Heredscreen Carrier Screening Service - Diseases List
基康檢隱性疾病基因檢測服務- 疾病列表

cordlife 康盛人生基因檢測 Heredscreen 基 康 檢

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No.	Related Diseases	相關疾病	Gene 基因
1	Gamma-aminobutyric acid transaminase deficiency	γ-氨基丁酸轉氨酶缺乏症	ABAT
2	Ichthyosis, congenital, autosomal recessive 4A	魚鱗病，先天性，體染色體隱性遺傳 4A	ABCA12
3	Cone-rod dystrophy 3	錐桿營養不良3	ABCA4
4	Cholestasis, progressive familial intrahepatic 2	膽汁淤積，進行性家族性肝內 2	ABCB11
5	Gallbladder disease 1	膽囊疾病1	ABCB4
6	Pseudoxanthoma elasticum	彈性假黃瘤	ABCC6
7	Diabetes mellitus, permanent neonatal 3, with or without neurologic features	糖尿病，永久性新生兒 3，有或沒有神經系統特徵	ABCC8
8	ACADM DEFICIENCY	學術缺陷	ACADM
9	ACADS DEFICIENCY	ACADS缺陷	ACADS
10	VLCAD deficiency	VLCAD 缺陷	ACADVL
11	Peroxisomal acyl-CoA oxidase deficiency	過氧化物酶體醯基輔酶A氧化酶缺乏症	ACOX1
12	Severe combined immunodeficiency due to ADA deficiency	ADA 缺乏導致的嚴重聯合免疫缺陷	ADA
13	Ehlers-Danlos syndrome, dermatosparaxis type	埃勒斯-當洛斯綜合徵，皮膚錯亂型	ADAMTS2
14	Usher syndrome, type 2C	亞瑟綜合症，2C 型	ADGRV1
15	Aspartylglucosaminuria	天門冬胺酸葡萄糖尿	AGA
16	Glycogen storage disease IIIa/b	肝醣累積病 IIIa/b	AGL
17	Primary hyperoxaluria type 1	原發性高草酸尿症第 1 型	AGXT
18	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase	S-腺苷同型半胱氨酸水解酶缺乏的高蛋氨酸血症	AHCY
19	Joubert syndrome 3	朱伯特症候群3	AHI1
20	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia	自體免疫性多內分泌病綜合徵，I型，伴或不伴可逆性幹骺端發育不良	AIRE
21	Sjogren-Larsson syndrome	乾燥-拉爾森症候群	ALDH3A2
22	Epilepsy, early-onset, 4, vitamin B6-dependent	癲癇，早發，4，維生素 B6 依賴性	ALDH7A1
23	Fructose intolerance, hereditary	果糖不耐受，遺傳性	ALDOB
24	Alstrom syndrome	阿爾斯特羅姆氏症候群	ALMS1
25	Ichthyosis, congenital, autosomal recessive 2	魚鱗病，先天性，體染色體隱性遺傳 2	ALOX12B
26	Ichthyosis, congenital, autosomal recessive 3	魚鱗病，先天性，體染色體隱性遺傳 3	ALOXE3
27	Hypophosphatasia	低磷酸酯酶症	ALPL
28	Persistent Mullerian duct syndrome, type I	持續性苗勒氏管綜合徵，I 型	AMHR2
29	Autosomal recessive spinocerebellar ataxia 10	體染色體隱性遺傳脊髓小腦共濟失調 10	ANO10
30	Hyaline fibromatosis syndrome	透明纖維瘤病症候群	ANTXR2
31	Hermansky-Pudlak syndrome 2	赫曼斯基-普德拉克氏症候群 2	AP3B1
32	Diabetes insipidus, nephrogenic, 2	腎性尿崩症，2	AQP2
33	Canavan disease	卡納萬病	ARSA
34	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	VI 型黏多醣貯積症（Maroteaux-Lamy）	ARSB
35	Farber lipogranulomatosis	法伯脂肪肉芽腫病	ASAH1
36	Canavan disease	卡納萬病	ASPA
37	Citrullinemia type I	I型瓜胺酸血症	ASS1
38	Ataxia-telangiectasia	共濟失調毛細血管擴張	ATM
39	Kufor-Rakeb syndrome/ Spastic paraplegia 78, autosomal recessive	Kufor-Rakeb 症候群/痙攣性截癱 78，體染色體隱性遺傳	ATP13A2
40	Renal tubular acidosis, distal, 3, with or without sensorineural hearing loss	力損失	ATP6V0A4
41	Renal tubular acidosis with progressive nerve deafness	腎小管性酸中毒伴隨進行性神經性耳聾	ATP6V1B1
42	Wilson disease	威爾森氏症	ATP7B
43	Cholestasis, progressive familial intrahepatic 1	膽汁淤積，進行性家族性肝內 1	ATP8B1
44	Meckel-Gruber syndrome	梅克爾-格魯伯氏症候群	B9D2
45	Bardet-Biedl syndrome 1	Bardet-Biedl 症候群 1	BBS1
46	Bardet-Biedl syndrome 10	Bardet-Biedl 症候群 10	BBS10
47	Bardet-Biedl syndrome 12	Bardet-Biedl 症候群 12	BBS12
48	Retinitis pigmentosa 74	色素性視網膜炎 74	BBS2
49	Bardet-Biedl syndrome 4	Bardet-Biedl 症候群 4	BBS4
50	Bardet-Biedl syndrome 9	Bardet-Biedl 症候群 9	BBS9
51	Deficiency of butyrylcholinesterase	丁醯膽鹼酯酶缺乏	BCHE
52	Maple syrup urine disease, type Ia	楓糖尿病，Ia 型	BCKDHA
53	Maple syrup urine disease, type Ib	楓糖尿病，Ib 型	BCKDHB
54	GRACILE syndrome	格雷西症候群	BCS1L
55	Bloom syndrome	布魯姆症候群	BLM
56	Osteogenesis imperfecta, type XIII	成骨不全，XIII 型	BMP1
57	Fanconi anemia, complementation group J	範可尼貧血，補充組 J	BRIP1
58	Bartter syndrome	巴特綜合症	BSND
59	Biotinidase deficiency	生物素酶缺乏症	BTD
60	Desbuquois dysplasia 1	Desbuquois發育不良1	CANT1
61	Muscular dystrophy, limb-girdle, autosomal recessive 1	肌肉營養不良症，肢帶，體染色體隱性遺傳 1	CAPN3
62	Ventricular tachycardia, catecholaminergic polymorphic, 2 & 3	室性心搏過速，兒茶酚胺能多態性，2 和 3	CASQ2
63	Lipodystrophy, congenital generalized, type 4	先天性全身性脂肪營養不良，4 型	CAVIN1
64	Homocystinuria, B6-responsive and nonresponsive types Thrombosis, hyperhor	血栓形成、高同型半胱氨酸血症	CBS
65	Intellectual disability, autosomal recessive 3	智力障礙，體染色體隱性遺傳 3	CC2D1A
66	Progressive pseudorheumatoid dysplasia	進行性假類風濕性發育不良	CCN6
67	Immunodeficiency 25	免疫缺陷25	CD247
68	Dyserythropoietic anemia, congenital, type Ia	先天性 Ia 型紅血球生成不良性貧血	CDAN1
69	Deafness, autosomal recessive 12	耳聾，體染色體隱性遺傳 12	CDH23
70	syndrome 25	25	CEP104
71	Joubert syndrome 31	朱伯特症候群 31	CEP120
72	Microcephaly 9, primary, autosomal recessive / Seckel syndrome 5	5	CEP152
73	Congenital bilateral absence of vas deferens	先天性雙側輸精管缺如	CFTR

74	Myasthenic syndrome, congenital, 6, presynaptic	先天性肌無力症，6，突觸前	CHAT
75	Choroideremia	無脈絡膜血症	CHM
76	Deafness, autosomal recessive 48	耳聾，體染色體隱性遺傳 48	CIB2
77	Bare lymphocyte syndrome, type II, complementation group A	裸淋巴細胞綜合徵，II 型，互補 A 組	CIITA
78	Hypophosphatemic rickets	低血磷性佝僂病	CLCN5
79	Ceroid lipofuscinosis, neuronal, 3	蠟質脂褐質沉著症，神經元，3	CLN3
80	Ceroid lipofuscinosis, neuronal, 5	蠟質脂褐質沉著症，神經元，5	CLN5
81	Ceroid lipofuscinosis, neuronal, 8	蠟質脂褐質沉著症，神經元，8	CLN8
82	Retinitis pigmentosa 61	色素性視網膜炎 61	CLRN1
83	Achromatopsia 2	全色盲2	CNGA3
84	Achromatopsia 3	全色盲3	CNGB3
85	Pitt-Hopkins like syndrome 1	皮特霍普金斯樣綜合症1	CNTNAP2
86	Mitochondrial complex IV deficiency, nuclear type 17	粒線體複合體 IV 缺陷，核型 17	COA8
87	Deafness, autosomal recessive 53	耳聾，體染色體隱性遺傳 53	COL11A2
88	Steel syndrome	鋼鐵綜合症	COL27A1
89	Alport syndrome 3B, autosomal recessive	Alport 症候群 3B，體染色體隱性遺傳	COL4A3
90	Alport syndrome 2, autosomal recessive	Alport 症候群 2，體染色體隱性遺傳	COL4A4
91	Epidermolysis bullosa dystrophica, autosomal recessive	營養不良性大皰性表皮鬆解症，體染色體隱性遺傳	COL7A1
92	Spastic ataxia 10, autosomal recessive	痙攣性共濟失調 10，體染色體隱性遺傳	COQ4
93	Mitochondrial complex IV deficiency, nuclear type 3	粒線體複合體 IV 缺陷，核型 3	COX10
94	Mitochondrial complex IV deficiency, nuclear type 7	粒線體複合體 IV 缺陷，核型 7	COX6B1
95	Joubert syndrome 17	朱伯特症候群 17	CPLANE1
96	Carbamoylphosphate synthetase I deficiency	氨基甲醯磷酸合成酶 I 缺乏症	CPS1
97	Carnitine palmitoyltransferase I deficiency	肉鹼棕櫚醯轉移酶 I 缺乏症	CPT1A
98	Carnitine palmitoyl transferase II deficiency, infantile	肉鹼棕櫚醯轉移酶 II 缺乏症，嬰兒	CPT2
99	Leber congenital amaurosis 8	萊伯先天性黑蒙 8	CRB1
100	Osteogenesis imperfecta, type VII	成骨不全，VII 型	CRTAP
101	Cystinosis, nephropathic	胱胺酸沉著症，腎臟病	CTNS
102	Galactosialidosis	半乳糖唾液酸貯積症	CTSA
103	Haim-Munk syndrome / Papillon-Lefevre syndrome	Haim-Munk 綜合症 / Papillon-Lefevre 綜合徵	CTSC
104	Ceroid lipofuscinosis, neuronal, 10	蠟質脂褐質沉著症，神經元，10	CTSD
105	Ceroid lipofuscinosis, neuronal, 13, Kufs type	蠟質脂褐質沉著症，神經元，13，Kufs 型	CTSF
106	Pyknodysostosis	緻密性骨不全	CTSK
107	Chronic granulomatous disease 4	慢性肉芽腫病4	CYBA
108	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	先天性腎上腺增生，由於 11-β-羥化酶缺乏	CYP11B1
109	Hypoaldosteronism, congenital	先天性醛固酮減少症	CYP11B2
110	17-alpha-hydroxylase/17,20-lyase deficiency	17-α-羥化酶/17,20-裂解酶缺乏症	CYP17A1
111	Aromatase deficiency	芳香酶缺乏症	CYP19A1
112	Anterior segment dysgenesis 6, multiple subtypes	眼前節發育不全6，多種亞型	CYP1B1
113	Spastic paraplegia 5A, autosomal recessive	痙攣性截癱 5A，體染色體隱性遺傳	CYP7B1
114	Maple syrup urine disease, type II	楓糖尿病，II 型	DBT
115	Woodhouse-Sakati syndrome	伍德豪斯-薩卡蒂氏症候群	DCAF17
116	Omenn syndrome	預兆症候群	DCLRE1C
117	Xeroderma pigmentosum, group E, DDB-negative subtype	色素性乾皮症，E 組，DDB 陰性亞型	DDB2
118	Aromatic L-amino acid decarboxylase deficiency	芳香族L-胺基酸脫羧酶缺乏症	DDC
119	Portal hypertension, noncirrhotic	門脈高壓，非肝硬化	DGUOK
120	Smith-Lemli-Opitz syndrome	史密斯-萊姆利-奧皮茨綜合徵	DHCR7
121	Dyskeratosis congenita, X-linked recessive	先天性角化不良，X連鎖隱性遺傳	DKC1
122	Pyruvate dehydrogenase E2 deficiency	丙酮酸脫氫酶 E2 缺乏症	DLAT
123	Dihydrolipoamide dehydrogenase deficiency	二氫硫辛醯胺脫氫酶缺乏症	DLD
124	Spondylocostal dysostosis 1, autosomal recessive	脊椎肋骨發育不良 1，體染色體隱性遺傳	DLL3
125	Becker muscular dystrophy	貝克爾肌肉營養不良症	DMD
126	Ciliary dyskinesia, primary, 3, with or without situs inversus	原發性纖毛運動障礙，3 級，伴或不伴反位	DNAH5
127	Ciliary dyskinesia, primary, 9, with or without situs inversus	原發性纖毛運動障礙，9，伴隨或不伴隨反位	DNAI2
128	Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections	高 IgE 症候群 2，體染色體隱性遺傳，伴隨反覆感染	DOCK8
129	Fetal akinesia deformation sequence 3 / Congenital myasthenic syndrome 10	胎兒運動不能變形序列3/先天性肌無力症候群10	DOK7
130	Congenital disorder of glycosylation, type Im	先天性糖基化障礙，Im 型	DOLK
131	Dihydropyrimidine dehydrogenase deficiency	二氫嘧啶脫氫酶缺乏症	DPYD
132	Hermansky-Pudlak syndrome 7	赫曼斯基-普德拉克氏症候群 7	DTNBP1
133	Short-rib thoracic dysplasia 3 with or without polydactyly	短肋胸廓發育不良 3 伴或不伴多指畸形	DYNC2H1
134	Muscular dystrophy, limb-girdle, autosomal recessive 2	肌肉營養不良症，肢帶，體染色體隱性遺傳 2	DYSF
135	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive	10B，少汗/毛髮/牙齒型，體染色體隱性遺傳	EDAR
136	Cutis laxa, autosomal recessive, type IB	皮膚鬆弛，體染色體隱性遺傳，IB 型	EFEMP2
137	Wolcott-Rallison syndrome	沃爾科特-拉里森症候群	EIF2AK3
138	Leukoencephalopathy with vanishing white matter 4, with or without ovarian failure	白質消失的白質腦病變 4，伴隨或不伴隨卵巢衰竭	EIF2B4
139	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure	白質消失的白質腦病變 5，伴隨或不伴隨卵巢衰竭	EIF2B5
140	Dysautonomia, familial	自主神經功能障礙，家族性	ELP1
141	Emery-Dreifuss muscular dystrophy 1, X-linked	X 連鎖 Emery-Dreifuss 肌肉失養症 1	EMD
142	Visceral neuropathy, familial, 1, autosomal recessive	內臟神經病變，家族性，1，體染色體隱性遺傳	ERBB3
143	Trichothiodystrophy 1, photosensitive	毛髮硫營養不良1型，光敏感性	ERCC2
144	Trichothiodystrophy 2, photosensitive	毛髮硫營養不良2型，光敏感性	ERCC3
145	Fanconi anemia, complementation group Q	範可尼貧血，補充Q組	ERCC4
146	Cerebrooculofacioskeletal syndrome 3	腦面骨骼症候群3	ERCC5
147	Cerebrooculofacioskeletal syndrome 1	腦面骨骼症候群1	ERCC6
148	Ellis-van Creveld syndrome	艾利斯-範克雷維爾德症候群	EVC
149	Ellis-van Creveld syndrome	艾利斯-範克雷維爾德症候群	EVC2
150	Retinitis pigmentosa 25	色素性視網膜炎 25	EYS
151	Factor XI deficiency	第 XI 因子缺乏症	F11

152	Hypoprothrombinemia	低凝血酶原血症	F2
153	Factor V deficiency	V因子缺乏症	F5
154	Factor VII deficiency	第七因子缺乏症	F7
155	Hemophilia B	乙型血友病	F9
156	Tyrosinemia type I	酪胺酸血症I型	FAH
157	Retinitis pigmentosa 28	色素性視網膜炎 28	FAM161A
158	Fanconi anemia, complementation group A	範可尼貧血，補充 A 組	FANCA
159	Fanconi anemia, complementation group C	範可尼貧血，補充 C 組	FANCC
160	Fanconi anemia, complementation group D2	範可尼貧血，補充組 D2	FANCD2
161	Fanconi anemia, complementation group E	範可尼貧血，補充 E 組	FANCE
162	Fanconi anemia, complementation group G	範可尼貧血，補充 G 組	FANCG
163	Fanconi anemia, complementation group I	範可尼貧血，補充 I 組	FANCI
164	Fanconi anemia, complementation group L	範可尼貧血，補充組 L	FANCL
165	Fumarase deficiency	延胡索酶缺乏症	FH
166	Emery-Dreifuss muscular dystrophy 6, X-linked	Emery-Dreifuss 肌肉失養症 6，X 連鎖	FHL1
167	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalie	肌肉營養不良症（先天性腦部和眼部異常），A 型，5	FKRP
168	Cardiomyopathy, dilated, autosomal recessive	擴張型心肌病變，體染色體隱性遺傳	FKTN
169	Fragile X syndrome	脆性X綜合症	FMR1
170	T-cell immunodeficiency, congenital alopecia, and nail dystrophy	T細胞免疫缺陷、先天性掉髮及指甲營養不良	FOXN1
171	Fraser syndrome 1	弗雷澤症候群1	FRAS1
172	Fraser syndrome 2	弗雷澤症候群2	FREM2
173	Friedreich ataxia	弗里德賴希共濟失調	FXN
174	Glycogen storage disease type Ia	Ia 型肝醣累積病	G6PC
175	Glucose-6-phosphate dehydrogenase deficiency	6-磷酸葡萄糖去氫酶缺乏症	G6PD
176	Glycogen storage disease II	肝醣累積病II	GAA
177	Krabbe disease	克拉伯病	GALC
178	Galactokinase deficiency with cataracts	半乳糖激酶缺乏症導致白內障	GALK1
179	Galactosemia	半乳糖血症	GALT
180	Gaucher disease, perinatal lethal	戈謝氏症，圍產期致命	GBA
181	Glycogen storage disease IV	肝醣貯積症IV	GBE1
182	Glutaric acidemia type I	I型戊二酸血症	GCDH
183	GTP Cyclohydrolase I Deficiency	GTP 環化水解酶 I 缺乏症	GCH1
184	Acromesomelic dysplasia 2A	頂體發育不良2A	GDF5
185	Laron dwarfism	拉倫侏儒症	GHR
186	Growth hormone deficiency, isolated, type IV	生長激素缺乏症，孤立性，IV 型	GHRHR
187	Deafness, autosomal recessive 1A	耳聾，體染色體隱性遺傳 1A	GJB2
188	Erythrokeratodermia variabilis et progressiva 1	變異性及進展性紅斑性角皮症 1	GJB3
189	Deafness, autosomal recessive 1B	耳聾，體染色體隱性遺傳 1B	GJB6
190	Fabry disease	法布瑞氏症	GLA
191	GM1-gangliosidosis, type I	GM1-神經節苷脂沉積症，I 型	GLB1
192	Glycine encephalopathy 1	甘氨酸腦病變1	GLDC
193	Lethal congenital contracture syndrome 1	致命性先天性攣縮綜合症1	GLE1
194	Nonaka myopathy	野中肌病變	GNE
195	Rhizomelic chondrodysplasia punctata, type 2	點狀根莖軟骨發育不良，2 型	GNPAT
196	Bernard-Soulier syndrome, type A1 (recessive)	Bernard-Soulier 綜合徵，A1 型（隱性）	GP1BA
197	Primary hyperoxaluria, type II	原發性高草酸尿症，II 型	GRHPR
198	Fraser syndrome 3	弗雷澤症候群3	GRIP1
199	Mucopolysaccharidosis VII	黏多醣貯積症VII	GUSB
200	Glycogen storage disease 0, liver	肝醣累積病 0，肝臟	GYS2
201	3-hydroxyacyl-CoA dehydrogenase deficiency	3-羥醯輔酶A脫氫酶缺乏症	HADH
202	LCHAD deficiency	LCHAD缺乏症	HADHA
203	Thalassemias, alpha-	地中海型貧血，α-	HBA1
204	Thalassemias, alpha-	地中海型貧血，α-	HBA2
205	Sickle cell anemia	鐮狀細胞性貧血	HBB
206	Tay-Sachs disease	泰-薩克斯病	HEXA
207	Sandhoff disease	桑德霍夫病	HEXB
208	Hemochromatosis, type 1	血色素沉著症，1 型	HFE
209	Alkaptonuria	黑酸尿症	HGD
210	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	3-羥基-3-甲基戊二醯輔酶A裂解酶缺陷	HMGCL
211	HMG-CoA synthase-2 deficiency	HMG-CoA合成酶2缺乏症	HMGCS2
212	Primary hyperoxaluria type III	原發性高草酸尿症III型	HOGA1
213	Hermansky-Pudlak syndrome 1	赫曼斯基-普德拉克氏症候群 1	HPS1
214	Hermansky-Pudlak syndrome 4	赫曼斯基-普德拉克氏症候群 4	HPS4
215	Pseudohermaphroditism, male, with gynecomastia	假兩性畸形，男性，伴隨男性女乳症	HSD17B3
216	D-bifunctional protein deficiency	D-雙功能蛋白缺乏症	HSD17B4
217	Mucopolysaccharidosis Ih/s	黏多醣沉積症 Ih/s	IDUA
218	Retinitis pigmentosa 80	色素性視網膜炎80	IFT140
219	Charcot-Marie-Tooth disease, axonal, type 2S	腓骨肌萎縮症，軸突，2S 型	IGHMBP2
220	Intellectual developmental disorder, X-linked 21	X 連鎖智能發展障礙 21	IL1RAPL1
221	Immunodeficiency 41 with lymphoproliferation and autoimmunity	免疫缺陷 41 伴隨淋巴球增生和自體免疫	IL2RA
222	Immunodeficiency 104, severe combined	免疫缺陷104，嚴重聯合	IL7R
223	Impaired intellectual development, truncal obesity, retinal dystrophy, and micropenis syndrome	智力發育受損、軀幹肥胖、視網膜營養不良與小陰莖綜合症	INPP5E
224	Glanzmann thrombasthenia 1	格蘭茨曼血小板無力症1	ITGA2B
225	Glanzmann thrombasthenia 2	格蘭茲曼血小板無力症2	ITGB3
226	Developmental and epileptic encephalopathy 35	發育性和癲癇性腦病變 35	ITPA
227	Isovaleryl-CoA dehydrogenase deficiency	異戊醯輔酶A脫氫酶缺乏症	IVD
228	Thyroid dyshormonogenesis 4	甲狀腺激素生成異常 4	IYD
229	Bartter syndrome, type 2	巴特氏症候群，2 型	KCNJ1

230	Hyperinsulinemic hypoglycemia, familial, 2	高胰島素性低血糖，家族性，2	KCNJ11
231	Muscular dystrophy, congenital, merosin deficient or partially deficient	先天性肌肉營養不良症，merosin 缺乏或部分缺乏	LAMA2
232	Epidermolysis bullosa, junctional 2A, intermediate	大皰性表皮鬆解症，交界 2A，中間	LAMA3
233	Epidermolysis bullosa, junctional 1A, intermediate	大皰性表皮鬆解症，交界 1A，中間	LAMB3
234	Epidermolysis bullosa, junctional 3A, intermediate	大皰性表皮鬆解症，交界 3A，中間	LAMC2
235	Muscular dystrophy-dystroglycanopathy	肌肉營養不良症-肌肉營養不良症	LARGE1
236	Leber congenital amaurosis 5	萊伯先天性黑蒙 5	LCA5
237	Hypercholesterolemia, familial LDL cholesterol level QTL2	高膽固醇血症、家族性 LDL 膽固醇水平 QTL2	LDLR
238	Hypercholesterolemia, familial, 4	高膽固醇血症，家族性，4	LDLRAP1
239	Leydig cell hypoplasia	間質細胞發育不全	LHCGR
240	Schwartz-Jampel syndrome, type 2	Schwartz-Jampel 綜合徵，2 型	LIFR
241	Cholesteryl ester storage disease	膽固醇酯貯積病	LIPA
242	Deafness, autosomal recessive 77	耳聾，體染色體隱性遺傳 77	LOXHD1
243	Woolly hair, autosomal recessive 1, with or without hypotrichosis	羊毛狀毛髮，體染色體隱性遺傳 1，伴或不伴少毛症	LPAR6
244	Lipoprotein lipase deficiency	脂蛋白脂肪酶缺乏症	LPL
245	Donnai-Barrow syndrome	唐奈-巴羅氏症候群	LRP2
246	Chediak-Higashi syndrome	切迪亞克-東氏症候群	LYST
247	Retinitis pigmentosa 62	色素性視網膜炎 62	MAK
248	Mannosidosis, alpha-, types I and II	甘露糖苷沉積症，α-，I 型和 II 型	MAN2B1
249	3-Methylcrotonyl-CoA carboxylase 1 deficiency	3-甲基巴豆醯輔酶A羧化酶1缺乏症	MCCC1
250	3-Methylcrotonyl-CoA carboxylase 2 deficiency	3-甲基巴豆醯輔酶A羧化酶2缺乏症	MCCC2
251	Mucolipidosis IV	粘脂沉積症IV	MCOLN1
252	Microcephaly 1, primary, autosomal recessive	小頭畸形 1，原發性，體染色體隱性遺傳	MCPH1
253	Intellectual developmental disorder, X-linked syndromic, Lubs type	智能發育障礙，X連鎖綜合徵，Lubs型	MECP2
254	Familial Mediterranean fever, autosomal recessive	家族性地中海熱，體染色體隱性遺傳	MEFV
255	Megalencephalic leukoencephalopathy with subcortical cysts 1	伴隨皮質下囊腫的巨腦白質腦病變 1	MLC1
256	Malonyl-CoA decarboxylase deficiency	丙二醯輔酶A脫羧酶缺乏症	MLYCD
257	Methylmalonic aciduria, vitamin B12-responsivem cblA type	甲基丙二酸尿症，維生素 B12 反應性 cblA 型	MMAA
258	Methylmalonic aciduria, vitamin B12-responsive, cblB type	甲基丙二酸尿症，維生素 B12 反應性，cblB 型	MMAB
259	Methylmalonic aciduria and homocystinuria, cblC type	甲基丙二酸尿症和高胱胺酸尿症，cblC 型	MMACHC
260	Methylmalonic acidemia mut(0) type	甲基丙二酸血症mut(0)型	MMUT
261	Congenital disorder of glycosylation, type Ib	先天性糖基化障礙，Ib 型	MPI
262	Combined immunodeficiency and megaloblastic anemia with or without hyperhomocysteinemia	合併免疫缺陷和巨幼細胞性貧血伴或不伴高同型半胱氨酸血症	MTHFD1
263	Homocystinuria due to MTHFR deficiency	MTHFR 缺乏導致的同型半胱氨酸尿症	MTHFR
264	Charcot-Marie-Tooth disease, type 4B1	腓骨肌萎縮症，4B1 型	MTMR2
265	Homocystinuria-megaloblastic anemia, cblG complementation type	同型半胱胺酸尿症-巨幼細胞性貧血，cblG 互補型	MTR
266	Homocystinuria-megaloblastic anemia, cbl E type	同型半胱胺酸尿症-巨幼細胞性貧血，cbl E 型	MTRR
267	Abetalipoproteinemia	無β脂蛋白血症	MTTP
268	Mevalonate kinase deficiency	甲羥戊酸激酶缺乏症	MVK
269	Deafness, autosomal recessive 3	耳聾，體染色體隱性遺傳 3	MYO15A
270	Deafness, autosomal recessive 2	耳聾，體染色體隱性遺傳2	MYO7A
271	Infantile liver failure syndrome 2	嬰兒肝衰竭綜合症2	NBAS
272	Nijmegen breakage syndrome	奈梅亨斷裂症候群	NBN
273	Chronic granulomatous disease 2	慢性肉芽腫病2	NCF2
274	Chronic granulomatous disease 3	慢性肉芽腫病3	NCF4
275	Mitochondrial complex I deficiency, nuclear type 10	粒線體複合體 I 缺陷，核型 10	NDUFAF2
276	Mitochondrial complex I deficiency, nuclear type 17	粒線體複合體 I 缺陷，核型 17	NDUFAF6
277	Nemaline myopathy 2, autosomal recessive	線狀肌病變 2，體染色體隱性遺傳	NEB
278	Sialidosis	唾液酸貯積症	NEU1
279	Ichthyosis, congenital, autosomal recessive 6	魚鱗病，先天性，體染色體隱性遺傳 6	NIPAL4
280	Hydatidiform mole, recurrent, 1	葡萄胎，復發性，1	NLRP7
281	Niemann-Pick disease	尼曼-匹克病	NPC1
282	Joubert syndrome 4/ Joubert syndrome 4/Nephronophthisis 1, juvenile	腎結核 1，青少年	NPHP1
283	syndrome	腎結核 3 / 腎-肝-胰發育不良 1 / Meckel-Gruber 綜合徵	NPHP3
284	Nephrotic syndrome, type 1	腎病綜合徵，1 型	NPHS1
285	Nephrotic syndrome, type 2	2 型腎病症候群	NPHS2
286	Gyrate atrophy of choroid and retina with or without ornithinemia	伴隨或不伴隨鳥氨酸血症的脈絡膜和視網膜迴旋萎縮	OAT
287	Albinism, oculocutaneous, type II	白化病，眼皮膚，II 型	OCA2
288	3-methylglutaconic aciduria, type III	3-甲基戊烯酸尿症，III 型	OPA3
289	X-linked intellectual disability-cerebellar hypoplasia syndrome	X連鎖智能障礙-小腦發育不全綜合徵	OPHN1
290	Osteopetrosis, autosomal recessive 5	骨石症，體染色體隱性遺傳 5	OSTM1
291	Auditory neuropathy, autosomal recessive	聽神經病，體染色體隱性遺傳	OTOF
292	Osteogenesis imperfecta, type VIII	成骨不全，VIII 型	P3H1
293	Phenylketonuria	苯酮尿症	PAH
294	CADH Deficiency	CADH缺乏症	PCBD1
295	Propionic acidemia	丙酸血症	PCCA
296	Propionic acidemia	丙酸血症	PCCB
297	Deafness, autosomal recessive 23	耳聾，體染色體隱性遺傳 23	PCDH15
298	Lacticacidemia due to PDX1 deficiency	PDX1缺乏引起的乳酸血症	PDHX
299	Prolidase deficiency	脯胺酸酶缺乏症	PEPD
300	Heimler syndrome 1	海姆勒氏症候群1	PEX1
301	Peroxisome biogenesis disorder 11A (Zellweger)	過氧化物酶體生物發生障礙 11A（Zellweger）	PEX13
302	Peroxisome biogenesis disorder 13A (Zellweger)	過氧化物酶體生物發生障礙 13A（Zellweger）	PEX14
303	Peroxisome biogenesis disorder 9B	過氧化物酶體生物合成障礙 9B	PEX7
304	Glycogen storage disease VII	肝醣貯積病VII	PFKM
305	Immunodeficiency 23	免疫缺陷23	PGM3
306	Refsum disease, Classic	Refsum病，經典	PHYH
307	Deafness, autosomal recessive 59	耳聾，體染色體隱性遺傳 59	PJVK

308	Polycystic kidney disease 4, with or without hepatic disease	多囊性腎4，伴或不伴肝病	PKHD1
309	Infantile neuroaxonal dystrophy	嬰兒神經軸突營養不良	PLA2G6
310	Nephrotic syndrome, type 3	腎病綜合徵，3 型	PLCE1
311	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	Ehlers-Danlos 綜合徵，脊柱後側凸型，1	PLOD1
312	Congenital disorder of glycosylation, type Ia	先天性糖基化障礙，Ia 型	PMM2
313	Immunodeficiency due to purine nucleoside phosphorylase deficiency	嘌呤核苷磷酸化酶缺乏導致的免疫缺陷	PNP
314	Mitochondrial DNA depletion syndrome 4A (Alpers type)	粒線體 DNA 耗竭症候群 4A（Alpers 型）	POLG
315	Xeroderma pigmentosum, variant type	色素性乾皮病，變異型	POLH
316	Leukodystrophy, hypomyelinating, 11	腦白質營養不良，髓鞘形成不足，11	POLR1C
317	Retinitis pigmentosa 76	色素性視網膜炎 76	POMGNT1
318	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1	肌肉營養不良-肌肉營養不良症（先天性腦部和眼部異常），A 型，1	POMT1
319	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2	肌肉營養不良-肌肉營養不良症（先天性腦部和眼部異常），A 型，2	POMT2
320	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	比克斯勒氏症候群	POR
321	Ceroid lipofuscinosis, neuronal, 1	蠟質脂褐質沉著症，神經元，1	PPT1
322	Myasthenic syndrome	肌無力症候群	PREPL
323	Epilepsy, progressive myoclonic 1B	癲癇，進行性肌陣攣 1B	PRICKLE1
324	Immunodeficiency 26, with or without neurologic abnormalities	免疫缺陷 26，伴隨或不伴隨神經系統異常	PRKDC
325	Pituitary hormone deficiency	腦下垂體激素缺乏	PROP1
326	Pituitary hormone deficiency	腦下垂體激素缺乏	PROP1
327	Phosphoribosylpyrophosphate synthetase superactivity	磷酸核糖焦磷酸合成酶超活性	PRPS1
328	Immunodeficiency 105, severe combined	免疫缺陷105，嚴重聯合	PTPRC
329	Breasts andor nipples, aplasia or hypoplasia of, 2	乳房和/或乳頭發育不全或發育不全，2	PTPRF
330	6-Pyruvoyl-tetrahydropterin synthase deficiency	6-丙酮醯四氫蝶呤合成酶缺乏症	PTS
331	Glycogen storage disease V	肝醣貯積症V	PYGM
332	Quinoid dihydropteridine reductase deficiency	醌二氫蝶啶還原酶缺乏症	QDPR
333	Carpenter syndrome	卡本特症候群	RAB23
334	Omenn syndrome	預兆症候群	RAG1
335	Congenital myasthenic syndrome	先天性肌無力症候群	RAPSN
336	Pontoneocerebellar hypoplasia	腦橋小腦發育不全	RARS2
337	MHC class II deficiency, complementation group B	MHC II 類缺陷，互補 B 組	RFXANK
338	Cartilage-hair hypoplasia	軟骨毛髮發育不全	RMRP
339	Aicardi-Goutieres syndrome 4	艾卡迪-古蒂埃症候群 4	RNASEH2A
340	Joubert syndrome 7	朱伯特症候群 7	RPGRIP1L
341	Retinoschisis	視網膜劈裂	RS1
342	Dyskeratosis congenita, autosomal recessive 5	先天性角化不良，體染色體隱性遺傳 5	RTEL1
343	Spastic ataxia, Charlevoix-Saguenay type	痙攣性共濟失調，夏洛瓦-薩格奈型	SACS
344	Oguchi disease-1	大口病-1	SAG
345	Aicardi-Goutieres syndrome 5	艾卡迪-古蒂埃症候群 5	SAMHD1
346	Shwachman-Diamond syndrome 1	舒瓦赫曼-戴蒙德症候群 1	SBDS
347	Epilepsy, progressive myoclonic 4, with or without renal failure	癲癇，進行性肌陣攣 4 級，伴隨或不伴隨腎衰竭	SCARB2
348	Mitochondrial complex IV deficiency, nuclear type 4	粒線體複合體 IV 缺陷，核 4 型	SCO1
349	Bardet-Biedl syndrome 16	Bardet-Biedl 症候群 16	SDCCAG8
350	Pontocerebellar hypoplasia type 2D	2D型腦橋小腦發育不全	SEPSECS
351	Emphysema due to AAT deficiency	AAT缺乏導致的肺氣腫	SERPINA1
352	Muscular dystrophy, limb-girdle, autosomal recessive 3	肌肉營養不良症，肢帶，體染色體隱性遺傳 3	SGCA
353	Muscular dystrophy, limb-girdle, autosomal recessive 4	肌肉營養不良症，肢帶，體染色體隱性遺傳 4	SGCB
354	Muscular dystrophy, limb-girdle, autosomal recessive 6	肌肉營養不良症，肢帶，體染色體隱性遺傳 6	SGCD
355	Muscular dystrophy, limb-girdle, autosomal recessive 5	肌肉營養不良症，肢帶，體染色體隱性遺傳 5	SGCG
356	Mucopolysaccharidosis type III (Sanfilippo A)	III 型黏多醣貯積症（Sanfilippo A）	SGSH
357	Charcot-Marie-Tooth disease, type 4C	腓骨肌萎縮症，4C 型	SH3TC2
358	Gitelman syndrome	吉特曼症候群	SLC12A3
359	Agenesis of the corpus callosum with peripheral neuropathy	胼胝體發育不全伴隨週邊神經病變	SLC12A6
360	Sialic acid storage disorder, infantile	嬰兒唾液酸儲存障礙	SLC17A5
361	Thiamine-responsive megaloblastic anemia syndrome	硫胺素反應性巨幼細胞貧血綜合症	SLC19A2
362	Carnitine deficiency, systemic primary	肉鹼缺乏症，全身性原發性	SLC22A5
363	Citrullinemia, type II, neonatal-onset	瓜胺酸血症，II 型，新生兒發病	SLC25A13
364	Atelosteogenesis, type II	軟骨形成，II 型	SLC26A2
365	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	耳聾，體染色體隱性遺傳 4，伴隨前庭導水管擴大	SLC26A4
366	Arterial tortuosity syndrome	動脈迂曲綜合症	SLC2A10
367	Glycogen storage disease Ib	肝醣累積病 Ib	SLC37A4
368	Cystinuria	胱氨酸尿症	SLC3A1
369	Albinism, oculocutaneous, type IV	白化病，眼皮膚，IV 型	SLC45A2
370	Cerebral creatine deficiency syndrome 1	腦肌酸缺乏症候群1	SLC6A8
371	Lysinuric protein intolerance	賴氨酸尿蛋白不耐受	SLC7A7
372	Cystinuria	胱氨酸尿症	SLC7A9
373	Schimke immuno-osseous dysplasia	Schimke免疫性骨發育不良	SMARCAL1
374	Spinal muscular atrophy	脊髓性肌肉萎縮症	SMN1
375	Niemann-Pick disease	尼曼-匹克病	SMPD1
376	Leber congenital amaurosis 3	萊伯先天性黑蒙3	SPATA7
377	Charcot-Marie-Tooth disease, axonal, type 2X	腓骨肌萎縮症，軸突，2X 型	SPG11
378	Spastic paraplegia 7, autosomal recessive	痙攣性截癱 7，體染色體隱性遺傳	SPG7
379	Netherton syndrome	內瑟頓症候群	SPINK5
380	Pseudovaginal perineoscrotal hypospadias	假性陰道會陰陰囊尿道下裂	SRD5A2
381	Lipoid adrenal hyperplasia	腎上腺類脂增生	STAR
382	Deafness, autosomal recessive 16	耳聾，體染色體隱性遺傳 16	STRC
383	Deafness, autosomal recessive 76	耳聾，體染色體隱性遺傳 76	SYNE4
384	Encephalopathy, progressive, with amyotrophy and optic atrophy	進行性腦病變，伴隨肌肉萎縮和視神經萎縮	TBCE
385	Joubert syndrome 13	朱伯特症候群 13	TCTN1

386	Neuropathy, hereditary sensory and autonomic, type IX, with developmental delay	型，伴隨發育遲緩	TECPR2
387	Hemochromatosis, type 3	血色素沉著症，3 型	TFR2
388	Thyroid dyshormonogenesis 3	甲狀腺激素生成異常 3	TG
389	Segawa syndrome, recessive	瀨川綜合徵，隱性遺傳	TH
390	Hypercholanemia, familial 1	高膽鹼血症，家族性 1	TJP2
391	Mitochondrial DNA depletion syndrome 2 (myopathic type)	粒線體 DNA 耗竭症候群 2（肌肉型）	TK2
392	Deafness, autosomal recessive 7	耳聾，體染色體隱性遺傳 7	TMC1
393	Joubert syndrome 2	朱伯特症候群2	TMEM216
394	Joubert syndrome 14	朱伯特症候群 14	TMEM237
395	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2	粒線體複合體 V（ATP 合成酶）缺陷，核型 2	TMEM70
396	Deafness, autosomal recessive 8/10	耳聾，體染色體隱性遺傳 8/10	TMPRSS3
397	Osteopetrosis, autosomal recessive 2	骨石症，體染色體隱性遺傳 2	TNFSF11
398	Ehlers-Danlos syndrome, classic-like, 1	埃勒斯-當洛斯綜合徵，經典樣，1	TNXB
399	Thyroid dyshormonogenesis 2A	甲狀腺激素生成異常 2A	TPO
400	Ceroid lipofuscinosis, neuronal, 2	蠟質脂褐質沉著症，神經元，2	TPP1
401	Aicardi-Goutieres syndrome 1, dominant and recessive	Aicardi-Goutieres 症候群 1，顯性和隱性	TREX1
402	Muscular dystrophy, limb-girdle, autosomal recessive 8	肌肉營養不良症，肢帶，體染色體隱性遺傳 8	TRIM32
403	Mulibrey nanism	穆利布雷奈米主義	TRIM37
404	Hypothyroidism, congenital, nongoitrous, 1	甲狀腺功能低下症，先天性，非甲狀腺腫，1	TSHR
405	Cardiomyopathy, dilated	擴張型心肌病變	TTN
406	Ataxia with isolated vitamin E deficiency	共濟失調伴隨孤立性維生素 E 缺乏	TTPA
407	Leber congenital amaurosis 15	萊伯先天性黑蒙 15	TULP1
408	Albinism, oculocutaneous, type IB	白化病，眼皮膚，IB 型	TYR
409	Johanson-Blizzard syndrome	約翰遜-暴風雪綜合症	UBR1
410	Gilbert syndrome	吉爾伯特症候群	UGT1A1
411	Intellectual developmental disorder, X-linked syndromic 14	智力發展障礙，X 連鎖症候群 14	UPF3B
412	Deafness, autosomal recessive 18A	耳聾，體染色體隱性遺傳 18A	USH1C
413	Retinitis pigmentosa 39	色素性視網膜炎 39	USH2A
414	Cerebellar hypoplasia, impaired intellectual development, and dysequilibrium syndrome	小腦發育不全、智力發展受損和平衡失調症候群 1	VLDLR
415	Cohen syndrome	科恩症候群	VPS13B
416	Neutropenia, severe congenital, 5, autosomal recessive	遺傳	VPS45
417	Pontocerebellar hypoplasia, type 2E	腦橋小腦發育不全，2E 型	VPS53
418	von Willebrand disease, type 3	馮維勒布蘭德病，3 型	VWF
419	Thrombocytopenia, X-linked	X連鎖血小板減少症	WAS
420	Deafness, autosomal recessive 31	耳聾，體染色體隱性遺傳 31	WHRN
421	Schopf-Schulz-Passarge syndrome	Schopf-Schulz-Passarge 綜合徵	WNT10A
422	Werner syndrome	維爾納症候群	WRN
423	Spinocerebellar ataxia, autosomal recessive 12	脊髓小腦性共濟失調，體染色體隱性遺傳 12	WWOX
424	Xeroderma pigmentosum, group A	色素性乾皮病，A 組	XPA
425	Xeroderma pigmentosum	著色性乾皮病	XPC
426	Immunodeficiency 48	免疫缺陷 48	ZAP70
427	Spastic paraplegia 15, autosomal recessive	痙攣性截癱 15，體染色體隱性遺傳	ZFYVE26
428	Brittle cornea syndrome 1	脆性角膜綜合症1	ZNF469