衛生福利部國民健康署

Health Promotion Administration, Ministry of Health and Welfare

113 年 3 月罕見疾病通報個案統計表

Statistical Report of Rare Disease Confirmed Cases in Taiwan, MAR, 2024

統計期間:個案數至 113 年 3 月 31 日及死亡數至 112 年 12 月 31 日

Statistical Period: The number of cases until 31th MAR 2024 and deaths until 31th DEC 2023

製表時間：113 年 4 月 17 日

Tabulation Date: 17th APR 2024

分類序號 No.	疾病名稱 The name of each disorder or disease	ICD10 碼 ICD-10-CM	個案數 The Number of Cases	死亡數 The Number of Deaths
A 先天性代謝異常 Inborn errors of metabolism				
◎ A1 尿素循環代謝異常 Urea cycle disorders				
01	Congenital urea cycle disorders(先天性尿素循環代謝障礙)	E72.20	83	27
02	Citrullinemia(瓜胺酸血症)	E72.23	136	3
03	Nitroacetylglutamate synthetase deficiency,NAG synthetase deficiency(乙醯穀胺酸合成酶缺乏症)	E72.29	0	0
04	Ornithine transcarbamylase deficiency(鳥胺酸氨甲醯基轉移酶缺乏症)	E72.4	29	5
05	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome(高鳥胺酸血症-高氨血症-高瓜胺酸血症症候群)	E72.4	3	0
◎ A2 胺基酸/有機酸代謝異常 Disorders of amino acid/organic acid metabolism				
01	Amino acid metabolic disorders(Aminoacidopathies)(胺基酸代謝疾病)	E72.8	14	3
02	Homocystinuria(高胱胺酸血症)	E72.11	37	1
03	Hypermethioninemia(高甲硫胺酸血症)	E72.19	31	1
04	Nonketotic hyperglycinemia(非酮性高甘胺酸血症)	E72.51	28	17
05	Phenylketouria(苯酮尿症)	E70.0	315	2
06	Tetrahydrobiopterin deficiency(四氫基喋呤缺乏症)	E70.1	6	0
07	Hereditary tyrosinemia(遺傳性高酪胺酸血症)	E70.21	11	1
08	Maple syrup urine disease(楓糖尿症)	E71.0	41	12
09	Organic acidemias(有機酸血症)	E71.118	28	4
10	Isovaleric academia(異戊酸血症)	E71.110	14	2
11	Glutaric aciduria typeI、II(戊二酸尿症，第一型、第二型)	type I: E72.3 type II : E71.313	118	11
12	Propionic academia(丙酸血症)	E71.121	17	5
13	Methylmalonic acidemia (甲基丙二酸血症)	E71.120	72	13
14	3-Hydroxy-3-methylglutaric acidemia (3-羥基-3-甲基戊二酸血症)	E71.118	7	0

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15	PAH type PKU combine with Sucrase-isomaltase deficiency(典型苯酮尿症合併蔗糖酶同麥芽糖酶缺乏症)	E74.31+ E70.0	2	0
16	Hyperlysinemia(高離胺酸血症)	E72.3	0	0
17	Histidinemia(組胺酸血症)	E70.41	0	0
18	3-Methylcrotonyl-CoA carboxylase deficiency(三甲基巴豆醯輔酶 A 羧化酵素缺乏症)	E71.19	77	0
19	Multiple carboxylase deficiency(多發性羧化酶缺乏症)	D81.819	10	0
20	Hyperprolinemia(高脯胺酸血症)	E72.59	2	0
21	Aromatic L-amino acid decarboxylase deficiency(芳香族 L-胺基酸類脫羧基酶缺乏症)	E70.9	69	21
22	Tyrosine hydroxylase deficiency(酪胺酸羥化酶缺乏症)	E70.20	15	1
23	Cobalamin C defect (Methylmalonic aciduria and Homocystinuria, cbl C type)(甲基丙二酸血症併高胱胺酸血症, Cbl C 型)	E71.120+ E72.11	2	0
24	Primary hyperoxaluria(原發性高草酸鹽尿症)	E72.53	1	0
25	Alkaptonuria(黑尿症)	E70.29	4	0
◎ A3 溶小體儲積症 Lysosomal storage disorders				
01	Gaucher disease (高雪氏症)	E75.22	43	9
02	GM1/GM2 gangliosidosis(GM1/GM2 神經節苷脂儲積症)	GM1: E75.19 GM2: E75.00	8	6
03	Fabry disease(Fabry 氏症)	E75.21	619	62
04	Niemann-Pick disease(Niemann-Pick 氏症，鞘髓磷脂儲積症)	E75.240: Type A E75.241: Type B E75.242: Type C E75.243: Type D E75.248: other E75.249 : unspecified	37	14
05	Metachromatic leukodystrophy(MLD)(MLD 症候群)	E75.25	22	13
06	Globoid cell leukodystrophy(Krabbe's disease)(球細胞腦白質失養症)	E75.23	2	0
07	Infantile form lysosomal acid lipase deficiency (Wolman disease)(嬰兒型溶酶體酸性脂肪酶缺乏症(又稱伍爾曼氏症))	E75.5	1	0
08	Cystinosis(胱胺酸血症)	E72.04	7	0

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09	Mucopolysaccharidoses(黏多醣症)	Type1: E76.01 E76.02 E76.03 Type2: E76.1 Other: E76.210 E76.211 E76.219 E76.22 E76.29 Unspecified:E76.3	215	85
10	Fucosidosis(岩藻糖代謝異常(儲積症))	E77.1	5	1
11	Sialidosis(涎酸酵素缺乏症)	E77.1	37	8
12	Mucolipidosis(黏脂質症)	type I: E77.1 typeII、III: E77.0 type IV :E75.11	13	1
13	Neuronal ceroid lipofuscinosis(神經元蠟樣脂褐質儲積症)	E75.4	11	4
14	Multiple sulfatase deficiency(多發性硫酸脂酶缺乏症)	E75.29	2	0
◎ A4 碳水化合物代謝異常 Disorders of carbohydrate metabolism				
01	Galactosemia(半乳糖血症)	E74.21	32	0
02	Glycogen storage disease(肝醣儲積症)	E74.09: type 0 E74.01: Type I E74.02: type II E74.03: type III E74.09: type IV E74.04: type V E74.09: type VI-XI E74.01: Von Gierke's	267	55
03	Glut(Glucose transport)1 deficiency syndrome(腦血管屏障葡萄糖輸送缺陷)	E74.8	11	0

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04	Transaldolase deficiency(轉醛醇酶缺乏症)	E74.8	1	0
◎ A5 脂肪酸氧化異常 Disorders of fatty acid oxidation				
01	Fatty acid oxidation defect(脂肪酸氧化作用缺陷)	E71.30 E71.310 E71.311 E71.312 E71.313 E71.314 E71.318 E71.32 E71.39	50	6
02	Carnitine deficiency syndrome, primary(原發性肉鹼缺乏症)	E71.41	220	8
03	Medium-chain acyl-coenzyme A dehydrogenase deficiency(MCAD)(中鏈脂肪酸去氫酵素缺乏症)	E71.311	9	0
04	Short-chain acyl-CoA dehydrogenase deficiency(短鏈脂肪酸去氫酶缺乏症)	E71.312	8	1
◎ A6 粒線體異常 Mitochondrial disorders				
01	Mitochondrial defect(粒線體缺陷)	E88.40	413	100
02	Kearns-Sayre syndrome(Kearns-Sayre 氏症候群)	H49.811 H49.812 H49.813 H49.819	6	2
03	Leigh disease(Leigh 氏童年期腦脊髓病變)	G31.82	32	15
04	MELAS(MELAS 症候群)	E88.41	128	40
05	Mitochondrial neurogastrointestinal encephalopathy syndrome(MNGIE 症候群粒線體性神經胃腸腦病變症候群)	E88.49	4	1
06	Pyruvate dehydrogenase deficiency(丙酮酸鹽脫氫酶缺乏症)	E74.4	8	1
07	Barth syndrome(巴氏症候群)	E78.71	4	0
08	Leber's hereditary optic neuropathy(雷伯氏遺傳性視神經病變)	H47.22	10	0
◎ A7 維生素代謝異常 Disorders of vitamin metabolism				
01	Biotinidase deficiency(生物素酶缺乏症)	D81.810	9	0
◎ A8 膽固醇及脂質代謝異常 Disorders of cholesterol and lipid metabolism				
01	Homozygous familial hypercholesterolemia(同合子家族性高膽固醇血症)	E78.0	66	2
02	Familial hyperchylomicronemia(家族性高乳糜微粒血症)	E78.3	16	0
03	Sitosterolemia(豆固醇血症(植物性))	E78.0	10	0
04	Congenital generalized lipodystrophy(先天性全身脂質營養不良症)	E88.1	33	15
05	Cerebrotendinous xanthomatosis(腦腱性黃瘤症)	E75.5	8	2
◎ A9 金屬代謝異常 Disorders of metal metabolism				

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01	Wilson's disease(威爾森氏症)	E83.01	735	75
02	Menkes syndrome(Menkes 症候群)	E83.09	18	10
03	Molybdenum cofactor deficiency(鉬輔酶缺乏症)	E61.5	4	2
◎A10 過氧化體異常 Peroxisomal disorders				
01	Zellweger syndrome(Zellweger 氏症候群)	E71.510	10	5
02	Adrenoleukodystrophy(腎上腺腦白質失養症)	E71.511 E71.520 E71.521 E71.528 E71.529	92	38
03	Rhizomelic chondrodysplasia punctata(肢近端型點狀軟骨發育不良)	E71.540	2	0
◎A11 其他代謝異常 Other metabolic disorders				
01	Porphyria(紫質症)	E80.20 E80.21 E80.29	134	9
02	Lesch-Nyhan syndrome(Lesch-Nyhan 氏症候群)	E79.1	15	3
03	Sulfite oxidase deficiency(亞硫酸鹽氧化酶缺乏)	E72.19	12	9
04	Carbohydrate-deficiency glycoprotein syndrome(碳水化合缺乏醣蛋白症候群)	E77.8	20	2
05	Trimethylaminuria(三甲基胺尿症)	E72.52	15	0
06	Hypophosphatasia(低磷酸酯酶症)	E83.39 E83.31	6	1
07	Beta-Ketothiolase deficiency(Beta 硫解酶缺乏症)	E71.19	0	0
08	Cerebral Creatine deficiency(大腦肌酸缺乏症)	E72.8	3	0
09	Thiamine metabolism dysfunction syndromes(硫胺素(維生素 B1)代謝功能障礙症候群)	E51.8	2	0
B 腦部或神經系統異常 Disorders of the brain or nervous system				
01	Multiple sclerosis, MS/Neuromyelitis optica spectrum disorders, NMOSD(多發性硬化症/泛視神經脊髓炎)	G35/ G36.0	2,531	255
02	Amyotrophic lateral sclerosis (ALS) (肌萎縮性側索硬化症)	G12.21	2,014	1,302
03	Ataxia telangiectasia(共濟失調微血管擴張症候群)	G11.3	17	4
04	Huntington disease(又稱 Huntington's chorea) (亨丁頓氏舞蹈症)	G10	506	184
05	Rett syndrome(雷特氏症)	F84.2	180	10
06	Spinal muscular atrophy(脊髓性肌肉萎縮症)	G12.0 G12.1	541	97
07	Spinocerebellar ataxia (脊髓小腦退化性動作協調障礙)	G11.1	1,566	525
08	Tuberous sclerosis(結節性硬化症)	Q85.1	781	47
09	Congenital insensitivity to pain with anhidrosis(CIPA)(先天性	L74.4	12	2

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	痛不敏感症合併無汗症)			
10	Neurofibromatosis type II(神經纖維瘤症候群第二型)	Q85.02	105	24
11	Alexander disease (Alexander 氏病)	E75.29	10	3
12	Stiffperson syndrome(僵體症候群)	G25.82	27	6
13	Hereditary spastic paraplegia(遺傳性痙攣性下身麻痺)	G11.4	221	9
14	Joubert syndrome(Joubert 氏症候群(家族性小腦蚓部發育不全))	Q04.3	17	2
15	Pelizaeus-Merzbacher disease(Pelizaeus-Merzbacher 氏症(慢性兒童型腦硬化症))	E75.29	29	2
16	Charcot Marie Tooth disease(夏柯-馬利-杜斯氏症)	G60.0	584	23
17	Kennedy disease(甘迺迪氏症(脊髓延髓性肌肉萎縮症))	G12.20 G12.21 G12.22 G12.29	131	14
18	Familial amyloidotic polyneuropathy(家族性澱粉樣多發性神經病變)	E85.1	294	89
19	Moebius syndrome(Moebius 症候群)	Q87.0	19	7
20	Mcleod syndrome(Mcleod 症候群)	Q97.8 Q98.8	2	1
21	Aicardi-Goutieres syndrome(Aicardi-Goutieres 症候群)	G31.89	3	1
22	Methyl CpG binding protein 2 Duplication Syndrome(MECP2 Duplication Syndrome)(MECP2 綜合症候群)	Q99.8	11	0
23	Dravet syndrome (DS)(Dravet 症候群)	G40.803 G40.804	109	1
24	Vanishing white matter disease(腦白質消失症)	G37.8	1	1
25	Pantothenate kinase associated neurodegeneration (PKAN) (泛酸鹽激酶關聯之神經退化性疾病)	G23.0	19	3
26	Phospholipsa A2-associated neurodegeneration(PLAN)(磷脂質脂解酶 A2 關聯之神經退化性疾病)	G23.0	4	0
27	Pitt-Hopkins syndrome(皮特-霍普金斯症候群)	Q87.0	6	0
28	Beta-Propeller protein-associated neurodegeneration (BPAN) (Beta 螺旋狀蛋白關聯之神經退化疾病)	G23.0	3	0
29	Infantile-onset ascending hereditary spastic paralysis, IAHSP(嬰兒型上行性遺傳性痙攣性麻痺)	G12.2	2	0
30	Congenital central hypoventilation syndrome(先天性中樞性換氣不足症候群)	G47.35	15	1
31	Von Hippel-Lindau disease(Von Hippel-Lindau 症候群)	Q85.8	37	3
C 呼吸循環系統異常 Disorders of the respiratory/circulation system				
01	Idiopathic infantile arterial calcification(特發性嬰兒動脈硬化	Q28.8	2	1

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	症)			
02	Cystic fibrosis (囊狀纖維化症)	E84.9	22	7
03	Idiopathic or Heritable pulmonary arterial hypertension (IPAH or HPAH）(特發性或遺傳性肺動脈高壓)	I27.0	601	222
04	Holt-Oram syndrome(Holt-Oram 氏症候群)	Q87.2	6	0
05	Andersen syndrome(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))	E74.09	6	0
06	Hereditary hemorrhagic telangiectasia(遺傳性出血性血管擴張症)	I78.0	38	8
07	Asphyxiating thoracic dystrophy(窒息性胸腔失養症)	Q77.2	1	1
D 消化系統異常 Disorders of the digestive system				
01	Progressive familial intrahepatic cholestasis(PFIC)(進行性家族性肝內膽汁滯留症)	K83.1	21	5
02	Inborn errors of bile acid synthesis(先天性膽酸合成障礙)	E78.70	4	0
03	α1- Antitrypsin deficiency(α1-抗胰蛋白酶缺乏症)	E88.01	0	0
04	Congenital interstitial cell of Cajal hyperplasia with neuronal intestinal dysplasia(先天性 Cajal 氏間質細胞增生合併腸道神經元發育異常)	Q43.8	1	1
05	Alagille syndrome(阿拉吉歐症候群)	Q44.7	17	2
06	Tricho-hepato-enteric syndrome(髮-肝-腸症候群)	Q89.7	5	2
E 腎臟泌尿系統異常 Disorders of the renal/urinary system				
01	Lowe syndrome(Lowe 氏症候群)	E72.03	25	3
02	Bartter's syndrome(Bartter 氏症候群)	E26.81	86	2
03	Autosomal recessive polycystic kidney disease(體染色體隱性多囊性腎臟疾病)	Q61.19	9	2
04	Alport syndrome(亞伯氏症候群)	Q87.81	6	1
F 皮膚系統異常 Disorders of the cutaneous system				
01	Hereditary epidermolysis bullosa (遺傳性表皮分解性水泡症)	Q81.0 Q81.1 Q81.2 Q81.8 Q81.9	150	10
02	Ichthyosis, lamellar recessive(層狀魚鱗癬(自體隱性遺傳型))	Q80.2	21	5
03	Collodion baby(膠膜兒)	Q80.2	1	0
04	Harlequin ichthyosis(斑色魚鱗癬)	Q80.4	4	1
05	Bullous Congenital ichthyosiform erythoderma(epidermolytic hyperkeratosis)(水泡型先天性魚鱗癬樣紅皮症(表皮鬆解性角化過度症))	Q80.3	31	2

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06	Ectodermal dysplasias(外胚層增生不良症)	Q82.4	106	2
07	Meleda disease(Meleda 島病)	Q82.8	13	1
08	Darier's disease(Darier 氏症(毛囊角化病))	Q82.8	43	3
09	Dyskeratosis congenita(先天性角化不全症)	Q82.8	3	1
10	Diffuse non-epidermolytic palmoplantar keratoderma type Unna-Thost(皮膚過度角化症雅司病)	Q82.8	13	0
11	Incontinentia pigmenti(色素失調症)	Q82.3	71	1
G 肌肉系統異常 Disorders of the muscular system				
01	Duchenne muscular dystrophy(裘馨氏肌肉失養症)	G71.0	433	143
02	Nemaline rod myopathy(Nemaline 線狀肌肉病變)	G71.2	34	4
03	Schwartz Jampel syndrome(Schwartz Jampel 氏症候群)	G71.13	4	0
04	Myotonic dystrophy(肌肉強直症)	G71.11	302	89
05	Facioscapulohumeral muscular dystrophy(面肩胛肱肌失養症)	G71.0	195	11
06	Myotubular myopathy(肌小管病變)	G71.2	24	5
07	Becker muscular dystrophy(貝克型肌肉失養症)	G71.0	69	13
08	Freeman-Sheldon syndrome(Freeman-Sheldon 氏症候群)	Q87.0	3	0
09	Limb-girdle muscular dystrophy(肢帶型肌失養症)	G71.0	130	8
10	Congenital muscular dystrophy(先天性肌失養症)	G71.0	48	2
11	Central core disease(中心軸空肌病)	G71.2	16	0
12	Multiminicore disease(多微小軸空肌病)	G71.2	4	0
13	Emery–Dreifuss muscular dystrophy (EDMD) (Emery–Dreifuss 肌失養症)	G71.0	8	1
14	GNE myopathy(GNE 遠端肌病變)	G71.8	11	0
15	Stormorken syndrome(史托摩根症候群)	D69.8	2	0
H 骨及軟骨異常 Disorders of bone and cartilage				
01	Achondroplasia(軟骨發育不全症)	Q77.4	435	16
02	Osteogenesis imperfecta(成骨不全症)	Q78.0	413	21
03	Primary Paget disease(原發性變形性骨炎)	M88.0 M88.1 M88.811 M88.812 M88.819 M88.821 M88.822 M88.829 M88.831 M88.832 M88.839 M88.841 M88.842 M88.849 M88.851	4	0

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		M88.852 M88.859 M88.861 M88.862 M88.869 M88.871 M88.872 M88.879 M88.88 M88.89 M88.9		
04	Cleidocraninal dysplasia(鎖骨顱骨發育異常)	Q74.0	50	1
05	Fibrodysplasia ossificans progressiva(進行性骨化性肌炎)	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 M61.172 M61.173 M61.174 M61.175 M61.176 M61.177 M61.178 M61.179 M61.18 M61.19	11	2
06	Split-hand/ Split-foot malformation(SHFM)(裂手裂足症)	Q71.60 Q71.61 Q71.62 Q71.63 Q72.70 Q72.71 Q72.72 Q72.73	13	0
07	Osteopetrosis(骨質石化症)	Q78.2	29	10

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08	Pseudoachondroplastic dysplasia(假性軟骨發育不全)	Q77.8	19	2
09	Multiple epiphyseal dysplasia(多發性骨骺發育不全症)	Q78.3	14	0
10	Craniometaphyseal dysplasia(顱骨幹骺端發育不良)	Q78.8	2	0
11	Cerebro-Costo-Mandibular syndrome(腦肋小頜症候群)	Q87.89	3	0
12	Crouzon syndrome(Crouzon 氏症候群)	Q75.1	79	4
13	Pfeiffer syndrome(Pfeiffer 氏症候群)	Q87.0	7	4
I 結締組織異常 Disorders of the connective tissue				
01	Ehlers Danlos syndromeIV(先天結締組織異常第四型)	Q79.6	24	1
J 血液系統異常 Disorders of the hematologic system				
01	Thalassemia major(重型海洋性貧血)	D56.0 D56.1	404	29
02	Thrombasthenia(血小板無力症)	D69.1	33	1
03	Homozygous proetin C deficiency(同基因合子蛋白質 C 缺乏症)	D68.59	6	1
04	Paroxysmal nocturnal hemoglobinuria(陣發性夜間血紅素尿症)	D59.5	149	27
05	Congenital thrombotic thrombocytopenic purpura(先天性血栓性血小板低下紫斑症)	M31.1	1	0
K 免疫系統異常 Disorders of the immune system				
01	Chronic primary granulomatous disease(原發性慢性肉芽腫病)	D71	29	4
02	Congenital Hyper IgE syndrome(先天性高免疫球蛋白 E 症候群)	D82.4	17	4
03	Bruton's agammaglobulinemia(布魯頓氏低免疫球蛋白血症)	D80.0	25	2
04	Wiskott- Aldrich syndrome(Wiskott- Aldrich 氏症候群)	D82.0	25	10
05	Severe combined immunodeficiency(嚴重複合型免疫缺乏症)	D81.0 D81.1 D81.2 D81.9	28	6
06	Complement component 8 deficiency(補體成份 8 缺乏症)	D84.1	2	0
07	IPEX syndrome(IPEX 症候群)	E31.0	1	1
08	Hyper-IgM syndrome(高免疫球蛋白 M 症候群)	D80.5	11	1
09	Interferon γ receptor 1 deficiency (γ 干擾素受體 1 缺陷)	D84.8	6	0
10	Hereditary angioedema (HAE)(遺傳性血管性水腫)	D84.1	42	0
11	Netherton syndrome(Netherton 症候群)	Q80.3	9	1
12	Atypical hemolytic uremic syndrome(非典型性尿毒溶血症候群)	D59.3	85	23
L 內分泌系統異常 Disorders of the endocrine system				
01	Kenny-Caffey syndrome(Kenny-Caffey 氏症候群)	Q87.1	1	0

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02	Pseudohypoparathyroidism(假性副甲狀腺低能症)	E20.1	74	0
03	X-linked hypophosphatemic rickets(性聯遺傳型低磷酸鹽佝僂症)	E83.31	149	5
04	Laron syndrome(Laron Dwarfism)(Laron 氏侏儒症候群)	E34.3	5	0
05	Bardet-Biedl syndrome(Bardet-Biedl 氏症候群)	Q87.89	48	5
06	Alsrtom Syndrome(Alstrom 氏症候群)	Q87.89	28	6
07	Persistent hyperinsulinemic hypoglycemia of infancy (PHHI)(持續性幼兒型胰島素過度分泌低血糖症)	E16.1	80	7
08	Wolfram syndrome, DIDMOAD(Wolfram 氏症候群)	E88.9	13	6
09	McCune Albright syndrome(McCune Albright 氏症候群)	Q78.1	26	1
10	Campomelic dysplasia with autosomal sex reversal(短指發育不良及性別顛倒)	Q99.8	1	1
11	ACTH resistance(腎上腺皮促素抗性)	E27.49	1	0
12	25-Hydroxyvitamin D 1-alpha-hydroxylase deficiency(第一型遺傳性維生素 D 依賴型佝僂症)	E83.32	17	0
13	Congenital adrenal hypoplasia(先天性腎上腺發育不全)	Q89.1	26	0
14	Kallmann syndrome(Kallmann 氏症候群)	E23.0	60	1
15	Permanent neonatal diabetes mellitus(永久性新生兒糖尿病)	P70.2	1	0
16	MIRAGE syndrome(MIRAGE 症候群)	Q89.8	2	1
M 先天畸形/症候群 Congenital malformations/syndromes				
01	Aarskog-Scott syndrome(Aarskog-Scott 氏症候群)	Q87.1	12	1
02	Waardenburg syndrome (瓦登伯格氏症候群)	E70.8	92	3
03	Apert syndrome(愛伯特氏症)	Q87.0	33	2
04	Smith-Lemli-Opitz syndrome(Smith-Lemli-Opitz 氏症候群)	E78.72	5	3
05	Larsen syndrome(Larsen 氏症候群(顎裂-先天性脫位症候群))	Q74.8	3	1
06	Beckwith Wiedemann syndrome(Beckwith Wiedemann 氏症候群)	Q87.3	83	3
07	Fraser syndrome(Fraser 氏症候群)	Q87.0	1	0
08	Multiple pterygium syndrome(多發性翼狀膜症候群)	Q79.8	10	2
09	Cornelia de Lange syndrome(Cornelia de Lange 氏症候群)	Q87.1	78	9
10	Hallerman-Streiff syndrome(海勒曼-史德萊夫氏症候群)	Q87.0	6	1
11	Kabuki syndrome(Kabuki 症候群)	Q89.8	86	2
12	Oto-Palato-Digital syndrome(耳-齶-指(趾)症候群)	Q87.0	4	0
13	Conradi-Hunermann syndrome(Conradi-Hunermann 氏症候群)	Q77.3	2	0
14	Treacher Collins syndrome(Treacher Collins 氏症候群)	Q75.4	28	0
15	Robinow syndrome(Robinow 氏症候群)	Q87.1	4	1

分類序號 No.	疾病名稱 The name of each disorder or disease	ICD10 碼 ICD-10-CM	個案數 The Number of Cases	死亡數 The Number of Deaths
16	Nail-Patella syndrome(指(趾)甲髕骨症候群)	Q87.2	6	0
17	Cardiofaciocutaneous syndrome(CFC 症候群)	Q87.89	20	0
18	Peters-Plus syndrome(Peters-Plus 症候群)	Q13.4	2	0
19	Nager Syndrome(Nager 症候群)	Q75.4	3	0
20	CHARGE syndrome(CHARGE 症候群)	Q89.8	35	2
21	White-Sutton syndrome(懷特-薩頓症候群)	Q99.8 F84.8 F78	1	0
22	Costello syndrome(克斯提洛氏彈性蛋白缺陷症)	Q87.89	8	1
23	Ayme-Gripp syndrome(Ayme-Gripp 症候群)	Q87.89	2	0
24	Coffin-Lowry syndrome(Coffin-Lowry 症候群)	Q89.8	3	1
25	Myhre syndrome(Myhre 症候群)	Q87.89	4	0
26	Sensenbrenner syndrome(森森布倫納症候群)	Q87.5	1	0
27	Keppen-Lubinsky syndrome(克片-魯賓斯基症候群)	E88.1	1	0
28	Angelman syndrome(Angelman 氏症候群)	Q93.5	105	5
29	DiGeorge syndrome(DiGeorge 症候群)	D82.1	250	22
30	Prader-Willi syndrome(Prader-Willi 氏症候群)	Q87.1	347	35
31	WAGR syndrome (Wilms' tumor-aniridia-genitourinary anomalies-mental retardation)(威爾姆氏腫瘤、無虹膜、性器異常、智能障礙症候群(WAGR 症候群))	Q87.89	12	2
32	Miller Dieker syndrome(Miller Dieker 症候群)	Q93.88	24	8
33	Rubinstein-Taybi syndrome(Rubinstein-Taybi 氏症候群)	Q87.2	52	0
34	Williams syndrome(威廉斯氏症候群)	Q93.89	316	9
35	Branchio-Oto-Renal syndrome (BOR syndrome) (Branchio-Oto-Renal 症候群(BOR 症候群))	Q87.89	9	0
36	Proteus syndrome(普洛提斯症候群)	Q87.1	2	0
37	Cockayne syndrome(Cockayne 氏症候群)	Q87.1	37	21
38	Hutchinson Gilford progeria syndrome(早老症)	E34.8	13	5
39	Schaaf-Yang syndrome(Schaaf-Yang 症候群)	Q87.1	1	0
N 眼睛異常 Eye disorders				
01	Stargardt's disease(Stargardt's 氏症)	H35.50	10	0
02	Occult macular dystrophy；OMD(隱匿性黃斑部失養症)	H35.50	2	0
03	Leber congenital amaurosis(萊伯氏先天性黑矇症)	H35.50	7	0
Z 其他未分類或不明原因 Unclassified or unknown				
	總計		**21,221**	**4,252**

註：1.個案數係指自 89 年 2 月公布施行罕見疾病防治及藥物法後，該罕見疾病累計所通報之個案數，無扣除死亡者。

2.本表僅包含我國籍個案。

Note: Number of cases was promulgated and enforced of the Regulations for Rare Disease Control and Orphan Drug Act from February 2000 onwards. The cumulative number of reported cases of Rare Diseases were not deducted from the number of deaths.