No.	基因	疾病名稱(英文)	疾病名稱(中文)	帶因率	v1.0 (SOFIVA)	v2.0 (Baylor)	v3.0 (Baylor)
1	CFTR	Cystic fibrosis	囊腫纖維症	1 in 25	●	●	●
2	FMR1	Fragile X syndrome	X染色體脆折症	1 in 250	●	◎	◎
3	HBA1	Alpha-thalassemia	甲型海洋性貧血	1 in 25	●	●	●
4	HBA2	Alpha-thalassemia	甲型海洋性貧血	1 in 25	●	●	●
5	HBB	Beta hemoglobinopathies	乙型海洋性貧血	1 in 129	●	●	●
6	SMN1/SMN2	Spinal muscular atrophy	脊髓性肌肉萎縮症	1 in 54	●	●	●
7	ABCC8	Familial hyperinsulinism, ABCC8-related	ABCC8基因相關家族性胰島素過多症	1 in 112		●	●
8	ABCD1	Adrenoleukodystrophy, X-linked	性聯遺傳腎上腺腦白質失養症	1 in 10500		◎	◎
9	ACADM	Medium chain acyl-CoA dehydrogenase deficiency	中鏈醯輔酶A去氫酶缺乏症	1 in 354		●	●
10	ACADVL	Very long-chain acyl-CoA dehydrogenase deficiency	極長鏈醯輔酶A去氫酶缺乏症	1 in 86		●	●
11	ADA	Adenosine deaminase deficiency	腺苷脫氨酶缺乏症	1 in 224		●	●
12	ADAMTS2	Ehlers-Danlos syndrome,dermatosparaxis type	先天性結締組織異常（埃勒斯-當洛斯症候群）	<1 in 500		●	●
13	AGA	Aspartylglycosaminuria	天冬氨酰葡萄糖胺尿症	<1 in 500		●	●
14	AGL	Glycogen storage disease, type III	肝醣儲積症第3型	1 in 159		●	●
15	AGXT	Primary hyperoxaluria, type I	原發性高草酸尿症第1型	1 in 158		●	●
16	ALDH3A2	Sjogren-Larsson syndrome	鳩拉二氏症候群	1 in 223		●	●
17	ALDOB	Hereditary fructose intolerance	遺傳性果糖不耐症	1 in 55		●	●
18	ALPL	Hypophosphatasia	低磷酸酯酶症	1 in 158		●	●
19	AMT	Glycine encephalopathy, AMT-related	AMT基因相關甘氨酸腦病/非酮性高甘胺酸血症	1 in 262		●	●
20	ARSA	Metachromatic leukodystrophy, ARSA-related	ARSA基因相關異染性腦白質退化症	1 in 100		●	●
21	ASL	Argininosuccinic aciduria	精胺丁二酸酵素缺乏症	1 in 133		●	●
22	ASPA	Canavan disease	卡納萬病（海綿狀腦白質營養不良症）	<1 in 500		●	●
23	ASS1	Citrullinemia, type I	瓜胺酸血症第一型	1 in 119		●	●
24	ATM	Ataxia-telangiectasia	共濟失調微血管擴張症候群	1 in 100		●	●
25	ATP7B	Wilson disease	威爾森氏症	1 in 90		●	●
26	BBS1	Bardet-Biedl syndrome 1	巴德-畢德氏症候群第1型	1 in 265		●	●
27	BBS10	Bardet-Biedl syndrome 10	巴德-畢德氏症候群第10型	1 in 447		●	●
28	BBS2	Bardet-Biedl syndrome 2	巴德-畢德氏症候群第2型	<1 in 500		●	●
29	BCKDHA	Maple syrup urine disease, type 1A	楓糖尿症第1A型	1 in 321		●	●
30	BCKDHB	Maple syrup urine disease, type 1B	楓糖尿症第1B型	1 in 364		●	●
31	BCS1L	GRACILE syndrome	GRACILE 症候群	1 in 111		●	●
32	BLM	Bloom syndrome	布盧姆症候群	<1 in 500		●	●
33	BTD	Biotinidase deficiency	生物素酶缺乏症	1 in 109		●	●
34	BTK	X-linked agammaglobulinemia	性聯遺傳無γ球蛋白血症	<1 in 500		◎	◎
35	CAPN3	Limb-girdle muscular dystrophy, type 2A	肢帶型肌肉失養症第2A型	<1 in 500		●	●
36	CBS	Homocystinuria, CBS-related	CBS基因相關高胱胺酸尿症	<1 in 500		●	●
37	CDH23	Usher syndrome, type 1D	尤塞氏症候群第1D型	1 in 202		●	●
38	CHAT	Congenital myasthenic syndrome, CHAT-related	CHAT基因相關先天性肌無力症候群	<1 in 500		●	●
39	CHRNE	Congenital myasthenic syndrome, CHRNE-related	CHRNE基因相關先天性肌無力症候群	<1 in 500		●	●
40	CLN3	Neuronal ceroid lipofuscinosis, CLN3-related	CLN3基因相關-神經元蠟樣脂褐質儲積症	1 in 145		●	●
41	CLN5	Neuronal ceroid lipofuscinosis, CLN5-related	CLN5基因相關-神經元蠟樣脂褐質儲積症	1 in 317		●	●
42	CLN6	Neuronal ceroid lipofuscinosis, CLN6-related	CLN6基因相關-神經元蠟樣脂褐質儲積症	1 in 261		●	●
43	CLN8	Neuronal ceroid lipofuscinosis, CLN8-related	CLN8基因相關-神經元蠟樣脂褐質儲積症	1 in 349		●	●
44	CLRN1	Usher syndrome, type 3A	尤塞氏症候群第3A型	<1 in 500		●	●
45	COL4A3	Alport syndrome, COL4A3-related	COL4A3基因相關亞伯氏症候群	1 in 323		●	●
46	CPT1A	Carnitine palmitoyltransferase I deficiency	肉鹼棕櫚醯基轉移酶缺乏第一型	<1 in 500		●	●
47	CPT2	Carnitine palmitoyltransferase II deficiency	肉鹼棕櫚醯基轉移酶缺乏症第2型	<1 in 500		●	●
48	CTNS	Cystinosis	胱胺酸症	1 in 158		●	●
49	CTSK	Pycnodysostosis	緻密性成骨不全症	1 in 439		●	●
50	CYBB	Chronic granulomatous disease, X-linked	性聯遺傳慢性肉芽腫疾病	1 in 150000		◎	◎
51	CYP1B1	Primary congenital glaucoma	原發性嬰幼兒型青光眼	1 in 74		●	●
52	CYP21A2	Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency	21-羥化酶缺乏引起之先天性腎上腺增生症	1 in 61		●	●
53	CYP27A1	Cerebrotendinous xanthomatosis	腦腱性黃瘤症	1 in 115		●	●
54	DBT	Maple syrup urine disease, type 2	楓糖尿症第2型	1 in 321		●	●
55	DCLRE1C	Omenn syndrome	歐門氏症候群	<1 in 500		●	●
56	DHCR7	Smith-Lemli-Opitz syndrome	Smith-Lemli-Opitz 症候群	1 in 100		●	●
57	DHDDS	Retinitis pigmentosa 59	視網膜色素病變第59型	<1 in 500		●	●
58	DLD	Dihydrolipoamide dehydrogenase deficiency	二氫硫辛醯胺脫氫酶缺乏症	<1 in 500		●	●
59	DMD	Duchenne/Becker muscular dystrophy, X-linked	性聯遺傳杜興氏 / 貝克氏肌肉失養症	1 in 4200		◎	◎
60	DOK7	Congenital myasthenic syndrome, DOK7-related	DOK7相關肌無力症候群	1 in 454		●	●
61	DPYD	Dihydropyrimidine dehydrogenase deficiency	二氫嘧啶脫氫酶缺乏症	1 in 20		●	●
62	EIF2B5	Leukoencephalopathy with vanishing white matter 5	腦白質病伴隨白質消失症第5型	<1 in 500		●	●
63	ELP1	Familial dysautonomia	家族性自主神經失調症	<1 in 500		●	●
64	ETHE1	Ethylmalonic encephalopathy	乙基丙二酸腦病變	<1 in 500		●	●
65	FAH	Tyrosinemia, type I	酪胺酸血症第1型	1 in 100		●	●
66	FANCC	Fanconi anemia, complementation group C	范可尼氏貧血症C型	<1 in 500		●	●
67	FH	Fumarase deficiency	延胡索酸酶缺乏症	<1 in 500		●	●
68	FKTN	Fukuyama congenital muscular dystrophy	福山型先天性肌肉失養症	<1 in 500		●	●

No.	基因	疾病名稱(英文)	疾病名稱(中文)	帶因率	v1.0 (SOFIVA)	v2.0 (Baylor)	v3.0 (Baylor)
69	G6PC1	Glycogen storage disease, type Ia	肝醣儲積症第1型	1 in 177		●	●
70	G6PD	Glucose-6-phosphate dehydrogenase deficiency	葡萄糖-六-磷酸鹽去氫酵素缺乏症 (蠶豆症)	1 in 30		◎	◎
71	GAA	Glycogen storage disease, type II / Pompe disease	肝醣儲積症第2型(龐貝氏症)	1 in 132		●	●
72	GALC	Krabbe disease	Krabbe氏症（球細胞腦白質失養症）	1 in 150		●	●
73	GALT	Galactosemia	半乳糖血症	1 in 110		●	●
74	GBA	Gaucher disease	高雪氏症	1 in 153		●	●
75	GCDH	Glutaric acidemia, type I	戊二酸血症第1型	1 in 112		●	●
76	GJB2	Nonsyndromic hearing loss and deafness (DFNB) 1	非症候群型遺傳性聽損第1型	1 in 42		●	●
77	GLB1	GLB1-related disorders	GLB1基因相關疾病	1 in 158		●	●
78	GLDC	Glycine encephalopathy / Nonketotic hyperglycinemia	甘氨酸腦病/非酮性高甘胺酸血症	1 in 135		●	●
79	GNE	GNE myopathy	GNE肌病變	<1 in 500		●	●
80	GNPTAB	Mucolipidosis II and mucolipidosis III alpha/ beta	黏脂質症第2型和第3型	1 in 158		●	●
81	GRHPR	Primary hyperoxaluria, type II	原發性高草酸尿症第2型	<1 in 500		●	●
82	HADHA	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	長鏈3-羥酰基輔酶A脫氫酶缺乏症	1 in 138		●	●
83	HEXA	Tay-Sachs disease	戴薩克斯症（家族性黑矇癡呆症）	1 in 250		●	●
84	HEXB	Sandhoff disease	山德霍夫症	1 in 278		●	●
85	HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	3-羥基-3-甲基戊二酸尿症	<1 in 500		●	●
86	HPS3	Hermansky-Pudlak syndrome, type 3	Hermansky-Pudlak氏症候群第3型	<1 in 500		●	●
87	HSD17B4	D-bifunctional protein deficiency	D型雙功能蛋白缺乏症	1 in 158		●	●
88	IDUA	Mucopolysaccharidosis, type I / Hurler syndrome	黏多醣症第1型 (賀勒氏症)	1 in 158		●	●
89	IL2RG	Severe combined immunodeficiency, X-linked	性聯遺傳遺傳嚴重免疫缺陷	1 in 69000		◎	◎
90	IVD	Isovaleric acidemia	異戊酸血症	1 in 250		●	●
91	LAMA3	Junctional epidermolysis bullosa, LAMA3-related	LAMA3基因相關接合型表皮溶解水皰症	<1 in 500		●	●
92	LAMB3	Junctional epidermolysis bullosa, LAMB3-related	LAMB3基因相關接合型表皮溶解水皰症	1 in 407		●	●
93	LAMC2	Junctional epidermolysis bullosa, LAMC2-related	LAMC2基因相關接合型表皮溶解水皰症	<1 in 500		●	●
94	LRPPRC	Mitochondrial complex IV deficiency, nuclear type 5 / Leigh syndrome, French-Canadian type	第5型核基因粒線體複合體IV缺乏症/法國-加拿大型亞急性壞死性腦脊髓病	<1 in 500		●	●
95	MAN2B1	Alpha-mannosidosis	α型甘露糖症	<1 in 500		●	●
96	MCOLN1	Mucolipidosis IV	黏脂質症第4型	<1 in 500		●	●
97	MLC1	Megalencephalic leukoencephalopathy with subcortical cysts	腦白質病伴隨皮層下囊腫	<1 in 500		●	●
98	MMACHC	Combined methylmalonic aciduria and homocystinuria, cblC type / Cobalamin C deficiency	合併型甲基丙二酸尿症與高胱胺酸尿症cblC型/鈷胺素C缺乏症	1 in 138		●	●
99	MPI	Congenital disorder of glycosylation, type Ib	先天性醣基化障礙第1B型	<1 in 500		●	●
100	MPL	Congenital amegakaryocytic thrombocytopenia	先天性巨核細胞缺乏血小板低下症	1 in 415		●	●
101	MTTP	Abetalipoproteinemia	無β脂蛋白血症	<1 in 500		●	●
102	MYO7A	Usher syndrome, type 1B	尤塞氏症候群第1B型	1 in 206		●	●
103	NBN	Nijmegen breakage syndrome	奈梅亨破損症候群	<1 in 500		●	●
104	NEB	Nemaline myopathy 2	線狀體肌肉病變第2型	1 in 224		●	●
105	NPC1	Niemann-Pick disease, type C1	尼曼匹克症C1型	1 in 282		●	●
106	NPHP1	NPHP1 nephronophthisis-related ciliopathies	NPHP1基因相關腎元發育不全相關纖毛病	1 in 202		●	●
107	NPHS1	Steroid resistant nephrotic syndrome, type 1	類固醇抗藥型腎病症候群第1型	1 in 377		●	●
108	NPHS2	Steroid-resistant nephrotic syndrome, type 2	類固醇抗藥型腎病症候群第2型	<1 in 500		●	●
109	OCRL	Lowe syndrome, X-linked	性聯遺傳Lowe症候群	1 in 375000		◎	◎
110	OTC	Ornithine transcarbamylase deficiency, X-linked	性聯遺傳鳥胺酸氨甲醯基轉移酶缺乏症	1 in 30000		◎	◎
111	PAH	Phenylalanine hydroxylase deficiency	苯丙氨酸羥化酶缺乏症	1 in 65		●	●
112	PC	Pyruvate carboxylase deficiency	丙酮酸酸羧化酶缺乏症	1 in 250		●	●
113	PCCA	Propionic acidemia, PCCA-related	PCCA基因相關丙酸血症	1 in 224		●	●
114	PCCB	Propionic acidemia, PCCB-related	PCCB基因相關丙酸血症	1 in 224		●	●
115	PCDH15	Usher syndrome, type 1F	尤塞氏症候群第1F型	1 in 395		●	●
116	PEX1	Zellweger spectrum disorders, PEX1-related	PEX1基因相關趙華格氏症	<1 in 500		●	●
117	PEX2	Zellweger spectrum disorders, PEX2-related	PEX2基因相關Zellweger氏症候群	<1 in 500		●	●
118	PEX7	Rhizomelic chondrodysplasia punctata, type 1	肢端型點狀軟骨發育不良第1型	<1 in 500		●	●
119	PHGDH	Phosphoglycerate dehydrogenase deficiency	磷酸甘油酸脫氫酶缺乏症	<1 in 500		●	●
120	PKHD1	Autosomal recessive polycystic kidney disease	胎兒型體染色體隱性遺傳多囊性腎臟病	1 in 144		●	●
121	PLA2G6	PLA2G6-associated neurodegeneration	PLA2G6基因相關神經退化性疾病	<1 in 500		●	●
122	PMM2	Congenital disorder of glycosylation, type Ia	先天性醣基化障礙第1A型	1 in 124		●	●
123	POLG	POLG-related disorders	POLG基因相關疾病	1 in 50		●	●
124	POMGNT1	Muscular dystrophy-dystroglycanopathy, type A, 3	先天性肌肉失養症伴隨醣基化功能缺陷A3型	1 in 462		●	●
125	PPT1	Neuronal ceroid lipofuscinosis, PPT1-related	PPT1基因相關神經元蠟樣脂褐質儲積症	1 in 368		●	●
126	PROP1	Combined pituitary hormone deficiency, type 2	複合性腦下垂體激素缺乏症第2型	1 in 141		●	●
127	PTS	6-pyruvoyl-tetrahydropterin synthase deficiency	6-丙酮醯四氫生物蝶呤合成酶缺乏症	<1 in 500		●	●
128	RAPSN	Congenital myasthenic syndrome, RAPSN-related	RAPSN基因相關先天性肌無力症候群	1 in 252		●	●
129	RMRP	Cartilage-hair hypoplasia	軟骨 - 毛髮發育不良症	<1 in 500		●	●
130	RTEL1	Dyskeratosis congenita, RTEL1-related	RTEL1基因相關先天性角化不全症	<1 in 500		●	●
131	SACS	Spastic ataxia, Charlevoix-Saguenay type	痙攣性共濟失調症（查爾瓦克斯-薩格奈型）	<1 in 500		●	●
132	SERPINA1	Alpha-1 antitrypsin deficiency	α1-抗胰蛋白酶缺乏症	1 in 38		●	●

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133	SGCA	Limb-girdle muscular dystrophy, type 3	肢帶型肌肉失養症第3型	<1 in 500		●	●
134	SGCB	Limb-girdle muscular dystrophy, type 4	肢帶型肌肉失養症第4型	<1 in 500		●	●
135	SGCG	Limb-girdle muscular dystrophy, type 5	肢帶型肌肉失養症第5型	1 in 354		●	●
136	SGSH	Mucopolysaccharidosis, type IIIA / Sanfilippo syndrome A	黏多醣症第三A型/聖菲利柏氏症A型	1 in 415		●	●
137	SLC12A6	Agenesis of the corpus callosum with peripheral neuropathy	胼胝體發育不全伴周邊神經病變	<1 in 500		●	●
138	SLC17A5	Free sialic acid storage disorders	游離唾液酸儲積症	<1 in 500		●	●
139	SLC22A5	Carnitine deficiency, systemic primary	原發性肉鹼缺乏症	1 in 200		●	●
140	SLC25A13	Citrin deficiency / Citrullinemia, type II	Citrin缺乏症/瓜胺酸血症第2型	<1 in 500		●	●
141	SLC25A15	Ornithine translocase deficiency	鳥胺酸轉位酶缺乏症	<1 in 500		●	●
142	SLC26A2	Skeletal dysplasia, SLC26A2-related	SLC26A2基因相關骨骼發育異常	1 in 158		●	●
143	SLC26A4	Pendred syndrome	Pendred症候群	1 in 80		●	●
144	SLC35A3	Arthrogryposis, mental retardation, and seizures	關節彎曲,智能發展遲緩及癲癇發作	<1 in 500		●	●
145	SLC37A4	Glycogen storage disease, type Ib / IIw	肝醣儲積症第Ib/Iiw型	1 in 354		●	●
146	SLC7A7	Lysinuric protein intolerance	賴胺酸尿症蛋白不耐症	<1 in 500		●	●
147	SMPD1	Niemann-Pick disease, types A/B	尼曼匹克症A/B型	1 in 196		●	●
148	SUMF1	Multiple sulfatase deficiency	多發性硫酸脂酶缺乏症	<1 in 500		●	●
149	TGM1	Autosomal recessive congenital ichthyosis	體染色體隱性遺傳魚鱗癬	1 in 301		●	●
150	TH	Tyrosine hydroxylase deficiency	酪胺酸羥化酶缺乏症	<1 in 500		●	●
151	TMEM216	Joubert syndrome 2	Joubert 氏症候群第2型（家族性小腦蚓部發育不全）	<1 in 500		●	●
152	TPP1	Neuronal ceroid lipofuscinosis, TPP1-related	TPP1基因相關-神經元蠟樣脂褐質儲積症第2型	1 in 314		●	●
153	TTPA	Ataxia with isolated vitamin E deficiency	共濟失調合併維生素E缺乏症	<1 in 500		●	●
154	USH1C	Usher syndrome, type 1C	尤塞氏症候群第1C型	1 in 353		●	●
155	USH2A	Usher syndrome, type 2A	尤塞氏綜合症第2A型	1 in 126		●	●
156	WAS	Wiskott-Aldrich syndrome, X-linked	性聯遺傳Wiskott–Aldrich症候群	1 in 187500		◎	◎
157	AAAS	Triple A syndrome	Triple A症候群	<1 in 500			●
158	ABAT	GABA-transaminase deficiency	GABA轉胺酶缺乏症	<1 in 500			●
159	ABCA12	Congenital ichthyosis, ABCA12-related	ABCA12基因相關先天性魚鱗癬	<1 in 500			●
160	ABCA3	Surfactant dysfunction, ABCA3-related	ABCA3基因相關肺表面活性劑代謝異常症	1 in 116			●
161	ABCA4	ABCA4-related disorders	ABCC4基因相關疾病	1 in 45			●
162	ABCB11	Progressive familial intrahepatic cholestasis 2	進行性家族性肝內膽汁滯留症第2型	1 in 158			●
163	ABCB4	Progressive familial intrahepatic cholestasis 3	進行性家族性肝內膽汁滯留症第3型	1 in 194			●
164	ABCC2	Dubin-Johnson syndrome	杜賓 - 強森症候群	1 in 158			●
165	ABCC6	Pseudoxanthoma elasticum	彈性纖維假黃瘤	1 in 79			●
166	ACAD9	Mitochondrial complex I deficiency,ACAD9-related	ACAD9相關粒線體複合物I缺乏症	<1 in 500			●
167	ACAT1	Beta-ketothiolase deficiency	β-酮硫解酶缺乏症	1 in 347			●
168	ACOX1	Peroxisomal acyl-CoA oxidase deficiency	過氧化體醯輔酶A氧化酶缺乏症	<1 in 500			●
169	ACSF3	Combined malonic and methylmalonic aciduria	丙二酸及甲基丙二酸綜合酸血症	1 in 86			●
170	ADAMTSL4	ADAMTSL4-related eye disorders	ADAMTSL4基因相關眼部疾病	<1 in 500			●
171	ADGRG1	Bilateral frontoparietal polymicrogyria	雙側額頂葉多小腦迴症	<1 in 500			●
172	ADGRV1	Usher syndrome, type 2C	尤塞氏綜合症第2C型	1 in 176			●
173	AFF2	Fragile XE syndrome	X染色體脆折症E型	1 in 18750			◎
174	AGPS	Rhizomelic chondrodysplasia punctata, type 3	近端型點狀軟骨發育不良第3型	<1 in 500			●
175	AHI1	Joubert syndrome 3	Joubert 氏症候群第3型	1 in 176			●
176	AIFM1	Combined oxidative phosphorylation deficiency 6	氧化磷酸化缺失症第6型	<1 in 500			◎
177	AIPL1	Leber congenital amaurosis 4	萊伯氏先天性黑矇症第4型	1 in 387			●
178	AIRE	Autoimmune polyglandular syndrome, type 1	自體免疫多腺體症候群第1型	1 in 354			●
179	ALDH7A1	Pyridoxine-dependent epilepsy	吡哆醇依賴性癲癇症	1 in 158			●
180	ALG1	Congenital disorder of glycosylation, type Ik	先天糖基化疾病第Ik型	<1 in 500			●
181	ALG13	Developmental and epileptic encephalopathy 36	發展性與癲癇性腦病變第36型	<1 in 750000			◎
182	ALG6	Congenital disorder of glycosylation, type Ic	先天糖基化疾病第1C型	<1 in 500			●
183	ALMS1	Alstrom syndrome	阿爾斯特倫症候群	<1 in 500			●
184	AMN	Imerslund-Gräsbeck syndrome 2	伊莫斯隆德-格拉斯貝克症候群 2 型	<1 in 500			●
185	ANO10	Autosomal recessive spinocerebellar ataxia, type 10	體染色體隱性遺傳小腦萎縮症第10型	1 in 93			●
186	AP1S1	MEDNIK syndrome	MEDNIK症候群	<1 in 500			●
187	AP3B1	Hermansky-Pudlak syndrome, type 2	Hermansky-Pudlak症候群第2型	1 in 158			●
188	AQP2	Nephrogenic diabetes insipidus	腎因型尿崩症	<1 in 500			●
189	AR	Androgen insensitivity syndrome	雄激素不敏感症候群	1 in 5000			◎
190	ARG1	Argininemia	精胺酸血症	<1 in 500			●
191	ARL13B	Joubert syndrome 8	oubert 氏症候群第8型	<1 in 500			●
192	ARL6	Bardet-Biedl syndrome 3	巴德-畢德氏症候群第3型	<1 in 500			●
193	ARSB	Mucopolysaccharidosis, type VI /Maroteaux-Lamy syndrome	黏多醣症第6型（馬洛托-拉米氏症）	1 in 291			●
194	ARSL	X-linked chondrodysplasia punctata 1	性聯遺傳點狀軟骨發育不良第1型	<1 in 500			◎
195	ARX	X-linked developmental disorders, ARX-related	ARX基因相關性聯遺傳發育型障礙	1 in 250000			◎
196	ASAH1	Farber lipogranulomatosis	法伯脂肪肉芽腫病	<1 in 500			●
197	ASNS	Asparagine synthetase deficiency	天門冬醯胺合成缺乏症	<1 in 500			●

No.	基因	疾病名稱(英文)	疾病名稱(中文)	帶因率	v1.0 (SOFIVA)	v2.0 (Baylor)	v3.0 (Baylor)
198	ATP6V1B1	Renal tubular acidosis and deafness, ATP6V1B1-related	ATP6V1B1基因相關腎小管性酸中毒合併聽力損失症	<1 in 500			●
199	ATP7A	Menkes disease	Menkes氏症候群	1 in 26000			◎
200	ATP8B1	Progressive familial intrahepatic cholestasis 1 and benign familial intrahepatic cholestasis 1	進行性家族性肝內膽汁滯留症第 1 型與良性復發性肝內膽汁滯留症第 1 型	1 in 53			●
201	ATRX	Alpha-thalassemia intellectual disability syndrome, X-linked	性聯遺傳α地中海型貧血智力障礙症候群	<1 in 750000			◎
202	AVPR2	AVPR2-related disorders	AVPR2基因相關疾病	1 in 63000			◎
203	B9D1	Joubert syndrome 27	Joubert 氏症候群第27型	<1 in 500			●
204	B9D2	Joubert syndrome 34	Joubert 氏症候群第34型	<1 in 500			●
205	BBS12	Bardet-Biedl syndrome 12	巴德-畢德氏症候群第12型	<1 in 500			●
206	BBS4	Bardet-Biedl syndrome 4	巴德-畢德氏症候群第4型	<1 in 500			●
207	BBS5	Bardet-Biedl syndrome 5	巴德-畢德氏症候群第5型	<1 in 500			●
208	BBS7	Bardet-Biedl syndrome 7	巴德-畢德氏症候群第7型	<1 in 500			●
209	BBS9	Bardet-Biedl syndrome 9	巴德-畢德氏症候群第9型	<1 in 500			●
210	BCHE	Pseudocholinesterase deficiency	假性膽鹼酯酶缺乏症	1 in 53			●
211	BLOC1S3	Hermansky-Pudlak syndrome, type 8	Hermansky-Pudlak症候群第8型	<1 in 500			●
212	BLOC1S6	Hermansky-Pudlak syndrome, type 9	Hermansky-Pudlak症候群第9型	<1 in 500			●
213	BMP1	Osteogenesis imperfecta, type XIII	成骨不全症第13型	<1 in 500			●
214	BRIP1	Fanconi anemia, complementation group J	范可尼氏貧血症J型	1 in 500			●
215	BSND	Bartter syndrome, type 4A	Bartter氏症候群第4型	<1 in 500			●
216	C2CD3	Orofaciodigital syndrome, type XIV	口腔 - 顏面 - 指（趾）發育異常症候群第14型	<1 in 500			●
217	CAD	Developmental and epileptic encephalopathy 50	發展性與癲癇性腦病變第50型	<1 in 500			●
218	CANT1	Desbuquois dysplasia, type I	Desbuquois氏發育不全症第1型	<1 in 500			●
219	CASQ2	Catecholaminergic polymorphic ventricular tachycardia, type 2	兒茶酚胺敏感性多形性心室頻脈第2型	1 in 224			●
220	CC2D1A	Autosomal recessive intellectual developmental disorder, type 3	體染色體隱性遺傳智能發展障礙症候群	<1 in 500			●
221	CC2D2A	Joubert syndrome 9	Joubert 氏症候群第9型	<1 in 500			●
222	CCDC103	Primary ciliary dyskinesia, CCDC103-related	CDC103基因相關原發性纖毛運動障礙症	1 in 217			●
223	CCDC39	Primary ciliary dyskinesia, CCDC39-related	CDC39基因相關原發性纖毛運動障礙症	1 in 144			●
224	CCDC88C	Congenital hydrocephalus 1	先天性水腦症第1型	1 in 137			●
225	CCN6	Progressive pseudorheumatoid dysplasia	進行性假性類風濕性骨發育不良症	<1 in 500			●
226	CD3D	Severe combined immunodeficiency, CD3D-related	CD3D基因相關重度複合型免疫缺陷症	<1 in 500			●
227	CD3E	Severe combined immunodeficiency, CD3E-related	CD3E基因相關嚴重複合型免疫缺陷症	<1 in 500			●
228	CD40	Hyper-IgM syndrome, type 3	高免疫球蛋白M症候群第3型	<1 in 500			●
229	CD40LG	X-Linked hyper IgM syndrome	性聯遺傳高免疫球蛋白M症候群	1 in 52500			◎
230	CD59	CD59-mediated hemolytic anemia	CD59 介導性溶血性貧血	<1 in 500			●
231	CEP104	Joubert syndrome 25	Joubert 氏症候群第25型	<1 in 500			●
232	CEP120	Joubert syndrome 31	Joubert 氏症候群第31型	<1 in 500			●
233	CEP152	CEP152-related disorders	CEP152基因相關疾病	<1 in 500			●
234	CEP290	Leber congenital amaurosis, CEP290-related / CEP290-related conditions	CEP290相關萊伯氏先天性黑矇症	1 in 185			●
235	CEP41	Joubert syndrome 15	Joubert 氏症候群第15型	<1 in 500			●
236	CERKL	Retinitis pigmentosa 26	視網膜色素病變第26型	1 in 137			●
237	CHM	Choroideremia, X-linked	性聯遺傳脈絡膜缺失症	1 in 25000			◎
238	CHRNG	Multiple pterygium syndrome, lethal type	致死型多發性翼狀膜症候群	1 in 50			●
239	CIB2	Usher syndrome, type 1J	尤塞氏綜合症第1J型	<1 in 500			●
240	CIITA	Bare lymphocyte syndrome, type II	裸淋巴球症候群第2型	<1 in 500			●
241	CLCN1	Myotonia congenita	先天性肌強直症	1 in 158			●
242	CLCN5	Dent disease	Dent氏病	<1 in 500			◎
243	CNGB3	Achromatopsia, CNGB3-related	CNGB3基因相關色彩感應失能症	1 in 98			●
244	COL11A2	COL11A2-related disorders	COL11A2基因相關疾病	<1 in 500			●
245	COL17A1	Junctional epidermolysis bullosa, COL17A1-related	COL17A1基因相關接合型表皮溶解水皰症	<1 in 500			●
246	COL27A1	Steel syndrome	史迪爾氏症候群	<1 in 500			●
247	COL4A4	Alport syndrome, COL4A4-related	COL4A4基因相關亞伯氏症候群	1 in 353			●
248	COL4A5	Alport syndrome, COL4A5-related, X-linked	COL4A5基因相關性聯遺傳亞伯氏症候群	1 in 47000			◎
249	COL7A1	Dystrophic epidermolysis bullosa, COL7A1-related	COL7A1基因相關表皮溶解水皰症	1 in 370			●
250	COLQ	Congenital myasthenic syndrome, COLQ-related	COLQ基因相關先天性肌無力症候群	1 in 430			●
251	COX15	Mitochondrial complex IV deficiency, nuclear type 6	核基因第 6 型粒線體呼吸鏈複合體 IV 缺乏症	<1 in 500			●
252	CPLANE1	Joubert syndrome 17	Joubert 氏症候群第17型	1 in 423			●
253	CPS1	Carbamoyl phosphate synthetase I deficiency	氨甲醯磷酸合成酶缺失症第1型	<1 in 500			●
254	CRB1	CRB1-related retinal dystrophies	CRB1基因相關視網膜失養症	1 in 112			●
255	CRPPA	Muscular dystrophy-dystroglycanopathy, type A, 7	先天性肌肉失養症伴隨醣基化功能缺陷A7型	1 in 371			●
256	CRTAP	Osteogenesis imperfecta, type VII	成骨不全症第7型	<1 in 500			●
257	CSPP1	Joubert syndrome 21	Joubert 氏症候群第21型	<1 in 500			●
258	CTSA	Galactosialidosis	半乳糖唾液酸儲積症	<1 in 500			●
259	CTSC	CTSC-related disorders	CTSC基因相關疾病	1 in 250			●
260	CTSD	Neuronal ceroid lipofuscinosis, CTSD-related	CTSD基因相關神經元蠟樣脂褐質儲積症	<1 in 500			●

No.	基因	疾病名稱(英文)	疾病名稱(中文)	帶因率	v1.0 (SOFIVA)	v2.0 (Baylor)	v3.0 (Baylor)
261	CYBA	Chronic granulomatous disease 4	慢性肉芽腫疾病第4型	<1 in 500			●
262	CYP11A1	Congenital adrenal insufficiency, CYP11A1-related	CYP11A1基因相關先天性腎上腺功能不足	1 in 114			●
263	CYP11B1	Congenital adrenal hyperplasia (CAH) due to 11-beta-hydroxylase deficiency	11-β-羥化酶缺乏引起之先天性腎上腺增生症	1 in 158			●
264	CYP11B2	Corticosterone methyloxidase deficiency	皮質酮甲基氧化酶缺乏症	<1 in 500			●
265	CYP17A1	Congenital adrenal hyperplasia (CAH) due to 17-alpha-hydroxylase deficiency	17-α-羥化酶缺乏引起之先天性腎上腺增生症	<1 in 500			●
266	CYP19A1	Aromatase deficiency	芳香化酶缺乏症	<1 in 500			●
267	CYP27B1	Vitamin D-dependent rickets, type 1A	維生素D依賴型佝僂症第1A型	1 in 22			●
268	CYP7B1	CYP7B1-related disorders	CYP7B1基因相關疾病	1 in 338			●
269	DCAF17	Woodhouse-Sakati syndrome	Woodhouse–Sakati症候群	<1 in 500			●
270	DCX	DCX-related disorders	DCX基因相關疾病	<1 in 750000			◎
271	DDX11	Warsaw breakage syndrome	Warsaw染色體斷裂症候群	<1 in 500			●
272	DGAT1	DGAT1 deficiency	DGAT1缺乏症	<1 in 500			●
273	DGUOK	Deoxyguanosine kinase deficiency / Mitochondrial DNA depletion syndrome 3	脫氧鳥苷激酶缺乏症/第3型粒線體DNA耗竭症	<1 in 500			●
274	DIS3L2	Perlman syndrome	珀爾曼症候群	<1 in 500			●
275	DKC1	Dyskeratosis congenita, X-linked	性聯遺傳先天性角化不全症	<1 in 750000			◎
276	DLL3	Spondylocostal dysostosis 1	脊椎肋骨發育不全第1型	1 in 289			●
277	DNAH11	Primary ciliary dyskinesia, DNAH11-related	DNAH11基因相關原發性纖毛運動障礙症	1 in 144			●
278	DNAH5	Primary ciliary dyskinesia, DNAH5-related	DNAH5基因相關原發性纖毛運動障礙症	1 in 120			●
279	DNAI1	Primary ciliary dyskinesia, DNAI1-related	DNAI1基因相關原發性纖毛運動障礙症	1 in 182			●
280	DNAI2	Primary ciliary dyskinesia, DNAI2-related	DNAI2基因相關原發性纖毛運動障礙症	<1 in 500			●
281	DNMT3B	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	免疫缺陷 - 著絲粒不穩定 - 面部異常症候群1型	<1 in 500			●
282	DUOX2	Thyroid dyshormonogenesis 6	甲狀腺激素生成障礙第6型	1 in 55			●
283	DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly	伴或不伴多指畸形的短肋胸廓發育不良 3 型	1 in 67			●
284	DYSF	Limb-girdle muscular dystrophy, type 2B	肢帶型肌肉失養症第2B型	1 in 311			●
285	EDA	Hypohidrotic ectodermal dysplasia, X-linked	性聯遺傳少汗性外胚層增生不良症	1 in 3800			◎
286	EIF2AK3	Wolcott-Rallison syndrome	Wolcott-Rallison 症候群	<1 in 500			●
287	EIF2B1	Leukoencephalopathy with vanishing white matter 1	腦白質病伴隨白質消失症第1型	<1 in 500			●
288	EIF2B2	Leukoencephalopathy with vanishing white matter 2	腦白質病伴隨白質消失症第2型	<1 in 500			●
289	EIF2B3	Leukoencephalopathy with vanishing white matter 3	腦白質病伴隨白質消失症第3型	<1 in 500			●
290	EIF2B4	Leukoencephalopathy with vanishing white matter 4	腦白質病伴隨白質消失症第4型	<1 in 500			●
291	EMD	Emery-Dreifuss muscular dystrophy, X-linked	埃勒斯-當洛斯症候群	1 in 250000			◎
292	EPG5	EPG5-related disorder	EPG5基因相關疾病	<1 in 500			●
293	ERCC2	ERCC2-related conditions	ERCC2基因相關病症	<1 in 500			●
294	ERCC6	Cerebrooculofacioskeletal syndrome 1 / Cockayne syndrome, type B	大腦-眼眶-臉部-骨骼異常症候群第1型/柯凱因氏症候群第B型	<1 in 500			●
295	ERCC8	Cockayne syndrome, type A	柯凱因氏症候群第A型	<1 in 500			●
296	ESCO2	Roberts-SC phocomelia syndrome	Roberts-SC型海豹肢症候群	<1 in 500			●
297	ETFA	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIA	多重醯基輔酶A去氫酶缺乏症 / 戊二酸尿症第IIA型	<1 in 500			●
298	ETFB	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIB	多重醯基輔酶A去氫酶缺乏症 / 戊二酸尿症第IIB型	1 in 408			●
299	ETFDH	Multiple acyl-CoA dehydrogenase deficiency / Glutaric aciduria, type IIC	多重醯基輔酶A去氫酶缺乏症 / 戊二酸尿症第IIC型	1 in 250			●
300	EVC	Ellis-van Creveld syndrome	埃利偉氏症候群	1 in 345			●
301	EVC2	Ellis-van Creveld syndrome	埃利偉氏症候群	1 in 122			●
302	EXOSC3	Pontocerebellar hypoplasia, type 1B	腦橋小腦發育不全第 1B 型	<1 in 500			●
303	EYS	Retinitis pigmentosa 25	視網膜色素病變第25型	1 in 129			●
304	F11	Factor XI deficiency / Hemophilia C	第十一凝血因子缺乏症 / C 型血友病	1 in 92			●
305	F2	Factor II deficiency / Prothrombin deficiency	第二凝血因子缺乏症 / 凝血酶原缺乏症	1 in 33			●
306	F5	Factor V deficiency	第五凝血因子缺乏症	1 in 12			●
307	F8	Factory VIII deficiency / Hemophilia A	第八凝血因子缺乏症 / A 型血友病	1 in 20000			◎
308	F9	Factor IX deficiency / Hemophilia B	第九凝血因子缺乏症 / B 型血友病	1 in 10000			◎
309	FAM161A	Retinitis pigmentosa 28	視網膜色素病變第28型	1 in 289			●
310	FANCA	Fanconi anemia, complementation group A	范可尼氏貧血症A型	1 in 345			●
311	FANCB	Fanconi anemia, complementation group B	范可尼氏貧血症B型	<1 in 750000			◎
312	FANCD2	Fanconi anemia, complementation group D2	范可尼氏貧血症D2型	<1 in 500			●
313	FANCE	Fanconi anemia, complementation group E	范可尼氏貧血症E型	<1 in 500			●
314	FANCF	Fanconi anemia, complementation group F	范可尼氏貧血症F型	<1 in 500			●
315	FANCG	Fanconi anemia, complementation group G	范可尼氏貧血症G型	<1 in 500			●
316	FANCI	Fanconi anemia, complementation group I	范可尼氏貧血症I型	<1 in 500			●
317	FANCL	Fanconi anemia, complementation group L	范可尼氏貧血症L型	<1 in 500			●
318	FBP1	Fructose-1,6-bisphosphatase deficiency	果糖-1,6-雙磷酸酶缺乏症	<1 in 500			●
319	FBXO7	Parkinson disease 15	帕金森氏症第15型	<1 in 500			●
320	FHL1	FHL1-related disorders	FHL1基因相關疾病	<1 in 750000			◎
321	FKBP10	Osteogenesis imperfecta, type XI	成骨不全症第 11 型	<1 in 500			●

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322	FKRP	Limb-girdle muscular dystrophy, type 2I / Muscular dystrophy-dystroglycanopathy, type A, 5	肢帶型肌肉失養症第2I型 /先天性肌肉失養症伴隨醣基化功能缺陷A5型	1 in 158			●
323	FMO3	Trimethylaminuria	三甲胺尿症	1 in 139			●
324	FOXN1	Severe combined immunodeficiency, FOXN1-related	FOXN1基因相關嚴重複合型免疫缺陷症	<1 in 500			●
325	FOXP3	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked	性聯遺傳免疫失調 - 多內分泌腺病 - 腸病症候群	<1 in 750000			◎
326	FOXRED1	Mitochondrial complex I deficiency, nuclear type 19	第19型核基因型粒線體呼吸鏈複合體I缺乏症	<1 in 500			●
327	FRAS1	Fraser syndrome, type 1	Fraser症候群第1型	1 in 316			●
328	FREM2	Fraser syndrome, type 2	Fraser症候群第2型	<1 in 500			●
329	FUCA1	Fucosidosis	岩糖酵素缺乏症(儲積症)	<1 in 500			●
330	FXN	Friedreich ataxia	弗里德賴希共濟失調症	1 in 250			●
331	G6PC3	G6PC3 deficiency	G6PC3 缺乏症	<1 in 500			●
332	GALE	Galactosemia, type III / Galactose epimerase deficiency	半乳糖血症第3型/半乳糖異構酶缺乏症	1 in 132			●
333	GALK1	Galactosemia, type II / Galactokinase deficiency	半乳糖激酶缺乏症	1 in 112			●
334	GALNS	Mucopolysaccharidosis, type IVA / Morquio syndrome	黏多醣症第4型/Morquio症候群	1 in 307			●
335	GALNT3	Hyperphosphatemic familial tumoral calcinosis	高磷血症家族性腫瘤性鈣質沉著症	<1 in 500			●
336	GAMT	Guanidinoacetate methyltransferase deficiency	胍基乙酸甲基轉移酶缺乏症	<1 in 500			●
337	GATM	Arginine:glycine amidinotransferase deficiency	精胺酸甘胺酸胺基轉移酶缺乏症	<1 in 500			●
338	GBE1	Glycogen storage disease, type IV / Adult polyglucosan body disease	肝醣儲積症第4型/成人型葡萄糖多聚體病	1 in 387			●
339	GCH1	GCH1-related disorders	GCDH1基因相關疾病	<1 in 500			●
340	GDF5	GDF5-related disorders	GDF5基因相關疾病	<1 in 500			●
341	GFM1	Combined oxidative phosphorylation deficiency 1	結合性氧化磷酸化缺乏症第1型	<1 in 500			●
342	GHR	Laron syndrome	萊倫氏症候群	1 in 167			●
343	GJB1	Charcot-Marie-Tooth disease, type 1X	性聯遺傳進行性腓骨肌萎縮症	1 in 7000			◎
344	GLA	Fabry disease, X-linked	法布瑞氏症	1 in 1500			◎
345	GLE1	Lethal congenital contracture syndrome 1	致死先天性攣縮症候群第1型	<1 in 500			●
346	GNPAT	Rhizomelic chondrodysplasia punctata, type 2	肢近端型點狀軟骨發育不良第2型	<1 in 500			●
347	GNPTG	Mucolipidosis III gamma	黏脂質症第三型 Gamma	<1 in 500			●
348	GNS	Mucopolysaccharidosis, type IIID / Sanfilippo syndrome D	黏多醣症第 IIID 型/聖菲利柏氏症 D 型	<1 in 500			●
349	GORAB	Geroderma osteodysplastica	老年樣皮膚營養不良及骨發育不良	<1 in 500			●
350	GP1BA	Bernard-Soulier syndrome, type A	伯納德 - 蘇里爾症候群第A型	<1 in 500			●
351	GP9	Bernard-Soulier syndrome, type C	伯納德 - 蘇里爾症候群第C型	<1 in 500			●
352	GRIP1	Fraser syndrome, type 3	Fraser症候群第3型	1 in 83			●
353	GSS	Glutathione synthetase deficiency	麩胱甘肽合成酶缺乏症	<1 in 500			●
354	GUCY2D	Leber congenital amaurosis 1	萊伯氏先天性黑矇症第1型	1 in 200			●
355	GUSB	Mucopolysaccharidosis, type VII / Sly syndrome	黏多醣症第7型/Sly症候群	<1 in 500			●
356	HADH	3-hydroxyacyl-CoA dehydrogenase deficiency	3-羥醯基輔酶A去氫酶缺乏症	<1 in 500			●
357	HADHB	Mitochondrial trifunctional protein deficiency, HADHB-related	HADHB基因相關粒線體三功能蛋白缺乏症	1 in 146			●
358	HAMP	Hereditary hemochromatosis, type 2B	遺傳性血鐵沉積症（血色素沉著病）第2B型	<1 in 500			●
359	HAX1	Congenital neutropenia, HAX1-related	HAX1基因相關先天性嗜中性白血球減少症	<1 in 500			●
360	HCFC1	Methylmalonic aciduria and homocystinuria, cblX type	甲基丙二酸血症與同型半胱氨酸尿症 cblX 型	<1 in 750000			◎
361	HGD	Alkaptonuria	黑尿症	1 in 250			●
362	HGSNAT	Mucopolysaccharidosis, type IIIC / Sanfilippo syndrome C	黏多醣症第3C型（聖菲利柏氏症）	1 in 482			●
363	HJV	Hereditary hemochromatosis, type 2	遺傳性血鐵沉積症（血色素沉著病）第2型	<1 in 500			●
364	HLCS	Holocarboxylase synthetase deficiency	多發性羧化酶缺乏症	<1 in 500			●
365	HMOX1	Heme oxygenase 1 deficiency	第一型血基質氧化酶缺乏症	<1 in 500			●
366	HOGA1	Primary hyperoxaluria, type III	原發性高草酸尿症第3型	1 in 309			●
367	HPD	Tyrosinemia, type III	酪胺酸血症第3型	<1 in 500			●
368	HPRT1	HPRT1-related disorders	HPRT1基因相關疾病	1 in 285000			◎
369	HPS1	Hermansky-Pudlak syndrome, type 1	Hermansky-Pudlak氏症候群第1型	<1 in 500			●
370	HPS4	Hermansky-Pudlak syndrome, type 4	Hermansky-Pudlak氏症候群第4型	<1 in 500			●
371	HPS5	Hermansky-Pudlak syndrome, type 5	Hermansky-Pudlak氏症候群第5型	<1 in 500			●
372	HPS6	Hermansky-Pudlak syndrome, type 6	Hermansky-Pudlak氏症候群第6型	<1 in 500			●
373	HSD17B10	HSD10 disease	HSD10 疾病	<1 in 750000			◎
374	HSD17B3	17-beta-hydroxysteroid dehydrogenase deficiency, type III	17-β-羥基類固醇脫氫酶缺乏症第3型	<1 in 500			●
375	HSD3B2	3-beta-hydroxysteroid dehydrogenase deficiency, type II	3-β-羥基類固醇脫氫酶缺乏引起之先天性腎上腺增生第2型	<1 in 500			●
376	HYAL1	Mucopolysaccharidosis, type IX / Hyaluronidase deficiency	黏多醣症第9型/玻尿酸酵素缺乏症	<1 in 500			●
377	HYLS1	Hydrolethalus syndrome	水腦致死性症候群	1 in 455			●
378	IDS	Mucopolysaccharidosis, type II / Hunter syndrome	黏多醣症第2型（韓特氏症）	1 in 60000			◎
379	IGHMBP2	IGHMBP2-related disorders	IGHMBP2基因相關疾病	<1 in 500			●
380	IKBKB	Severe combined immunodeficiency, IKBKB-related	IKBKB基因相關嚴重複合型免疫缺陷症	<1 in 500			●

No.	基因	疾病名稱(英文)	疾病名稱(中文)	帶因率	v1.0 (SOFIVA)	v2.0 (Baylor)	v3.0 (Baylor)
381	IL7R	Severe combined immunodeficiency, IL7R-related	IL7R基因相關嚴重複合型免疫缺陷症	1 in 388			●
382	INPP5E	Joubert syndrome 1	Joubert 氏症候群第1型	1 in 264			●
383	INVS	Nephronophthisis 2	腎單位腎癆第2型	1 in 373			●
384	IQCB1	Senior-Loken syndrome 5	Senior-Løken症候群第5型	<1 in 500			●
385	ITGA6	Junctional epidermolysis bullosa, ITGA6-related	ITGA6基因相關接合型表皮溶解水皰症	1 in 159			●
386	ITGB3	ITGB3-related disorders	ITGB3基因相關疾病	<1 in 500			●
387	ITGB4	Junctional epidermolysis bullosa, ITGB4-related	ITGB4基因相關接合型表皮溶解水皰症	<1 in 500			●
388	JAK3	Severe combined immunodeficiency, JAK3-related	JAK3基因相關嚴重複合型免疫缺陷症	1 in 299			●
389	KCNJ1	Bartter syndrome, type 2	巴特症候群第2型	<1 in 500			●
390	KCNJ11	Familial hyperinsulinism, KCNJ11-related	KCNJ11基因相關家族性高胰島素血症	<1 in 500			●
391	L1CAM	L1 syndrome	L1症候群	1 in 22500			◎
392	LAMA2	LAMA2 muscular dystrophy	LAMA2基因相關肌肉失養症	1 in 87			●
393	LARGE1	Muscular dystrophy-dystroglycanopathy, type A, 6	先天性肌肉失養症伴隨醣基化功能缺陷A6型	<1 in 500			●
394	LCA5	Leber congenital amaurosis 5	萊伯氏先天性黑矇症第5型	<1 in 500			●
395	LDLR	Familial hypercholesterolemia, LDLR-related	LDLR基因相關家族性高膽固醇血症	<1 in 500			●
396	LDLRAP1	Familial hypercholesterolemia, LDLRAP1-related	LDLRAP1基因相關家族性高膽固醇血症	<1 in 500			●
397	LHX3	Combined pituitary hormone deficiency, type 3	結合性腦下垂體激素缺乏症第3型	<1 in 500			●
398	LIFR	Stuve-Wiedemann syndrome	Stuve-Wiedemann 症候群	<1 in 500			●
399	LIG4	LIG4 syndrome	LIG4症候群	<1 in 500			●
400	LIPA	Lysosomal acid lipase deficiency	溶酶體酸性脂肪酶缺乏症	<1 in 500			●
401	LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type	甲基丙二酸尿症併高胱胺酸血症cblF型	<1 in 500			●
402	LOXHD1	Nonsyndromic hearing loss and deafness (DFNB) 77	非症候群型遺傳性聽損第77型	<1 in 500			●
403	LPL	Lipoprotein lipase deficiency	脂蛋白解脂酶缺乏症	<1 in 500			●
404	LRAT	LRAT-related disorders	LRAT基因相關疾病	<1 in 500			●
405	LRP2	Donnai-Barrow syndrome	Donnai–Barrow症候群	1 in 213			●
406	LYST	Chediak-Higashi syndrome	Chediak-Higashi症候群	<1 in 500			●
407	MAK	Retinitis pigmentosa 62	視網膜色素病變第62型	<1 in 500			●
408	MANBA	Beta-mannosidosis	Beta-甘露糖症	<1 in 500			●
409	MCCC1	3-methylcrotonyl-CoA carboxylase 1 deficiency	3-甲基巴豆醯輔酵素羧化酵素缺乏症第1型	1 in 147			●
410	MCCC2	3-methylcrotonyl-CoA carboxylase 2 deficiency	3-甲基巴豆醯輔酵素羧化酵素缺乏症第2型	1 in 120			●
411	MCEE	Methylmalonyl-CoA epimerase deficiency	甲基丙二醯輔酶A表異構酶缺乏症	<1 in 500			●
412	MCPH1	Autosomal recessive primary microcephaly 1	體染色體隱性原發性小頭症第1型	1 in 146			●
413	MECR	MECR-related neurologic disorder	MECR基因相關神經系統疾病	<1 in 500			●
414	MED17	Infantile cerebral and cerebellar atrophy	嬰兒期大腦與小腦萎縮症	<1 in 500			●
415	MEFV	Familial Mediterranean fever	家族性地中海熱	1 in 115			●
416	MESP2	Spondylothoracic dysostosis and spondylocostal dysostosis 2	脊椎胸廓發育不良與脊椎肋骨發育不全症第2型	1 in 224			●
417	MFSD8	Neuronal ceroid lipofuscinosis, MFSD8-related	MFSD8基因相關神經元蠟樣脂褐質儲積症	<1 in 500			●
418	MID1	X-linked Opitz G/BBB syndrome	性聯遺傳Opitz G/BBB症候群	1 in 500000			◎
419	MKKS	Bardet-Biedl syndrome 6	巴爾代 - 畢德症候群第6型	1 in 219			●
420	MKS1	MKS1-related disorders	MKS1基因相關疾病	1 in 260			●
421	MLYCD	Malonyl-CoA decarboxylase deficiency	丙二醯輔酶A脫羧酶缺乏症	<1 in 500			●
422	MMAA	Methylmalonic aciduria, MMAA-related	MMAA基因相關甲基丙二酸血症	1 in 316			●
423	MMAB	Methylmalonic aciduria, MMAB-related	MMAB基因相關甲基丙二酸血症	1 in 456			●
424	MMADHC	Combined methylmalonic aciduria and homocystinuria, cblD type / Cobalamin D deficiency	合併型甲基丙二酸尿症與高胱胺酸尿症cblD型/鈷胺素D缺乏症	<1 in 500			●
425	MMUT	Methylmalonic aciduria, MMUT-related	MMUT基因相關甲基丙二酸血症	1 in 383			●
426	MOCS1	Molybdenum cofactor deficiency of complementation group A	鉬輔酶缺乏症A型	<1 in 500			●
427	MOCS2	Molybdenum cofactor deficiency of complementation group B	鉬輔酶缺乏症B型	1 in 236			●
428	MPV17	MPV17-related mitochondrial DNA (mtDNA) maintenance defect	粒線體DNA缺乏症候群第6型（肝腦病變型）	<1 in 500			●
429	MRE11	Ataxia-telangiectasia-like disorder 1	類共濟失調性微血管擴張症	<1 in 500			●
430	MTHFR	Homocystinuria caused by methylenetetrahydrofolate reductase (MTHFR) deficiency	亞甲基四氫葉酸還原酶（MTHFR）缺乏症	1 in 158			●
431	MTM1	X-linked myotubular myopathy	性聯遺傳肌小管肌病變	<1 in 750000			◎
432	MTR	Homocystinuria-megaloblastic anemia, cblG type	高胱胺酸尿症合併巨芽細胞貧血cblG型	1 in 224			●
433	MTRR	Homocystinuria, type cblE	高胱胺酸尿症cblE型	<1 in 500			●
434	MUSK	Congenital myasthenic syndrome, MUSK-related	MUSK相關先天性肌無力症候群	<1 in 500			●
435	MVK	Mevalonic aciduria / Hyper-IgD syndrome	甲羥戊酸激酶缺乏症/高IgD症候群	1 in 167			●
436	MYO15A	Nonsyndromic hearing loss and deafness (DFNB) 3	非症候群型遺傳性聽損第3型	1 in 117			●
437	NAGA	Schindler disease	Schindler疾病	1 in 94			●
438	NAGLU	Mucopolysaccharidosis, type IIIB / Sanfilippo syndrome B	黏多醣症第IIIB型/Sanfilippo症候群B型	<1 in 500			●
439	NAGS	N-acetylglutamate synthase deficiency	N-乙醯穀胺酸合成酶缺乏症	<1 in 500			●
440	NCF2	Chronic granulomatous disease 2	慢性肉芽腫症第2型	1 in 500			●
441	NDRG1	Charcot-Marie-Tooth disease, type 4D	進行性腓骨肌萎縮症第4D型	<1 in 500			●
442	NDUFAF2	Mitochondrial complex I deficiency,nuclear type 10	第10型核基因型粒線體複合體I缺乏症	<1 in 500			●
443	NDUFAF5	Mitochondrial complex I deficiency,nuclear type 16	第16型核基因型粒線體複合體I缺乏症	<1 in 500			●

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444	NDUFAF6	Mitochondrial complex I deficiency,nuclear type 17	第17型核基因型粒線體複合體I缺乏症	<1 in 500			●
445	NDUFS4	Mitochondrial complex I deficiency,nuclear type 1	第1型核基因型粒線體複合體I缺乏症	1 in 423			●
446	NDUFS6	Mitochondrial complex I deficiency,nuclear type 9	第9型核基因型粒線體複合體I缺乏症	<1 in 500			●
447	NDUFS7	Mitochondrial complex I deficiency,nuclear type 3	第3型核基因型粒線體複合體I缺乏症	<1 in 500			●
448	NDUFV1	Mitochondrial complex I deficiency,nuclear type 4	第4型核基因型粒線體複合體I缺乏症	<1 in 500			●
449	NEU1	Sialidosis	唾液酸儲積症	<1 in 500			●
450	NGLY1	Congenital disorder of deglycosylation, type 1	先天性醣基化障礙第1型	1 in 274			●
451	NPC2	Niemann-Pick disease, type C2	尼曼匹克症C2型	<1 in 500			●
452	NPHP3	NPHP3 nephronophthisis-related ciliopathies	NPHP3基因相關腎元發育不全相關纖毛病	<1 in 500			●
453	NPHP4	NPHP4 nephronophthisis-related ciliopathies	NPHP4基因相關腎元發育不全相關纖毛病	<1 in 500			●
454	NR0B1	X-linked congenital adrenal hypoplasia	性聯遺傳先天性腎上腺發育不良	1 in 52500			◎
455	NR2E3	Enhanced S-cone syndrome	增強型S-圓錐症候群	1 in 204			●
456	NSMCE3	Lung disease, immunodeficiency, and chromosome breakage syndrome (LICS)	肺部疾病合併免疫缺陷與染色體斷裂症候群	<1 in 500			●
457	NTRK1	Congenital insensitivity to pain with anhidrosis	先天性痛不敏感症合併無汗症	<1 in 500			●
458	OAT	Ornithine aminotransferase deficiency	鳥胺酸胺基轉移酶缺乏症	<1 in 500			●
459	OCA2	Oculocutaneous albinism, type II	眼睛皮膚白化症第2型	1 in 76			●
460	OPA3	3-methylglutaconic aciduria, type III / Costeff syndrome	3-甲基戊二酸血症第3型/Costeff 症候群	<1 in 500			●
461	OSTM1	Osteopetrosis, OSTM1-related	OSTM1基因相關骨質石化症	<1 in 500			●
462	OTOA	Nonsyndromic hearing loss and deafness (DFNB) 22	非症候群型遺傳性聽損第22型	<1 in 500			●
463	OTOF	Nonsyndromic hearing loss and deafness (DFNB) 9	非症候群型遺傳性聽損第9型	1 in 96			●
464	P3H1	Osteogenesis imperfecta, type VIII	成骨不全症第 8 型	<1 in 500			●
465	PANK2	Pantothenate kinase-associated neurodegeneration	泛酸鹽激酶關聯之神經退化性疾病	1 in 289			●
466	PCBD1	Pterin-4 alpha-carbinolamine dehydratase (PCD) deficiency	喋呤甲醇胺脫水酶缺乏症	<1 in 500			●
467	PCNT	Microcephalic osteodysplastic primordial dwarfism, type II	原始侏儒症第2型	<1 in 500			●
468	PDHA1	Pyruvate dehydrogenase E1-alpha deficiency	丙酮酸鹽脫氫酶缺乏症E1-alpha型	<1 in 750000			◎
469	PDHB	Pyruvate dehydrogenase E1-beta deficiency	丙酮酸鹽脫氫酶缺乏症E1-beta型	<1 in 500			●
470	PEPD	Prolidase deficiency	脯氨酸肽酶缺乏症	1 in 242			●
471	PET100	Mitochondrial complex IV deficiency, nuclear type 12	第12型核基因粒線體複合體IV缺乏症	1 in 452			●
472	PEX10	Zellweger spectrum disorders, PEX10-related	PEX10基因相關Zellweger氏症候群	1 in 406			●
473	PEX12	Zellweger spectrum disorders, PEX12-related	PEX12基因相關Zellweger氏症候群	<1 in 500			●
474	PEX13	Zellweger spectrum disorder, PEX13-related	PEX13基因相關趙葦格氏症	<1 in 500			●
475	PEX16	Zellweger spectrum disorder, PEX16-related	PEX16基因相關趙葦格氏症	<1 in 500			●
476	PEX26	Zellweger spectrum disorders, PEX26-related	PEX26基因相關趙葦格氏症	<1 in 500			●
477	PEX5	Zellweger spectrum disorder, PEX5-related	PEX5基因相關趙葦格氏症	1 in 408			●
478	PEX6	Zellweger spectrum disorders, PEX6-related	PEX6基因相關Zellweger氏症候群	1 in 280			●
479	PFKM	Glycogen storage disease, type VII	肝醣儲積症第7型	<1 in 500			●
480	PGM3	PGM3-congenital disorder of glycosylation / Immunodeficiency 23	PGM3基因相關醣基化先天異常/免疫缺陷症第23型	<1 in 500			●
481	PHKB	Glycogen storage disease, type IXb	肝醣儲積症Ixb型	<1 in 500			●
482	PHKG2	Glycogen storage disease, type IXc	肝醣儲積症Ixc型	<1 in 500			●
483	PHYH	Refsum disease	Refsum病	<1 in 500			●
484	PIGN	PIGN-related disorders	PIGN基因相關疾病	<1 in 500			●
485	PJVK	Nonsyndromic hearing loss and deafness (DFNB) 59	非症候群型遺傳性聽損第59型	<1 in 500			●
486	PLCE1	Steroid-resistant nephrotic syndrome, type 3	類固醇抗藥型腎病症候群第3型	<1 in 500			●
487	PLEKHG5	PLEKHG5-related disorders	PLEKHG5基因相關疾病	<1 in 500			●
488	PLOD1	PLOD1-related kyphoscoliotic Ehlers-Danlos syndrome	PLOD1基因相關脊柱後側彎型Ehlers-Danlos症候群	1 in 158			●
489	PLP1	PLP1-related disorders	PLP1基因相關疾病	1 in 37500			◎
490	PNPO	Pyridoxamine 5’ -phosphate oxidase deficiency	吡哆胺5'-磷酸氧化酶缺乏症	<1 in 500			●
491	POLH	Xeroderma pigmentosum, variant type (XP-V)	色素性乾皮症變異型（XP-V）	1 in 37500			●
492	POMT1	Muscular dystrophy-dystroglycanopathy, type A, 1	先天性肌肉失養症伴隨醣基化功能缺陷A1型	1 in 275			●
493	POMT2	Muscular dystrophy-dystroglycanopathy, type A, 2	先天性肌肉失養症伴隨醣基化功能缺陷A2型	1 in 465			●
494	POR	Cytochrome P450 oxidoreductase deficiency	細胞色素P450氧化還原酶缺乏症	1 in 370			●
495	POU1F1	Combined or isolated pituitary hormone deficiency, type 1	複合性或單一性腦下垂體激素缺乏症第1型	<1 in 500			●
496	PRCD	Retinitis pigmentosa 36	視網膜色素病變第36型	<1 in 500			●
497	PRDM5	Brittle cornea syndrome 2	角膜脆弱症候群第2型	<1 in 500			●
498	PRF1	Familial hemophagocytic lymphohistiocytosis 2	家族性噬血性淋巴組織細胞增生症第2型	<1 in 500			●
499	PRPS1	PRPS1-related disorders	PRPS1基因相關症狀	1 in 500000			◎
500	PSAP	Metachromatic leukodystrophy due to saposin B deficiency	Saposin B缺乏型異染性腦白質失養症	<1 in 500			●
501	PTPRC	Severe combined immunodeficiency, PTPRC-related	PTPRC基因相關嚴重複合型免疫缺陷症	<1 in 500			●
502	PUS1	Myopathy, lactic acidosis, and sideroblastic anemia	肌病變合併乳酸性酸中毒及鐵粒幼紅血球性貧血	<1 in 500			●
503	PYGM	Glycogen storage disease, type V	肝醣儲積症第5型	1 in 191			●
504	QDPR	Dihydropteridine reductase (DHPR) deficiency	二氫蝶啶還原酶缺乏症	<1 in 500			●
505	RAB23	Carpenter syndrome	卡本特氏症候群	<1 in 500			●

No.	基因	疾病名稱(英文)	疾病名稱(中文)	帶因率	v1.0 (SOFIVA)	v2.0 (Baylor)	v3.0 (Baylor)
506	RAG1	Severe combined immunodeficiency, RAG1-related	RAG1相關嚴重複合型免疫缺陷症	1 in 245			●
507	RAG2	Severe combined immunodeficiency, RAG2-related	RAG2相關嚴重複合型免疫缺陷症	<1 in 500			●
508	RARS2	Pontocerebellar hypoplasia, type 6	橋腦小腦發育不全症第6型	<1 in 500			●
509	RDH12	Leber congenital amaurosis 13	萊伯氏先天性黑矇症第13型	1 in 456			●
510	RLBP1	RLBP1-related retinopathies	RLBP1基因相關視網膜病變	1 in 296			●
511	RNASEH2A	Aicardi-Goutieres syndrome 4	Aicardi–Goutières症候群第4型	<1 in 500			●
512	RNASEH2B	Aicardi-Goutieres syndrome 2	Aicardi–Goutières症候群第2型	<1 in 500			●
513	RNASEH2C	Aicardi-Goutieres syndrome 3	Aicardi-Goutieres症候群第3型	<1 in 500			●
514	RP2	Retinitis pigmentosa 2	視網膜色素病變第2型	1 in 58000			◎
515	RPE65	Leber congenital amaurosis 2	萊伯氏先天性黑矇症第2型	1 in 228			●
516	RPGR	Retinitis pigmentosa 3	視網膜色素病變第3型	1 in 20000			◎
517	RPGRIP1L	Ciliopathies, RPGRIP1L-related	RPGRIP1L基因相關纖毛病	1 in 259			●
518	RS1	Juvenile retinoschisis, X-linked	性聯遺傳先天性視網膜裂損症	1 in 2500			◎
519	RXYLT1	Muscular dystrophy-dystroglycanopathy, type A, 10	先天性肌肉失養症伴隨醣基化功能缺陷A10型	<1 in 500			●
520	RYR1	RYR1-related disorders	RYR1基因相關疾病	1 in 150			●
521	SAMD9	Normophosphatemic familial tumoral calcinosis	正常血磷型家族性腫瘤樣鈣化症	<1 in 500			●
522	SAMHD1	Aicardi-Goutieres syndrome 5	Aicardi-Goutieres症候群第5型	<1 in 500			●
523	SBDS	Shwachman-Diamond syndrome	史黛氏症	1 in 145			●
524	SCARB2	Action myoclonus renal failure syndrome	動作性肌陣攣 - 腎衰竭症候群	<1 in 500			●
525	SCO2	Mitochondrial complex IV deficiency, nuclear type 2	第2型核基因型粒線體複合體 IV缺乏症	1 in 149			●
526	SEC23B	Congenital dyserythropoietic anemia, type II	先天性紅血球生成異常性貧血第二型	<1 in 500			●
527	SEPSECS	Progressive cerebello-cerebral atrophy	進行性小腦大腦萎縮症	<1 in 500			●
528	SGCD	Limb-girdle muscular dystrophy, type 6	肢帶型肌肉失養症第6型	<1 in 500			●
529	SKIC2	Trichohepatoenteric syndrome 2	髮 - 肝 - 腸症候群第2型	<1 in 500			●
530	SLC12A1	Bartter syndrome, type 1	巴特症候群第1型	1 in 360			●
531	SLC12A3	Gitelman syndrome	吉特曼症候群	1 in 100			●
532	SLC19A2	Thiamine-responsive megaloblastic anemia syndrome	硫胺素反應性巨芽細胞貧血症候群	<1 in 500			●
533	SLC19A3	Biotin-thiamine-responsive basal ganglia disease	生物素-硫胺素反應性基底核疾病	1 in 120			●
534	SLC1A4	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly	合併胼胝體變薄與進行性小頭症之痙攣性四肢麻痺症候群	<1 in 500			●
535	SLC25A20	Carnitine-acylcarnitine translocase deficiency	肉鹼醯基肉鹼轉位酶缺乏症	<1 in 500			●
536	SLC26A3	Congenital secretory chloride diarrhea 1	先天性分泌性氯化物腹瀉第1型	<1 in 500			●
537	SLC27A4	Ichthyosis prematurity syndrome	早產兒魚鱗癬症候群	<1 in 500			●
538	SLC38A8	Foveal hypoplasia 2	黃斑部發育不全第2型	<1 in 500			●
539	SLC39A4	Acrodermatitis enteropathica	腸病變性肢端皮膚炎	1 in 354			●
540	SLC45A2	Oculocutaneous albinism, type IV	眼皮膚白化症第4型	1 in 158			●
541	SLC4A11	Corneal dystrophy and perceptive deafness syndrome	角膜失養症和感音性失聰症	<1 in 500			●
542	SLC4A4	Renal tubular acidosis	腎小管性酸中毒	<1 in 500			●
543	SLC5A5	Thyroid dyshormonogenesis 1	甲狀腺激素生成障礙第1型	<1 in 500			●
544	SLC6A8	Creatine transporter defect, SLC6A8-related, X-linked / Cerebral creatine deficiency syndrome	SLC6A8基因相關肌酸轉運缺陷症/大腦肌酸缺乏症候群	1 in 20600			◎
545	SMARCAL1	Schimke immunoosseous dysplasia	chimke免疫骨發育不全症	<1 in 500			●
546	SNAP29	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma (CEDNIK) syndrome	CEDNIK症候群（腦發育不全 - 神經病變 - 魚鱗癬 - 掌蹠角化症候群）	<1 in 500			●
547	SPG11	SPG11-related disorders	SPG11基因相關疾病	1 in 141			●
548	SPR	Sepiapterin reductase deficiency	墨蝶呤還原酶缺乏症	<1 in 500			●
549	SRD5A2	5-alpha-reductase deficiency	5-α還原酶缺乏症	<1 in 500			●
550	ST3GAL5	Amish infantile epilepsy syndrome	Amish 嬰兒癲癇症候群	<1 in 500			●
551	STAR	Lipoid congenital adrenal hyperplasia	先天性脂肪性腎上腺皮質增生症	<1 in 500			●
552	STX11	Familial hemophagocytic lymphohistiocytosis 4	家族性噬血性淋巴組織細胞增生症第4型	1 in 354			●
553	STXBP2	Familial hemophagocytic lymphohistiocytosis 5	家族性噬血性淋巴組織細胞增生症第5型	1 in 296			●
554	SUOX	Isolated sulfite oxidase deficiency	單純性亞硫酸鹽氧化酶缺乏症	<1 in 500			●
555	SURF1	SURF1-related disorders	SURF1基因相關疾病	1 in 316			●
556	SYNE4	Nonsyndromic hearing loss and deafness (DFNB) 76	非症候群型遺傳性聽損第76型	<1 in 500			●
557	TAFAZZIN (formerly TAZ)	Barth syndrome	巴氏症候群	1 in 225000			◎
558	TANGO2	Recurrent metabolic crises with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration	反覆代謝危機合併橫紋肌溶解、心律不整與神經退化	<1 in 500			●
559	TAT	Tyrosinemia, type II	酪胺酸血症第2型	<1 in 500			●
560	TBCD	Early-onset progressive encephalopathy with brain atrophy and thin corpus callosum	早發性進行性腦病變伴隨腦萎縮及胼胝體變薄	<1 in 500			●
561	TBCE	TBCE-related disorders	TBCE基因相關疾病	<1 in 500			●
562	TCIRG1	Osteopetrosis, infantile malignant, TCIRG1-related	TCIRG1基因相關嬰兒型惡性骨質石化症	1 in 316			●
563	TCN2	Transcobalamin II deficiency	轉鈷胺素 II 缺乏症	<1 in 500			●
564	TECPR2	TECPR2-related hereditary sensory and autonomic neuropathy with intellectual disability	TECPR2基因相關遺傳性感覺與自主神經病變合併智能障礙	<1 in 500			●
565	TERT	Dyskeratosis congenita spectrum disorders	先天性角化不良症譜系疾病	<1 in 500			●
566	TF	Atransferrinemia	無鐵傳遞蛋白質貧血	1 in 116			●
567	TFR2	Hereditary hemochromatosis, type 3	遺傳性血鐵沉積症（血色素沉著病）第3型	<1 in 500			●
568	TG	Thyroid dyshormonogenesis 3	甲狀腺激素生成障礙第3型	1 in 217			●

No.	基因	疾病名稱(英文)	疾病名稱(中文)	帶因率	v1.0 (SOFIVA)	v2.0 (Baylor)	v3.0 (Baylor)
569	TJP2	Progressive familial intrahepatic cholestasis 4	進行性家族性肝內膽汁滯留症第4型	<1 in 500			●
570	TK2	TK2-related mitochondrial disorders	TK2基因相關粒線體疾病	<1 in 500			●
571	TMC1	Nonsyndromic hearing loss and deafness (DFNB) 7	非症候群型遺傳性聽損第7型	1 in 96			●
572	TMEM67	TMEM67-related disorders	TMEM67基因相關疾病	1 in 216			●
573	TMPRSS3	Nonsyndromic hearing loss and deafness (DFNB) 8	非症候群型遺傳性聽損第8型	1 in 96			●
574	TNXB	TNXB-related classical-like Ehlers-Danlos syndrome	TNXB基因相關類典型Ehlers-Danlos症候群	1 in 28			●
575	TPO	Thyroid dyshormonogenesis 2A	甲狀腺激素生成障礙第2A型	1 in 227			●
576	TREX1	TREX1-related disorders	TREX1基因相關疾病	<1 in 500			●
577	TRIM32	TRIM32-related disorders	TRIM32基因相關疾病	<1 in 500			●
578	TRIM37	Mulibrey nanism	侏儒症合併肌肉、肝、腦、眼異常	<1 in 500			●
579	TRMU	Acute infantile liver failure	嬰兒急性肝衰竭	<1 in 500			●
580	TSEN2	Pontocerebellar hypoplasia, type 2B	橋腦小腦發育不全症第2B型	<1 in 500			●
581	TSEN54	Pontocerebellar hypoplasia, types 4 and 2A	橋腦小腦發育不全症第2A型和第4型	<1 in 500			●
582	TSFM	Combined oxidative phosphorylation deficiency 3	結合性氧化磷酸化缺乏症第3型	<1 in 500			●
583	TSHB	Congenital hypothyroidism, TSHB-related	TSHB基因相關先天性甲狀腺功能低下	<1 in 500			●
584	TSHR	Congenital hypothyroidism, TSHR-related	TSHR基因相關先天性甲狀腺機能低下症	1 in 322			●
585	TTC37	Trichohepatoenteric syndrome 1	毛髮 - 肝臟 - 腸道症候群第1型	1 in 381			●
586	TTC8	Bardet-Biedl syndrome 8	巴德-畢德氏症候群第8型	<1 in 500			●
587	TULP1	TULP1-related disorders	TULP1基因相關疾病	1 in 186			●
588	TYMP	Mitochondrial DNA depletion syndrome 1, MNGIE type	第1型粒線體DNA耗竭症候群（MNGIE型）	<1 in 500			●
589	TYR	Oculocutaneous albinism, type I	眼睛皮膚白化症第1型	1 in 20			●
590	TYRP1	Oculocutaneous albinism, type III	眼睛皮膚白化症第3型	1 in 488			●
591	UBA1	X-linked infantile spinal muscular atrophy	性聯遺傳嬰兒型脊髓性肌肉萎縮症	<1 in 750000			◎
592	UBR1	Johanson-Blizzard syndrome	Johanson–Blizzard症候群	1 in 250			●
593	UNC13D	Familial hemophagocytic lymphohistiocytosis 3	家族性噬血性淋巴組織細胞增生症第3型	<1 in 500			●
594	UPB1	Beta-ureidopropionase deficiency	β-尿基丙酸酶缺乏症	<1 in 500			●
595	VDR	Vitamin D-resistant rickets, type 2A	維生素D依賴性佝僂病第2A型	<1 in 500			●
596	VLDLR	VLDLR-associated cerebellar hypoplasia	VLDLR基因相關小腦發育不全症	<1 in 500			●
597	VPS11	Hypomyelinating leukodystrophy 12	低髓鞘腦白質失養症第12型	<1 in 500			●
598	VPS13A	Choreoacanthocytosis	舞蹈症-棘狀紅細胞增多症	<1 in 500			●
599	VPS13B	Cohen syndrome	科恩症候群	<1 in 500			●
600	VPS45	Severe congenital neutropenia 5	嚴重先天性嗜中性白血球低下症第5型	<1 in 500			●
601	VPS53	Pontocerebellar hypoplasia, type 2E	橋腦小腦發育不全症第2E型	<1 in 500			●
602	VRK1	Pontocerebellar hypoplasia, type 1A	小腦發育不全症第1A 型	<1 in 500			●
603	VSX2	Microphthalmia / Anophthalmia	先天性小眼球症/無眼球症	<1 in 500			●
604	WNT10A	Odonto-onycho-dermal dysplasia / Schopf-Schulz-Passarge syndrome	牙 - 指甲 - 皮膚發育不良症候群 / Schöpf–Schulz–Passarge症候群	1 in 305			●
605	WRN	Werner syndrome	Werner症候群	1 in 224			●
606	WWOX	WWOX deficiency	WWOX缺乏症	<1 in 500			●
607	XPA	Xeroderma pigmentosum, group A	色素性乾皮症A型	<1 in 750000			●
608	XPC	Xeroderma pigmentosum, group C	著色性乾皮症C組	1 in 38000			●
609	ZBTB24	Immunodeficiency-centromeric instability-facial anomalies syndrome 2	免疫缺陷 - 著絲點不穩定 - 顏面異常症候群第2型	<1 in 500			●
610	ZFYVE26	Spastic paraplegia, type 15	痙攣性下半身麻痺第15型	<1 in 500			●
611	ZIC3	X-linked heterotaxy-1	性聯遺傳內臟異位症第1型	1 in 375000			◎
612	ZNF469	Brittle cornea syndrome 1	角膜脆弱症候群第1型	<1 in 500			●

◎：男性版本無檢測X染色體性聯遺傳(XL)疾病。由於帶因篩檢目的為找出「本身無症狀、但可能將隱性致病基因遺傳給下一代」的健康父母」，男性若有相關臨床症狀應尋求專科醫師評估進行「診斷式檢測」。