財團法人台北病理中心

罕見先天代謝異常疾病種類

代號	胺基酸類	中文名稱	辨識標記
*1	Phenylketonuria	苯酮尿症	PHE，TYR，PHE/TYR
*2	Homocystinuria	高胱胺酸尿症	MET
*3	Maple Syrup Urine Disease	楓糖尿症	LEU/ILE，VAL
4	Tyrosinemia	酪胺酸血症	TYR
5	Citrullinemia	瓜胺酸血症	CIT
6	Argininemia	精胺酸血症	ARG
7	Hyperammonemia，Hyperornithinemia Homocitrullinuria Syndrome	高氨血症	ORN
	脂肪酸類		
8	Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	極長鏈脂肪酸代謝異常	C14，C14.1，C16
9	Long Chain hydroxyl Acyl-CoA Dehydrogenase Deficiency (LCHAD)	長鏈脂肪酸代謝異常	C16OH，C18OH，C18.1OH
*10	Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	中鏈脂肪酸代謝異常	C6，C8，C10，C8/C10
11	Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)	短鏈脂肪酸代謝異常	C4，C6
12	Carnitine/acylcarnitine Tanslocase Deficiency	肉鹼穿透障礙	C16，C18，C18.1
13	Carnitine Palmitoyl Transferase Deficiency Type I (CPT-I)	肉鹼結合酵素缺乏症第一型	C0 (free carnitine higher) C16，C18，C18.1 (lower)
14	Carnitine Palmitoyl Transferase Deficiency Type II (CPT-II)	肉鹼結合酵素缺乏症第二型	C14，C16，C18，C18.1
15	Carnitine Transporter Defect	肉鹼吸收障礙	C0，C2 (lower)
*16	Multiple Acyl-CoA Dehydrogenase Deficiency (GA-I)	戊二酸血症第一型	C5DC

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代號	有機酸類	中文名稱	辨識標記
17	Multiple Acyl-CoA Dehydrogenase Deficiency (GA-II)	戊二酸血症第二型	C4，C5，C8，C8.1，C12，C14，C16
*18	Methylmalonic Acidemia (MMA)	甲基丙二酸血症	C3，C4DC，C3/C2
19	Propionic Acidemia (PA)	丙酸血症	C3，C3/C2
*20	Isovaleric Acidemia (IVA)	異戊酸血症	C5
21	3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	3-甲基巴豆醯輔酶A羧化酶缺乏症	C5OH
22	3-Hydroxy-3-Methylglutaric Aciduria (HMG)	3-羥基-3-甲基戊二酸尿症	C5OH
23	Trifunctional protein deficiency	三功能蛋白缺乏症	C16OH，C18OH，C18.1OH
24	Ethylmalonic Acidemia	乙基丙二酸血症	C4，C5
25	Malonic Acidemia	丙二酸血症	C3DC
26	Isobutyryl CoA dehydrogenase deficiency	Isobutyryl CoA 去氫酶缺乏症	C14.1
	非串聯質譜儀篩檢項目		
*27	Congenital Hypothyroidism (CHT)	先天性甲狀腺功能低下症	TSH
*28	Congenital Adrenal Hyper (CAH)	先天性腎上腺增生症	17-OHP
*29	Gucose-6-Phosphate Dehydrogenase Deficiency (G6PD)	葡萄糖 6 磷酸鹽去氫酶缺乏症	G6PD
*30	Galactosemia	半乳糖血症	Total Galactose

備註：疾病代號前有*者為國健局指定項目.

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