二百零五項罕見疾病 ICD-9-CM 診斷代碼

修正案總說明

現行公告罕見疾病共二百零七項，其中二百零五項疾病代碼係依 ICD-9-CM 編碼；因應衛生福利部中央健康保險署公告全民健康保險特約醫院自一百零五年一月一日起，門、住診診斷及處置代碼全面單軌申報二零一四年版 ICD-10-CM/PCS。為周延罕見疾病之 ICD-10-CM 疾病編碼，爰擬具罕見疾病 ICD-9-CM 診斷代碼修正案，並自一百零五年一月一日生效。

205 項罕見疾病 ICD-9-CM 診斷代碼修正案對照表

分類	序號	中文病名（僅供參考）	英文病名(縮寫)	修正名稱	現行名稱
				ICD-10-CM 診斷代碼	ICD-9-CM 診斷代碼
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	Congenit al Urea cycle disorders	E72.20	270.6
	02	瓜胺酸血症	Citrullinemia	E72.23	270.6
	03	乙醯穀胺酸合成酶缺乏症	Nitroacetylglutamate synthetase deficiency,NAG synthetase deficiency	E72.29	270.6
	04	鳥胺酸氨甲醯基轉移酶缺乏症	Ornithine transcarbamylase deficiency	E72.4	270.6
	05	高鳥胺酸血症-高氨血症-高瓜胺酸血症症候群	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	E72.4	270.6
◎ 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)	E72.8	270.9
	02	高胱胺酸血症	Homocystinuria	E72.11	270.4
	03	高甲硫胺酸血症	Hypermethioninemia	E72.19	270.4
	04	非酮性高甘胺酸血症	Nonketotic hyperglycinemia	E72.51	270.7
	05	苯酮尿症	Phenylketouria	E70.0	270.1
	06	四氫基喋呤缺乏症	Tetrahydrobiopterin deficiency	E70.1	270.1
	07	遺傳性高酪胺酸血症	Hereditary tyrosinemia	E70.21	270.2
	08	楓糖尿症	Maple syrup urine disease	E71.0	270.3
	09	有機酸血症	Organic acidemias	E71.118	270.9
	10	異戊酸血症	Isovaleric academia	E71.110	270.3

	11	戊二酸尿症，第一型、第二型	Glutaric aciduria type Ⅰ、Ⅱ	type Ⅰ:E72.3 、type11:E71.313	270.9
	12	丙酸血症	Propionic academia	E71.121	270.3
	13	甲基丙二酸血症	Methylmalonic acidemia	E71.120	270.3
	14	3-氫基-3-甲基戊二酸血症	3-Hydroxy-3-methyl-glutaric acidemia	E71.118	270.9
	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	E74.31+E70.0	271.3+270.1
	16	高離氨基酸血症	Hyperlysinemia	E72.3	270.7
	17	組胺酸血症	Histidinemia	E70.41	270.5
	18	三甲基巴豆醯輔酶A羧化酵素缺乏症	3-Methylcrotonyl-CoA carboxylase deficiency	E71.19	270.9
	19	多發性羧化酶缺乏症	Multiple carboxylase deficiency	D81.819	270.9
	20	高脯胺酸血症	Hyperprolinemia	E72.59	270.8
	21	芳香族 L-胺基酸類脫羧基酶缺乏症	Aromatic L-amino acid decarboxylase deficiency	E70.9	270.2
	22	酪胺酸羥化酶缺乏症	Tyrosine hydroxylase deficiency	E70.20	270.2
◎ A3 脂質儲積					
A3	01	高雪氏症	Gaucher’s disease	E75.22	272.7
	02	GM1/GM2 神經節苷脂儲積症	GM1/GM2 gangliosidosis	GM1:E75.19 GM2:E75.00	330.1
	03	Fabry 氏症	Fabry disease	E75.21	272.7
	04	Niemann-Pick 氏症，鞘髓磷脂儲積症	Niemann-Pick disease	E75.240:Type A E75.241:Type B E75.242:Type C E75.243:Type D E75.248:other E75.249:unspecified	272.7
	05	MLD 症候群	Metachromatic Leukodystrophy (MLD)	E75.25	330.0
	06	球細胞腦白質失養症	Globoid Cell Leukodystrophy (Krabbe’s disease)	E75.23	330.0
◎A4 碳水化合物代謝異常					
A4	01	半乳糖血症	Galactosemia	E74.21	271.1

	02	肝醣儲積症	Glycogen storage disease	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	271.0
	03	腦血管屏障葡萄糖輸送缺陷	Glut（Glucose Transport）1 deficiency syndrome	E74.8	271.8
◎ A5 脂肪酸氧化異常					
	01	脂肪酸氧化作用缺陷	Fatty acid oxidation defect	E71.30 E71.310 E71.311 E71.312 E71.313 E71.314 E71.318 E71.32 E71.39	277.8
	02	原發性肉鹼缺乏症	Carnitine deficiency syndrome, primary	E71.41	272.9
	03	中鏈脂肪酸去氫酵素缺乏症	Medium-chain acyl-coenzyme A dehydrogenase deficiency（MCAD）	E71.311	277.8
	04	短鏈脂肪酸去氫酶缺乏症	Short-chain acyl-CoA dehydrogenase deficiency	E71.312	277.8
◎ A6 粒線體代謝異常					
A6	01	粒線體缺陷	Mitochondrial defect	E88.40	277.9
	02	Kearns-Sayre 氏症候群	Kearns-Sayre syndrome	H49.811 H49.812 H49.813 H49.819	277.8
	03	Leigh 氏童年期腦脊髓病變	Leigh disease	G31.82	330.8
	04	MELAS 症候群	MELAS	E88.41	758.89
	05	MNGIE 症候群粒線體性神經胃腸腦病變症候群	Mitochondrial Neurogastrointestinal Encephalopathy Syndrome	E88.89	277.9
	06	丙酮酸鹽脫氫酶缺乏症	Pyruvate dehydrogenase deficiency	E74.4	271.8
	07	巴氏症候群	Barth Syndrome	E78.71	759.89
◎ A7 溶小體代謝異常					
A7	01	胱胺酸血症	Cystinosis	E72.04	270.0

	02	黏多醣症	Mucopolysaccharidoses	E76.3	277.5
	03	岩藻糖代謝異常（儲積症）	Fucosidosis	E77.1	271.8
	04	涎酸酵素缺乏症	Sialidosis	E77.1	272.7
	05	黏脂質症	Mucolipidosis	type I:E77.1 type II、III:E77.0 type IV:E75.11	272.7
	06	神經元蠟樣脂褐質儲積症	Neuronal ceroid lipofuscinosis	E75.4	330.1
◎ 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	E78.0	272.0
	02	家族性高乳糜微粒血症	Familial Hyperchylomicronemia	E78.3	272.3
	03	豆固醇血症 〈植物性〉	Sitosterolemia	E78.0	272.0
◎A9 礦物離子缺陷					
A9	01	威爾森氏症	Wilson’s disease	E83.01	275.1
	02	Menkes 症候群	Menkes syndrome	E83.09	759.89
	03	鉬輔酶缺乏症	Molybdenum cofactor deficiency	E61.5	277.8
◎ A10 過氧化體代謝異常					
A10	01	Zellweger 氏症候群	Zellweger syndrome	E71.510	277.9
	02	腎上腺腦白質失養症	Adrenoleukodystrophy	E71.511 E71.520 E71.521 E71.528 E71.529	272.7
	03	肢近端型點狀軟骨發育不良	Rhizomelic Chondrodysplasia Punctata	E71.540	277.8
◎ A11 其他代謝異常					
	01	紫質症	Porphyria	E80.20 E80.21 E80.29	277.1
	02	Lesch-Nyhan 氏症候群	Lesch-Nyhan syndrome	E79.1	277.2
	03	亞硫酸鹽氧化酶缺乏	Sulfite oxidase deficiency	E72.19	270.0
	04	碳水化合缺乏醣蛋白症候群	Carbohydrate-deficiency glycoprotein syndrome	E77.8	277.9

	05	臭魚症	Trimethylaminuria	E72.52	277.8
	06	先天性全身脂質營養不良症	Congenital generalized lipodystrophy	E88.1	272.6
	07	腦腱性黃瘤症	Cerebrotendinous Xanthomatosis	E75.5	272.7
	08	低磷酸酯酶症	Hypophosphatasia	E83.39 E83.31	275.3
	09	Beta 硫解酶缺乏症	Beta-Ketothiolase Deficiency	E71.19	270.3
B 腦部或神經系統病變					
B1	01	多發性硬化症	Multiple sclerosis	G35	340
	02	肌萎縮性側索硬化症	Amyotrophic lateral sclerosis (ALS)	G12.21	335.20
	03	共濟失調微血管擴張症候群	Ataxia telangiectasia	G11.3	334.8
	04	亨丁頓氏舞蹈症	Huntington disease(又稱 Huntington's chorea)	G10	333.4
	05	瑞特氏症候群	Rett syndrome	F84.2	330.8
	06	脊髓性肌肉萎縮症	Spinal muscular atrophy	G12.9	335.10
	07	脊髓小腦退化性動作協調障礙	Spinocerebellar ataxia	G11.1	334.3
	08	結節性硬化症	Tuberous sclerosis	Q85.1	759.5
	09	先天性痛不敏感症合併無汗症	Congenital insensitivity to pain with anhidrosis (CIPA)	L74.4	705.0
	10	神經纖維瘤症候群第二型	Neurofibromatosis type Ⅱ	Q85.02	237.72
	11	Alexander 氏病	Alexander disease	E75.29	331.89
	12	僵體症候群	Stiffperson syndrome	G25.82	333.91
	13	遺傳性痙攣性下身麻痺	Hereditary spastic paraplegia	G11.4	334.1
	14	Joubert 氏症候群（家族性小腦蚓部發育不全）	Joubert syndrome	Q04.3	759.89
	15	Pelizaeus-Merzbacher 氏症（慢性兒童型腦硬化症）	Pelizaeus-Merzbacher Disease	E75.29	330.0
	16	Charcot Maire Tooth 氏症（進行性神經性腓骨萎縮症）	Charcot Marie Tooth Disease	G60.0	356.1

	17	甘迺迪氏症(脊髓延髓性肌肉萎縮症)	Kennedy Disease	G12.20 G12.21 G12.22 G12.29	335.8
	18	家族性澱粉樣多發性神經病變	Familial Amyloidotic Polyneuropathy	E85.1	277.3+357.4
	19	Moebius 症候群	Moebius syndrome	Q87.0	352.6
	20	Mcleod 症候群	Mcleod syndrome	J43.0	758.81
	21	Aicardi-Goutieres 症候群	Aicardi-Goutieres syndrome	G31.89	330.0
	22	普洛提斯症候群	Proteus Syndrome	Q87.3	759.89
	23	MECP2 綜合症候群	Methyl CpG binding protein 2 Duplication Syndrome (MECP2 Duplication Syndrome)	Q99.8	330.8
	24	腦肋小頜症候群	Cerebro-Costo-Mandibular Syndrome	Q87.89	759.89
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	Q28.8	747.89
	02	囊狀纖維化症	Cystic fibrosis	E84.9	277.00
	03	原發性肺動脈高壓	Primary Pulmonary Hypertension (PPH)	I27.0	416.0
	04	Holt-Oram 氏症候群	Holt-Oram Syndrome	Q87.2	759.89
	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	E74.09	359.3+426.89
	06	遺傳性出血性血管擴張症	Hereditary Hemorrhagic Telangiectasia	I78.0	448.0
	07	窒息性胸腔失養症	Asphyxiating thoracic dystrophy	Q77.2	756.4
	08	先天性中樞性換氣不足症候群	Congenital Central Hypoventilation Syndrome	G47.35	327.25
D 消化系統病變					
	01	進行性家族性肝內膽汁滯留症	Progressive intrahepatic cholestasis,PFIC	K83.1	751.69
	02	先天性膽酸合成障礙	Inborn errors of bile acid synthesis	E78.70	277.9
	03	α1-抗胰蛋白酶缺乏症	α1- Antitrypsin deficiency	E88.01	277.6
	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	Q43.8	750.5

	05	阿拉吉歐症候群	Alagille Syndrome	Q44.7	759.89
E	腎臟泌尿系統病變				
E1	01	Lowe 氏症候群	Lowe syndrome	E72.03	270.8
	02	Bartter 氏症候群	Bartter’s syndrome	E26.81	255.1
	03	體染色體隱性多囊性腎臟疾病	Autosomal recessive polycystic kidney disease	Q61.19	753.14
F	皮膚病變				
F1	01	遺傳性表皮分解性水泡症	Hereditary epidermolysis bullosa	Q81.9	Q81.9
	02	層狀魚鱗癬（自體隱性遺傳型）	Ichthyosis, lamellar recessive	Q80.2	Q80.2
	03	膠膜兒	Collodion baby	Q80.2	Q80.2
	04	斑色魚鱗癬	Harlequin ichthyosis	Q80.4	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 erythoderma （epidermolytic hyperkeratosis）	Q80.3	Q80.3
	06	外胚層增生不良症	Ectodermal Dysplasias	Q82.4	Q82.4
	07	Meleda 島病	Meleda disease	Q82.8	Q82.8
	08	Darier 氏症（毛囊角化病）	Darier’s disease	Q82.8	Q82.8
	09	先天性角化不全症	Dyskeratosis Congenita	Q82.8	Q82.8
	10	皮膚過度角化症雅司病	Diffuse Non-epidermolytic Palmoplantar Keratoderma type Unna-Thost	Q82.8	Q82.8
	11	色素失調症	Incontinentia Pigmenti	Q82.3	Q82.3
	12	Netherton 症候群	Netherton Syndrome	Q80.3	Q80.3
G	肌肉病變				
G1	01	裘馨氏肌肉失養症	Duchenne muscular dystrophy	G71.0	G71.0
	02	Nemaline 線狀肌肉病變	Nemaline Rod Myopathy	G71.2	G71.2
	03	Schwartz Jampel 氏症候群	Schwartz Jampel syndrome	G71.13	G71.13
	04	肌肉強直症	Myotonic dystrophy	G71.11	G71.11
	05	面肩胛肱肌失養症	Facioscapulohumeral muscular dystrophy	G71.0	G71.0

	06	肌小管病變	Myotubular Myopathy	G71.2	G71.2
	07	貝克型肌肉失養症	Becker Muscular Dystrophy	G71.0	G71.0
	08	Freeman-Sheldon 氏症候群	Freeman-Sheldon syndrome	Q87.0	Q87.0
	09	肢帶型肌失養症	Limb-girdle muscular dystrophy	G71.0	G71.0
H	骨及軟骨病變				
H1	01	軟骨發育不全症	Achondroplasia	Q77.4	756.4
	02	成骨不全症	Osteogenesis imperfecta	Q78.0	756.51
	03	原發性變形性骨炎	Primary Paget disease	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 M88.852 M88.859 M88.861 M88.862 M88.869 M88.871 M88.872 M88.879 M88.88 M88.89 M88.9	731.0
	04	鎖骨顱骨發育異常	Cleidocraninal dysplasia	Q74.0	755.59
	05	進行性骨化性肌炎	Fibrodysplasia Ossificans Progressiva	M61.10 M61.111 M61.112 M61.119 M61.121 M61.122 M61.129 M61.131	728.11

				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	
	06	裂手裂足症	Split-hand/ Split-foot malformation（SHFM）	Q71.60 Q71.61 Q71.62 Q71.63 Q72.70 Q72.71 Q72.72 Q72.73	hand755.58 foot755.67
	07	骨質石化症	Osteopetrosis	Q78.2	756.52
	08	假性軟骨發育不全	Pseudoachondroplastic dysplasia	Q77.8	756.4
	09	多發性骨骺發育不全症	Multiple Epiphyseal Dysplasia	Q78.3	756.56
I　結締組織病變					
I1	01	先天結締組織異常第四型	Ehlers Danlos syndrome IV	Q79.6	756.83
J　血液疾病					
	01	重型海洋性貧血	Thalassemia major	D56.0 D56.1	282.4
	02	血小板無力症	Thrombasthenia	D69.1	287.1

	03	同基因合子蛋白質C缺乏症	Homozygous proetin C deficiency	D68.59	273.3
	04	陣發性夜間血紅素尿症	Paroxysmal Nocturnal Hemoglobinuria	D59.5	283.2
	05	非典型性尿毒溶血症候群	Atypical Hemolytic Uremic Syndrome	D59.3	283.11
K	免疫疾病				
K1	01	原發性慢性肉芽腫病	Chronic primary granulomatous disease	D71	288.1
	02	先天性高免疫球蛋白E症候群	Congenital Hyper IgE syndrome	D82.4	288.1
	03	布魯頓氏低免疫球蛋白血症	Bruton’s agammaglobulinemia	D80.0	279.04
	04	Wiskott-Aldrich氏症候群	Wiskott- Aldrich Syndrome	D82.0	279.12
	05	嚴重複合型免疫缺乏症	Severe combined immunodeficiency	D81.0 D81.1 D81.2 D81.9	279.2
	06	補體成份8缺乏症	Complement Component 8 deficiency	D84.1	279.8
	07	IPEX 症候群	IPEX Syndrome	E31.0	759.89
	08	高免疫球蛋白M症候群	Hyper-IgM syndrome	D80.5	279.05
	09	γ 干擾素受體1缺陷	Interferon γ receptor 1 deficiency	D84.8	279.4
L	內分泌疾病				
L1	01	Kenny-Caffey氏症候群	Kenny-Caffey syndrome	Q87.1	759.89
	02	假性副甲狀腺低能症	Pseudohypoparathyroidism	E20.1	275.49
	03	性連遺傳型低磷酸鹽佝僂症	X-linked hypophosphatemic rickets	E83.31	275.3
	04	Laron 氏侏儒症候群	Laron syndrome （Laron Dwarfism）	E34.3	259.4
	05	Bardet-Biedl氏症候群	Bardet-Biedl syndrome	Q87.89	759.89
	06	Alstrom氏症候群	Alsrtom Syndrome	Q87.89	759.2
	07	持續性幼兒型胰島素過度分泌低血糖症	Persistent hyperinsulinemic hypoglycemia of infancy (PHHI)	E16.1	251.1
	08	Wolfram氏症候群	Wolfram syndrome，DIDMOAD	E88.9	277.9
	09	McCune Albright氏症候群	McCune Albright syndrome	Q78.1	756.59

	10	短指發育不良及性別顛倒	Campomelic dysplasia with autosomal sex reversal	Q99.8	758.89
	11	腎上腺皮促素抗性	ACTH resistance	E27.49	253.4
	12	1α-羥化酶缺乏症候群	1α-hydroxylase deficiency	E25.0	268.0
	13	先天性腎上腺發育不全	Congenital adrenal hypoplasia	Q89.1	759.1
	14	Kallmann 氏症候群	Kallmann syndrome	E23.0	253.4
M	先天畸形症候群				
M1	01	Aarskog-Scott 氏症候群	Aarskog-Scott syndrome	Q87.1	759.89
	02	瓦登伯格氏症候群	Waardenburg syndrome	E70.8	270.2
	03	愛伯特氏症	Apert syndrome	Q87.0	755.55
	04	Smith-Lemli-Opitz 氏症候群	Smith-Lemli-Opitz syndrome	E78.72	759.89
	05	Larsen 氏症候群（顎裂-先天性脫位症候群）	Larsen syndrome	Q74.8	755.8
	06	Beckwith Wiedemann 氏症候群	Beckwith Wiedemann syndrome	Q87.3	759.89
	07	Crouzon 氏症候群	Crouzon syndrome	Q75.1	756.0
	08	Fraser 氏症候群	Fraser syndrome	Q87.0	759.89
	09	多發性翼狀膜症候群	Multiple pterygium syndrome	Q79.8	759.89
	10	Cornelia de Lange 氏症候群	Cornelia de Lange syndrome	Q87.1	759.89
	11	海勒曼-史德萊夫氏症候群	Hallerman-Streiff Syndrome	Q87.0	756.0
	12	歌舞伎症候群	Kabuki syndrome	Q89.8	759.89
	13	耳-齶-指（趾）症候群	Oto-Palato-Digital syndrome	Q87.0	759.89
	14	Conradi-Hunermann 氏症候群	Conradi-Hunermann syndrome	Q77.3	756.59
	15	Treacher Collins 氏症候群	Treacher Collins Syndrome	Q75.4	756.0
	16	Robinow 氏症候群	Robinow Syndrome	Q87.1	759.89
	17	Pfeiffer 氏症候群	Pfeiffer syndrome	Q87.0	755.55
	18	泛酸鹽激酶關聯之神經退化性疾病	Pantothenate Kinase Associated Neurodegeneration（PKAN）	G23.0	277.9

	19	指（趾）甲髕骨症候群	Nail-Patella Syndrome	Q87.2	756.89
	20	CFC 症候群	Cardiofaciocutaneous Syndrome	Q87.89	759.89
	21	Peters-Plus 症候群	Peters-Plus syndrome	Q13.4	743.44
	22	Nager 症候群	Nager Syndrome	Q75.4	756.0
N	染色體異常				
N1	01	Angelman 氏症候群	Angelman syndrome	Q93.5	759.89
	02	DiGeorge’s 症候群	DiGeorge’s syndrome	D82.1	279.11
	03	Prader-Willi 氏症候群	Prader-Willi syndrome	Q87.1	759.81
	04	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）	Q87.89	759.89
	05	Miller Dieker 症候群	Miller Dieker syndrome	Q93.88	742.2
	06	Rubinstein-Taybi 氏症候群	Rubinstein-Taybi syndrome	Q87.2	759.89
	07	威廉斯氏症候群	Williams Syndrome	Q93.89	759.89
	08	Von Hippel－Lindau 症候群	Von Hippel－Lindau disease	Q85.8	759.6
	09	Branchio-Oto-Renal Syndrome（BOR Syndrome）	Branchio-Oto-Renal 症候群（BOR 症候群）	Q87.89	759.89
Z	其他未分類或不明原因				
Z1	01	Cockayne 氏症候群	Cockayne syndrome	Q87.1	759.89
	02	早老症	Hutchinson Gilford progeria syndrome	E34.8	259.8
	03	髮-肝-腸症候群	Tricho-hepato-enteric syndrome	Q89.7	759.7
	04	Stargardt’s 氏症	Stargardt’s disease	H35.50	362.75