Skin

Gene panel

Gene panel information

Gene panel	**Skin**
Version	5
Total genes	414
Activation date	Friday 16 june 2023
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
AAAS	99.43 %	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
AAGAB	99.98 %	614888	Keratoderma, palmoplantar, punctate type IA, 148600 (3), Autosomal dominant
ABCA12	99.81 %	607800	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500 (3), Autosomal recessive; Ichthyosis, congenital, autosomal recessive 4A, 601277 (3), Autosomal recessive
ABCB6	99.29 %	605452	Dyschromatosis universalis hereditaria 3, 615402 (3), Autosomal dominant; [Blood group, Langereis system], 111600 (3); Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 (3), Autosomal dominant; Microphthalmia/coloboma 7, 614497 (3), Autosomal dominant
ABCC9	99.79 %	601439	Cardiomyopathy, dilated, 1O, 608569 (3), Autosomal dominant; Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850 (3), Autosomal dominant; ?Atrial fibrillation, familial, 12, 614050 (3), Autosomal dominant; Intellectual disability and myopathy syndrome, 619719 (3), Autosomal recessive
ABCG8	99.84 %	605460	Sitosterolemia 1, 210250 (3), Autosomal recessive; {Gallbladder disease 4}, 611465 (3)
ABHD5	99.8 %	604780	Chanarin-Dorfman syndrome, 275630 (3), Autosomal recessive
ADAM10	99.63 %	602192	{Alzheimer disease 18, susceptibility to}, 615590 (3); Reticulate acropigmentation of Kitamura, 615537 (3), Autosomal dominant
ADAR	98.86 %	146920	Dyschromatosis symmetrica hereditaria, 127400 (3), Autosomal dominant; Aicardi-Goutieres syndrome 6, 615010 (3), Autosomal recessive
ADGRE2	98.06 %	606100	Vibratory urticaria, 125630 (3), Autosomal dominant
AGPAT2	99.75 %	603100	Lipodystrophy, congenital generalized, type 1, 608594 (3), Autosomal recessive
AHSG	99.97 %	138680	?Alopecia-intellectual disability syndrome 1, 203650 (3), Autosomal recessive
AKT2	99.9 %	164731	Diabetes mellitus, type II, 125853 (3), Autosomal dominant; Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900 (3), Autosomal dominant
ALAS2	99.87 %	301300	Anemia, sideroblastic, 1, 300751 (3), X-linked recessive; Protoporphyria, erythropoietic, X-linked, 300752 (3), X-linked
ALDH3A2	99.66 %	609523	Sjogren-Larsson syndrome, 270200 (3), Autosomal recessive
ALOX12B	99.65 %	603741	Ichthyosis, congenital, autosomal recessive 2, 242100 (3), Autosomal recessive
ALOXE3	99.8 %	607206	Ichthyosis, congenital, autosomal recessive 3, 606545 (3), Autosomal recessive
ALX4	99.89 %	605420	Parietal foramina 2, 609597 (3), Autosomal dominant; {Craniosynostosis 5, susceptibility to}, 615529 (3), Autosomal dominant; Frontonasal dysplasia 2, 613451 (3), Autosomal recessive
ANAPC1	72.56 %	608473	Rothmund-Thomson syndrome, type 1, 618625 (3), Autosomal recessive
ANTXR1	99.77 %	606410	GAPO syndrome, 230740 (3), Autosomal recessive; {?Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant
ANTXR2	99.56 %	608041	Hyaline fibromatosis syndrome, 228600 (3), Autosomal recessive

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AP1B1	99.9 %	600157	Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 (3), Autosomal recessive
AP1S1	98.44 %	603531	MEDNIK syndrome, 609313 (3), Autosomal recessive
AP3B1	99.78 %	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AP3D1	99.68 %	607246	?Hermansky-Pudlak syndrome 10, 617050 (3), Autosomal recessive
APCDD1	99.82 %	607479	Hypotrichosis 1, 605389 (3), Autosomal dominant
AQP5	99.72 %	600442	Palmoplantar keratoderma, Bothnian type, 600231 (3), Autosomal dominant
ARHGAP31	99.98 %	610911	Adams-Oliver syndrome 1, 100300 (3), Autosomal dominant
ARSL	99.68 %	300180	Chondrodysplasia punctata, X-linked recessive, 302950 (3), X-linked recessive
ATP2A2	99.86 %	108740	Acrokeratosis verruciformis, 101900 (3), Autosomal dominant; Darier disease, 124200 (3), Autosomal dominant; {Rhabdomyolysis, susceptibility to, 2}, 621236 (3), Autosomal dominant
ATP2C1	99.95 %	604384	Hailey-Hailey disease, 169600 (3), Autosomal dominant
ATP6V1B2	99.89 %	606939	Zimmermann-Laband syndrome 2, 616455 (3), Autosomal dominant; Deafness, congenital, with onychodystrophy, autosomal dominant, 124480 (3), Autosomal dominant
ATP7A	99.79 %	300011	Occipital horn syndrome, 304150 (3), X-linked recessive; Neuronopathy, distal hereditary motor, X-linked, 300489 (3), X-linked recessive; Menkes disease, 309400 (3), X-linked recessive
ATR	99.75 %	601215	Seckel syndrome 1, 210600 (3), Autosomal recessive; ?Cutaneous telangiectasia and cancer syndrome, familial, 614564 (3), Autosomal dominant
AXIN2	99.8 %	604025	Colorectal cancer, somatic, 114500 (3); Oligodontia-colorectal cancer syndrome, 608615 (3), Autosomal dominant
BANF1	97.05 %	603811	Nestor-Guillermo progeria syndrome, 614008 (3), Autosomal recessive
BCS1L	99.9 %	603647	GRACILE syndrome, 603358 (3), Autosomal recessive; Mitochondrial complex III deficiency, nuclear type 1, 124000 (3), Autosomal recessive; Bjornstad syndrome, 262000 (3), Autosomal recessive
BLM	99.87 %	604610	Bloom syndrome, 210900 (3), Autosomal recessive
BLOC1S3	99.88 %	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S6	99.66 %	604310	Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
BMS1	99.41 %	611448	?Aplasia cutis congenita, nonsyndromic, 107600 (3), Autosomal dominant
BRAF	99.53 %	164757	Melanoma, malignant, somatic, 155600 (3); LEOPARD syndrome 3, 613707 (3), Autosomal dominant; Cardiofaciocutaneous syndrome, 115150 (3), Autosomal dominant; Adenocarcinoma of lung, somatic, 211980 (3); Noonan syndrome 7, 613706 (3), Autosomal dominant; Colorectal cancer, somatic, 114500 (3); Nonsmall cell lung cancer, somatic, 211980 (3)
BSCL2	99.73 %	606158	Lipodystrophy, congenital generalized, type 2, 269700 (3), Autosomal recessive; Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112 (3), Autosomal dominant; Silver spastic paraplegia syndrome, 270685 (3), Autosomal dominant; Encephalopathy, progressive, with or without lipodystrophy, 615924 (3), Autosomal recessive
BTD	99.93 %	609019	Biotinidase deficiency, 253260 (3), Autosomal recessive
C1QA	99.8 %	120550	C1q deficiency 1, 613652 (3), Autosomal recessive
C1QB	98.46 %	120570	C1q deficiency 2, 620321 (3), Autosomal recessive
C1QC	99.81 %	120575	C1q deficiency 3, 620322 (3), Autosomal recessive
C5	99.89 %	120900	C5 deficiency, 609536 (3), Autosomal recessive; [Eculizumab, poor response to], 615749 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
CARD11	99.86 %	607210	B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive
CARD14	99.87 %	607211	Psoriasis 2, 602723 (3), Autosomal dominant; Pityriasis rubra pilaris, 173200 (3), Autosomal dominant
CASP10	99.48 %	601762	Autoimmune lymphoproliferative syndrome, type II, 603909 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Lymphoma, non-Hodgkin, somatic, 605027 (3)
CASP14	99.6 %	605848	Ichthyosis, congenital, autosomal recessive 12, 617320 (3), Autosomal recessive
CASP8	99.81 %	601763	{Breast cancer, protection against}, 114480 (3), Somatic mutation, Autosomal dominant; ?Caspase 8 lymphadenopathy syndrome, 607271 (3), Autosomal recessive; Hepatocellular carcinoma, somatic, 114550 (3); {Lung cancer, protection against}, 211980 (3), Somatic mutation, Autosomal dominant
CAST	99.89 %	114090	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295 (3), Autosomal recessive
CAV1	99.66 %	601047	Lipodystrophy, congenital generalized, type 3, 612526 (3), Autosomal recessive; Pulmonary hypertension, primary, 3, 615343 (3), Autosomal dominant; Lipodystrophy, familial partial, type 7, 606721 (3), Autosomal dominant
CAVIN1	99.8 %	603198	Lipodystrophy, congenital generalized, type 4, 613327 (3), Autosomal recessive
CBL	99.82 %	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563 (3), Autosomal dominant; ?Juvenile myelomonocytic leukemia, 607785 (3), Somatic mutation, Autosomal dominant
CDH1	99.69 %	192090	Ovarian cancer, somatic, 167000 (3); Blepharocheilodontic syndrome 1, 119580 (3), Autosomal dominant; Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215 (3), Autosomal dominant; Endometrial carcinoma, somatic, 608089 (3); Breast cancer, lobular, somatic, 114480 (3)
CDH3	99.86 %	114021	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553 (3), Autosomal recessive; Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 (3), Autosomal recessive
CDKN1B	99.99 %	600778	Multiple endocrine neoplasia, type IV, 610755 (3), Autosomal dominant
CDKN1C	99.99 %	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant
CDSN	99.91 %	602593	Hypotrichosis 2, 146520 (3), Autosomal dominant; Peeling skin syndrome 1, 270300 (3), Autosomal recessive
CERS3	99.83 %	615276	Ichthyosis, congenital, autosomal recessive 9, 615023 (3), Autosomal recessive
CHST8	99.98 %	610190	*No OMIM phenotypes*
CHUK	99.58 %	600664	?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339 (3), Autosomal recessive; ?Cocoon syndrome, 613630 (3), Autosomal recessive
CIB1	99.85 %	602293	{Epidermodysplasia verruciformis, susceptibility to, 3}, 618267 (3), Autosomal recessive
CIDEC	99.82 %	612120	?Lipodystrophy, familial partial, type 5, 615238 (3), Autosomal recessive
CLCF1	99.86 %	607672	Cold-induced sweating syndrome 2, 610313 (3), Autosomal recessive
CLCN7	99.83 %	602727	Hypopigmentation, organomegaly, and delayed myelination and development, 618541 (3), Autosomal dominant; Osteopetrosis, autosomal recessive 4, 611490 (3), Autosomal recessive; Osteopetrosis, autosomal dominant 2, 166600 (3), Autosomal dominant
CLDN1	99.87 %	603718	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 (3), Autosomal recessive
CLDN10	99.86 %	617579	HELIX syndrome, 617671 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
COG6	99.89 %	606977	Shaheen syndrome, 615328 (3), Autosomal recessive; Congenital disorder of glycosylation, type III, 614576 (3), Autosomal recessive
COL17A1	99.88 %	113811	Epithelial recurrent erosion dystrophy, 122400 (3), Autosomal dominant; Epidermolysis bullosa, junctional 4, intermediate, 619787 (3), Autosomal recessive
COL18A1	99.79 %	120328	Knobloch syndrome, type 1, 267750 (3), Autosomal recessive; Glaucoma, primary closed-angle, 618880 (3), Autosomal dominant
COL7A1	99.82 %	120120	Nail disorder, nonsyndromic congenital, 8, 607523 (3), Autosomal dominant; Epidermolysis bullosa dystrophica, Bart type, 132000 (3), Autosomal dominant; Epidermolysis bullosa dystrophica inversa, 226600 (3), Autosomal recessive; Epidermolysis bullosa dystrophica, autosomal recessive, 226600 (3), Autosomal recessive; Epidermolysis bullosa, pretibial, 131850 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa dystrophica, autosomal dominant, 131750 (3), Autosomal dominant; Transient bullous of the newborn, 131705 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa pruriginosa, 604129 (3), Autosomal recessive, Autosomal dominant; Epidermolysis bullosa dystrophica, localisata variant, 226600 (3), Autosomal recessive
COX7B	99.97 %	300885	Linear skin defects with multiple congenital anomalies 2, 300887 (3), X-linked dominant
CPN1	99.77 %	603103	Carboxypeptidase N deficiency, 212070 (3), Autosomal recessive
CST6	99.24 %	601891	?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535 (3), Autosomal recessive
CSTA	99.8 %	184600	Peeling skin syndrome 4, 607936 (3), Autosomal recessive
CTLA4	99.88 %	123890	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100 (3), Autosomal dominant
CTSB	99.94 %	116810	Keratolytic winter erythema, 148370 (4), Autosomal dominant
CTSC	99.79 %	602365	Periodontitis 1, juvenile, 170650 (3), Autosomal recessive; Haim-Munk syndrome, 245010 (3), Autosomal recessive; Papillon-Lefevre syndrome, 245000 (3), Autosomal recessive
CYLD	99.49 %	605018	Brooke-Spiegler syndrome, 605041 (3), Autosomal dominant; Cylindromatosis, familial, 132700 (3), Autosomal dominant; Trichoepithelioma, multiple familial, 1, 601606 (3), Autosomal dominant; ?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132 (3), Autosomal dominant
CYP11A1	99.76 %	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
CYP11B1	99.96 %	610613	Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant; Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive
CYP21A2	98.92 %	613815	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive
CYP26C1	99.58 %	608428	Focal facial dermal dysplasia 4, 614974 (3), Autosomal recessive
CYP4F22	99.69 %	611495	Ichthyosis, congenital, autosomal recessive 5, 604777 (3), Autosomal recessive
DCAF17	99.65 %	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DCLRE1C	99.78 %	605988	Severe combined immunodeficiency, Athabascan type, 602450 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
DCPS	99.81 %	610534	Al-Raqad syndrome, 616459 (3), Autosomal recessive
DCT	99.97 %	191275	Oculocutaneous albinism, type VIII, 619165 (3), Autosomal recessive
DDB2	99.98 %	600811	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
DDR2	98.65 %	191311	Warburg-Cinotti syndrome, 618175 (3), Autosomal dominant; Spondylometaepiphyseal dysplasia, short limb-hand type, 271665 (3), Autosomal recessive
DHCR24	99.28 %	606418	Desmosterolosis, 602398 (3), Autosomal recessive
DIP2B	98.94 %	611379	Intellectual developmental disorder, autosomal dominant, FRA12A type, 136630 (3), Autosomal dominant
DKC1	99.67 %	300126	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DLL4	99.71 %	605185	Adams-Oliver syndrome 6, 616589 (3), Autosomal dominant
DOCK6	99.4 %	614194	Adams-Oliver syndrome 2, 614219 (3), Autosomal recessive
DOCK8	99.75 %	611432	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700 (3), Autosomal recessive
DPH1	99.89 %	603527	Developmental delay with short stature, dysmorphic facial features, and sparse hair, 616901 (3), Autosomal recessive
DSC2	99.51 %	125645	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476 (3), Autosomal recessive, Autosomal dominant; Arrhythmogenic right ventricular dysplasia 11, 610476 (3), Autosomal recessive, Autosomal dominant
DSG1	99.9 %	125670	Keratosis palmoplantaris striata I, AD, 148700 (3), Autosomal dominant; Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 (3), Autosomal recessive
DSG3	99.91 %	169615	Blistering, acantholytic, of oral and laryngeal mucosa, 619226 (3), Autosomal recessive
DSG4	99.88 %	607892	Hypotrichosis 6, 607903 (3), Autosomal recessive
DSP	99.97 %	125647	Arrhythmogenic right ventricular dysplasia 8, 607450 (3), Autosomal dominant; Epidermolysis bullosa, lethal acantholytic, 609638 (3), Autosomal recessive; Keratosis palmoplantaris striata II, 612908 (3), Autosomal dominant; Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821 (3), Autosomal dominant; Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 (3), Autosomal recessive
DST	99.87 %	113810	Neuropathy, hereditary sensory and autonomic, type VI, 614653 (3), Autosomal recessive; Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425 (3), Autosomal recessive
DSTYK	99.39 %	612666	Spastic paraplegia 23, autosomal recessive, 270750 (3), Autosomal recessive; Congenital anomalies of kidney and urinary tract 1, 610805 (3), Autosomal dominant
DTNBP1	99.86 %	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
EBP	99.7 %	300205	MEND syndrome, 300960 (3), X-linked recessive; Chondrodysplasia punctata, X-linked dominant, 302960 (3), X-linked dominant
ECM1	99.76 %	602201	Urbach-Wiethe disease, 247100 (3), Autosomal recessive
EDA	99.51 %	300451	Tooth agenesis, selective, X-linked 1, 313500 (3), X-linked dominant; Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 (3), X-linked recessive
EDAR	99.34 %	604095	[Hair morphology 1, hair thickness], 612630 (3); 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
EDARADD	99.95 %	606603	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 (3), Autosomal recessive; Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 (3), Autosomal dominant

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EDN3	99.75 %	131242	Waardenburg syndrome, type 4B, 613265 (3), Autosomal recessive, Autosomal dominant; {Hirschsprung disease, susceptibility to, 4}, 613712 (3), Autosomal dominant
EDNRA	99.92 %	131243	{Migraine, resistance to}, 157300 (3), Autosomal dominant; Mandibulofacial dysostosis with alopecia, 616367 (3), Autosomal dominant
EDNRB	99.98 %	131244	{Hirschsprung disease, susceptibility to, 2}, 600155 (3), Autosomal dominant; ?ABCD syndrome, 600501 (3), Autosomal recessive; Waardenburg syndrome, type 4A, 277580 (3), Autosomal recessive, Autosomal dominant
ELOVL1	99.89 %	611813	Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 (3), Autosomal recessive, Autosomal dominant
ENPP1	99.81 %	173335	{Obesity, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Hypophosphatemic rickets, autosomal recessive, 2, 613312 (3), Autosomal recessive; {Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853 (3), Autosomal dominant; Arterial calcification, generalized, of infancy, 1, 208000 (3), Autosomal recessive; Cole disease, 615522 (3), Autosomal dominant
EOGT	98.82 %	614789	Adams-Oliver syndrome 4, 615297 (3), Autosomal recessive
EPG5	99.86 %	615068	Vici syndrome, 242840 (3), Autosomal recessive
ERCC2	99.66 %	126340	Xeroderma pigmentosum, group D, 278730 (3), Autosomal recessive; Trichothiodystrophy 1, photosensitive, 601675 (3), Autosomal recessive; ?Cerebrooculofacioskeletal syndrome 2, 610756 (3), Autosomal recessive
ERCC3	99.76 %	133510	Trichothiodystrophy 2, photosensitive, 616390 (3), Autosomal recessive; Xeroderma pigmentosum, group B, 610651 (3), Autosomal recessive
ERCC4	99.73 %	133520	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760 (3), Autosomal recessive; XFE progeroid syndrome, 610965 (3), Autosomal recessive; Xeroderma pigmentosum, group F, 278760 (3), Autosomal recessive; Fanconi anemia, complementation group Q, 615272 (3), Autosomal recessive
ERCC5	99.95 %	133530	Xeroderma pigmentosum, group G, 278780 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 3, 616570 (3), Autosomal recessive; Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 (3), Autosomal recessive
ERCC6	99.56 %	609413	UV-sensitive syndrome 1, 600630 (3), Autosomal recessive; Cerebrooculofacioskeletal syndrome 1, 214150 (3), Autosomal recessive; ?De Sanctis-Cacchione syndrome, 278800 (3), Autosomal recessive; Cockayne syndrome, type B, 133540 (3), Autosomal recessive; {Macular degeneration, age-related, susceptibility to, 5}, 613761 (3); Premature ovarian failure 11, 616946 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
ERCC8	99.6 %	609412	UV-sensitive syndrome 2, 614621 (3), Autosomal recessive; Cockayne syndrome, type A, 216400 (3), Autosomal recessive
EVC	99.73 %	604831	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; ?Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EVC2	99.79 %	607261	Ellis-van Creveld syndrome, 225500 (3), Autosomal recessive; Weyers acrofacial dysostosis, 193530 (3), Autosomal dominant
EXPH5	99.88 %	612878	Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028 (3), Autosomal recessive
F12	99.88 %	610619	Angioedema, hereditary, 3, 610618 (3), Autosomal dominant; Factor XII deficiency, 234000 (3), Autosomal recessive
FAM111B	99.91 %	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FAS	99.94 %	134637	Squamous cell carcinoma, burn scar-related, somatic (3); Autoimmune lymphoproliferative syndrome, type IA, 601859 (3), Autosomal dominant; {Autoimmune lymphoproliferative syndrome}, 601859 (3), Autosomal dominant

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FASLG	99.88 %	134638	Autoimmune lymphoproliferative syndrome, type IB, 601859 (3), Autosomal dominant; {Lung cancer, susceptibility to}, 211980 (3), Somatic mutation, Autosomal dominant
FDPS	100 %	134629	Porokeratosis 9, multiple types, 616631 (3), Autosomal dominant
FECH	99.96 %	612386	Protoporphyria, erythropoietic, 1, 177000 (3), Autosomal recessive
FERMT1	99.85 %	607900	Kindler syndrome, 173650 (3), Autosomal recessive
FGF5	99.96 %	165190	Trichomegaly, 190330 (3), Autosomal recessive
FGFR1	99.77 %	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FGFR3	99.87 %	134934	Muenke syndrome, 602849 (3), Autosomal dominant; SADDAN, 616482 (3), Autosomal dominant; Hypochondroplasia, 146000 (3), Autosomal dominant; Thanatophoric dysplasia, type II, 187601 (3), Autosomal dominant; Nevus, epidermal, somatic, 162900 (3); CATSHL syndrome, 610474 (3), Autosomal recessive, Autosomal dominant; Thanatophoric dysplasia, type I, 187600 (3), Autosomal dominant; Spermatocytic seminoma, somatic, 273300 (3); Bladder cancer, somatic, 109800 (3); LADD syndrome 2, 620192 (3), Autosomal dominant; Achondroplasia, 100800 (3), Autosomal dominant; Cervical cancer, somatic, 603956 (3); Colorectal cancer, somatic, 114500 (3); Crouzon syndrome with acanthosis nigricans, 612247 (3), Autosomal dominant
FLG	95.36 %	135940	Ichthyosis vulgaris, 146700 (3), Autosomal recessive, Autosomal dominant; {Dermatitis, atopic, susceptibility to, 2}, 605803 (3), Autosomal dominant
FLG2	99.88 %	616284	Peeling skin syndrome 6, 618084 (3), Autosomal recessive
FOXN1	99.44 %	600838	T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806 (3), Autosomal dominant; T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 (3), Autosomal recessive; ?T-cell immunodeficiency with thymic aplasia, 242700 (3), Autosomal recessive
FZD6	99.92 %	603409	Nail disorder, nonsyndromic congenital, 1, 161050 (3), Autosomal recessive
GBA	93.64 %	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Autosomal dominant, Multifactorial
GINS1	99.96 %	610608	Immunodeficiency 55, 617827 (3), Autosomal recessive
GJA1	100 %	121014	Erythrokeratodermia variabilis et progressiva 3, 617525 (3), Autosomal dominant; Craniometaphyseal dysplasia, autosomal recessive, 218400 (3), Autosomal recessive; Oculodentodigital dysplasia, 164200 (3), Autosomal dominant; Palmoplantar keratoderma with congenital alopecia, 104100 (3), Autosomal dominant; Syndactyly, type III, 186100 (3), Autosomal dominant; Oculodentodigital dysplasia, autosomal recessive, 257850 (3), Autosomal recessive
GJB2	100 %	121011	Keratoderma, palmoplantar, with deafness, 148350 (3), Autosomal dominant; Deafness, autosomal recessive 1A, 220290 (3), Digenic dominant, Autosomal recessive; Deafness, autosomal dominant 3A, 601544 (3), Autosomal dominant; Hystrix-like ichthyosis with deafness, 602540 (3), Autosomal dominant; Bart-Pumphrey syndrome, 149200 (3), Autosomal dominant; Keratitis-ichthyosis-deafness syndrome, 148210 (3), Autosomal dominant; Vohwinkel syndrome, 124500 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
GJB3	100 %	603324	Deafness, digenic, GJB2/GJB3, 220290 (3), Digenic dominant, Autosomal recessive; Erythrokeratodermia variabilis et progressiva 1, 133200 (3), Autosomal recessive, Autosomal dominant; Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644 (3), Autosomal dominant
GJB4	100 %	605425	Erythrokeratodermia variabilis et progressiva 2, 617524 (3), Autosomal dominant
GJB6	100 %	604418	Ectodermal dysplasia 2, Clouston type, 129500 (3), Autosomal dominant; Deafness, autosomal dominant 3B, 612643 (3), Autosomal dominant; Deafness, autosomal recessive 1B, 612645 (3), Autosomal recessive; Deafness, digenic GJB2/GJB6, 220290 (3), Digenic dominant, Autosomal recessive
GPNMB	99.81 %	604368	Amyloidosis, primary localized cutaneous, 3, 617920 (3), Autosomal recessive
GPR143	99.66 %	300808	Ocular albinism, type I, Nettleship-Falls type, 300500 (3), X-linked; Nystagmus 6, congenital, X-linked, 300814 (3), X-linked recessive
GREM2	100 %	608832	Tooth agenesis, selective, 9, 617275 (3), Autosomal dominant
GRHL2	99.68 %	608576	Deafness, autosomal dominant 28, 608641 (3), Autosomal dominant; Ectodermal dysplasia/short stature syndrome, 616029 (3), Autosomal recessive; Corneal dystrophy, posterior polymorphous, 4, 618031 (3), Autosomal dominant
GRIN2B	99.93 %	138252	Developmental and epileptic encephalopathy 27, 616139 (3), Autosomal dominant; Intellectual developmental disorder, autosomal dominant 6, with or without seizures, 613970 (3), Autosomal dominant
GTF2E2	99.98 %	189964	Trichothiodystrophy 6, nonphotosensitive, 616943 (3), Autosomal recessive
GTF2H5	99.97 %	608780	Trichothiodystrophy 3, photosensitive, 616395 (3), Autosomal recessive
HCCS	99.85 %	300056	Linear skin defects with multiple congenital anomalies 1, 309801 (3), X-linked dominant
HOXC13	99.63 %	142976	Ectodermal dysplasia 9, hair/nail type, 614931 (3), Autosomal recessive
HPGD	99.85 %	601688	?Digital clubbing, isolated congenital, 119900 (3), Autosomal recessive; Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100 (3), Autosomal recessive; Cranioosteoarthropathy, 259100 (3), Autosomal recessive
HPS1	99.94 %	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS3	99.85 %	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
HPS4	99.84 %	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS5	99.85 %	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
HPS6	99.81 %	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
HR	99.88 %	602302	Atrichia with papular lesions, 209500 (3), Autosomal recessive; Alopecia universalis, 203655 (3), Autosomal recessive
HRAS	99.97 %	190020	Bladder cancer, somatic, 109800 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Congenital myopathy with excess of muscle spindles, 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); Costello syndrome, 218040 (3), Autosomal dominant
HRURF	98.12 %	619257	Hypotrichosis 4, 146550 (3), Autosomal dominant
HSPA9	99.8 %	600548	Even-plus syndrome, 616854 (3), Autosomal recessive; Anemia, sideroblastic, 4, 182170 (3), Autosomal dominant
IFT122	99.49 %	606045	Cranioectodermal dysplasia 1, 218330 (3), Autosomal recessive
IFT43	99.83 %	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

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
IKBKG	54.61 %	300248	Incontinentia pigmenti, 308300 (3), X-linked dominant; Ectodermal dysplasia and immunodeficiency 1, 300291 (3), X-linked recessive; Immunodeficiency 33, 300636 (3), X-linked recessive; Autoinflammatory disease, systemic, X-linked, 301081 (3), X-linked
IL1RN	99.11 %	147679	Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852 (3), Autosomal recessive; {Gastric cancer risk after H. pylori infection}, 613659 (3); {Microvascular complications of diabetes 4}, 612628 (3); Interleukin 1 receptor antagonist deficiency, 612852 (3), Autosomal recessive
IL2RA	99.89 %	147730	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367 (3), Autosomal recessive; {Diabetes, mellitus, insulin-dependent, susceptibility to, 10}, 601942 (3)
IL31RA	99.86 %	609510	?Amyloidosis, primary localized cutaneous, 2, 613955 (3), Autosomal dominant
IL36RN	99.77 %	605507	Psoriasis 14, pustular, 614204 (3), Autosomal recessive
ITGA3	99.11 %	605025	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748 (3), Autosomal recessive
ITGA6	99.34 %	147556	Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817 (3), Autosomal recessive
ITGB4	99.89 %	147557	Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730 (3), Autosomal recessive; Epidermolysis bullosa, junctional 5A, intermediate, 619816 (3), Autosomal recessive
ITPR2	99.06 %	600144	?Anhidrosis, isolated, with normal sweat glands, 106190 (3), Autosomal recessive
JUP	99.59 %	173325	Naxos disease, 601214 (3), Autosomal recessive; ?Arrhythmogenic right ventricular dysplasia 12, 611528 (3), Autosomal dominant
KANK2	99.94 %	614610	Nephrotic syndrome, type 16, 617783 (3), Autosomal recessive; Palmoplantar keratoderma and woolly hair, 616099 (3), Autosomal recessive
KCNH1	99.86 %	603305	Zimmermann-Laband syndrome 1, 135500 (3), Autosomal dominant; Temple-Baraitser syndrome, 611816 (3), Autosomal dominant
KCTD1	99.84 %	613420	Scalp-ear-nipple syndrome, 181270 (3), Autosomal dominant
KDF1	99.25 %	616758	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 (3), Autosomal dominant
KDSR	99.92 %	136440	Erythrokeratodermia variabilis et progressiva 4, 617526 (3), Autosomal recessive
KEAP1	99.63 %	606016	*No OMIM phenotypes*
KIT	99.6 %	164920	Gastrointestinal stromal tumor, familial, 606764 (3), Autosomal dominant, Isolated cases; Mastocytosis, cutaneous, 154800 (3), Autosomal dominant; Piebaldism, 172800 (3), Autosomal dominant; Germ cell tumors, somatic, 273300 (3); Mastocytosis, systemic, somatic, 154800 (3); Leukemia, acute myeloid, somatic, 601626 (3)
KITLG	98.53 %	184745	Hyperpigmentation with or without hypopigmentation, 145250 (3), Autosomal dominant; Waardenburg syndrome, type 2F, 619947 (3), Autosomal recessive; Deafness, autosomal dominant 69, unilateral or asymmetric, 616697 (3), Autosomal dominant; [Skin/hair/eye pigmentation 7, blond/brown hair], 611664 (3)
KLHL24	99.87 %	611295	Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236 (3), Autosomal recessive; Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294 (3), Autosomal dominant
KRAS	97.26 %	190070	Gastric cancer, somatic, 613659 (3); Oculoectodermal syndrome, somatic, 600268 (3); Breast cancer, somatic, 114480 (3); Noonan syndrome 3, 609942 (3), Autosomal dominant; RAS-associated autoimmune leukoproliferative disorder, 614470 (3), Autosomal dominant; Arteriovenous malformation of the brain, somatic, 108010 (3); Lung cancer, somatic, 211980 (3); Pancreatic carcinoma, somatic, 260350 (3); Leukemia, acute myeloid, somatic, 601626 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Cardiofaciocutaneous syndrome 2, 615278 (3), Autosomal dominant; Bladder cancer, somatic, 109800 (3)

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
KREMEN1	99.75 %	609898	Ectodermal dysplasia 13, hair/tooth type, 617392 (3), Autosomal recessive
KRT1	99.7 %	139350	Ichthyosis, annular epidermolytic 2, 620148 (3), Autosomal dominant; Palmoplantar keratoderma, nonepidermolytic, 600962 (3), Autosomal dominant; Epidermolytic hyperkeratosis 1, 113800 (3), Autosomal dominant; Palmoplantar keratoderma, epidermolytic, 2, 620411 (3), Autosomal dominant; Keratosis palmoplantaris striata III, 607654 (3); Ichthyosis histrix, Curth-Macklin type, 146590 (3), Autosomal dominant
KRT10	99.88 %	148080	Ichthyosis, annular epidermolytic 1, 607602 (3), Autosomal dominant; Epidermolytic hyperkeratosis 2B, autosomal recessive, 620707 (3), Autosomal recessive; Epidermolytic hyperkeratosis 2A, autosomal dominant, 620150 (3), Autosomal dominant; ?Ichthyosis histrix, Lambert type, 146600 (3), Autosomal dominant; Ichthyosis with confetti, 609165 (3), Autosomal dominant
KRT13	99.91 %	148065	White sponge nevus 2, 615785 (3), Autosomal dominant
KRT14	99.88 %	148066	Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001 (3), Autosomal recessive; Epidermolysis bullosa simplex 1C, localized, 131800 (3), Autosomal dominant; Dermatopathia pigmentosa reticularis, 125595 (3), Autosomal dominant; Epidermolysis bullosa simplex 1A, generalized severe, 131760 (3), Autosomal dominant; Naegeli-Franceschetti-Jadassohn syndrome, 161000 (3), Autosomal dominant; Epidermolysis bullosa simplex 1B, generalized intermediate, 131900 (3), Autosomal dominant
KRT16	99.95 %	148067	Palmoplantar keratoderma, nonepidermolytic, focal, 613000 (3), Autosomal dominant; Pachyonychia congenita 1, 167200 (3), Autosomal dominant
KRT17	99.95 %	148069	Steatocystoma multiplex, 184500 (3), Autosomal dominant; Pachyonychia congenita 2, 167210 (3), Autosomal dominant
KRT2	99.68 %	600194	Ichthyosis bullosa of Siemens, 146800 (3), Autosomal dominant
KRT25	99.2 %	616646	Woolly hair, autosomal recessive 3, 616760 (3), Autosomal recessive
KRT3	98.91 %	148043	Meesmann corneal dystrophy 2, 618767 (3), Autosomal dominant
KRT5	99.91 %	148040	Epidermolysis bullosa simplex 2A, generalized severe, 619555 (3), Autosomal dominant; Dowling-Degos disease 1, 179850 (3), Autosomal dominant; Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960 (3), Autosomal dominant; Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599 (3), Autosomal recessive; Epidermolysis bullosa simplex 2B, generalized intermediate, 619588 (3), Autosomal dominant; Epidermolysis bullosa simplex 2C, localized, 619594 (3), Autosomal dominant; Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352 (3), Autosomal dominant
KRT6A	99.86 %	148041	Pachyonychia congenita 3, 615726 (3), Autosomal dominant
KRT6B	99.83 %	148042	Pachyonychia congenita 4, 615728 (3), Autosomal dominant
KRT6C	94.58 %	612315	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735 (3), Autosomal dominant
KRT7	99.54 %	148059	*No OMIM phenotypes*
KRT71	99.93 %	608245	?Hypotrichosis 13, 615896 (3), Autosomal dominant
KRT74	98.52 %	608248	Woolly hair, autosomal dominant, 194300 (3), Autosomal dominant; ?Hypotrichosis 3, 613981 (3), Autosomal dominant; ?Ectodermal dysplasia 7, hair/nail type, 614929 (3), Autosomal recessive
KRT81	75.62 %	602153	Monilethrix 2, 621169 (3), Autosomal dominant
KRT83	99.99 %	602765	Monilethrix 3, 621170 (3), Autosomal dominant; Erythrokeratodermia variabilis et progressiva 5, 617756 (3), Autosomal recessive
KRT85	99.74 %	602767	Ectodermal dysplasia 4, hair/nail type, 602032 (3), Autosomal recessive
KRT86	75.47 %	601928	Monilethrix 1, 158000 (3), Autosomal dominant
KRT9	99.72 %	607606	Palmoplantar keratoderma, epidermolytic, 1, 144200 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
LAMA3	99.76 %	600805	Epidermolysis bullosa, junctional 2A, intermediate, 619783 (3), Autosomal recessive; Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660 (3), Autosomal recessive; Epidermolysis bullosa, junctional 2B, severe, 619784 (3), Autosomal recessive
LAMB3	99.71 %	150310	Epidermolysis bullosa, junctional 1B, severe, 226700 (3), Autosomal recessive; Epidermolysis bullosa, junctional 1A, intermediate, 226650 (3), Autosomal recessive; Amelogenesis imperfecta, type IA, 104530 (3), Autosomal dominant
LAMC2	99.5 %	150292	Epidermolysis bullosa, junctional 3B, severe, 619786 (3), Autosomal recessive; Epidermolysis bullosa, junctional 3A, intermediate, 619785 (3), Autosomal recessive
LBR	99.84 %	600024	Pelger-Huet anomaly, 169400 (3), Autosomal dominant; ?Reynolds syndrome, 613471 (3), Autosomal dominant; Rhizomelic skeletal dysplasia with or without Pelger-Huet anomaly, 618019 (3), Autosomal recessive; Greenberg skeletal dysplasia, 215140 (3), Autosomal recessive
LEMD3	98.11 %	607844	Buschke-Ollendorff syndrome, 166700 (3), Autosomal dominant; Osteopoikilosis with or without melorheostosis, 166700 (3), Autosomal dominant
LIPE	99.9 %	151750	Lipodystrophy, familial partial, type 6, 615980 (3), Autosomal recessive
LIPH	99.91 %	607365	Hypotrichosis 7, 604379 (3), Autosomal recessive; Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 (3), Autosomal recessive
LMNA	99.84 %	150330	Mandibuloacral dysplasia, 248370 (3), Autosomal recessive; Heart-hand syndrome, Slovenian type, 610140 (3), Autosomal dominant; Cardiomyopathy, dilated, 1A, 115200 (3), Autosomal dominant; Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516 (3), Autosomal recessive; Restrictive dermopathy 2, 619793 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2B1, 605588 (3), Autosomal recessive; Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350 (3), Autosomal dominant; Hutchinson-Gilford progeria, 176670 (3), Autosomal dominant; Lipodystrophy, familial partial, type 2, 151660 (3), Autosomal dominant; Muscular dystrophy, congenital, 613205 (3), Autosomal dominant; Malouf syndrome, 212112 (3), Autosomal dominant
LMNB2	99.74 %	150341	Microcephaly 27, primary, autosomal dominant, 619180 (3), Autosomal dominant; ?Epilepsy, progressive myoclonic, 9, 616540 (3), Autosomal recessive; {Lipodystrophy, partial, acquired, susceptibility to}, 608709 (3), Autosomal dominant
LMX1B	99.92 %	602575	Focal segmental glomerulosclerosis 10, 256020 (3), Autosomal dominant; Nail-patella syndrome, 161200 (3), Autosomal dominant
LORICRIN	99.92 %	152445	Vohwinkel syndrome with ichthyosis, 604117 (3), Autosomal dominant
LPAR6	99.99 %	609239	Hypotrichosis 8, 278150 (3), Autosomal recessive; Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 (3), Autosomal recessive
LRMDA	99 %	614537	Albinism, oculocutaneous, type VII, 615179 (3), Autosomal recessive
LRP1	99.42 %	107770	?Keratosis pilaris atrophicans, 604093 (3), Autosomal recessive; Developmental dysplasia of the hip 3, 620690 (3), Autosomal dominant
LSS	99.85 %	600909	Hypotrichosis 14, 618275 (3), Autosomal recessive; Cataract 44, 616509 (3), Autosomal recessive; Alopecia-intellectual disability syndrome 4, 618840 (3), Autosomal recessive
LYST	99.87 %	606897	Chediak-Higashi syndrome, 214500 (3), Autosomal recessive
LZTR1	99.3 %	600574	Noonan syndrome 2, 605275 (3), Autosomal recessive; Noonan syndrome 10, 616564 (3), Autosomal dominant; {Schwannomatosis-2, susceptibility to}, 615670 (3), Autosomal dominant
MAP2K1	99.43 %	176872	Cardiofaciocutaneous syndrome 3, 615279 (3), Autosomal dominant; Melorheostosis, isolated, somatic mosaic, 155950 (3)
MAP2K2	99.93 %	601263	Cardiofaciocutaneous syndrome 4, 615280 (3), Autosomal dominant
MAPK1	99.73 %	176948	Noonan syndrome 13, 619087 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
MAPRE2	99.74 %	605789	Symmetric circumferential skin creases, congenital, 2, 616734 (3), Autosomal dominant
MARS1	99.43 %	156560	Spastic paraplegia 70, autosomal recessive, 620323 (3), Autosomal recessive; Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant
MBTPS2	99.85 %	300294	Keratosis follicularis spinulosa decalvans, X-linked, 308800 (3), X-linked recessive; Osteogenesis imperfecta, type XIX, 301014 (3), X-linked recessive; IFAP syndrome with or without BRESHECK syndrome, 308205 (3), X-linked recessive; ?Olmsted syndrome, X-linked, 300918 (3), X-linked recessive
MC1R	99.47 %	155555	[Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (3); [Skin/hair/eye pigmentation 2, red hair/fair skin], 266300 (3), Autosomal recessive; [Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300 (3), Autosomal recessive; {Melanoma, cutaneous malignant, 5}, 613099 (3); {Albinism, oculocutaneous, type II, modifier of}, 203200 (3), Autosomal recessive; {UV-induced skin damage}, 266300 (3), Autosomal recessive
MC2R	99.99 %	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive
MC4R	99.96 %	155541	Obesity (BMIQ20), 618406 (3), Autosomal recessive, Autosomal dominant; {Obesity, resistance to (BMIQ20)}, 618406 (3), Autosomal recessive, Autosomal dominant
MCM4	99.77 %	602638	Immunodeficiency 54, 609981 (3), Autosomal recessive
MEN1	99.91 %	613733	Lipoma, somatic (3); Angiofibroma, somatic (3); Multiple endocrine neoplasia 1, 131100 (3), Autosomal dominant; Carcinoid tumor of lung (3); Adrenal adenoma, somatic (3); Parathyroid adenoma, somatic (3)
MFN2	99.82 %	608507	Lipomatosis, multiple symmetric, with or without peripheral neuropathy, 151800 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260 (3), Autosomal dominant; Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087 (3), Autosomal recessive; Hereditary motor and sensory neuropathy VIA, 601152 (3), Autosomal dominant
MITF	99.9 %	156845	Waardenburg syndrome, type 2A, 193510 (3), Autosomal dominant; {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 (3); Tietz albinism-deafness syndrome, 103500 (3), Autosomal dominant; COMMAD syndrome, 617306 (3), Autosomal recessive
MLPH	99.88 %	606526	Griscelli syndrome, type 3, 609227 (3), Autosomal recessive
MPDU1	99.91 %	604041	Congenital disorder of glycosylation, type If, 609180 (3), Autosomal recessive
MPLKIP	98.89 %	609188	Trichothiodystrophy 4, nonphotosensitive, 234050 (3), Autosomal recessive
MSMO1	99.89 %	607545	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834 (3), Autosomal recessive
MSX1	99.87 %	142983	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 (3), Autosomal dominant; Ectodermal dysplasia 3, Witkop type, 189500 (3), Autosomal dominant; Orofacial cleft 5, 608874 (3), Autosomal dominant
MVD	99.56 %	603236	Porokeratosis 7, multiple types, 614714 (3), Autosomal dominant
MYO5A	99.86 %	160777	Griscelli syndrome, type 1, 214450 (3), Autosomal recessive
NCSTN	98.83 %	605254	Acne inversa, familial, 1, 142690 (3), Autosomal dominant
NDUFB11	98.75 %	300403	Linear skin defects with multiple congenital anomalies 3, 300952 (3), X-linked dominant; ?Mitochondrial complex I deficiency, nuclear type 30, 301021 (3), X-linked
NECTIN1	99.95 %	600644	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 (3), Autosomal recessive; Orofacial cleft 7, 225060 (3), Autosomal recessive
NECTIN4	99.17 %	609607	Ectodermal dysplasia-syndactyly syndrome 1, 613573 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
NF1	99.83 %	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
NFKBIA	99.86 %	164008	Ectodermal dysplasia and immunodeficiency 2, 612132 (3), Autosomal dominant
NHP2	100 %	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
NIPAL4	99.97 %	609383	Ichthyosis, congenital, autosomal recessive 6, 612281 (3), Autosomal recessive
NLRC4	99.89 %	606831	?Familial cold autoinflammatory syndrome 4, 616115 (3), Autosomal dominant; Autoinflammation with infantile enterocolitis, 616050 (3), Autosomal dominant
NLRP1	99.92 %	606636	{Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579 (3); ?Respiratory papillomatosis, juvenile recurrent, congenital, 618803 (3), Autosomal recessive; Autoinflammation with arthritis and dyskeratosis, 617388 (3), Autosomal recessive, Autosomal dominant; Palmoplantar carcinoma, multiple self-healing, 615225 (3), Autosomal dominant
NLRP12	99.92 %	609648	Familial cold autoinflammatory syndrome 2, 611762 (3), Autosomal dominant
NLRP3	99.95 %	606416	CINCA syndrome, 607115 (3), Autosomal dominant; Familial cold inflammatory syndrome 1, 120100 (3), Autosomal dominant; Keratoendothelitis fugax hereditaria, 148200 (3), Autosomal dominant; Deafness, autosomal dominant 34, with or without inflammation, 617772 (3), Autosomal dominant; Muckle-Wells syndrome, 191900 (3), Autosomal dominant
NNT	99.79 %	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive
NOP10	99.98 %	606471	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400 (3), Autosomal dominant; ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425 (3), Autosomal recessive; ?Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NOTCH1	99.8 %	190198	Adams-Oliver syndrome 5, 616028 (3), Autosomal dominant; Aortic valve disease 1, 109730 (3), Autosomal dominant
NR5A1	99.09 %	184757	46XX sex reversal 4, 617480 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant; Adrenocortical insufficiency, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant
NRAS	99.69 %	164790	Noonan syndrome 6, 613224 (3), Autosomal dominant; ?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (3); Melanocytic nevus syndrome, congenital, somatic, 137550 (3); Epidermal nevus, somatic, 162900 (3); Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (3); Thyroid carcinoma, follicular, somatic, 188470 (3); Neurocutaneous melanosis, somatic, 249400 (3); Colorectal cancer, somatic, 114500 (3)
NSDHL	99.66 %	300275	CK syndrome, 300831 (3), X-linked recessive; CHILD syndrome, 308050 (3), X-linked dominant
OCA2	97.93 %	611409	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 (3), Autosomal recessive; [Skin/hair/eye pigmentation 1, blond/brown hair], 227220 (3), Autosomal recessive; Albinism, brown oculocutaneous, 203200 (3), Autosomal recessive; Albinism, oculocutaneous, type II, 203200 (3), Autosomal recessive
OSMR	99.8 %	601743	Amyloidosis, primary localized cutaneous, 1, 105250 (3), Autosomal dominant
PADI3	98.72 %	606755	Uncombable hair syndrome, 191480 (3), Autosomal recessive
PARN	99.09 %	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PAX3	99.62 %	606597	Craniofacial-deafness-hand syndrome, 122880 (3), Autosomal dominant; Waardenburg syndrome, type 3, 148820 (3), Autosomal recessive, Autosomal dominant; Waardenburg syndrome, type 1, 193500 (3), Autosomal dominant; Rhabdomyosarcoma 2, alveolar, 268220 (3), Somatic mutation
PAX6	99.91 %	607108	Optic nerve hypoplasia, 165550 (3), Autosomal dominant; Cataract with late-onset corneal dystrophy, 106210 (3), Autosomal dominant; Microphthalmia/coloboma 12, 120200 (3), Autosomal dominant; ?Coloboma of optic nerve, 120430 (3), Autosomal dominant; Aniridia, 106210 (3), Autosomal dominant; Anterior segment dysgenesis 5, multiple subtypes, 604229 (3), Autosomal dominant; ?Morning glory disc anomaly, 120430 (3), Autosomal dominant; Foveal hypoplasia 1, 136520 (3), Autosomal dominant; Keratitis, 148190 (3), Autosomal dominant
PDGFRB	99.81 %	173410	Premature aging syndrome, Penttinen type, 601812 (3), Autosomal dominant; ?Ocular pterygium-digital keloid dysplasia syndrome, 621091 (3), Autosomal dominant; Kosaki overgrowth syndrome, 616592 (3), Autosomal dominant; Myofibromatosis, infantile, 1, 228550 (3), Autosomal dominant; Basal ganglia calcification, idiopathic, 4, 615007 (3), Autosomal dominant
PEPD	99.42 %	613230	Prolidase deficiency, 170100 (3), Autosomal recessive
PERP	99.9 %	609301	Erythrokeratodermia variabilis et progressiva 7, 619209 (3), Autosomal recessive; Olmsted syndrome 2, 619208 (3), Autosomal dominant
PEX7	99.68 %	601757	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3), Autosomal recessive; Peroxisome biogenesis disorder 9B, 614879 (3), Autosomal recessive
PGM3	99.78 %	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PHGDH	99.34 %	606879	Neu-Laxova syndrome 1, 256520 (3), Autosomal recessive; Phosphoglycerate dehydrogenase deficiency, 601815 (3), Autosomal recessive
PHYH	99.98 %	602026	Refsum disease, 266500 (3), Autosomal recessive
PIEZO1	99.83 %	611184	[ER blood group system], 620207 (3), Autosomal recessive; Lymphatic malformation 6, 616843 (3), Autosomal recessive; Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 (3), Autosomal dominant
PIGL	99.79 %	605947	CHIME syndrome, 280000 (3), Autosomal recessive
PIGT	99.83 %	610272	?Paroxysmal nocturnal hemoglobinuria 2, 615399 (3), Somatic mutation, Autosomal dominant; Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398 (3), Autosomal recessive
PKP1	99.71 %	601975	Ectodermal dysplasia/skin fragility syndrome, 604536 (3), Autosomal recessive
PLCD1	99.81 %	602142	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 (3), Autosomal recessive, Autosomal dominant
PLCG2	99.91 %	600220	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878 (3), Autosomal dominant; Familial cold autoinflammatory syndrome 3, 614468 (3), Autosomal dominant
PLEC	99.95 %	601282	?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487 (3), Autosomal recessive; Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670 (3), Autosomal recessive; Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138 (3), Autosomal recessive; Epidermolysis bullosa simplex 5A, Ogna type, 131950 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 (3), Autosomal recessive
PLG	99.61 %	173350	Dysplasminogenemia, 217090 (3), Autosomal recessive; Angioedema, hereditary, 4, 619360 (3), Autosomal dominant; Plasminogen deficiency, type I, 217090 (3), Autosomal recessive
PLIN1	99.57 %	170290	Lipodystrophy, familial partial, type 4, 613877 (3), Autosomal dominant
PMVK	96.34 %	607622	Porokeratosis 1, multiple types, 175800 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
PNPLA1	99.92 %	612121	Ichthyosis, congenital, autosomal recessive 10, 615024 (3), Autosomal recessive
PNPLA2	99.9 %	609059	Neutral lipid storage disease with myopathy, 610717 (3), Autosomal recessive
PNPLA6	99.93 %	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive
POFUT1	99.85 %	607491	Dowling-Degos disease 2, 615327 (3), Autosomal dominant
POGLUT1	99.82 %	615618	Dowling-Degos disease 4, 615696 (3), Autosomal dominant; Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 (3), Autosomal recessive
POLA1	99.61 %	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	99.77 %	603968	Xeroderma pigmentosum, variant type, 278750 (3), Autosomal recessive
POMC	99.85 %	176830	{Obesity, early-onset, susceptibility to}, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial; Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive
POMP	99.97 %	613386	Proteasome-associated autoinflammatory syndrome 2, 618048 (3), Autosomal dominant; Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952 (3), Autosomal recessive
PORCN	99.81 %	300651	Focal dermal hypoplasia, 305600 (3), X-linked dominant
PPARG	99.93 %	601487	{Diabetes, type 2}, 125853 (3), Autosomal dominant; Insulin resistance, severe, digenic, 125853 (3), Autosomal dominant; Lipodystrophy, familial partial, type 3, 604367 (3), Autosomal dominant; Obesity, severe, 601665 (3), Autosomal recessive, Autosomal dominant, Multifactorial
PPOX	98.87 %	600923	Variegate porphyria, childhood-onset, 620483 (3), Autosomal recessive; Variegate porphyria, 176200 (3), Autosomal dominant
PPP1CB	99.51 %	600590	Noonan syndrome-like disorder with loose anagen hair 2, 617506 (3), Autosomal dominant
PRKCD	99.51 %	176977	Autoimmune lymphoproliferative syndrome, type III, 615559 (3), Autosomal recessive
PRKD1	99.92 %	605435	Congenital heart defects and ectodermal dysplasia, 617364 (3), Autosomal dominant
PSAT1	99.75 %	610936	Neu-Laxova syndrome 2, 616038 (3), Autosomal recessive; Phosphoserine aminotransferase deficiency, 610992 (3), Autosomal recessive
PSENEN	99.87 %	607632	Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 (3), Autosomal dominant
PSMB8	99.74 %	177046	Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 (3), Autosomal recessive
PTCH1	99.87 %	601309	Basal cell nevus syndrome 1, 109400 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3); Holoprosencephaly 7, 610828 (3), Autosomal dominant
PTCH2	99.8 %	603673	Medulloblastoma, somatic, 155255 (3); Basal cell carcinoma, somatic, 605462 (3)
PTEN	99.87 %	601728	{Glioma susceptibility 2}, 613028 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant
PTPN11	99.84 %	176876	Noonan syndrome 1, 163950 (3), Autosomal dominant; LEOPARD syndrome 1, 151100 (3), Autosomal dominant; Metachondromatosis, 156250 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, somatic, 607785 (3)
PTPRF	99.57 %	179590	?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001 (3), Autosomal recessive
RAB27A	99.97 %	603868	Griscelli syndrome, type 2, 607624 (3), Autosomal recessive
RAD50	99.81 %	604040	Nijmegen breakage syndrome-like disorder, 613078 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
RAF1	99.91 %	164760	Cardiomyopathy, dilated, 1NN, 615916 (3), Autosomal dominant; Noonan syndrome 5, 611553 (3), Autosomal dominant; LEOPARD syndrome 2, 611554 (3), Autosomal dominant
RAG1	100 %	179615	Omenn syndrome, 603554 (3), Autosomal recessive; Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (3)
RAG2	99.99 %	179616	Severe combined immunodeficiency, B cell-negative, 601457 (3), Autosomal recessive; Combined cellular and humoral immune defects with granulomas, 233650 (3), Autosomal recessive; Omenn syndrome, 603554 (3), Autosomal recessive
RBM28	99.92 %	612074	?Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 (3), Autosomal recessive
RBPJ	99.83 %	147183	Adams-Oliver syndrome 3, 614814 (3), Autosomal dominant
RECQL4	99.91 %	603780	Baller-Gerold syndrome, 218600 (3), Autosomal recessive; Rothmund-Thomson syndrome, type 2, 268400 (3), Autosomal recessive; RAPADILINO syndrome, 266280 (3), Autosomal recessive
RET	99.85 %	164761	{Hirschsprung disease, susceptibility to, 1}, 142623 (3), Autosomal dominant; Multiple endocrine neoplasia IIA, 171400 (3), Autosomal dominant; {Hirschsprung disease, protection against}, 142623 (3), Autosomal dominant; Medullary thyroid carcinoma, 155240 (3), Autosomal dominant; Pheochromocytoma, 171300 (3), Autosomal dominant; Multiple endocrine neoplasia IIB, 162300 (3), Autosomal dominant
RHBDF2	99.74 %	614404	Tylosis with esophageal cancer, 148500 (3), Autosomal dominant
RHOA	97.54 %	165390	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727 (3)
RIN2	99.88 %	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 (3), Autosomal recessive
RIPK4	99.68 %	605706	CHAND syndrome, 214350 (3), Autosomal recessive; Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 (3), Autosomal recessive
RIT1	99.11 %	609591	Noonan syndrome 8, 615355 (3), Autosomal dominant
RMRP	98.59 %	157660	Anauxetic dysplasia 1, 607095 (3), Autosomal recessive; Metaphyseal dysplasia without hypotrichosis, 250460 (3), Autosomal recessive; Cartilage-hair hypoplasia, 250250 (3), Autosomal recessive
RNF113A	99.98 %	300951	Trichothiodystrophy 5, nonphotosensitive, 300953 (3), X-linked
RPL21	98.37 %	603636	Hypotrichosis 12, 615885 (3), Autosomal dominant
RSPO1	99.45 %	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
RSPO4	96.41 %	610573	Anonychia congenita, 206800 (3), Autosomal recessive
RTEL1	99.92 %	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 (3), Autosomal dominant
SAMHD1	99.94 %	606754	?Chilblain lupus 2, 614415 (3), Autosomal dominant; Aicardi-Goutieres syndrome 5, 612952 (3), Autosomal recessive
SASH1	99.79 %	607955	Dyschromatosis universalis hereditaria 1, 127500 (3), Autosomal dominant; ?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SCN9A	99.74 %	603415	Erythermalgia, primary, 133020 (3), Autosomal dominant; Insensitivity to pain, congenital, 243000 (3), Autosomal recessive; Small fiber neuropathy, 133020 (3), Autosomal dominant; Paroxysmal extreme pain disorder, 167400 (3), Autosomal dominant; Neuropathy, hereditary sensory and autonomic, type IID, 243000 (3), Autosomal recessive
SDR9C7	99.91 %	609769	Ichthyosis, congenital, autosomal recessive 13, 617574 (3), Autosomal recessive
SERPINB7	99.95 %	603357	Palmoplantar keratoderma, Nagashima type, 615598 (3), Autosomal recessive
SERPINB8	99.76 %	601697	Peeling skin syndrome 5, 617115 (3), Autosomal recessive
SERPING1	99.9 %	606860	Angioedema, hereditary, 1 and 2, 106100 (3), Autosomal recessive, Autosomal dominant; Complement component 4, partial deficiency of, 120790 (3), Autosomal dominant
SGPL1	99.71 %	603729	RENI syndrome, 617575 (3), Autosomal recessive
SHOC2	99.83 %	602775	Noonan syndrome-like with loose anagen hair 1, 607721 (3), Autosomal dominant
SKIV2L	99.43 %	600478	Trichohepatoenteric syndrome 2, 614602 (3), Autosomal recessive
SLC17A9	99.77 %	612107	Porokeratosis 8, disseminated superficial actinic type, 616063 (3), Autosomal dominant
SLC24A5	99.96 %	609802	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750 (3), Autosomal recessive; Albinism, oculocutaneous, type VI, 113750 (3), Autosomal recessive
SLC27A4	99.85 %	604194	Ichthyosis prematurity syndrome, 608649 (3), Autosomal recessive
SLC29A3	99.61 %	612373	Histiocytosis-lymphadenopathy plus syndrome, 602782 (3), Autosomal recessive
SLC39A4	99.85 %	607059	Acrodermatitis enteropathica, 201100 (3), Autosomal recessive
SLC45A2	99.82 %	606202	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, black/nonblack hair], 227240 (3), Autosomal recessive; Albinism, oculocutaneous, type IV, 606574 (3), Autosomal recessive; [Skin/hair/eye pigmentation 5, dark/fair skin], 227240 (3), Autosomal recessive
SLURP1	99.55 %	606119	Meleda disease, 248300 (3), Autosomal recessive
SMARCA2	99.82 %	600014	Nicolaides-Baraitser syndrome, 601358 (3), Autosomal dominant; Blepharophimosis-impaired intellectual development syndrome, 619293 (3), Autosomal dominant
SMARCAD1	99.68 %	612761	Basan syndrome, 129200 (3), Autosomal dominant; Huriez syndrome, 181600 (3), Autosomal dominant; Adermatoglyphia, 136000 (3), Autosomal dominant
SNAI2	99.97 %	602150	*No OMIM phenotypes*
SNAP29	99.51 %	604202	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3), Autosomal recessive
SNRPE	98.33 %	128260	Hypotrichosis 11, 615059 (3), Autosomal dominant
SOS1	99.57 %	182530	Noonan syndrome 4, 610733 (3), Autosomal dominant; Fibromatosis, gingival, 1, 135300 (3), Autosomal dominant
SOS2	99.81 %	601247	Noonan syndrome 9, 616559 (3), Autosomal dominant
SOX10	99.99 %	602229	Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant
SOX18	99.84 %	601618	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3), Autosomal recessive; Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 (3), Autosomal dominant
SPINK5	99.75 %	605010	Netherton syndrome, 256500 (3), Autosomal recessive
SPINT1	99.97 %	605123	*No OMIM phenotypes*
SPRED1	99.97 %	609291	Legius syndrome, 611431 (3), Autosomal dominant
SRD5A3	99.98 %	611715	Kahrizi syndrome, 612713 (3), Autosomal recessive; Congenital disorder of glycosylation, type Iq, 612379 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
SREBF1	99.51 %	184756	Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016 (3), Autosomal dominant; Mucoepithelial dysplasia, hereditary, 158310 (3), Autosomal dominant
ST14	99.8 %	606797	Ichthyosis, congenital, autosomal recessive 11, 602400 (3), Autosomal recessive
ST3GAL5	99.49 %	604402	Salt and pepper developmental regression syndrome, 609056 (3), Autosomal recessive
STAT3	99.7 %	102582	Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
STIM1	99.89 %	605921	Myopathy, tubular aggregate, 1, 160565 (3), Autosomal dominant; Stormorken syndrome, 185070 (3), Autosomal dominant; Immunodeficiency 10, 612783 (3), Autosomal recessive
STK11	99.95 %	602216	Melanoma, malignant, somatic, 155600 (3); Pancreatic cancer, somatic, 260350 (3); Peutz-Jeghers syndrome, 175200 (3), Autosomal dominant; Testicular tumor, somatic, 273300 (3)
STS	99.8 %	300747	Ichthyosis, X-linked, 308100 (3), X-linked recessive
SUFU	99.9 %	607035	{Meningioma, familial, susceptibility to}, 607174 (3), Autosomal dominant; Joubert syndrome 32, 617757 (3), Autosomal recessive; Basal cell nevus syndrome 2, 620343 (3); {Medulloblastoma}, 155255 (3), Somatic mutation, Autosomal recessive, Autosomal dominant
SULT2B1	99.62 %	604125	Ichthyosis, congenital, autosomal recessive 14, 617571 (3), Autosomal recessive
SUMF1	99.88 %	607939	Multiple sulfatase deficiency, 272200 (3), Autosomal recessive
TARS1	99.82 %	187790	Trichothiodystrophy 7, nonphotosensitive, 618546 (3), Autosomal recessive
TAT	99.94 %	613018	Tyrosinemia, type II, 276600 (3), Autosomal recessive
TCHH	99.71 %	190370	?Uncombable hair syndrome 3, 617252 (3), Autosomal recessive
TERC	99.98 %	602322	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant
TERT	99.86 %	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant
TGM1	99.11 %	190195	Ichthyosis, congenital, autosomal recessive 1, 242300 (3), Autosomal recessive
TGM3	99.96 %	600238	?Uncombable hair syndrome 2, 617251 (3), Autosomal recessive
TGM5	99.86 %	603805	Peeling skin syndrome 2, 609796 (3), Autosomal recessive
TINF2	99.94 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TMC6	99.86 %	605828	{Epidermodysplasia verruciformis, susceptibility to, 1}, 226400 (3), Autosomal recessive
TMC8	99.72 %	605829	{Epidermodysplasia verruciformis, susceptibility to, 2}, 618231 (3), Autosomal recessive
TP63	99.8 %	603273	Premature ovarian failure 21, 620311 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
TREX1	100 %	606609	Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315 (3), Autosomal dominant; Aicardi-Goutieres syndrome 1, dominant and recessive, 225750 (3), Autosomal recessive, Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant; Chilblain lupus, 610448 (3), Autosomal dominant
TRPM1	99.09 %	603576	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216 (3), Autosomal recessive
TRPM4	99.8 %	606936	Progressive familial heart block, type IB, 604559 (3), Autosomal dominant; Erythrokeratodermia variabilis et progressiva 6, 618531 (3), Autosomal dominant
TRPS1	99.98 %	604386	Trichorhinophalangeal syndrome, type III, 190351 (3), Autosomal dominant; Trichorhinophalangeal syndrome, type I, 190350 (3), Autosomal dominant
TRPV3	99.9 %	607066	?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400 (3), Autosomal dominant; Olmsted syndrome 1, 614594 (3), Autosomal dominant
TSPEAR	99.99 %	612920	Tooth agenesis, selective, 10, 620173 (3), Autosomal recessive; ?Deafness, autosomal recessive 98, 614861 (3), Autosomal recessive; Ectodermal dysplasia 14, hypohidrotic/hair/tooth/nail type, 618180 (3), Autosomal recessive
TTC37	99.79 %	614589	Trichohepatoenteric syndrome 1, 222470 (3), Autosomal recessive
TUBB	99.77 %	191130	Symmetric circumferential skin creases, congenital, 1, 156610 (3), Autosomal dominant; Cortical dysplasia, complex, with other brain malformations 6, 615771 (3), Autosomal dominant
TWIST2	99.99 %	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	99.17 %	606448	?Glucocorticoid deficiency 5, 617825 (3), Autosomal recessive
TYRP1	99.57 %	115501	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (3); Albinism, oculocutaneous, type III, 203290 (3), Autosomal recessive
UBA2	99.78 %	613295	ACCES syndrome, 619959 (3), Autosomal dominant
UBR1	99.84 %	605981	Johanson-Blizzard syndrome, 243800 (3), Autosomal recessive
UROD	98.6 %	613521	Porphyria, hepatoerythropoietic, 176100 (3), Autosomal recessive, Autosomal dominant; Porphyria cutanea tarda, 176100 (3), Autosomal recessive, Autosomal dominant
UROS	99.98 %	606938	Porphyria, congenital erythropoietic, 263700 (3), Autosomal recessive
USB1	99.94 %	613276	Poikiloderma with neutropenia, 604173 (3), Autosomal recessive
UVSSA	99.92 %	614632	UV-sensitive syndrome 3, 614640 (3), Autosomal recessive
VCX	93.22 %	300229	*No OMIM phenotypes*
VCX3A	80.19 %	300533	*No OMIM phenotypes*
VPS33B	99.9 %	608552	Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009 (3), Autosomal recessive; Cholestasis, progressive familial intrahepatic, 12, 620010 (3), Autosomal recessive; Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 (3), Autosomal recessive
WDR19	99.62 %	608151	Nephronophthisis 13, 614377 (3), Autosomal recessive; Cranioectodermal dysplasia 4, 614378 (3), Autosomal recessive; Senior-Loken syndrome 8, 616307 (3), Autosomal recessive; Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 (3), Autosomal recessive; ?Spermatogenic failure 72, 619867 (3), Autosomal recessive
WDR35	99.87 %	613602	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 (3), Autosomal recessive; Cranioectodermal dysplasia 2, 613610 (3), Autosomal recessive
WNT10A	99.77 %	606268	Schopf-Schulz-Passarge syndrome, 224750 (3), Autosomal recessive; Tooth agenesis, selective, 4, 150400 (3), Autosomal recessive, Autosomal dominant; Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
WRAP53	99.62 %	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
XPA	99.78 %	611153	Xeroderma pigmentosum, group A, 278700 (3), Autosomal recessive
XPC	99.93 %	613208	Xeroderma pigmentosum, group C, 278720 (3), Autosomal recessive
ZMPSTE24	97.94 %	606480	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3), Autosomal recessive; Restrictive dermopathy 1, 275210 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2025-11-12**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.