

罕見疾病基金會服務罕見疾病分類明細表 (2019 獎學金專用)

| 01、胺基酸有機酸代謝異常 |   |
|---------------|---|
| 0101          | 苯酮尿症<br>Phenylketouria(PKU)   |
| 0102          | 高胱氨酸血症<br>Homocystinuria  |
| 0103          | 遺傳性高酪氨酸血症<br>Hereditary tyrosinemia   |
| 0104          | 高甲硫氨酸血症<br>Methionine adenosyltransferase deficiency ,MET                           |
| 0105          | 楓糖尿症<br>Maple syrup urine disease (MSUD)  |
| 0106          | 非酮性高甘氨酸血症<br>Nonketotic hyperglycinemia   |
| 0107          | 胱氨酸症<br>Cystinosis  |
| 0108          | 苯酮尿症-四氫喋呤缺乏症<br>(Phenylketonuria)-(Tetrahydrobiopterin deficiency)                  |
| 0110          | 高離胺基酸血症<br>Hyperlysinemia   |
| 0111          | 組胺酸血症<br>Histidinemia   |
| 02、尿素循環代謝異常   |   |
| 0201          | 瓜胺酸血症<br>Citrullinemia  |
| 0202          | 鳥胺酸氨甲醯基轉移酶缺乏症<br>Ornithine transcarbamylase deficiency                              |
| 0203          | 乙醯穀胺酸合成酶缺乏症<br>Nitroacetylglutamate synthetase deficiency (NAG)                     |
| 03、其他代謝異常     |   |
| 0301          | 肝醣儲積症 (type I~type IV)<br>Glycogen storage disease (type I~type IV)                 |
| 0302          | 黏多糖症 (type I ~ type VI)<br>Mucopolysaccharidoses(type I ~ type VI)                  |
| 0303          | 高雪氏症<br>Gaucher's disease   |
| 0304          | Fabry 氏症 (法布瑞氏症)<br>Fabry Disease   |
| 0305          | 尼曼匹克症<br>Niemann-Pick Disease   |
| 0306          | 短鏈脂肪酸去氫酶缺乏症<br>Short-chain acyl-CoA dehydrogenase deficiency                        |
| 0307          | 腎上腺腦白質失養症<br>Adrenoleukodystrophy (ALD)   |
| 0308          | 脂肪酸氧化作用缺陷<br>Fatty acid oxidation defect  |
| 0309          | 亞硫酸鹽氧化酶缺乏<br>Sulfite oxidase deficiency   |
| 0310          | 遺傳性果糖不耐症,果糖尿症<br>Fructose intolerance, hereditary                                   |
| 0311          | 岩藻糖代謝異常 (儲積症)<br>Fucosidosis  |
| 0312          | 原發性肉鹼缺乏症<br>Carnitine deficiency syndrome, primary                                  |
| 0313          | MLD 症候群<br>Metachromatic Leukodystrophy (MLD)                                       |
| 0314          | 粒線體缺陷<br>Mitochondrial defect   |
| 0315          | 紫質症<br>porphyria  |
| 0112          | 甲基丙二酸血症<br>Methylmalonic acidemia (MMA)   |
| 0113          | 異戊酸血症<br>Isovaleric acidemia (IVA)  |
| 0114          | 丙酸血症<br>Propionic acidemia (PA)   |
| 0115          | 戊二酸血症,第一、二型<br>Glutaric aciduria type I, II   |
| 0116          | 白胺酸代謝異常<br>3-Hydroxy-3-methyl-glutaric acidemia                                     |
| 0117          | 三甲基巴豆醯輔酶 A 核化酵素缺乏症<br>3-Methylcrotony-CoA carboxylase deficiency                    |
| 0118          | 多發性羧化酶缺乏症 (生物素酵素缺乏症)<br>Multiple carboxylase deficiency                             |
| 0119          | 高脯胺酸血症<br>Hyperprolinemia   |
| 0120          | 芳香族 L-胺基酸類脫羧基酶缺乏症<br>Aromatic L-amino acid decarboxylase deficiency                 |
| 0121          | 甲基丙二酸血症併高胱氨酸血症(Cb1 C 型)<br>Homocystinuria, Cb1C type)                               |
| 0204          | 精胺丁二酸酵素缺乏症<br>Argininosuccinic aciduria   |
| 0205          | 高鳥胺酸血症-高安血症-高瓜胺酸血症候群<br>Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome |
| 0206          | 精胺丁二酸酵素缺乏症<br>Argininosuccinic Aciduria   |
| 0320          | 黏脂質症<br>Mucopolidosis   |
| 0321          | (其他未分類之代謝異常疾病)<br>Carbohydrate-deficiencyglycoprotein syndrome                      |
| 0322          | 碳水化合物缺乏糖蛋白症候群<br>Trimethylaminuria  |
| 0323          | 臭魚症<br>Congenital generalized Lipodystrophy   |
| 0324          | 先天性全身脂質營養不良症<br>Medium-chain acyl-coenzyme A dehydrogenase deficiency (MCAD)        |
| 0325          | 中鏈脂肪酸去氫酶缺乏症<br>Pyruvate dehydrogenase deficiency                                    |
| 0326          | 丙酮酸鹽脫氫酶缺乏症<br>Cerebrotendinous Xanthomatosis  |
| 0327          | 腦髓性黃瘤症<br>Glut(Glucose Transport) 1 Deficiency Syndrome                             |
| 0328          | 腦血管屏障葡萄糖輸送缺陷<br>Rhizomelic Chondrodysplasia Punctata (RCDP)                         |
| 0329          | 肢近端型點狀軟骨發育不良<br>Sifosterolemia  |
| 0330          | 豆腐渣血症<br>Molybdenum cofactor deficiency   |
| 0331          | 鉛輔酶缺乏症<br>Hypophosphatasia  |
| 0332          | 低磷酸酯酶症<br>Globoid Cell Leukodystrophy   |
| 0333          | 球細胞腦白質失養症<br>Barth Syndrome   |
| 0334          | 巴氏症候群   |

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|-------------------|------------------------|---|------|--|---|
| 0316              | 威爾森氏症                  | Wilson's disease                                    | 0335 | Beta 硫解酶缺乏症  | Beta-Ketothiolase 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 | 0337 | 多發性硫酸酯酶缺乏症   | 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 Hypertensio,PPH                   | 0407 | Andersen 氏症候群 (心節律障礙暨週期性<br>麻痺症候群; 鉀離子通道病變疾病)  | 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元發育異常<br>Congenital Interstitial Cell of Cajal Hyperplasia with 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 nephrogenicdiabetes insipidus              | 0604 | 家族性低血鈣症  | 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 | 神經元蠟樣脂褐質沉積症  | 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              | 亨汀頓氏舞蹈症                | Huntington disease(又稱 Huntington's chorea)          | 0723 | 酪胺酸經化酶缺乏症  | 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 | 泛酸鹽激酶關聯之神經退化性疾病  | Pantothenate Kinase Associated Neurodegeneration , PKAN |
| 0712              | Charcot-Marie-Tooth 氏症 | Charcot-Marie-Tooth Disease                         | 0731 | Moebius 症候群  | Moebius Syndrome  |

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| 0713           | GM1/GM2 神經節苷脂儲積症     | GM1/GM2 gangliosidosis                           | McLeod 症候群                    | McLeod Syndrome  |
| 0714           | Lesch-Nyhan 氏症候群     | Lesch-Nyhan syndrome                             | Aicardi-Goutieres 症候群         | Aicardi-Goutieres Syndrome   |
| 0715           | 共濟失調微血管擴張症候群         | Ataxia telangiectasia                            | 普洛提斯症候群                       | Proteus Syndrome   |
| 0716           | 涎酸酵素缺乏症              | Sialidosis                                       | MECTP2 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 | 腦肋/小頷症候群                      | Cerebro-Costo-Mandibular Syndrome                                  |
| 0718           | 下視丘功能障礙症候群           | Hypothalamic dysfunction syndrome                | Dravet 症候群                    | Dravet Syndrome  |
| 0719           | Miller Dieker 症候群    | Miller Dieker syndrome                           | 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                 | 嬰兒型全身性玻璃樣變性                   | Infantile systemic hyalinosis                                      |
| 0802           | 鱗狀魚鱗癬 (自體隱性遺傳型)      | Ichthyosis, lamellar recessive                   | Meleda 島病                     | Meleda disease   |
| 0803           | 外胚層增生不良症             | Ectodermal Dysplasias                            | Darier 氏病 (毛囊角化症)             | Darier's disease   |
| 0804           | 膠膜兒                  | Collodion baby                                   | 先天性角化不全症                      | Dyskeratosis Congenita   |
| 0805           | 斑色魚鱗癬                | Harlequin ichthyosis                             | 皮膚過度角化症雅司病                    | Diffuse Non-epidermolytic Palmoplantar Keratoderma type Unna-Thost |
| 0806           | 水泡型先天性魚鱗癬樣紅皮症        | Bullous Congenital ichthyosiform erythroderma    | Netherton 症候群                 | Netherton Syndrome   |
| 0807           | 色素失調症                | Incontinentia pigmenti                           | 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             | 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)                | Freeman-Sheldon 氏症候群          | Freeman-Sheldon syndrome   |
| 0903           | 肌中央軸空病               | Central core myopathy                            | 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                            | 先天性肌失養症                       | Congenital Muscular Dystrophy                                      |
| 0905           | Schwartz Jampel 氏症候群 | Schwartz Jampel syndrome                         | 多微小軸空肌病                       | Multiminicore Disease  |
| 0906           | 肌肉強直症                | Myotonic dystrophy                               | Emery-Dreifuss 肌失養症           | Emery-Dreifuss Muscular Dystrophy                                  |
| 0907           | 其他型肌肉萎縮症             |  | GENE 遠端肌病變                    | GENE myopathy  |
| 0908           | 肌小管病變                | Myotubular myopathy                              | 史托摩根症候群                       | Stormorken syndrome  |
| 0909           | 面肩胛肱肌失養症             | Facioscapulohumeral muscular dystrophy           |                               |  |
| <b>10、骨頭病變</b> |                      |  |                               |  |
| 1001           | 成骨不全症 (玻璃娃娃)         | Osteogenesis imperfecta                          | 骨骼發育異常                        | Spondyloepiphyseal Dysplasia(SED)                                  |
| 1002           | 軟骨發育不全症(小兒)          | Achondroplasia                                   | 裂手裂足症                         | Split-hand/ Split-foot malformation (SHFM)                         |
| 1003           | 骨質石化症 (大理石寶)         | Osteopetrosis                                    | 假性軟骨發育不全                      | Pseudoachondroplastic dysplasia                                    |
| 1004           | 進行性骨化性肌炎             | Fibrodysplasia Ossificans Progressiva            | 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                          | Hypochondroplasia   |
| 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)                 | Waardenburg syndrome                     | 1104 | 畢耳氏症候群                            | Beals Syndrome  |
| <b>12、造血功能異常</b>   |                               |  |      |                                   |   |
| 1202               | 重型海洋性貧血                       | 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貧血                    | Diamond Blackfan Anemia   |
| 1204               | 同基因合子蛋白質 C 缺乏症                | Homozygous proetin C deficiency          | 1208 | 非典型性性尿毒溶血症候群                      | 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 | 補體成分 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 B 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               | 嚴重複合免疫缺陷症                     | 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 $\alpha$ -羥化酶缺乏症候群              | 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 | 克斯提洛氏彈性蛋白缺陷症(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-Posis-Epicanthus Inversus Syndrome |
| 1604                | Cornelia de Lange 氏症候群           | Cornelia de Lange syndrome                       | 1618 | 歌舞伎症候群                 | Kabuki make-up syndrome                             |
| 1605                | X 脆折症                            | Fragile X syndrome                               | 1619 | 耳-髖-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)甲發育症候群             | 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                       | Treacher 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、好吃寶寶) | 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 | 先天性靜脈脈畸形骨肥大症候群         | 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 256 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 2019 年 2 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。