Skin_Disorders panel

versie	V3 (335 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AAAS	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
AAGAB	614888	Keratoderma, palmoplantar, punctate type IA, 148600 (3), Autosomal dominant
ABCA12	607800	Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive
ABCB6	605452	[Blood group, Langereis system], 111600 (3); Dyschromatosis universalis hereditaria 3, 615402 (3), Autosomal dominant; Microphthalmia, isolated, with coloboma 7, 614497 (3), Autosomal dominant; Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 (3), Autosomal dominant
ABHD5	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ADAM10	602192	{Alzheimer disease 18, susceptibility to}, 615590 (3); Reticulate acropigmentation of Kitamura, 615537 (3), Autosomal dominant
ADAR	146920	Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive; Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant
ADGRE2	606100	Vibratory urticaria, 125630 (3), Autosomal dominant
ALDH3A2	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALOX12B	603741	Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive
ALOXE3	607206	Ichthyosis, congenital, autosomal recessive 3, 606545 (3), Autosomal recessive
ALX4	605420	{Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive; Parietal foramina 2, 609597 (3), Autosomal dominant
ANTXR1	606410	GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
ANTXR2	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive
AP1S1	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
AP3B1	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3D1	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
APCDD1	607479	Hypotrichosis 1, 605389 (3), Autosomal dominant
AQP5	600442	Palmoplantar keratoderma, Bothnian type, 600231 (3), Autosomal dominant
ARHGAP31	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARSL (ARSE)	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ATP2A2	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant

ATP6V1B2	606939	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant; Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant
ATP7A	300011	Menkes disease, 309400 (3), X-linked recessive; Occipital horn syndrome, 304150 (3), X-linked recessive; Spinal muscular atrophy, distal, X-linked 3, 300489 (3), X-linked recessive
ATR	601215	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant; Seckel syndrome 1, 210600 (3), Autosomal recessive
AXIN2	604025	Colorectal cancer, somatic, 114500 (3); Oligodontia-colorectal cancer syndrome, 608615 (3), Autosomal dominant
BANF1	603811	Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive
BCS1L	603647	Bjornstad syndrome, 262000 (3), Autosomal recessive; GRACILE syndrome, 603358 (3), Autosomal recessive; Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive
BLM	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLOC1S3	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S6	604310	?Hermansky-pudlak syndrome 9, 614171 (3), Autosomal recessive
BMS1	611448	?Aplasia cutis congenita, nonsyndromic, 107600 (3), Autosomal dominant
BRAF	164757	Adenocarcinoma of lung, somatic, 211980 (3); Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Colorectal cancer, somatic (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Melanoma, malignant, somatic (3); Nonsmall cell lung cancer, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant
C1QA	120550	C1q deficiency, 613652 (3), Autosomal recessive
C1QB	120570	C1q deficiency, 613652 (3), Autosomal recessive
C1QC	120575	C1q deficiency, 613652 (3), Autosomal recessive
C5	120900	C5 deficiency, 609536 (3); [Eculizumab, poor response to], 615749 (3), Autosomal dominant
CARD11	607210	B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant
CARD14	607211	Pityriasis rubra pilaris, 173200 (3), Autosomal dominant; Psoriasis 2, 602723 (3), Autosomal dominant
CASP10	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
CASP14	605848	Ichthyosis, congenital, autosomal recessive 12, 617320 (3), Autosomal recessive

CASP8	601763	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; {Breast cancer, protection against}, 114480 (3), Autosomal dominant, Somatic mutation; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Autosomal dominant, Somatic mutation
CAST	114090	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295 (3), Autosomal recessive
CBL	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
CD151	602243	[Blood group, Raph], 179620 (3); Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3)
CDH1	192090	Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; {Breast cancer, lobular}, 114480 (3), Autosomal dominant, Somatic mutation; Endometrial carcinoma, somatic, 608089 (3); Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; Ovarian cancer, somatic, 167000 (3); {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant, Somatic mutation
CDH3	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive; Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive
CDKN1B	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant
CDKN1C	600856	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; IMAGE syndrome, 614732 (3), Autosomal dominant
CDSN	602593	Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive
CERS3	615276	Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive
CHST8	610190	?Peeling skin syndrome 3, 616265 (3), Autosomal recessive
CHUK	600664	Cocoon syndrome, 613630 (3)
CLDN1	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
CLDN10	617579	HELIX syndrome, 617671 (3), Autosomal recessive
COG6	606977	Congenital disorder of glycosylation, type IIl, 614576 (3), Autosomal recessive; Shaheen syndrome, 615328 (3), Autosomal recessive
COL17A1	113811	Epidermolysis bullosa, junctional, localisata variant, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant
COL18A1	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive

COL7A1	120120	EBD inversa, 226600 (3), Autosomal recessive; EBD, Bart type, 132000 (3), Autosomal dominant; EBD, localisata variant (3); Epidermolysis bullosa dystrophica, AD, 131750 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, AR, 226600 (3), Autosomal recessive; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant; Toenail dystrophy, isolated, 607523 (3), Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant
COX7B	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
CPN1	603103	Carboxypeptidase N deficiency, 212070 (3), Autosomal recessive
CST6	601891	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535 (3), Autosomal recessive
CSTA	184600	Peeling skin syndrome 4, 607936 (3), Autosomal recessive
CTLA4	123890	Autoimmune lymphoproliferative syndrome, type V, 616100 (3), Autosomal dominant; {Celiac disease, susceptibility to, 3}, 609755 (3); {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
CTSC	602365	Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive; Periodontitis 1, juvenile, 170650 (3), Autosomal recessive
CYLD	605018	Brooke-Spiegler syndrome, 605041 (3), Autosomal dominant; Cylindromatosis, familial, 132700 (3), Autosomal dominant; Trichoepithelioma, multiple familial, 1, 601606 (3), Autosomal dominant
CYP11A1	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive; Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive
CYP26C1	608428	Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive
CYP4F22	611495	Ichthyosis, congenital, autosomal recessive 5, 604777 (3), Autosomal recessive
DCAF17	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DCLRE1C	605988	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive
DCPS	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
DDB2	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
DHCR24	606418	Desmosterolosis, 602398 (3), Autosomal recessive

DIP2B	611379	Mental retardation, FRA12A type, 136630 (3), Autosomal dominant
DKC1	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLL4	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
DOCK6	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
DPH1	603527	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive
DSC2	125645	Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal recessive, Autosomal dominant; Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal recessive, Autosomal dominant
DSG1	125670	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive; Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant
DSG3	169615	No OMIM phenotype
DSG4	607892	Hypotrichosis 6, 607903 (3), Autosomal recessive
DSP	125647	Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Skin fragility-woolly hair syndrome, 607655 (3), Autosomal recessive
DST	113810	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 (3), Autosomal recessive; ?Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive
DSTYK	612666	Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant; Spastic paraplegia 23, 270750 (3), Autosomal recessive
DTNBP1	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
EBP	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
EDA	300451	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive; Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant
EDAR	604095	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 (3), Autosomal dominant; Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 (3), Autosomal recessive; [Hair morphology 1, hair thickness], 612630 (3)
EDARADD	606603	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant; Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive

EDN3	131242	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant; Waardenburg syndrome, type 4B, 613265 (3), Autosomal recessive, Autosomal dominant
EDNRA	131243	Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant; {Migraine, resistance to}, 157300 (3), Autosomal dominant
EDNRB	131244	ABCD syndrome, 600501 (3), Autosomal recessive; {Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; Waardenburg syndrome, type 4A, 277580 (3), Autosomal recessive, Autosomal dominant
ELOVL4	605512	Ichthyosis, spastic quadriplegia, and mental retardation, 614457 (3), Autosomal recessive; Spinocerebellar ataxia 34, 133190 (3), Autosomal dominant; Stargardt disease 3, 600110 (3), Autosomal dominant
ENPP1	173335	Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
EOGT	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
EPG5	615068	Vici syndrome, 242840 (3), Autosomal recessive
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
ERCC3	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC4	133520	Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive
ERCC5	133530	Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6	609413	Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; UV-sensitive syndrome 1, 600630 (3), Autosomal recessive
ERCC8	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive

EVC	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EXPH5	612878	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028 (3), Autosomal recessive
F12	610619	Angioedema, hereditary, type III, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive
FAM111B	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FAS	134637	Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant; Squamous cell carcinoma, burn scar-related, somatic (3)
FASLG	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal dominant, Somatic mutation
FECH	612386	Protoporphyria, erythropoietic, 1, 177000 (3), Autosomal recessive
FERMT1	607900	Kindler syndrome, 173650 (3), Autosomal recessive
FGFR1	136350	Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3); Hartsfield syndrome, 615465 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant
FGFR3	134934	Achondroplasia, 100800 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Muenke syndrome, 602849 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); SADDAN, 616482 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant
FLG	135940	{Dermatitis, atopic, susceptibility to, 2}, 605803 (3); Ichthyosis vulgaris, 146700 (3), Autosomal dominant
FOXN1	600838	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive
FZD6	603409	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157 (3), Autosomal recessive

GBA	606463	Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; {Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
GINS1	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
GJA1	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Hypoplastic left heart syndrome 1, 241550 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant
GJB2	121011	Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Autosomal recessive; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant
GJB3	603324	Deafness, autosomal dominant 2B, 612644 (3), Autosomal dominant; Deafness, autosomal dominant, with peripheral neuropathy (3); Deafness, autosomal recessive (3); Deafness, digenic, GJB2/GJB3, 220290 (3), Autosomal recessive; Erythrokeratodermia variabilis et progressiva 1, 133200 (3), Autosomal recessive, Autosomal dominant
GJB4	605425	Erythrokeratodermia variabilis et progressiva 2, 617524 (3), Autosomal dominant
GJB6	604418	Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Autosomal recessive; Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant
GPR143	300808	Nystagmus 6, congenital, X-linked, 300814 (3); Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked
GREM2	608832	Tooth agenesis, selective, 9, 617275 (3), Autosomal dominant
GRHL2	608576	Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant; Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive

GRIN2B	138252	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3), Autosomal dominant
GTF2E2	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
GTF2H5	608780	Trichothiodystrophy 3, photosensitive, 616395 (3)
HCCS	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HOXC13	142976	Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive
HPGD	601688	Cranioosteoarthropathy, 259100 (3), Autosomal recessive; Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive
HPS1	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS3	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
HPS4	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS5	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
HPS6	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
HR	602302	Alopecia universalis, 203655 (3), Autosomal recessive; Atrichia with papular lesions, 209500 (3), Autosomal recessive; Hypotrichosis 4, 146550 (3), Autosomal dominant
HRAS	190020	Bladder cancer, somatic, 109800 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant; Costello syndrome, 218040 (3), Autosomal dominant; Nevus sebaceous or woolly hair nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Spitz nevus or nevus spilus, somatic, 137550 (3); Thyroid carcinoma, follicular, somatic, 188470 (3)
HSPA9	600548	Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant; Even-plus syndrome, 616854 (3), Autosomal recessive
IFT122	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT43	614068	?Cranioectodermal dysplasia 3, 614099 (3), Autosomal recessive; ?Retinitis pigmentosa 81, 617871 (3), Autosomal recessive; Short-rib thoracic dysplasia 18 with polydactyly, 617866 (3), Autosomal recessive
IKBKG	300248	Ectodermal dysplasia and immunodeficiency 1, 300291 (3); Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3); Immunodeficiency 33, 300636 (3), X-linked recessive; Immunodeficiency, isolated, 300584 (3); Incontinentia pigmenti, 308300 (3), X-linked dominant; Invasive pneumococcal disease, recurrent isolated, 2, 300640 (3)
IL2RA	147730	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3); Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive
IL31RA	609510	?Amyloidosis, primary localized cutaneous, 2, 613955 (3), Autosomal dominant
ITGA3	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748 (3), Autosomal recessive

ITGA6	147556	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 (3), Autosomal recessive
ITGB4	147557	Epidermolysis bullosa of hands and feet, 131800 (3), Autosomal dominant; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3), Autosomal recessive
ITPR2	600144	?Anhidrosis, isolated, with normal sweat glands, 106190 (3), Autosomal recessive
JUP	173325	Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant; Naxos disease, 601214 (3), Autosomal recessive
KCNH1	603305	Temple-Baraitser syndrome, 611816 (3), Autosomal dominant; Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant
KCTD1	613420	Scalp-ear-nipple syndrome, 181270 (3), Autosomal dominant
KDF1	616758	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 (3), Autosomal dominant
KDSR	136440	Erythrokeratodermia variabilis et progressiva 4, 617526 (3), Autosomal recessive
KEAP1	606016	No OMIM phenotype
KIT	164920	Gastrointestinal stromal tumor, familial, 606764 (3), Autosomal dominant, Isolated cases; Germ cell tumors, somatic, 273300 (3); Leukemia, acute myeloid, somatic, 601626 (3); Mastocytosis, cutaneous, 154800 (3), Autosomal dominant; Mastocytosis, systemic, somatic, 154800 (3); Piebaldism, 172800 (3), Autosomal dominant
KITLG	184745	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 (3), Autosomal dominant; Hyperpigmentation with or without hypopigmentation, 145250 (3), Autosomal dominant; [Skin/hair/eye pigmentation 7, blond/brown hair], 611664 (3)
KLHL24	611295	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294 (3), Autosomal dominant
KRAS	190070	Arteriovenous malformation of the brain, somatic, 108010 (3); Bladder cancer, somatic, 109800 (3); Breast cancer, somatic, 114480 (3); Cardiofaciocutaneous syndrome 2, 615278 (3); Gastric cancer, somatic, 137215 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant, Somatic mutation; Lung cancer, somatic, 211980 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; Oculoectodermal syndrome, somatic, 600268 (3); Pancreatic carcinoma, somatic, 260350 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)
KREMEN1	609898	Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive

KRT1	139350	Epidermolytic hyperkeratosis, 113800 (3), Autosomal recessive, Autosomal dominant; Ichthyosis histrix, Curth-Macklin type, 146590 (3), Autosomal dominant; Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3), Autosomal dominant; Keratosis palmoplantaris striata III, 607654 (3); Palmoplantar keratoderma, epidermolytic, 144200 (3), Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, 600962 (3), Autosomal dominant
KRT10	148080	Epidermolytic hyperkeratosis, 113800 (3), Autosomal recessive, Autosomal dominant; Ichthyosis with confetti, 609165 (3), Autosomal dominant; Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3), Autosomal dominant
KRT13	148065	White sponge nevus 2, 615785 (3), Autosomal dominant
KRT14	148066	Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant
KRT16	148067	Pachyonychia congenita 1, 167200 (3), Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, focal, 613000 (3), Autosomal dominant
KRT17	148069	Pachyonychia congenita 2, 167210 (3), Autosomal dominant; Steatocystoma multiplex, 184500 (3), Autosomal dominant
KRT2	600194	Ichthyosis bullosa of Siemens, 146800 (3), Autosomal dominant
KRT3	148043	Meesmann corneal dystrophy, 122100 (3), Autosomal dominant
KRT5	148040	Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3), Autosomal dominant; Epidermolysis bullosa simplex, Koebner type, 131900 (3), Autosomal dominant; Epidermolysis bullosa simplex, Weber-Cockayne type, 131800 (3), Autosomal dominant; Epidermolysis bullosa simplex, recessive 1, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex-MCR, 609352 (3); Epidermolysis bullosa simplex-MP, 131960 (3), Autosomal dominant
KRT6A	148041	Pachyonychia congenita 3, 615726 (3)
KRT6B	148042	Pachyonychia congenita 4, 615728 (3)
KRT6C	612315	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735 (3), Autosomal dominant
KRT7	148059	No OMIM phenotype
KRT74	608248	?Ectodermal dysplasia 7, hair/nail type, 614929 (3), Autosomal recessive; ?Hypotrichosis 3, 613981 (3), Autosomal dominant; Woolly hair, autosomal dominant, 194300 (3), Autosomal dominant
KRT83	602765	Erythrokeratodermia variabilis et progressiva 5, 617756 (3), Autosomal recessive; Monilethrix, 158000 (3), Autosomal dominant
KRT85	602767	Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive
KRT9	607606	Palmoplantar keratoderma, epidermolytic, 144200 (3), Autosomal dominant

LAMA3	600805	Epidermolysis bullosa, generalized atrophic benign, 226650 (3), Autosomal recessive; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Laryngoonychocutaneous syndrome, 245660 (3), Autosomal recessive
LAMB3	150310	Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant; Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
LAMC2	150292	Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
LBR	600024	Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive; Pelger-Huet anomaly, 169400 (3), Autosomal dominant; Pelger-Huet anomaly with mild skeletal anomalies, 618019 (3); ?Reynolds syndrome, 613471 (3), Autosomal dominant
LIPH	607365	Hypotrichosis 7, 604379 (3), Autosomal recessive; Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3), Autosomal recessive
LIPN	613924	Ichthyosis, congenital, autosomal recessive 8, 613943 (3), Autosomal recessive
LMNA	150330	Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal recessive, Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant; Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
LMNB2	150341	?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant
LOR	152445	Vohwinkel syndrome with ichthyosis, 604117 (3), Autosomal dominant
LPAR6	609239	Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive
LRMDA	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
LRP1	107770	?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive
LYST	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
LZTR1	600574	Noonan syndrome 10, 616564 (3), Autosomal dominant; Noonan syndrome 2, 605275 (3), Autosomal recessive; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant

MBTPS2	300294	IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive
MC1R	155555	{Albinism, oculocutaneous, type II, modifier of}, 203200 (3), Autosomal recessive; [Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3); {Melanoma, cutaneous malignant, 5}, 613099 (3); [Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3), Autosomal recessive; [Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3), Autosomal recessive; {UV-induced skin damage}, 266300 (3), Autosomal recessive
MC2R	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive
MC4R	155541	Obesity (BMIQ20), 618406 (3); {Obesity, resistence to (BMIQ20)}, 618306 (3)
MCM4	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
MEN1	613733	Adrenal adenoma, somatic (3); Angiofibroma, somatic (3); Carcinoid tumor of lung (3); Lipoma, somatic (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant; Parathyroid adenoma, somatic (3)
MITF	156845	COMMAD syndrome, 617306 (3), Autosomal recessive; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; Waardenburg syndrome/ocular albinism, digenic, 103470 (3), Autosomal dominant
MLPH	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
MMP1	120353	COPD, rate of decline of lung function in, 606963 (3); {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600 (3), Autosomal recessive
MPDU1	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MPLKIP	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
MSX1	142983	Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Orofacial cleft 5, 608874 (3); Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant
MYO5A	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
NECTIN1	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
NECTIN4	609607	Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive
NF1	613113	Leukemia, juvenile myelomonocytic, 607785 (3), Autosomal dominant, Somatic mutation; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant; Watson syndrome, 193520 (3), Autosomal dominant
NFKBIA	164008	Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant

NIPAL4	609383	Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive
NLRC4	606831	Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant; ?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant
NLRP1	606636	Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant; {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3)
NLRP12	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
NLRP3	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NNT	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive
NOP10	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NOTCH1	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
NR5A1	184757	Adrenocortical insufficiency, 612964 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant; 46, XX sex reversal 4, 617480 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant
NRAS	164790	Colorectal cancer, somatic, 114500 (3); Epidermal nevus, somatic, 162900 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Neurocutaneous melanosis, somatic, 249400 (3); Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3)
NSDHL	300275	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive
OCA2	611409	Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive
OSMR	601743	Amyloidosis, primary localized cutaneous, 1, 105250 (3), Autosomal dominant

PAX3	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant
PAX6	607108	Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3); Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; ?Coloboma, ocular, 120200 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Optic nerve hypoplasia, 165550 (3), Autosomal dominant
PDGFRB	173410	Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myeloproliferative disorder with eosinophilia, 131440 (4), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant
PEPD	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PEX7	601757	Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive; Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
PGM3	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PHGDH	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHYH	602026	Refsum disease, 266500 (3), Autosomal recessive
PIEZO1	611184	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant; Lymphatic malformation 6, 616843 (3), Autosomal recessive
PIGL	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PIGT	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
PKP1	601975	Ectodermal dysplasia/skin fragility syndrome, 604536 (3)
PLCD1	602142	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 (3), Autosomal recessive, Autosomal dominant
PLCG2	600220	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant
PLEC	601282	Epidermolysis bullosa simplex with muscular dystrophy, 226670 (3), Autosomal recessive; ?Epidermolysis bullosa simplex with nail dystrophy, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive

PMVK	607622	Porokeratosis 1, multiple types, 175800 (3), Autosomal dominant
PNPLA1	612121	Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive
PNPLA2	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
PNPLA6	603197	Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive
POFUT1	607491	Dowling-Degos disease 2, 615327 (3), Autosomal dominant
POGLUT1	615618	Dowling-Degos disease 4, 615696 (3), Autosomal dominant; ?Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 (3), Autosomal recessive
POLA1	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive; Van Esch-O'Driscoll syndrome, 301030 (3), X-linked recessive
POLH	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
POMC	176830	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive; {Obesity, early-onset, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
POMP	613386	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive; Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant
PORCN	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
PPOX	600923	Porphyria variegata, 176200 (3), Autosomal dominant
PRKCD	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
PRKD1	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
PSAT1	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSENEN	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
PSMB8	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive
PTCH1	601309	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTEN	601728	Cowden syndrome 1, 158350 (3), Autosomal dominant; {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3)

PTPN11	176876	LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3); Metachondromatosis, 156250 (3), Autosomal dominant; Noonan syndrome 1, 163950 (3), Autosomal dominant
PTPRF	179590	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001 (3), Autosomal recessive
RAB27A	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAD50	604040	Nijmegen breakage syndrome-like disorder, 613078 (3)
RAF1	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3), Autosomal dominant
RAG1	179615	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3); Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive
RAG2	179616	Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive
RBM28	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
RBPJ	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
RECQL4	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive; Rothmund-Thomson syndrome, 268400 (3), Autosomal recessive
RET	164761	Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; {Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant
RHBDF2	614404	Tylosis with esophageal cancer, 148500 (3), Autosomal dominant
RIN2	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive
RIT1	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RMRP	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive
RSPO1	609595	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 (3), Autosomal recessive; Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 (3), Autosomal recessive

SCN9A	603415	{Dravet syndrome, modifier of}, 607208 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 7, 613863 (3), Autosomal dominant; Erythermalgia, primary, 133020 (3), Autosomal dominant; Febrile seizures, familial, 3B, 613863 (3), Autosomal dominant; HSAN2D, autosomal recessive, 243000 (3), Autosomal recessive; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Small fiber neuropathy, 133020 (3), Autosomal dominant
SDR9C7	609769	Ichthyosis, congenital, autosomal recessive 13, 617574 (3), Autosomal recessive
SERPINB7	603357	Palmoplantar keratoderma, Nagashima type, 615598 (3), Autosomal recessive
SERPINB8	601697	Peeling skin syndrome 5, 617115 (3), Autosomal recessive
SERPING1	606860	Angioedema, hereditary, types I and II, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
SGPL1	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
SHOC2	602775	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
SLC24A5	609802	Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive; [Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive
SLC27A4	604194	Ichthyosis prematurity syndrome, 608649 (3)
SLC29A3	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC39A4	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
SLC45A2	606202	Albinism, oculocutaneous, type IV, 606574 (3); [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive
SLURP1	606119	Meleda disease, 248300 (3), Autosomal recessive
SMARCA2	600014	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant
SMARCAD1	612761	Adermatoglyphia, 136000 (3), Autosomal dominant; Basan syndrome, 129200 (3), Autosomal dominant; Huriez syndrome, 181600 (3), Autosomal dominant
SNAI2	602150	Piebaldism, 172800 (3), Autosomal dominant; Waardenburg syndrome, type 2D, 608890 (3), Autosomal recessive
SNAP29	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
SNRPE	128260	Hypotrichosis 11, 615059 (3), Autosomal dominant
SOS1	182530	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
SOS2	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant

SOX10	602229	PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
SOX18	601618	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3), Autosomal recessive; Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 (3), Autosomal dominant
SPINK5	605010	Netherton syndrome, 256500 (3), Autosomal recessive
SPINT1	No OMIM gene	No OMIM phenotype
SPRED1	609291	Legius syndrome, 611431 (3), Autosomal dominant
SRD5A3	611715	Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive; Kahrizi syndrome, 612713 (3), Autosomal recessive
ST14	606797	Ichthyosis, congenital, autosomal recessive 11, 602400 (3), Autosomal recessive
ST3GAL5	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
STAT3	102582	Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant; Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant
STIM1	605921	Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant
STK11	602216	Melanoma, malignant, somatic (3); Pancreatic cancer, somatic, 260350 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3)
STS	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive
SULT2B1	604125	Ichthyosis, congenital, autosomal recessive 14, 617571 (3), Autosomal recessive
SUMF1	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
TAT	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TERC	602322	{Aplastic anemia}, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant; {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743 (3), Autosomal dominant
TGM1	190195	Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive
TGM3	600238	?Uncombable hair syndrome 2, 617251 (3), Autosomal recessive
TGM5	603805	Peeling skin syndrome 2, 609796 (3), Autosomal recessive
TINF2	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant

TP63	603273	ADULT syndrome, 103285 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant
TRPM1	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3)
TRPS1	604386	Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant
TRPV3	607066	Olmsted syndrome, 614594 (3), Autosomal dominant; ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 (3), Autosomal dominant
TWIST2	607556	Ablepharon-macrostomia syndrome, 200110 (3), Autosomal dominant; Barber-Say syndrome, 209885 (3), Autosomal dominant; Focal facial dermal dysplasia 3, Setleis type, 227260 (3), Autosomal recessive
TXNRD2	606448	?Glucocorticoid deficiency 5, 617825 (3), Autosomal recessive
TYRP1	115501	Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive; [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3)
UBR1	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
UROD	613521	Porphyria cutanea tarda, 176100 (3), Autosomal recessive, Autosomal dominant; Porphyria, hepatoerythropoietic, 176100 (3), Autosomal recessive, Autosomal dominant
UROS	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
USB1	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
VCX	300229	No OMIM phenotype
VCX3A	300533	No OMIM phenotype
WDR19	608151	?Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Nephronophthisis 13, 614377 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive
WDR35	613602	Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive; Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive
WNT10A	606268	Odontoonychodermal dysplasia, 257980 (3), Autosomal recessive; Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal recessive, Autosomal dominant
XPA	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPC	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
ZMPSTE24	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: Sept 30, 2019

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.