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

No. 編號	*GENE 基因*	DISORDER	疾病
1	*HMGCL*	3-hydroxy-3-methylglutarayl-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	*ABCC8*	ABCC8-related disorders	ABCC8相關疾病
3	*MTTP*	Abetalipoproteinemia	無β脂蛋白血症
4	*ACAD9*	ACAD9 deficiency	醯輔酶A去氫酶9缺乏症
5	*CNGB3*	Achromatopsia (CNGB3-related)	全色盲(CNGB3相關)
6	*SLC39A4*	Acrodermatitis enteropathica	腸源性肢端皮炎
7	*ADA*	Adenosine deaminase deficiency	腺苷脱氨酶缺乏症
8	*SAMHD1*	Aicardi-Goutieres syndrome (SAMHD1-related)	Aicardi-Goutieres綜合症(SAMHD1相關)
9	*CYP11B2*	Aldosterone synthase deficiency	醛固酮合成酶缺乏症
10	*MAN2B1*	Alpha-mannosidosis	α-甘露糖苷貯積症
11	*HBA1*	Alpha-thalassemia	甲型地中海貧血
12	*HBA2*		
13	*ATRX*	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	*COL4A3*	Alport Syndrome (COL4A3-related)	亞伯氏綜合症(COL4A3相關)
15	*COL4A4*	Alport Syndrome (COL4A4-related)	亞伯氏綜合症(COL4A4相關)
16	*COL4A5*	Alport Syndrome, X-linked (COL4A5-related)	性聯遺傳亞伯氏綜合症(COL4A5相關)
17	*ALMS1*	Alström syndrome	阿爾斯特倫綜合症
18	*SLC12A6*	Andermann syndrome	安德曼綜合症
19	*ARG1*	Arginase deficiency	精氨酸酶缺乏症
20	*ASL*	Argininosuccinic aciduria	精氨基琥珀酸尿症
21	*CYP19A1*	Aromatase deficiency	芳香化酶缺乏症
22	*ASNS*	Asparagine synthetase deficiency	天門冬醯胺合成酶缺乏症
23	*AGA*	Aspartylglucosaminuria	天門冬氨酸葡萄糖胺尿症
24	*TTPA*	Ataxia with vitamin E deficiency	共濟失調伴維他命E缺乏症
25	*ATM*	Ataxia-telangiectasia	共濟失調微血管擴張症
26	*AIRE*	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	*LOXHD1*	Autosomal recessive deafness 77	常染色體隱性77型失聰
28	*SACS*	Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS)	Charlevoix-Saguenay常染色體隱性痙攣性共濟失調
29	*BBS10*	Bardet-Biedl syndrome (BBS10-related)	巴德-畢德氏綜合症(BBS10相關)
30	*BBS12*	Bardet-Biedl syndrome (BBS12-related)	巴德-畢德氏綜合症(BBS12相關)
31	*BSND*	Bartter syndrome type 4A	巴特氏綜合症第4A型
32	*BBS1*	BBS1-related disorders	BBS1相關疾病

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

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

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93	*FKTN*	FKTN-related disorders	FKTN相關疾病
94	*FMR1*	Fragile X syndrome	脆性X綜合症
95	*FH*	Fumarate hydratase deficiency	延胡索酸酶缺乏症
96	*GALK1*	Galactokinase deficiency galactosemia	半乳糖激酶缺乏症
97	*GALT*	Galactosemia (GALT-related)	半乳糖血症(GALT相關)
98	*GBA*	Gaucher disease	高雪氏症
99	*SLC12A3*	Gitelman syndrome (SLC12A3-related)	吉特曼綜合症(SLC12A3相關)
100	*GJB2*	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	*GLE1*	GLE1-related disorders	GLE1相關疾病
102	*GCDH*	Glutaric acidemia type I	戊二酸血症1型
103	*ETFA*	Glutaric acidemia type IIA	戊二酸血症2A型
104	*ETFDH*	Glutaric acidemia type IIC	戊二酸血症2C型
105	*AMT*	Glycine encephalopathy (AMT-related)	甘氨酸腦病(AMT相關)
106	*GLDC*	Glycine encephalopathy (GLDC-related)	甘氨酸腦病(GLDC相關)
107	*G6PC1*	Glycogen storage disease type IA	肝醣儲積症1A型
108	*SLC37A4*	Glycogen storage disease type IB	肝醣儲積症1B型
109	*GAA*	Glycogen storage disease type II (Pompe disease)	肝醣儲積症2型(龐貝氏症)
110	*AGL*	Glycogen storage disease type III	肝醣儲積症3型
111	*GBE1*	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	*PYGM*	Glycogen storage disease type V	肝醣儲積症5型
113	*PFKM*	Glycogen storage disease type VII	肝醣儲積症7型
114	*BCS1L*	GRACILE syndrome/BCS1L-related disorders	GRACILE綜合症/BCS1L相關疾病
115	*GAMT*	Guanidinoacetate methyltransferase deficiency	胍基乙酸甲基轉移酶缺乏症
116	*HBB*	HBB-related hemoglobinopathies	HBB相關血紅蛋白病
117	*ALDOB*	Hereditary fructose intolerance	遺傳性果糖不耐症
118	*HJV*	Hereditary hemochromatosis type 2 (HJV-related)	遺傳性血色素沉著症2型(HJV相關)
119	*TFR2*	Hereditary hemochromatosis type 3	遺傳性血色素沉著症3型
120	*HPS1*	Hermansky-Pudlak syndrome type 1	Hermansky-Pudlak綜合症1型
121	*HPS3*	Hermansky-Pudlak syndrome type 3	Hermansky-Pudlak綜合症3型
122	*HLCS*	Holocarboxylase synthetase deficiency	多發性羧化酶缺乏症
123	*CBS*	Homocystinuria due to CBS deficiency	胱硫醚β合成酶缺乏性高胱氨酸尿症
124	*MTHFR*	Homocystinuria due to MTHFR deficiency	高胱胺酸尿症(因缺乏亞甲基四氫葉酸還原酶)
125	*MTRR*	Homocystinuria, cobalamin E type	高胱胺酸尿症(鈷胺素E型)
126	*HSD17B4*	HSD17B4-related disorders	HSD17B4相關疾病

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

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

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

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211	*PHGDH*	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	*PKHD1*	Polycystic kidney disease (PKHD1-related)	多囊性腎病變(PKHD1相關)
213	*ADGRG1*	Polymicrogyria (ADGRG1-related)	多小腦回(ADGRG1相關)
214	*POMGNT1*	POMGNT1-related disorders	POMGNT1相關疾病
215	*RARS2*	Pontocerebellar hypoplasia (RARS2-related)	橋腦小腦發育不全(RARS2相關)
216	*SEPSECS*	Pontocerebellar hypoplasia (SEPSECS-related)	橋腦小腦發育不全(SEPSECS相關)
217	*MED17*	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	*SLC22A5*	Primary carnitine deficiency	原發性肉鹼缺乏症
219	*DNAH5*	Primary Ciliary Dyskinesia (DNAH5-related)	原發性纖毛運動障礙(DNAH5相關)
220	*DNAI1*	Primary Ciliary Dyskinesia (DNAI1-related)	原發性纖毛運動障礙(DNAI1相關)
221	*DNAI2*	Primary Ciliary Dyskinesia (DNAI2-related)	原發性纖毛運動障礙(DNAI2相關)
222	*AGXT*	Primary hyperoxaluria type 1	原發性高草酸尿症1型
223	*GRHPR*	Primary hyperoxaluria type 2	原發性高草酸尿症2型
224	*HOGA1*	Primary hyperoxaluria type 3	原發性高草酸尿症3型
225	*ABCB11*	Progressive familial intrahepatic cholestasis type 2	進行性家族性肝內膽汁滯留症2型
226	*PCCA*	Propionic acidemia (PCCA-related)	丙酸血症(PCCA相關)
227	*PCCB*	Propionic acidemia (PCCB-related)	丙酸血症(PCCB相關)
228	*PRPS1*	PRPS1-related disorders	PRPS1相關疾病
229	*PSAP*	PSAP-related disorders	PSAP相關疾病
230	*CTSK*	Pycnodysostosis	緻密性成骨不全症
231	*PC*	Pyruvate carboxylase deficiency	丙酮酸羧化酶缺乏症
232	*PDHA1*	Pyruvate dehydrogenase complex deficiency (PDHA1-related)	丙酮酸鹽脫氫酶缺乏症(PDHA1相關)
233	*PDHB*	Pyruvate dehydrogenase complex deficiency (PDHB-related)	丙酮酸鹽脫氫酶缺乏症(PDHB相關)
234	*RAPSN*	RAPSN-related disorders	RAPSN相關疾病
235	*ATP6V1B1*	Renal tubular acidosis with deafness (ATP6V1B1-related)	腎小管酸中毒伴失聰(ATP6V1B1相關)
236	*EYS*	Retinitis pigmentosa 25	視網膜色素病變25型
237	*CERKL*	Retinitis pigmentosa 26	視網膜色素病變26型
238	*FAM161A*	Retinitis Pigmentosa 28	視網膜色素病變28型
239	*PEX7*	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	*AGPS*	Rhizomelic chondrodysplasia punctata type 3	肢近端型點狀軟骨發育不良3型

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

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

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