

# 【基因分析費用一覽表】

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※使用說明：

- 1.依疾病名稱之英文字母排列
- 2.以藍色底標示之基因可申請國健補助

疾病名稱	收費碼	檢驗項目	金額	比例	總價
3-甲基巴豆醯輔酶A羧化酵素缺乏症 (3-methylcrotonyl-CoA-carboxylase deficiency, 3MCC)	96188S50	MCCC1 (Self-pay)	12160	80%	15200
	96188S51	MCCC1 Tx. fee (Self-pay)	3040	20%	
	96188S52	MCCC2 (Self-pay)	10880	80%	13600
	96188S53	MCCC2 Tx. fee (Self-pay)	2720	20%	
軟骨發育不全症 (Acondroplasia)	96188SA8	Acondroplasia之FGFR3 hot spot (Self-pay)	1200	80%	1500
	96188SA9	Acondroplasia之FGFR3 hot spot Tx. fee (Self-pay)	300	20%	
急性間歇性紫質症 (Acute intermittent porphyria, AIP)	96188S10	HMBS (Self-pay)	8960	80%	11200
	96188S11	HMBS Tx. fee (Self-pay)	2240	20%	
芳香族L-胺基酸類脫羧酶缺乏症 (Aromatic L-amino acid decarboxylase deficiency, AADC)	96188S98	DDC (Self-pay)	8960	80%	11200
	96188S99	DDC Tx. fee (Self-pay)	2240	20%	
生物素酶缺乏症 (Biotinidase deficiency)	96188S60	BTD (Self-pay)	2560	80%	3200
	96188S61	BTD Tx. fee (Self-pay)	640	20%	
肉鹼棕櫚醯基轉移酶1缺乏症 (Carnitine palmitoyltransferase 1 deficiency)	96188SC4	CPT1A (Self-pay)	12160	80%	15200
	96188SC5	CPT1A Tx. fee (Self-pay)	3040	20%	
瓜胺酸血症第1型 (Citrullinemia type I)	96188S56	ASS1 (Self-pay)	10240	80%	12800
	96188S57	ASS1 Tx. fee (Self-pay)	2560	20%	
瓜胺酸血症第2型 (Citrullinemia type II)	96188S58	SLC25A13 (Self-pay)	11520	80%	14400
	96188S59	SLC25A13 Tx. fee (Self-pay)	2880	20%	
先天性腎上腺增生症 (Congenital adrenal hyperplasia, CAH)	96188SC8	CYP21A2 (Self-pay)	9920	80%	12400
	96188SC9	CYP21A2 Tx. fee (Self-pay)	2480	20%	
Dopa-反應型肌張力不全症 (Dopa-Responsive Dystonia)	96188SB8	SPR (Self-pay)	1920	80%	2400
	96188SB9	SPR Tx. fee (Self-pay)	480	20%	
法布瑞氏症 (Fabry disease)	96188S16	GLA (Self-pay)	4480	80%	5600
	96188S17	GLA Tx. fee (Self-pay)	1120	20%	
	96188S03	Fabry IVS4 point mutation 基因分析 (Self-pay)	1200	80%	1500
	96188S55	Fabry IVS4 point mutation Tx. fee (Self-pay)	300	20%	
家族性脂蛋白脂酶缺乏症 (Familial lipoprotein lipase deficiency)	96188SD6	LPL (Self-pay)	6400	80%	8000
	96188SD7	LPL Tx. fee (Self-pay)	1600	20%	

半乳糖血症第1型 (Galactosemia type I)	96188S66	GALT (Self-pay)	5120	80%	6400
	96188S67	GALT Tx. fee (Self-pay)	1280	20%	
半乳糖血症第2型 (Galactosemia type II)	96188S68	GALK1 (Self-pay)	5120	80%	6400
	96188S69	GALK1 Tx. fee (Self-pay)	1280	20%	
高雪氏症 (Gaucher disease)	96188S28	GBA (Self-pay)	7680	80%	9600
	96188S29	GBA Tx. fee (Self-pay)	1920	20%	
葡萄糖-六-磷酸鹽去氫酵素缺乏症 (Glucose-6-phosphate dehydrogenase deficiency, G6PD)	96188SD4	G6PD (Self-pay)	8320	80%	10400
	96188SD5	G6PD Tx. fee (Self-pay)	2080	20%	
戊二酸血症 第一型 (Glutaric aciduria type I, GA1)	96188S78	GCDH (Self-pay)	7680	80%	9600
	96188S79	GCDH Tx. fee (Self-pay)	1920	20%	
高胱氨酸尿症 (Homocystinuria)	96188S80	CBS (Self-pay)	10880	80%	13600
	96188S81	CBS Tx. fee (Self-pay)	2720	20%	
	96188S82	MTHFR 2 hot spots (Self-pay)	1280	80%	1600
	96188S83	MTHFR 2 hot spots Tx. fee (Self-pay)	320	20%	
甲狀腺機能低下症 (Hypothyroidism)	96188SC2	TG (Self-pay)	30720	80%	38400
	96188SC3	TG Tx. fee (Self-pay)	7680	20%	
異戊酸血症 (Isovaleric acidemia, IVA)	96188S84	IVD (Self-pay)	7040	80%	8800
	96188S85	IVD Tx. fee (Self-pay)	1760	20%	
楓糖尿症 (Maple Syrup Urine Disease, MSUD)	96188S86	DBT (Self-pay)	7040	80%	8800
	96188S87	DBT Tx. fee (Self-pay)	1760	20%	
馬凡氏症 (Marfan syndrome)	96188S22	FBN1 (Self-pay)	26320	80%	32900
	96188S23	FBN1 Tx. fee (Self-pay)	6580	20%	
	96188S24	TGFBR2 (Self-pay)	7200	80%	9000
	96188S25	TGFBR2 Tx. fee (Self-pay)	1800	20%	
中鏈脂肪酸去氫酶缺乏症 (medium-chain acyl-CoA dehydrogenase deficiency)	96188S88	ACADM (Self-pay)	7680	80%	9600
	96188S89	ACADM Tx. fee (Self-pay)	1920	20%	
甲基丙二酸血症 (Methylmalonic acidmia, MMA)	96188S90	MUT (Self-pay)	8320	80%	10400
	96188S91	MUT Tx. fee (Self-pay)	2080	20%	
	96188S92	MMACHC (Self-pay)	2560	80%	3200
	96188S93	MMACHC Tx. fee (Self-pay)	640	20%	

粒線體疾病之粒線體基因點突變分析 (mitochondrial disease)  A3243G、G3460A、A8344G、 T8993G、T8993C、T10158C、 T10191C、C11777A、G11778A、 T12706C、G13513A、G14459A、 T14484C、T14487C	96188SA0	mtDNA單一點突變分析(Self-pay)	640	80%	800
	96188SA1	mtDNA單一點突變分析檢體處理費(Self-pay)	160	20%	
	96188SA2	mtDNA 3個位點組合分析(Self-pay)	1600	80%	2000
	96188SA3	mtDNA 3個位點組合分析檢體處理費(Self-pay)	400	20%	
	96188SA4	mtDNA 5個位點組合分析(Self-pay)	2400	80%	3000
	96188SA5	mtDNA 5個位點組合分析檢體處理費(Self-pay)	600	20%	
粒線體疾病之粒線體基因缺失分析 (mt DNA 4977 bp deletion)	96188SA6	mtDNA 4977 bp deletion (Self-pay)	640	80%	800
	96188SA7	mtDNA 4977 bp deletion Tx. fee (Self-pay)	160	20%	
粒線體疾病 (mitochondrial disease) mtDNA全基因定序	96188SE0	mtDNA (Self-pay)	20000	80%	25000
	96188SE1	mtDNA Tx. fee (Self-pay)	5000	20%	
mtDNA全基因定序大多是找到沒意義的SNP，耗費大量人力物力，與牛醫師討論後決議，目前以做上面所列的14個突變點為主，較有臨床上實際意義，又可申請國健補助。					
黏多醣症第二型 (Mucopolysaccharidosis II)	96188SC6	IDS (Self-pay)	5760	80%	7200
	96188SC7	IDS Tx. fee (Self-pay)	1440	20%	
多發性羧化酶缺乏症 (Multiple carboxylase deficiency)	96188S62	HLCS (Self-pay)	7680	80%	9600
	96188S63	HLCS Tx. fee (Self-pay)	1920	20%	
多發性內分泌瘤病 (Multiple endocrine neoplasia) 陳明晃醫師轉診來的病人會做此基因	96188SD2	MEN1 (Self-pay)	6400	80%	8000
	96188SD3	MEN1 Tx. fee (Self-pay)	1600	20%	
尼曼匹克症 A型 (Niemann-Pick disease type A)	96188S32	SMPD1 (Self-pay)	3840	80%	4800
	96188S33	SMPD1 Tx. fee (Self-pay)	960	20%	
尼曼匹克症C型 (Niemann-Pick disease type C)	96188S34	NPC1 (Self-pay)	12000	80%	15000
	96188S35	NPC1 Tx. fee (Self-pay)	3000	20%	
鳥胺酸氨甲醯基轉移酶缺乏症 (Ornithine transcarbamylase deficiency)	96188S94	OTC (Self-pay)	6400	80%	8000
	96188S95	OTC Tx. fee (Self-pay)	1600	20%	
持續性幼兒型胰島素過度 分泌低血糖症(Persistent hyperinsulinemic hypoglycemia of infancy, PHHD)	96188S64	GLUD1 (Self-pay)	8320	80%	10400
	96188S65	GLUD1 Tx. fee (Self-pay)	2080	20%	
苯酮尿症 (Phenylketouria, PKU)	96188S70	PAH (Self-pay)	8320	80%	10400
	96188S71	PAH Tx. fee (Self-pay)	2080	20%	
	96188S72	PTS (Self-pay)	3840	80%	4800
	96188S73	PTS Tx. fee (Self-pay)	960	20%	
	96188S74	QDPR (Self-pay)	4480	80%	5600
	96188S75	QDPR Tx. fee (Self-pay)	1120	20%	
	96188S76	GCH1 (Self-pay)	3840	80%	4800
	96188S77	GCH1 Tx. fee (Self-pay)	960	20%	

龐貝氏症 (Pompe disease)	96188S18	GAA (Self-pay)	12000	80%	15000
	96188S19	GAA Tx. fee (Self-pay)	3000	20%	
原發性肉鹼缺乏症 (Primary carnitine deficiency syndrome)	96188S26	SLC22A5 (Self-pay)	6400	80%	8000
	96188S27	SLC22A5 Tx. fee (Self-pay)	1600	20%	
豆固醇血症 (Sitosterolemia)	96188S36	ABCG5 (Self-pay)	8320	80%	10400
	96188S37	ABCG5 Tx. fee (Self-pay)	2080	20%	
	96188S38	ABCG8 (Self-pay)	8320	80%	10400
	96188S39	ABCG8 Tx. fee (Self-pay)	2080	20%	
短鏈脂肪酸去氫酶缺乏症 (Short-chain acyl-CoA dehydrogenase deficiency)	96188S20	ACADS (Self-pay)	6400	80%	8000
	96188S21	ACADS Tx. fee (Self-pay)	1600	20%	
甲狀腺過氧化物酶缺乏症 (Thyroid peroxidase deficiency)	96188SC0	TPO (Self-pay)	10880	80%	13600
	96188SC1	TPO Tx. fee (Self-pay)	2720	20%	
臭魚症 (Trimethylaminuria)	96188S40	FMO3 (Self-pay)	5760	80%	7200
	96188S41	FMO3 Tx. fee (Self-pay)	1440	20%	
酪胺酸羥化酶缺乏症 (Tyrosine hydroxylase deficiency)	96188S96	TH (Self-pay)	8960	80%	11200
	96188S97	TH Tx. fee (Self-pay)	2240	20%	
極長鏈脂肪酸去氫酶缺乏症 (Very long-chain acyl-CoA dehydrogenase deficiency, VLCADD)	96188SD8	ACADVL (Self-pay)	12800	80%	16000
	96188SD9	ACADVL Tx. fee (Self-pay)	3200	20%	
逢希伯-林道症候群 (Von Hippel-Lindau syndrome) 陳明晃醫師轉診來的病人會做此基因	96188SD0	VHL (Self-pay)	1920	80%	2400
	96188SD1	VHL Tx. fee (Self-pay)	480	20%	
多項基因外顯子組合基因檢驗 (只做單點，或是家族成員僅做 proband 的突變點時使用)	96188SB0	4 exons 組合 (Self-pay)	2560	80%	3200
	96188SB1	4 exons 組合 Tx. fee (Self-pay)	640	20%	
	96188SB2	3 exons 組合 (Self-pay)	1920	80%	2400
	96188SB3	3 exons 組合 Tx. fee (Self-pay)	480	20%	
	96188SB4	2 exons 組合 (Self-pay)	1280	80%	1600
	96188SB5	2 exons 組合 Tx. fee (Self-pay)	320	20%	
	96188SB6	1 exon (Self-pay)	640	80%	800
	96188SB7	1 exon Tx. fee (Self-pay)	160	20%	