

公告之罕見疾病分類序號彙總表

107/5/30(依據衛生福利部 107 年 5 月 30 日衛授國字第 1070401231 號)

| 分類序號 | 中文病名 (僅供參考) | 英文病名(縮寫) | ICD-9- CM 編碼 | ICD-10- CM 編碼 | 備註 |
|--|-------------|-----------------------|---|------------------|---|
| A. 先天性代謝異常 | | | | | |
| ◎A1 尿素循環代謝異常 Urea cycle disorders (高血氨症) | | | | | |
| A1 | 01 | 先天性尿素循環代謝障礙 | Congenital Urea cycle disorders | 270.6 | E72.20 |
| | 02 | 瓜胺酸血症 | Citrullinemia | 270.6 | E72.23 |
| | 03 | 乙醯穀胺酸合成酶缺乏症 | Nitroacetylglutamate synthetase deficiency, NAG synthetase deficiency | 270.6 | E72.29 |
| | 04 | 鳥胺酸氨甲醯基轉移酶缺乏症 | Ornithine transcarbamylase deficiency | 270.6 | E72.4 |
| | 05 | 高鳥胺酸血症-高氨血症-高瓜胺酸血症症候群 | Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome | 270.6 | E72.4 |
| ◎A2 胺基酸/有機酸代謝異常 Amino acid metabolic disorders / Organic acidemias | | | | | |
| A2 | 01 | 胺基酸代謝疾病 | Amino acid metabolic disorders(Aminoacidopathies) | 270.9 | E72.8 |
| | 02 | 高胱胺酸血症 | Homocystinuria | 270.4 | E72.11 |
| | 03 | 高甲硫胺酸血症 | Hypermethioninemia | 270.4 | E72.19 |
| | 04 | 非酮性高甘胺酸血症 | Nonketotic hyperglycinemia | 270.7 | E72.51 |
| | 05 | 苯酮尿症 | Phenylketouria | 270.1 | E70.0 |
| | 06 | 四氫基喋呤缺乏症 | Tetrahydrobiopterin deficiency | 270.1 | E70.1 |
| | 07 | 遺傳性高酪胺酸血症 | Hereditary tyrosinemia | 270.2 | E70.21 |
| | 08 | 楓糖尿症 | Maple syrup urine disease | 270.3 | E71.0 |
| | 09 | 有機酸血症 | Organic acidemias | 270.9 | E71.118 |
| | 10 | 異戊酸血症 | Isovaleric academia | 270.3 | E71.110 |
| | 11 | 戊二酸尿症，第一型、第二型 | Glutaric aciduria type I、II | 270.9 | type I : E72.3 type II : E71.313 |
| | 12 | 丙酸血症 | Propionic academia | 270.3 | E71.121 |

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|-----------|--------------------------|---|------------------------|--------------------|--|--|
| 13 | 甲基丙二酸血症 | Methylmalonic acidemia | 270.3 | E71.120 | | |
| 14 | 3-氫基-3-甲基戊二酸血症 | 3-Hydroxy-3-methyl-glutaric acidemia | 270.9 | E71.118 | | |
| 15 | 典型苯酮尿症合併蔗糖酶同麥芽糖酶缺乏症 | PAH type PKU combine with Sucrase-isomaltase deficiency | 271.3 + 270.1 | E74.31+E 70.0 | | |
| 16 | 高離氨基酸血症 | Hyperlysinemia | 270.7 | E72.3 | | |
| 17 | 組胺酸血症 | Histidinemia | 270.5 | E70.41 | | |
| 18 | 三甲基巴豆醯輔酶 A 羧化酵素缺乏症 | 3-Methylcrotonyl-CoA carboxylase deficiency | 270.9 | E71.19 | | |
| 19 | 多發性羧化酶缺乏症 | Multiple carboxylase deficiency | 270.9 | D81.819 | | |
| 20 | 高脯胺酸血症 | Hyperprolinemia | 270.8 | E72.59 | | |
| 21 | 芳香族 L-胺基酸類脫羧基酶缺乏症 | Aromatic L-amino acid decarboxylase deficiency | 270.2 | E70.9 | | |
| 22 | 酪胺酸羥化酶缺乏症 | Tyrosine hydroxylase deficiency | 270.2 | E70.20 | | |
| 23 | 甲基丙二酸血症併高胱胺酸血症 (Cb1 C 型) | Cobalamin C defect (Methylmalonic aciduria and Homocystinuria, cb1C type) | | E71.120+ E72.11 | | |
| ◎ A3 脂質儲積 | | | | | | |
| A3 | 01 | 高雪氏症 | Gaucher's disease | 272.7 | E75.22 | |
| | 02 | GM1/GM2 神經節苷脂儲積症 | GM1/GM2 gangliosidosis | 330.1 | GM1 : E75.19 GM2 : E75.00 | |
| | 03 | Fabry 氏症 | Fabry disease | 272.7 | E75.21 | |
| | 04 | Niemann-Pick 氏症，鞘髓磷脂儲積症 | Niemann-Pick disease | 272.7 | type A : E75.240 type B : E75.241 type C : E75.242 type D : E75.243 other : E75.248 | |

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|---------------|----|----------------------------------|--|-----------------|--|----|
| | | | | | unspedif ied : E75.249 | |
| | 05 | MLD 症候群 | Metachromatic Leukodystrophy (MLD) | 330.0 | E75.25 | |
| | 06 | 球細胞腦白質失養症 | Globoid Cell Leukodystrophy (Krabbe' s disease) | 330.0 | E75.23 | |
| | 07 | 嬰兒型溶酶體酸性脂肪 酶缺乏症 (又稱伍爾曼氏 症) | Infantile form Lysosomal Acid Lipase Deficiency (Wolman Disease) | 272.7 | E75.5 | |
| ◎A4 碳水化合物代謝異常 | | | | | | |
| A4 | 01 | 半乳糖血症 | Galactosemia | 271.1 | E74.21 | |
| | 02 | 肝醣儲積症 | Glycogen storage disease | 271.0 | type 0 : E74.09 type I : E74.01 type II : E74.02 type III : E74.03 type IV : E74.09 type V : E74.04 type VI- IX : E74.09 Von Gierke's :E74.01 | |
| | 03 | 腦血管屏障葡萄糖輸送 缺陷 | Glut (Glucose Transport) l deficiency syndrome | 271.8 | E74.8 | |

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| ◎ A5 脂肪酸氧化異常 | | | | | |
| A5 | 01 | 脂肪酸氧化作用缺陷 | Fatty acid oxidation defect | 277.8 | E71.30 E71.310 E71.311 E71.312 E71.313 E71.314 E71.318 E71.32 E71.39 |
| | 02 | 原發性肉鹼缺乏症 | Carnitine deficiency syndrome, primary | 272.9 | E71.41 |
| | 03 | 中鏈脂肪酸去氫酵素缺乏症 | Medium-chain acyl-coenzyme A dehydrogenase deficiency (MCAD) | 277.8 | E71.311 |
| | 04 | 短鏈脂肪酸去氫酶缺乏症 | Short-chain acyl-CoA dehydrogenase deficiency | 277.8 | E71.312 |
| ◎ A6 粒線體代謝異常 | | | | | |
| A6 | 01 | 粒線體缺陷 | Mitochondrial defect | 277.9 | E88.40 |
| | 02 | Kearns-Sayre 氏症候群 | Kearns-Sayre syndrome | 277.8 | H49.811 H49.812 H49.813 H49.819 |
| | 03 | Leigh 氏童年期腦脊髓病變 | Leigh disease | 330.8 | G31.82 |
| | 04 | MELAS 症候群 | MELAS | 758.89 | E88.41 |
| | 05 | MNGIE 症候群粒線體性神經胃腸腦病變症候群 | Mitochondrial Neurogastrointestinal Encephalopathy Syndrome | 277.9 | E88.49 |
| | 06 | 丙酮酸鹽脫氫酶缺乏症 | Pyruvate dehydrogenase deficiency | 271.8 | E74.4 |
| | 07 | 巴氏症候群 | Barth Syndrome | 759.89 | E78.71 |
| ◎ A7 溶小體代謝異常 | | | | | |
| A7 | 01 | 胱胺酸血症 | Cystinosis | 270.0 | E72.04 |
| | 02 | 黏多醣症 | Mucopolysaccharidoses | 277.5 | Type I : |

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| | | | | | E76. 01 E76. 02 E76. 03 Type II : E76. 1 Other : E76. 210 E76. 211 E76. 219 E76. 22 E76. 29 Unspecif ied : E76. 3 | |
| | 03 | 岩藻糖代謝異常 (儲積症) | Fucosidosis | 271. 8 | E77. 1 | |
| | 04 | 涎酸酵素缺乏症 | Sialidosis | 272. 7 | E77. 1 | |
| | 05 | 黏脂質症 | Mucopolipidosis | 272. 7 | type I : E77. 1 type II、III : E77. 0 type IV : E75. 11 | |
| | 06 | 神經元蠟樣脂褐質儲積症 | Neuronal ceroid lipofuscinosis | 330. 1 | E75. 4 | |
| | 07 | 多發性硫酸脂酶缺乏症 | Multiple sulfatase deficiency | | E75. 29 | |
| ◎ A8 膽固醇及脂質代謝異常 Cholesterol and Lipid metabolism | | | | | | |
| A8 | 01 | 同合子家族性高膽固醇血症 | Homozygous familial hypercholesterolemia | 272. 0 | E78. 0 | |
| | 02 | 家族性高乳糜微粒血症 | Familial Hyperchylomicronemia | 272. 3 | E78. 3 | |
| | 03 | 豆固醇血症 〈植物性〉 | Sitosterolemia | 272. 0 | E78. 0 | |

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| ◎A9 礦物離子缺陷 | | | | | | |
| A9 | 01 | 威爾森氏症 | Wilson's disease | 275.1 | E83.01 | |
| | 02 | Menkes 症候群 | Menkes syndrome | 759.89 | E83.09 | |
| | 03 | 鉬輔酶缺乏症 | Molybdenum cofactor deficiency | 277.8 | E61.5 | |
| ◎A10 過氧化體代謝異常 | | | | | | |
| A10 | 01 | Zellweger 氏症候群 | Zellweger syndrome | 277.9 | E71.510 | |
| | 02 | 腎上腺腦白質失養症 | Adrenoleukodystrophy | 272.7 | E71.511 E71.520 E71.521 E71.528 E71.529 | |
| | 03 | 肢近端型點狀軟骨發育不良 | Rhizomelic Chondrodysplasia Punctata | 277.8 | E71.540 | |
| ◎A11 其他代謝異常 | | | | | | |
| A11 | 01 | 紫質症 | Porphyria | 277.1 | E80.20 E80.21 E80.29 | |
| | 02 | Lesch-Nyhan 氏症候群 | Lesch-Nyhan syndrome | 277.2 | E79.1 | |
| | 03 | 亞硫酸鹽氧化酶缺乏 | Sulfite oxidase deficiency | 270.0 | E72.19 | |
| | 04 | 碳水化合物缺乏糖蛋白症候群 | Carbohydrate-deficiency glycoprotein syndrome | 277.9 | E77.8 | |
| | 05 | 三甲基胺尿症 | Trimethylaminuria | 277.8 | E72.52 | 修正中文病名 |
| | 06 | 先天性全身脂質營養不良症 | Congenital generalized lipodystrophy | 272.6 | E88.1 | |
| | 07 | 腦腱性黃瘤症 | Cerebrotendinous Xanthomatosis | 272.7 | E75.5 | |
| | 08 | 低磷酸酯酶症 | Hypophosphatasia | 275.3 | E83.39 E83.31 | |
| | 09 | Beta 硫解酶缺乏症 | Beta-Ketothiolase Deficiency | 270.3 | E71.19 | |
| | 10 | 生物素酶缺乏症 | Biotinidase Deficiency | 277.6 | D81.810 | |
| B 腦部或神經系統病變 | | | | | | |

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| B1 | 01 | 多發性硬化症 | Multiple sclerosis | 340 | G35 | |
| | 02 | 肌萎縮性側索硬化症 | Amyotrophic lateral sclerosis (ALS) | 335.20 | G12.21 | |
| | 03 | 共濟失調微血管擴張症候群 | Ataxia telangiectasia | 334.8 | G11.3 | |
| | 04 | 亨丁頓氏舞蹈症 | Huntington disease(又稱 Huntington's chorea) | 333.4 | G10 | |
| | 05 | 瑞特氏症候群 | Rett syndrome | 330.8 | F84.2 | |
| | 06 | 脊髓性肌肉萎縮症 | Spinal muscular atrophy | 335.10 | G12.9 | |
| | 07 | 脊髓小腦退化性動作協調障礙 | Spinocerebellar ataxia | 334.3 | G11.1 | |
| | 08 | 結節性硬化症 | Tuberous sclerosis | 759.5 | Q85.1 | |
| | 09 | 先天性痛不敏感症合併無汗症 | Congenital insensitivity to pain with anhidrosis (CIPA) | 705.0 | L74.4 | |
| | 10 | 神經纖維瘤症候群第二型 | Neurofibromatosis type II | 237.72 | Q85.02 | |
| | 11 | Alexander 氏病 | Alexander disease | 331.89 | E75.29 | |
| | 12 | 僵體症候群 | Stiffperson syndrome | 333.91 | G25.82 | |
| | 13 | 遺傳性痙攣性下身麻痺 | Hereditary spastic paraplegia | 334.1 | G11.4 | |
| | 14 | Joubert 氏症候群 (家族性小腦蚓部發育不全) | Joubert syndrome | 759.89 | Q04.3 | |
| | 15 | Pelizaeus-Merzbacher 氏症 (慢性兒童型腦硬化症) | Pelizaeus-Merzbacher Disease | 330.0 | E75.29 | |
| | 16 | Charcot Maire Tooth 氏症 (進行性神經性腓骨萎縮症) | Charcot Marie Tooth Disease | 356.1 | G60.0 | |
| | 17 | 甘迺迪氏症 (脊髓延髓性肌肉萎縮症) | Kennedy Disease | 335.8 | G12.20 G12.21 G12.22 G12.29 | |
| | 18 | 家族性澱粉樣多發性神經病變 | Familial Amyloidotic Polyneuropathy | 277.3 + 357.4 | E85.1 | |
| | 19 | Moebius 症候群 | Moebius syndrome | 352.6 | Q87.0 | |
| | 20 | McLeod 症候群 | McLeod syndrome | 758.81 | Q97.8 | |

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| | | | | | Q98.8 | |
| | 21 | Aicardi-Goutieres 症候群 | Aicardi-Goutieres syndrome | 330.0 | G31.89 | |
| | 22 | 普洛提斯症候群 | Proteus Syndrome | 759.89 | Q87.3 | |
| | 23 | MECP2 綜合症候群 | Methyl CpG binding protein 2 Duplication Syndrome (MECP2 Duplication Syndrome) | 330.8 | Q99.8 | |
| | 24 | 腦肋小頷症候群 | Cerebro-Costo-Mandibular Syndrome | 759.89 | Q87.89 | |
| | 25 | Dravet 症候群 | Dravet Syndrome, DS | | G40.311 | 新增 |
| | 26 | 腦白質消失症 | Vanishing White Matter Disease | | G37.8 | 新增 |
| C 呼吸循環系統病變 | | | | | | |
| C1 | 01 | 特發性嬰兒動脈硬化症 | Idiopathic Infantile Arterial Calcification | 747.89 | Q28.8 | |
| | 02 | 囊狀纖維化症 | Cystic fibrosis | 277.00 | E84.9 | |
| | 03 | 原發性肺動脈高壓 | Primary Pulmonary Hypertension (PPH) | 416.0 | I27.0 | |
| | 04 | Holt-Oram 氏症候群 | Holt-Oram Syndrome | 759.89 | Q87.2 | |
| | 05 | 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) | Andersen syndrome | 359.3 + 426.89 | E74.09 | |
| | 06 | 遺傳性出血性血管擴張症 | Hereditary Hemorrhagic Telangiectasia | 448.0 | I78.0 | |
| | 07 | 窒息性胸腔失養症 | Asphyxiating thoracic dystrophy | 756.4 | Q77.2 | |
| | 08 | 先天性中樞性換氣不足症候群 | Congenital Central Hypoventilation Syndrome | 327.25 | G47.35 | |
| D 消化系統病變 | | | | | | |
| D1 | 01 | 進行性家族性肝內膽汁滯留症 | Progressive intrahepatic cholestasis, PFIC | 751.69 | K83.1 | |
| | 02 | 先天性膽酸合成障礙 | Inborn errors of bile acid synthesis | 277.9 | E78.70 | |
| | 03 | α 1-抗胰蛋白酶缺乏症 | α 1- Antitrypsin deficiency | 277.6 | E88.01 | |

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| 04 | 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 Dysplasia | 750.5 | Q43.8 | | |
| 05 | 阿拉吉歐症候群 | Alagille Syndrome | 759.89 | Q44.7 | | |
| E 腎臟泌尿系統病變 | | | | | | |
| E1 | 01 | Lowe 氏症候群 | Lowe syndrome | 270.8 | E72.03 | |
| | 02 | Bartter 氏症候群 | Bartter's syndrome | 255.1 | E26.81 | |
| | 03 | 體染色體隱性多囊性腎臟疾病 | Autosomal recessive polycystic kidney disease | 753.14 | Q61.19 | |
| F 皮膚病變 | | | | | | |
| F1 | 01 | 遺傳性表皮分解性水泡症 | Hereditary epidermolysis bullosa | 757.39 | Q81.9 | |
| | 02 | 層狀魚鱗癬 (自體隱性遺傳型) | Lchthyosis, lamellar recessive | 757.1 | Q80.2 | |
| | 03 | 膠膜兒 | Collodion baby | 757.1 | Q80.2 | |
| | 04 | 斑色魚鱗癬 | Harlequin ichthyosis | 757.1 | Q80.4 | |
| | 05 | 水泡型先天性魚鱗癬樣紅皮症 (表皮鬆解性角化過度症) | Bullous Congenital ichthyosiform erythroderma (epidermolytic hyperkeratosis) | 757.1 | Q80.3 | |
| | 06 | 外胚層增生不良症 | Ectodermal Dysplasias | 757.31 | Q82.4 | |
| | 07 | Meleda 島病 | Meleda disease | 757.39 | Q82.8 | |
| | 08 | Darier 氏症 (毛囊角化病) | Darier's disease | 757.39 | Q82.8 | |
| | 09 | 先天性角化不全症 | Dyskeratosis Congenita | 757.39 | Q82.8 | |
| | 10 | 皮膚過度角化症雅司病 | Diffuse Non-epidermolytic Palmoplantar Keratoderma type Unna-Thost | 757.39 | Q82.8 | |
| | 11 | 色素失調症 | Incontinentia Pigmenti | 757.33 | Q82.3 | |
| | 12 | Netherton 症候群 | Netherton Syndrome | 757.1 | Q80.3 | |
| G 肌肉病變 | | | | | | |
| G1 | 01 | 裘馨氏肌肉失養症 | Duchenne muscular | 359.1 | G71.0 | |

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| | | | dystrophy | | | |
| | 02 | Nemaline 線狀肌肉病變 | Nemaline Rod Myopathy | 359.0 | G71.2 | |
| | 03 | Schwartz Jampel 氏症候群 | Schwartz Jampel syndrome | 756.89 | G71.13 | |
| | 04 | 肌肉強直症 | Myotonic dystrophy | 359.2 | G71.11 | |
| | 05 | 面肩胛肱肌失養症 | Facioscapulohumeral muscular dystrophy | 359.1 | G71.0 | |
| | 06 | 肌小管病變 | Myotubular Myopathy | 359.0 | G71.2 | |
| | 07 | 貝克型肌肉失養症 | Becker Muscular Dystrophy | 359.1 | G71.0 | |
| | 08 | Freeman-Sheldon 氏症候群 | Freeman-Sheldon syndrome | 759.89 | Q87.0 | |
| | 09 | 肢帶型肌失養症 | Limb-girdle muscular dystrophy | 359.1 | G71.0 | |
| | 10 | 先天性肌失養症 | Congenital Muscular Dystrophy | 359.0 | G71.0 | |
| | 11 | 中心軸空肌病 | Central Core Disease | 359.0 | G71.2 | |
| | 12 | 多微小軸空肌病 | Multiminicore Disease | 359.0 | G71.2 | |
| | 13 | Emery - Dreifuss 肌失養症 | Emery-Dreifuss Muscular Dystrophy (EDMD) | | G71.0 | 修正分類 序號 |
| H 骨及軟骨病變 | | | | | | |
| H1 | 01 | 軟骨發育不全症 | Achondroplasia | 756.4 | Q77.4 | |
| | 02 | 成骨不全症 | Osteogenesis imperfecta | 756.51 | Q78.0 | |
| | 03 | 原發性變形性骨炎 | Primary Paget disease | 731.0 | M88.0 M88.1 M88.811 M88.812 M88.819 M88.821 M88.822 M88.829 M88.831 M88.832 M88.839 M88.841 | |

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| | | | | | M88.842 M88.849 M88.851 M88.852 M88.859 M88.861 M88.862 M88.869 M88.871 M88.872 M88.879 M88.88 M88.89 M88.9 | |
| | 04 | 鎖骨顱骨發育異常 | Cleidocranial dysplasia | 755.59 | Q74.0 | |
| | 05 | 進行性骨化性肌炎 | Fibrodysplasia Ossificans Progressiva | 728.11 | M61.10 M61.111 M61.112 M61.119 M61.121 M61.122 M61.129 M61.131 M61.132 M61.139 M61.141 M61.142 M61.143 M61.144 M61.145 M61.146 M61.151 M61.152 M61.159 M61.161 M61.162 M61.169 M61.171 | |

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|----------|----|--------------------|---|----------------------------------|--|----|
| | | | | | M61.172 M61.173 M61.174 M61.175 M61.176 M61.177 M61.178 M61.179 M61.18 M61.19 | |
| | 06 | 裂手裂足症 | Split-hand/ Split-foot malformation (SHFM) | hand755. 58 foot755. 67 | Q71.60 Q71.61 Q71.62 Q71.63 Q72.70 Q72.71 Q72.72 Q72.73 | |
| | 07 | 骨質石化症 | Osteopetrosis | 756.52 | Q78.2 | |
| | 08 | 假性軟骨發育不全 | Pseudoachondroplastic dysplasia | 756.4 | Q77.8 | |
| | 09 | 多發性骨骺發育不全症 | Multiple Epiphyseal Dysplasia | 756.56 | Q78.3 | |
| I 結締組織病變 | | | | | | |
| I1 | 01 | 先天結締組織異常第四 型 | Ehlers Danlos syndrome IV | 756.83 | Q79.6 | |
| J 血液疾病 | | | | | | |
| J1 | 01 | 重型海洋性貧血 | Thalassemia major | 282.4 | D56.0 D56.1 | |
| | 02 | 血小板無力症 | Thrombasthenia | 287.1 | D69.1 | |
| | 03 | 同基因合子蛋白質 C 缺乏 症 | Homozygous protein C deficiency | 273.3 | D68.59 | |
| | 04 | 陣發性夜間血紅素尿症 | Paroxysmal Nocturnal Hemoglobinuria | 283.2 | D59.5 | |
| | 05 | 非典型性尿毒溶血症候 群 | Atypical Hemolytic Uremic Syndrome | 283.11 | D59.3 | |

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|---------|-------------|---------------------------|--|---|----------------------------------|------------|
| K 免疫疾病 | | | | | | |
| K1 | 01 | 原發性慢性肉芽腫病 | Chronic primary granulomatous disease | 288.1 | D71 | |
| | 02 | 先天性高免疫球蛋白 E 症 候群 | Congenital Hyper IgE syndrome | 288.1 | D82.4 | |
| | 03 | 布魯頓氏低免疫球蛋白 血症 | Bruton' s agammaglobulinemia | 279.04 | D80.0 | |
| | 04 | Wiskott- Aldrich 氏症候 群 | Wiskott- Aldrich Syndrome | 279.12 | D82.0 | |
| | 05 | 嚴重複合型免疫缺乏症 | Severe combined immunodeficiency | 279.2 | D81.0 D81.1 D81.2 D81.9 | |
| | 06 | 補體成份 8 缺乏症 | Complement Component 8 deficiency | 279.8 | D84.1 | |
| | 07 | IPEX 症候群 | IPEX Syndrome | 759.89 (279.8 , 569.89 , 259.8, 758.89) | E31.0 | |
| | 08 | 高免疫球蛋白 M 症候群 | Hyper-IgM syndrome | 279.05 | D80.5 | |
| | 09 | γ 干擾素受體 1 缺陷 | Interferon γ receptor 1 deficiency | 279.4 | D84.8 | |
| | 10 | 遺傳性血管性水腫 | Hereditary Angioedema (HAE) | | D84.1 | 修正分類 序號 |
| L 內分泌疾病 | | | | | | |
| L1 | 01 | Kenny-Caffey 氏症候群 | Kenny-Caffey syndrome | 759.89 | Q87.1 | |
| | 02 | 假性副甲狀腺低能症 | Pseudohypoparathyroidism | 275.49 | E20.1 | |
| | 03 | 性連遺傳型低磷酸鹽佝 僂症 | X-linked hypophosphatemic rickets | 275.3 | E83.31 | |
| | 04 | Laron 氏侏儒症候群 | Laron syndrome (Laron Dwarfism) | 259.4 | E34.3 | |
| | 05 | Bardet-Biedl 氏症候群 | Bardet-Biedl syndrome | 759.89 | Q87.89 | |
| | 06 | Alstrom 氏症候群 | Alstrom Syndrome | 759.2 | Q87.89 | |

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|------------------|----------------------|--|-----------------------------|--------------|---------------------------------------|--|
| 07 | 持續性幼兒型胰島素過度分泌低血糖症 | Persistent hyperinsulinemic hypoglycemia of infancy (PHHI) | 251.1 | E16.1 | | |
| 08 | Wolfram 氏症候群 | Wolfram syndrome , DIDMOAD | 277.9 | E88.9 | | |
| 09 | McCune Albright 氏症候群 | McCune Albright syndrome | 756.59 | Q78.1 | | |
| 10 | 短指發育不良及性別顛倒 | Campomelic dysplasia with autosomal sex reversal | 758.89 | Q99.8 | | |
| 11 | 腎上腺皮促素抗性 | ACTH resistance | 253.4 | E27.49 | | |
| 12 | 第一型遺傳性維生素 D 依賴型佝僂症 | 25-Hydroxyvitamin D1-Alpha-Hydroxylase Deficiency | 268.0 | E83.32 | 修正 ICD-10-CM 編碼、修正 中英文病 名 | |
| 13 | 先天性腎上腺發育不全 | Congenital adrenal hypoplasia | 759.1 | Q89.1 | | |
| 14 | Kallmann 氏症候群 | Kallmann syndrome | 253.4 | E23.0 | | |
| 15 | 永久性新生兒糖尿病 | Permanent Neonatal Diabetes Mellitus | 775.1 | P70.2 | | |
| M 先天畸形症候群 | | | | | | |
| M1 | 01 | Aarskog-Scott 氏症候群 | Aarskog-Scott syndrome | 759.89 | Q87.1 | |
| | 02 | 瓦登伯格氏症候群 | Waardenburg syndrome | 270.2 | E70.8 | |
| | 03 | 愛伯特氏症 | Apert syndrome | 755.55 | Q87.0 | |
| | 04 | Smith-Lemli-Opitz 氏症候群 | Smith-Lemli-Opitz syndrome | 759.89 | E78.72 | |
| | 05 | Larsen 氏症候群 (顎裂-先天性脫位症候群) | Larsen syndrome | 755.8 | Q74.8 | |
| | 06 | Beckwith Wiedemann 氏症候群 | Beckwith Wiedemann syndrome | 759.89 | Q87.3 | |
| | 07 | Crouzon 氏症候群 | Crouzon syndrome | 756.0 | Q75.1 | |
| | 08 | Fraser 氏症候群 | Fraser syndrome | 759.89 | Q87.0 | |
| | 09 | 多發性翼狀膜症候群 | Multiple pterygium syndrome | 759.89 | Q79.8 | |
| | 10 | Cornelia de Lange 氏症候群 | Cornelia de Lange syndrome | 759.89 | Q87.1 | |

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|---------|------------------------|---|---|------------------------|--------|--|
| 11 | 海勒曼-史德萊夫氏症候群 | Hallerman-Streiff Syndrome | 756.0 | Q87.0 | | |
| 12 | 歌舞伎症候群 | Kabuki syndrome | 759.89 | Q89.8 | | |
| 13 | 耳-齶-指(趾)症候群 | Oto-Palato-Digital syndrome | 759.89 | Q87.0 | | |
| 14 | Conradi-Hunermann 氏症候群 | Conradi-Hunermann syndrome | 756.59 | Q77.3 | | |
| 15 | Treacher Collins 氏症候群 | Treacher Collins Syndrome | 756.0 | Q75.4 | | |
| 16 | Robinow 氏症候群 | Robinow Syndrome | 759.89 | Q87.1 | | |
| 17 | Pfeiffer 氏症候群 | Pfeiffer syndrome | 755.55 | Q87.0 | | |
| 18 | 泛酸鹽激酶關聯之神經退化性疾病 | Pantothenate Kinase Associated Neurodegeneration (PKAN) | 277.9 | G23.0 | | |
| 19 | 指(趾)甲髕骨症候群 | Nail-Patella Syndrome | 756.89 | Q87.2 | | |
| 20 | CFC 症候群 | Cardiofaciocutaneous Syndrome | 759.89 | Q87.89 | | |
| 21 | Peters-Plus 症候群 | Peters-Plus syndrome | 743.44 | Q13.4 | | |
| 22 | Nager 症候群 | Nager Syndrome | 756.0 | Q75.4 | | |
| 23 | CHARGE 症候群 | CHARGE Syndrome | 759.89 | Q89.8 | | |
| 24 | 懷特-薩頓症候群 | White-Sutton syndrome | | Q99.88 F84.8 F78 | | |
| N 染色體異常 | | | | | | |
| N1 | 01 | Angelman 氏症候群 | Angelman syndrome | 759.89 | Q93.5 | |
| | 02 | DiGeorge's 症候群 | DiGeorge's syndrome | 279.11 | D82.1 | |
| | 03 | Prader-Willi 氏症候群 | Prader-Willi syndrome | 759.81 | Q87.1 | |
| | 04 | 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) | W A G R syndrome (Wilms' tumor-Aniridia-Genitourinary Anomalies-mental Retardation) | 759.89 | Q87.89 | |
| | 05 | Miller Dieker 症候群 | Miller Dieker syndrome | 742.2 | Q93.88 | |
| | 06 | Rubinstein-Taybi 氏症候群 | Rubinstein-Taybi syndrome | 759.89 | Q87.2 | |

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|--------------|----|-------------------------------------|--|-----------------|------------------|----|
| | 07 | 威廉斯氏症候群 | Williams Syndrome | 759.89 | Q93.89 | |
| | 08 | Von Hippel - Lindau 症候群 | Von Hippel - Lindau disease | 759.6 | Q85.8 | |
| | 09 | Branchio-Oto-Renal 症候群 (BOR 症候群) | Branchio-Oto-Renal Syndrome (BOR Syndrome) | 759.89 | Q87.89 | |
| Z 其他未分類或不明原因 | | | | | | |
| Z1 | 01 | Cockayne 氏症候群 | Cockayne syndrome | 759.89 | Q87.1 | |
| | 02 | 早老症 | Hutchinson Gilford progeria syndrome | 259.8 | E34.8 | |
| | 03 | 髮-肝-腸症候群 | Tricho-hepato-enteric syndrome | 759.7 | Q89.7 | |
| | 04 | Stargardt's 氏症 | Stargardt's disease | 362.75 | H35.50 | |
| | 05 | 隱匿性黃斑部失養症 | Occult Macular Dystrophy ;OMD | 362.76 | H35.50 | |

※黃色底列疾病為 107/5/30 函文新增