公告「罕見疾病名單暨 ICD-10-CM 編碼一覽表」部分規定

分類	序號	中文病名（僅供參考）	英文病名(縮寫)	ICD-10-CM 編碼
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
◎A3 溶小體儲積症 Lysosomal storage disorders				
A3	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 E75.244:Type A/B E75.248:other E75.249:unspecified
	14	多發性硫酸脂酶缺乏症	Multiple sulfatase deficiency	E75.26
◎A4 碳水化合物代謝異常 Disorders of carbohydrate metabolism				
A4	03	腦血管屏障葡萄糖輸送缺陷	Glut (Glucose transport)1 deficiency syndrome	E74.810
	04	轉醛醇酶缺乏症	Transaldolase deficiency	E74.89
◎A8 膽固醇及脂質代謝異常 Disorders of cholesterol and lipid metabolism				
A8	01	同合子家族性高膽固醇血症	Homozygous familial hypercholesterolemia	E78.01
	03	豆固醇血症(植物性)	Sitosterolemia	E78.00 E78.01
◎A11 其他代謝異常 Other metabolic disorders				
A11	04	先天性醣基化障礙	Congenital disorder of glycosylation, CDG	E77.8
B.腦部或神經系統異常 Disorders of the brain or nervous system				
	07	脊髓小腦退化性動作協調障礙	Spinocerebellar ataxia	G11.9
	17	甘迺迪氏症(脊髓延髓性肌肉萎縮症)	Kennedy disease	G12.20 G12.24 G12.29
	23	Dravet 症候群	Dravet syndrome, DS	G40.833 G40.834

	32	Basilicata-Akhtar症候群	Basilicata-Akhtar syndrome	F78.A9
G.肌肉系統異常 Disorders of the muscular system				
G1	01	裘馨氏肌肉失養症	Duchenne muscular dystrophy	G71.01
	02	Nemaline 線狀肌肉病變	Nemaline rod myopathy	G71.21
	05	面肩胛肱肌失養症	Facioscapulohumeral muscular dystrophy	G71.02
	06	肌小管病變	Myotubular myopathy	G71.220
	07	貝克型肌肉失養症	Becker muscular dystrophy	G71.01
	09	肢帶型肌失養症	Limb-girdle muscular dystrophy	G71.031 G71.032 G71.033 G71.0340 G71.0341 G71.0342 G71.0349 G71.035 G71.038 G71.039
	10	先天性肌失養症	Congenital muscular dystrophy	G71.09
	11	中心軸空肌病	Central core disease	G71.29
	12	多微小軸空肌病	Multiminicore disease	G71.29
	13	Emery-Dreifuss 肌失養症	Emery-Dreifuss muscular dystrophy (EDMD)	G71.00 G71.09
I.結締組織異常 Disorders of the connective tissue				
I1	01	先天結締組織異常第四型	Ehlers Danlos syndrome IV	Q79.63
J.血液系統異常 Disorders of the hematologic system				
	05	先天性血栓性血小板低下紫斑症	Congenital thrombotic thrombocytopenic purpura	M31.19
K.免疫系統異常 Disorders of the immune system				
K1	05	嚴重複合型免疫缺乏症	Severe combined immunodeficiency	D81.0 D81.1 D81.2 D81.31 D81.9
	09	γ 干擾素受體 1 缺陷	Interferon γ receptor 1 deficiency	D84.89
	11	Netherton 症候群	Netherton syndrome	Q80.8
	12	非典型性尿毒溶血症候群	Atypical hemolytic uremic syndrome	D59.32 D59.39
L.內分泌系統異常 Disorders of the endocrine system				

L1	04	Laron 氏侏儒症候群	Laron syndrome (Laron Dwarfism)	E34.321
M.先天畸形/症候群 Congenital malformations/syndromes				
M1	01	Aarskog-Scott 氏症候群	Aarskog-Scott syndrome	Q87.19
	02	瓦登伯格氏症候群	Waardenburg syndrome	Q87.89
	09	Cornelia de Lange 氏症候群	Cornelia de Lange syndrome	Q87.19
	15	Robinow 氏症候群	Robinow syndrome	Q87.89
	28	Angelman 氏症候群	Angelman syndrome	Q93.51
	30	Prader-Willi 氏症候群	Prader-Willi syndrome	Q87.11
	34	威廉斯氏症候群	Williams syndrome	Q93.82
	37	Cockayne 氏症候群(柯凱因氏症候群)	Cockayne syndrome	Q87.89
	39	Schaaf-Yang 症候群	Schaaf-Yang syndrome	Q87.19