公告罕見疾病名單暨 ICD-10-CM 編碼一覽表部分規定修正規定

分類	序號	中文病名（僅供參考）	英文病名(縮寫)	ICD-10-CM 診斷代碼
A.先天性代謝異常 Inborn errors of metabolism				
◎A2 胺基酸/有機酸代謝異常 Disorders of amino acid/organic acid metabolism				
A2	01	胺基酸代謝疾病	Amino acid metabolic disorders (Aminoacidopathies)	E72.9 E70.9 E72.10 E72.89 E71.2 E70.89
	21	芳香族 L-胺基酸類脫羧基酶缺乏症	Aromatic L-amino acid decarboxylase deficiency	E70.81
◎A3 溶小體儲積症 Lysosomal storage disorders				
A3	02	GM1/GM2 神經節苷脂儲積症	GM1/GM2 gangliosidosis	GM1: E75.19 GM2: E75.00 E75.09 E75.01 E75.02
	09	黏多醣症	Mucopolysaccharidoses	Type I Hurler's syndrome E76.01 Type I Hurler-Scheie syndrome E76.02 Type I Scheie syndrome E76.03 Type II Hunter syndrome E76.1 Type III Sanfilippo syndrome E76.22 Type IVA　Morquio syndrome E76.210 Type IVB Morquio syndrome E76.211 Type IV Other Morquio syndrome E76.219 Other MPS E76.29 Unspecified MPS E76.3
◎A4 碳水化合物代謝異常 Disorders of carbohydrate metabolism				
A4	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
◎A11 其他代謝異常 Other metabolic disorders				
A11	08	大腦肌酸缺乏症	Cerebral creatine deficiency	E72.89
	10	嘌呤合成代謝異常	Disorders of purine biosynthesis metabolism	E79.8 E79.9
B.腦部或神經系統異常 Disorders of the brain or nervous system				

B1	07	脊髓小腦退化性動作協調障礙	Spinocerebellar ataxia	G11.10 G11.11 G11.19 G11.2 G11.8 G11.9
	11	Alexander 氏病	Alexander disease	G31.89
	21	Aicardi-Goutieres 症候群	Aicardi-Goutieres syndrome	E79.8
	24	腦白質消失症	Vanishing white matter disease	G11.8
	29	嬰兒型上行性遺傳性痙攣性麻痺	Infantile-onset ascending hereditary spastic paralysis, IAHSP	G12.20 G12.24 G12.29
	31	Von Hippel-Lindau 症候群	Von Hippel-Lindau disease	Q85.83
	32	Basilicata-Akhtar 症候群	Basilicata-Akhtar syndrome	F78.A9 F84.8
C.呼吸循環系統異常 Disorders of the respiratory/circulation system				
C1	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	G72.3
D.消化系統異常 Disorders of the digestive system				
D1	06	髮-肝-腸症候群	Tricho-hepato-enteric syndrome	Q89.7 K52.89
G.肌肉系統異常 Disorders of the muscular system				
G1	10	先天性肌失養症	Congenital muscular dystrophy	G71.20
	13	Emery-Dreifuss 肌失養症	Emery-Dreifuss muscular dystrophy (EDMD)	G71.00 G71.038 G71.09
	14	GNE 遠端肌病變	GNE myopathy	G71.8 G71.9
H.骨及軟骨異常 Disorders of bone and cartilage				
H1	09	多發性骨骺發育不全症	Multiple epiphyseal dysplasia	Q77.8
J.血液系統異常 Disorders of the hematologic system				
J1	05	先天性血栓性血小板低下紫斑症	Congenital thrombotic thrombocytopenic purpura	D69.42
L.內分泌系統異常 Disorders of the endocrine system				
L1	01	Kenny-Caffey 氏症候群	Kenny-Caffey syndrome	Q87.19
	08	Wolfram 氏症候群	Wolfram syndrome, DIDMOAD	E34.8

	10	短指發育不良及性別顛倒	Campomelic dysplasia with autosomal sex reversal	Q87.19
M.先天畸形/症候群 Congenital malformations/syndromes				
M1	15	Robinow 氏症候群	Robinow syndrome	Q87.19
	21	懷特-薩頓症候群	White-Sutton syndrome	Q87.0
	37	Cockayne 氏症候群(柯凱因氏症候群)	Cockayne syndrome	Q87.19