

罕見疾病基金會服務罕見疾病病類明細表 (2020 獎學金專用)

01、胺基酸有機酸代謝異常

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|------|--------------|--|------|-------------------------|---|
| 0101 | 苯酮尿症 | Phenylketouria(PKU) | 0113 | 異戊酸血症 | Isovaleric acidemia (IVA) |
| 0102 | 高胱胺酸血症 | Homocystinuria | 0114 | 丙酸血症 | Propionic acidemia (PA) |
| 0103 | 遺傳性高酪胺酸血症 | Hereditary tyrosinemia | 0115 | 戊二酸血症，第一、二型 | Glutaric aciduria type I, II |
| 0104 | 高甲硫胺酸血症 | Methionine adenosyltransferase deficiency ,MET | 0116 | 白胺酸代謝異常 | 3-Hydroxy-3-methyl-glutaric acidemia |
| 0105 | 楓糖尿症 | Maple syrup urine disease (MSUD) | 0117 | 三甲基巴豆腦輔酶 A 梭化酵素缺乏症 | 3-Methylcrotony-CoA carboxylase deficiency |
| 0106 | 非酮性高甘胺酸血症 | Nonketotic hyperglycinemia | 0118 | 多發性羧化酶缺乏症 (生物素酵素缺乏症) | Multiple carboxylase deficiency |
| 0107 | 胱胺酸症 | Cystinosis | 0119 | 高脯胺酸血症 | Hyperprolinemia |
| 0108 | 苯酮尿症-四氫葉啉缺乏症 | (Phenylketonuria)-(Tetrahydrobiopterin deficiency) | 0120 | 芳香族 L-胺基酸類脫羧基酶缺乏症 | Aromatic L-amino acid decarboxylase deficiency |
| 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) | Cobalamin C Defect (Methylmalonic Aciduria and Homocystinuria, CblC type) |
| 0111 | 組胺酸血症 | Histidinemia | 0122 | 黑尿症 | Alkaptonuria |
| 0112 | 甲基丙二酸血症 | Methylmalonic acidemia (MMA) | | | |

02、尿素循環代謝異常

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| 0201 | 瓜胺酸血症 | Citrullinemia | 0204 | 精胺丁二酸酵素缺乏症 | Argininosuccinic aciduria |
| 0202 | 鳥胺酸氨甲醯基轉移酶缺乏症 | Orotidine transcarbamylase deficiency | 0205 | 高鳥胺酸血症-高安血症-高瓜胺酸血症候群 | Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome |
| 0203 | 乙醯穀胺酸合成酶缺乏症 | Nitroacetylglutamate synthetase deficiency (NAG) | 0206 | 精胺丁二酸酵素缺乏症 | Argininosuccinic Aciduria |

03、其他代謝異常

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| 0301 | 肝醣儲積症 (type I~type IV) | Glycogen storage disease (type I~type IV) | 0320 | 黏脂質症 | Mucopolidosis |
| 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 | Carbohydrate-deficiencyglycoprotein syndrome |
| 0304 | Fabry 氏症 (法布瑞氏症) | Fabry Disease | 0323 | 臭魚症 | Trimethylaminuria |
| 0305 | 尼曼匹克症 | Niemann-Pick Disease | 0324 | 先天性全身脂質營養不良症 | Congenital generalized Lipodystrophy |
| 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 | Medium-chain acyl-Coenzyme A dehydrogenase deficiency (MCAD) |
| 0307 | 腎上腺腦白質失養症 | Adrenoleukodystrophy (ALD) | 0326 | 丙酮酸鹽脫氫酶缺乏症 | Pyruvate dehydrogenase deficiency |
| 0308 | 脂肪酸氧化作用缺陷 | Fatty acid oxidation defect | 0327 | 腦腱性黃瘤症 | Cerebrotendinous Xanthomatosis |
| 0309 | 亞硫酸鹽氧化酶缺乏 | Sulfite oxidase deficiency | 0328 | 腦血管屏障葡萄糖輸送缺陷 | Glut(Glucose Transport) 1 Deficiency Syndrome |
| 0310 | 遺傳性果糖不耐症; 果酸尿症 | Fructose intolerance, hereditary | 0329 | 肢近端型點狀軟骨發育不良 | Rinzomeic Chondrodysplasia Punctata (RCDP) |
| 0311 | 岩藻糖代謝異常 (儲積症) | Fucosidosis | 0330 | 豆固醇血症 | Sitosterolemia |
| 0312 | 原發性肉鹼缺乏症 | Carnitine deficiency syndrome, primary | 0331 | 鉅輔酶缺乏症 | Molybdenum cofactor deficiency |
| 0313 | MLD 症候群 | Metachromatic Leukodystrophy (MLD) | 0332 | 低磷酸酯酶症 | Hypophosphatasia |
| 0314 | 粒線體缺陷 | Mitochondrial defect | 0333 | 球細胞腦白質失養症 | Globoid Cell Leukodystrophy |

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| 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 | 持續性幼年型胰島素過度分泌 低血糖症 | Persistent hyperinsulinemic hypoglycemia of infancy | 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 |
| 04、心肺功能失調 | | | | | |
| 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 (心節律障礙暨週期性 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 | | | |
| 05、消化系統失調 | | | | | |
| 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 | 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 |
| 06、泌尿系統失調 | | | | | |
| 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 | Bartter 氏症候群 | Bartter's syndrome |
| 07、腦部或神經病變 | | | | | |
| 0701 | 毛樣腦血管疾病 | Moya moya disease | 0720 | 神經元蠟樣脂褐質儲積症 | 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 | 酪胺酸羥化酶缺乏症 | 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 | 泛酸鹽激酶相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 | 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 | Staidosis | 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 | 腦肋小鎖症候群 | 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 |
| 08、皮膚病變 | | | | | |
| 0801 | 遺傳性表皮分解性水疱症 | Hereditary epidermolysis bullosa | 0809 | 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 | 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 | 水泡型先天性魚鱗癬樣紅皮症 | Bullous Congenital ichthyiform 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 | | | |
| 09、肌肉病變 | | | | | |
| 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 | 面肩胛肱肌失養症 | Facioscapulohumeral muscular dystrophy | | | |
| 10、骨頭病變 | | | | | |
| 1001 | 成骨不全症(玻璃娃娃) | Osteogenesis imperfecta | 1008 | 骨骼發育異常 | Spondyloepiphyseal Dysplasia(SED) |
| 1002 | 軟骨發育不全症(小小人兒) | Achondroplasia | 1009 | 裂手裂足症 | Split-hand/ Split-foot malformation (SHFM) |
| 1003 | 骨質石化症(大理石寶寶) | Osteopetrosis | 1010 | 假性軟骨發育不全 | Pseudochondroplastic 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 | 多發性母髌發育不全症 | Multiple Epiphyseal Dysplasia |
| 1006 | 鎖骨顛骨發育異常 | Cleidocranial dysplasia | 1013 | 次軟骨發育不全症 | Hypochondroplasia |

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| 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 |
| 11、結締組織病變 | | | | | |
| 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 |
| 12、造血功能異常 | | | | | |
| 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 | α 1-抗胰蛋白酶缺乏症 | α 1- Antitrypsin deficiency | 1209 | 蛋白質 S 缺乏症 | Protein S Deficiency |
| 13、免疫疾病 | | | | | |
| 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 | γ 干擾素受體 1 缺陷 | Interferon γ 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 |
| 14、內分泌疾病 | | | | | |
| 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 (W A G R 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 | 1α -羥化酶缺乏症候群 | 1α -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 |
| 15、不正常細胞增生瘤 | | | | | |
| 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 | Lymphangi leiomyomatosis(LAM) |
| 1504 | 神經母細胞瘤 | Neuroblastoma | 1507 | 達希伯-林道症候群 | Von Hippel-Lindau(VHL) |
| 16、外觀異常 | | | | | |
| 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-Ptosis-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-齶-指(趾)症候群 | Oto-Palato-Digital syndrome |

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| 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 (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 | 多發性翼狀腺症候群 | Multiple pterygium syndrome | 1627 | 懷特-薩頓症候群 | White-Sutton Syndrome |
| 1614 | 努南氏症 | Noonan syndrome | | | |
| 17、染色體異常 | | | | | |
| 1701 | Prader-Willi 氏症候群 (小胖威利、好吃費寶) | 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 | | | |
| 18、其他分類或不明原因 | | | | | |
| 1801 | 早老症 | Hutchinson Gilford progeria syndrome | 1809 | 先天性靜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 | Stargard' s 氏症 | Stargard' s disease |
| 1804 | 髮一肝一腸症候群 | 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。