FamilyCODE Gene List
福碼基因列表

System 系統	Diseases 疾病	Gene 基因
77 Inherited Metabolic Diseases 77 種遺傳性代謝病	Medium-chain acyl-CoA dehydrogenase deficiency 中鏈乙酰輔酶 A 脫氫酶缺乏症	ACADM
	Short-chain acyl-CoA dehydrogenase deficiency 短鏈酰基輔酶 A 脫氫酶缺乏症	ACADS
	Very long-chain acyl-CoA dehydrogenase deficiency 極長鏈酰基輔酶 A 脫氫酶缺乏症	ACADVL
	Beta-ketothiolase deficiency β-酮硫解酶缺乏症	ACAT1
	Aspartylglucosaminuria 天冬氨酰葡糖胺尿症	AGA
	Glycogen storage disease type III 糖原貯積病 III 型	AGL
	Sjögren-Larsson syndrome Sjogren-Larsson 綜合症	ALDH3A2
	Hereditary fructose intolerance 遺傳性果糖不耐受症	ALDOB
	Congenital disorder of glycosylation 先天性糖基化障礙	ALG6, DPM1, MOGS, SLC35C1, B4GALT1, SLC35A1, ALG1, DOLK, PMM2, MPI, DPAGT1
	Hypophosphatasia 低磷酸酯酶症	ALPL
	Arginase deficiency 精氨酸酶缺乏症	ARG1
	Metachromatic leukodystrophy 異染性腦白質營養不良	ARSA
	Mucopolysaccharidosis type VI 粘多醣貯積症 VI 型	ARSB
	Argininosuccinate lyase deficiency 精氨琥珀酸裂解酶缺乏症	ASL
	Citrullinemia type I 瓜氨酸血症 I 型	ASS1
	Wilson disease Wilson 病	ATP7B
	Bardet-Biedl syndrome types 1, 2 and 10 巴-比二氏綜合症 1，2 及 10 型	BBS1, BBS10, BBS2
	Maple syrup urine disease types Ia, Ib and II 楓糖尿症 Ia 型、Ib 型和 II 型	BCKDHA, BCKDHB, DBT
	Biotinidase deficiency 生物素酶缺乏症	BTD

	Homocystinuria 高胱氨酸尿症	CBS
	Dihydrolipoamide dehydrogenase deficiency 二氫硫辛酰胺脫氫酶缺乏症	DLD
	Carnitine palmitoyltransferase I deficiency 肉鹼棕櫚酰轉移酶 I 缺乏症	CPT1A
	Carnitine palmitoyltransferase II deficiency 晚發型肉鹼棕櫚酰轉移酶 II 缺乏症	CPT2
	Cystinosis 胱氨酸貯積症	CTNS
	Neuronal ceroid lipofuscinosis 神經元蠟樣質脂褐質沉積病	CTSD, MFSD8, CLN3, CLN5, CLN6, CLN8, PPT1, TPP1
	Cerebrotendinous xanthomatosis 腦腱黃瘤病	CYP27A1
	Smith-Lemli-Opitz syndrome Smith-Lemli-Opitz 綜合症	DHCR7
	Dihydropyrimidine dehydrogenase deficiency 二氫嘧啶脫氫酶缺乏症	DPYD
	Glutaric acidemia type II 戊二酸血症 II 型	ETFA, ETFDH, ETFB
	Ethylmalonic encephalopathy 乙基丙二酸腦病	ETHE1
	Tyrosinemia type I 酪氨酸血症 I 型	FAH
	Glucose-6-phosphate dehydrogenase deficiency 葡萄糖-6-磷酸脫氫酶缺乏症	G6PD
	Glycogen storage disease type II 糖原貯積病 II 型	GAA
	Krabbe disease Krabbe 病	GALC
	Mucopolysaccharidosis type IV, subtype A 粘多醣貯積症 IV A 型	GALNS
	Galactosemia type I 半乳糖血症 I 型	GALT
	Glycogen storage disease type IV 糖原貯積病 IV 型	GBE1
	Glutaric acidemia type I 戊二酸血症 I 型	GCDH
	Mucopolysaccharidosis type VII 粘多醣貯積症 VII 型	GUSB
	Fabry disease Fabry 病	GLA
	GLB1-related disorders GM1 神經節苷脂貯積症及黏多醣貯積症 IVB 型	GLB1
	Glycine encephalopathy 甘氨酸腦病	GLDC, AMT

	Mucolipidosis II alpha/beta and Mucolipidosis III alpha/beta 黏脂質貯積症 II α/II β 型和 III α/III β 型	GNPTAB
	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency 長鏈 3-羥酰基輔酶 A 脫氫酶缺乏病	HADHA
	Hexosaminidase A deficiency, including Tay-Sachs disease Tay-Sachs 病	HEXA
	Sandhoff disease Sandhoff 病	HEXB
	Holocarboxylase synthetase deficiency 羧化全酶合成酶缺乏症	HLCS
	3-hydroxy-3-methylglutaryl-CoA lyase deficiency 3-羥基-3-甲基戊二酰輔酶 A 裂解酶缺乏症	HMGCL
	Mucopolysaccharidosis type II 粘多醣貯積症 II 型	IDS
	Mucopolysaccharidosis type I 粘多醣貯積症 I 型	IDUA
	Isovaleric academia 異戊酸血症	IVD
	Lysosomal acid lipase deficiency 溶酶體酸性脂肪酶缺乏症	LIPA
	Alpha-mannosidosis α-甘露糖苷貯積症	MAN2B1
	Mucolipidosis type IV 黏脂質貯積症 IV 型	MCOLN1
	MLC1-related megalencephalic leukoencephalopathy with subcortical cysts MLC1 相關巨腦性腦白質營養不良伴皮質下囊腫	MLC1
	Isolated methylmalonic academia 單純型甲基丙二酸血症	MMAB, MMAA, MUT
	Methylmalonic acidemia with homocystinuria type cblC 甲基丙二酸血症伴同型半胱氨酸血症 cblC 型	MMACHC
	Abetalipoproteinemia 無 β-脂蛋白血症	MTTP
	Ornithine transcarbamylase deficiency 鳥氨酸氨甲酰基轉移酶缺乏症	OTC
	Phenylalanine hydroxylase deficiency 苯丙氨酸羥化酶缺乏症	PAH
	Pyruvate carboxylase deficiency 丙酮酸羧化酶缺乏症	PC
	Propionic academia 丙酸血症	PCCA, PCCB
	Peroxisome biogenesis disorders, Zellweger syndrome spectrum 過氧化物酶體生物合成障礙及澤爾韋格綜合症譜系	PEX6, PEX1
	Rhizomelic chondrodysplasia punctata type 1 肢近端型點狀軟骨發育不良 I 型	PEX7
	BH4-deficient hyperphenylalaninemia 四氫生物喋呤缺乏症	PTS, QDPR, GCH1
	Glycogen storage disease type V 糖原貯積病 V 型	PYGM

	Mucopolysaccharidosis type III 粘多醣貯積症 III 型	SGSH, NAGLU, HGSNAT, GNS
	Sialic acid storage disease, including Salla disease 唾液酸貯積症（包括 Salla 病）	SLC17A5
	Systemic primary carnitine deficiency 原發性肉鹼缺乏症	SLC22A5
	Citrin deficiency 希特林蛋白缺乏症	SLC25A13
	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome 高鳥氨酸血症-高氨血症-同型瓜氨酸尿症綜合症	SLC25A15
	Glycogen storage disease type I, subtypes Ia and Ib 糖原貯積病 I 型	SLC37A4, G6PC
	Lysinuric protein intolerance 賴氨酸尿性蛋白不耐受症	SLC7A7
	Niemann-Pick disease types A, B, C1 and C2 尼曼-匹克病 A，B，C1 和 C2 型	SMPD1, NPC1, NPC2
	Multiple sulfatase deficiency 多種硫酸酯酶缺乏症	SUMF1
	Tyrosine hydroxylase deficiency 酪氨酸羥化酶缺乏症	TH
	Ataxia with vitamin E deficiency 共濟失調伴維生素 E 缺乏症	TTPA
15 Inherited Blood Immune Diseases 15 遺傳性血液免疫疾病	X-linked adrenoleukodystrophy X 連鎖腎上腺腦白質營養不良	ABCD1
	Adenosine deaminase deficiency 腺苷脫氨酶缺乏症	ADA
	X-linked agammaglobulinemia X 連鎖無丙種球蛋白血症	BTK
	Immunodeficiency with hyper-IgM type 1 高 IgM 免疫缺陷病 I 型	CD40LG
	X-linked chronic granulomatous disease X 連鎖慢性肉芽腫性疾病	CYBB
	Fanconi anemia, complementation group C 範可尼貧血互補群 C 型	FANCC
	Autosomal recessive severe congenital neutropenia 常染色體隱性遺傳重症先天性中性白細胞減少症	G6PC3, HAX1
	Alpha-thalassemia α-地中海貧血	HBA1/HBA2
	Beta-hemoglobinopathies, including beta-thalassemia and sickle cell disease β-血紅蛋白病（包括 β 地中海貧血和鐮刀型細胞貧血症)	HBB
	Chediak-Higashi syndrome Chédiak-Higashi 綜合症	LYST
	Congenital amegakaryocytic thrombocytopenia 先天性純巨核細胞再生障礙性血小板減少症	MPL
	Autosomal recessive chronic granulomatous disease 常染色體隱性遺傳慢性肉芽腫性疾病	NCF1, CYBA, NCF2
	Familial hemophagocytic lymphohistiocytosis 家族性噬血細胞性淋巴組織細胞增多症	PRF1, UNC13D

	X-linked lymphoproliferative syndrome type 1 X 連鎖淋巴增殖綜合徵 I 型	SH2D1A
	WAS-related disorders (including Wiskott-Aldrich syndrome, X-linked congenital neutropenia and X-linked thrombocytopenia) WAS 相關疾病（包括威斯科特—奧爾德里奇綜合徵，X 連鎖先天性嗜中性白血球減少症和 X 連鎖血小板減少症）	WAS
12 Inherited Neurological/ Muscular Disorders 12 種遺傳性神經/肌肉疾病	Ataxia-telangiectasia 共濟失調性毛細血管擴張症	ATM
	Limb-girdle muscular dystrophy type 2, subtypes 2A, 2C, 2D and 2E 肢帶型肌營養不良 2A，2C，2D 和 2E 亞型	CAPN3, SGCG, SGCA, SGCB
	Congenital myasthenic syndrome 先天性肌無力綜合症	CHRNE, COLQ, DOK7, RAPSN, CHAT, CHRNA1
	Leukoencephalopathy with vanishing white matter 白質消融性白質腦病	EIF2B5
	Congenital dystroglycanopathies, including Fukuyama congenital muscular dystrophy, muscle-eye-brain disease and Walker-Warburg syndrome 先天性肌營養不良蛋白聚醣病變(包括 Fukuyana 型先天性肌營養不良, 肌-眼-腦疾病，和 Walker-Warburg 綜合徵）	FKTN, LARGE, POMGNT1, POMT1, POMT2
	Nonaka myopathy Nonaka 肌病	GNE
	L1 syndrome L1 綜合症	L1CAM
	PLA2G6-associated neurodegeneration PLA2G6 相關神經退行性病變（包括嬰兒神經軸索營養不良）	PLA2G6
	POLG-related disorders POLG 相關疾病	POLG
	Autosomal recessive spastic ataxia of Charlevoix-Saguenay 常染色體隱性遺傳 Charlevoix-Saguenay 痙攣性共濟失調	SACS
	Hereditary motor and sensory neuropathy with agenesis of the corpus callosum 遺傳性運動感覺性神經病伴有胼胝體發育不全	SLC12A6
	Spinal muscular atrophy 脊髓性肌萎縮	SMN1
8 Inherited Sensory Organ Disorders 8 種遺傳五官疾病	Usher syndrome type 3A Usher 綜合症 3A 型	CLRN1
	CYP1B1-related glaucoma CYP1B1 相關青光眼	CYP1B1
	GJB2-related nonsyndromic hearing loss type DFNB1 GJB2 相關非綜合徵性聽力損失 1 型	GJB2
	X-linked Ocular albinism X 連鎖眼皮膚白化病	GPR143
	Usher syndrome type 1 Usher 綜合症 1 型	MYO7A, USH1C, CDH23, PCDH15, USH1G
	Pendred syndrome and non-syndromic deafness type DFNB4 耳聾-甲狀腺腫綜合徵/常染色體隱性遺傳非綜合徵性聽力損失 DFNB4 型	SLC26A4

	Oculocutaneous albinism types 1A, 1B, 2 and 4 眼皮膚白化病 1A，1B，2 和 4 型	TYR, OCA2, SLC45A2
	Usher syndrome type 2A Usher 綜合症 2A 型	USH2A
4 Inherited Endocrine /Urinary Diseases 4 種遺傳內分泌/泌尿疾病	Familial hyperinsulinism 家族性胰島功能亢進	ABCC8, KCNJ11
	Laron syndrome Laron 綜合症	GHR
	X-linked adrenal hypoplasia congenital X 連鎖先天性腎上腺發育不全	NR0B1
	Combined pituitary hormone deficiency types 1, 2 and 3 聯合性垂體激素缺乏症 1，2 和 3 型	POU1F1, PROP1, LHX3
19 Multi-system and Other inherited diseases 19 種多系統綜合症及其他遺傳疾病	Ehlers-Danlos syndrome, cardiac valvular form 心臟瓣膜型 Ehlers-Danlos 綜合症	COL1A2
	Autosomal recessive congenital ichthyosis type 4 常染色體隱性遺傳先天性魚鱗病 4 型	ABCA12
	Autosomal recessive epidermolysis bullosa dystrophica 營養不良性大皰性表皮鬆懈症	COL7A1
	Ehlers-Danlos syndrome type VI Ehlers-Danlos 綜合症 VI 型	PLOD1
	Autosomal recessive congenital ichthyosis type 1 常染色體隱性遺傳先天性魚鱗癬 I 型	TGM1
	Primary hyperoxaluria 原發性高草酸尿症	AGXT, GRHPR
	Autosomal recessive Alport syndrome 常染色體隱性遺傳 Alport 綜合症	COL4A3, COL4A4
	Nephrotic syndrome types 1 and 2 腎病綜合徵 1 和 2 型	NPHS1, NPHS2
	Polycystic kidney disease, autosomal recessive type 常染色體隱性遺傳多囊腎病	PKHD1
	Cystic fibrosis 囊性纖維化	CFTR
	Alpha-1-antitrypsin deficiency α1-抗胰蛋白酶缺乏症	SERPINA1
	ABCB11 deficiency, including progressive familial intrahepatic cholestasis type 2 and benign recurrent intrahepatic cholestasis type 2 ABCB11 缺乏症（包括進行性家族性肝內膽汁淤積症 2 型和良性複發性肝內膽汁淤積 2 型）	ABCB11
	ATP8B1 deficiency, including progressive familial intrahepatic cholestasis type 1 and benign recurrent intrahepatic cholestasis type 1 ATP8B1 缺乏症（包括進行性家族性肝內膽汁淤積症 1 型和良性複發性肝內膽汁淤積 1 型）	ATP8B1
	SLC26A2-related disorders, including atelosteogenesis type II, achondrogenesis type IB, diastrophic dysplasia and multiple epiphyseal dysplasia-4 SLC26A2 相關疾病（包括骨發育不全 II 型，軟骨成長不全 IB 型，骨畸形發育不良和多發性骨骺發育）	SLC26A2

	Shwachman-Diamond syndrome Shwachman-Diamond 綜合症	SBDS
	Joubert syndrome Joubert 綜合症	AHI1, CEP290
	Nijmegen breakage syndrome Nijmegen 斷裂綜合症	NBN
	Lowe syndrome Lowe 綜合症	OCRL
	Mitochondrial trifunctional protein deficiency 線粒體三功能蛋白缺乏症	HADHB

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