

衛生福利部國民健康署

108 年 7 月罕見疾病通報個案統計表

統計期間:個案數至 108 年 7 月 31 日 / 死亡數至 108 年 7 月 31 日

製表時間: 108 年 8 月 15 日

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
A 先天性代謝異常				
◎A1 尿素循環代謝異常 Urea cycle disorders (高血氨症)				
01	Congenital urea cycle disorders(先天性尿素循環代謝障礙)	E72.20	75	26
02	Citrullinemia(瓜胺酸血症)	E72.23	84	2
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(鳥胺酸氮甲醯基轉移酶缺乏症)	E72.4	22	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	2	0
◎ A2 胺基酸/有機酸代謝異常 Amino acid metabolic disorders / Organic acidemias				
01	Amino acid metabolic disorders(Aminoacidopathies)(胺基酸代謝疾病)	E72.8	11	3
02	Homocystinuria(高胱胺酸血症)	E72.11	33	1
03	Hypermethioninemia(高甲硫胺酸血症)	E72.19	29	1
04	Nonketotic hyperglycinemia(非酮性高甘胺酸血症)	E72.51	27	15
05	Phenylketouria(苯酮尿症)	E70.0	281	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	39	10
09	Organic acidemias(有機酸血症)	E71.118	22	1
10	Isovaleric academia(異戊酸血症)	E71.110	13	2
11	Glutaric aciduria type I、II(戊二酸尿症,第一型、第二型)	E72.3 E71.313	84	13
12	Propionic academia(丙酸血症)	E71.121	17	4
13	Methylmalonic acidemia(甲基丙二酸血症)	E71.120	62	12
14	3-Hydroxy-3-methyl-glutaric acidemia(3-氫基-3-甲基戊二酸血症)	E71.118	6	0
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(高離氨基酸血症)	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	E71.19	60	0

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
	輔酶 A 羧化酵素缺乏症)			
19	Multiple carboxylase deficiency(多發性羧化酶缺乏症)	D81.819	7	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	47	15
22	Tyrosine hydroxylase deficiency(酪胺酸羧化酶缺乏症)	E70.20	11	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	1	0
◎ A3 脂質儲積				
01	Gaucher's disease(高雪氏症)	E75.22	40	6
02	GM1/GM2 gangliosidosis(GM1/GM2 神經節苷脂儲積症)	E75.19 E75.00	6	6
03	Fabry disease(Fabry 氏症)	E75.21	361	27
04	Niemann-Pick disease(Niemann-Pick 氏症，鞘髓磷脂儲積症)	E75.240 E75.241 E75.242 E75.243 E75.248 E75.249	30	12
05	Metachromatic Leukodystrophy (MLD) (MLD 症候群)	E75.25	20	11
06	Globoid Cell Leukodystrophy (Krabbe's disease)(球細胞腦白 質失養症)	E75.23	1	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
◎ A4 碳水化合物代謝異常				
01	Galactosemia(半乳糖血症)	E74.21 E74.09 E74.01	25	0
02	Glycogen storage disease(肝醣儲積症)	E74.02 E74.03 E74.09 E74.04	226	45
03	Glut (Glucose Transport) 1 deficiency syndrome(腦血管屏障 葡萄糖輸送缺陷)	E74.8	6	0
◎ A5 脂肪酸氧化異常				
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	41	3

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
02	Carnitine deficiency syndrome, primary(原發性肉鹼缺乏症)	E71.41	163	7
03	Medium-chain acyl-coenzyme A dehydrogenase deficiency (MCAD) (中鏈脂肪酸去氫酵素缺乏症)	E71.311	8	0
04	Short-chain acyl-CoA dehydrogenase deficiency(短鏈脂肪酸去氫酶缺乏症)	E71.312	7	1
◎ A6 粒線體代謝異常				
01	Mitochondrial defect(粒線體缺陷)	E88.40	325	66
		H49.811		
02	Kearns-Sayre syndrome(Kearns-Sayre 氏症候群)	H49.812 H49.813 H49.819	4	2
03	Leigh disease(Leigh 氏童年期腦脊髓病變)	G31.82	26	11
04	MELAS(MELAS 症候群)	E88.41	97	26
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	4	1
07	Barth Syndrome(巴氏症候群)	E78.71	3	0
◎ A7 溶小體代謝異常				
01	Cystinosis(胱胺酸血症)	E72.04	5	0
02	Mucopolysaccharidoses(黏多醣症)	E76.3	183	75
03	Fucosidosis(岩藻糖代謝異常 (儲積症))	E77.1	2	1
04	Sialidosis(涎酸酵素缺乏症)	E77.1	31	7
		E77.1		
05	Mucopolipidosis(黏脂質症)	E77.0	10	0
		E75.11		
06	Neuronal ceroid lipofuscinosis(神經元蠟樣脂褐質儲積症)	E75.4	5	4
07	Multiple sulfatase deficiency(多發性硫酸脂酶缺乏症)	E75.29	2	0
◎ A8 膽固醇及脂質代謝異常 Cholesterol and Lipid metabolism				
01	Homozygous familial hypercholesterolemia(同合子家族性高膽固醇血症)	E78.0	53	2
02	Familial Hyperchylomicronemia(家族性高乳糜微粒血症)	E78.3	14	0
03	Sitosterolemia(豆固醇血症 〈植物性〉)	E78.0	5	0
◎ A9 礦物離子缺陷				
01	Wilson's disease(威爾森氏症)	E83.01	620	47
02	Menkes syndrome(Menkes 症候群)	E83.09	14	8
03	Molybdenum cofactor deficiency(鉬輔酶缺乏症)	E61.5	3	1
◎ A10 過氧化體代謝異常				
01	Zellweger syndrome(Zellweger 氏症候群)	E71.510	4	2
02	Adrenoleukodystrophy(腎上腺腦白質失養症)	E71.511 E71.520	68	26

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
		E71.521		
		E71.528		
		E71.529		
03	Rhizomelic Chondrodysplasia Punctata(肢近端型點狀軟骨發育不良)	E71.540	1	0
◎A11 其他代謝異常				
		E80.20		
01	Porphyria(紫質症)	E80.21	111	5
		E80.29		
02	Lesch-Nyhan syndrome(Lesch-Nyhan 氏症候群)	E79.1	14	3
03	Sulfite oxidase deficiency(亞硫酸鹽氧化酶缺乏)	E72.19	11	5
04	Carbohydrate-deficiency glycoprotein syndrome(碳水化合物缺乏醣蛋白症候群)	E77.8	11	2
05	Trimethylaminuria(三甲基胺尿症)	E72.52	11	0
06	Congenital generalized lipodystrophy(先天性全身脂質營養不良症)	E88.1	29	11
07	Cerebrotendinous Xanthomatosis(腦腱性黃瘤症)	E75.5	6	2
08	Hypophosphatasia(低磷酸酯酶症)	E83.39	4	1
		E83.31		
09	Beta-Ketothiolase Deficiency(Beta 硫解酶缺乏症)	E71.19	0	0
10	Biotinidase Deficiency(生物素酶缺乏症)	D81.810	6	0
B 腦部或神經系統病變				
01	Multiple sclerosis(多發性硬化症)	G35	1,864	150
02	Amyotrophic lateral sclerosis (ALS) (肌萎縮性側索硬化症)	G12.21	1419	810
03	Ataxia telangiectasia(共濟失調微血管擴張症候群)	G11.3	12	4
04	Huntington disease(又稱 Huntington's chorea) (亨丁頓氏舞蹈症)	G10	376	103
05	Rett syndrome(瑞特氏症候群)	F84.2	151	5
06	Spinal muscular atrophy(脊髓性肌肉萎縮症)	G12.9	477	79
07	Spinocerebellar ataxia (脊髓小腦退化性動作協調障礙)	G11.1	1,301	339
08	Tuberous sclerosis(結節性硬化症)	Q85.1	646	29
09	Congenital insensitivity to pain with anhidrosis (CIPA) (先天性痛不敏感症合併無汗症)	L74.4	10	2
10	Neurofibromatosis type II(神經纖維瘤症候群第二型)	Q85.02	80	15
11	Alexander disease (Alexander 氏病)	E75.29	5	1
12	Stiffperson syndrome(僵體症候群)	G25.82	17	3
13	Hereditary spastic paraplegia(遺傳性痙攣性下身麻痺)	G11.4	125	3
14	Joubert syndrome(Joubert 氏症候群(家族性小腦蚓部發育不全))	Q04.3	10	1
15	Pelizaeus-Merzbacher Disease(Pelizaeus-Merzbacher 氏症(慢性兒童型腦硬化症))	E75.29	17	0

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
16	Charcot Marie Tooth Disease(Charcot Maire Tooth 氏症(進行性神經性腓骨萎縮症))	G60.0	372	10
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	88	7
18	Familial Amyloidotic Polyneuropathy(家族性澱粉樣多發性神經病變)	E85.1	136	54
19	Moebius syndrome(Moebius 症候群)	Q87.0	19	7
20	Mcleod syndrome(Mcleod 症候群)	J43.0	2	1
21	Aicardi-Goutieres syndrome(Aicardi-Goutieres 症候群)	G31.89	1	0
22	Proteus Syndrome(普洛提斯症候群)	Q87.3	2	0
23	Methyl CpG binding protein 2 Duplication Syndrome (MECP2 Duplication Syndrome) (MECP2 綜合症候群)	Q99.8	6	0
24	Cerebro-Costo-Mandibular Syndrome(腦肋小頷症候群)	Q87.89	3	0
25	Dravet Syndrome (DS)(Dravet 症候群)	G40.311	65	1
26	Vanishing White Matter Disease(腦白質消失症)	G37.8	1	1

C 呼吸循環系統病變

01	Idiopathic Infantile Arterial Calcification(特發性嬰兒動脈硬化症)	Q28.8	2	1
02	Cystic fibrosis (囊狀纖維化症)	E84.9	20	5
03	Primary Pulmonary Hypertension (PPH)(原發性肺動脈高壓)	I27.0	497	144
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	3	0
06	Hereditary Hemorrhagic Telangiectasia(遺傳性出血性血管擴張症)	I78.0	27	7
07	Asphyxiating thoracic dystrophy(窒息性胸腔失養症)	Q77.2	0	0
08	Congenital Central Hypoventilation Syndrome(先天性中樞性換氣不足症候群)	G47.35	10	0

D 消化系統病變

01	Progressive intrahepatic cholestasis (PFIC) (進行性家族性肝內膽汁滯留症)	K83.1	19	5
02	Inborn errors of bile acid synthesis(先天性膽酸合成障礙)	E78.70	3	0
03	α 1- Antitrypsin deficiency(α 1-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	15	0

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
E 腎臟泌尿系統病變				
01	Lowe syndrome(Lowe 氏症候群)	E72.03	20	2
02	Bartter's syndrome(Bartter 氏症候群)	E26.81	81	0
03	Autosomal recessive polycystic kidney disease(體染色體隱性多囊性腎臟疾病)	Q61.19	6	1
F 皮膚病變				
01	Hereditary epidermolysis bullosa (遺傳性表皮分解性水泡症)	Q81.9	100	7
02	Lchthyosis, lamellar recessive(層狀魚鱗癬 (自體隱性遺傳型))	Q80.2	21	2
03	Collodion baby(膠膜兒)	Q80.2	1	0
04	Harlequin ichthyosis(斑色魚鱗癬)	Q80.4	2	1
05	Bullous Congenital ichthyosiform erythroderma (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	26	1
06	Ectodermal Dysplasias(外胚層增生不良症)	Q82.4	92	0
07	Meleda disease(Meleda 島病)	Q82.8	10	1
08	Darier's disease(Darier 氏症 (毛囊角化病))	Q82.8	38	2
09	Dyskeratosis Congenita(先天性角化不全症)	Q82.8	2	0
10	Diffuse Non-epidermolytic Palmoplantar Keratoderma type Unna-Thost(皮膚過度角化症雅司病)	Q82.8	10	0
11	Incontinentia Pigmenti(色素失調症)	Q82.3	55	0
12	Netherton Syndrome(Netherton 症候群)	Q80.3	6	1
G 肌肉病變				
01	Duchenne muscular dystrophy(裘馨氏肌肉失養症)	G71.0	374	90
02	Nemaline Rod Myopathy(Nemaline 線狀肌肉病變)	G71.2	29	2
03	Schwartz Jampel syndrome(Schwartz Jampel 氏症候群)	G71.13	3	0
04	Myotonic dystrophy(肌肉強直症)	G71.11	209	52
05	Facioscapulohumeral muscular dystrophy(面肩胛肱肌失養症)	G71.0	138	8
06	Myotubular Myopathy(肌小管病變)	G71.2	17	4
07	Becker Muscular Dystrophy(貝克型肌肉失養症)	G71.0	51	7
08	Freeman-Sheldon syndrome(Freeman-Sheldon 氏症候群)	Q87.0	1	0
09	Limb-girdle muscular dystrophy(肢帶型肌失養症)	G71.0	71	1
10	Congenital Muscular Dystrophy(先天性肌失養症)	G71.0	21	0
11	Central Core Disease(中心軸空肌病)	G71.2	5	0
12	Multiminicore Disease(多微小軸空肌病)	G71.2	1	0
13	Emery-Dreifuss Muscular Dystrophy (EDMD)(Emery-Dreifuss 肌失養症)	G71.0	2	0
14	GNE myopathy(GNE 遠端肌病變)	G71.8	2	0

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
15	Stormorken syndrome(史托摩根症候群)	D69.8	1	0
H 骨及軟骨病變				
01	Achondroplasia(軟骨發育不全症)	Q77.4	402	10
02	Osteogenesis imperfecta(成骨不全症)	Q78.0	358	12
		M88.0		
		M88.1		
		M88.811		
		M88.812		
		M88.819		
		M88.821		
		M88.822		
		M88.829		
		M88.831		
		M88.832		
		M88.839		
		M88.841		
03	Primary Paget disease(原發性變形性骨炎)	M88.842	3	0
		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	Cleidocranial dysplasia(鎖骨顱骨發育異常)	Q74.0	41	1
		M61.10		
		M61.111		
		M61.112		
		M61.119		
		M61.121		
		M61.122		
		M61.129		
		M61.131		
		M61.132		
		M61.139		
		M61.141		
05	Fibrodysplasia Ossificans Progressiva(進行性骨化性肌炎)	M61.142	11	1
		M61.143		
		M61.144		
		M61.145		
		M61.146		
		M61.151		
		M61.152		
		M61.159		
		M61.161		
		M61.162		
		M61.169		
		M61.171		
		M61.172		

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
		M61.173		
		M61.174		
		M61.175		
		M61.176		
		M61.177		
		M61.178		
		M61.179		
		M61.18		
		M61.19		
		Q71.60		
		Q71.61		
		Q71.62		
06	Split-hand/ Split-foot malformation (SHFM) (裂手裂足症)	Q71.63	13	0
		Q72.70		
		Q72.71		
		Q72.72		
		Q72.73		
07	Osteopetrosis(骨質石化症)	Q78.2	22	8
08	Pseudoachondroplastic dysplasia(假性軟骨發育不全)	Q77.8	13	0
09	Multiple Epiphyseal Dysplasia(多發性骨骺發育不全症)	Q78.3	10	0
I 結締組織病變				
01	Ehlers Danlos syndromeIV(先天結締組織異常第四型)	Q79.6	21	0
J 血液疾病				
01	Thalassemia major(重型海洋性貧血)	D56.0	388	18
		D56.1		
02	Thrombasthenia(血小板無力症)	D69.1	30	1
03	Homozygous proetin C deficiency(同基因合子蛋白質 C 缺乏症)	D68.59	6	0
04	Paroxysmal Nocturnal Hemoglobinuria(陣發性夜間血紅素尿症)	D59.5	121	11
05	Atypical Hemolytic Uremic Syndrome(非典型性尿毒溶血症候群)	D59.3	22	4
K 免疫疾病				
01	Chronic primary granulomatous disease(原發性慢性肉芽腫病)	D71	25	3
02	Congenital Hyper IgE syndrome(先天性高免疫球蛋白 E 症候群)	D82.4	14	4
03	Bruton's agammaglobulinemia(布魯頓氏低免疫球蛋白血症)	D80.0	20	2
04	Wiskott- Aldrich Syndrome(Wiskott- Aldrich 氏症候群)	D82.0	22	8
		D81.0		
05	Severe combined immunodeficiency(嚴重複合型免疫缺乏症)	D81.1	20	4
		D81.2		
		D81.9		
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	8	1

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
09	Interferon γ receptor 1 deficiency (γ 干擾素受體 1 缺陷)	D84.8	3	0
10	Hereditary Angioedema (HAE)(遺傳性血管性水腫)	D84.1	9	0

L 內分泌疾病

01	Kenny-Caffey syndrome(Kenny-Caffey 氏症候群)	Q87.1	1	0
02	Pseudohypoparathyroidism(假性副甲狀腺低能症)	E20.1	65	0
03	X-linked hypophosphatemic rickets(性連遺傳型低磷酸鹽佝僂症)	E83.31	128	3
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	37	3
06	Alstrom Syndrome(Alstrom 氏症候群)	Q87.89	19	4
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	74	6
08	Wolfram syndrome(Wolfram 氏症候群)	E88.9	10	5
09	McCune Albright syndrome(McCune Albright 氏症候群)	Q78.1	22	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	16	0
13	Congenital adrenal hypoplasia(先天性腎上腺發育不全)	Q89.1	19	0
14	Kallmann syndrome(Kallmann 氏症候群)	E23.0	52	1
15	Permanent Neonatal Diabetes Mellitus(永久性新生兒糖尿病)	P70.2	1	0

M 先天畸形症候群

01	Aarskog-Scott syndrome(Aarskog-Scott 氏症候群)	Q87.1	11	0
02	Waardenburg syndrome (瓦登伯格氏症候群)	E70.8	78	2
03	Apert syndrome(愛伯特氏症)	Q87.0	31	1
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	2	1
06	Beckwith Wiedemann syndrome(Beckwith Wiedemann 氏症候群)	Q87.3	62	1
07	Crouzon syndrome(Crouzon 氏症候群)	Q75.1	70	4
08	Fraser syndrome(Fraser 氏症候群)	Q87.0	1	0
09	Multiple pterygium syndrome(多發性翼狀膜症候群)	Q79.8	10	2
10	Cornelia de Lange syndrome(Cornelia de Lange 氏症候群)	Q87.1	61	6
11	Hallerman-Streiff Syndrome(海勒曼-史德萊夫氏症候群)	Q87.0	6	1
12	Kabuki syndrome(歌舞伎症候群)	Q89.8	57	2
13	Oto-Palato-Digital syndrome(耳-齶-指 (趾) 症候群)	Q87.0	4	0

分類序號	疾病名稱	ICD10 碼	個案數	死亡數
14	Conradi-Hunermann syndrome(Conradi-Hunermann 氏症候群)	Q77.3	2	0
15	Treacher Collins Syndrome(Treacher Collins 氏症候群)	Q75.4	24	0
16	Robinow Syndrome(Robinow 氏症候群)	Q87.1	5	1
17	Pfeiffer syndrome(Pfeiffer 氏症候群)	Q87.0	7	4
18	Pantothenate Kinase Associated Neurodegeneration (PKAN) (泛酸鹽激酶關聯之神經退化性疾病)	G23.0	13	3
19	Nail-Patella Syndrome(指(趾)甲髕骨症候群)	Q87.2	4	0
20	Cardiofaciocutaneous Syndrome(CFC 症候群)	Q87.89	6	0
21	Peters-Plus syndrome(Peters-Plus 症候群)	Q13.4	2	0
22	Nager Syndrome(Nager 症候群)	Q75.4	2	0
23	CHARGE Syndrome(CHARGE 症候群)	Q89.8	9	0
24	White-Sutton syndrome(懷特-薩頓症候群)	Q99.8 F84.8 F78.0	1	0
25	Costello syndrome(克斯提洛氏彈性蛋白缺陷症)	Q87.89	2	0
N 染色體異常				
01	Angelman syndrome(Angelman 氏症候群)	Q93.5	80	1
02	DiGeorge's syndrome(DiGeorge's 症候群)	D82.1	206	16
03	Prader-Willi syndrome(Prader-Willi 氏症候群)	Q87.1	319	29
04	W A G R 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 (W A G R 症候群))	Q87.89	10	0
05	Miller Dieker syndrome(Miller Dieker 症候群)	Q93.88	19	5
06	Rubinstein-Taybi syndrome(Rubinstein-Taybi 氏症候群)	Q87.2	39	0
07	Williams Syndrome(威廉斯氏症候群)	Q93.89	264	4
08	Von Hippel-Lindau disease(Von Hippel-Lindau 症候群)	Q85.8	21	3
09	Branchio-Oto-Renal Syndrome (BOR Syndrome) (Branchio-Oto-Renal 症候群 (BOR 症候群))	Q87.89	6	0
Z 其他未分類或不明原因				
01	Cockayne syndrome(Cockayne 氏症候群)	Q87.1	29	21
02	Hutchinson Gilford progeria syndrome(早老症)	E34.8	7	5
03	Tricho-hepato-enteric syndrome(髮-肝-腸症候群)	Q87.9	2	1
04	Stargardt's disease(Stargardt's 氏症)	H35.50	0	0
05	Occult Macular Dystrophy ; OMD(隱匿性黃斑部失養症)	H35.50	2	0
總計			16,295	2,740

註：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。