, 247100

EDA	100,0%	100,0%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	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	100,0%	100,0%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDN3	100,0%	100,0%	Waardenburg syndrome, type 4B, 613265
EDNRA	100,0%	100,0%	Mandibulofacial dysostosis with alopecia, 616367
EDNRB	100,0%	100,0%	ABCD syndrome, 600501 Waardenburg syndrome, type 4A, 277580
EFEMP2	100,0%	100,0%	Cutis laxa, autosomal recessive, type IB, 614437
EFNB1	100,0%	100,0%	Craniofrontonasal dysplasia, 304110
EIF2AK3	100,0%	100,0%	Wolcott-Rallison syndrome, 226980
ELN	100,0%	100,0%	Cutis laxa, autosomal dominant, 123700 Supravalvar aortic stenosis, 185500
ELOVL1	100,0%	100,0%	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527
ELOVL4	100,0%	100,0%	Spinocerebellar ataxia 34, 133190 Stargardt disease 3, 600110 Ichthyosis, spastic quadriplegia, and mental retardation, 614457
ENAM	100,0%	100,0%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ENG	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 1, 187300
ENPP1	100,0%	99,9%	Hypophosphatemic rickets, autosomal recessive, 2, 613312 Arterial calcification, generalized, of infancy, 1, 208000 Cole disease, 615522
EPG5	100,0%	100,0%	Vici syndrome, 242840
EPS8L3	100,0%	100,0%	?Hypotrichosis 5, 612841
ERCC2	100,0%	100,0%	Xeroderma pigmentosum, group D, 278730 Trichothiodystrophy 1, photosensitive, 601675 ?Cerebrooculofacioskeletal syndrome 2, 610756
ERCC3	100,0%	100,0%	Trichothiodystrophy 2, photosensitive, 616390 Xeroderma pigmentosum, group B, 610651
ERCC4	100,0%	100,0%	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 XFE progeroid syndrome, 610965 Xeroderma pigmentosum, group F, 278760 Fanconi anemia, complementation group Q, 615272

ERCC5	100,0%	100,0%	Xeroderma pigmentosum, group G, 278780 Cerebrooculofacioskeletal syndrome 3, 616570 Xeroderma pigmentosum, group G/Cockayne syndrome, 278780
ERCC6	100,0%	100,0%	UV-sensitive syndrome 1, 600630 Cerebrooculofacioskeletal syndrome 1, 214150 ?De Sanctis-Cacchione syndrome, 278800 Cockayne syndrome, type B, 133540 Premature ovarian failure 11, 616946
ERCC8	100,0%	100,0%	UV-sensitive syndrome 2, 614621 Cockayne syndrome, type A, 216400
EVC	100,0%	99,8%	Ellis-van Creveld syndrome, 225500 ?Weyers acrofacial dysostosis, 193530
EVC2	100,0%	100,0%	Ellis-van Creveld syndrome, 225500 Weyers acrofacial dysostosis, 193530
EXPH5	100,0%	100,0%	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028
F13A1	100,0%	100,0%	Factor XIIIA deficiency, 613225
FAM111B	100,0%	100,0%	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704
FAM20A	100,0%	100,0%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM20C	100,0%	100,0%	Raine syndrome, 259775
FAM83G	100,0%	100,0%	No OMIM Disease ID
FAM83H	100,0%	100,0%	Amelogenesis imperfecta, type IIIA, 130900
FANCA	100,0%	100,0%	Fanconi anemia, complementation group A, 227650
FANCB	100,0%	100,0%	Fanconi anemia, complementation group B, 300514
FANCC	97,3%	97,3%	Fanconi anemia, complementation group C, 227645
FANCD2	98,8%	98,8%	Fanconi anemia, complementation group D2, 227646
FANCE	100,0%	100,0%	Fanconi anemia, complementation group E, 600901
FANCF	100,0%	100,0%	Fanconi anemia, complementation group F, 603467
FANCG	100,0%	100,0%	Fanconi anemia, complementation group G, 614082
FANCI	100,0%	100,0%	Fanconi anemia, complementation group I, 609053
FANCL	100,0%	100,0%	Fanconi anemia, complementation group L, 614083
FANCM	100,0%	100,0%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FAT4	100,0%	100,0%	Van Maldergem syndrome 2, 615546 Hennekam lymphangiectasia-lymphedema syndrome 2, 616006
FBLN5	91,8%	91,8%	Cutis laxa, autosomal recessive, type IA, 219100 Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 Macular degeneration, age-related, 3, 608895

			Neuropathy, hereditary, with or without age-related macular degeneration, 608895 ?Cutis laxa, autosomal dominant 2, 614434
FDPS	100,0%	100,0%	Porokeratosis 9, multiple types, 616631
FECH	100,0%	100,0%	Protoporphyria, erythropoietic, 1, 177000
FERMT1	100,0%	100,0%	Kindler syndrome, 173650
FGF10	100,0%	100,0%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF23	100,0%	100,0%	Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993 Hypophosphatemic rickets, autosomal dominant, 193100
FGF3	100,0%	100,0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF5	100,0%	100,0%	Trichomegaly, 190330
FGF8	100,0%	100,0%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100,0%	100,0%	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	100,0%	100,0%	Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Jackson-Weiss syndrome, 123150 Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Apert syndrome, 101200 Pfeiffer syndrome, 101600 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,
FGFR3	100,0%	100,0%	Muenke syndrome, 602849 SADDAN, 616482 Hypochondroplasia, 146000 LADD syndrome, 149730 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 Achondroplasia, 100800 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247
FH	100,0%	100,0%	Leiomyomatosis and renal cell cancer, 150800 Fumarase deficiency, 606812
FKBP10	100,0%	100,0%	Osteogenesis imperfecta, type XI, 610968 Bruck syndrome 1, 259450
FKBP14	100,0%	100,0%	Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557
FLCN	100,0%	100,0%	Birt-Hogg-Dube syndrome, 135150 Colorectal cancer, somatic, 114500 Pneumothorax, primary spontaneous, 173600 Renal carcinoma, chromophobe, somatic, 144700
FLG	100,0%	100,0%	Ichthyosis vulgaris, 146700
FLG2	99,9%	99,9%	Peeling skin syndrome 6, 618084
FLT4	100,0%	100,0%	Hemangioma, capillary infantile, somatic, 602089 Lymphatic malformation 1, 153100 Congenital heart defects, multiple types, 7, 618780
FNIP1	100,0%	100,0%	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705
FOXC2	100,0%	100,0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	100,0%	100,0%	Bamforth-Lazarus syndrome, 241850
FOXN1	100,0%	100,0%	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705
FOXP3	100,0%	100,0%	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790
FREM1	100,0%	100,0%	Manitoba oculotrichoanal syndrome, 248450 Bifid nose with or without anorectal and renal anomalies, 608980 Trigonocephaly 2, 614485
FUCA1	100,0%	100,0%	Fucosidosis, 230000
FZD6	100,0%	100,0%	Nail disorder, nonsyndromic congenital, 1, 161050
GALNS	100,0%	100,0%	Mucopolysaccharidosis IVA, 253000
GALNT3	100,0%	100,0%	Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900
GAN	100,0%	100,0%	Giant axonal neuropathy-1, 256850

GATA2	100,0%	100,0%	Emberger syndrome, 614038 Immunodeficiency 21, 614172
GDF2	100,0%	100,0%	Telangiectasia, hereditary hemorrhagic, type 5, 615506
GDF5	100,0%	100,0%	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 Brachydactyly, type A1, C, 615072
GGCX	100,0%	100,0%	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450 Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842
GJA1	100,0%	100,0%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GJB2	100,0%	100,0%	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	100,0%	100,0%	Deafness, digenic, GJB2/GJB3, 220290 Deafness, autosomal dominant 2B, 612644 Erythrokeratodermia variabilis et progressiva 1, 133200 Deafness, autosomal recessive, Deafness, autosomal dominant, with peripheral neuropathy,
GJB4	100,0%	100,0%	Erythrokeratodermia variabilis et progressiva 2, 617524
GJB6	100,0%	100,0%	Ectodermal dysplasia 2, Clouston type, 129500 Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290

GJC2	99,9%	99,5%	Lymphatic malformation 3, 613480 ?Spastic paraplegia 44, autosomal recessive, 613206 Leukodystrophy, hypomyelinating, 2, 608804
GLA	91,3%	91,3%	Fabry disease, cardiac variant, 301500 Fabry disease, 301500
GLB1	100,0%	100,0%	GM1-gangliosidosis, type I, 230500 GM1-gangliosidosis, type III, 230650 Mucopolysaccharidosis type IVB (Morquio), 253010 GM1-gangliosidosis, type II, 230600
GLMN	100,0%	100,0%	Glomuvenous malformations, 138000
GMPPA	100,0%	100,0%	Alacrima, achalasia, and mental retardation syndrome, 615510
GNA11	100,0%	100,0%	Hypocalciuric hypercalcemia, type II, 145981 Hypocalcemia, autosomal dominant 2, 615361
GNA14	100,0%	100,0%	No OMIM Disease ID
GNAQ	100,0%	100,0%	Capillary malformations, congenital, 1, somatic, mosaic, 163000 Sturge-Weber syndrome, somatic, mosaic, 185300
GNAS	83,9%	82,0%	ACTH-independent macronodular adrenal hyperplasia, 219080 Pituitary adenoma 3, multiple types, somatic, 617686 Pseudohypoparathyroidism Ic, 612462 Pseudohypoparathyroidism Ia, 103580 Osseous heteroplasia, progressive, 166350 Pseudohypoparathyroidism Ib, 603233 McCune-Albright syndrome, somatic, mosaic, 174800 Pseudopseudohypoparathyroidism, 612463
GORAB	100,0%	100,0%	Geroderma osteodysplasticum, 231070
GPNMB	95,5%	95,5%	Amyloidosis, primary localized cutaneous, 3, 617920
GPR143	100,0%	100,0%	Ocular albinism, type I, Nettleship-Falls type, 300500 Nystagmus 6, congenital, X-linked, 300814
GRHL2	100,0%	100,0%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
GRHL3	100,0%	100,0%	van der Woude syndrome 2, 606713
GSN	100,0%	100,0%	Amyloidosis, Finnish type, 105120
GTF2E2	100,0%	100,0%	Trichothiodystrophy 6, nonphotosensitive, 616943
GTF2H5	72,5%	72,5%	Trichothiodystrophy 3, photosensitive, 616395
HCCS	100,0%	100,0%	Linear skin defects with multiple congenital anomalies 1, 309801
HDAC8	96,6%	96,0%	Cornelia de Lange syndrome 5, 300882
HERC2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 38, 615516

HLCS	100,0%	100,0%	Holocarboxylase synthetase deficiency, 253270
HMBS	100,0%	100,0%	Porphyria, acute intermittent, nonerythroid variant, 176000 Porphyria, acute intermittent, 176000
HMGB3	100,0%	100,0%	?Microphthalmia, syndromic 13, 300915
HOXC13	100,0%	100,0%	Ectodermal dysplasia 9, hair/nail type, 614931
HPGD	100,0%	100,0%	?Digital clubbing, isolated congenital, 119900 Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 Cranioosteoarthropathy, 259100
HPS1	100,0%	100,0%	Hermansky-Pudlak syndrome 1, 203300
HPS3	100,0%	100,0%	Hermansky-Pudlak syndrome 3, 614072
HPS4	100,0%	100,0%	Hermansky-Pudlak syndrome 4, 614073
HPS5	100,0%	100,0%	Hermansky-Pudlak syndrome 5, 614074
HPS6	100,0%	100,0%	Hermansky-Pudlak syndrome 6, 614075
HR	100,0%	100,0%	Atrichia with papular lesions, 209500 Alopecia universalis, 203655
HRAS	100,0%	100,0%	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	90,4%	90,3%	CARASIL syndrome, 600142 Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779
HYAL1	100,0%	100,0%	Mucopolysaccharidosis type IX, 601492
IDUA	100,0%	100,0%	Mucopolysaccharidosis Is, 607016 Mucopolysaccharidosis Ih/s, 607015 Mucopolysaccharidosis Ih, 607014
IFT122	100,0%	100,0%	Cranioectodermal dysplasia 1, 218330
IFT43	100,0%	100,0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IKBKG	100,0%	100,0%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636
IL17RA	100,0%	100,0%	Immunodeficiency 51, 613953
IL17RD	100,0%	100,0%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IL1RN	100,0%	100,0%	Interleukin 1 receptor antagonist deficiency, 612852

IL31RA	100,0%	100,0%	?Amyloidosis, primary localized cutaneous, 2, 613955
IL36RN	100,0%	100,0%	Psoriasis 14, pustular, 614204
INSR	100,0%	100,0%	Rabson-Mendenhall syndrome, 262190 Leprechaunism, 246200 Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 Hyperinsulinemic hypoglycemia, familial, 5, 609968
IRF4	100,0%	100,0%	No OMIM Disease ID
IRF6	100,0%	100,0%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome 1, 119300
ISG15	100,0%	100,0%	Immunodeficiency 38, 616126
ITGA3	100,0%	100,0%	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748
ITGA6	100,0%	100,0%	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817
ITGB4	100,0%	100,0%	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 Epidermolysis bullosa, junctional 5A, intermediate, 619816
ITGB6	100,0%	100,0%	Amelogenesis imperfecta, type IH, 616221
JUP	100,0%	100,0%	Naxos disease, 601214 ?Arrhythmogenic right ventricular dysplasia 12, 611528
KANK2	100,0%	100,0%	Nephrotic syndrome, type 16, 617783 Palmoplantar keratoderma and woolly hair, 616099
KAT6B	100,0%	100,0%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KCNH1	98,7%	98,7%	Zimmermann-Laband syndrome 1, 135500 Temple-Baraitser syndrome, 611816
KCNK9	97,3%	97,3%	Birk-Barel syndrome, 612292
KDF1	100,0%	100,0%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDSR	100,0%	100,0%	Erythrokeratodermia variabilis et progressiva 4, 617526
KIF11	100,0%	100,0%	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950
KIT	100,0%	100,0%	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	100,0%	100,0%	Hyperpigmentation with or without hypopigmentation, 145250 Deafness, autosomal dominant 69, unilateral or asymmetric, 616697
KLHL24	100,0%	100,0%	Epidermolysis bullosa simplex 6, generalized, with scarring and hair loss, 617294
KLK4	100,0%	100,0%	Amelogenesis imperfecta, type IIA1, 204700

KLLN	100,0%	100,0%	Cowden syndrome 4, 615107
KMT2D	100,0%	100,0%	Kabuki syndrome 1, 147920
KRAS	100,0%	100,0%	Gastric cancer, somatic, 613659 Oculoectodermal syndrome, somatic, 600268 Breast cancer, somatic, 114480 Noonan syndrome 3, 609942 RAS-associated autoimmune leukoproliferative disorder, 614470 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 Bladder cancer, somatic, 109800
KRT1	100,0%	100,0%	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 Epidermolytic hyperkeratosis, 113800 Palmoplantar keratoderma, nonepidermolytic, 600962 Keratosis palmoplantaris striata III, 607654 Palmoplantar keratoderma, epidermolytic, 144200 Ichthyosis histrix, Curth-Macklin type, 146590
KRT10	100,0%	100,0%	Epidermolytic hyperkeratosis, 113800 Ichthyosis with confetti, 609165 Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602
KRT13	100,0%	100,0%	White sponge nevus 2, 615785
KRT14	100,0%	100,0%	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-Franceschetti-Jadassohn syndrome, 161000 Epidermolysis bullosa simplex 1B, generalized intermediate, 131900
KRT16	100,0%	100,0%	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 Pachyonychia congenita 1, 167200
KRT17	100,0%	100,0%	Steatocystoma multiplex, 184500 Pachyonychia congenita 2, 167210
KRT2	100,0%	100,0%	Ichthyosis bullosa of Siemens, 146800
KRT4	100,0%	100,0%	White sponge nevus 1, 193900
KRT5	100,0%	100,0%	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	100,0%	100,0%	Pachyonychia congenita 3, 615726
KRT6B	100,0%	100,0%	Pachyonychia congenita 4, 615728
KRT6C	100,0%	99,9%	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735
KRT71	100,0%	100,0%	?Hypotrichosis 13, 615896
KRT74	100,0%	100,0%	Woolly hair, autosomal dominant, 194300 ?Hypotrichosis 3, 613981 ?Ectodermal dysplasia 7, hair/nail type, 614929
KRT75	100,0%	100,0%	No OMIM Disease ID
KRT81	100,0%	100,0%	Monilethrix, 158000
KRT83	100,0%	100,0%	Monilethrix, 158000 Erythrokeratodermia variabilis et progressiva 5, 617756
KRT85	100,0%	100,0%	Ectodermal dysplasia 4, hair/nail type, 602032
KRT86	100,0%	100,0%	Monilethrix, 158000
KRT9	100,0%	100,0%	Palmoplantar keratoderma, epidermolytic, 144200
LAMA3	100,0%	100,0%	Epidermolysis bullosa, junctional 2A, intermediate, 619783 Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660 Epidermolysis bullosa, junctional 2B, severe, 619784
LAMB3	100,0%	100,0%	Epidermolysis bullosa, junctional 1B, severe, 226700 Epidermolysis bullosa, junctional 1A, intermediate, 226650 Amelogenesis imperfecta, type IA, 104530
LAMC2	100,0%	100,0%	Epidermolysis bullosa, junctional 3B, severe, 619786 Epidermolysis bullosa, junctional 3A, intermediate, 619785
LAMTOR2	100,0%	100,0%	Immunodeficiency due to defect in MAPBP-interacting protein, 610798
LDHA	100,0%	100,0%	Glycogen storage disease XI, 612933
LDLRAP1	100,0%	100,0%	Hypercholesterolemia, familial, 4, 603813
LEMD3	100,0%	100,0%	Buschke-Ollendorff syndrome, 166700 Osteopoikilosis with or without melorheostosis, 166700
LIPH	100,0%	100,0%	Hypotrichosis 7, 604379 Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379
LIPN	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 8, 613943
LMBRD1	96,1%	96,1%	Methylmalonic aciduria and homocystinuria, cblF type, 277380

LMNA	100,0%	100,0%	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	100,0%	100,0%	Focal segmental glomerulosclerosis 10, 256020 Nail-patella syndrome, 161200
LONP1	100,0%	100,0%	CODAS syndrome, 600373
LORICRIN	100,0%	100,0%	Vohwinkel syndrome with ichthyosis, 604117
LPAR6	100,0%	100,0%	Hypotrichosis 8, 278150 Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150
LPIN2	100,0%	100,0%	Majeed syndrome, 609628
LRMDA	99,6%	99,6%	Albinism, oculocutaneous, type VII, 615179
LSS	100,0%	100,0%	Hypotrichosis 14, 618275 Cataract 44, 616509 Alopecia-intellectual disability syndrome 4, 618840
LTBP3	100,0%	100,0%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
LTBP4	100,0%	100,0%	Cutis laxa, autosomal recessive, type IC, 613177
LYST	100,0%	100,0%	Chediak-Higashi syndrome, 214500
LYZ	100,0%	100,0%	Amyloidosis, renal, 105200
MAP2K1	100,0%	100,0%	Cardiofaciocutaneous syndrome 3, 615279 Melorheostosis, isolated, somatic mosaic, 155950
MAP2K2	100,0%	100,0%	Cardiofaciocutaneous syndrome 4, 615280
MBTPS2	100,0%	100,0%	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
MED12	100,0%	100,0%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Hardikar syndrome, 301068 Opitz-Kaveggia syndrome, 305450

MEFV	96,4%	96,4%	Neutrophilic dermatosis, acute febrile, 608068 Familial Mediterranean fever, AR, 249100 Familial Mediterranean fever, AD, 134610
MGP	100,0%	100,0%	Keutel syndrome, 245150
MITF	100,0%	100,0%	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306
MLH1	100,0%	100,0%	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 Muir-Torre syndrome, 158320 Mismatch repair cancer syndrome 1, 276300
MLPH	100,0%	100,0%	Griscelli syndrome, type 3, 609227
MMACHC	100,0%	100,0%	Methylmalonic aciduria and homocystinuria, cblC type, 277400
MMP2	100,0%	100,0%	Multicentric osteolysis, nodulosis, and arthropathy, 259600
MMP20	100,0%	100,0%	Amelogenesis imperfecta, type IIA2, 612529
MPLKIP	100,0%	100,0%	Trichothiodystrophy 4, nonphotosensitive, 234050
MRE11	100,0%	100,0%	Ataxia-telangiectasia-like disorder 1, 604391
MSH2	100,0%	100,0%	Muir-Torre syndrome, 158320 Colorectal cancer, hereditary nonpolyposis, type 1, 120435 Mismatch repair cancer syndrome 2, 619096
MSX1	100,0%	100,0%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874
MTOR	100,0%	100,0%	Focal cortical dysplasia, type II, somatic, 607341 Smith-Kingsmore syndrome, 616638
MUTYH	100,0%	100,0%	Adenomas, multiple colorectal, 608456 Gastric cancer, somatic, 613659
MVD	100,0%	100,0%	Porokeratosis 7, multiple types, 614714
MVK	90,5%	90,5%	Hyper-IgD syndrome, 260920 Porokeratosis 3, multiple types, 175900 Mevalonic aciduria, 610377
MYH8	100,0%	100,0%	Carney complex variant, 608837 Trismus-pseudocamptodactyly syndrome, 158300
MYO5A	100,0%	100,0%	Griscelli syndrome, type 1, 214450
NAA10	100,0%	100,0%	Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855

NAGA	100,0%	100,0%	Schindler disease, type I, 609241 Kanzaki disease, 609242 Schindler disease, type III, 609241
NBAS	100,0%	100,0%	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800 Infantile liver failure syndrome 2, 616483
NCF1	100,0%	100,0%	Chronic granulomatous disease 1, autosomal recessive, 233700
NCSTN	100,0%	100,0%	Acne inversa, familial, 1, 142690
NDUFB11	100,0%	99,9%	Linear skin defects with multiple congenital anomalies 3, 300952 ?Mitochondrial complex I deficiency, nuclear type 30, 301021
NECTIN1	100,0%	100,0%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NECTIN4	100,0%	100,0%	Ectodermal dysplasia-syndactyly syndrome 1, 613573
NEK11	100,0%	100,0%	No OMIM Disease ID
NEK9	100,0%	100,0%	?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262 Nevus comedonicus, somatic, 617025 Lethal congenital contracture syndrome 10, 617022
NF1	100,0%	100,0%	Watson syndrome, 193520 Leukemia, juvenile myelomonocytic, 607785 Neurofibromatosis, familial spinal, 162210 Neurofibromatosis, type 1, 162200 Neurofibromatosis-Noonan syndrome, 601321
NFKBIA	100,0%	100,0%	Ectodermal dysplasia and immunodeficiency 2, 612132
NHP2	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 2, 613987
NIPAL4	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 6, 612281
NIPBL	100,0%	100,0%	Cornelia de Lange syndrome 1, 122470
NLRP1	100,0%	100,0%	?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 Autoinflammation with arthritis and dyskeratosis, 617388 Palmoplantar carcinoma, multiple self-healing, 615225
NLRP12	100,0%	100,0%	Familial cold autoinflammatory syndrome 2, 611762
NLRP3	100,0%	100,0%	CINCA syndrome, 607115 Familial cold inflammatory syndrome 1, 120100 Keratoendothelitis fugax hereditaria, 148200 Deafness, autosomal dominant 34, with or without inflammation, 617772 Muckle-Wells syndrome, 191900
NME1	100,0%	100,0%	No OMIM Disease ID
NOD2	100,0%	100,0%	Blau syndrome, 186580
NOP10	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 1, 224230

NOTCH1	100,0%	100,0%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
NRAS	100,0%	100,0%	Noonan syndrome 6, 613224 ?RAS-associated autoimmune lymphoproliferative syndrome type IV, 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	100,0%	100,0%	Sotos syndrome, 117550
NSDHL	100,0%	100,0%	CK syndrome, 300831 CHILD syndrome, 308050
OCA2	100,0%	100,0%	Albinism, brown oculocutaneous, 203200 Albinism, oculocutaneous, type II, 203200
ODAM	100,0%	100,0%	No OMIM Disease ID
ODAPH	100,0%	100,0%	Amelogenesis imperfecta, type IIA4, 614832
OFD1	100,0%	100,0%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OSMR	100,0%	100,0%	Amyloidosis, primary localized cutaneous, 1, 105250
PADI3	100,0%	100,0%	Uncombable hair syndrome, 191480
PAH	100,0%	100,0%	Phenylketonuria, 261600
PALB2	100,0%	100,0%	Fanconi anemia, complementation group N, 610832
PAX3	100,0%	100,0%	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220
PAX9	100,0%	100,0%	Tooth agenesis, selective, 3, 604625
PCNA	100,0%	100,0%	?Ataxia-telangiectasia-like disorder 2, 615919
PDGFB	100,0%	100,0%	Meningioma, SIS-related, 607174 Basal ganglia calcification, idiopathic, 5, 615483 Dermatofibrosarcoma protuberans, 607907
PDGFRB	100,0%	100,0%	Premature aging syndrome, Penttinen type, 601812 Kosaki overgrowth syndrome, 616592 Myofibromatosis, infantile, 1, 228550 Basal ganglia calcification, idiopathic, 4, 615007

PEPD	100,0%	100,0%	Prolidase deficiency, 170100
PERP	100,0%	100,0%	Erythrokeratodermia variabilis et progressiva 7, 619209 Olmsted syndrome 2, 619208
PEX7	91,3%	91,3%	Rhizomelic chondrodysplasia punctata, type 1, 215100 Peroxisome biogenesis disorder 9B, 614879
PHEX	100,0%	99,9%	Hypophosphatemic rickets, X-linked dominant, 307800
PHGDH	100,0%	100,0%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PHYH	100,0%	100,0%	Refsum disease, 266500
PIEZO1	100,0%	100,0%	Lymphatic malformation 6, 616843 Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380
PIGA	100,0%	100,0%	Paroxysmal nocturnal hemoglobinuria, somatic, 300818 Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868 Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072
PIGN	98,8%	98,8%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PIK3CA	100,0%	100,0%	CLOVE syndrome, somatic, 612918 Hepatocellular carcinoma, somatic, 114550 Breast cancer, somatic, 114480 Ovarian cancer, somatic, 167000 Colorectal cancer, somatic, 114500 CLAPO syndrome, somatic, 613089 Keratosis, seborrheic, somatic, 182000 Nevus, epidermal, somatic, 162900 Gastric cancer, somatic, 613659 Nonsmall cell lung cancer, somatic, 211980 Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501 Cowden syndrome 5, 615108 Macrodactyly, somatic,,
PITX2	100,0%	100,0%	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PKP1	100,0%	100,0%	Ectodermal dysplasia/skin fragility syndrome, 604536
PLCD1	100,0%	100,0%	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600
PLCG2	100,0%	100,0%	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 Familial cold autoinflammatory syndrome 3, 614468
PLEC	100,0%	100,0%	?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	100,0%	100,0%	Dysplasminogenemia, 217090 Angioedema, hereditary, 4, 619360 Plasminogen deficiency, type I, 217090
PLIN1	100,0%	100,0%	Lipodystrophy, familial partial, type 4, 613877
PLOD1	100,0%	100,0%	Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400
PLOD3	100,0%	100,0%	Lysyl hydroxylase 3 deficiency, 612394
PMS2	100,0%	100,0%	Colorectal cancer, hereditary nonpolyposis, type 4, 614337 Mismatch repair cancer syndrome 4, 619101
PMVK	100,0%	100,0%	Porokeratosis 1, multiple types, 175800
PNPLA1	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 10, 615024
PNPLA2	100,0%	100,0%	Neutral lipid storage disease with myopathy, 610717
POC1A	100,0%	100,0%	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813
POFUT1	100,0%	100,0%	Dowling-Degos disease 2, 615327
POGLUT1	100,0%	100,0%	Dowling-Degos disease 4, 615696 ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232
POLD1	100,0%	100,0%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381
POLH	100,0%	100,0%	Xeroderma pigmentosum, variant type, 278750
POLR1C	83,0%	82,8%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	100,0%	100,0%	Treacher Collins syndrome 2, 613717
POLR3A	100,0%	100,0%	Wiedemann-Rautenstrauch syndrome, 264090 Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100,0%	100,0%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381 Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POMC	100,0%	100,0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POMP	100,0%	100,0%	Proteasome-associated autoinflammatory syndrome 2, 618048 Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952
PORCN	100,0%	100,0%	Focal dermal hypoplasia, 305600
POT1	100,0%	100,0%	No OMIM Disease ID
PPOX	100,0%	100,0%	Porphyria variegata, 176200
PQBP1	100,0%	100,0%	Renpenning syndrome, 309500
PRKAR1A	100,0%	100,0%	Pigmented nodular adrenocortical disease, primary, 1, 610489 Acrodysostosis 1, with or without hormone resistance, 101800 Carney complex, type 1, 160980

			Myxoma, intracardiac, 255960 Adrenocortical tumor, somatic,
PSEN1	100,0%	100,0%	Pick disease, 172700 Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 Dementia, frontotemporal, 600274 ?Acne inversa, familial, 3, 613737 Cardiomyopathy, dilated, 1U, 613694 Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 Alzheimer disease, type 3, 607822
PSENEN	100,0%	100,0%	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736
PSMB8	100,0%	100,0%	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040
PSTPIP1	100,0%	100,0%	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416
PTCH1	100,0%	100,0%	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
PTCH2	100,0%	100,0%	Medulloblastoma, somatic, 155255 Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462
PTDSS1	100,0%	100,0%	Lenz-Majewski hyperostotic dwarfism, 151050
PTEN	100,0%	100,0%	Cowden syndrome 1, 158350 Lhermitte-Duclos disease, 158350 Prostate cancer, somatic, 176807 Macrocephaly/autism syndrome, 605309
PTHLH	100,0%	100,0%	Brachydactyly, type E2, 613382
PTPN11	100,0%	100,0%	Noonan syndrome 1, 163950 LEOPARD syndrome 1, 151100 Metachondromatosis, 156250 Leukemia, juvenile myelomonocytic, somatic, 607785
PTPN14	100,0%	100,0%	Choanal atresia and lymphedema, 613611
PTPRF	100,0%	100,0%	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001
PYCR1	100,0%	100,0%	Cutis laxa, autosomal recessive, type IIIB, 614438 Cutis laxa, autosomal recessive, type IIB, 612940
RAB23	100,0%	100,0%	Carpenter syndrome, 201000
RAB27A	100,0%	100,0%	Griscelli syndrome, type 2, 607624
RAD21	100,0%	100,0%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RAD50	100,0%	100,0%	Nijmegen breakage syndrome-like disorder, 613078

RAF1	100,0%	100,0%	Cardiomyopathy, dilated, 1NN, 615916 Noonan syndrome 5, 611553 LEOPARD syndrome 2, 611554
RAG1	100,0%	100,0%	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	100,0%	100,0%	Severe combined immunodeficiency, B cell-negative, 601457 Combined cellular and humoral immune defects with granulomas, 233650 Omenn syndrome, 603554
RAI1	100,0%	100,0%	Smith-Magenis syndrome, 182290
RBBP8	100,0%	100,0%	Seckel syndrome 2, 606744 Jawad syndrome, 251255 Pancreatic carcinoma, somatic,
RBM28	100,0%	100,0%	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079
RBP4	100,0%	100,0%	Microphthalmia, isolated, with coloboma 10, 616428 Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147
RBPJ	100,0%	100,0%	Adams-Oliver syndrome 3, 614814
RECQL4	100,0%	100,0%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
RHBDF2	100,0%	100,0%	Tylosis with esophageal cancer, 148500
RHOA	80,7%	80,7%	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727
RIN2	100,0%	100,0%	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075
RIPK4	100,0%	100,0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RMRP	NC	NC	Anauxetic dysplasia 1, 607095 Metaphyseal dysplasia without hypotrichosis, 250460 Cartilage-hair hypoplasia, 250250
RNASEH2A	100,0%	100,0%	Aicardi-Goutieres syndrome 4, 610333
RNASEH2B	91,0%	91,0%	Aicardi-Goutieres syndrome 2, 610181
RNASEH2C	100,0%	100,0%	Aicardi-Goutieres syndrome 3, 610329
RNU4ATAC	NC	NC	Roifman syndrome, 616651 Lowry-Wood syndrome, 226960 Microcephalic osteodysplastic primordial dwarfism, type I, 210710
ROGDI	100,0%	100,0%	Kohlschutter-Tonz syndrome, 226750
RPL21	100,0%	100,0%	Hypotrichosis 12, 615885

RSPO1	100,0%	100,0%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RSPO4	100,0%	100,0%	Anonychia congenita, 206800
RTEL1	100,0%	100,0%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
RUNX2	100,0%	100,0%	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
SAMD9	100,0%	100,0%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SAMHD1	100,0%	100,0%	?Chilblain lupus 2, 614415 Aicardi-Goutieres syndrome 5, 612952
SART3	100,0%	100,0%	No OMIM Disease ID
SASH1	100,0%	100,0%	Dyschromatosis universalis hereditaria 1, 127500 ?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373
SAT1	100,0%	100,0%	No OMIM Disease ID
SATB2	100,0%	100,0%	Glass syndrome, 612313
SCN10A	100,0%	100,0%	Episodic pain syndrome, familial, 2, 615551
SCN11A	100,0%	100,0%	Episodic pain syndrome, familial, 3, 615552 Neuropathy, hereditary sensory and autonomic, type VII, 615548
SCN9A	100,0%	100,0%	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	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 13, 617574
SEC23B	100,0%	100,0%	?Cowden syndrome 7, 616858 Dyserythropoietic anemia, congenital, type II, 224100
SERPINA12	100,0%	100,0%	No OMIM Disease ID
SERPINA3	100,0%	100,0%	Alpha-1-antichymotrypsin deficiency, Cerebrovascular disease, occlusive,
SERPINB7	100,0%	100,0%	Palmoplantar keratoderma, Nagashima type, 615598
SERPINB8	100,0%	100,0%	Peeling skin syndrome 5, 617115
SERPING1	100,0%	100,0%	Angioedema, hereditary, 1 and 2, 106100 Complement component 4, partial deficiency of, 120790

SERPINH1	100,0%	100,0%	Osteogenesis imperfecta, type X, 613848
SGPL1	100,0%	100,0%	Nephrotic syndrome, type 14, 617575
SHOC2	100,0%	100,0%	Noonan syndrome-like with loose anagen hair 1, 607721
SKI	100,0%	100,0%	Shprintzen-Goldberg syndrome, 182212
SKIV2L	100,0%	100,0%	Trichohepatoenteric syndrome 2, 614602
SLC17A9	100,0%	100,0%	Porokeratosis 8, disseminated superficial actinic type, 616063
SLC24A4	100,0%	100,0%	Amelogenesis imperfecta, type IIA5, 615887
SLC24A5	100,0%	100,0%	Albinism, oculocutaneous, type VI, 113750
SLC26A2	100,0%	100,0%	Epiphyseal dysplasia, multiple, 4, 226900 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050
SLC27A4	100,0%	100,0%	Ichthyosis prematurity syndrome, 608649
SLC29A3	100,0%	100,0%	Histiocytosis-lymphadenopathy plus syndrome, 602782
SLC2A10	100,0%	100,0%	Arterial tortuosity syndrome, 208050
SLC39A13	100,0%	100,0%	Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350
SLC39A4	100,0%	100,0%	Acrodermatitis enteropathica, 201100
SLC45A2	100,0%	100,0%	Albinism, oculocutaneous, type IV, 606574
SLC4A4	100,0%	100,0%	Renal tubular acidosis, proximal, with ocular abnormalities, 604278
SLC6A19	100,0%	100,0%	Iminoglycinuria, digenic, 242600 Hartnup disorder, 234500 Hyperglycinuria, 138500
SLC7A7	100,0%	100,0%	Lysinuric protein intolerance, 222700
SLCO2A1	100,0%	100,0%	Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100 Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441
SLURP1	100,0%	100,0%	Meleda disease, 248300
SLX4	100,0%	100,0%	Fanconi anemia, complementation group P, 613951
SMAD3	100,0%	100,0%	Loeys-Dietz syndrome 3, 613795
SMARCA2	98,4%	98,2%	Nicolaides-Baraitser syndrome, 601358 Blepharophimosis-impaired intellectual development syndrome, 619293
SMARCA4	100,0%	100,0%	Coffin-Siris syndrome 4, 614609
SMARCAD1	100,0%	100,0%	Basan syndrome, 129200 Huriez syndrome, 181600 Adermatoglyphia, 136000
SMARCAL1	100,0%	100,0%	Schimke immunoosseous dysplasia, 242900

SMARCB1	100,0%	100,0%	Rhabdoid tumors, somatic, 609322 Coffin-Siris syndrome 3, 614608
SMO	100,0%	100,0%	Pallister-Hall-like syndrome, 241800 Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	100,0%	100,0%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	100,0%	100,0%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SNAP29	100,0%	100,0%	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528
SNRPE	100,0%	100,0%	Hypotrichosis 11, 615059
SNX10	100,0%	99,9%	Osteopetrosis, autosomal recessive 8, 615085
SOS1	100,0%	100,0%	Noonan syndrome 4, 610733 ?Fibromatosis, gingival, 1, 135300
SOX10	100,0%	100,0%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX18	99,9%	99,3%	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940
SOX2	100,0%	100,0%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SP7	100,0%	100,0%	Osteogenesis imperfecta, type XII, 613849
SPINK5	100,0%	100,0%	Netherton syndrome, 256500
SPINT2	100,0%	100,0%	Diarrhea 3, secretory sodium, congenital, syndromic, 270420
SPRED1	100,0%	100,0%	Legius syndrome, 611431
SPRY4	100,0%	100,0%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRD5A3	100,0%	100,0%	Kahrizi syndrome, 612713 Congenital disorder of glycosylation, type Iq, 612379
ST14	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 11, 602400
ST3GAL5	98,7%	98,7%	Salt and pepper developmental regression syndrome, 609056
STAMBP	100,0%	100,0%	Microcephaly-capillary malformation syndrome, 614261
STAT3	100,0%	100,0%	Hyper-IgE recurrent infection syndrome, 147060 Autoimmune disease, multisystem, infantile-onset, 1, 615952
STAT5B	100,0%	100,0%	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 Leukemia, acute promyelocytic, somatic, 102578

STIM1	100,0%	100,0%	Myopathy, tubular aggregate, 1, 160565 Stormorken syndrome, 185070 Immunodeficiency 10, 612783
STING1	100,0%	100,0%	STING-associated vasculopathy, infantile-onset, 615934
STK11	100,0%	100,0%	Melanoma, malignant, somatic, 155600 Pancreatic cancer, somatic, 260350 Peutz-Jeghers syndrome, 175200 Testicular tumor, somatic, 273300
STK4	100,0%	100,0%	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868
STS	97,4%	97,3%	Ichthyosis, X-linked, 308100
SUFU	100,0%	100,0%	Joubert syndrome 32, 617757 Medulloblastoma, desmoplastic, 155255 Basal cell nevus syndrome, 109400
SULT2B1	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 14, 617571
SUMF1	100,0%	100,0%	Multiple sulfatase deficiency, 272200
TALDO1	100,0%	100,0%	Transaldolase deficiency, 606003
TAP1	100,0%	100,0%	Bare lymphocyte syndrome, type I, 604571
TAP2	100,0%	100,0%	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571
TAPBP	96,6%	96,6%	Bare lymphocyte syndrome, type I, 604571
TAT	100,0%	100,0%	Tyrosinemia, type II, 276600
TBC1D24	100,0%	100,0%	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	100,0%	100,0%	Ulnar-mammary syndrome, 181450
TCHH	100,0%	100,0%	?Uncombable hair syndrome 3, 617252
TCIRG1	100,0%	100,0%	Osteopetrosis, autosomal recessive 1, 259700
TEK	100,0%	100,0%	Venous malformations, multiple cutaneous and mucosal, 600195 Glaucoma 3, primary congenital, E, 617272
TERC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	83,7%	83,7%	No OMIM Disease ID
TERT	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742
TFAP2A	100,0%	100,0%	Branchiooculofacial syndrome, 113620

TGFB2	100,0%	100,0%	Loeys-Dietz syndrome 4, 614816
TGFBR1	100,0%	99,9%	Loeys-Dietz syndrome 1, 609192
TGFBR2	100,0%	100,0%	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
TGM1	100,0%	100,0%	Ichthyosis, congenital, autosomal recessive 1, 242300
TGM3	100,0%	100,0%	?Uncombable hair syndrome 2, 617251
TGM5	100,0%	100,0%	Peeling skin syndrome 2, 609796
TINF2	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
TMC6	100,0%	100,0%	Epidermodysplasia verruciformis, 226400
TMC8	100,0%	100,0%	Epidermodysplasia verruciformis 2, 618231
TMEM165	100,0%	100,0%	Congenital disorder of glycosylation, type IIk, 614727
TNFRSF11A	100,0%	99,7%	Osteopetrosis, autosomal recessive 7, 612301 Osteolysis, familial expansile, 174810
TNFRSF11B	100,0%	100,0%	Paget disease of bone 5, juvenile-onset, 239000
TNFRSF1A	92,8%	92,8%	Periodic fever, familial, 142680
TNFSF11	100,0%	100,0%	Osteopetrosis, autosomal recessive 2, 259710
TNXB	100,0%	100,0%	Ehlers-Danlos syndrome, classic-like, 1, 606408 Vesicoureteral reflux 8, 615963
TP63	100,0%	100,0%	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	100,0%	100,0%	No OMIM Disease ID
TREX1	100,0%	100,0%	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 Chilblain lupus, 610448
TRIM32	100,0%	100,0%	?Bardet-Biedl syndrome 11, 615988 Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TRIM37	98,7%	98,7%	Mulibrey nanism, 253250
TRPM4	100,0%	100,0%	Progressive familial heart block, type IB, 604559 Erythrokeratodermia variabilis et progressiva 6, 618531

TRPS1	100,0%	100,0%	Trichorhinophalangeal syndrome, type III, 190351 Trichorhinophalangeal syndrome, type I, 190350
TRPV3	97,1%	97,1%	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 Olmsted syndrome 1, 614594
TSC1	100,0%	100,0%	Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-1, 191100 Lymphangioleiomyomatosis, 606690
TSC2	100,0%	100,0%	Lymphangioleiomyomatosis, somatic, 606690 ?Focal cortical dysplasia, type II, somatic, 607341 Tuberous sclerosis-2, 613254
TSPEAR	100,0%	100,0%	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TTC37	100,0%	100,0%	Trichohepatoenteric syndrome 1, 222470
TTI2	100,0%	100,0%	Intellectual developmental disorder, autosomal recessive 39, 615541
TWIST2	100,0%	100,0%	Ablepharon-macrostomia syndrome, 200110 Barber-Say syndrome, 209885 Focal facial dermal dysplasia 3, Setleis type, 227260
TYR	100,0%	100,0%	Albinism, oculocutaneous, type IB, 606952 Waardenburg syndrome/albinism, digenic, 103470 Albinism, oculocutaneous, type IA, 203100
TYRP1	100,0%	100,0%	Albinism, oculocutaneous, type III, 203290
UBE2A	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Nascimento type, 300860
UBR1	98,0%	98,0%	Johanson-Blizzard syndrome, 243800
UROD	100,0%	100,0%	Porphyria, hepatoerythropoietic, 176100 Porphyria cutanea tarda, 176100
UROS	100,0%	100,0%	Porphyria, congenital erythropoietic, 263700
USB1	100,0%	100,0%	Poikiloderma with neutropenia, 604173
UVSSA	100,0%	100,0%	UV-sensitive syndrome 3, 614640
VDR	99,9%	98,7%	Rickets, vitamin D-resistant, type IIA, 277440
VEGFC	100,0%	100,0%	Lymphatic malformation 4, 615907
VHL	100,0%	100,0%	Erythrocytosis, familial, 2, 263400 von Hippel-Lindau syndrome, 193300 Renal cell carcinoma, somatic, 144700 Pheochromocytoma, 171300 Hemangioblastoma, cerebellar, somatic,
VPS13B	99,5%	99,4%	Cohen syndrome, 216550
VPS33B	100,0%	100,0%	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085

WAS	100,0%	100,0%	Wiskott-Aldrich syndrome, 301000 Neutropenia, severe congenital, X-linked, 300299 Thrombocytopenia, X-linked, intermittent, 313900 Thrombocytopenia, X-linked, 313900
WDR19	100,0%	100,0%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR35	100,0%	100,0%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR72	96,9%	96,9%	Amelogenesis imperfecta, type IIA3, 613211
WIPF1	100,0%	100,0%	Wiskott-Aldrich syndrome 2, 614493
WNT10A	100,0%	100,0%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100,0%	100,0%	Tooth agenesis, selective, 8, 617073 Split-hand/foot malformation 6, 225300
WNT5A	100,0%	100,0%	Robinow syndrome, autosomal dominant 1, 180700
WNT7A	100,0%	100,0%	Fuhrmann syndrome, 228930 Ulna and fibula, absence of, with severe limb deficiency, 276820
WRAP53	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 3, 613988
WRN	100,0%	100,0%	Werner syndrome, 277700
XPA	100,0%	100,0%	Xeroderma pigmentosum, group A, 278700
XPC	100,0%	100,0%	Xeroderma pigmentosum, group C, 278720
XYLT1	100,0%	99,7%	Desbuquois dysplasia 2, 615777
XYLT2	96,7%	96,7%	Spondyloocular syndrome, 605822
YWHAZ	100,0%	100,0%	No OMIM Disease ID
ZBTB20	100,0%	100,0%	Primrose syndrome, 259050
ZMPSTE24	100,0%	100,0%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy 1, 275210
ZNF469	100,0%	100,0%	Brittle cornea syndrome 1, 229200
ZNF592	100,0%	100,0%	No OMIM Disease ID
ZNF750	100,0%	100,0%	?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.
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.
TWIST is the chemistry used for WES analysis.

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 coverage denoting NC are non-protein coding genes.
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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors