

分類序號	中文病名	英文病名(縮寫)	ICD-9-CM	ICD-10-CM																																												
A.先天性代謝異常																																																
◎A1尿素循環代謝異常 Urea cycle disorders (高血氮症)																																																
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																																												
	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+E70.0																																												
	16	高離胺基酸血症	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	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	◎ 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 unspedified : 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
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◎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 ) 1 deficiency syndrome	271.8	E74.8
◎ 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	E88.89
	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	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
◎ 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
◎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	碳水化合物缺乏醣蛋白症候群	Carbohydrate-deficiency glycoprotein syndrome	277.9	E77.8
	05	臭魚症	Trimethylaminuria	277.8	E72.52
	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
B 腦部或神經系統病變					
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)	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	J43.0
	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

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	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	α1-抗胰蛋白酶缺乏症	α1- Antitrypsin deficiency	277.6	E88.01
	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	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	Barter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	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	鱗狀魚鱗癬（自體隱性遺傳）	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)	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 dystrophy	359.1	G71.0
	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

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 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
	04	鎖骨顛骨發育異常	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 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 proetin 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
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 ,	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
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
	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	1α-羥化酶缺乏症候群	1α-hydroxylase deficiency	268.0	E25.0
	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
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
	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)甲骰骨症候群	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
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 (WA GR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
	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