公告罕見疾病名單暨 ICD-10-CM 編碼一覽表部分規定修正草案對照表

修正規定					現行規定					說明
分類	序號	中文病名（僅供參考）	英文病名	ICD-10-CM 診斷代碼	分類	序號	中文病名（僅供參考）	英文病名	ICD-10-CM 診斷代碼	
A.先天性代謝異常 Inborn errors of metabolism					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 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)	E72.9 E70.9 E72.10 E72.89 E71.2 E70.89	A2	01	胺基酸代謝疾病	Amino acid metabolic disorders (Aminoacidopathies)	E72.9 E70.9 E72.10 E72.89	修正 ICD-10-CM 編碼
	21	芳香族 L-胺基酸類脫羧基酶缺乏症	Aromatic L-amino acid decarboxylase deficiency	E70.81		21	芳香族 L-胺基酸類脫羧基酶缺乏症	Aromatic L-amino acid decarboxylase deficiency	E70.9	
◎A3 溶小體儲積症 Lysosomal storage disorders					◎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	A3	02	GM1/GM2 神經節苷脂儲積症	GM1/GM2 gangliosidosis	GM1: E75.19 GM2: E75.00	修正 ICD-10-CM 編碼
	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		09	黏多醣症	Mucopolysaccharidoses	Type1: E76.01 E76.02 E76.03 Type2: E76.1 Other: E76.210 E76.211 E76.219 E76.22 E76.29 Unspecified: E76.3	

				Other MPS E76.29 Unspecified MPS E76.3						
◎A4 碳水化合物代謝異常 Disorders of carbohydrate metabolism					◎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	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 E74.01:Von Gierke's	修正 ICD-10-CM 編碼-刪除 E74.01Von Gierke's
◎A11 其他代謝異常 Other metabolic disorders					◎A11 其他代謝異常 Other metabolic disorders					
A11	08	大腦肌酸缺乏症	Cerebral creatine deficiency	E72.89	A11	08	大腦肌酸缺乏症	Cerebral creatine deficiency	E72.8	修正 ICD-10-CM 編碼
	10	嘌呤合成代謝異常	Disorders of purine biosynthesis metabolism	E79.8 E79.9						新增罕病
B.腦部或神經系統異常 Disorders of the brain or nervous system					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	B1	07	脊髓小腦退化性動作協調障礙	Spinocerebellar ataxia	G11.9	修正 ICD-10-CM 編碼
	11	Alexander 氏病	Alexander disease	G31.89		11	Alexander 氏病	Alexander disease	E75.29	
	21	Aicardi-Goutieres 症候群	Aicardi-Goutieres syndrome	E79.8		21	Aicardi-Goutieres 症候群	Aicardi-Goutieres syndrome	G31.89	
	24	腦白質消失症	Vanishing white matter disease	G11.8		24	腦白質消失症	Vanishing white matter disease	G37.8	
	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		29	嬰兒型上行性遺傳性痙攣性麻痺	Infantile-onset ascending hereditary spastic paralysis, IAHSP	G12.2	
	31	Von Hippel-Lindau 症候群	Von Hippel-Lindau disease	Q85.83		31	Von Hippel-Lindau症候群	Von Hippel-Lindau disease	Q85.8	
	32	Basilicata-Akhtar 症候群	Basilicata-Akhtar syndrome	F78.A9 F84.8		32	Basilicata-Akhtar 症候群	Basilicata-Akhtar syndrome	F78.A9	
C.呼吸循環系統異常 Disorders of the respiratory/circulation system					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	Andersen syndrome	G72.3	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	Andersen syndrome	E74.09	修正 ICD-10-CM 編碼

		變)					變)			
D.消化系統異常 Disorders of the digestive system					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	D1	06	髮-肝-腸症候群	Tricho-hepato-enteric syndrome	Q89.7	修正 ICD-10-CM 編碼
G.肌肉系統異常 Disorders of the muscular system					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	G1	10	先天性肌失養症	Congenital muscular dystrophy	G71.09	修正 ICD-10-CM 編碼
	13	Emery-Dreifuss 肌失養症	Emery-Dreifuss muscular dystrophy (EDMD)	G71.00 G71.038 G71.09		13	Emery-Dreifuss 肌失養症	Emery-Dreifuss muscular dystrophy (EDMD)	G71.00 G71.09	
	14	GNE 遠端肌病變	GNE myopathy	G71.8 G71.9		14	GNE 遠端肌病變	GNE myopathy	G71.8	
H.骨及軟骨異常 Disorders of bone and cartilage					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	H1	09	多發性骨骺發育不全症	Multiple epiphyseal dysplasia	Q78.3	修正 ICD-10-CM 編碼
J.血液系統異常 Disorders of the hematologic system					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	J1	05	先天性血栓性血小板低下紫斑症	Congenital thrombotic thrombocytopenic purpura	M31.19	修正 ICD-10-CM 編碼
L.內分泌系統異常 Disorders of the endocrine system					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	L1	01	Kenny-Caffey 氏症候群	Kenny-Caffey syndrome	Q87.1	修正 ICD-10-CM 編碼
	08	Wolfram 氏症候群	Wolfram syndrome, DIDMOAD	E34.8		08	Wolfram 氏症候群	Wolfram syndrome, DIDMOAD	E88.9	
	10	短指發育不良及性別顛倒	Campomelic dysplasia with autosomal sex reversal	Q87.19		10	短指發育不良及性別顛倒	Campomelic dysplasia with autosomal sex reversal	Q99.8	
M.先天畸形/症候群 Congenital malformations/syndromes					M.先天畸形/症候群 Congenital malformations/syndromes					
M1	15	Robinow 氏症候群	Robinow syndrome	Q87.19	M1	15	Robinow 氏症候群	Robinow syndrome	Q87.89	修正 ICD-10-CM 編碼
	21	懷特-薩頓症候群	White-Sutton syndrome	Q87.0		21	懷特-薩頓症候群	White-Sutton syndrome	Q99.8 F84.8 F78	
	37	Cockayne 氏症候群(柯凱因氏症候群)	Cockayne syndrome	Q87.19		37	Cockayne 氏症候群(柯凱因氏症候群)	Cockayne syndrome	Q87.89	