

<b>SkinDisorders panel</b>
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<b>versie</b>	19-Feb-2019 (333 genen)	Centrum voor Medische Genetica Gent
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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	GAP0 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

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<i>ARHGAP31</i>	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
<i>ARSE</i>	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
<i>ATP2A2</i>	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant
<i>ATP6V1B2</i>	606939	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant; Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant
<i>ATP7A</i>	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
<i>ATR</i>	601215	?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant; Seckel syndrome 1, 210600 (3), Autosomal recessive
<i>AXIN2</i>	604025	Colorectal cancer, somatic, 114500 (3); Oligodontia-colorectal cancer syndrome, 608615 (3), Autosomal dominant
<i>BANF1</i>	603811	Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive
<i>BCS1L</i>	603647	Bjornstad syndrome, 262000 (3), Autosomal recessive; GRACILE syndrome, 603358 (3); Leigh syndrome, 256000 (3), Autosomal recessive, Mitochondrial; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive
<i>BLM</i>	604610	Bloom syndrome, 210900 (3), Autosomal recessive
<i>BLOC1S3</i>	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
<i>BLOC1S6</i>	604310	?Hermansky-pudlak syndrome 9, 614171 (3), Autosomal recessive
<i>BMS1</i>	611448	?Aplasia cutis congenita, nonsyndromic, 107600 (3), Autosomal dominant
<i>BRAF</i>	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); Non-small cell lung cancer, somatic (3); Noonan syndrome 7, 613706 (3), Autosomal dominant
<i>C1QA</i>	120550	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QB</i>	120570	C1q deficiency, 613652 (3), Autosomal recessive
<i>C1QC</i>	120575	C1q deficiency, 613652 (3), Autosomal recessive
<i>C5</i>	120900	C5 deficiency, 609536 (3); [Eculizumab, poor response to], 615749 (3), Autosomal dominant
<i>CARD11</i>	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
<i>CARD14</i>	607211	Pityriasis rubra pilaris, 173200 (3), Autosomal dominant; Psoriasis 2, 602723 (3), Autosomal dominant

<i>CASP10</i>	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
<i>CASP14</i>	605848	Ichthyosis, congenital, autosomal recessive 12, 617320 (3), Autosomal recessive
<i>CASP8</i>	601763	?Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3), Autosomal recessive; {Breast cancer, protection against}, 114480 (3), Autosomal dominant; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Autosomal recessive
<i>CAST</i>	114090	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295 (3), Autosomal recessive
<i>CBL</i>	165360	?Juvenile myelomonocytic leukemia, 607785 (3), Autosomal dominant, Somatic mutation; Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant
<i>CD151</i>	602243	[Blood group, Raph], 179620 (3); Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3)
<i>CDH1</i>	192090	Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; {Breast cancer, lobular}, 114480 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; Ovarian carcinoma, somatic, 167000 (3); {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant
<i>CDH3</i>	114021	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive; Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive
<i>CDKN1B</i>	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant
<i>CDKN1C</i>	600856	Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant; IMAGE syndrome, 614732 (3), Autosomal dominant
<i>CDSN</i>	602593	Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive
<i>CERS3</i>	615276	Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive
<i>CHST8</i>	610190	?Peeling skin syndrome 3, 616265 (3), Autosomal recessive
<i>CHUK</i>	600664	Cocoon syndrome, 613630 (3)
<i>CLDN1</i>	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
<i>CLDN10</i>	617579	HELIX syndrome, 617671 (3), Autosomal recessive
<i>COG6</i>	606977	Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive; Shaheen syndrome, 615328 (3), Autosomal recessive

<i>COL17A1</i>	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
<i>COL18A1</i>	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive
<i>COL7A1</i>	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
<i>COX7B</i>	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
<i>CPN1</i>	603103	Carboxypeptidase N deficiency, 212070 (3), Autosomal recessive
<i>CST6</i>	601891	No OMIM phenotype
<i>CSTA</i>	184600	Peeling skin syndrome 4, 607936 (3), Autosomal recessive
<i>CTLA4</i>	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
<i>CTSC</i>	602365	Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive; Periodontitis 1, juvenile, 170650 (3), Autosomal recessive
<i>CYP11A1</i>	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
<i>CYP11B1</i>	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive; Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant
<i>CYP21A2</i>	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
<i>CYP26C1</i>	608428	Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive
<i>CYP4F22</i>	611495	Ichthyosis, congenital, autosomal recessive 5, 604777 (3), Autosomal recessive
<i>DCAF17</i>	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
<i>DCLRE1C</i>	605988	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, Athabaskan type, 602450 (3), Autosomal recessive
<i>DCPS</i>	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive

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<i>DDB2</i>	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive
<i>DHCR24</i>	606418	Desmosterolosis, 602398 (3), Autosomal recessive
<i>DIP2B</i>	611379	Mental retardation, FRA12A type, 136630 (3), Autosomal dominant
<i>DKC1</i>	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
<i>DLL4</i>	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
<i>DOCK6</i>	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
<i>DOCK8</i>	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
<i>DPH1</i>	603527	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901 (3), Autosomal recessive
<i>DSC2</i>	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
<i>DSG1</i>	125670	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive; Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant
<i>DSG3</i>	169615	No OMIM phenotype
<i>DSG4</i>	607892	Hypotrichosis 6, 607903 (3), Autosomal recessive
<i>DSP</i>	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
<i>DST</i>	113810	Epidermolysis bullosa simplex, autosomal recessive 2, 615425 (3), Autosomal recessive; ?Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive
<i>DSTYK</i>	612666	Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant; Spastic paraplegia 23, 270750 (3), Autosomal recessive
<i>DTNBP1</i>	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
<i>EBP</i>	300205	Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant; MEND syndrome, 300960 (3), X-linked recessive
<i>EDA</i>	300451	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive; Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant
<i>EDAR</i>	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)

<i>EDARADD</i>	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
<i>EDN3</i>	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
<i>EDNRA</i>	131243	Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant; {Migraine, resistance to}, 157300 (3), Autosomal dominant
<i>EDNRB</i>	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
<i>ELOVL4</i>	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
<i>ENPP1</i>	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 dominant; {Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>EOGT</i>	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
<i>EPG5</i>	615068	Vici syndrome, 242840 (3), Autosomal recessive
<i>ERCC2</i>	126340	?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive
<i>ERCC3</i>	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
<i>ERCC4</i>	133520	Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive; ?XFE progeroid syndrome, 610965 (3); Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive
<i>ERCC5</i>	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

<i>ERCC6</i>	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 recessive; {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
<i>ERCC8</i>	609412	Cockayne syndrome, type A, 216400 (3), Autosomal recessive; UV-sensitive syndrome 2, 614621 (3), Autosomal recessive
<i>EVC</i>	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EVC2</i>	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
<i>EXPH5</i>	612878	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028 (3), Autosomal recessive
<i>F12</i>	610619	Angioedema, hereditary, type III, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive
<i>FAM111B</i>	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
<i>FAS</i>	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)
<i>FASLG</i>	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Autosomal recessive
<i>FECH</i>	612386	Protoporphyrinemia, erythropoietic, 1, 177000 (3), Autosomal recessive
<i>FERMT1</i>	607900	Kindler syndrome, 173650 (3), Autosomal recessive
<i>FGFR1</i>	136350	Encephalocraniocutaneous lipomatosis, 613001 (3), Somatic mosaicism; 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
<i>FGFR3</i>	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
<i>FLG</i>	135940	{Dermatitis, atopic, susceptibility to, 2}, 605803 (3); Ichthyosis vulgaris, 146700 (3), Autosomal dominant
<i>FOXP1</i>	600838	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive
<i>FZD6</i>	603409	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157 (3), Autosomal recessive
<i>GBA</i>	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), Isolated cases, Multifactorial
<i>GINS1</i>	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
<i>GJA1</i>	121014	Atrioventricular septal defect 3, 600309 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Erythrokeratoderma 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
<i>GJB2</i>	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



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<i>GJB3</i>	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; Erythrokeratoderma variabilis et progressiva 1, 133200 (3), Autosomal recessive, Autosomal dominant
<i>GJB4</i>	605425	Erythrokeratoderma variabilis et progressiva 2, 617524 (3), Autosomal dominant
<i>GJB6</i>	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
<i>GPR143</i>	300808	Nystagmus 6, congenital, X-linked, 300814 (3); Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked
<i>GREM2</i>	608832	Tooth agenesis, selective, 9, 617275 (3), Autosomal dominant
<i>GRHL2</i>	608576	Corneal dystrophy, posterior polymorphous, 4, 618031 (3); Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive
<i>GRIN2B</i>	138252	Epileptic encephalopathy, early infantile, 27, 616139 (3), Autosomal dominant; Mental retardation, autosomal dominant 6, 613970 (3), Autosomal dominant
<i>GTF2E2</i>	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
<i>GTF2H5</i>	608780	Trichothiodystrophy 3, photosensitive, 616395 (3)
<i>HCCS</i>	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
<i>HOXC13</i>	142976	Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive
<i>HPGD</i>	601688	Cranioosteoarthropathy, 259100 (3), Autosomal recessive; Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive
<i>HPS1</i>	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
<i>HPS3</i>	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
<i>HPS4</i>	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
<i>HPS5</i>	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
<i>HPS6</i>	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
<i>HR</i>	602302	Alopecia universalis, 203655 (3), Autosomal recessive; Atrichia with papular lesions, 209500 (3), Autosomal recessive; Hypotrichosis 4, 146550 (3), Autosomal dominant

<i>HRAS</i>	190020	{Bladder cancer, somatic}, 109800 (3); Congenital myopathy with excess of muscle spindles, 218040 (3), Autosomal dominant, Isolated cases; Costello syndrome, 218040 (3), Autosomal dominant, Isolated cases; {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)
<i>HSPA9</i>	600548	Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant; Even-plus syndrome, 616854 (3), Autosomal recessive
<i>IFT122</i>	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
<i>IFT43</i>	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
<i>IKBKG</i>	300248	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 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)
<i>IL2RA</i>	147730	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3); Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive
<i>IL31RA</i>	609510	?Amyloidosis, primary localized cutaneous, 2, 613955 (3), Autosomal dominant
<i>ITGA3</i>	605025	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748 (3), Autosomal recessive
<i>ITGA6</i>	147556	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 (3), Autosomal recessive
<i>ITGB4</i>	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
<i>ITPR2</i>	600144	?Anhidrosis, isolated, with normal sweat glands, 106190 (3), Autosomal recessive
<i>JUP</i>	173325	Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant; Naxos disease, 601214 (3), Autosomal recessive
<i>KCNH1</i>	603305	Temple-Baraitser syndrome, 611816 (3), Autosomal dominant; Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant
<i>KCTD1</i>	613420	Scalp-ear-nipple syndrome, 181270 (3), Autosomal dominant
<i>KDF1</i>	616758	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 (3), Autosomal dominant
<i>KDSR</i>	136440	Erythrokeratoderma variabilis et progressiva 4, 617526 (3), Autosomal recessive

<i>KEAP1</i>	606016	No OMIM phenotype
<i>KIT</i>	164920	Gastrointestinal stromal tumor, familial, 606764 (3), Autosomal dominant, Isolated cases; Germ cell tumors, somatic, 273300 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant; Mastocytosis, cutaneous, 154800 (3), Autosomal dominant; Mastocytosis, systemic, somatic, 154800 (3); Piebaldism, 172800 (3), Autosomal dominant
<i>KITLG</i>	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)
<i>KLHL24</i>	611295	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294 (3), Autosomal dominant
<i>KRAS</i>	190070	Arteriovenous malformation of the brain, somatic, 108010 (3); Bladder cancer, somatic, 109800 (3); Breast cancer, somatic, 114480 (3); Cardiodiagnostic syndrome 2, 615278 (3); Gastric cancer, somatic, 137215 (3); Leukemia, acute myeloid, 601626 (3), Autosomal dominant; Lung cancer, somatic, 211980 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; Pancreatic carcinoma, somatic, 260350 (3); RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3)
<i>KREMEN1</i>	609898	Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive
<i>KRT1</i>	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
<i>KRT10</i>	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
<i>KRT13</i>	148065	White sponge nevus 2, 615785 (3), Autosomal dominant
<i>KRT14</i>	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

<i>KRT16</i>	148067	Pachyonychia congenita 1, 167200 (3), Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, focal, 613000 (3), Autosomal dominant
<i>KRT17</i>	148069	Pachyonychia congenita 2, 167210 (3), Autosomal dominant; Steatocystoma multiplex, 184500 (3), Autosomal dominant
<i>KRT2</i>	600194	Ichthyosis bullosa of Siemens, 146800 (3), Autosomal dominant
<i>KRT3</i>	148043	Meesmann corneal dystrophy, 122100 (3), Autosomal dominant
<i>KRT5</i>	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
<i>KRT6A</i>	148041	Pachyonychia congenita 3, 615726 (3)
<i>KRT6B</i>	148042	Pachyonychia congenita 4, 615728 (3)
<i>KRT6C</i>	612315	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735 (3), Autosomal dominant
<i>KRT7</i>	148059	No OMIM phenotype
<i>KRT74</i>	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
<i>KRT83</i>	602765	Erythrokeratoderma variabilis et progressiva 5, 617756 (3), Autosomal recessive; Monilethrix, 158000 (3), Autosomal dominant
<i>KRT85</i>	602767	Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive
<i>KRT9</i>	607606	Palmoplantar keratoderma, epidermolytic, 144200 (3), Autosomal dominant
<i>LAMA3</i>	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
<i>LAMB3</i>	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
<i>LAMC2</i>	150292	Epidermolysis bullosa, junctional, Herlitz type, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional, non-Herlitz type, 226650 (3), Autosomal recessive
<i>LBR</i>	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

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<i>LIPH</i>	607365	Hypotrichosis 7, 604379 (3), Autosomal recessive; Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3), Autosomal recessive
<i>LIPN</i>	613924	Ichthyosis, congenital, autosomal recessive 8, 613943 (3), Autosomal recessive
<i>LMNA</i>	150330	Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, AD, 181350 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, AR, 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; Muscular dystrophy, limb-girdle, type 1B, 159001 (3), Autosomal dominant; Restrictive dermopathy, lethal, 275210 (3), Autosomal recessive
<i>LMNB2</i>	150341	?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant
<i>LOR</i>	152445	Vohwinkel syndrome with ichthyosis, 604117 (3), Autosomal dominant
<i>LPAR6</i>	609239	Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive
<i>LRMDA</i>	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
<i>LRP1</i>	107770	?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive
<i>LYST</i>	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
<i>LZTR1</i>	600574	Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
<i>MBTPS2</i>	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
<i>MC1R</i>	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
<i>MC2R</i>	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive

<i>MC4R</i>	155541	Obesity, autosomal dominant, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
<i>MCM4</i>	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
<i>MEN1</i>	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)
<i>MITF</i>	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
<i>MLPH</i>	606526	Griselli syndrome, type 3, 609227 (3), Autosomal recessive
<i>MMP1</i>	120353	COPD, rate of decline of lung function in, 606963 (3); {Epidermolysis bullosa dystrophica, autosomal recessive, modifier of}, 226600 (3), Autosomal recessive
<i>MPDU1</i>	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
<i>MPLKIP</i>	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
<i>MSX1</i>	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
<i>MYO5A</i>	160777	Griselli syndrome, type 1, 214450 (3), Autosomal recessive
<i>NECTIN1</i>	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
<i>NECTIN4</i>	609607	Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive
<i>NF1</i>	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
<i>NFKBIA</i>	164008	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132 (3), Autosomal dominant
<i>NIPAL4</i>	609383	Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive
<i>NLRC4</i>	606831	Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant; ?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant

<i>NLRP1</i>	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)
<i>NLRP12</i>	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
<i>NLRP3</i>	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
<i>NNT</i>	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive
<i>NOP10</i>	606471	Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
<i>NOTCH1</i>	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
<i>NR5A1</i>	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
<i>NRAS</i>	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)
<i>NSDHL</i>	300275	CHILD syndrome, 308050 (3), X-linked dominant; CK syndrome, 300831 (3), X-linked recessive
<i>OCA2</i>	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
<i>OSMR</i>	601743	Amyloidosis, primary localized cutaneous, 1, 105250 (3), Autosomal dominant
<i>PAX3</i>	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Autosomal recessive; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant

<i>PAX6</i>	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
<i>PDGFRB</i>	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
<i>PEPD</i>	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
<i>PEX7</i>	601757	Peroxisome biogenesis disorder 9B, 614879 (3); Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive
<i>PGM3</i>	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
<i>PHGDH</i>	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
<i>PHYH</i>	602026	Refsum disease, 266500 (3), Autosomal recessive
<i>PIEZO1</i>	611184	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant; Lymphedema, hereditary, III, 616843 (3), Autosomal recessive
<i>PIGL</i>	605947	CHIME syndrome, 280000 (3), Autosomal recessive
<i>PIGT</i>	610272	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive; ?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Autosomal dominant, Somatic mutation
<i>PKP1</i>	601975	Ectodermal dysplasia/skin fragility syndrome, 604536 (3)
<i>PLCD1</i>	602142	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 (3), Autosomal recessive, Autosomal dominant
<i>PLCG2</i>	600220	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant
<i>PLEC</i>	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, type 2Q, 613723 (3), Autosomal recessive
<i>PMVK</i>	607622	Porokeratosis 1, multiple types, 175800 (3), Autosomal dominant
<i>PNPLA1</i>	612121	Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive



<i>PNPLA2</i>	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
<i>PNPLA6</i>	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
<i>POFUT1</i>	607491	Dowling-Degos disease 2, 615327 (3), Autosomal dominant
<i>POGLUT1</i>	615618	Dowling-Degos disease 4, 615696 (3), Autosomal dominant; ?Muscular dystrophy, limb-girdle, type 2Z, 617232 (3), Autosomal recessive
<i>POLA1</i>	312040	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220 (3), X-linked recessive
<i>POLH</i>	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
<i>POMC</i>	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
<i>POMP</i>	613386	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
<i>PORCN</i>	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
<i>PPOX</i>	600923	Porphyria variegata, 176200 (3), Autosomal dominant
<i>PRKCD</i>	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
<i>PRKD1</i>	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
<i>PSAT1</i>	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; ?Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
<i>PSENE1</i>	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
<i>PSMB8</i>	177046	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040 (3), Autosomal recessive
<i>PTCH1</i>	601309	Basal cell carcinoma, somatic, 605462 (3); Basal cell nevus syndrome, 109400 (3), Autosomal dominant; Holoprosencephaly 7, 610828 (3), Autosomal dominant
<i>PTEN</i>	601728	Bannayan-Riley-Ruvalcaba syndrome, 153480 (3); Cowden syndrome 1, 158350 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); {Glioma susceptibility 2}, 613028 (3); Lhermitte-Duclos syndrome, 158350 (3), Autosomal dominant; Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant; Malignant melanoma, somatic, 155600 (3); {Meningioma}, 607174 (3), Autosomal dominant; PTEN hamartoma tumor syndrome (3); {Prostate cancer, somatic}, 176807 (3); Squamous cell carcinoma, head and neck, somatic, 275355 (3); VATER association with

		macrocephaly and ventriculomegaly, 276950 (3), Autosomal recessive
<i>PTPN11</i>	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
<i>PTPRF</i>	179590	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001 (3), Autosomal recessive
<i>RAB27A</i>	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
<i>RAD50</i>	604040	Nijmegen breakage syndrome-like disorder, 613078 (3)
<i>RAF1</i>	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3); Noonan syndrome 5, 611553 (3), Autosomal dominant
<i>RAG1</i>	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
<i>RAG2</i>	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
<i>RBM28</i>	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
<i>RBPJ</i>	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
<i>RECQL4</i>	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive; Rothmund-Thomson syndrome, 268400 (3), Autosomal recessive
<i>RET</i>	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
<i>RHBDF2</i>	614404	Tylosis with esophageal cancer, 148500 (3), Autosomal dominant

<i>RIN2</i>	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive
<i>RIT1</i>	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
<i>RMRP</i>	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive
<i>RSPO1</i>	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
<i>SCN9A</i>	603415	{Dravet syndrome, modifier of}, 607208 (3), Autosomal dominant; Epilepsy, generalized, with febrile seizures plus, type 7, 613863 (3), Autosomal dominant; Erythralgia, 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
<i>SDR9C7</i>	609769	Ichthyosis, congenital, autosomal recessive 13, 617574 (3), Autosomal recessive
<i>SERPINB7</i>	603357	Palmoplantar keratoderma, Nagashima type, 615598 (3), Autosomal recessive
<i>SERPINB8</i>	601697	Peeling skin syndrome 5, 617115 (3), Autosomal recessive
<i>SERPING1</i>	606860	Angioedema, hereditary, types I and II, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
<i>SGPL1</i>	603729	Nephrotic syndrome 14, 617575 (3), Autosomal recessive
<i>SHOC2</i>	602775	Noonan-like syndrome with loose anagen hair, 607721 (3), Autosomal dominant
<i>SLC24A5</i>	609802	Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive; [Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive
<i>SLC27A4</i>	604194	Ichthyosis prematurity syndrome, 608649 (3)
<i>SLC29A3</i>	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
<i>SLC39A4</i>	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
<i>SLC45A2</i>	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
<i>SLURP1</i>	606119	Meleda disease, 248300 (3), Autosomal recessive
<i>SMARCA2</i>	600014	Nicolaidis-Baraitser syndrome, 601358 (3), Autosomal dominant

<i>SMARCAD1</i>	612761	Adermatoglyphia, 136000 (3), Autosomal dominant; Basan syndrome, 129200 (3), Autosomal dominant
<i>SNAI2</i>	602150	Piebaldism, 172800 (3), Autosomal dominant; Waardenburg syndrome, type 2D, 608890 (3), Autosomal recessive
<i>SNAP29</i>	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
<i>SNRPE</i>	128260	Hypotrichosis 11, 615059 (3), Autosomal dominant
<i>SOS1</i>	182530	?Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant; Noonan syndrome 4, 610733 (3), Autosomal dominant
<i>SOS2</i>	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
<i>SOX10</i>	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
<i>SOX18</i>	601618	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3), Autosomal recessive; Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 (3), Autosomal dominant
<i>SPINK5</i>	605010	Netherton syndrome, 256500 (3), Autosomal recessive
<i>SPINT1</i>	No OMIM gene	No OMIM phenotype
<i>SPRED1</i>	609291	Legius syndrome, 611431 (3), Autosomal dominant
<i>SRD5A3</i>	611715	Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive; Kahrizi syndrome, 612713 (3), Autosomal recessive
<i>ST14</i>	606797	Ichthyosis, congenital, autosomal recessive 11, 602400 (3), Autosomal recessive
<i>ST3GAL5</i>	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
<i>STAT3</i>	102582	Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant; Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant
<i>STIM1</i>	605921	Immunodeficiency 10, 612783 (3), Autosomal recessive; Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant
<i>STK11</i>	602216	Melanoma, malignant, somatic (3); Pancreatic cancer, 260350 (3), Autosomal dominant, Somatic mutation, Multifactorial; Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3)
<i>STS</i>	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive
<i>SULT2B1</i>	604125	Ichthyosis, congenital, autosomal recessive 14, 617571 (3), Autosomal recessive
<i>SUMF1</i>	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
<i>TAT</i>	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
<i>TERC</i>	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

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<i>TGM1</i>	190195	Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive
<i>TGM3</i>	600238	?Uncombable hair syndrome 2, 617251 (3), Autosomal recessive
<i>TGM5</i>	603805	Peeling skin syndrome 2, 609796 (3), Autosomal recessive
<i>TINF2</i>	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
<i>TP63</i>	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, 129400 (3), Autosomal dominant; Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant
<i>TRPM1</i>	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3)
<i>TRPV3</i>	607066	Olmsted syndrome, 614594 (3), Autosomal dominant; ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 (3), Autosomal dominant
<i>TWIST2</i>	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
<i>TXNRD2</i>	606448	?Glucocorticoid deficiency 5, 617825 (3), Autosomal recessive
<i>TYRP1</i>	115501	Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive; [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3)
<i>UBR1</i>	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
<i>UROD</i>	613521	Porphyria cutanea tarda, 176100 (3), Autosomal recessive, Autosomal dominant; Porphyria, hepatoerythropoietic, 176100 (3), Autosomal recessive, Autosomal dominant
<i>UROS</i>	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
<i>USB1</i>	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
<i>VCX</i>	300229	No OMIM phenotype
<i>VCX3A</i>	300533	No OMIM phenotype
<i>WDR19</i>	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
<i>WDR35</i>	613602	Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive; Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive

<i>WNT10A</i>	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
<i>XPA</i>	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
<i>XPC</i>	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
<i>ZMPSTE24</i>	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: July 04, 2018

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.