

Carrier Screening - 302 Genes
攜帶者篩查 - 302種基因

No. 編號	GENE 基因	DISORDER	疾病
1	<i>HMGCL</i>	3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency	3-羥基-3-甲基戊二酰輔酶A(HMG-CoA)裂解酶缺乏症
2	<i>ABCC8</i>	ABCC8-related disorders	ABCC8相關疾病
3	<i>MTTP</i>	Abetalipoproteinemia	無β脂蛋白血症
4	<i>ACAD9</i>	ACAD9 deficiency	醯輔酶A去氫酶9缺乏症
5	<i>CNGB3</i>	Achromatopsia (CNGB3-related)	全色盲(CNGB3相關)
6	<i>SLC39A4</i>	Acrodermatitis enteropathica	腸源性肢端皮炎
7	<i>ADA</i>	Adenosine deaminase deficiency	腺苷脫氨酶缺乏症
8	<i>SAMHD1</i>	Aicardi-Goutieres syndrome (SAMHD1-related)	Aicardi-Goutieres綜合症(SAMHD1相關)
9	<i>CYP11B2</i>	Aldosterone synthase deficiency	醛固酮合成酶缺乏症
10	<i>MAN2B1</i>	Alpha-mannosidosis	α-甘露糖苷貯積症
11	<i>HBA1</i>	Alpha-thalassemia	甲型地中海貧血
12	<i>HBA2</i>		
13	<i>ATRX</i>	Alpha-thalassemia X-linked intellectual disability syndrome	伴甲型地中海貧血性聯遺傳智力低下綜合症
14	<i>COL4A3</i>	Alport Syndrome (COL4A3-related)	亞伯氏綜合症(COL4A3相關)
15	<i>COL4A4</i>	Alport Syndrome (COL4A4-related)	亞伯氏綜合症(COL4A4相關)
16	<i>COL4A5</i>	Alport Syndrome, X-linked (COL4A5-related)	性聯遺傳亞伯氏綜合症(COL4A5相關)
17	<i>ALMS1</i>	Alström syndrome	阿爾斯特倫綜合症
18	<i>SLC12A6</i>	Andermann syndrome	安德曼綜合症
19	<i>ARG1</i>	Arginase deficiency	精氨酸酶缺乏症
20	<i>ASL</i>	Argininosuccinic aciduria	精氨基琥珀酸尿症
21	<i>CYP19A1</i>	Aromatase deficiency	芳香化酶缺乏症
22	<i>ASNS</i>	Asparagine synthetase deficiency	天門冬醯胺合成酶缺乏症
23	<i>AGA</i>	Aspartylglucosaminuria	天門冬氨酸葡萄糖胺尿症
24	<i>TTPA</i>	Ataxia with vitamin E deficiency	共濟失調伴維他命E缺乏症
25	<i>ATM</i>	Ataxia-telangiectasia	共濟失調微血管擴張症
26	<i>AIRE</i>	Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia	自體免疫性多內分泌病變伴陰道念珠菌感染伴外胚層增生不良症
27	<i>LOXHD1</i>	Autosomal recessive deafness 77	常染色體隱性77型失聰
28	<i>SACS</i>	Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	Charlevoix-Saguenay常染色體隱性痙攣性共濟失調
29	<i>BBS10</i>	Bardet-Biedl syndrome (BBS10-related)	巴德-畢德氏綜合症(BBS10相關)
30	<i>BBS12</i>	Bardet-Biedl syndrome (BBS12-related)	巴德-畢德氏綜合症(BBS12相關)
31	<i>BSND</i>	Bartter syndrome type 4A	巴特氏綜合症第4A型
32	<i>BBS1</i>	BBS1-related disorders	BBS1相關疾病

33	<i>BBS2</i>	BBS2-related disorders	BBS2相關疾病
34	<i>ACAT1</i>	Beta-ketothiolase deficiency	β -酮硫解酶缺乏症
35	<i>BLM</i>	Bloom syndrome	布隆氏綜合症
36	<i>ASPA</i>	Canavan disease	海綿狀腦白質營養不良症
37	<i>CPS1</i>	Carbamoylphosphate synthetase I deficiency	氨甲酰磷酸合成酶I缺乏症
38	<i>CPT1A</i>	Carnitine palmitoyltransferase I deficiency	肉鹼棕櫚醯基轉移酶I缺乏症
39	<i>CPT2</i>	Carnitine palmitoyltransferase II deficiency	肉鹼棕櫚醯基轉移酶II缺乏症
40	<i>RAB23</i>	Carpenter syndrome (RAB23-related)	Carpenter綜合症(RAB23相關)
41	<i>RMRP</i>	Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders	軟骨-毛髮發育不全-異常增生譜系疾病
42	<i>CYP27A1</i>	Cerebrotendinous xanthomatosis	腦腱性黃瘤症
43	<i>CFTR</i>	CFTR-related disorders (including cystic fibrosis)	CFTR相關疾病(包括囊狀纖維化)
44	<i>NDRG1</i>	Charcot-Marie-Tooth disease (NDRG1-related)	進行性神經性腓骨萎縮症(NDRG1相關)
45	<i>GJB1</i>	Charcot-Marie-Tooth disease, X-linked (GJB1-related)	進行性神經性腓骨萎縮症性聯遺傳(GJB1相關)
46	<i>VPS13A</i>	Chorea-acanthocytosis	舞蹈棘狀紅血球症
47	<i>CHM</i>	Choroideremia	脈絡膜缺失症
48	<i>CYBA</i>	Chronic granulomatous disease (CYBA-related)	慢性肉芽腫病(CYBA相關)
49	<i>CYBB</i>	Chronic granulomatous disease (CYBB-related)	慢性肉芽腫病(CYBB相關)
50	<i>SLC25A13</i>	Citrin deficiency	希特林蛋白缺陷症
51	<i>ASS1</i>	Citrullinemia type 1	瓜胺酸血症第1型
52	<i>ERCC8</i>	Cockayne syndrome type A	柯凱因氏綜合症A型
53	<i>ERCC6</i>	Cockayne syndrome type B	柯凱因氏綜合症B型
54	<i>VPS13B</i>	Cohen syndrome	科恩綜合症
55	<i>ACSF3</i>	Combined malonic and methylmalonic aciduria (ACSF3-related)	丙二酸及甲基丙二酸聯合尿症(ACSF3相關)
56	<i>GFM1</i>	Combined oxidative phosphorylation deficiency (GFM1-related)	結合性氧化磷酸化缺乏症(GFM1相關)
57	<i>TSFM</i>	Combined oxidative phosphorylation deficiency (TSFM-related)	結合性氧化磷酸化缺乏症(TSFM相關)
58	<i>LHX3</i>	Combined pituitary hormone deficiency (LHX3-related)	結合性腦下垂體賀爾蒙缺失(LHX3相關)
59	<i>PROP1</i>	Combined pituitary hormone deficiency (PROP1-related)	結合性腦下垂體賀爾蒙缺失(PROP1相關)
60	<i>CYP11B1</i>	Congenital adrenal hyperplasia due to 11-beta-hydroxylase-deficiency	先天性腎上腺增生症-11 β 羥化酶缺失症
61	<i>CYP21A2</i>	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	先天性腎上腺增生症-21羥化酶缺失症
62	<i>HSD3B2</i>	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase	先天性腎上腺增生症3 β 羥基類固醇脫氫酶缺乏症2型

		type II deficiency	
63	<i>MPL</i>	Congenital amegakaryocytic thrombocytopenia	先天性巨核細胞缺乏血小板低下症
64	<i>ALG6</i>	Congenital disorder of glycosylation (ALG6-related)	先天性糖基化疾病(ALG6相關)
65	<i>MPI</i>	Congenital disorder of glycosylation (MPI-related)	先天性糖基化疾病(MPI相關)
66	<i>PMM2</i>	Congenital disorder of glycosylation (PMM2-related)	先天性糖基化疾病(PMM2相關)
67	<i>TGM1</i>	Congenital ichthyosis (TGM1-related)	先天性魚鱗癬(TGM1相關)
68	<i>NTRK1</i>	Congenital insensitivity to pain with anhidrosis	先天性痛覺不敏感併無汗症
69	<i>CHRNE</i>	Congenital myasthenic syndrome (CHRNE-related)	先天性肌無力綜合症(CHRNE相關)
70	<i>SLC4A11</i>	Corneal dystrophy and perceptive deafness	角膜失養和感音性失聰症
71	<i>CYP17A1</i>	CYP17A1-related disorders	CYP17A1相關疾病
72	<i>CTNS</i>	Cystinosis	胱胺酸症
73	<i>DHDDS</i>	DHDDS-related disorders	DHDDS相關疾病
74	<i>DLD</i>	Dihydrolipoamide dehydrogenase deficiency (DLD)	二氫硫辛醯胺脫氫酶缺乏症
75	<i>DMD</i>	DMD-related dystrophinopathy	裘馨氏肌肉萎縮症(DMD相關)
76	<i>DYSF</i>	Dysferlinopathy	肌鐵蛋白缺陷型肌營養不良症
77	<i>COL7A1</i>	Dystrophic epidermolysis bullosa (COL7A1-related)	營養不良性大疱性表皮鬆解症(COL7A1相關)
78	<i>ADAMTS2</i>	Ehlers-Danlos syndrome, dermatosparaxis type	埃勒斯當洛斯綜合症(皮膚脆裂型)
79	<i>EVC</i>	Ellis-van Creveld syndrome (EVC-related)	埃利偉氏綜合症(EVC相關)
80	<i>EVC2</i>	Ellis-van Creveld syndrome (EVC2-related)	埃利偉氏綜合症(EVC2相關)
81	<i>EMD</i>	Emery-Dreifuss muscular dystrophy (EMD-related)	Emery-Dreifuss肌肉萎縮症(EMD相關)
82	<i>NR2E3</i>	Enhanced S-cone syndrome/retinitis pigmentosa 37	增強型S錐綜合症/視網膜色素病變37型
83	<i>ETHE1</i>	Ethylmalonic encephalopathy	乙基丙二酸腦病變
84	<i>GLA</i>	Fabry disease	法布瑞氏症
85	<i>F9</i>	Factor IX deficiency (Hemophilia B)	凝血因子IX缺乏症(乙型血友病)
86	<i>ELP1</i>	Familial dysautonomia	家族性自主神經失調症
87	<i>LDLR</i>	Familial hypercholesterolemia (LDLR-related)	家族性高膽固醇血症(LDLR相關)
88	<i>LDLRAP1</i>	Familial hypercholesterolemia (LDLRAP1-related)	家族性高膽固醇血症(LDLRAP1相關)
89	<i>FANCA</i>	Fanconi anemia type A	范科尼貧血A型
90	<i>FANCC</i>	Fanconi anemia type C	范科尼貧血C型
91	<i>FANCG</i>	Fanconi anemia type G	范科尼貧血G型
92	<i>FKRP</i>	FKRP-related disorders	FKRP相關疾病

93	<i>FKTN</i>	FKTN-related disorders	FKTN相關疾病
94	<i>FMR1</i>	Fragile X syndrome	脆性X綜合症
95	<i>FH</i>	Fumarate hydratase deficiency	延胡索酸酶缺乏症
96	<i>GALK1</i>	Galactokinase deficiency galactosemia	半乳糖激酶缺乏症
97	<i>GALT</i>	Galactosemia (GALT-related)	半乳糖血症(GALT相關)
98	<i>GBA</i>	Gaucher disease	高雪氏症
99	<i>SLC12A3</i>	Gitelman syndrome (SLC12A3-related)	吉特曼綜合症(SLC12A3相關)
100	<i>GJB2</i>	GJB2-related DFNB1 nonsyndromic hearing loss and deafness	GJB2相關DFNB1非綜合症性聽力損失和耳聾
101	<i>GLE1</i>	GLE1-related disorders	GLE1相關疾病
102	<i>GCDH</i>	Glutaric acidemia type I	戊二酸血症1型
103	<i>ETFA</i>	Glutaric acidemia type IIA	戊二酸血症2A型
104	<i>ETFDH</i>	Glutaric acidemia type IIC	戊二酸血症2C型
105	<i>AMT</i>	Glycine encephalopathy (AMT-related)	甘氨酸腦病(AMT相關)
106	<i>GLDC</i>	Glycine encephalopathy (GLDC-related)	甘氨酸腦病(GLDC相關)
107	<i>G6PC</i>	Glycogen storage disease type IA	肝醣儲積症1A型
108	<i>SLC37A4</i>	Glycogen storage disease type IB	肝醣儲積症1B型
109	<i>GAA</i>	Glycogen storage disease type II (Pompe disease)	肝醣儲積症2型(龐貝氏症)
110	<i>AGL</i>	Glycogen storage disease type III	肝醣儲積症3型
111	<i>GBE1</i>	Glycogen storage disease type IV/ adult polyglucosan body disease	肝醣儲積症4型(成人葡萄糖多聚體病)
112	<i>PYGM</i>	Glycogen storage disease type V	肝醣儲積症5型
113	<i>PFKM</i>	Glycogen storage disease type VII	肝醣儲積症7型
114	<i>BCS1L</i>	GRACILE syndrome/BCS1L-related disorders	GRACILE綜合症/BCS1L相關疾病
115	<i>GAMT</i>	Guanidinoacetate methyltransferase deficiency	胍基乙酸甲基轉移酶缺乏症
116	<i>HBB</i>	HBB-related hemoglobinopathies	HBB相關血紅蛋白病
117	<i>ALDOB</i>	Hereditary fructose intolerance	遺傳性果糖不耐症
118	<i>HJV</i>	Hereditary hemochromatosis type 2 (HJV-related)	遺傳性血色素沉著症2型(HJV相關)
119	<i>TFR2</i>	Hereditary hemochromatosis type 3	遺傳性血色素沉著症3型
120	<i>HPS1</i>	Hermansky-Pudlak syndrome type 1	Hermansky-Pudlak綜合症1型
121	<i>HPS3</i>	Hermansky-Pudlak syndrome type 3	Hermansky-Pudlak綜合症3型
122	<i>HLCS</i>	Holocarboxylase synthetase deficiency	多發性羧化酶缺乏症
123	<i>CBS</i>	Homocystinuria due to CBS deficiency	胱硫醚β合成酶缺乏性高胱氨酸尿症
124	<i>MTHFR</i>	Homocystinuria due to MTHFR deficiency	高胱氨酸尿症(因缺乏亞甲基四氫葉酸還原酶)
125	<i>MTRR</i>	Homocystinuria, cobalamin E type	高胱氨酸尿症(鈷胺素E型)
126	<i>HSD17B4</i>	HSD17B4-related disorders	HSD17B4相關疾病

127	<i>HYLS1</i>	Hydrolethalus syndrome type 1	Hydrolethalus綜合症1型
128	<i>SLC25A15</i>	Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome	高鳥氨酸血症-高氨血症-同型瓜氨酸尿綜合症
129	<i>EDA</i>	Hypohidrotic ectodermal dysplasia (EDA-related)	少汗性外胚層發育不良症(EDA相關)
130	<i>ALPL</i>	Hypophosphatasia	低磷酸酯酶症
131	<i>GNE</i>	Inclusion body myopathy 2	包涵體肌炎2型
132	<i>IVD</i>	Isovaleric acidemia	異戊酸血症
133	<i>TMEM216</i>	Joubert syndrome 2/TMEM216-related disorders	茹貝爾綜合症2型/TMEM216相關疾病
134	<i>LAMB3</i>	Junctional epidermolysis bullosa (LAMB3-related)	接合性表皮溶解水皰症(LAMB3相關)
135	<i>LAMC2</i>	Junctional epidermolysis bullosa (LAMC2-related)	接合性表皮溶解水皰症(LAMC2相關)
136	<i>KCNJ11</i>	KCNJ11-related disorders	KCNJ11相關疾病
137	<i>GALC</i>	Krabbe disease	球細胞腦白質失養症(Krabbe病)
138	<i>LAMA2</i>	LAMA2-related muscular dystrophy	LAMA2相關肌肉失養症
139	<i>LAMA3</i>	LAMA3-related disorders	LAMA3相關疾病
140	<i>CEP290</i>	Leber congenital amaurosis 10/ CEP290-related disorders	萊伯氏先天性黑矇症10型/CEP290相關疾病
141	<i>RDH12</i>	Leber congenital amaurosis 13	萊伯氏先天性黑矇症13型
142	<i>LCA5</i>	Leber congenital amaurosis 5	萊伯氏先天性黑矇症5型
143	<i>CRB1</i>	Leber congenital amaurosis 8/CRB1-related disorders	萊伯氏先天性黑矇症8型/CRB1相關疾病
144	<i>LRPPRC</i>	Leigh syndrome, French Canadian type	Leigh綜合症法裔加拿大型
145	<i>EIF2B5</i>	Leukoencephalopathy with vanishing white matter (EIF2B5-related)	白質消融性白質腦病(EIF2B5相關)
146	<i>CAPN3</i>	Limb-girdle muscular dystrophy type 2A (calpainopathy)	肢帶型肌肉失養症2A型(鈣蛋白酶病)
147	<i>SGCG</i>	Limb-girdle muscular dystrophy type 2C	肢帶型肌肉失養症2C型
148	<i>SGCA</i>	Limb-girdle muscular dystrophy type 2D	肢帶型肌肉失養症2D型
149	<i>SGCB</i>	Limb-girdle muscular dystrophy type 2E	肢帶型肌肉失養症2E型
150	<i>STAR</i>	Lipoid congenital adrenal hyperplasia	先天性類脂性腎上腺皮質增生症
151	<i>LPL</i>	Lipoprotein lipase deficiency	脂蛋白酶脂解酵素缺乏症
152	<i>HADHA</i>	Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency	長鏈3-羥烷基輔酶A脫氫酶缺乏症
153	<i>SLC7A7</i>	Lysinuric protein intolerance	賴氨酸尿蛋白不耐受症
154	<i>LIPA</i>	Lysosomal acid lipase deficiency	溶酶體酸性脂肪酶缺乏症
155	<i>CIITA</i>	Major histocompatibility complex class II deficiency (CIITA-related)	主要組織相容性複合體II類分子缺陷(CIITA相關)
156	<i>BCKDHA</i>	Maple syrup urine disease (MSUD) type 1A	楓糖尿症1A型
157	<i>BCKDHB</i>	Maple syrup urine disease (MSUD)	楓糖尿症1B型

		type 1B	
158	<i>DBT</i>	Maple syrup urine disease (MSUD) type 2	楓糖尿症2型
159	<i>ACADM</i>	Medium chain acyl-CoA dehydrogenase (MCAD) deficiency	中鏈醯輔酶A去氫酶缺乏症
160	<i>MLC1</i>	Megalencephalic leukoencephalopathy with subcortical cysts type 1	巨腦性腦白質病伴皮層下囊腫1型
161	<i>ATP7A</i>	Menkes disease/ATP7A-related disorders	孟克斯氏症/ATP7A相關疾病
162	<i>ARSA</i>	Metachromatic leukodystrophy (ARSA-related)	異染性腦白質營養不良(ARSA相關)
163	<i>MMAA</i>	Methylmalonic acidemia (MMAA-related)	甲基丙二酸血症(MMAA相關)
164	<i>MMAB</i>	Methylmalonic acidemia (MMAB-related)	甲基丙二酸血症(MMAB相關)
165	<i>MUT</i>	Methylmalonic acidemia (MUT-related)	甲基丙二酸血症(MUT相關)
166	<i>MMACHC</i>	Methylmalonic acidemia with homocystinuria, cobalamin C type	甲基丙二酸血症併高胱胺酸血症(鈷胺素C型)
167	<i>MMADHC</i>	Methylmalonic acidemia with homocystinuria, cobalamin D type	甲基丙二酸血症併高胱胺酸血症(鈷胺素D型)
168	<i>VSX2</i>	Microphthalmia /clinical anophthalmia (VSX2-related)	小眼畸形/臨床無眼畸形(VSX2相關)
169	<i>NDUFAF5</i>	Mitochondrial complex I deficiency/ Leigh syndrome (NDUFAF5-related)	粒線體酵素複合物I缺乏症/Leigh綜合症(NDUFAF5相關)
170	<i>NDUFS6</i>	Mitochondrial complex I deficiency/ Leigh syndrome (NDUFS6-related)	粒線體酵素複合物I缺乏症/Leigh綜合症(NDUFS6相關)
171	<i>MPV17</i>	Mitochondrial DNA depletion syndrome (MPV17-related)	粒線體DNA耗竭綜合症(MPV17相關)
172	<i>PUS1</i>	Mitochondrial myopathy and sideroblastic anemia 1	粒線體肌病和鐵粒細胞性貧血1型
173	<i>TYMP</i>	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	粒線體神經胃腸腦病變症
174	<i>MKS1</i>	MKS1-related disorders	MKS1相關疾病
175	<i>GNPTAB</i>	Mucopolipidosis type II/III (GNPTAB-related)	黏脂質症2/3型(GNPTAB相關)
176	<i>GNPTG</i>	Mucopolipidosis type III (GNPTG-related)	黏脂質症3型(GNPTG相關)
177	<i>MCOLN1</i>	Mucopolipidosis type IV	黏脂質症4型
178	<i>IDUA</i>	Mucopolysaccharidosis type I	黏多醣症1型
179	<i>IDS</i>	Mucopolysaccharidosis type II (Hunter syndrome)	黏多醣症2型(韓特氏綜合症)
180	<i>SGSH</i>	Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome)	黏多醣症3A型(聖菲利柏氏綜合症A型)
181	<i>NAGLU</i>	Mucopolysaccharidosis type IIIB (Sanfilippo B syndrome)	黏多醣症3B型(聖菲利柏氏綜合症B型)
182	<i>HGSNAT</i>	Mucopolysaccharidosis type IIIC (Sanfilippo C syndrome)/retinitis pigmentosa 73	黏多醣症3C型(聖菲利柏氏綜合症C型)/視網膜色素病變73型

183	<i>GNS</i>	Mucopolysaccharidosis type IIID (Sanfilippo D syndrome)	黏多醣症3D型(聖菲利柏氏綜合症D型)
184	<i>GLB1</i>	Mucopolysaccharidosis type IVB (Morquio B syndrome)/GM1 gangliosidosis	黏多醣症4B型(莫奎歐綜合症B型)/GM1神經節苷脂儲積症
185	<i>ARSB</i>	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	黏多醣症6型(Maroteaux-Lamy綜合症)
186	<i>HYAL1</i>	Mucopolysaccharidosis type IX	黏多醣症9型
187	<i>SUMF1</i>	Multiple sulfatase deficiency	多發性硫酸脂酶缺乏症
188	<i>NAGS</i>	N-Acetylglutamate synthase deficiency	N-乙醯穀胺酸合成酶缺乏症
189	<i>NEB</i>	Nemaline myopathy 2	桿狀體肌症-2型
190	<i>AQP2</i>	Nephrogenic diabetes insipidus (AQP2-related)	腎性尿崩症(AQP2相關)
191	<i>NPHS1</i>	Nephrotic syndrome/ congenital Finnish nephrosis (NPHS1-related)	腎病綜合症(NPHS1相關)
192	<i>NPHS2</i>	Nephrotic syndrome/steroid-resistant nephrotic syndrome (NPHS2-related)	類固醇抗性腎病綜合症(NPHS2相關)
193	<i>TPP1</i>	Neuronal ceroid-lipofuscinosis (TPP1-related)	神經元蠟樣脂褐質沉著症(TPP1相關)
194	<i>CLN3</i>	Neuronal ceroid-lipofuscinosis (CLN3-related)	神經元蠟樣脂褐質沉著症(CLN3相關)
195	<i>CLN5</i>	Neuronal ceroid-lipofuscinosis (CLN5-related)	神經元蠟樣脂褐質沉著症(CLN5相關)
196	<i>CLN6</i>	Neuronal ceroid-lipofuscinosis (CLN6-related)	神經元蠟樣脂褐質沉著症(CLN6相關)
197	<i>MFSD8</i>	Neuronal ceroid-lipofuscinosis (MFSD8-related)	神經元蠟樣脂褐質沉著症(MFSD8相關)
198	<i>PPT1</i>	Neuronal ceroid-lipofuscinosis (PPT1-related)	神經元蠟樣脂褐質沉著症(PPT1相關)
199	<i>CLN8</i>	Neuronal ceroid-lipofuscinosis/ Northern epilepsy (CLN8-related)	神經元蠟樣脂褐質沉著症/北方癲癇型(CLN8相關)
200	<i>SMPD1</i>	Niemann-Pick disease type A/B	尼曼匹克症A/B型
201	<i>NPC1</i>	Niemann-Pick disease type C (NPC1-related)	尼曼匹克症C型(NPC1相關)
202	<i>NPC2</i>	Niemann-Pick disease type C (NPC2-related)	尼曼匹克症C型(NPC2相關)
203	<i>NBN</i>	Nijmegen breakage syndrome	奈梅亨破損綜合症
204	<i>OPA3</i>	OPA3-related conditions	OPA3相關疾病
205	<i>OAT</i>	Ornithine aminotransferase deficiency	鳥胺酸酮酸轉胺酶缺乏症
206	<i>OTC</i>	Ornithine transcarbamylase (OTC) deficiency	鳥胺酸氨甲醯基轉移酶缺乏症
207	<i>TCIRG1</i>	Osteopetrosis (TCIRG1-related)	骨質石化症(TCIRG1相關)
208	<i>SLC26A4</i>	Pendred syndrome	Pendred氏綜合症
209	<i>ACOX1</i>	Peroxisomal acyl-CoA oxidase deficiency	過氧化物酶酰基輔酶A氧化酶缺乏症
210	<i>PAH</i>	Phenylalanine hydroxylase deficiency (including Phenylketonuria (PKU))	苯丙氨酸羥化酶缺乏症(包括苯酮尿症)

211	<i>PHGDH</i>	Phosphoglycerate dehydrogenase deficiency/Neu-Laxova syndrome type 1	磷酸甘油酸脫氫酶缺乏症/Neu-Laxova綜合症1型
212	<i>PKHD1</i>	Polycystic kidney disease (PKHD1-related)	多囊性腎病變(PKHD1相關)
213	<i>ADGRG1</i>	Polymicrogyria (ADGRG1-related)	多小腦回(ADGRG1相關)
214	<i>POMGNT1</i>	POMGNT1-related disorders	POMGNT1相關疾病
215	<i>RARS2</i>	Pontocerebellar hypoplasia (RARS2-related)	橋腦小腦發育不全(RARS2相關)
216	<i>SEPSECS</i>	Pontocerebellar hypoplasia (SEPSECS-related)	橋腦小腦發育不全(SEPSECS相關)
217	<i>MED17</i>	Postnatal progressive microcephaly with seizures and brain atrophy/ Infantile cerebral and cerebellar atrophy (MED17-related)	產後進行性小頭畸形伴癲癇發作和腦萎縮/嬰兒腦和小腦萎縮(MED17相關)
218	<i>SLC22A5</i>	Primary carnitine deficiency	原發性肉鹼缺乏症
219	<i>DNAH5</i>	Primary Ciliary Dyskinesia (DNAH5-related)	原發性纖毛運動障礙(DNAH5相關)
220	<i>DNAI1</i>	Primary Ciliary Dyskinesia (DNAI1-related)	原發性纖毛運動障礙(DNAI1相關)
221	<i>DNAI2</i>	Primary Ciliary Dyskinesia (DNAI2-related)	原發性纖毛運動障礙(DNAI2相關)
222	<i>AGXT</i>	Primary hyperoxaluria type 1	原發性高草酸尿症1型
223	<i>GRHPR</i>	Primary hyperoxaluria type 2	原發性高草酸尿症2型
224	<i>HOGA1</i>	Primary hyperoxaluria type 3	原發性高草酸尿症3型
225	<i>ABCB11</i>	Progressive familial intrahepatic cholestasis type 2	進行性家族性肝內膽汁滯留症2型
226	<i>PCCA</i>	Propionic acidemia (PCCA-related)	丙酸血症(PCCA相關)
227	<i>PCCB</i>	Propionic acidemia (PCCB-related)	丙酸血症(PCCB相關)
228	<i>PRPS1</i>	PRPS1-related disorders	PRPS1相關疾病
229	<i>PSAP</i>	PSAP-related disorders	PSAP相關疾病
230	<i>CTSK</i>	Pycnodysostosis	緻密性成骨不全症
231	<i>PC</i>	Pyruvate carboxylase deficiency	丙酮酸羧化酶缺乏症
232	<i>PDHA1</i>	Pyruvate dehydrogenase complex deficiency (PDHA1-related)	丙酮酸鹽脫氫酶缺乏症(PDHA1相關)
233	<i>PDHB</i>	Pyruvate dehydrogenase complex deficiency (PDHB-related)	丙酮酸鹽脫氫酶缺乏症(PDHB相關)
234	<i>RAPSN</i>	RAPSN-related disorders	RAPSN相關疾病
235	<i>ATP6V1B1</i>	Renal tubular acidosis with deafness (ATP6V1B1-related)	腎小管酸中毒伴失聰(ATP6V1B1相關)
236	<i>EYS</i>	Retinitis pigmentosa 25	視網膜色素病變25型
237	<i>CERKL</i>	Retinitis pigmentosa 26	視網膜色素病變26型
238	<i>FAM161A</i>	Retinitis Pigmentosa 28	視網膜色素病變28型
239	<i>PEX7</i>	Rhizomelic chondrodysplasia punctata type 1/ Refsum disease (PEX7-related)	肢近端型點狀軟骨發育不良1型/雷弗素姆病(PEX7相關)
240	<i>AGPS</i>	Rhizomelic chondrodysplasia punctata type 3	肢近端型點狀軟骨發育不良3型

241	<i>ESCO2</i>	Roberts syndrome	羅伯氏綜合症
242	<i>RPE65</i>	RPE65-related disorders	RPE65相關疾病
243	<i>RPGRIP1L</i>	RPGRIP1L-related disorders	RPGRIP1L相關疾病
244	<i>RTEL1</i>	RTEL-1-related disorders	RTEL-1相關疾病
245	<i>HEXB</i>	Sandhoff disease	Sandhoff病/GM2神經節苷脂儲積症
246	<i>SMARCAL1</i>	Schimke immuno-osseous dysplasia	Schimke免疫骨發育不良
247	<i>DCLRE1C</i>	Severe combined immune deficiency (DCLRE1C-related)	嚴重聯合免疫缺陷症(DCLRE1C相關)
248	<i>RAG2</i>	Severe combined immunodeficiency (RAG2-related)	嚴重聯合免疫缺陷症(RAG2相關)
249	<i>VPS45</i>	Severe congenital neutropenia due to VPS45-deficiency	嚴重先天性嗜中性球減少症(因VPS45缺失)
250	<i>HAX1</i>	Severe congenital neutropenia type 3	嚴重先天性嗜中性球減少症3型
251	<i>SLC17A5</i>	Sialic acid storage disorders	唾液酸貯積症
252	<i>ALDH3A2</i>	Sjögren-Larsson syndrome	Sjögren-Larsson氏綜合症
253	<i>SLC26A2</i>	SLC26A2-related disorders	SLC26A2相關疾病
254	<i>SLC35A3</i>	SLC35A3-related disorders	SLC35A3相關疾病
255	<i>DHCR7</i>	Smith-Lemli-Opitz syndrome	Smith-Lemli-Opitz綜合症
256	<i>ZFYVE26</i>	Spastic paraplegia type 15	痙攣性下身麻痺15型
257	<i>TECPR2</i>	Spastic paraplegia type 49	痙攣性下身麻痺49型
258	<i>SMN1</i>	Spinal muscular atrophy	脊髓性肌肉萎縮症
259	<i>MESP2</i>	Spondylothoracic dysostosis	脊椎肋骨發育不全
260	<i>COL27A1</i>	Steel Syndrome	鋼鐵綜合症
261	<i>LIFR</i>	Stüve-Wiedemann syndrome	Stüve-Wiedemann綜合症
262	<i>HEXA</i>	Tay-Sachs disease/hexosaminidase A deficiency	戴薩克斯症/己醯胺酶A缺乏症
263	<i>PTS</i>	Tetrahydrobiopterin deficiency (PTS-related)	四氫生物蝶呤缺乏症(PTS相關)
264	<i>TRMU</i>	Transient infantile liver failure	急性新生兒肝衰竭
265	<i>TH</i>	Tyrosine hydroxylase deficiency	酪胺酸羥化酶缺乏症
266	<i>FAH</i>	Tyrosinemia type I	酪胺酸血症1型
267	<i>TAT</i>	Tyrosinemia type II	酪胺酸血症2型
268	<i>MYO7A</i>	Usher syndrome type IB/ MYO7A-related disorders	尤塞氏綜合症1B型/MYO7A相關疾病
269	<i>USH1C</i>	Usher syndrome type IC/ USH1C-related disorders	尤塞氏綜合症1C型/USH1C相關疾病
270	<i>CDH23</i>	Usher syndrome type ID	尤塞氏綜合症1D型
271	<i>PCDH15</i>	Usher syndrome type IF/ PCDH15-related disorders	尤塞氏綜合症1F型/PCDH15相關疾病
272	<i>USH2A</i>	Usher syndrome type IIA/ USH2A-related disorders	尤塞氏綜合症2A型/USH2A相關疾病
273	<i>CLRN1</i>	Usher syndrome type IIIA	尤塞氏綜合症3A型
274	<i>ACADVL</i>	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	特長鏈醯輔酶A去氫酶缺乏症
275	<i>VRK1</i>	VRK1-related disorders	VRK1相關疾病

276	<i>ATP7B</i>	Wilson disease	威爾森氏症
277	<i>WNT10A</i>	WNT10A-related disorders	WNT10A相關疾病
278	<i>ABCD1</i>	X-linked adrenoleukodystrophy	性聯遺傳腎上腺白質營養不良
279	<i>SLC6A8</i>	X-linked creatine transporter deficiency	性聯遺傳肌酸轉運載體缺乏症
280	<i>RS1</i>	X-linked juvenile retinoschisis	性聯遺傳視網膜裂損症
281	<i>MTM1</i>	X-linked myotubular myopathy	性聯遺傳肌小管病變
282	<i>IL2RG</i>	X-linked severe combined immunodeficiency (X-SCID)	性聯遺傳嚴重聯合免疫缺陷症
283	<i>XPA</i>	Xeroderma pigmentosum complementation group A	著色性乾皮症-A型
284	<i>XPC</i>	Xeroderma pigmentosum complementation group C	著色性乾皮症-C型
285	<i>PEX1</i>	Zellweger spectrum disorder (PEX1-related)	柴爾維格氏症(PEX1相關)
286	<i>PEX10</i>	Zellweger spectrum disorder (PEX10-related)	柴爾維格氏症(PEX10相關)
287	<i>PEX12</i>	Zellweger spectrum disorder (PEX12-related)	柴爾維格氏症(PEX12相關)
288	<i>PEX2</i>	Zellweger spectrum disorder (PEX2-related)	柴爾維格氏症(PEX2相關)
289	<i>PEX6</i>	Zellweger spectrum disorder (PEX6-related)	柴爾維格氏症(PEX6相關)
290	<i>BTD</i>	Biotinidase deficiency	生物素酶缺乏症
291	<i>F11</i>	Factor XI deficiency (Hemophilia C)	第11凝血因子缺乏症(血友病C)
292	<i>SLC25A20</i>	Carnitine-acylcarnitine translocase deficiency	肉碱-酰基肉碱轉位酶缺乏症
293	<i>MKKS</i>	Bardet-Biedl syndrome 6	巴德-畢德氏綜合症(6型)
294	<i>G6PD</i>	Glucose-6-phosphate dehydrogenase (G6PD) deficiency	6-磷酸葡萄糖脫氫酶(G6PD)缺乏症(蠶豆症)
295	<i>GP1BA</i>	GP1BA-related conditions	GP1BA相關疾病
296	<i>GP9</i>	Bernard-Soulier syndrome (GP9-related)	Bernard-Soulier綜合症(GP9相關)
297	<i>HADH</i>	3-hydroxyacyl-CoA dehydrogenase deficiency	3-羥基輔酶A脫氫酶缺乏症
298	<i>HADHB</i>	Mitochondrial trifunctional protein deficiency, HADHA-related	線粒體三功能蛋白缺乏症 (HADHA-相關)
299	<i>MCCC1</i>	3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC1-related)	三甲基巴豆醯輔酶A梭化酶缺乏症(MCC1相關)
300	<i>MCCC2</i>	3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCC2-related)	三甲基巴豆醯輔酶A梭化酶缺乏症(MCC2相關)
301	<i>MEFV</i>	Familial mediterranean fever	家族性地中海熱
302	<i>SERPINA1</i>	Alpha-1 antitrypsin deficiency	α1-抗胰蛋白酶缺乏症