

罕見疾病基金會服務罕見疾病類明細表 (2020 獎學金專用)

| 01、胺基酸有機酸代謝異常 |                         |  |  |
|---------------|-------------------------|--|--|
| 0101          | 苯酮尿症                    | Phenylketouria(PKU)                                | 0113 異戊酸血症   |
| 0102          | 高胱胺酸血症                  | Homocystinuria                                     | 0114 丙酸血症  |
| 0103          | 遺傳性高酪胺酸血症               | Hereditary tyrosinemia                             | 0115 戊二酸血症，第一、二型   |
| 0104          | 高甲硫胺酸血症                 | Methionine adenosyltransferase deficiency ,MET     | 0116 白胺酸代謝異常   |
| 0105          | 楓糖尿症                    | Maple syrup urine disease (MSUD)                   | 0117 三甲基巴豆輔酶 A 梭化酵素缺乏症   |
| 0106          | 非酮性高甘胺酸血症               | Nonketotic hyperglycinemia                         | 0118 多發性胺化酶缺乏症 (生物素酵素缺乏症)  |
| 0107          | 胱胺酸症                    | Cystinosis   | 0119 高脯胺酸血症  |
| 0108          | 苯酮尿症- 四氫基喋呤缺乏症          | (Phenylketonuria)-(Tetrahydrobiopterin deficiency) | 0120 芳香族 L-胺基酸類脫羧基酶缺乏症   |
| 0110          | 高離胺基酸血症                 | Hyperlysinemia                                     | 0121 甲基丙二酸血症併高胱胺酸血症(Cbl C 型)   |
| 0111          | 組胺酸血症                   | Histidinemia                                       | 0122 黑尿症   |
| 0112          | 甲基丙二酸血症                 | Methylmalonic acidemia (MMA)                       |  |
| 02、尿素循環代謝異常   |                         |  |  |
| 0201          | 瓜胺酸血症                   | Citrullinemia                                      | 0204 精胺丁二酸酵素缺乏症  |
| 0202          | 鳥胺酸甲醯基轉移酶缺乏症            | Omithine transcarbamylase deficiency               | 0205 高鳥胺酸血症-高安血症-高瓜胺酸血症候群  |
| 0203          | 乙醯胺酸合成酶缺乏症              | Nitroacetylglutamate synthetase deficiency (NAG)   | 0206 精胺丁二酸酵素缺乏症  |
| 03、其他代謝異常     |                         |  |  |
| 0301          | 肝醣儲積症 (type I-type IV)  | Glycogen storage disease (type I-type IV)          | 0320 黏脂質症  |
| 0302          | 黏多糖症 (type I ~ type VI) | Mucopolysaccharidoses (type I ~ type VI)           | 0321 (其他未分類之代謝異常疾病)  |
| 0303          | 高雪氏症                    | Gaucher's disease                                  | 0322 碳水化合物缺乏醣蛋白症候群   |
| 0304          | Fabry 氏症 (法布瑞氏症)        | Fabry Disease                                      | 0323 臭魚症   |
| 0305          | 尼曼匹克症                   | Niemann-Pick Disease                               | 0324 先天性全身脂質營養不良症  |
| 0306          | 短鏈脂肪酸去氫酶缺乏症             | Short-chain acyl-CoA dehydrogenase deficiency      | 0325 中鏈脂肪酸去氫酶缺乏症   |
| 0307          | 腎上腺腦白質失養症               | Adrenoleukodystrophy (ALD)                         | 0326 丙酮酸鹽脫氫酶缺乏症  |
| 0308          | 脂肪酸氧化作用缺陷               | Fatty acid oxidation defect                        | 0327 腦腱性黃瘤症  |
| 0309          | 亞硫酸鹽氧化酶缺乏症              | Sulfite oxidase deficiency                         | 0328 腦血管屏障葡萄糖輸送缺陷  |
| 0310          | 遺傳性果糖不耐症, 果糖尿症          | Fructose intolerance, hereditary                   | 0329 肢近端型點狀軟骨發育不良  |
| 0311          | 岩藻糖代謝異常 (儲積症)           | Fucosidosis  | 0330 豆腐腦血症   |
| 0312          | 原發性肉鹼缺乏症                | Carnitine deficiency syndrome, primary             | 0331 鉅輔酶缺乏症  |
| 0313          | MLD 症候群                 | Metachromatic Leukodystrophy (MLD)                 | 0332 低磷酸酯酶症  |
| 0314          | 粒線體缺陷                   | Mitochondrial defect                               | 0333 球細胞腦白質失養症   |
|               |                         |  | 0334 Isovaleric acidemia (IVA)   |
|               |                         |  | 0335 Propionic acidemia (PA)   |
|               |                         |  | 0336 Glutaric aciduria type I, II  |
|               |                         |  | 0337 3-Hydroxy-3-methyl-glutaric acidemia                                      |
|               |                         |  | 0338 3-Methylcrotony-CoA carboxylase deficiency                                |
|               |                         |  | 0339 Multiple carboxylase deficiency   |
|               |                         |  | 0340 Hyperprolinemia   |
|               |                         |  | 0341 Aromatic L-amino acid decarboxylase deficiency                            |
|               |                         |  | 0342 Cobalamin C Defect (Methylmalonic Aciduria and Homocystinuria, CblC type) |
|               |                         |  | 0343 Alkaptonuria  |
|               |                         |  | 0344 Argininosuccinic aciduria   |
|               |                         |  | 0345 Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome               |
|               |                         |  | 0346 Argininosuccinic Aciduria   |
|               |                         |  | 0347 Mucopolipidosis   |
|               |                         |  | 0348 Carbohydrate-deficiencyglycoprotein syndrome                              |
|               |                         |  | 0349 Trimethylaminuria   |
|               |                         |  | 0350 Congenital generalized Lipodystrophy                                      |
|               |                         |  | 0351 Medium-chain acyl-coenzyme A dehydrogenase deficiency (MCAD)              |
|               |                         |  | 0352 Pyruvate dehydrogenase deficiency   |
|               |                         |  | 0353 Cerebrotendinous Xanthomatosis  |
|               |                         |  | 0354 Glut(Glucose Transport) 1 Deficiency Syndrome                             |
|               |                         |  | 0355 Rhizomelic Chondrodysplasia Punctata (RCDP)                               |
|               |                         |  | 0356 Spherosterolemia  |
|               |                         |  | 0357 Molybdenum cofactor deficiency  |
|               |                         |  | 0358 Hypophosphatasia  |
|               |                         |  | 0359 Globoid Cell Leukodystrophy   |



|                   |                        |  |      |   |   |
|-------------------|------------------------|--|------|---|---|
| 0315              | 紫質症                    | porphyria  | 0334 | 巴氏症候群                                       | Barth Syndrome  |
| 0316              | 威爾森氏症                  | Wilson's disease   | 0335 | Beta 硫醇酶缺乏症                                 | Beta-Kerithiolase Deficiency  |
| 0317              | 先天性高乳酸血症               | Congenital hyperlactic acidemia  | 0336 | 嬰兒型溶酶體酸性脂肪酶缺乏症                              | Infantile form Lysosomal Acid Lipase Deficiency                                     |
| 0318              | 持續性幼兒型胰島素過度分泌<br>低血糖症  | Persistent hyperinsulinemic hypoglycemia of infancy<br>Low blood sugar | 0337 | 多發性硫酸脂酶缺乏症                                  | Multiple Sulfatase Deficiency   |
| 0319              | 半乳糖血症                  | Galactosemia   | 0338 | 生物素酶缺乏症                                     | Biotinidase Deficiency  |
| <b>04、心肺功能失調</b>  |                        |  |      |   |   |
| 0401              | 原發性肺血鐵質沉積症             | Primary Pulmonary hemosiderosis  | 0406 | Holt-Oram 氏症候群                              | Holt-Oram Syndrome  |
| 0402              | 原發性肺動脈高壓症              | Primary Pulmonary Hypertension, PPH                                    | 0407 | Andersen 氏症候群 (心節律障礙週期性<br>麻痺症候群；鈣離子通道病變疾病) | Andersen's syndrome   |
| 0403              | Alstrom 氏症候群           | Alstrom Syndrome   | 0408 | 窒息性胸腔失養症                                    | Asphyxiating thoracic dystrophy   |
| 0404              | 特發性嬰兒動脈硬化              | Idiopathic Infantile Arterial Calcification                            | 0409 | 先天性中樞性換氣不足症候群                               | Congenital Central Hypoventilation Syndrome   |
| 0405              | 囊狀纖維化                  | Cystic fibrosis  |      |   |   |
| <b>05、消化系統失調</b>  |                        |  |      |   |   |
| 0501              | 進行性家族性肝內膽汁滯留症          | Progressive intrahepatic cholestasis, PFIC                             | 0503 | 先天性 Cajal 氏間質細胞增生合併腸道神經<br>元發育異常            | Congenital Interstitial Cell of Cajal Hyperplasia with<br>Neuronal Intestinal Dyspl |
| 0502              | 先天性膽酸合成障礙              | Inborn errors of bile acid synthesis                                   | 0504 | 阿拉吉歐症候群                                     | Alagille Syndrome   |
| <b>06、泌尿系統失調</b>  |                        |  |      |   |   |
| 0601              | 腎因型尿崩症                 | X-linked nephrogenic diabetes insipidus                                | 0604 | 家族性低血鉀症                                     | Hypokalemia, familial   |
| 0602              | 性聯遺傳型低磷酸鹽佝僂症           | X-linked hypophosphatemic rickets                                      | 0605 | 自體隱性遺傳多囊性腎疾病                                | Autosomal recessive polycystic kidney disease                                       |
| 0603              | Lowe 氏症候群              | Lowe syndrome  | 0606 | Barter 氏症候群                                 | Barter's syndrome   |
| <b>07、腦部或神經病變</b> |                        |  |      |   |   |
| 0701              | 毛樣腦血管疾病                | Moya moya disease  | 0720 | 神經元巔癩脂褐質儲積症                                 | Neuronal ceroid lipofuscinosis  |
| 0702              | 胼胝體發育不全症               | Agenesis of corpus callosum  | 0721 | Alexander 氏病                                | Alexander disease   |
| 0703              | 脊髓小腦退化性動作協調障礙          | Spinocerebellar ataxia   | 0722 | 僵體症候群                                       | Stiffperson syndrome  |
| 0704              | 亨丁頓氏舞蹈症                | Huntington disease(又稱 Huntington's chorea)                             | 0723 | 酪胺酸羧化酶缺乏症                                   | Tyrosine hydroxylase deficiency   |
| 0705              | 結節性硬化症                 | Tuberous sclerosis   | 0724 | Wolfram 氏症候群                                | Wolfram syndrome , DIDMOAD  |
| 0706              | 多發性硬化症                 | Multiple sclerosis   | 0725 | 遺傳性痙攣性下身麻痺                                  | Hereditary spastic Paraplegia   |
| 0707              | Zellweger 氏症候群         | Zellweger syndrome   | 0726 | Joubert 氏症候群(家族性小腦蚓部發育不全)                   | Joubert syndrome  |
| 0708              | 瑞特氏症候群                 | Rett syndrome  | 0727 | Pelizaeus-Merzbacher 氏症(慢性兒童型腦硬<br>化症)      | Pelizaeus-Merzbacher Disease  |
| 0709              | 脊髓性肌肉萎縮症               | Spinal muscular atrophy  | 0728 | 甘迺迪氏症 (脊髓延髓性肌肉萎縮症)                          | Kennedy Disease   |
| 0710              | Menkes 氏症候群            | Menkes disease   | 0729 | 家族性澱粉樣多發性神經病變                               | Familial Amyloidotic Polyneuropathy   |
| 0711              | 肌萎縮性側索硬化症(漸凍人)         | Amyotrophic lateral sclerosis (ALS)                                    | 0730 | 泛酸鹽激酶相關聯之神經退化性疾病                            | Pantothenate Kinase Associated<br>Neurodegeneration , PKAN                          |
| 0712              | Charcot-Marie-Tooth 氏症 | Charcot-Marie-Tooth Disease  | 0731 | Moebius 症候群                                 | Moebius Syndrome  |



|                |                      |  |      |                         |  |
|----------------|----------------------|--|------|-------------------------|--|
| 0713*          | GMI/GM2 神經節苷脂儲積症     | GMI/GM2 gangliosidosis                           | 0732 | McLeod 症候群              | McLeod Syndrome  |
| 0714           | Lesch-Nyhan 氏症候群     | Lesch-Nyhan syndrome                             | 0733 | Aicardi-Goutieres 症候群   | Aicardi-Goutieres Syndrome   |
| 0715           | 共濟失調微血管擴張症候群         | Ataxia telangiectasia                            | 0734 | 普洛提斯症候群                 | Proteus Syndrome   |
| 0716           | 涎酸酵素缺乏症              | Sialidosis                                       | 0735 | MECP2 綜合症候群             | Methyl CpG binding protein 2 Duplication Syndrome                  |
| 0717           | 先天性痛不敏感症合併無汗症        | Congenital insensitivity to pain with anhidrosis | 0736 | 腦肋小頰症候群                 | Cerebro-Costo-Mandibular Syndrome                                  |
| 0718           | 下視丘功能障礙症候群           | Hypothalamic dysfunction syndrome                | 0737 | Dravet 症候群              | Dravet Syndrome  |
| 0719           | Miller Dieker 症候群    | Miller Dieker syndrome                           | 0738 | 腦白質消失症                  | Vanishing White Matter Disease                                     |
| <b>08、皮膚病變</b> |                      |  |      |                         |  |
| 0801           | 遺傳性表皮分解性水皰症          | Hereditary epidermolysis bullosa                 | 0809 | 嬰兒型全身性玻璃樣變性             | Infantile systemic hyalinosis                                      |
| 0802           | 層狀魚鱗癬(自體隱性遺傳型)       | Ichthyosis, lamellar recessive                   | 0810 | Meleda 島病               | Meleda disease   |
| 0803           | 外胚層增生不良症             | Ectodermal Dysplasias                            | 0811 | Darier 氏病 (毛囊角化症)       | Darier's disease   |
| 0804           | 膠膜兒                  | Collodion baby                                   | 0812 | 先天性角化不全症                | Dyskeratosis Congenita   |
| 0805           | 斑色魚鱗癬                | Harlequin ichthyosis                             | 0813 | 皮膚過度角化症雅司病              | Diffuse Non-epidermolytic Palmoplantar Keratoderma type Unna-Thost |
| 0806           | 水泡型先天性鱗鱗樣紅皮症         | Bullous Congenital ichthyosiform erythroderma    | 0814 | Netherton 症候群           | Netherton Syndrome   |
| 0807           | 色素失調症                | Incontinentia pigmenti                           | 0815 | 先天性巨大型黑色素痣              | Giant Congenital Melanocytic Nevus                                 |
| 0808           | 眼睛皮膚白化症              | Oculocutaneous albinism                          |      |                         |  |
| <b>09、肌肉病變</b> |                      |  |      |                         |  |
| 0901           | 遺傳性細胞漿內體肌病變          | Hereditary cytoplasmic body myopathy             | 0910 | 貝克型肌肉失養症                | Becker Muscular Dystrophy(BMD)                                     |
| 0902           | 裘馨氏肌肉萎縮症             | Duchenne muscular dystrophy (DMD)                | 0911 | Freeman-Sheldon 氏症候群    | Freeman-Sheldon syndrome   |
| 0903           | 肌中央軸空病               | Central core myopathy                            | 0912 | 肢帶型肌失養症(第2A型、第2B型、第2D型) | Limb-girdle muscular dystrophy(type 2A、2B、2D)                      |
| 0904           | Nemaline 線狀肌肉病變      | Nemaline Rod Myopathy                            | 0913 | 先天性肌失養症                 | Congenital Muscular Dystrophy                                      |
| 0905           | Schwartz Jampel 氏症候群 | Schwartz Jampel syndrome                         | 0914 | 多微小軸空肌病                 | Multiminicore Disease  |
| 0906           | 肌肉強直症                | Myotonic dystrophy                               | 0915 | Emery-Dreifuss 肌失養症     | Emery-Dreifuss Muscular Dystrophy                                  |
| 0907           | 其他型肌肉萎縮症             |  | 0916 | GNE 遠端肌病變               | GNE myopathy   |
| 0908           | 肌小管病變                | Myotubular myopathy                              | 0917 | 史托摩根症候群                 | Stormorken syndrome  |
| 0909           | 面高胛肱肌失養症             | Facioscapulohumeral muscular dystrophy           |      |                         |  |
| <b>10、骨頭病變</b> |                      |  |      |                         |  |
| 1001           | 成骨不全症 (玻璃娃娃)         | Osteogenesis imperfecta                          | 1008 | 骨路發育異常                  | Spondyloepiphyseal Dysplasia(SED)                                  |
| 1002           | 軟骨發育不全症(小人兒)         | Achondroplasia                                   | 1009 | 裂手裂足症                   | Split-hand/ Split-foot malformation (SHFM)                         |
| 1003           | 骨質石化症(大理石寶)          | Osteopetrosis                                    | 1010 | 假性軟骨發育不全                | Pseudoachondroplastic dysplasia                                    |
| 1004           | 進行性骨化性肌炎             | Fibrodysplasia Ossificans Progressiva            | 1011 | Conradi-Hunermann 氏症候群  | Conradi-Hunermann syndrome   |
| 1005           | 原發性變形性骨炎             | Primary Paget disease                            | 1012 | 多發性骨骺發育不全症              | Multiple Epiphyseal Dysplasia                                      |
| 1006           | 鎖骨顛骨發育異常             | Cleidocranial dysplasia                          | 1013 | 次軟骨發育不全症                | Hypochoondroplasia   |

|                    |                               |  |      |                                    |  |
|--------------------|-------------------------------|--|------|------------------------------------|--|
| 1007               | McCune Albright 氏症候群(纖維性骨失養症) | McCune Albright syndrome                 | 1014 | 先天頸椎病變                             | Klippel-Feil Syndrome  |
| <b>11、結締組織病變</b>   |                               |  |      |                                    |  |
| 1101               | 馬凡氏症(蜘蛛人症)                    | Marfan syndrome                          | 1103 | 先天結締組織異常第四型                        | Ehlers Danlos syndrome IV  |
| 1102               | 瓦登伯格氏症候群(藍眼珠)                 | Wardenburg syndrome                      | 1104 | 畢耳氏症候群                             | Beals Syndrome   |
| <b>12、造血功能異常</b>   |                               |  |      |                                    |  |
| 1202               | 重型海洋性贫血                       | Thalassemia major                        | 1206 | 陣發性夜間血紅素尿症                         | Paroxysmal Nocturnal Hemoglobinuria  |
| 1203               | 血小板無力症                        | Thrombasthenia                           | 1207 | 先天性純紅血球再生障礙性贫血                     | Diamond Blackfan Anemia  |
| 1204               | 同基因合子蛋白質 C 缺乏症                | Homozygous protein C deficiency          | 1208 | 非典型性尿毒溶血症候群                        | Atypical Hemolytic Uremic Syndrome   |
| 1205               | $\alpha$ -1-抗胰蛋白酶缺乏症          | $\alpha$ -1-Antitrypsin deficiency       | 1209 | 蛋白質 S 缺乏症                          | Protein S Deficiency   |
| <b>13、免疫疾病</b>     |                               |  |      |                                    |  |
| 1301               | 布魯頓氏低免疫球蛋白血症                  | Bruton's agammaglobulinemia              | 1306 | 補體成份 8 缺乏症                         | Complement Component 8 deficiency  |
| 1302               | 原發性慢性肉芽腫病                     | Chronic primary granulomatous disease    | 1307 | IPEX 症候群                           | IPEX Syndrome  |
| 1303               | 先天性高免疫球蛋白 E 症候群               | Congenital Hyper IgE syndrome            | 1308 | 高免疫球蛋白 M 症候群                       | Hyper-IgM Syndrome   |
| 1304               | Wiskott-Aldrich 氏症候群          | Wiskott-Aldrich Syndrome                 | 1309 | $\gamma$ 干擾素受體 1 缺陷                | Interferon $\gamma$ receptor 1 deficiency  |
| 1305               | 嚴重複合型免疫缺乏症                    | Severe combined immunodeficiency         | 1310 | 遺傳性血管性水腫                           | Hereditary Angioedema  |
| <b>14、內分泌疾病</b>    |                               |  |      |                                    |  |
| 1401               | 先天性腎上腺發育不全(非增生症)              | Congenital adrenal hypoplasia            | 1407 | Kenny-Caffey 氏症候群                  | Kenny-Caffey syndrome  |
| 1402               | 假性副甲狀腺低能症                     | Pseudohypoparathyroidism                 | 1408 | 威爾姆氏腫瘤、無虹膜、性器異常、智能障礙症候群 (WAGR 症候群) | WAGR Syndrome (Wilms' tumor-Aniridia-Genitourinary Anomalies-mental Retardation) |
| 1403               | 同合子家族性高膽固醇血症                  | Homozygous familial hypercholesterolemia | 1409 | 腎上腺皮促素抗性                           | ACTH resistance  |
| 1404               | 家族性高乳糜微粒血症                    | Familial hyperchylomicronemia            | 1410 | $1\alpha$ -羟化酶缺乏症候群                | $1\alpha$ -hydroxylase deficiency  |
| 1405               | 肢端肥大症(大肢症)                    | Acromegaly                               | 1411 | Kallmann 氏症候群                      | Kallmann syndrome  |
| 1406               | Laron 氏侏儒症候群                  | Laron syndrome (Laron dwarfism)          | 1412 | 永久性新生兒糖尿病                          | Permanent Neonatal Diabetes Mellitus   |
| <b>15、不正常細胞增生瘤</b> |                               |  |      |                                    |  |
| 1501               | 神經纖維瘤症候群第二型                   | Neurofibromatosis Type II                | 1505 | Beckwith Wiedemann 氏症候群            | Beckwith Wiedemann syndrome  |
| 1503               | 視網膜母細胞瘤                       | Retinoblastoma                           | 1506 | 淋巴血管平滑肌肉增生症                        | Lymphangiomyomatosis(LAM)  |
| 1504               | 神經母細胞瘤                        | Neuroblastoma                            | 1507 | 達希伯-林道症候群                          | Von Hippel-Lindau (VHL)  |
| <b>16、外觀異常</b>     |                               |  |      |                                    |  |
| 1601               | 愛伯特氏症                         | Apert syndrome                           | 1615 | 克斯提各氏彈性蛋白缺陷症(小黑人症)                 | Costello Syndrome  |
| 1602               | Crouzon 氏症候群                  | Crouzon Syndrome                         | 1616 | Fraser 氏症                          | Fraser syndrome  |
| 1603               | 羅素-西弗氏症                       | Russell-Silver syndrome                  | 1617 | 先天性家族性臉口狹小症                        | Blepharophimosis-Prosis-Epicanthus Inversus Syndrome                             |
| 1604               | Comelia de Lange 氏症候群         | Comelia de Lange syndrome                | 1618 | 歌舞伎症候群                             | Kabuki make-up syndrome  |
| 1605               | X 脆折症                         | Fragile X syndrome                       | 1619 | 耳-顴-指(趾)症候群                        | Oto-Palato-Digital syndrome  |

|                     |                                  |  |      |                        |                                       |
|---------------------|----------------------------------|--|------|------------------------|---------------------------------------|
| 1606                | CHARGE 聯合畸形                      | CHARGE association                               | 1620 | Robinow 氏症候群           | Robinow Syndrome                      |
| 1607                | Aarskog-Scott 氏症候群               | Aarskog-Scott syndrome                           | 1621 | Pfeiffer 氏症候群          | Pfeiffer Syndrome                     |
| 1608                | Smith-Lemli-Opitz 症候群            | Smith-Lemli-Opitz syndrome                       | 1622 | 指(趾)甲贅肉症候群             | Nail-Patella Syndrome                 |
| 1609                | Bardet-Biedl 氏症候群                | Bardet-Biedl syndrome                            | 1623 | CFC 症候群                | Cardiofaciocutaneous Syndrome         |
| 1610                | Larsen 氏症候群<br>(顎裂-先天性脫位症候群)     | Larsen syndrome                                  | 1624 | Peter-Plus 症候群         | Peter-Plus Syndrome                   |
| 1611                | 皮爾羅賓氏症                           | Pierre Robin Syndrome                            | 1625 | Nager 症候群              | Nager Syndrome                        |
| 1612                | 崔卻·柯林斯氏症候群                       | Treachet Collins syndrome                        | 1626 | Coffin-Siris 症候群       | Coffin-Siris syndrome                 |
| 1613                | 多發性翼狀膜症候群                        | Multiple pterygium syndrome                      | 1627 | 懷特-薩頓症候群               | White-Sutton Syndrome                 |
| 1614                | 努南氏症                             | Noonan syndrome                                  |      |                        |                                       |
| <b>17、染色體異常</b>     |                                  |  |      |                        |                                       |
| 1701                | Prader-Willi 氏症候群<br>(小胖威利、好吃寶費) | Prader-Willi syndrome                            | 1706 | Rubinstein-Taybi 氏症候群  | Rubinstein-Taybi syndrome             |
| 1702                | Angelman 氏症候群(快樂玩偶)              | Angelman syndrome                                | 1707 | Branchio-Oto-Renal 症候群 | Branchio-Oto-Renal Syndrome           |
| 1703                | 威廉斯氏症                            | Williams Syndrome                                | 1708 | Kleefstra 症候群          | Kleefstra Syndrome                    |
| 1704                | DiGeorge's 症候群(狄喬治氏症)            | DiGeorge's disease                               |      |                        |                                       |
| <b>18、其他分類或不明原因</b> |                                  |  |      |                        |                                       |
| 1801                | 早老症                              | Hutchinson Gilford progeria syndrome             | 1809 | 先天性靜脈畸形肥大症候群           | Klippel-Trenaunay syndrome            |
| 1802                | Cockayne 氏(柯凱因氏)症候群              | Cockayne syndrome                                | 1810 | 遺傳性出血性血管擴張症            | Hereditary Hemorrhagic Telangiectasia |
| 1803                | 海勒曼·史德萊夫氏症候群                     | Hallermann-Streiff syndrome                      | 1811 | Stargardt' s 氏症        | Stargardt' s disease                  |
| 1804                | 髮-肝-腸症候群                         | Tricho-hepato-enteric syndrome                   | 1812 | 先天性無虹膜                 | aniridia                              |
| 1805                | 先天性水痘症候群                         | Congenital Varicella Syndrome                    | 1813 | Kohlmeier-Degos 綜合症    | Kohlmeier-Degos Disease               |
| 1806                | 成人型早老症                           | Werner Syndrome                                  | 1814 | 隱匿性黃斑部失養症              | Occult Macular Dystrophy              |
| 1808                | 短指發育不良及性別顛倒                      | Campomelic dysplasia with autosomal sex reversal |      |                        |                                       |

\* 本表為本會自行分類，皆為目前基金會服務之所有罕見疾病之疾病種類(共 257 種)，由於涵蓋一些目前政府尚未公告或在審查中卻急需協助之罕病，所以本會之分類名單原則上會比衛福部公告(目前截至 2020 年 4 月共 223 種)的罕病種類還多，未來將視實際需要不定期進行更新。

