Dermatology

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Single Gene

Code	Diseases	Gene
sCe5/0007	Acne inversa familial type 3	PSEN1
sCe5/0008	Acrodermatitis enteropathica	SLC39A4
sCe5/0015	Albinism, oculocutaneous nonsyndromic	SLC24A5
sCe5/0016	Albinism, oculocutaneous type 1A	TYR
sCe5/0017	Albinism, oculocutaneous type 1B	TYR
sCe5/0018	Albinism, oculocutaneous type 2	OCA2
sCe5/0019	Albinism, oculocutaneous type 3	TYRP1
sCe5/0020	Albinism, oculocutaneous type 4	SLC45A2
sCe5/0021	Albinism, oculocutaneous type 5	C10ORF11
sCe5/0022	Alopecia universalis	HR
sCe5/0038	Amyloidosis, primary localized cutaneous, type 1	OSMR
sCe5/0039	Amyloidosis, primary localized cutaneous, type 2	IL31RA
sCe5/0056	Atrichia with papular lesions	HR
sCe5/0076	Buschke-Ollendorff syndrome	LEMD3
sCe5/0077	C1q deficiency	C1QA
sCe5/0090	Cole disease	ENPP1
sCe5/0102	Cutaneous telangiectasia and cancer syndrome, familial	ATR
sCe5/0103	Cutis laxa type 1A, autosomal recessive	FBLN5
sCe5/0104	Cutis laxa type 1B, autosomal recessive	EFEMP2
sCe5/0105	Cutis laxa type 1C, autosomal recessive	LTBP4
sCe5/0106	Cutis laxa type 2, autosomal dominant	FBLN5
sCe5/0107	Cutis laxa type 2A, autosomal recessive	ATP6V0A2
sCe5/0108	Cutis laxa type 2B, autosomal recessive	PYCR1
sCe5/0109	Cutis laxa type 3A, autosomal recessive	ALDH18A1
sCe5/0110	Cutis laxa type 3B, autosomal recessive	PYCR1

sCe5/0111	Cutis laxa, autosomal dominant	ELN
sCe5/0116	Dermatitis, atopic type 2	FLG
sCe5/0117	Dermatopathia pigmentosa reticularis	KRT14
sCe5/0121	Dyschromatosis universalis hereditaria type 3	ABCB6
sCe5/0122	Dyskeratosis congenita, autosomal dominant type 1	TERC
sCe5/0123	Dyskeratosis congenita, autosomal recessive type 1	NOP10
sCe5/0124	Dyskeratosis congenita, autosomal recessive type 2	NHP2
sCe5/0125	Dyskeratosis congenita, autosomal recessive type 4/ autosomal dominant type 2	TERT
sCe5/0126	Dyskeratosis congenita, autosomal recessive type 5	RTEL1
sCe5/0127	Dyskeratosis congenita, autosomal recessive type 6	PARN
sCe5/0128	Dyskeratosis congenita, autosomal recessive type 7	ACD
sCe5/0129	Dyskeratosis congenita, X-linked	DKC1
sCe5/0131	Ectodermal dysplasia type 4, hair/nail type	KRT85
sCe5/0132	Ectodermal dysplasia, ectrodactyly, and macular dystrophy	CDH3
sCe5/0133	Ectodermal dysplasia, hidrotic	GJB6
sCe5/0134	Ectodermal dysplasia, hypohidrotic, autosomal recessive	EDAR
sCe5/0135	Ectodermal dysplasia, hypohidrotic, autosomal recessive	EDARADD
sCe5/0136	Ectodermal dysplasia, hypohidrotic, with immune deficiency	IKBKG
sCe5/0137	Ectodermal dysplasia, hypohidrotic, X-linked	EDA
sCe5/0138	Ectodermal dysplasia/skin fragility syndrome	PKP1
sCe5/0139	Ehlers-Danlos syndrome type 1/2	COL5A1
sCe5/0140	Ehlers-Danlos syndrome type 1/2	COL5A2
sCe5/0141	Ehlers-Danlos syndrome type 3	COL3A1
sCe5/0142	Ehlers-Danlos syndrome type 3	TNXB
sCe5/0143	Ehlers-Danlos syndrome type 4	COL5A1
sCe5/0144	Ehlers-Danlos syndrome type 4	COL3A1
sCe5/0145	Ehlers-Danlos syndrome type 6	PLOD1
sCe5/0146	Ehlers-Danlos syndrome type 7A	COL1A1
sCe5/0147	Ehlers-Danlos syndrome type 7B	COL1A2

sCe5/0148	Ehlers-Danlos syndrome type 7C	ADAMTS2
sCe5/0149	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss	FKBP14
sCe5/0150	Ehlers-Danlos syndrome, musculocontractural type 1	CHST14
sCe5/0151	Ehlers-Danlos syndrome, musculocontractural type 2	DSE
sCe5/0152	Ehlers-Danlos syndrome, progeroid type 1	B4GALT7
sCe5/0153	Ehlers-Danlos syndrome, progeroid type, type 2	B3GALT6
sCe5/0155	Epidermolysis bullosa dystrophica	COL7A1
sCe5/0156	Epidermolysis bullosa dystrophica, autosomal recessive, modifier of	MMP1
sCe5/0157	Epidermolysis bullosa junctionalis with pyloric atresia	ITGA6
sCe5/0158	Epidermolysis bullosa junctionalis with pyloric atresia	ITGB4
sCe5/0159	Epidermolysis bullosa simplex	KRT5
sCe5/0160	Epidermolysis bullosa simplex with pyloric atresia	PLEC
sCe5/0161	Epidermolysis bullosa simplex, autosomal recessive type 1	KRT14
sCe5/0162	Epidermolysis bullosa simplex, autosomal recessive type 2	DST
sCe5/0163	Epidermolysis bullosa simplex, Dowling-Meara type	KRT14
sCe5/0164	Epidermolysis bullosa simplex, Koebner type	KRT14
sCe5/0165	Epidermolysis bullosa simplex, Ogna type	PLEC
sCe5/0166	Epidermolysis bullosa simplex, Weber-Cockayne type	KRT14
sCe5/0167	Epidermolysis bullosa, generalized atrophic benign	LAMA3
sCe5/0168	Epidermolysis bullosa, junctional	LAMC2
sCe5/0169	Epidermolysis bullosa, junctional	COL17A1
sCe5/0170	Epidermolysis bullosa, junctional, Herlitz type	LAMA3
sCe5/0171	Epidermolysis bullosa, junctional, Herlitz type	LAMB3
sCe5/0172	Epidermolysis bullosa, junctional, non-Herlitz type	LAMB3
sCe5/0173	Epidermolysis bullosa, lethal acantholytic	DSP
sCe5/0174	Epidermolysis bullosa, nonspecific, autosomal recessive	EXPH5
sCe5/0175	Epidermolytic hyperkeratosis	KRT1
sCe5/0176	Epidermolytic hyperkeratosis	KRT10
sCe5/0177	Epidermolytic palmoplantar keratoderma	KRT9

sCe5/0182	Erythrokeratodermia variabilis et progressive	GJB3
sCe5/0183	Erythrokeratodermia variabilis et progressive	GJB4
sCe5/0194	Focal dermal hypoplasia	PORCN
sCe5/0197	Geroderma osteodysplasticum	GORAB
sCe5/0207	Griscelli syndrome type 1	MYO5A
sCe5/0208	Griscelli syndrome type 3	MLPH
sCe5/0220	Hyaline fibromatosis syndrome	ANTXR2
sCe5/0232	Hypotrichosis type 11	SNRPE
sCe5/0233	Hypotrichosis type 12	RPL21
sCe5/0234	Hypotrichosis type 13	KRT71
sCe5/0235	Hypotrichosis type 2	CDSN
sCe5/0236	Hypotrichosis type 3	KRT74
sCe5/0237	Hypotrichosis type 4	HR
sCe5/0238	Hypotrichosis type 6	DSG4
sCe5/0239	Hypotrichosis type 7	LIPH
sCe5/0240	Hypotrichosis type 8	LPAR6
sCe5/0241	Hypotrichosis-lymphedema-telangiectasia syndrome	SOX18
sCe5/0242	Ichthyosiform erythroderma, congenital, nonbullous type 1	ALOXE3
sCe5/0243	Ichthyosiform erythroderma, congenital, nonbullous type 1	NIPAL4
sCe5/0244	Ichthyosis congenital, autosomal recessive, PNPLA1 related	PNPLA1
sCe5/0245	Ichthyosis congenital, Harlequin fetus type	ABCA12
sCe5/0246	Ichthyosis follicularis, atricia, and photophobia syndrome	MBTPS2
sCe5/0247	Ichthyosis prematurity syndrome	SLC27A4
sCe5/0248	Ichthyosis vulgaris	FLG
sCe5/0249	Ichthyosis, bullous type	KRT2
sCe5/0250	Ichthyosis, congenital, autosomal recessive type 1	TGM1
sCe5/0251	Ichthyosis, congenital, autosomal recessive, type 11	ST14
sCe5/0252	Ichthyosis, congenital, autosomal recessive, type 2	ALOX12B
sCe5/0253	Ichthyosis, congenital, autosomal recessive, type 9	CERS3

sCe5/0254	Ichthyosis, lamellar type 2	ABCA12
sCe5/0255	Ichthyosis, lamellar type 3	CYP4F22
sCe5/0256	Ichthyosis, lamellar type 4	LIPN
sCe5/0257	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis	CLDN1
sCe5/0258	Ichthyosis, spastic quadriplegia, and mental retardation	ELOVL4
sCe5/0259	Ichthyosis, X-linked	STS
sCe5/0300	Incontinentia pigmenti type 2	IKBKG
sCe5/0309	Keratoderma, palmoplantar, punctate type 1A	AAGAB
sCe5/0310	Keratosis follicularis spinulosa declavans, X-linked	MBTPS2
sCe5/0311	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma	POMP
sCe5/0312	Keratosis palmoplantaris striata type 1	DSG1
sCe5/0313	Keratosis palmoplantaris striata type 2	DSP
sCe5/0314	Kindler syndrome	FERMT1
sCe5/0315	Kindler syndrome	FBLIM1
sCe5/0316	Laryngoonychocutaneous syndrome	LAMA3
sCe5/0318	Legius syndrome	SPRED1
sCe5/0331	Mal de Meleda	SLURP1
sCe5/0332	Mandibuloacral dysplasia with type B lipodystrophy	ZMPSTE24
sCe5/0337	MEDNIK syndrome	AP1S1
sCe5/0342	Muckle-wells syndrome	NLRP3
sCe5/0348	Naegeli-Franceschetti-Jadassohn syndrome	KRT14
sCe5/0351	Netherton syndrome	SPINK5
sCe5/0352	Neurofibromatosis type 1	NF1
sCe5/0353	Neurofibromatosis type 1 -like syndrome	SPRED1
sCe5/0354	Neurofibromatosis type 2	NF2
sCe5/0362	Olmsted syndrome	TRPV3
sCe5/0404	Palmoplantar keratoderma, nonepidermolytic, focal	KRT16
sCe5/0405	Papillon-Lefevre syndrome	CTSC
sCe5/0406	Peeling skin syndrome type 1	CDSN

sCe5/0407	Peeling skin syndrome type 2	TGM5
sCe5/0408	Peeling skin syndrome type 3	CHST8
sCe5/0409	Peeling skin syndrome type 4	CSTA
sCe5/0410	Piebaldism	KIT
sCe5/0411	Piebaldism	SNAI2
sCe5/0412	Pityriasis rubra pilaris	CARD14
sCe5/0414	Porokeratosis type 3, disseminated superficial actinic	MVK
sCe5/0415	Porphyria cutanea tarda	UROD
sCe5/0417	Pseudoxanthoma elasticum	ABCC6
sCe5/0418	Pseudoxanthoma elasticum, forme fruste	ABCC6
sCe5/0419	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency	GGCX
sCe5/0420	Psoriasis susceptibility type 11	IL12B
sCe5/0421	Psoriasis type 2	CARD14
sCe5/0422	Psoriasis, generalized pustular	IL36RN
sCe5/0423	Pterygium syndrome	CHRNG
sCe5/0431	Restrictive dermopathy, lethal	LMNA
sCe5/0432	Restrictive dermopathy, lethal	ZMPSTE24
sCe5/0439	Sarcoidosis, early-onset	NOD2
sCe5/0448	Skin fragility-woolly hair syndrome	DSP
sCe5/0449	Skin hair eye pigmentation type 6	SLC24A4
sCe5/0452	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like	SLC39A13
sCe5/0461	Steatocystoma multiplex	KRT17
sCe5/0462	Stiff skin syndrome	FBN1
sCe5/0467	Telangiectasia hereditary hemorrhagic type 5	GDF2
sCe5/0468	Telangiectasia, hereditary hemorrhagic, of Rendu, Osler and Weber type 1	ENG
sCe5/0469	Telangiectasia, hereditary hemorrhagic, type 2	ACVRL1
sCe5/0470	Terminal osseous dysplasis	FLNA
sCe5/0471	Trichodontoosseous syndrome	DLX3
sCe5/0472	Trichohepatoenteric syndrome type 1	TTC37

sCe5/0473	Trichohepatoenteric syndrome type 2	SKIV2L
sCe5/0474	Trichorhinophalangeal syndrome type 1	TRPS1
sCe5/0475	Trichothiodystrophy	ERCC3
sCe5/0476	Trichothiodystrophy	ERCC2
sCe5/0477	Trichothiodystrophy	GTF2H5
sCe5/0478	Trichothiodystrophy, nonphotosensitive type 1	MPLKIP
sCe5/0480	Tylosis with esophageal cancer	RHBDF2
sCe5/0481	UV-sensitive syndrome type 1	ERCC6
sCe5/0482	UV-sensitive syndrome type 3	UVSSA
sCe5/0484	Vasculopathy, infantile-onset, TMEM173/STING related	TMEM173
sCe5/0486	Vohwinkel syndrome with ichthyosis	LOR
sCe5/0487	Waardenburg syndrome type 1	PAX3
sCe5/0488	Waardenburg syndrome type 2E	SOX10
sCe5/0489	Waardenburg syndrome type 4C	SOX10
sCe5/0493	Wrinkly skin syndrome	ATP6V0A2
sCe5/0494	Xeroderma pigmentosum, group A	XPA
sCe5/0495	Xeroderma pigmentosum, group C	XPC
sCe5/0496	Xeroderma pigmentosum, group D	ERCC2
sCe5/0497	Xeroderma pigmentosum, group E, DDB-negative subtype	DDB2
sCe5/0498	Xeroderma pigmentosum, group F	ERCC4
sCe5/0499	Xeroderma pigmentosum, group G	ERCC5
sCe5/0500	Xeroderma pigmentosum, variant type	POLH
sCe5/0501	XFE progeroid syndrome	ERCC4

Panel

Code	product name	TAT-day
	Clinical Exome Whole exome Solo	
pCe5/0007	Congenital ichthyosis panel	25
pCe5/0009	Cutis laxa panel	25
pCe5/0010	Ehlers-Danlos syndrome and related disorders panel	25
pCe5/0011	Epidermolysis bullosa panel	25
pCe5/0014	Ichthyosis extended panel	25
pCe5/0015	Melanoma panel	25
pCe5/0016	Neurofibromatosis panel	25
pCe5/0017	Nonsyndromic hypotrichosis panel	25
pCe5/0023	Waardenburg syndrome panel	25