

| 疾病列表  | 疾病列表   | 基因       | 遺傳模式     | 帶因率        |
|---|--|----------|----------|------------|
|   | Achalasia-Addisonianism-Alacrima Syndrome                    | AAAS     |          |            |
|   | Harlequin ichthyosis   | ABCA12   |          |            |
|   | Stargardt Disease, Type 1                                    | ABCA4    |          |            |
| 進行性家族性肝內膽汁滯留症                                   | Progressive Familial Intrahepatic Cholestasis, Type 2        | ABCB11   | AR       | 1/112      |
|   | Progressive Familial Intrahepatic Cholestasis, Type 3        | ABCB4    |          |            |
|   | Pseudoxanthoma elasticum                                     | ABCC6    |          |            |
| 家族性胰島素過多症-ABCC8 型                               | Familial Hyperinsulinism, ABCC8-Related                      | ABCC8    | AR       | 1/112      |
| 性聯遺傳腎上腺腦白質失養症                                   | Adrenoleukodystrophy, X-Linked                               | ABCD1    | X-Linked | 1/21,000   |
| 醃輔酶 A 去氫酶 9 缺乏症                                 | Mitochondrial Complex I Deficiency, ACAD9-Related            | ACAD9    | AR       | <1/500     |
| 中鏈脂肪酸去氫酶缺乏症                                     | Medium Chain Acyl-CoA Dehydrogenase Deficiency               | ACADM    | AR       | 1/69       |
| 短鏈脂肪酸去氫酶缺乏症                                     | Short Chain Acyl-CoA Dehydrogenase Deficiency                | ACADS    | AR       | 1/85       |
| 短支鏈醃輔酶 A 去氫酶缺乏症                                 | Short/branched chain acyl-CoA dehydrogenase                  | ACADSB   | AR       | 1/368      |
| 極長鏈醃輔酶 A 去氫酶缺乏症                                 | Very Long-Chain Acyl-CoA Dehydrogenase Deficiency            | ACADVL   | AR       | 1/118      |
| Beta 硫解酶缺乏症                                     | Beta-Ketothiolase Deficiency                                 | ACAT1    | AR       | <1/500     |
| 過氧化物酶酰基輔酶 A 氧化酶缺乏症                              | Acyl-CoA Oxidase I Deficiency                                | ACOX1    | AR       | <1/500     |
| 丙二酸及甲基丙二酸血症                                     | Combined Malonic and Methylmalonic Aciduria                  | ACSF3    | AR       | <1/500     |
| 腺嘌呤脫氨酶缺乏症                                       | Severe Combined Immunodeficiency, ADA-Related                | ADA      | AR       | 1/224      |
| 膠原蛋白髮育異常埃勒斯-當洛二氏症候群                             | Ehlers-Danlos Syndrome, Type VIIC                            | ADAMTS2  | AR       | <1/500     |
| 雙側額頂多小腦迴畸形症                                     | Bilateral Frontoparietal Polymicrogyria                      | ADGRG1   | AR       | <1/500     |
| 天門冬醃胺葡萄糖胺尿症                                     | Aspartylglucosaminuria                                       | AGA      | AR       | <1/500     |
| 肝醃儲積症第三型  | Glycogen Storage Disease, Type III (Cori/Forbes)             | AGL      | AR       | 1/158      |
| 肢近端型點狀軟骨發育不良第三型                                 | Rhizomelic Chondrodysplasia Punctata, Type 3                 | AGPS     | AR       | <1/500     |
| 原發性高草酸鹽尿症-1型                                    | Hyperoxaluria, Primary, Type 1                               | AGXT     | AR       | 1/120      |
| 自體免疫多腺體症候群 I 型                                  | Autoimmune polyendocrinopathy syndrome, type I               | AIRE     | AR       | 1/150      |
| Sjögren-Larsson 症候群                             | Sjogren-Larsson Syndrome                                     | ALDH3A2  | AR       | 1/250      |
|   | Pyridoxine-dependent epilepsy                                | ALDH7A1  |          |            |
| 遺傳性果糖不耐症  | Hereditary Fructose Intolerance                              | ALDOB    | AR       | 1/122      |
| 先天性醃基化疾病-1c型                                    | Congenital Disorder of Glycosylation, Type 1C                | ALG6     | AR       | <1/500     |
| Alstrom 氏症候群                                    | Alstrom Syndrome   | ALMS1    | AR       | 1/500      |
| 低磷酸酯酶症  | Hypophosphatasia, ALPL-Related                               | ALPL     | AR       | 1/158      |
|   | Persistent Müllerian duct syndrome type 1                    | AMH      |          |            |
|   | Persistent Müllerian duct syndrome type 2                    | AMHR2    |          |            |
| 非酮性高甘胺酸血症 -AMT 型                                | Glycine Encephalopathy, AMT-Related                          | AMT      | AR       | 1/373      |
|   | Mental retardation, enteropathy, deafness, peripheral neuro  | AP1S1    |          |            |
| 腎性尿崩症   | Familial Nephrogenic Diabetes Insipidus, AQP2-Related        | AQP2     | AR       | <1/500     |
|   | Androgen insensitivity syndrome, X-Linked                    | AR       |          |            |
| 精胺酸酶缺乏症   | Argininemia  | ARG1     | AR       | 1/296      |
| 異染性白質退化症  | Metachromatic Leukodystrophy, ARSA-Related                   | ARSA     | AR       | 1/100      |
| 黏多醃症-6 型(馬洛托 - 拉米氏症)                            | Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)              | ARSB     | AR       | 1/250      |
| 精胺丁二酸酶缺乏症                                       | Argininosuccinate Lyase Deficiency                           | ASL      | AR       | 1/132      |
| 天門冬醃胺合成缺乏症                                      | Asparagine Synthetase Deficiency                             | ASNS     | AR       | <1/500     |
| 卡那凡氏症   | Canavan Disease  | ASPA     | AR       | 1/300      |
| 瓜胺酸血症   | Citrullinemia, Type 1  | ASS1     | AR       | 1/119      |
| 共濟失調微血管擴張性症候群                                   | Ataxia-Telangiectasia  | ATM      | AR       | 1/100      |
| 腎小管酸中毒伴隨耳聾                                      | Renal Tubular Acidosis and Deafness, ATP6V1B1-Related        | ATP6V1B1 | AR       | <1/500     |
| Menkes 氏症候群                                     | Menkes Syndrome, X-Linked                                    | ATP7A    | X-Linked | 1/50,000   |
| 威爾森氏症   | Wilson Disease   | ATP7B    | AR       | 1/87       |
|   | Progressive Familial Intrahepatic Cholestasis, Type 1        | ATP8B1   |          |            |
| 甲型海洋性貧血 -性聯遺傳智力障礙症候群                            | Alpha-Thalassemia Intellectual Disability Syndrome, X-Linked | ATRX     | X-Linked | <1/250,000 |
| Bardet-Biedl 氏症候群-1 型                           | Bardet-Biedl Syndrome 1                                      | BBS1     | AR       | 1/367      |
| Bardet-Biedl 氏症候群-10 型                          | Bardet-Biedl Syndrome 10                                     | BBS10    | AR       | 1/395      |
| Bardet-Biedl 氏症候群-12 型                          | Bardet-Biedl Syndrome 12                                     | BBS12    | AR       | 1/791      |
| Bardet-Biedl 氏症候群-2 型                           | Bardet-Biedl Syndrome 2                                      | BBS2     | AR       | 1/621      |
|   | Bardet-Biedl Syndrome 4                                      | BBS4     |          |            |
|   | Bardet-Biedl Syndrome 9                                      | BBS9     |          |            |
| 假性膽鹼酯酶缺乏症                                       | Pseudocholinesterase Deficiency                              | BCHE     | AR       | 1/28       |
| 楓糖尿症-Ia 型                                       | Maple Syrup Urine Disease, Type 1A                           | BCKDHA   | AR       | 1/321      |
| 楓糖尿症-Ib 型                                       | Maple Syrup Urine Disease, Type 1B                           | BCKDHB   | AR       | 1/364      |
| Björnstad 症候群; Gracile 症候群; 粒線體 Complex III 缺乏症 | GRACILE Syndrome   | BCS1L    | AR       | <1/500     |
| 布隆氏症候群  | Bloom Syndrome   | BLM      | AR       | 1/800      |
|   | Fanconi anemia, Group J                                      | BRIP1    |          |            |
| Bartter 氏症候群                                    | Bartter syndrome, Type 4a                                    | BSND     | AR       | 1/500      |
| 生物素酵素缺乏症  | Biotinidase Deficiency                                       | BTD      | AR       | 1/124      |
|   | Isolated growth hormone deficiency, Type III, X-linked       | BTK      |          |            |
|   | Desbuquois dysplasia 1                                       | CANT1    |          |            |
| 肢帶型肌肉失養症-2A型                                    | Limb-Girdle Muscular Dystrophy, Type 2A                      | CAPN3    | AR       | <1/500     |
| 兒茶酚胺敏感性多形性室性心博過速 (CPVT)-CASQ2 型                 | Catecholaminergic polymorphic ventricular tachycardia        | CASQ2    | AR       | 1/224      |
| 高胱胺酸尿症  | Homocystinuria, CBS-Related                                  | CBS      | AR       | 1/224      |
|   | Mental retardation, autosomal recessive 3                    | CC2D1A   |          |            |
| 尤塞氏綜合症-1D 型                                     | Usher Syndrome, Type 1D                                      | CDH23    | AR       | 1/285      |
| Bardet-Biedl 氏症候群-14 型                          | Leber Congenital Amaurosis, Type CEP290                      | CEP290   | AR       | 1/190      |
| 視網膜色素變性-26 型                                    | Retinitis Pigmentosa 26                                      | CERKL    | AR       | 1/148      |
| 囊狀纖維化   | Cystic Fibrosis  | CFTR     | AR       | 1/32       |
| 脈絡膜缺失症  | Choroideremia, X-Linked                                      | CHM      | X-Linked | 1/25,000   |
| 先天性肌無力症候群-CHRNE 型                               | Congenital Myasthenic Syndrome, CHRNE-Related                | CHRNE    | AR       | 1/408      |
| 多發性翼狀膜症候群                                       | Escobar Syndrome   | CHRNA3   | AR       | <1/500     |

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| 第二型裸淋巴細胞綜合症                | Bare Lymphocyte Syndrome, CIITA-Related                        | CIITA   | AR       | <1/500    |
| 神經元蠟樣脂褐質沉著疾病<br>-CLN3 型    | Ceroid Lipofuscinosis, Neuronal, 3                             | CLN3    | AR       | 1/230     |
| 神經元蠟樣脂褐質沉著疾病<br>-CLN5 型    | Ceroid Lipofuscinosis, Neuronal, 5                             | CLN5    | AR       | <1/500    |
| 神經元蠟樣脂褐質沉著疾病<br>-CLN6 型    | Ceroid Lipofuscinosis, Neuronal, 6                             | CLN6    | AR       | <1/500    |
| 神經元蠟樣脂褐質沉著疾病<br>-CLN8 型    | Ceroid Lipofuscinosis, Neuronal, 8 (a.k.a. Northern Epilepsy)  | CLN8    | AR       | <1/500    |
| 尤塞氏綜合症-3A 型                | Usher Syndrome, Type 3   | CLRN1   | AR       | 1/500     |
|                            | Achromatopsia, CNGA3-Related                                   | CNGA3   |          |           |
| 色彩感應失能症                    | Achromatopsia, CNGB3-Related                                   | CNGB3   | AR       | 1/87      |
|                            | Fibrochondrogenesis type 2                                     | COL11A2 |          |           |
| 艾柏症候群-COL4A3型              | Alport Syndrome, COL4A3-Related                                | COL4A3  | AR       | 1/267     |
| 艾柏症候群-COL4A4型              | Alport Syndrome, COL4A4-Related                                | COL4A4  | AR       | 1/267     |
| 艾柏症候群-COL4A5型              | Alport Syndrome, X-Linked                                      | COL4A5  | X-Linked | 1/139     |
| 表皮分解性水皰症                   | Dystrophic Epidermolysis Bullosa, COL7A1-Related               | COL7A1  | AR       | 1/196     |
| 甲西先磷酸合成酶缺乏症                | Carbamoyl Phosphate Synthetase I Deficiency                    | CPS1    | AR       | 1/570     |
| 肉鹼結合酶缺乏症第一型                | Carnitine Palmitoyltransferase IA Deficiency                   | CPT1A   | AR       | 1/354     |
| 肉鹼結合酶缺乏症第二型                | Carnitine Palmitoyltransferase II Deficiency                   | CPT2    | AR       | <1/500    |
| 萊伯氏先天性黑矇症-8型; 色素性視網膜炎-12 型 | Leber congenital amaurosis 8                                   | CRB1    | AR       | 1/104     |
| 胱胺酸血症                      | Cystinosis   | CTNS    | AR       | 1/158     |
|                            | Papillon-Lefevre Syndrome                                      | CTSC    |          |           |
|                            | Ceroid Lipofuscinosis, Neuronal, 10 (CLN10 Disease)            | CTSD    |          |           |
| 緻密性成骨不全症                   | Pycnodysostosis  | CTSK    | AR       | <1/500    |
| 慢性肉芽腫病                     | Chronic Granulomatous Disease, CYBA-Related                    | CYBA    | AR       | 1/224     |
| 性聯遺傳慢性肉芽腫病                 | Chronic Granulomatous Disease, X-Linked                        | CYBB    | X-Linked | 1/149,254 |
| 先天性腎上腺增生症<br>-11b 羥化酶缺乏症   | Congenital Adrenal Hyperplasia, 11-beta-hydroxylase-deficient  | CYP11B1 | AR       | 1/158     |
| 皮質酮甲基氧化酶缺乏症                | Corticosterone Methyloxidase Deficiency                        | CYP11B2 | AR       | <1/500    |
| 先天性腎上腺增生症<br>-17a 羥化酶缺乏症   | Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficient | CYP17A1 | AR       | 1/500     |
| 芳香環轉化酶缺乏症                  | Aromatase Deficiency   | CYP19A1 | AR       | <1/500    |
| 原發先天性青光眼                   | Primary Congenital Glaucoma                                    | CYP1B1  | AR       | 1/50      |
| 先天性腎上腺增生症<br>-21 羥化酶缺乏症    | Congenital Adrenal Hyperplasia, 21-hydroxylase-deficient       | CYP21A2 | AR       | 1/61      |
| 腦髓性黃瘤症                     | Cerebrotendinous Xanthomatosis                                 | CYP27A1 | AR       | 1/500     |
|                            | Vitamin D-dependent rickets type 1A                            | CYP27B1 |          |           |
| 楓糖尿症-II 型                  | Maple Syrup Urine Disease, Type 2                              | DBT     | AR       | 1/481     |
| 嚴重複合型免疫缺乏症伴隨離子輻射           | Severe Combined Immunodeficiency, Type Athabaskan              | DCLRE1C | AR       | <1/500    |
|                            | Xeroderma Pigmentosum Group E                                  | DDB2    |          |           |
| Smith-Lemli-Opitz 症候群      | Smith-Lemli-Opitz Syndrome                                     | DHCR7   | AR       | 1/30      |
| 視網膜色素變性-59 型               | Retinitis Pigmentosa 59  | DHDDS   | AR       | 1/296     |
|                            | Dyskeratosis congenita, X-Linked                               | DKC1    |          |           |
| 二氫硫辛醯胺脫氫酶(E3)缺乏症           | Dihydrolipoamide Dehydrogenase Deficiency                      | DLD     | AR       | 1/500     |
| 裘馨氏肌肉失養症                   | Duchenne/Becker Muscular Dystrophy                             | DMD     | X-Linked | 1/2,350   |
| 原發性纖毛運動障礙<br>-DNAH5 型      | Ciliary Dyskinesia, Primary 3                                  | DNAH5   | AR       | 1/142     |
| 原發性纖毛運動障礙<br>-DNAI1 型      | Ciliary Dyskinesia, Primary 1                                  | DNAI1   | AR       | 1/230     |
| 原發性纖毛運動障礙<br>-DNAI2 型      | Ciliary Dyskinesia, Primary 9                                  | DNAI2   | AR       | 1/447     |
| 原發性纖毛運動障礙<br>-DNAL1 型      | Ciliary Dyskinesia, Primary, 16                                | DNAL1   | AR       | <1/500    |
|                            | Congenital Myasthenic Syndrome, DOK7-Related                   | DOK7    |          |           |
| 二氫嘧啶脫氫酶缺乏症                 | Dihydropyrimidine Dehydrogenase Deficiency                     | DPYD    | AR       | <1/500    |
| 肢帶型肌肉失養症-2B型               | Limb-Girdle Muscular Dystrophy, Type 2B                        | DYSF    | AR       | <1/500    |
| 汗性外胚層增生不良症                 | Hypohidrotic Ectodermal Dysplasia, X-Linked                    | EDA     | X-Linked | 1/14,167  |
|                            | Hypohidrotic Ectodermal Dysplasia                              | EDAR    |          |           |
| Wolcott-Rallison 症候群       | Wolcott-Rallison Syndrome                                      | EIF2AK3 | AR       | <1/500    |
| 腦白質病伴隨白質消失症                | Leukoencephalopathy with Vanishing White Matter                | EIF2B5  | AR       | <1/500    |
|                            | Dysautonomia, familial (IKBKAP or ELP1)                        | IKBKAP  |          |           |
| 肌失養症                       | Emery-Dreifuss Muscular Dystrophy 1, X-Linked                  | EMD     | X-Linked | 1/81,967  |
| 著色性乾皮症                     | Xeroderma Pigmentosum Group D                                  | ERCC2   |          |           |
| 著色性乾皮症                     | Xeroderma Pigmentosum Group B                                  | ERCC3   |          |           |
| 著色性乾皮症                     | Xeroderma Pigmentosum Group F                                  | ERCC4   |          |           |
| 著色性乾皮症                     | Xeroderma pigmentosum Group G                                  | ERCC5   |          |           |
| B 型柯凱因氏症候群                 | Cockayne syndrome, type B                                      | ERCC6   | AR       | 1/500     |
| A 型柯凱因氏症候群                 | Cockayne syndrome, type A                                      | ERCC8   | AR       | 1/822     |
| Roberts 症候群                | Roberts Syndrome   | ESCO2   | AR       | <1/500    |
| 戊二酸尿症-IIA 型                | Glutaric Acidemia, Type 2A                                     | ETFPA   | AR       | 1/500     |
| 戊二酸尿症-IIB 型                | Glutaric Acidemia, Type 2B                                     | ETFB    | AR       | 1/500     |
| 戊二酸尿症-IIC 型                | Glutaric Acidemia, Type 2C                                     | ETFDH   | AR       | 1/250     |
| 乙基丙二酸腦病變                   | Ethylmalonic Encephalopathy                                    | ETHE1   | AR       | <1/500    |
| 埃利偉氏症候群-EVC型               | Ellis-van Creveld Syndrome, EVC-Related                        | EVC     | AR       | 1/142     |
| 埃利偉氏症候群-EVC2型              | Ellis-van Creveld Syndrome, EVC2-related                       | EVC2    | AR       | 1/240     |
| 橋腦小腦發育不全-1B型               | Pontocerebellar Hypoplasia, Type 1B                            | EXOSC3  | AR       | <1/500    |
| 視網膜色素變性-25 型               | Retinitis Pigmentosa 25  | EYS     | AR       | 1/66      |
| 第十一凝血因子缺乏症                 | Factor XI deficiency   | F11     | AR       | 1/500     |
|                            | Prothrombin deficiency   | F2      |          |           |
| 血友病 A 型                    | Hemophilia A   | F8      | X-Linked | 1/3,250   |
| 血友病 B 型                    | Hemophilia B   | F9      | X-Linked | 1/15,000  |

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| 高酪胺酸血症-1 型                                     | Tyrosinemia, Type I                                    | FAH     | AR       | 1/99     |
| 視網膜色素變性-28 型                                   | Retinitis Pigmentosa 28                                | FAM161A | AR       | 1/296    |
| Fanconi 氏貧血 A 型                                | Fanconi Anemia, Group A                                | FANCA   | AR       | 1/239    |
| Fanconi 氏貧血 C 型                                | Fanconi Anemia, Group C                                | FANCC   | AR       | 1/535    |
| Fanconi 氏貧血 G 型                                | Fanconi Anemia, Group G                                | FANCG   | AR       | 1/632    |
| 延胡索酸缺乏症  | Fumarase Deficiency                                    | FH      | AR       | <1/500   |
| 肌肉失養症醣基化功能缺陷-FKRP 型                            | Limb-Girdle Muscular Dystrophy, Type 2I                | FKRP    | AR       | 1/158    |
| 肌肉失養症醣基化功能缺陷-FKTN 型                            | Walker-Warburg Syndrome, FKTN-Related                  | FKTN    | AR       | <1/500   |
| 肝醣儲積症第 1A 型                                    | Glycogen Storage Disease, Type IA                      | G6PC    | AR       | 1/177    |
| 葡萄糖-六-磷酸鹽去氫酶缺乏症<br>(蠶豆症)                       | Glucose-6-Phosphate Dehydrogenase Deficiency*          | G6PD    | X-Linked | 1/7      |
| 龐貝氏症(肝醣儲積症第二型)                                 | Glycogen Storage Disease, Type II (Pompe Disease)      | GAA     | AR       | 1/100    |
| Krabbe 氏症(球細胞腦白質失養症)                           | Krabbe Disease   | GALC    | AR       | 1/158    |
| 半乳糖異構酶缺乏症                                      | Galactose epimerase deficiency                         | GALE    | AR       | <1/500   |
| 半乳糖激酶缺乏症                                       | Galactokinase Deficiency (Galactosemia, Type II)       | GALK1   | AR       | 1/110    |
| 黏多醣症-4A 型(莫奎歐氏症)                               | Mucopolysaccharidosis, Type IVA                        | GALNS   | AR       | 1/224    |
|  | Hyperphosphatemic familial tumoral calcinosis          | GALNT3  |          |          |
| 半乳糖血症  | Galactosemia   | GALT    | AR       | 1/110    |
| 胍基乙酸鹽甲基轉移酶缺乏症                                  | Guanidinoacetate Methyltransferase Deficiency          | GAMT    | AR       | 1/371    |
| 高雪氏症   | Gaucher Disease  | GBA     | AR       | 1/77     |
| 肝醣儲積症第四型                                       | Glycogen Storage Disease, Type IV                      | GBE1    | AR       | 1/387    |
| 戊二酸血症-I 型                                      | Glutaric Acidemia, Type 1                              | GCDH    | AR       | 1/87     |
|  | Dopa-responsive dystonia                               | GCH1    |          |          |
|  | Grebe syndrome   | GDF5    |          |          |
| 氧化磷酸化缺乏症-GFM1 型                                | Combined Oxidative Phosphorylation Deficiency 1        | GFM1    | AR       | <1/500   |
|  | Isolated growth hormone deficiency, Type IA/II         | GH1     |          |          |
|  | Isolated growth hormone deficiency, Type IB            | GHRHR   |          |          |
| 性聯遺傳進行性神經性腓骨萎縮症-1 型                            | Charcot-Marie-Tooth Disease with Deafness, X-Linked    | GJB1    | X-Linked | 1/667    |
| 非症候群型遺傳性聽障<br>-GJB2 型                          | Non-Syndromic Hearing Loss (a.k.a. Connexin 26)        | GJB2    | AR       | 1/42     |
|  | Erythrokeratoderma variabilis et progressiva           | GJB3    |          |          |
| 非症候群型遺傳性聽障<br>-GJB6 型                          | Non-Syndromic Hearing Loss (a.k.a. Connexin 30)        | GJB6    | AR       | 1/423    |
| 法布瑞氏症  | Fabry Disease  | GLA     | X-Linked | 1/25,000 |
| GLB1 相關疾病：<br>- 黏多醣症-4B 型(莫奎歐氏症)<br>- 神經節苷脂儲積症 | Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis   | GLB1    | AR       | 1/134    |
| 非酮性高甘胺酸血症<br>-GLDC 型                           | Glycine Encephalopathy, GLDC-Related                   | GLDC    | AR       | 1/193    |
| 致死先天性攣縮綜合症                                     | Lethal Congenital Contracture Syndrome 1               | GLE1    | AR       | <1/500   |
| 包涵體肌肉病變  | Inclusion Body Myopathy 2                              | GNE     | AR       | <1/500   |
| 黏脂症第二型和第三型-alpha/beta                          | Mucopolysaccharidosis II/IIIA                          | GNPTAB  | AR       | <1/500   |
| 黏脂症第三型-gamma                                   | Mucopolysaccharidosis III gamma                        | GNPTG   | AR       | <1/500   |
| 黏多醣症-3D 型(聖菲利柏氏症)                              | Mucopolysaccharidosis, Type IIID (Sanfilippo D)        | GNS     | AR       | 1/500    |
|  | Geroderma osteodysplastica                             | GORAB   |          |          |
| 巨大血小板症候群-A1 型                                  | Bernard-Soulier Syndrome, Type A2                      | GP1BA   | AR       | 1/500    |
|  | Bernard-Soulier Syndrome, Type B                       | GP1BB   |          |          |
| 巨大血小板症候群-C 型                                   | Bernard-Soulier Syndrome, Type C                       | GP9     | AR       | 1/500    |
| 原發性高草酸鹽尿症-2 型                                  | Primary Hyperoxaluria, Type 2                          | GRHPR   | AR       | <1/500   |
|  | Leber congenital amaurosis 1                           | GUCY2D  |          |          |
| 黏多醣症-7 型                                       | Mucopolysaccharidosis, Type VII                        | GUSB    | AR       | 1/250    |
| 三功能蛋白缺乏症及長鏈 3-羥醯輔酶 A 脫氫酶缺乏症                    | Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency  | HADHA   | AR       | <1/500   |
|  | Trifunctional protein deficiency                       | HADHB   |          |          |
| 嚴重先天性中性粒細胞減少症<br>-HAX1 型                       | Congenital Neutropenia, HAX1-Related                   | HAX1    | AR       | 1/224    |
| 甲型海洋性貧血  | Alpha-Thalassemia                                      | HBA1    |          |          |
| 甲型海洋性貧血  | Alpha-Thalassemia                                      | HBA2    |          |          |
| 鐮刀型紅血球症;乙型海洋性貧血;血紅素 C 疾病                       | Beta-Hemoglobinopathies                                | HBB     | AR       | 1/158    |
| 戴薩克斯症  | Tay-Sachs Disease                                      | HEXA    | AR       | 1/300    |
| Sandoff 症<br>(成年型 GM2 神經節苷脂儲積症)                | Sandhoff Disease                                       | HEXB    | AR       | 1/600    |
| 血鐵沉著症  | Hemochromatosis, Type 1                                | HFE     |          |          |
| 血鐵沉著症  | Hemochromatosis, Type 2A                               | HFE2    |          |          |
| 黑尿症  | Alkaptonuria   | HGD     | AR       | 1/250    |
| 黏多醣症-3C 型(聖菲利柏氏症)                              | Mucopolysaccharidosis, Type IIIC (Sanfilippo C)        | HGSNAT  | AR       | 1/434    |
| 多發性羧化酶缺乏症                                      | Holocarboxylase Synthetase Deficiency                  | HLCS    | AR       | 1/500    |
| 3-羥基-3-甲基戊二酸血症                                 | 3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency | HMGCL   | AR       | <1/500   |
|  | Heme Oxygenase-1 Deficiency                            | HMOX1   |          |          |
| 原發性高草酸鹽尿症-3 型                                  | Primary Hyperoxaluria, Type 3                          | HOGA1   | AR       | 1/184    |
|  | Tyrosinemia, Type III                                  | HPD     |          |          |
| Hermansky-Pudlak 症候群-1 型                       | Hermansky-Pudlak Syndrome 1                            | HPS1    | AR       | 1/354    |
| Hermansky-Pudlak 症候群-3 型                       | Hermansky-Pudlak Syndrome 3                            | HPS3    | AR       | 1/354    |
|  | Hermansky-Pudlak syndrome 4                            | HPS4    |          |          |
|  | 17-beta hydroxysteroid dehydrogenase 3 deficiency      | HSD17B3 |          |          |
| D-雙功能蛋白缺乏症                                     | D-Bifunctional Protein Deficiency                      | HSD17B4 | AR       | 1/158    |
| 先天性腎上腺增生症<br>-3b 羥化酶缺乏症                        | 3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency | HSD3B2  | AR       | <1/500   |
| Hydrolethalus 症候群                              | Hydrolethalus Syndrome                                 | HYLS1   | AR       | <1/500   |
| 黏多醣症-2 型(韓特氏症)                                 | Mucopolysaccharidosis, Type II (Hunter Syndrome)       | IDS     | X-Linked | 1/50,000 |
| 黏多醣症-1 型(賀勒氏症)                                 | Mucopolysaccharidosis, Type I (Hurler Syndrome)        | IDUA    | AR       | <1/500   |
| 性聯遺傳嚴重複合型免疫缺乏症                                 | Severe Combined Immunodeficiency, X-Linked             | IL2RG   | X-Linked | 1/25,000 |
|  | Glanzmann thrombasthenia                               | ITGB3   |          |          |

| 疾病列表                                       | 疾病列表   | 基因      | 遺傳模式     | 帶因率       |
|--|--|---------|----------|-----------|
| 異戊酸血症                                      | Isovaleric Acidemia  | IVD     | AR       | 1/167     |
| 永久性新生兒糖尿病                                  | Congenital Hyperinsulinism, KCNJ11-Related                   | KCNJ11  | AR       | 1/423     |
| 肌肉失養症-LAMA2 型                              | LAMA2-related Muscular Dystrophy                             | LAMA2   | AR       | <1/500    |
| 接合型表皮溶解水皰症-LAMA3 型                         | Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related      | LAMA3   | AR       | 1/781     |
| 接合型表皮溶解水皰症-LAMB3 型                         | Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related      | LAMB3   | AR       | 1/781     |
| 接合型表皮溶解水皰症-LAMC2 型                         | Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related      | LAMC2   | AR       | 1/781     |
| 萊伯氏先天性黑矇症-5型                               | Leber Congenital Amaurosis, Type LCA5                        | LCA5    | AR       | 1/500     |
|  | Familial Hypercholesterolemia, LDLR-Related                  | LDLR    |          |           |
|  | Familial Hypercholesterolemia, LDLRAP1-Related               | LDLRAP1 |          |           |
|  | Leydig cell hypoplasia                                       | LHCGR   |          |           |
| Stuve-Wiedemann 症候群                        | Stuve-Wiedemann Syndrome                                     | LIFR    | AR       | <1/500    |
| 溶酶體酸性脂肪酶缺乏症                                | Lysosomal Acid Lipase Deficiency                             | LIPA    | AR       | <1/500    |
|  | Woolly Hair/Hypotrichosis Syndrome                           | LIPH    |          |           |
| 非症候群型遺傳性聽障<br>-LOXHD1 型                    | Deafness, Autosomal Recessive 77                             | LOXHD1  | AR       | 1/500     |
|  | Lipoprotein Lipase Deficiency                                | LPL     |          |           |
| Leigh 症候群-complex IV (COX) 酶缺乏             | Leigh Syndrome, French-Canadian Type                         | LRPPRC  | AR       | 1/447     |
| Chediak-Higashi 症候群                        | Chediak-Higashi syndrome                                     | LYST    | AR       | <1/500    |
| 甘露糖症                                       | Alpha-Mannosidosis   | MAN2B1  | AR       | 1/354     |
|  | Hypermethioninemia   | MAT1A   |          |           |
| 三甲基巴豆醯輔酶 A 羧化酶缺乏症                          | 3-Methylcrotonyl-CoA Carboxylase 1 Deficiency                | MCCC1   | AR       | 1/95      |
| 三甲基巴豆醯輔酶 A 羧化酶缺乏症                          | 3-Methylcrotonyl-CoA Carboxylase 2 Deficiency                | MCCC2   | AR       | 1/95      |
| 黏脂症第四型                                     | Mucopolysaccharidosis, Type IV                               | MCOLN1  | AR       | 1/300     |
|  | RETT Syndrome  | MECP2   |          |           |
| 產後進展性小頭畸形伴隨癲癇及腦萎縮                          | Microcephaly, postnatal progressive, with seizures and brain | MED17   | AR       | <1/500    |
| 家族性地中海熱病                                   | Familial Mediterranean Fever                                 | MEFV    | AR       | 1/20      |
| 脊椎肋骨發育不全                                   | Spondylothoracic Dysostosis, MESP2-Related                   | MESP2   | AR       | <1/500    |
| 神經元蠟樣脂褐質沉著疾病<br>-MFSD8 型                   | Ceroid Lipofuscinosis, Neuronal, 7                           | MFSD8   | AR       | <1/500    |
|  | Bardet-Biedl Syndrome 6                                      | MKKS    |          |           |
| Joubert 氏症候群-28型                           | Meckel-Gruber Syndrome, Type 1                               | MKS1    | AR       | 1/260     |
| 巨腦性腦白質病伴有皮層下囊腫第一型                          | Megalencephalic Leukoencephalopathy with Subcortical Cy      | MLC1    | AR       | <1/500    |
|  | Malonyl-CoA decarboxylase deficiency                         | MLYCD   |          |           |
| 甲基丙二酸血症-cb1A型                              | Methylmalonic Aciduria, MMAA-Related                         | MMAA    | AR       | 1/301     |
| 甲基丙二酸血症-cb1B型                              | Methylmalonic Aciduria, MMAB-Related                         | MMAB    | AR       | 1/435     |
| 甲基丙二酸血症併高胱氨酸血症-cb1C 型                      | Methylmalonic Aciduria and Homocystinuria, Type cb1C         | MMACHC  | AR       | 1/134     |
| 甲基丙二酸血症併高胱氨酸血症-cb1D 型                      | Methylmalonic Aciduria and Homocystinuria, Type cb1D         | MMADHC  | AR       | <1/500    |
|  | Molybdenum cofactor deficiency                               | MOCS1   |          |           |
| 先天性醣基化疾病-1b型                               | Congenital Disorder of Glycosylation, Type 1B                | MPI     | AR       | <1/500    |
| 先天性無巨型細胞性血小板缺乏症                            | Congenital Amegakaryocytic Thrombocytopenia                  | MPL     | AR       | 1/102     |
| 肝腦病變型粒線體DNA 耗竭症候群<br>-MPV17 型              | Hepatocerebral Mitochondrial DNA Depletion Syndrome, M       | MPV17   | AR       | <1/500    |
|  | Ataxia-telangiectasia-like disorder 1                        | MRE11   |          |           |
|  | Homocystinuria due to Deficiency of MTHFR                    | MTHFR   |          |           |
| 性聯遺傳肌小管病變                                  | Myotubular Myopathy, X-Linked                                | MTM1    | X-Linked | 1/25,000  |
| 高胱氨酸血症-巨母紅血球性貧血<br>-cobalamin E 型          | Homocystinuria, Type cb1E                                    | MTRR    | AR       | <1/500    |
| 無β脂蛋白血症                                    | Abetalipoproteinemia   | MTTP    | AR       | <1/500    |
| 甲基丙二酸血症-MUT型                               | Methylmalonic Aciduria, Type mut(0)                          | MUT     | AR       | 1/195     |
| 聽損   | Deafness, autosomal recessive, 3                             | MYO15A  |          |           |
| 非症候群型遺傳性聽障<br>-MYO7A 型; 尤塞氏綜合症 1B 型        | Usher Syndrome, Type 1B                                      | MYO7A   | AR       | 1/206     |
| 黏多糖症-3B 型(聖菲利柏氏症)                          | Mucopolysaccharidosis, Type III B (Sanfilippo B)             | NAGLU   | AR       | <1/500    |
| N-乙酰穀胺酸合成酶缺乏症                              | N-acetylglutamate Synthase Deficiency                        | NAGS    | AR       | <1/500    |
| Nijmegen 斷裂症候群                             | Nijmegen Breakage Syndrome                                   | NBN     | AR       | 1/158     |
| 進行性神經性腓骨萎縮症-4D 型                           | Charcot-Marie-Tooth Disease type 4D                          | NDRG1   | AR       | 1/22      |
| 粒線體 Complex I 缺乏症(Leigh 症候<br>群)-NDUFAF5 型 | Mitochondrial Complex I Deficiency, NDUFAF5-Related          | NDUFAF5 | AR       | 1/447     |
|  | Mitochondrial complex I deficiency                           | NDUFS4  |          |           |
| 粒線體 Complex I 缺乏症(Leigh 症候<br>群)-NDUFAF6 型 | Mitochondrial Complex I Deficiency, NDUFS6-Related           | NDUFS6  | AR       | <1/500    |
| 線狀體肌肉病變                                    | Nemaline Myopathy, NEB-Related                               | NEB     | AR       | 1/112     |
|  | Sialidosis   | NEU1    |          |           |
|  | Hydatidiform Mole, Recurrent                                 | NLRP7   |          |           |
| 尼曼匹克症-C1 型                                 | Niemann-Pick Disease, Type C1/D                              | NPC1    | AR       | 1/194     |
| 尼曼匹克症-C2 型                                 | Niemann-Pick Disease, Type C2                                | NPC2    | AR       | 1/194     |
| Joubert 氏症候群-4 型                           | Juvenile Nephronophthisis                                    | NPHP1   | AR       | 1/480     |
| 先天性腎病症候群-1 型                               | Congenital Finnish Nephrosis                                 | NPHS1   | AR       | 1/289     |
| 先天性腎病症候群-2 型                               | Steroid-Resistant Nephrotic Syndrome                         | NPHS2   | AR       | 1/289     |
| 先天性腎上腺增生症-X染色體遺傳                           | Congenital Adrenal Hypoplasia, X-linked                      | NR0B1   | X-Linked | 1/6,250   |
| 網膜色素變性-37 型                                | Enhanced S-Cone Syndrome                                     | NR2E3   | AR       | 1/209     |
| 先天性痛覺不敏感合併無汗症                              | Congenital Insensitivity to Pain with Anhidrosis (CIPA)      | NTRK1   | AR       | <1/500    |
| 鳥胺酸酮酸轉胺酶缺乏症                                | Ornithine Aminotransferase Deficiency                        | OAT     | AR       | <1/500    |
| Lowe 氏症候群                                  | Lowe syndrome, X-Linked                                      | OCRL    | X-Linked | 1/250,000 |
| Costeff 症候群                                | Costeff Syndrome (3-Methylglutaconic Aciduria, Type 3)       | OPA3    | AR       | <1/500    |
| 鳥胺酸氨甲醯基轉移酶缺乏症                              | Ornithine Transcarbamylase Deficiency                        | OTC     | X-Linked | 1/7,000   |
| 苯酮尿症(苯丙氨酸羧化酶缺乏症)                           | Phenylketonuria  | PAH     | AR       | 1/93      |
| 泛酸鹽激活酵素關聯之神經退化性疾病                          | Pantothenate Kinase-Associated Neurodegeneration             | PANK2   | AR       | 1/289     |
| 丙酮酸羧化酶缺乏症                                  | Pyruvate Carboxylase Deficiency                              | PC      | AR       | 1/250     |
| 丙酸血症-PCCA 型                                | Propionic Acidemia, PCCA-Related                             | PCCA    | AR       | 1/224     |
| 丙酸血症-PCCB 型                                | Propionic Acidemia, PCCB-Related                             | PCCB    | AR       | 1/224     |

| 疾病列表  | 疾病列表  | 基因       | 遺傳模式     | 帶因率        |
|---|---|----------|----------|------------|
| 非症候群型遺傳性聽障<br>-PCDH15 型; 尤塞氏綜合症 1F 型                | Usher Syndrome, Type 1F                                   | PCDH15   | AR       | 1/395      |
| 丙酮酸鹽脫氫酶E1-alpha 缺乏症                                 | Pyruvate Dehydrogenase Deficiency, X-Linked               | PDHA1    | X-Linked | <1/250,000 |
| 丙酮酸鹽脫氫酶 E1-beta 缺乏症                                 | Pyruvate Dehydrogenase Deficiency, PDHB-Related           | PDHB     | AR       | <1/500     |
|   | Prolidase deficiency                                      | PEPD     |          |            |
|   | Cytochrome-c oxidase deficiency                           | PET100   |          |            |
| 柴爾維格氏症候群<br>-PEX1 型                                 | Peroxisome Biogenesis Disorder 1A (Zellweger)             | PEX1     | AR       | 1/147      |
| 柴爾維格氏症候群<br>-PEX10 型                                | Peroxisome Biogenesis Disorder 6A (Zellweger)             | PEX10    | AR       | 1/500      |
| 柴爾維格氏症候群<br>-PEX12 型                                | Peroxisome Biogenesis Disorder 3A (Zellweger)             | PEX12    | AR       | 1/373      |
| 柴爾維格氏症候群<br>-PEX2 型                                 | Peroxisome Biogenesis Disorder 5A (Zellweger)             | PEX2     | AR       | 1/500      |
| 柴爾維格氏症候群<br>-PEX6 型                                 | Peroxisome Biogenesis Disorder 4A (Zellweger)             | PEX6     | AR       | 1/280      |
| 肢近端型點狀軟骨發育不良第一型                                     | Rhizomelic Chondrodysplasia Punctata, Type 1              | PEX7     | AR       | 1/158      |
| 肝醣儲積症第七型  | Glycogen Storage Disease, Type VII                        | PFKM     | AR       | <1/500     |
| 磷酸脫氫酶缺乏症  | Phosphoglycerate Dehydrogenase Deficiency                 | PHGDH    | AR       | <1/500     |
|   | Multiple congenital anomalies-hypotonia-seizures syndrom  | PIGN     |          |            |
| 隱性多囊性腎疾病<br>-PKHD1 型                                | Polycystic Kidney Disease, Autosomal Recessive            | PKHD1    | AR       | 1/70       |
| 新生兒神經軸發育不良  | Infantile neuroaxonal dystrophy 1                         | PLA2G6   | AR       | 1/500      |
| 先天性醣基化疾病-1a型  | Congenital Disorder of Glycosylation, Type 1A, PMM2-Relat | PMM2     | AR       | <1/500     |
|   | Pyridoxal 5'-phosphate-dependent epilepsy                 | PNPO     |          |            |
| 漸進性外眼肌麻痺  | POLG-Related Disorders                                    | POLG     | AR       | 1/113      |
|   | Xeroderma pigmentosum Variant                             | POLH     |          |            |
| 肌肉失養症醣基化功能缺陷-網膜色素變性 76型                             | Muscle-Eye-Brain Disease, POMGNT1-Related                 | POMGNT1  | AR       | 1/462      |
|   | Cytochrome P450 oxidoreductase deficiency                 | POR      |          |            |
| 神經元蠟樣脂褐質沉著疾病<br>-PPT1 型                             | Ceroid Lipofuscinosis, Neuronal, 1                        | PPT1     | AR       | 1/368      |
|   | Myasthenic syndrome, congenital, 22                       | PREPL    |          |            |
| 下垂體激素缺乏症-2 型  | Combined Pituitary Hormone Deficiency 2                   | PROP1    | AR       | 1/45       |
| Arts 症候群; Rosenberg-Chutorian症候群;<br>PRPS1 基因相關症候群  | Arts syndrome, X-Linked                                   | PRPS1    | X-Linked | <1/250,000 |
| saposin-b 缺乏引發異染性白質退化症                              | Metachromatic Leukodystrophy, PSAP-Related                | PSAP     | AR       | <1/500     |
| 四氫基喋呤缺乏症  | 6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency    | PTS      | AR       | 1/354      |
| 粒線體病變合併鐵粒幼<br>紅細胞性貧血                                | Mitochondrial Myopathy and Sideroblastic Anemia (MLASA)   | PUS1     | AR       | <1/500     |
| 肝醣儲積症第五型  | Glycogen Storage Disease, Type V (McArdle Disease)        | PYGM     | AR       | <1/500     |
| Carpenter 症候群                                       | Carpenter Syndrome  | RAB23    | AR       | <1/500     |
| 歐門氏症候群-RAG1 型                                       | Omenn Syndrome, RAG1-Related                              | RAG1     | AR       | 1/137      |
| 歐門氏症候群-RAG2 型                                       | Omenn Syndrome, RAG2-Related                              | RAG2     | AR       | 1/137      |
| 先天性肌無力症候群-RAPSN 型                                   | Congenital Myasthenic Syndrome, RAPSN-Related             | RAPSN    | AR       | <1/500     |
| 橋腦小腦發育不全-6 型  | Pontocerebellar Hypoplasia, Type 1 and 6, RARS2-Related   | RARS2    | AR       | <1/500     |
| 萊伯氏先天性黑矇症-13 型                                      | Leber Congenital Amaurosis, Type RDH12                    | RDH12    | AR       | <1/500     |
|   | Retinal Dystrophies, RLBP1-Associated                     | RLBP1    |          |            |
| 軟骨頭髮發育不全; 幹骨后端的軟骨生成; 骨骼發育異常症<br>候群                  | Cartilage-Hair Hypoplasia                                 | RMRP     | AR       | <1/500     |
|   | Aicardi-Goutieres syndrome, RNASEH2C-related              | RNASEH2C |          |            |
| 萊伯氏先天性黑矇症-2型; 色素性視網膜炎-20 型                          | Leber Congenital Amaurosis 2                              | RPE65    | AR       | 1/228      |
| Meckel 症候群; Joubert 氏症候群                            | Ciliopathies, RPGRIP1L-Related                            | RPGRIP1L | AR       | 1/259      |
| 性聯遺傳視網膜裂損症/視覺黃斑症                                    | Juvenile Retinoschisis, X-Linked                          | RS1      | X-Linked | 1/2,500    |
| 先天性角化不全症-5 型  | Dyskeratosis Congenita, RTEL1-Related                     | RTEL1    | AR       | 1/500      |
| Autosomal Recessive遺傳痙攣性共濟失調症                       | Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay | SACS     | AR       | <1/500     |
| MIRAGE症候群   | MIRAGE syndrome   | SAMD9    |          |            |
| Aicardi-Goutieres 症候群                               | Aicardi-Goutières Syndrome                                | SAMHD1   | AR       | <1/500     |
|   | Shwachman-Diamond syndrome                                | SBDS     |          |            |
| 橋腦小腦發育不全-2D型  | Pontocerebellar Hypoplasia, Type 2D                       | SEPSECS  | AR       | <1/500     |
|   | Alpha-1-Antitrypsin Deficiency                            | SERPINA1 |          |            |
| 肢帶型肌肉失養症-2D型  | Limb-Girdle Muscular Dystrophy, Type 2D                   | SGCA     | AR       | <1/500     |
| 肢帶型肌肉失養症-2E型  | Limb-Girdle Muscular Dystrophy, Type 2E                   | SGCB     | AR       | 1/500      |
| 肢帶型肌肉失養症-2F型  | Limb-Girdle Muscular Dystrophy, Type 2F                   | SGCD     | AR       | <1/500     |
| 肢帶型肌肉失養症-2C型  | Limb-Girdle Muscular Dystrophy, Type 2C                   | SGCG     | AR       | 1/381      |
| 黏多醣症-3A 型(聖菲利柏氏症)                                   | Mucopolysaccharidosis, Type IIIA (Sanfilippo A)           | SGSH     | AR       | 1/454      |
| Gitelman 氏症候群                                       | Gitelman Syndrome   | SLC12A3  | AR       | 1/100      |
| 安德曼綜合症  | Agenesis of the Corpus Callosum with Peripheral Neuropat  | SLC12A6  | AR       | <1/500     |
| 唾液酸儲存症  | Salla Disease   | SLC17A5  | AR       | <1/500     |
|   | Megaloblastic Anemia Syndrome                             | SLC19A2  |          |            |
| 原發性肉鹼缺乏症  | Carnitine Deficiency                                      | SLC22A5  | AR       | 1/129      |
| Citrin 缺乏症  | Citrullinemia, Type II                                    | SLC25A13 | AR       | <1/500     |
| 高鳥氨酸-高血氨-高瓜胺酸綜合症候群(HHH症候群)                          | Hyperornithinemia-Hyperammonemia-Homocitrullinuria (H     | SLC25A15 | AR       | <1/500     |
| 肉鹼轉位酶缺乏症  | Carnitine-acylcarnitine translocase deficiency            | SLC25A20 | AR       | <1/500     |
| 軟骨生成不全症候群-1B 型; 畸型發育不良-II 型; 畸型發<br>育不良; 多發性骨骺發育不全症 | Achondrogenesis, Type 1B                                  | SLC26A2  | AR       | 1/158      |
| 先天性分泌性氯化物腹瀉   | Congenital Chloride Diarrhea                              | SLC26A3  | AR       | <1/500     |
| Pendred 氏症候群  | Pendred Syndrome  | SLC26A4  | AR       | 1/80       |
| 遠端關節攣縮,<br>智能障礙及癲癇症候群                               | Autism Spectrum, Epilepsy and Arthrogyriposis             | SLC35A3  | AR       | <1/500     |
| 肝醣儲積症第 1b 型   | Glycogen Storage Disease, Type IB                         | SLC37A4  | AR       | 1/158      |
| 腸道內鋅吸收不良症   | Acrodermatitis Enteropathica                              | SLC39A4  | AR       | <1/500     |
|   | Cystinuria, Type A  | SLC3A1   |          |            |

| 疾病列表  | 疾病列表  | 基因       | 遺傳模式         | 帶因率       |
|---|---|----------|--------------|-----------|
|   | Oculocutaneous albinism, Type 4                                     | SLC45A2  |              |           |
| 眼角膜內皮細胞失養症  | Corneal Dystrophy and Perceptive Deafness                           | SLC4A11  | AR           | <1/500    |
| 肌酸缺乏症候群   | Creatine Transporter Defect (Cerebral Creatine Deficiency Syndrome) | SLC6A8   | X-Linked     | 1/3,434   |
| Lysunyruc 蛋白質耐受不良症                                  | Lysinuric Protein Intolerance                                       | SLC7A7   | AR           | <1/500    |
|   | Cystinuria, Type B  | SLC7A9   |              |           |
| Schimke 免疫-骨發育不良                                    | Schimke Immunoosseous Dysplasia                                     | SMARCAL1 | AR           | 1/500     |
| 脊髓性肌肉萎縮症  | Spinal Muscular Atrophy   | SMN1     | AR           | 1/54      |
| 尼曼匹克症-A/B 型   | Niemann-Pick Disease, Types A/B                                     | SMPD1    | AR           | 1/250     |
|   | 5-alpha reductase deficiency  | SRD5A2   |              |           |
|   | GM3 synthase deficiency   | ST3GAL5  |              |           |
| 脂質先天性腎上腺發育不全  | Lipoid Congenital Adrenal Hyperplasia                               | STAR     | AR           | <1/500    |
|   | Deafness, autosomal recessive 16                                    | STRC     |              |           |
|   | Mitochondrial DNA depletion syndrome 5 (encephalomyopathy)          | SUCLA2   |              |           |
| 多發性硫酸脂酶缺乏症  | Multiple Sulfatase Deficiency                                       | SUMF1    | AR           | 1/500     |
| Leigh 症候群-SURF1型;<br>進行性神經性腓骨萎縮症-SURF1 型            | Leigh Syndrome  | SURF1    | AR           | <1/500    |
| 高酪胺血症-2 型   | Tyrosinemia, Type II  | TAT      | AR           | 1/250     |
| 骨質石化症-TCIRG1 型                                      | Osteopetrosis, Infantile Malignant, TCIRG1-Related                  | TCIRG1   | AR           | 1/250     |
| 痙攣性下身麻痺-49 型  | Hereditary Spastic Paraparesis, Type 49                             | TECPR2   | AR           | <1/500    |
| 血鐵沉積症-3 型   | Hemochromatosis, Type 3, TFR2-Related                               | TFR2     | AR           | <1/500    |
| 先天性魚鱗癬狀紅皮症  | Lamellar Ichthyosis, Type 1   | TGM1     | AR           | 1/224     |
| Segawa 症候群(瀨川氏病)                                    | Segawa Syndrome, TH-Related   | TH       | AR           | 1/224     |
|   | Deafness, autosomal dominant 36, autosomal recessive 7              | TMC1     |              |           |
| Joubert 氏症候群-2 型                                    | Joubert Syndrome 2 / Meckel Syndrome 2                              | TMEM216  | AR           | 1/141     |
| 甲狀腺素合成異常-TPO 型                                      | Congenital hypothyroidism   | TPO      | AR           | 1/373     |
| 神經元蠟樣脂褐質沉著疾病<br>-TPP1 型                             | Ceroid Lipofuscinosis, Neuronal, 2                                  | TPP1     | AR           | 1/252     |
|   | Aicardi-Goutieres syndrome, TREX1-related                           | TREX1    |              |           |
| 肢帶型肌肉失養症-2H型; Bardet-Biedl 氏症候群-11型                 | Bardet-Biedl syndrome 11  | TRIM32   | AR           | <1/500    |
|   | Mulibrey nanism syndrome  | TRIM37   |              |           |
| 急性新生兒肝衰竭  | Acute Infantile Liver Failure, TRMU-Related                         | TRMU     | AR           | <1/500    |
|   | Pontocerebellar hypoplasia  | TSEN54   |              |           |
| 氧化磷酸化缺乏症-TSFM 型                                     | Combined Oxidative Phosphorylation Deficiency 3                     | TSFM     | AR           | <1/500    |
| 先天性甲狀腺低功能症-TSHB 型                                   | Congenital hypothyroidism   | TSHB     | AR           | 1/500     |
|   | Hypothyroidism, congenital, nongoitrous, 1                          | TSHR     |              |           |
| 髮-肝-腸症候群  | Tricho-Hepato-Enteric Syndrome                                      | TTC37    | AR           | 1/500     |
|   | Familial dilated cardiomyopathy                                     | TTN      |              |           |
| 維他命 E 缺乏共濟失調症                                       | Ataxia with Vitamin E Deficiency                                    | TTPA     | AR           | <1/500    |
| MNGIE 症候群粒線體性神經胃腸腦病變症候群                             | Myoneurogastrointestinal Encephalopathy (MNGIE)                     | TYMP     | AR           | <1/500    |
|   | Oculocutaneous Albinism, Type 1                                     | TYR      |              |           |
|   | Oculocutaneous albinism, Type 3                                     | TYRP1    |              |           |
| 克果納傑氏症候群  | Crigler-Najjar Syndrome   | UGT1A1   | AR           | <1/500    |
|   | Beta-ureidopropionase deficiency                                    | UPB1     |              |           |
| 非症候群型遺傳性聽障<br>-USH1C 型; 尤塞氏綜合症 1C 型                 | Usher Syndrome, Type 1C   | USH1C    | AR           | 1/353     |
| 尤塞氏綜合症-2A 型   | Usher Syndrome, Type 2A   | USH2A    | AR           | 1/126     |
| 舞蹈棘狀紅血球症  | Choreo-acanthocytosis   | VPS13A   | AR           | <1/500    |
| 柯恩綜合症   | Cohen Syndrome  | VPS13B   | AR           | <1/500    |
| 嚴重先天性中性粒細胞減少症<br>-VPS45 型                           | Congenital Neutropenia, VPS45-Related                               | VPS45    | AR           | 1/224     |
|   | Pontocerebellar Hypoplasia, Type 2E                                 | VPS53    |              |           |
| 橋腦小腦發育不全-1A型  | Pontocerebellar Hypoplasia, Type 1A                                 | VRK1     | AR           | <1/500    |
| 小眼症   | Microphthalmia/Anophthalmia, VSX2-Related                           | VSX2     | AR           | 1/91      |
|   | Von Willebrand disease  | VWF      |              |           |
| Wiskott-Aldrich 氏症候群; 性聯遺傳血小板缺乏症; 低嗜嚴重先天性中性球症-WAS 型 | Wiskott-Aldrich syndrome, X-Linked                                  | WAS      | X-Linked     | 1/125,000 |
|   | Progressive Pseudorheumatoid Dysplasia                              | WISP3    |              |           |
| Schopf-Schulz-Passar ge 症候群                         | Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passar ge            | WNT10A   | AR           | <1/500    |
|   | Werner Syndrome   | WRN      |              |           |
| 著色性乾皮症-A 型  | Xeroderma pigmentosum Group A                                       | XPA      | AR           | 1/500     |
| 著色性乾皮症-C 型  | Xeroderma Pigmentosum Group C                                       | XPC      | AR           | 1/500     |
| 痙攣性下身麻痺-15 型  | Spastic Paraplegia Type 15  | ZFYVE26  | AR           | <1/500    |
| X 染色體脆折症  | Fragile X Syndrome  | FMR1     | X-Linked     | 1/151     |
| 非症候群型遺傳性聽障<br>-OTOF 型                               | Auditory Neuropathy   | OTOF     | AR           | <1/500    |
| 藥害性聽損   | non-syndromic sensorineural hearing loss                            | 12SrRNA  | Mitochondria | 1/500     |
| 腎小管發育不全   | Renal Tubular Dysgenesis  | AGT      | AR           | 1/80      |