單基因疾病列表

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
1	A2ML1	Noonan-like syndrome 類努南氏症候群	AD
2	ABCC8	Congenital hyperinsulinism 先天性高胰島素血症 Infantile diabetes mellitus 嬰兒型糖尿病	AD/AR
3	ABCD1	Adrenoleukodystrophy, ALD 腎上腺腦白質失養症	XL
4	ACAD8	Isobutyryl-CoA dehydrogenase deficiency, IBDD 異丁醯輔酶A去氫酶缺乏症	AR
5	ACAD9	Mitochondrial complex I deficiency 粒線體複合物I缺乏症	AR
6	ACADM	Medium-chain acyl-CoA dehydrogenase deficiency, MCAD deficiency 中鏈醯輔酶A去氫酶缺乏症	AR
7	ACADS	Short-chain acyl-CoA dehydrogenase deficiency, SCAD deficiency 短鏈醯輔酶A去氫酶缺乏症	AR
8	ACADSB	2-Methylbutyryl-CoA dehydrogenase deficiency 2-甲基丁醯輔酶A去氫酶缺乏症	AR
9	ACADVL	Very long chain acyl-CoA dehydrogenase deficiency, VLCAD deficiency 極長鏈醯輔酶A去氫酶缺乏症	AR
10	ACAT1	Beta-ketothiolase deficiency β-酮硫解酶缺乏症	AR
11	ACE	Renal tubular dysgenesis 腎小管發育不全	AR
12	ACVRL1	Hereditary hemorrhagic telangiectasia 遺傳性出血性之血管擴張症	AD
13	ADA	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
14	AGRN	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
15	AGT	Renal tubular dysgenesis 腎小管發育不全症	AR
16	AGTR1	Renal tubular dysgenesis 腎小管發育不全症	AR
17	AGXT	Hyperoxaluria, primary, type 1, HP1 原發性高草酸尿症第一型	AR
18	AHCY	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency 高甲硫胺酸血症伴隨S-腺苷-L-高半胺酸水解酵素缺乏症	AR
19	AK2	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
20	ALDH7A1	Pyridoxine-dependent epilepsy 維生素B7依賴性癲癇	AR
21	ALDOB	Hereditary fructose intolerance, HFI 遺傳性果糖不耐症	AR
22	ALG2	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
23	ALPL	Hypophosphatasia 低磷酸酶症 Pyridoxine-dependent epilepsy 維生素B6依賴性癲癇	AD/AR AR
24	ALX4	Parietal foramina 2 頂骨孔症第二型	AD
25	AMT	Glycine encephalopathy, GCE 甘氨酸腦病(非酮性高甘胺酸血症)	AR
26	ANKRD11	KBG syndrome KBG症候群	AD
27	ANOS1	Kallmann syndrome 1 Kallmann氏症候群第一型	XL
28	AP1S1	MEDNIK syndrome MEDNIK症候群	AR
29	APC	Familial adenomatous polyposis 家族腺瘤性瘜肉症	AD
30	APOB	Hypercholesterolemia, familial, 2 家族性高膽固醇血症	AD
31	AQP2	AVP resistance (formerly nephrogenic diabetes insipidus) 抗利尿激素抗性（舊稱腎性尿崩症）	AD/AR
32	AR	Androgen insensitivity syndrome 雄激素不敏感綜合症	XL

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
33	ARG1	Argininemia 精胺酸血症	AR
34	ARSA	Metachromatic leukodystrophy 異染性腦白質退化症	AR
35	ARSB	Mucopolysaccharidosis type 6 (Maroteaux-Lamy syndrome), MPS6 黏多醣症第六型（馬洛托-拉米氏症）	AR
36	ARX	Developmental and epileptic encephalopathy 1, EIEE1 發育性癲癇性腦病第一型 Lissencephaly, X-linked 2(XLAG Syndrome) X染色體連鎖無腦回畸形 Intellectual developmental disorder, X-linked X染色體連鎖智力障礙	XL
37	ASL	Argininosuccinic aciduria, ASA 精胺丁二酸酵素缺乏症	AR
38	ASPA	Canavan disease 卡納萬病（海綿狀腦白質營養不良症）	AR
39	ASS1	Citrullinemia type 1, CTLN1 瓜胺酸血症第一型	AR
40	ASXL1	Bohring-Opitz syndrome Bohring-Opitz症候群	AD
41	ATL1	Neuropathy, hereditary sensory, type ID 遺傳性感覺神經病變1D型 Hereditary spastic paraplegia 3A, autosomal dominant 遺傳性痙攣性下身麻痺	AD
42	ATM	Ataxia-telangiectasia 共濟失調微血管擴張症候群	AR
43	ATP7A	Menkes syndrome Menkes氏症候群	XL
44	ATP7B	Wilson disease, WND 威爾森氏症	AR
45	AUH	3-Methylglutaconyl-CoA Hydratase Deficiency 3-甲基戊烯二醯輔酶A水和酶缺乏症	AR
46	AVPR2	AVP resistance(nephrogenic diabetes insipidus) 抗利尿激素抗性（腎因型尿崩症）	XL
47	BCKDHA	Maple syrup urine disease 楓糖尿症	AR
48	BCKDHB	Maple syrup urine disease 楓糖尿症	AR
49	BLM	Bloom Syndrome 布隆氏症候群	AR
50	BLNK	Agammaglobulinemia 無丙種球蛋白血症	AR
51	BRAF	Leopard syndrome 3 LEOPARD 症候群第三型 Noonan syndrome 7 努南氏症候群第七型	AD
52	BTD	Biotinidase deficiency 生物素酶缺乏症	AR
53	BTK	Agammaglobulinemia 無丙種球蛋白血症	XL
54	CA5A	Carbonic anhydrase VA deficiency 碳酸酐酶VA缺乏症	AR
55	CACNA1C	Long QT syndrome 長QT症候群	AD
56	CACNA1D	Primary aldosteronism 原發性醛固酮增多症	AD
57	CAD	Early infantile epileptic encephalopathy 早期嬰兒癲癇性腦病	AR
58	CASR	Neonatal severe hyperparathyroidism 新生兒重度甲狀旁腺功能亢進	AD/AR
59	CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia 類努南症候群伴隨或不伴隨幼年型骨髓單核細胞白血病	AD
60	CBS	Homocystinuria (HCU) due to cystathionine beta-synthase deficiency 高胱胺酸尿症	AR
61	CC2D2A	COACH syndrome 2 COACH症候群第二型 Joubert syndrome 9 Joubert氏症候群第九型 Meckel syndrome 6 梅克爾症候群第六型	AR
62	CD247	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
63	CD3D	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR

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64	CD3E	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
65	CD40	Combined immunodeficiencies 複合免疫缺陷症	AR
66	CD40LG	Combined immunodeficiencies 複合免疫缺陷症	XL
67	CD79A	Agammaglobulinemia 無丙種球蛋白血症	AR
68	CD96	C syndrome(Opitz trigonocephaly syndrome) C綜合症（Opitz三角頭綜合症）	AD
69	CDKL5	CDKL5 deficiency disorder CDKL5缺乏症	XL
70	CEP55	Multinucleated neurons-anhydramnios-renal dysplasia-cerebellar hypoplasia-hydranencephaly syndrome(MARCH) 多核神經元-無羊水-腎發育不良-小腦發育不全-腦積水症候群	AR
71	CFC1	Heterotaxy, visceral, 2, autosomal 內臟異位症	AD
72	CFTR	Cystic fibrosis 囊腫性纖維症	AR
73	CHAT	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
74	CHD7	CHARGE syndrome CHARGE 症候群	AD
75	CHRNA1	Congenital myasthenic syndromes 先天性肌無力綜合症	AD/AR
76	CHRNB1	Congenital myasthenic syndromes 先天性肌無力綜合症	AD/AR
77	CHRND	Congenital myasthenic syndromes 先天性肌無力綜合症	AD/AR
78	CHRNE	Congenital myasthenic syndromes 先天性肌無力綜合症	AD/AR
79	CIITA	Combined immunodeficiencies 複合免疫缺陷症	AR
80	CLCN2	Hyperaldosteronism 原發性高醛固酮症	AD
81	CLCN7	Osteopetrosis, autosomal dominant 2 顯性遺傳骨質石化症第二型(大理石寶寶) Osteopetrosis, autosomal recessive 4 隱性遺傳骨質石化症第四型	AD AR
82	COL10A1	Metaphyseal chondrodysplasia, Schmid type Schmid型幹骺端軟骨發育不全	AD
83	COL11A1	Deafness, autosomal dominant 37 聽損 Fibrochondrogenesis 1 纖維軟骨增生 Marshall syndrome 馬歇爾症候群 Stickler syndrome, type II 斯蒂克勒症候群	AD AR AD AD
84	COL13A1	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
85	COL1A1	Osteogenesis Imperfecta type III or severe type IV 成骨不全症 第三型或嚴重第四型	AD
86	COL1A2	Osteogenesis Imperfecta type III or severe type IV 成骨不全症 第三型或嚴重第四型	AD
87	COL2A1	Achondrogenesis, type II; ACG2 or hypochondrogenesis 軟骨成長不全 Spondyloperipheral dysplasia 脊椎周圍發育不良 Spondyloepimetaphyseal dysplasia, Strudwick type 脊椎干骺端發育不良 Stickler syndrome, type I 斯蒂克勒綜合症第一型	AD
88	COL4A3	Alport syndrome type 3 亞伯氏症候群第3型	AD/AR
89	COL4A4	Alport syndrome type 2 亞伯氏症候群第2型	AR
90	COL4A5	Alport syndrome, X-linked 性聯遺傳亞伯氏症候群	XL
91	COL5A1	Ehlers-Danlos syndrome, classic type, 1 Ehlers-Danlos症候群 Fibromuscular dysplasia, multifocal 纖維肌性發育不良	AD

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92	COL5A2	Ehlers-Danlos syndrome, classic type, 2 Ehlers-Danlos症候群	AD
93	COLQ	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
94	COMT	Catechol-O-methyltransferase activity, variation in 兒茶酚胺氧位甲基轉移酶多態性	AR
95	COQ2	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
96	COQ4	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
97	COQ5	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
98	COQ6	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
99	COQ7	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
100	COQ8A	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
101	COQ9	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
102	CORO1A	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
103	CPS1	CPS1 deficiency CPS1缺乏症（氨基甲酰磷酸合成酶1缺乏症）	AR
104	CPT1A	Carnitine palmitoyltransferase I deficiency 肉鹼棕櫚醯基轉移酶缺乏症第一型	AR
105	CPT2	Carnitine palmitoyltransferase II deficiency, myopathic, stress-induced 肉鹼棕櫚酰轉移酶I缺乏症第二型（肌病型，應激誘發）	AD/AR
106	CREBBP	Menke-Hennekam syndrome 1 Menke-Hennekam症候群 Rubinstein-Taybi syndrome 1 魯賓斯坦-泰必氏綜合症	AD
107	CTNS	Cystinosis 胞囊症	AR
108	CYBA	Congenital defects of phagocyte number, function or both 吞噬細胞數量或功能先天缺陷	AR
109	CYBB	Congenital defects of phagocyte number, function or both 吞噬細胞數量或功能先天缺陷	XL
110	CYBC1	Congenital defects of phagocyte number, function or both 吞噬細胞數量或功能先天缺陷	AR
111	CYP11A1	Adrenal hyperplasia, congenital 先天性腎上腺增生	AR
112	CYP11B1	Hyperaldosteronism 原發性高醛固酮症	AD
113	CYP11B2	Hyperaldosteronism 原發性高醛固酮症 Hypoaldosteronism 低醛固酮症	AR
114	CYP21A2	Adrenal hyperplasia, congenital 先天性腎上腺增生	AR
115	CYP27B1	Vitamin D-dependent rickets type 1A 維生素D依賴型佝僂症第1A型	AR
116	DBT	Maple syrup urine disease 楓糖尿症	AR
117	DCLRE1C	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
118	DCX	Lissencephaly, X-linked 性聯遺傳平腦症	XL
119	DDC	AADC deficiency AADC缺乏症（芳香胺基酸脫羧酶缺乏症）	AR
120	DHCR24	Desmosterolosis 24-脫氫膽固醇症	AR
121	DHCR7	Smith-Lemli-Opitz syndrome 史密斯–萊姆利–歐畢茲症候群	AR
122	DLAT	Primary pyruvate dehydrogenase complex deficiency 原發性丙酮酸脫氫酶複合體缺乏症	AR
123	DLD	Dihydrolipoamide dehydrogenase deficiency 二氫硫辛醯胺脫氫酶缺乏症	AR
124	DMD	Duchenne Muscular Dystrophy 裘馨氏肌肉失養症	XL
125	DNAH9	Ciliary dyskinesia, primary, 40 原發性纖毛運動障礙	AR
126	DOCK8	Combined immunodeficiencies 複合免疫缺陷症	AR

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127	DOK7	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
128	DPAGT1	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
129	DSG2	Arrhythmogenic right ventricular dysplasia 10, ARVD 致心律失常性右心室發育不良 Cardiomyopathy, dilated, 1BB 擴張性心肌病變	AD/AR
130	DUOX2	Thyroid dyshormonogenesis 6 先天性甲狀腺低能症_甲狀腺素合成異常	AR
131	DUOXA2	Thyroid dyshormonogenesis 5 先天性甲狀腺低能症_甲狀腺素合成異常	AR
132	DYNC2H1	Short-rib thoracic dysplasia 3 with or without polydactyly 短肋胸廓發育不良合併或不合併多指症	AR
133	ECHS1	Mitochondrial short chain enoyl-CoA hydratase I deficiency & 3-hydroxyisobutryl-CoA hydrolase deficiency 線粒體短鏈烯醯-CoA水合酶I缺乏及3-羥基異丁酰-CoA水解酶缺乏症	AR
134	EDA	Ectodermal dysplasia 1, hypohidrotic, X-linked 外胚層發育不良第1型，低汗型，X連鎖	XL
135	EDAR	Hypohidrotic ectodermal dysplasia 少汗性外胚層發育不良症	AD/AR
136	EFNB1	Craniofrontonasal dysplasia 顱額鼻發育不良	XL
137	EFTUD2	Mandibulofacial dysostosis, Guion-Almeida type 下頜面骨發育不全症, Guion-Almeida型	AD
138	EIF2AK3	Infantile diabetes mellitus 嬰兒型糖尿病	AR
139	ELOVL4	Spinocerebellar ataxia 34 脊髓小腦萎縮症 34 型 Stargardt disease 3 Stargardt's氏症	AD
140	EMX2	Schizencephaly 裂腦畸形	AD
141	EP300	Menke-Hennekam syndrome 2 Menke-Hennekam症候群 Rubinstein-Taybi syndrome 2 魯賓斯坦-泰必氏綜合症	AD
142	ERBB3	Lethal congenital contracture syndrome 2 致死先天性攣縮症候群第二型	AR
143	ERF	Craniosynostosis 4 顱縫早閉	AD
144	ETFA	Glutaric acidemia type II 第二型戊二酸血症	AR
145	ETFB	Glutaric acidemia type II 第二型戊二酸血症	AR
146	ETFDH	Glutaric acidemia type II 第二型戊二酸血症	AR
147	EVC	Ellis-van Creveld syndrome 埃利偉氏症候群	AR
148	EVC2	Ellis-van Creveld syndrome 埃利偉氏症候群 Weyers acrofacial dysostosis Weyers面骨發育不全	AR AD
149	EXT1	Exostoses, multiple, type 1 遺傳性多發性骨軟骨瘤	AD
150	EXT2	Exostoses, multiple, type 2 遺傳性多發性骨軟骨瘤	AD
151	EYA1	Branchiootic syndrome type 1 Branchiootic 症候群第1型 Branchiootorenal syndrome type 1 Branchiootorenal 症候群第1型	AD
152	EZH2	Weaver syndrome 韋弗綜合症	AD
153	F10	Coagulation factor deficiencies 凝血因子缺乏症	AR
154	F11	Coagulation factor deficiencies 凝血因子缺乏症	AD/AR
155	F12	Coagulation factor deficiencies 凝血因子缺乏症	AR
156	F13A1	Coagulation factor deficiencies 凝血因子缺乏症	AR
157	F13B	Coagulation factor deficiencies 凝血因子缺乏症	AR

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158	F2	Coagulation factor deficiencies 凝血因子缺乏症	AR
159	F5	Coagulation factor deficiencies 凝血因子缺乏症	AR
160	F7	Coagulation factor deficiencies 凝血因子缺乏症	AR
161	F8	Hemophilia A 血友病A型 Coagulation factor deficiencies 凝血因子缺乏症	XL
162	F9	Coagulation factor deficiencies 凝血因子缺乏症	XL
163	FAH	Hereditary tyrosinemia type 1 遺傳性酪氨酸血症1型	AR
164	FAM111A	FAM111A-related skeletal dysplasia FAM111A相關骨骼發育不良	AD
165	FANCA	Fanconi anemia 范可尼氏貧血症	AR
166	FANCD2	Fanconi anemia 范可尼氏貧血症	AR
167	FAS	Diseases of immune dysregulation 免疫調節疾病	AD
168	FASLG	Diseases of immune dysregulation 免疫調節疾病	AD
169	FBN1	Marfan syndrome 馬凡氏症候群	AD
170	FBN2	Beals Syndrome / Congenital Contractural Arachnodactyly 畢耳氏症候群 / 先天性攣縮蜘蛛樣指 / 趾	AD
171	FBP1	Fructose-1,6-bisphosphatase deficiency 果糖-1,6-二磷酸酶缺乏症	AR
172	FECH	Protoporphyria, erythropoietic 紅血球原卟啉症	AR
173	FGA	Coagulation factor deficiencies 凝血因子缺乏症	AR
174	FGB	Coagulation factor deficiencies 凝血因子缺乏症	AR
175	FGF10	Lacrimo-auriculo-dento-digital syndrome LADD症候群	AD
176	FGF23	Hypophosphatemic rickets, autosomal dominant 低磷酸鹽佝僂症	AD
177	FGFR1	Encephalocraniocutaneous lipomatosis 腦顱皮膚脂肪增多症 Hartsfield syndrome Hartsfield 症候群 Hypogonadotropic hypogonadism 2 with or without anosmia 低促性腺激素性性腺功能減退症 Jackson-weiss syndrome Jackson-Weiss 症候群 Osteoglophonic dysplasia Osteoglophonic 發育不良 Pfeiffer syndrome Pfeiffer 症候群 Trigonocephaly 1 三角頭畸形第一型	AD
178	FGFR2	Apert Syndrome 亞伯氏症候群 Crouzon syndrome 克魯松氏症候群 Jackson-weiss syndrome Jackson-Weiss 症候群 Pfeiffer syndrome Pfeiffer 症候群 Lacrimoauriculodentodigital syndrome LADD 症候群	AD

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179	FGFR3	Lacrimoauriculodentodigital syndrome LADD 症候群 Camptodactyly, tall stature, and hearing loss syndrome 屈曲指合併身材高大及聽損 Crouzon syndrome with acanthosis nigricans 克魯松氏症候群合併黑色素棘皮症 Muenke syndrome Muenke 氏症候群 Achondroplasia, severe, with developmental delay and acanthosis nigricans 嚴重型軟骨發育不全合併發展遲緩及黑色素棘皮症 Thanatophoric dysplasia, type I 致死性畸胎侏儒症第一型 Thanatophoric dysplasia, type II 致死性畸胎侏儒症第二型	AD
180	FGG	Coagulation factor deficiencies 凝血因子缺乏症	AR
181	FLAD1	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency 由黃素腺嘌呤二核苷酸合成酶缺乏引起的脂肪儲積性肌病	AR
182	FLNB	Atelosteogenesis, type I and type III 骨骼發育不全第一型及第三型 Boomerang dysplasia 回飛鏢樣骨發育不全 Larsen syndrome Larsen氏症候群（顎裂-先天性脫位症候群）	AD
183	FLT4	Congenital heart defects, multiple types 先天性心臟缺陷 Lymphatic malformation 1 淋巴管畸形	AD
184	FNIP1	Agammaglobulinemia 無丙種球蛋白血症	AR
185	FOLR1	Cerebral folate transport deficiency 腦葉酸運輸缺陷	AR
186	FOXC2	Lymphedema-distichiasis syndrome 淋巴水腫-雙行睫綜合徵	AD
187	FOXG1	Rett syndrome 雷特氏症	AD
188	FOXL2	Blepharophimosis-Ptosis-Epicanthus Inversus Syndrome, type I and II 先天性家族性瞼口狹小症第一型及第二型 Premature ovarian failure 早發性卵巢衰竭	AD/AR
189	FOXN1	Thymic defects 胸腺缺陷	AR
190	FOXP3	Diseases of immune dysregulation 免疫調節疾病	XL
191	FREM1	Manitoba oculotrichoanal syndrome, MOTA MOTA 症候群 Bifid nose with or without anorectal and renal anomalies syndrome, BNAR BNAR 症候群 FREM1-related congenital anomalies of kidney and urinary tract, CAKUT 先天性腎臟與泌尿道異常 Trigonocephaly 顱縫早閉	AD/AR
192	G6PC1	Glycogen storage disease type 1A 肝醣儲積症第1型	AR
193	G6PD	Anemia, nonspherocytic hemolytic, due to G6PD deficiency G6PD缺乏症 (蠶豆症)	XL
194	GAA	Infantile-onset Pompe disease 嬰兒期發病Pompe病	AR
195	GALK1	Galactokinase deficiency 半乳糖激酶缺乏症	AR
196	GALNS	Mucopolysaccharidosis 4a (Morquio) 黏多醣症第4A型（摩可氏症）	AR
197	GALT	Classic galactosemia 經典型半乳糖血症	AR
198	GATA1	GATA1-positive transient abnormal myelopoiesis GATA1陽性暫時性異常骨髓增生症	Somatic
199	GATA4	Infantile diabetes mellitus 嬰兒型糖尿病	AD
200	GATA6	Infantile diabetes mellitus 嬰兒型糖尿病	AD
201	GBA(GBA1)	Neuronopathic Gaucher disease 神經型高雪氏症	AR

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
202	GCDH	Glutaricaciduria, type I 戊二酸血症第1型	AR
203	GCH1	Dopamine-responsive dystonia 多巴胺反應性肌張力不全症 Hyperphenylalaninemia, BH4-deficient, Type B B型四氫基喋呤缺乏之高苯丙胺酸血症	AD/AR AR
204	GCK	Congenital hyperinsulinism 先天性高胰島素血症 Infantile diabetes mellitus 嬰兒型糖尿病	AD AR
205	GFPT1	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
206	GFRA1	Renal hypodysplasia/aplasia 4, RHDA4 腎臟發育不良/發育不全	AR
207	GJB1	Charcot-Marie-Tooth disease, X-linked dominant 1 性聯遺傳進行性腓骨肌萎縮症	XL
208	GJB2	Sensorineural hearing loss 感覺神經性聽損	AD/AR
209	GJB6	Deafness, autosomal recessive 1B 體染色體隱性遺傳聽損第1B型	AD/AR
210	GLA	Fabry disease 法布瑞氏症	XL
211	GLB1	GM1-gangliosidosis type 1,2,3 GM1神經節苷脂儲積症第1型，第2型，第3型 Mucopolysaccharidosis type 4 黏多醣症第4型	AR
212	GLDC	Glycine encephalopathy 甘氨酸腦病	AR
213	GLI3	Greig cephalopolysyndactyly syndrome 格雷格頭顱多指趾症候群 Pallister-Hall syndrome Pallister-Hall 症候群 Polydactyly, postaxial, types A1 and B 軸後多指症 Polydactyly, preaxial, type IV 軸後多指症	AD
214	GLIS3	Infantile diabetes mellitus 嬰兒型糖尿病	AR
215	GLUD1	Congenital hyperinsulinism 先天性高胰島素血症	AD
216	GMPPB	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
217	GNA11	Hypocalcemia, autosomal dominant 2 顯性遺傳低血鈣症第二型 Hypocalciuric hypercalcemia, familial, type II 家族性低尿鈣性高血鈣症第二型	AD
218	GNMT	Glycine N-methyltransferase deficiency 甘氨酸N-甲基轉移酶缺乏症	AR
219	GOT2	Glutamic-oxaloacetic transaminase 2 deficiency 谷氨酸-草酰乙酸轉氨酶2缺乏症	AR
220	GPIHBP1	GPIHBP1 deficiency GPIHBP1缺乏症	AR
221	GPR143	Ocular albinism type 1 眼睛白化症第1型	XL
222	ADGRG1	Bilateral frontoparietal polymicrogyria, BFPP 雙側額頂葉多小腦迴症	AR
223	GUCY2C	Congenital sodium diarrhea 先天性鈉離子腹瀉	AD
224	GUSB	Mucopolysaccharidosis 7 (Sly) 黏多醣症第7型（史萊氏症）	AR
225	HADHA	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency and trifunctional protein deficiency 長鏈3-羥基酰基-CoA脫氫酶缺乏及三功能蛋白缺乏症	AR
226	HADHB	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency and trifunctional protein deficiency 長鏈3-羥基酰基-CoA脫氫酶缺乏及三功能蛋白缺乏症	AR
227	HBA1	Hemoglobin Bart's hydrops fetalis (subset of alpha thalassemia) 巴氏血紅素胎兒水腫（α-地中海型貧血的一種）	AD
228	HBA2	Hemoglobin Bart's hydrops fetalis (subset of alpha thalassemia) 巴氏血紅素胎兒水腫（α-地中海型貧血的一種）	AD
229	HBB	Beta Thalassemia β-地中海型貧血 Sickle cell disease 鐮刀型紅血球疾病	AR
230	HBG1	Hereditary persistence of fetal hemoglobin, HPFH 遺傳性胎兒血色素持續症	AD

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
231	HDAC8	Cornelia de Lange syndrome 5 Cornelia de Lange氏症候群（狄蘭氏症候群）	XL
232	HESX1	Combined pituitary hormone deficiency 複合性腦下垂體激素缺乏症	AD/AR
233	HEXA	Tay-Sachs disease 家族黑矇性癡呆症	AR
234	HFE	Hemochromatosis, type 1; HFE1 血鐵沈積症	AR
235	HIBCH	Mitochondrial short chain enoyl-CoA hydratase I deficiency & 3-hydroxyisobutryl-CoA hydrolase deficiency 線粒體短鏈烯醯-CoA水合酶I缺乏及3-羥基異丁酰-CoA水解酶缺乏症	AR
236	HLCS	Holocarboxylase synthetase deficiency 全羧化酶合成酶缺乏症	AR
237	HMGCL	HMG CoA lyase deficiency HMG-CoA裂解酶缺乏症	AR
238	HNF1A	Congenital hyperinsulinism 先天性高胰島素血症	AD
239	HNF4A	Congenital hyperinsulinism 先天性高胰島素血症	AD
240	HNRNPK	Au-Kline syndrome Au-Kline 症候群	AD
241	HPD	Hawkinsinuria 霍金尿症 Tyrosinemia Type 3 酪胺酸血症第3型	AD/AR
242	HPRT1	Lesch-Nyhan syndrome 萊希-尼亨症候群 Hyperuricemia, HRPT-related 高尿酸血症	XL
243	HRAS	Costello syndrome 克斯提洛氏彈性蛋白缺陷症	AD
244	HSD17B3	17-beta hydroxysteroid dehydrogenase 3 deficiency 17-β-羥基類固醇脫氫酶缺乏症第3型	AR
245	HSD3B2	Adrenal hyperplasia, congenital 先天性腎上腺增生	AR
246	HSPD1	Spastic paraplegia 13, autosomal dominant 顯性遺傳痙攣性下身麻痺	AD
247	IDH2	D-2-hydroxyglutaric aciduria 2 D-2-羥基戊二酸尿症第二型	AD
248	IDS	Mucopolysaccharidosis 2 (Hunter) 黏多醣症第2型（亨特氏症）	XL
249	IDUA	Mucopolysaccharidosis 1 (Hurler) 黏多醣症第1型（赫勒氏症）	AR
250	IER3IP1	Infantile diabetes mellitus 嬰兒型糖尿病	AR
251	IGF1R	Insulin-like growth factor I, resistance to 第一型類胰島素生長因子抗性	AD/AR
252	IGHM	Agammaglobulinemia 無丙種球蛋白血症	AR
253	IGHMBP2	Charcot-Marie-Tooth disease, axonal, type 2S, CMT2S 軸索型進行性腓骨肌萎縮症 Spinal muscular atrophy with respiratory distress type 1, SMARD1 脊髓性肌肉萎縮症伴呼吸窘迫第一型	AR
254	IGSF1	Central hypothyroidism 中樞性甲狀腺功能低下	XL
255	IKBKG	Combined immunodeficiencies 複合免疫缺陷症	XL
256	IKZF1	Combined immunodeficiencies 複合免疫缺陷症	AD
257	IL10RA	Diseases of immune dysregulation 免疫調節疾病	AR
258	IL10RB	Diseases of immune dysregulation 免疫調節疾病	AR
259	IL1RN	Autoinflammatory disorders 自體炎症性疾病	AR
260	IL2RG	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	XL
261	IL7R	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
262	INS	Infantile diabetes mellitus 嬰兒型糖尿病	AD/AR
263	IRAK4	Defects in intrinsic and innate immunity 內在及先天免疫缺陷	AR

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264	IRF6	Popliteal pterygium syndrome 1 腘窩翼狀膜症候群 Van der Woude syndrome Van der Woude 症候群（體染色體顯性遺傳唇顎裂症候群）	AD
265	ITGB2	Congenital defects of phagocyte number, function or both 吞噬細胞數量或功能先天缺陷	AR
266	IVD	Isovaleric acidemia 異戊酸血症	AR
267	JAG1	Alagille syndrome 1 阿拉吉歐症候群第一型	AD
268	JAK3	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
269	KAT6B	Genitopatellar syndrome 生殖器-髕骨症候群 Say-Barber-Biesecker-Young-Simpson syndrome(SBBYSS syndrome) SBBYSS 症候群	AD
270	KCNH2	Long QT syndrome 長QT症候群	AD
271	KCNJ11	Congenital hyperinsulinism 先天性高胰島素血症 Infantile diabetes mellitus 嬰兒型糖尿病	AD/AR
272	KCNJ5	Hyperaldosteronism 原發性高醛固酮症	AD
273	KCNQ1	Long QT syndrome 長QT症候群	AD
274	KCNQ2	Self-limited familial neonatal epilepsy, neonatal-onset developmental and epileptic encephalopathy 自限性家族性新生兒癲癇、新生兒起病發育及癲癇性腦病	AD
275	KCNQ3	Self-limited familial neonatal epilepsy, neonatal-onset developmental and epileptic encephalopathy 自限性家族性新生兒癲癇、新生兒起病發育及癲癇性腦病	AD
276	KDM6A	Kabuki syndrome 歌舞伎症候群	XL
277	KDR	Hemangioma, capillary infantile 嬰兒微血管瘤	AD
278	KIT	Gastrointestinal stromal tumor, GIST 胃腸道基質瘤 Piebaldism 斑狀白化症	AD
279	KLHL40	Nemaline myopathy 8, autosomal recessive 隱性遺傳性桿狀體肌病	AR
280	KMT2D	Kabuki syndrome 歌舞伎症候群	AD
281	KRAS	Noonan syndrome 1 努南氏症候群第1型	AD
282	LAMTOR2	Immunodeficiency due to defect in MAPBP-interacting protein MAPBP相互作用蛋白缺陷之免疫缺乏症	AR
283	LAT	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
284	LCK	Combined immunodeficiencies 複合免疫缺陷症	AR
285	LCP2	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
286	LDLR	Hypercholesterolemia, familial, 1 家族性高膽固醇血症	AD/AR
287	LHX3	Combined pituitary hormone deficiency 複合性腦下垂體激素缺乏症	AR
288	LHX4	Combined pituitary hormone deficiency 複合性腦下垂體激素缺乏症	AD
289	LIG4	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
290	LIPA	Lysosomal acid lipase deficiency 溶酶體酸性脂酶缺乏症	AR
291	PAFAH1B1	Lissencephaly 1 平腦症第一型 Subcortical laminar heterotopia 皮質下帶狀灰質異位症	AD

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
292	LMNA	Cardiomyopathy, dilated, 1A 擴張型心肌病變 1A型 Charcot-Marie-Tooth disease, type 2B1 進行性腓骨肌萎縮症 2B1型 Emery-Dreifuss muscular dystrophy 2, autosomal dominant Emery-Dreifuss肌失養症，體染色體顯性遺傳 Emery-Dreifuss muscular dystrophy 3, autosomal recessive Emery-Dreifuss肌失養症，體染色體隱性遺傳 Heart-hand syndrome, Slovenian type 斯洛維尼亞型心手症候群 Hutchinson-Gilford progeria 早老症 Lipodystrophy, familial partial, type 2 家族性部分性脂肪失養症 Malouf syndrome Malouf 症候群 Mandibuloacral dysplasia 下頷骨發育不良 Muscular dystrophy, congenital 先天性肌失養症 Restrictive dermopathy 2 拘束性皮膚病變	AD/AR
293	LMX1B	Nail-Patella Syndrome 指(趾)甲髕骨症候群	AD
294	LRBA	Diseases of immune dysregulation 免疫調節疾病	AR
295	LYST	Severe congenital neutropenias 嚴重先天性中性白血球缺乏症	AR
296	MAGED2	Bartter syndrome 巴特氏症候群	XL
297	MAP2K1	Cardiofaciocutaneous syndrome 3 Cardiofaciocutaneous 症候群第三型	AD
298	MAP2K2	Cardiofaciocutaneous syndrome 4 Cardiofaciocutaneous 症候群第四型	AD
299	MATN3	Epiphyseal dysplasia, multiple, 5 多發性骨骺發育不全第五型	AD
300	MCCC1	3-Methylcrotonyl-CoA carboxylase 1 deficiency, MCC1D (MCCC1-Related) 3-甲基巴豆醯輔酵素羧化酵素缺乏症第1型	AR
301	MCCC2	3-Methylcrotonyl-CoA carboxylase 2 deficiency, MCC2D (MCCC2-Related) 3-甲基巴豆醯輔酵素羧化酵素缺乏症第2型	AR
302	MCEE	Isolated methylmalonic acidemia 孤立型甲基丙二酸血症 (Vitamin B12-responsive) Methylmalonic acidemia 維生素B12反應型甲基丙二酸血症	AR
303	MECOM	Severe congenital thrombocytopenias 嚴重先天性血小板減少症	AD
304	MECP2	Neurodevelopmental conditions (i.e. Rett syndrome) 神經發展疾病（如瑞特氏症）	XL
305	MFN2	Charcot-Marie-Tooth disease, axonal, type 2A2A 軸索型進行性腓骨肌萎縮症 Charcot-Marie-Tooth disease, axonal, type 2A2B 軸索型進行性腓骨肌萎縮症 Hereditary motor and sensory neuropathy VIA 遺傳性肌肉感覺神經病變	AD/AR
306	MLYCD	Malonyl-CoA decarboxylase deficiency 丙二醯輔酶A脫羧酶缺乏症	AR
307	MMAA	Isolated methylmalonic acidemia 孤立型甲基丙二酸血症 (Vitamin B12-responsive) Methylmalonic acidemia 維生素B12反應型甲基丙二酸血症	AR
308	MMAB	Isolated methylmalonic acidemia 孤立型甲基丙二酸血症 (Vitamin B12-responsive) Methylmalonic acidemia 維生素B12反應型甲基丙二酸血症	AR
309	MMACHC	Methylmalonic aciduria and homocystinuria, cblC type 甲基丙二酸尿症合併高半胱胺酸尿症，cblC型	AR
310	MMADHC	Isolated methylmalonic acidemia 孤立型甲基丙二酸血症 (Vitamin B12-responsive) Methylmalonic acidemia 維生素B12反應型甲基丙二酸血症	AR
311	MMUT	Infantile-onset Pompe disease 嬰兒期發病Pompe病	AR
312	MNX1	Infantile diabetes mellitus 嬰兒型糖尿病	AD

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
313	MOCS1	Molybdenum cofactor deficiency 鉬輔因子缺乏症	AR
314	MPI	MPI-CDG MPI-CDG（磷酸甘露糖異構酶缺乏症）	AR
315	MPL	Severe congenital thrombocytopenias 嚴重先天性血小板減少症	AR
316	MRAP	ACTH signaling defects ACTH訊號缺陷	AR
317	MSN	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	XL
318	MSRB3	Genetic hearing loss 遺傳性聽力障礙	AR
319	MSX2	Parietal foramina 1 頂骨孔症第一型	AD
320	MTM1	Myotubular myopathy, X-linked, XLMTM 性聯遺傳肌小管病變	XL
321	MUSK	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
322	MYD88	Defects in intrinsic and innate immunity 內在及先天免疫缺陷	AR
323	MYH3	Arthrogryposis, distal, type 2A (Freeman-Sheldon syndrome) 遠端關節彎曲2A型（Freeman-Sheldon氏症候群） Arthrogryposis, distal, type 2B3 (Sheldon-Hall syndrome) 遠端關節彎曲2B型（Sheldon-Hall氏症候群） Contractures, pterygia, and spondylocarpostarsal fusion syndrome 1A 關節攣縮、翼狀胬肉合併脊柱腕跗骨融合症1A型 Contractures, pterygia, and spondylocarpotarsal fusion syndrome 1B 關節攣縮、翼狀胬肉合併脊柱腕跗骨融合症1B型	AD/AR
324	MYH7	Cardiomyopathy, dilated, 1S 擴張型心肌病變 Cardiomyopathy, hypertrophic, 1 肥厚性心肌病變 Congenital myopathy 7A, myosin storage, autosomal dominant 常染色體顯性遺傳肌球蛋白儲存性先天性肌病 7A型 Congenital myopathy 7B, myosin storage, autosomal recessive 體染色體隱性遺傳肌球蛋白儲積性先天性肌病 7B型 Left ventricular noncompaction 5 左心室緻密不全	AD/AR
325	MYO5B	Diarrhea with microvillus atrophy, with or without cholestasis 伴有或不伴有膽汁鬱積的微絨毛萎縮性腹瀉	AR
326	MYRF	Cardiac-urogenital syndrome 心臟泌尿生殖症候群 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization 伴可逆性髓鞘空泡化的輕度腦炎/腦病 Nanophthalmos 1 小眼症	AD
327	NAGS	N-acetylglutamate synthase deficiency N-乙醯穀氨酸合成酶缺乏症	AR
328	NANS	NANS-CDG, congenital disorder of glycosylation 先天性糖基化異常	AR
329	NCF1	Congenital defects of phagocyte number, function or both 吞噬細胞數量或功能先天缺陷	AR
330	NCF2	Congenital defects of phagocyte number, function or both 吞噬細胞數量或功能先天缺陷	AR
331	NCF4	Congenital defects of phagocyte number, function or both 吞噬細胞數量或功能先天缺陷	AR
332	NDE1	Lissencephaly 4 (with microcephaly) 平腦症第四型併小頭畸形 Microhydranencephaly 小頭積水性無腦畸形	AR
333	NDP	Norrie disease 諾里氏病 Exudative vitreoretinopathy 2, X-linked X-連鎖家族性滲出性玻璃體視網膜病變	XL
334	NEUROG3	Infantile diabetes mellitus 嬰兒型糖尿病	AR
335	NEXN	Cardiomyopathy 心肌病變	AD/AR
336	NF1	Neurofibromatosis type 1, NF1 神經纖維瘤第一型	AD
337	NF2	Neurofibromatosis type 2, NF2 神經纖維瘤第二型	AD
338	NHEJ1	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
339	NIPBL	Cornelia de Lange syndrome 1 Cornelia de Lange 氏症候群	AD
340	NKX2-5	Atrial septal defect 7, with or without AV conduction defects 心房中隔缺損第7型 Ventricular septal defect 3 室中隔缺損第3型 Tetralogy of Fallot 法洛氏四重症 Hypoplastic left heart syndrome 2 左心發育不全症候群第2型	AD
341	NLRC4	Autoinflammatory disorders 自體炎症性疾病	AD
342	NLRP3	Autoinflammatory disorders 自體炎症性疾病	AD
343	NOTCH2	Alagille syndrome 2 阿拉吉歐症候群第二型	AD
344	NPC1	Niemann-Pick disease type C1, NPC1 尼曼匹克症C1型	AR
345	NPC2	Niemann-Pick disease type C2, NPC2 尼曼匹克症C2型	AR
346	NPRL3	Epilepsy, familial focal, with variable foci 3 伴隨不定病灶之家族性局部癲癇第三型	AD
347	NR0B1	Adrenal hypoplasia 腎上腺發育不全	XL
348	NR3C2	Primary aldosteronism 原發性醛固酮增多症	AD
349	NR5A1	Adrenal hypoplasia 腎上腺發育不全	AD
350	NRAS	Noonan syndrome 6 努南氏症候群第六型	AD
351	NSD1	Sotos syndrome Sotos 症候群	AD
352	NSDHL	Congenital hemidysplasia with ichthyosiform erythroderma and limb defects(CHILD syndrome) 先天性半身發育異常合併魚鱗癬樣紅皮症及肢體缺陷 CK syndrome CK 症候群	XL
353	OAT	Ornithine Aminotransferase (OAT) deficiency 鳥胺酸氨基轉移酶（OAT）缺乏症	AR
354	OCRL	Dent disease 2 Dent病 Lowe syndrome Lowe氏症候群	XL
355	OTC	Ornithine transcarbamylase (OTC) deficiency 鳥胺酸氨甲醯基轉移酶（OTC）缺乏症	XL
356	OTOF	Sensorineural hearing loss 感覺神經性聽損	AR
357	OTX2	Combined pituitary hormone deficiency 複合性腦下垂體激素缺乏症	AD
358	OXCT1	Succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency 琥珀酰-CoA:3-酮酸CoA轉移酶（SCOT）缺乏症	AR
359	PAH	Phenylketonuria, PKU 苯酮尿症 Hyperphenylalaninaemia 高苯丙胺酸血症	AR
360	PAX1	Thymic defects 胸腺缺陷	AR
361	PAX2	Glomerulosclerosis, focal segmental, 7 局部性腎絲球硬化症 Papillorenal syndrome 腎性眼缺損症候群	AD
362	PAX6	Infantile diabetes mellitus 嬰兒型糖尿病	AD
363	PCBD1	Hyperphenylalaninemia, BH4-deficient, Type D D型四氫基喋呤缺乏之高苯丙胺酸血症	AR
364	PCCA	Propionic acidemia 丙酸血症	AR
365	PCCB	Propionic acidemia 丙酸血症	AR
366	PCDH19	Developmental and epileptic encephalopathy 9 發展性癲癇腦病變第九型	XL
367	PCSK1	ACTH signaling defects ACTH訊號缺陷	AR

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
368	PDHA1	Primary pyruvate dehydrogenase complex deficiency 原發性丙酮酸脫氫酶複合體缺乏症	XL
369	PDHB	Primary pyruvate dehydrogenase complex deficiency 原發性丙酮酸脫氫酶複合體缺乏症	AR
370	PDHX	Primary pyruvate dehydrogenase complex deficiency 原發性丙酮酸脫氫酶複合體缺乏症	AR
371	PDP1	Primary pyruvate dehydrogenase complex deficiency 原發性丙酮酸脫氫酶複合體缺乏症	AR
372	PDSS1	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
373	PDSS2	Primary CoQ10 deficiency 原發性輔酶Q10缺乏症	AR
374	PDX1	Infantile diabetes mellitus 嬰兒型糖尿病	AD
375	PEX1	Peroxisome biogenesis disorder(Zellweger syndrome) 過氧化物生成障礙疾病（Zellweger氏症候群）	AR
376	PGM1	PGM1-CDG PGM1-CDG（磷酸葡萄糖變位酶1缺乏症）	AR
377	PGM3	Combined immunodeficiencies 複合免疫缺陷症	AR
378	PHEX	Hypophosphatemic rickets, X-linked dominant 遺傳性低磷酸鹽佝僂症	XL
379	PHGDH	3-PGDH deficiency 3-磷酸甘油酸脫氫酶缺乏症	AR
380	PIEZO1	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema 遺傳性乾燥性紅血球增多症 Lymphatic malformation 6 淋巴管畸形第六型	AD/AR
381	PIK3CA	Megalencephaly-capillary malformation-polymicrogyria syndrome, MCAP 巨腦畸形-微血管畸形症候群	AD
382	PIK3R1	Agammaglobulinemia 7, autosomal recessive 隱性遺傳低免疫球蛋白症 Immunodeficiency 36 免疫缺乏症第36型 SHORT syndrome SHORT 症候群	AR AD AD
383	PKD1	Polycystic kidney disease 1 多囊性腎病	AD
384	PKD2	Polycystic kidney disease 2 多囊性腎病	AD
385	PKHD1	Autosomal recessive polycystic kidney disease 胎兒型體染色體隱性遺傳多囊性腎臟病	AR
386	PKLR	Human erythrocyte R-type pyruvate kinase deficiency 紅血球R型丙酮酸激酶缺乏症	AR
387	PROKR2	Kallmann syndrome Kallmann氏症候群	AD/AR
388	PLEC	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
389	PLPBP	Pyridoxine-dependent epilepsy 維生素B6依賴性癲癇	AR
390	PMM2	Carbohydrate Deficiency Glycoprotein Type I syndrome 碳水化合缺乏醣蛋白症候群第一型	AR
391	PNPO	Pyridoxine-dependent epilepsy 維生素B6依賴性癲癇	AR
392	POLR1C	4H Leukodystrophy 4H 腦白質失養症	AR
393	POLR3A	4H Leukodystrophy 4H 腦白質失養症	AR
394	POLR3B	4H Leukodystrophy 4H 腦白質失養症	AD/AR
395	POMC	ACTH signaling defects ACTH訊號缺陷	AR
396	POU1F1	Combined pituitary hormone deficiency 複合性腦下垂體激素缺乏症	AD/AR
397	POU4F3	Sensorineural hearing loss 感覺神經性聽損	AD
398	PRDX1	Maternal vitamin B12 deficiency (Methylmalonic aciduria and homocystinuria cblC type) 甲基丙二酸血症缺乏維生素B13	AR
399	PREPL	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
400	PRF1	Diseases of immune dysregulation 免疫調節疾病	AR

項次 No.	基因 gene	單基因疾病 Single Gene Disorder	遺傳模式 Inheritance
401	PRKDC	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
402	PRODH	Hyperprolinemia, type I 高脯胺酸血症	AD/AR
403	PROP1	Combined pituitary hormone deficiency 複合性腦下垂體激素缺乏症	AR
404	PRRT2	Self-limited familial neonatal epilepsy, neonatal-onset developmental and epileptic encephalopathy 自限性家族性新生兒癲癇、新生兒起病發育及癲癇性腦病	AD
405	PRSS1	Pancreatitis, hereditary 遺傳性胰臟炎	AD
406	PTF1A	Infantile diabetes mellitus 嬰兒型糖尿病	AR
407	PTPN11	Leopard syndrome 1 Leopard 症候群第一型 Metachondromatosis 混合性軟骨瘤病 Noonan syndrome 1 努南氏症候群第一型	AD
408	PTPRC	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
409	PTS	Hyperphenylalaninemia, BH4-deficient, Type A A型四氫基喋呤缺乏之高苯丙胺酸血症	AR
410	QDPR	Hyperphenylalaninemia, BH4-deficient, Type C C型四氫基喋呤缺乏之高苯丙胺酸血症	AR
411	RAB23	Carpenter syndrome 1 卡本特氏症候群第1型	AR
412	RAB27A	Diseases of immune dysregulation 免疫調節疾病	AR
413	RAC2	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AD/AR
414	RAD21	Cornelia de Lange syndrome 4 Cornelia de Lange 氏症候群	AD/AR
415	RAF1	Noonan syndrome 5 努南氏症候群第五型	AD
416	RAG1	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
417	RAG2	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
418	RAPSN	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
419	RBM8A	Severe congenital thrombocytopenias 嚴重先天性血小板減少症	AR
420	RELN	Lissencephaly 2 (Norman-Roberts type) 平腦症第二型（諾曼 - 羅伯茨綜合症）	AD/AR
421	REN	Renal tubular dysgenesis 腎小管發育不全症 Tubulointerstitial kidney disease, autosomal dominant, 4 體染色體顯性遺傳性腎小管間質性腎病	AD/AR
422	RET	Hirschsprung disease 先天性巨結腸症 Medullary thyroid carcinoma 甲狀腺髓質癌 Multiple endocrine neoplasia 多發性內分泌腫瘤 Pheochromocytoma 嗜鉻細胞瘤	AD
423	RFX5	Combined immunodeficiencies 複合免疫缺陷症	AR
424	RFX6	Infantile diabetes mellitus 嬰兒型糖尿病	AR
425	RFXANK	Combined immunodeficiencies 複合免疫缺陷症	AR
426	RFXAP	Combined immunodeficiencies 複合免疫缺陷症	AR
427	RIT1	Noonan syndrome 8 努南氏症候群第八型	AD
428	RNF213	Moyamoya disease 2 毛毛樣腦血管疾病（霧霧症）	AD/AR
429	RPE65	RPE65-related disease RPE65相關視網膜病變	AD/AR
430	RPL11	Diamond-Blackfan anemia 鑽石-布萊克凡貧血	AD

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431	RPL5	Diamond-Blackfan anemia 鑽石-布萊克凡貧血	AD
432	RPS10	Diamond-Blackfan anemia 鑽石-布萊克凡貧血	AD
433	RPS19	Diamond-Blackfan anemia 鑽石-布萊克凡貧血	AD
434	RPS24	Diamond-Blackfan anemia 鑽石-布萊克凡貧血	AD
435	RUNX2	Cleidocranial dysplasia 鎖骨顱骨成骨不全症	AD
436	SCN1A	Dravet Syndrome 卓飛症候群（Dravet症候群） Developmental and epileptic encephalopathy 6B, non-Dravet 發展性與癲癇性腦病變 Epilepsy, generalized, with febrile seizures plus, type 2 全面性癲癇合併熱性痙攣 Severe myoclonic epilepsy of infancy 嚴重嬰兒肌跳躍癲癇	AD
437	SCN2A	Developmental and epileptic encephalopathy 發育性及癲癇性腦病	AD
438	SCN4A	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
439	SCN5A	Long QT syndrome 長QT症候群	AD
440	SCN8A	Self-limited familial neonatal epilepsy, neonatal-onset developmental and epileptic encephalopathy 自限性家族性新生兒癲癇、新生兒起病發育及癲癇性腦病	AD
441	SCNN1A	Primary aldosteronism 原發性醛固酮增多症	AR
442	SCNN1B	Primary aldosteronism 原發性醛固酮增多症	AR
443	SCNN1G	Primary aldosteronism 原發性醛固酮增多症	AR
444	SERPING1	Hereditary angioedema types1 and 2, HAE 遺傳性血管性水腫第一型及第二型	AD/AR
445	SF3B4	Acrofacial dysostosis 1, Nager type 軸前面骨發育不全症候群（Nager症候群）	AD
446	SFTPC	Surfactant deficiency 肺表面活性劑缺乏症	AD
447	SH2D1A	Diseases of immune dysregulation 免疫調節疾病	XL
448	SHOC2	Noonan syndrome-like with loose anagen hair 1 類努南氏症候群合併生長期毛髮鬆動症	AD
449	SI	Congenital sucrase-isomaltase deficiency 先天性蔗糖-異麥芽糖酶缺乏症	AR
450	SKI	Shprintzen-Goldberg syndrome Shprintzen-Goldberg 症候群	AD
451	SLC13A5	Early infantile epileptic encephalopathy 早期嬰兒癲癇性腦病	AR
452	SLC16A2	MCT8 deficiency (Allan-Herndon-Dudley Syndrome) MCT8缺乏症（艾倫-赫恩登-達德利症候群）	XL
453	SLC18A3	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
454	SLC19A3	Biotin-thiamine responsive basal ganglia disease 生物素-硫胺素響應性基底核病	AR
455	SLC22A5	Primary Carnitine Deficiency 原發性肉鹼缺乏症	AR
456	SLC25A1	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
457	SLC25A13	Citrullinemia type 2 瓜胺酸血症第2型	AR
458	SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome 高鳥氨酸血症-高氨血症-同瓜氨酸尿症候群	AR
459	SLC25A20	CACT deficiency 肉鹼脂酰轉移酶缺乏症（CACT缺乏症）	AR
460	SLC25A24	Fontaine progeroid syndrome 方丹早衰症候群	AD

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461	SLC26A2	Achondrogenesis 1b 軟骨生成不全第1b型 Atelosteogenesis type 2 骨骼發育不全第二型 Epiphyseal dysplasia, multiple, 4 多發性骨骼發育不全第四型 Diastrophic dysplasia 畸型發育不良	AR
462	SLC26A3	Congenital chloride diarrhea 先天性氯離子腹瀉	AR
463	SLC26A4	Sensorineural hearing loss 感覺神經性聽損 Pendred syndrome Pendred氏症候群	AR
464	SLC2A1	Glucose transporter type 1 deficiency 葡萄糖轉運蛋白1型缺乏症	AD/AR
465	SLC2A2	Infantile diabetes mellitus 嬰兒型糖尿病	AD
466	SLC35A2	SLC35A2-CDG (Congenital disorder of glycosylation, type IIm) 先天性醣基化障礙	XL
467	SLC5A1	Glucose-galactose malabsorption 葡萄糖-半乳糖吸收不良	AR
468	SLC5A7	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
469	SLC9A3	Congenital sodium diarrhea 先天性鈉離子腹瀉	AR
470	SLURP1	Meleda disease Meleda島病	AR
471	SMAD4	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome 少年多發性/遺傳性出血性血管擴張症 Juvenile polyposis syndrome 年輕型多發息肉症候群 Myhre syndrome Myhre 症候群	AD
472	SMC1A	Cornelia de Lange syndrome 2 Cornelia de Lange氏症候群（狄蘭氏症候群） Developmental and epileptic encephalopathy 發展性癲癇腦病變	XL
473	SMC3	Cornelia de Lange syndrome 3 Cornelia de Lange氏症候群（狄蘭氏症候群）	AD
474	SMN1	Spinal muscular atrophy 脊髓性肌肉萎縮症	AR
475	SMPD1	Niemann-Pick disease type A, type B 尼曼匹克症A型，B型	AR
476	SNAP25	Congenital myasthenic syndromes 先天性肌無力綜合症	AD
477	SNRPB	Cerebro-costo-mandibular syndrome 腦肋小頜症候群	AD
478	SOD1	Amyotrophic lateral sclerosis 1 肌萎縮性側索硬化症第一型	AD/AR
479	SOS1	Noonan syndrome 4 努南氏症候群第四型	AD
480	SOS2	Noonan syndrome 9 努南氏症候群第九型	AD
481	SOX3	Combined pituitary hormone deficiency 複合性腦下垂體激素缺乏症	XL
482	SOX9	46XX/46XY sex reversal 性別顛倒 Campomelic dysplasia 短指發育不良 Campomelic dysplasia with autosomal sex reversal 短指發育不良及性別顛倒	AD
483	SPAST	Hereditary Spastic Paraplegia, HSP 遺傳性痙攣性下身麻痺	AD
484	SPECC1L	Teebi hypertelorism syndrome 1 Teebi氏眼距過寬症候群	AD
485	SPRED1	Legius syndrome 雷吉士症候群	AD
486	SRD5A2	Pseudovaginal perineoscrotal hypospadias(5-alpha-reductase deficiency) 假陰道會陰陰囊尿道下裂（5α-還原酶缺乏症）	AR
487	STAR	Adrenal hyperplasia, congenital 先天性腎上腺增生	AR

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488	STAT3	Infantile diabetes mellitus 嬰兒型糖尿病	AD
489	STX11	Diseases of immune dysregulation 免疫調節疾病	AR
490	STXBP1	STXBP1-related neonatal epilepsy STXBP1相關新生兒癲癇	AD/AR
491	STXBP2	Diseases of immune dysregulation 免疫調節疾病	AR
492	SUZ12	Imagawa-Matsumoto syndrome, IMMAS(SUZ12-related overgrowth disorder) Imagawa-Matsumoto 症候群（SUZ12-related 過度生長障礙）	AD
493	SYNGAP1	Intellectual developmental disorder, autosomal dominant 5 智能發展障礙	AD
494	SYT2	Congenital myasthenic syndromes 先天性肌無力綜合症	AD/AR
495	TAFAZZIN	Barth syndrome 巴氏症候群	XL
496	TARDBP	Amyotrophic lateral sclerosis 10 肌萎縮性側索硬化症第十型	AD
497	TAT	Tyrosinemia type 2 酪胺酸血症第二型	AR
498	TBX19	ACTH signaling defects ACTH訊號缺陷	AR
499	TBX5	Holt-Oram syndrome Holt-Oram氏症候群	AD
500	TCF12	Craniosynostosis 3 顱縫過早癒合症第三型	AD
501	TCF3	Agammaglobulinemia 無丙種球蛋白血症	AD/AR
502	TGFBR1	Loeys-Dietz syndrome 1, LDS Loeys-Dietz 症候群第一型	AD
503	TGFBR2	Loeys-Dietz syndrome 2, LDS Loeys-Dietz 症候群第二型	AD
504	TH	Dopa responsive dystonia, autosomal recessive Segawa syndrome, recessive Tyrosine hydroxylase deficiency 酪胺酸羥化酶缺乏症	AR
505	THPO	Severe congenital thrombocytopenias 嚴重先天性血小板減少症	AD/AR
506	TPO	Goitrous hypothyroidism 甲狀腺腫型甲狀腺功能低下	AR
507	TRAF7	Cardiac, facial, and digital anomalies with developmental delay 心臟、面部及手指/腳趾異常合併發展遲緩	AD
508	TRAPPC2	Spondyloepiphyseal dysplasia tarda 遲發性脊椎骨骺生成不良	XL
509	TRHR	Central hypothyroidism 中樞性甲狀腺功能低下	AR
510	TRMU	Liver failure, infantile, transient 嬰兒肝功能衰竭（暫時性）	AR
511	TRPM6	Hypomagnesemia Type I 低鎂血症1型	AR
512	TSC1	Tuberous sclerosis complex 結節性硬化症	AD
513	TSC2	Tuberous sclerosis complex 結節性硬化症	AD
514	TSHB	Central hypothyroidism 中樞性甲狀腺功能低下	AR
515	TSHR	Permanent neonatal hyperthyroidism 永久性新生兒甲狀腺功能亢進	AD
516	TTC7A	Severe combined immunodeficiencies 嚴重複合免疫缺陷症	AR
517	TTR	Amyloid polyneuropathy, familial 家族性澱粉樣多發性神經病變	AD
518	TUBA1A	Lissencephaly 3 平腦症第三型	AD
519	TWIST1	Saethre-Chotzen syndrome Saethre-Chotzen 氏症候群	AD
520	UBE3A	Angelman syndrome 天使症候群	AD
521	UGT1A1	Crigler–Najjar type 1 克果納傑氏症第1型 (高膽紅素血症)	AD/AR
522	UNC13D	Diseases of immune dysregulation 免疫調節疾病	AR
523	UROS	Protoporphyria, erythropoietic 紅血球原卟啉症	AR

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524	USH1C	Genetic hearing loss 遺傳性聽力障礙	AR
525	USP18	Autoinflammatory disorders 自體炎症性疾病	AR
526	VAMP1	Congenital myasthenic syndromes 先天性肌無力綜合症	AR
527	VAPB	Amyotrophic lateral sclerosis 8 肌萎縮性側索硬化症第八型	AD
528	VHL	Von Hippel-Lindau syndrome 逢希伯-林道症候群(視網膜小腦脊髓血管瘤症)	AD
529	VLDLR	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1, CAMRQ1(VLDLR cerebellar hypoplasia) 小腦發育不全、智能發展障礙與平衡失調症候群第一型（VLDLR 小腦發育不全）	AR
530	WAC	Desanto-Shinawi syndrome Desanto-Shinawi 症候群	AD
531	WAS	Combined immunodeficiencies 複合免疫缺陷症	XL
532	WDR62	Microcephaly 2, primary, with or without cortical malformations 原發性小頭畸形，伴有或不伴有皮質畸形	AR
533	WFS1	Wolfram syndrome 1 Wolfram氏症候群	AD/AR
534	XIAP	Diseases of immune dysregulation 免疫調節疾病	XL
535	ZAP70	Combined immunodeficiencies 複合免疫缺陷症	AR
536	ZEB2	Mowat-Wilson syndrome Mowat-Wilson 症候群	AD
537	ZFP57	Infantile diabetes mellitus 嬰兒型糖尿病	AD/AR
538	ZIC1	Structural brain anomalies with impaired intellectual development and craniosynostosis, BAIDCS 伴隨智力發育受損與顱縫早閉的腦部結構異常	AD
539	ZIC3	Congenital heart defects, nonsyndromic, multiple types, 1, X-linked 先天性心臟缺陷 Heterotaxy, visceral, 1, X-linked 臟器異位症候群 VACTERL association, X-linked VACTERL綜合徵	XL

說明：

遺傳模式 Inheritance

AD：體染色體顯性遺傳 Autosomal dominant

AR：體染色體隱性遺傳 Autosomal recessive

XL：X 染色體性聯遺傳 X-linked inheritance