愛唯美帶因篩檢 Carrier Screening

本表提供每個基因與其相應疾病為檢測結果為陰性時的殘留風險。所提供的數值是假設該疾病沒有家族史且無症狀。當機率等於或大於1/500時，提供了疾病的殘留風險值。對於機率小於1/500時的疾病，則認為殘留風險已大幅降低。

提供報告時，可從已發布的機率推斷出殘留風險值，殘留風險僅作為檢測結果陰性時其風險的參考，實際數值因個人的種族背景將有所不同。

標有*的基因，由於樣品特異性限制，無法計算出準確的殘留風險值。詳細的規範限制，請參閱檢測者報告的「局限性」部分說明。

Autosomal Recessive

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
1	3-BETA-HYDROXYSTEROID DEHYDROGENASE TYPE II DEFICIENCY (AR) 3-β-羥基類固醇脫氫酶缺乏症-2型	HSD3B2	全人種	< 1 in 500	99%	1 in 49901
2	3-HYDROXY-3-METHYLGLUTARYL-COENZYME A LYASE DEFICIENCY (AR) 3-羥基-3-甲基戊二酸血症	HMGCL	全人種	< 1 in 500	99%	1 in 49901
3	3-METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY (AR) 三甲基巴豆醯輔酶A梭化酶缺乏症-1型	MCCC1	全人種	1 in 147	99%	1 in 14601
4	3-METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY (AR) 三甲基巴豆醯輔酶A梭化酶缺乏症-2型	MCCC2	全人種	1 in 120	99%	1 in 11,901
5	3-PHOSPHOGLYCERATE DEHYDROGENASE DEFICIENCY (AR) 3-磷酸甘油酸去氫酶缺乏症	PHGDH	全人種	< 1 in 500	99%	1 in 49,901
6	6-PYRUVOYL-TETRAHYDROPTERIN SYNTHASE (PTPS) DEFICIENCY (AR) 6-丙酮醯四氫蝶呤合成酶（PTPS）缺乏症	PTS	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
7	ABETALIPOPROTEINEMIA (AR) 無β脂蛋白血症	MTTP	全人種	< 1 in 500	99%	1 in 49,901
8	ACHONDROGENESIS, TYPE 1B (AR) 軟骨發育不全症-1B 型	SLC26A2	全人種	1 in 158	99%	1 in 15,701
9	ACHROMATOPSIA, CNGB3-RELATED (AR) 色彩感應失能症-CNGB3相關	CNGB3	全人種	1 in 98	99%	1 in 9,701
10	ACRODERMATITIS ENTEROPATHICA (AR) 腸病變性肢端皮膚炎	SLC39A4	全人種	1 in 354	99%	1 in 35,301
11	ACUTE INFANTILE LIVER FAILURE, TRMU-RELATED (AR) 急性新生兒肝衰竭-TRMU相關	TRMU	全人種	< 1 in 500	99%	1 in 49,901
12	ACYL-COA OXIDASE I DEFICIENCY (AR) 過氧化物酶酰基輔酶A氧化酶缺乏症	ACOX1	全人種	< 1 in 500	99%	1 in 49,901
13	AICARDI-GOUTIERES SYNDROME (AR) Aicardi-Goutieres 症候群	SAMHD1	全人種	< 1 in 500	99%	1 in 49,901
14	ALKAPTONURIA (AR) 黑尿症	HGD	全人種	1 in 250	99%	1 in 24,900
15	ALPHA-1 ANTITRYPSIN DEFICIENCY (AR) α1-抗胰蛋白酶缺乏症	SERPINA1	全人種	1 in 38	99%	1 in 3,701
16	ALPHA-MANNOSIDOSIS (AR) α-甘露糖貯積症	MAN2B1	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
17	ALPHA-THALASSEMIA (AR) 甲型海洋性貧血	HBA1/ HBA2	全人種	1 in 25	99%	1 in 2,401
18	ALPORT SYNDROME, COL4A3-RELATED (AR) 亞伯氏症候群-COL4A3型	COL4A3	全人種	1 in 323	99%	1 in 32,201
19	ALPORT SYNDROME, COL4A4-RELATED (AR) 亞伯氏症候群-COL4A4型	COL4A4	全人種	1 in 353	99%	1 in 35,201
20	ALSTROM SYNDROME (AR) Alstrom症候群	ALMS1	全人種	< 1 in 500	99%	1 in 49,901
21	ANDERMANN SYNDROME (AR) Andermann 症候群	SLC12A6	全人種	< 1 in 500	99%	1 in 49,901
22	ARGININEMIA (AR) 精胺酸血症	ARG1	全人種	< 1 in 500	99%	1 in 49,901
23	ARGININOSUCCINATE LYASE DEFICIENCY (AR) 精胺丁二酸酶缺乏症	ASL	全人種	1 in 133	99%	1 in 13,201
24	AROMATASE DEFICIENCY (AR) 芳香環轉化酶缺乏症	CYP19A1	全人種	< 1 in 500	99%	1 in 49,901
25	AROMATIC LAMINO ACID DECARBOXYLASE DEFICIENCY (AR) 芳香族L-胺基酸類脫羧基酶缺乏症	DDC	全人種	< 1 in 500	99%	1 in 49,901
26	ASPARAGINE SYNTHETASE DEFICIENCY (AR) 天門冬醯胺酶合成缺乏症	ASNS	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
27	ASPARTYLGLYCOSAMINURIA (AR) 天冬氨醯葡萄糖胺尿症	AGA	全人種	< 1 in 500	99%	1 in 49,901
28	ATAXIA WITH VITAMIN E DEFICIENCY (AR) 共濟失調與維生素 E 缺乏症	TTPA	全人種	< 1 in 500	99%	1 in 49,901
29	ATAXIA-TELANGIECTASIA (AR) 共濟失調微血管擴張症候群	ATM	全人種	1 in 100	99%	1 in 9,901
30	AUTISM SPECTRUM, EPILEPSY AND ARTHROGRYPOSIS (AR) 自閉症譜系障礙-癲癇-關節攣縮症候群	SLC35A3	全人種	< 1 in 500	99%	1 in 49,901
31	AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 (AR) 自體免疫多腺體症候群-1 型	AIRE	全人種	1 in 354	99%	1 in 35,301
32	AUTOSOMAL RECESSIVE SPASTIC ATAXIA OF CHARLEVOIX-SAGUENAY (AR) 遺傳性痙攣性共濟失調症	SACS	全人種	< 1 in 500	99%	1 in 49,901
33	BARDET-BIEDL SYNDROME, BBS10-RELATED (AR) Bardet-Biedl氏症候群-BBS10相關	BBS10	全人種	1 in 447	99%	1 in 44,601
34	BARDET-BIEDL SYNDROME, BBS12-RELATED (AR) Bardet-Biedl氏症候群-BBS12相關	BBS12	全人種	< 1 in 500	99%	1 in 49,901
35	BARDET-BIEDL SYNDROME, BBS1-RELATED (AR) Bardet-Biedl氏症候群-BBS1相關	BBS1	全人種	1 in 265	99%	1 in 26,401
36	BARDET-BIEDL SYNDROME, BBS2-RELATED (AR) Bardet-Biedl氏症候群-BBS2相關	BBS2	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
37	BARE LYMPHOCYTE SYNDROME, CIITA-RELATED (AR) 裸淋巴球症候群-CIITA相關	CIITA	全人種	< 1 in 500	99%	1 in 49,901
38	BARTTER SYNDROME, BSND-RELATED (AR) Bartter 氏症候群-BSND相關	BSND	全人種	< 1 in 500	99%	1 in 49,901
39	BATTEN DISEASE, CLN3-RELATED (AR) 巴頓氏症-CLN3相關	CLN3	全人種	1 in 145	99%	1 in 14,401
40	BERNARD-SOULIER SYNDROME, TYPE A1 (AR) Bernard-Soulier症候群-A1型	GP1BA	全人種	< 1 in 500	99%	1 in 49,901
41	BERNARD-SOULIER SYNDROME, TYPE C (AR) Bernard-Soulier症候群-C型	GP9	全人種	< 1 in 500	99%	1 in 49,901
42	BETA-HEMOGLOBINOPATHIES (AR) β-血紅素疾病	HBB	全人種	1 in 129	99%	1 in 12,801
43	BETA-KETOTHIOLASE DEFICIENCY (AR) β-酮硫解酶缺乏症	ACAT1	全人種	1 in 347	99%	1 in 34,601
44	BILATERAL FRONTOPARIETAL POLYMICROGYRIA (AR) 雙側額頂葉多小腦迴畸形症	GPR56	全人種	< 1 in 500	99%	1 in 49,901
45	BIOTINIDASE DEFICIENCY (AR) 生物素酶缺乏症	BTD	全人種	1 in 120	99%	1 in 11,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
46	BLOOM SYNDROME (AR) Bloom症候群	BLM	全人種	< 1 in 500	99%	1 in 49,901
47	CANAVAN DISEASE (AR) 家族性軸突海綿退化	ASPA	全人種	< 1 in 500	99%	1 in 49,901
48	CARBAMOYL PHOSPHATE SYNTHETASE I DEFICIENCY (AR) 氨甲醯磷酸合成酶缺失症-1型	CPS1	全人種	< 1 in 500	99%	1 in 49,901
49	CARNITINE DEFICIENCY (AR) 原發性肉鹼缺乏症	SLC22A5	全人種	1 in 200	99%	1 in 19,901
50	CARNITINE PALMITOYLTRANSFERASE IA DEFICIENCY (AR) 肉鹼棕櫚醯轉移酶 1A 缺乏症	CPT1A	全人種	< 1 in 500	99%	1 in 49,901
51	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY (AR) 肉鹼棕櫚醯基轉移酶 II 缺乏症	CPT2	全人種	< 1 in 500	99%	1 in 49,901
52	CARPENTER SYNDROME (AR) Carpenter症候群	RAB23	全人種	< 1 in 500	99%	1 in 49,901
53	CARTILAGE-HAIR HYPOPLASIA (AR) 軟骨毛髮發育不全症	RMRP	全人種	< 1 in 500	99%	1 in 49,901
54	CEREBROTENDINOUS XANTHOMATOSIS (AR) 腦腱性黃瘤症	CYP27A1	全人種	1 in 115	99%	1 in 11,401
55	CHARCOT-MARIE-TOOTH-DISEASE, TYPE 4D (AR) 進行性神經性腓骨萎縮症-4D型	NDRG1	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
56	CHOREOACANTHOCYTOSIS (AR) 舞蹈棘狀紅血球症	VPS13A	全人種	< 1 in 500	99%	1 in 49,901
57	CHRONIC GRANULOMATOUS DISEASE, CYBA-RELATED (AR) 慢性肉芽腫病-CYBA相關	CYBA	全人種	< 1 in 500	99%	1 in 49,901
58	CILIOPATHIES, RPGRIP1L-RELATED (AR) RPGRIP1L相關疾病	RPGRIP1L	全人種	1 in 259	99%	1 in 25,801
59	CITRIN DEFICIENCY (AR) Citrin缺乏症	SLC25A13	全人種	< 1 in 500	99%	1 in 49,901
60	CITRULLINEMIA, TYPE 1 (AR) 瓜胺酸血症-1型	ASS1	全人種	1 in 119	99%	1 in 11,801
61	COHEN SYNDROME (AR) 科恩症候群	VPS13B	全人種	< 1 in 500	99%	1 in 49,901
62	COMBINED MALONIC AND METHYLMALONIC ACIDURIA (AR) 丙二酸及甲基丙二酸血症	ACSF3	全人種	1 in 86	99%	1 in 8,501
63	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 1 (AR) 結合性氧化磷酸化缺乏症-1型	GFM1	全人種	< 1 in 500	99%	1 in 49,901
64	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3 (AR) 結合性氧化磷酸化缺乏症-3型	TSFM	全人種	< 1 in 500	99%	1 in 49,901
65	COMBINED PITUITARY HORMONE DEFICIENCY-2 (AR) 結合性腦下垂體賀爾蒙缺失-2型	PROP1	全人種	1 in 141	99%	1 in 14,001

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
66	CONGENITAL ADRENAL HYPERPLASIA, 11-BETA-HYDROXYLASE DEFICIENCY (AR) 先天性腎上腺增生症-11β羥化酶缺失症	CYP11B1	全人種	1 in 158	99%	1 in 15,701
67	CONGENITAL ADRENAL HYPERPLASIA, 17-ALPHA-HYDROXYLASE DEFICIENCY (AR) 先天性腎上腺增生症-17α羥化酶缺失症	CYP17A1	全人種	< 1 in 500	99%	1 in 49,901
68	CONGENITAL ADRENAL HYPERPLASIA, 21-HYDROXYLASE DEFICIENCY (AR) 先天性腎上腺增生症-21羥化酶缺失症	CYP21A2	全人種	1 in 61	98%	1 in 3,001
69	CONGENITAL AMEGAKARYOCYTIC THROMBOCYTOPENIA (AR) 先天巨核細胞缺乏血小板低下症	MPL	全人種	1 in 415	99%	1 in 41,401
70	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, PMM2-Related (AR) 先天性醣基化疾病-1A型	PMM2	全人種	1 in 124	99%	1 in 12,301
71	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B (AR) 先天性醣基化疾病-1B型	MPI	全人種	< 1 in 500	99%	1 in 49,901
72	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C (AR) 先天性醣基化疾病-1C型	ALG6	全人種	< 1 in 500	99%	1 in 49,901
73	CONGENITAL FINNISH NEPHROSIS (AR) 先天性腎病變症候群	NPHS1	全人種	1 in 325	99%	1 in 32,401
74	CONGENITAL HYPERINSULINISM, KCNJ11-Related (AR) 先天性高胰島素血症-KCNJ11相關	KCNJ11	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
75	CONGENITAL INSENSITIVITY TO PAIN WITH ANHIDROSIS (CIPA) (AR) 先天性痛覺不敏感合併無汗症	NTRK1	全人種	< 1 in 500	99%	1 in 49,901
76	CONGENITAL MYASTHENIC SYNDROME, CHRNE-RELATED (AR) 先天性肌無力症候群-CHRNE相關	CHRNE	全人種	< 1 in 500	99%	1 in 49,901
77	CONGENITAL MYASTHENIC SYNDROME, RAPSN-RELATED (AR) 先天性肌無力症候群-RAPSN相關	RAPSN	全人種	1 in 252	99%	1 in 25,101
78	CONGENITAL NEUTROPENIA, HAX1-RELATED (AR) 先天性嗜中性白血球減少症-HAX1相關	HAX1	全人種	1 in 500	99%	Reduced
79	CONGENITAL NEUTROPENIA, VPS45-RELATED (AR) 先天性嗜中性白血球減少症-VPS45相關	VPS45	全人種	1 in 500	99%	Reduced
80	COQ4-RELATED CONDITIONS (AR) COQ4相關疾病	COQ4	全人種	< 1 in 500	99%	1 in 49,901
81	CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS (AR) 角膜失養和感音性失聰症	SLC4A11	全人種	< 1 in 500	99%	1 in 49,901
82	CORTICOSTERONE METHYLOXIDASE DEFICIENCY (AR) 皮質酮甲基氧化酶缺乏症	CYP11B2	全人種	< 1 in 500	99%	1 in 49,901
83	COSTEFF SYNDROME (3-METHYLGLUTACONIC ACIDURIA, TYPE 3) (AR) Costeff症候群	OPA3	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
84	CRB1-RELATED RETINAL DYSTROPHIES (AR) CRB1相關視網膜營養不良症	CRB1	全人種	1 in 112	99%	1 in 11,101
85	CYSTIC FIBROSIS (AR) 囊腫纖維症	CFTR	全人種	1 in 45	99%	1 in 4,401
86	CYSTINOSIS (AR) 胱胺酸血症	CTNS	全人種	1 in 158	99%	1 in 15,701
87	D-BIFUNCTIONAL PROTEIN DEFICIENCY (AR) D-雙功能蛋白缺乏症	HSD17B4	全人種	< 1 in 158	99%	1 in 15,701
88	DEAFNESS, AUTOSOMAL RECESSIVE 77 (AR) 遺傳性聽力障礙77型	LOXHD1	全人種	< 1 in 500	99%	1 in 49,901
89	DYSKERATOSIS CONGENITA, RTEL1-RELATED (AR) 先天性角化不全症-RTEL1相關	RTEL1	全人種	< 1 in 500	99%	1 in 49,901
90	DYSTROPHIC EPIDERMOLYSIS BULLOSA, COL7A1-Related (AR) 表皮分解性水皰症-COL7A1相關	COL7A1	全人種	1 in 370	99%	1 in 36,901
91	EHLERS-DANLOS SYNDROME, TYPE VII C (AR) 埃勒斯-當洛斯症候群 -VII C型	ADAMTS2	全人種	< 1 in 500	99%	1 in 49,901
92	ELLIS-VAN CREVELD SYNDROME, EVC2-RELATED (AR) 埃利偉氏症候群-EVC2相關	EVC2	全人種	1 in 122	99%	1 in 12,101
93	ELLIS-VAN CREVELD SYNDROME, EVC-RELATED (AR) 埃利偉氏症候群-EVC相關	EVC	全人種	1 in 345	99%	1 in 34,401

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
94	ENHANCED S-CONE SYNDROME (AR) 增強型S錐體症候群	NR2E3	全人種	1 in 204	99%	1 in 20,301
95	ERCC6-RELATED DISORDERS (AR) ERCC6相關疾病	ERCC6	全人種	< 1 in 500	99%	1 in 49,901
96	ERCC8-RELATED DISORDERS (AR) ERCC8相關疾病	ERCC8	全人種	< 1 in 500	99%	1 in 49,901
97	ETHYLMALONIC ENCEPHALOPATHY (AR) 乙基丙二酸腦病變	ETHE1	全人種	< 1 in 500	99%	1 in 49,901
98	F2-RELATED CONDITIONS (AR) F2相關疾病	F2	全人種	1 in 33	99%	1 in 3,201
99	F5-RELATED CONDITIONS (AR) F5相關疾病	F5	全人種	1 in 12	99%	1 in 1,101
100	FACTOR XI DEFICIENCY (AR) 第11凝血因子缺乏症	F11	全人種	1 in 92	99%	1 in 9,101
101	FAMILIAL DYSAUTONOMIA (AR) 家族性自主神經失調症-IKBKAP相關	IKBKAP	全人種	< 1 in 500	99%	1 in 49,901
102	FAMILIAL HYPERCHOLESTEROLEMIA, LDLRAP1-RELATED (AR) 家族性高膽固醇血症-LDLRAP相關	LDLRAP1	全人種	< 1 in 500	99%	1 in 49,901
103	FAMILIAL HYPERCHOLESTEROLEMIA, LDLR-RELATED (AR) 家族性高膽固醇血症-LDLR相關	LDLR	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
104	FAMILIAL HYPERINSULINISM, ABCC8-RELATED (AR) (母非帶因者；但父是帶因者時，猶太裔有1in540殘餘風險) 家族性胰島素過多症-ABCC8相關	ABCC8	全人種	1 in 112	99%	1 in 11,101
105	FAMILIAL MEDITERRANEAN FEVER (AR) 家族性地中海熱	MEFV	全人種	1 in 115	99%	1 in 11,401
106	FAMILIAL NEPHROGENIC DIABETES INSIPIDUS, AQP2-RELATED (AR) 家族性腎性尿崩症-AQP2相關	AQP2	全人種	< 1 in 500	99%	1 in 49,901
107	FANCONI ANEMIA, GROUP A (AR) Fanconi氏貧血-A型	FANCA	全人種	1 in 345	99%	1 in 34,401
108	FANCONI ANEMIA, GROUP C (AR) Fanconi氏貧血-C型	FANCC	全人種	1 in 1,053	99%	1 in 105,201
109	FANCONI ANEMIA, GROUP G (AR) Fanconi氏貧血-G型	FANCG	全人種	< 1 in 500	99%	1 in 49,901
110	FUMARASE DEFICIENCY (AR) 延胡索酸酶缺乏症	FH	全人種	< 1 in 500	99%	1 in 49,901
111	GALACTOKINASE DEFICIENCY (GALACTOSEMIA, TYPE II) (AR) 第二型半乳糖血症	GALK1	全人種	1 in 122	99%	1 in 12,101
112	GALACTOSEMIA (AR) 半乳糖血症	GALT	全人種	1 in 110	99%	1 in 10,901
113	GAUCHER DISEASE (AR) 高雪氏症	GBA	全人種	1 in 153	95%	1 in 3,041

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
114	GITELMAN SYNDROME (AR) Gitelman症候群	SLC12A3	全人種	1 in 100	99%	1 in 9,901
115	GLUTARIC ACIDEMIA, TYPE 1 (AR) 戊二酸血症-1型	GCDH	全人種	1 in 112	99%	1 in 11,101
116	GLUTARIC ACIDEMIA, TYPE 2A (AR) 戊二酸血症-2A型	ETFA	全人種	< 1 in 500	99%	1 in 49,901
117	GLUTARIC ACIDEMIA, TYPE 2C (AR) 戊二酸血症-2C型	ETFDH	全人種	1 in 250	99%	1 in 24,901
118	GLYCINE ENCEPHALOPATHY, AMT-RELATED (AR) 非酮性高甘胺酸血症-AMT相關	AMT	全人種	1 in 262	99%	1 in 26,101
119	GLYCINE ENCEPHALOPATHY, GLDC-RELATED (AR) 非酮性高甘胺酸血症-GLDC相關	GLDC	全人種	1 in 135	99%	1 in 13,401
120	GLYCOGEN STORAGE DISEASE TYPE 5 (McArdle Disease) (AR) 肝醣儲積症-5型 (麥卡德爾氏病)	PYGM	全人種	1 in 191	99%	1 in 19,001
121	GLYCOGEN STORAGE DISEASE, TYPE 1a (AR) 肝醣儲積症-1A型	G6PC	全人種	1 in 177	99%	1 in 17,601
122	GLYCOGEN STORAGE DISEASE, TYPE 1b (AR) 肝醣儲積症-1B型	SLC37A4	全人種	1 in 354	99%	1 in 35,301
123	GLYCOGEN STORAGE DISEASE, TYPE 2 (POMPE DISEASE) (AR) 肝醣儲積症-2型 (龐貝氏症)	GAA	全人種	1 in 132	99%	1 in 13,101

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
124	GLYCOGEN STORAGE DISEASE, TYPE 3 (AR) 肝醣儲積症-3型	AGL	全人種	1 in 159	99%	1 in 15,801
125	GLYCOGEN STORAGE DISEASE, TYPE 4 (AR) 肝醣儲積症-4型	GBE1	全人種	1 in 387	99%	1 in 38,601
126	GLYCOGEN STORAGE DISEASE, TYPE 7 (AR) 肝醣儲積症-7型	PFKM	全人種	< 1 in 500	99%	1 in 49,901
127	GRACILE SYNDROME (AR) Gracll 症候群	BCS1L	全人種	1 in 111	99%	1 in 11,001
128	GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY (AR) 胍基乙酸甲基轉移酶缺乏症	GAMT	全人種	< 1 in 500	99%	1 in 49,901
129	HEMOCHROMATOSIS TYPE 2A (AR) 血鐵沉積症-2A型	HFE2	全人種	< 1 in 500	99%	1 in 49,901
130	HEMOCHROMATOSIS, TYPE 3, TFR2-Related (AR) 血鐵沉積症-3型	TFR2	全人種	< 1 in 500	99%	1 in 49,901
131	HEPATOCEREBRAL MITOCHONDRIAL DNA DEPLETION SYNDROME, MPV17-RELATED (AR) 肝腦病變型粒線體DNA耗竭症候群-MPV17相關	MPV17	全人種	< 1 in 500	99%	1 in 49,901
132	HEREDITARY FRUCTOSE INTOLERANCE (AR) 遺傳性果糖不耐症	ALDOB	全人種	1 in 55	99%	1 in 5,401
133	HEREDITARY HEMOCHROMATOSIS TYPE 1 (AR) 血鐵沉積症-1型	HFE	全人種	1 in 10	99%*	1 in 901*

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
134	HEREDITARY SPASTIC PARAPARESIS, TYPE 49 (AR) 痙攣性下身麻痺-49型	TECPR2	全人種	< 1 in 500	99%	1 in 49,901
135	HERMANSKY-PUDLAK SYNDROME, HPS1-RELATED (AR) Hermansky-Pudlak 症候群-HPS1相關	HPS1	全人種	< 1 in 500	99%	1 in 49,901
136	HERMANSKY‐PUDLAK SYNDROME, HPS3-RELATED (AR) Hermansky-Pudlak 症候群-HPS3相關	HPS3	全人種	< 1 in 500	99%	1 in 49,901
137	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY (AR) 全羧化酶合成酶缺乏症	HLCS	全人種	< 1 in 500	99%	1 in 49,901
138	HOMOCYSTINURIA DUE TO DEFICIENCY OF MTHFR (AR) 高胱胺酸尿症-MTHFR缺乏型	MTHFR	全人種	1 in 158	99%	1 in 15,701
139	HOMOCYSTINURIA, CBS-RELATED (AR) 高胱胺酸尿症-CBS相關	CBS	全人種	1 in 224	99%	1 in 22,301
140	HOMOCYSTINURIA, Type cblE (AR) 高胱胺酸尿症- cblE型	MTRR	全人種	< 1 in 500	99%	1 in 49,901
141	HYDROLETHALUS SYNDROME (AR) Hydrolethalus症候群	HYLS1	全人種	1 in 455	99%	1 in 45,401
142	HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA (HHH SYNDROME) (AR) 鳥氨酸-高血氨-高瓜胺酸綜合症候群	SLC25A15	全人種	< 1 in 500	99%	1 in 49,901
143	HYPOPHOSPHATASIA,ALPL-RELATED (AR) 低磷酸酯酶症	ALPL	全人種	1 in 158	95%	1 in 15,701

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
144	INCLUSION BODY MYOPATHY 2 (AR) 包涵體肌炎	GNE	全人種	< 1 in 500	99%	1 in 49,901
145	INFANTILE CEREBRAL AND CEREBELLAR ATROPHY (AR) 產後進展性小頭畸形伴癲癇/腦萎縮	MED17	全人種	< 1 in 500	99%	1 in 49,901
146	ISOVALERIC ACIDEMIA (AR) 異戊酸血症	IVD	全人種	1 in 250	99%	1 in 24,901
147	JOUBERT SYNDROME 2 / MECKEL SYNDROME 2 (AR) Joubert 症候群-2型/Meckel 症候群-2型	TMEM216	全人種	< 1 in 500	99%	1 in 49,901
148	JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMB3-RELATED (AR) 接合性表皮溶解水皰症-LAMB3相關	LAMB3	全人種	1 in 407	99%	1 in 40,601
149	JUNCTIONAL EPIDERMOLYSIS BULLOSA, LAMC2-RELATED (AR) 接合性表皮溶解水皰症-LAMC2相關	LAMC2	全人種	1 in 500	99%	1 in 49,901
150	JUNCTIONAL EPIDERMOLYSIS BULLOSA/LARYNGOONYCHOCUTANEOUS SYNDROME,LAMA3RELATED (AR) 接合性表皮溶解水皰症-LAMA3相關	LAMA3	全人種	< 1 in 500	99%	1 in 49,901
151	KRABBE DISEASE (AR) Krabbe氏症	GALC	全人種	1 in 150	99%	1 in 14,901
152	LAMELLAR ICHTHYOSIS, TYPE 1 (AR) 層狀魚鱗癬-1型	TGM1	全人種	1 in 301	95%	1 in 30,001
153	LEBER CONGENITAL AMAUROSIS 2 (AR) 萊伯氏先天性黑矇症-2型	RPE65	全人種	1 in 228	99%	1 in 22,701

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
154	LEBER CONGENITAL AMAUROSIS, TYPE CEP290 (AR) 萊伯氏先天性黑矇症-CEP290型	CEP290	全人種	1 in 185	99%	1 in 18,401
155	LEBER CONGENITAL AMAUROSIS, TYPE LCA5 (AR) 萊伯氏先天性黑矇症- LCA5型	LCA5	全人種	< 1 in 500	99%	1 in 49,901
156	LEBER CONGENITAL AMAUROSIS, TYPE RDH12 (AR) 萊伯氏先天性黑矇症-RDH12 型	RDH12	全人種	1 in 456	99%	1 in 45,501
157	LEIGH SYNDROME, FRENCH-CANADIAN TYPE (AR) Leigh症候群-法裔加拿大型	LRPPRC	全人種	< 1 in 500	99%	1 in 49,901
158	LETHAL CONGENITAL CONTRACTURE SYNDROME 1 (AR) 致死先天性攣縮症候群	GLE1	全人種	< 1 in 500	99%	1 in 49,901
159	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER (AR) 腦白質病伴隨白質消失症	EIF2B5	全人種	< 1 in 500	99%	1 in 49,901
160	LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2A (AR) 肢帶型肌肉失養症-2A 型	CAPN3	全人種	< 1 in 500	99%	1 in 49,901
161	LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2B (AR) 肢帶型肌失養症 -2B 型	DYSF	全人種	1 in 311	99%	1 in 31,001
162	LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2C (AR) 肢帶型肌肉失養症-2C 型	SGCG	全人種	1 in 354	99%	1 in 35,301

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
163	LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2D (AR) 肢帶型肌肉失養症-2D 型	SGCA	全人種	< 1 in 500	99%	1 in 49,901
164	LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2E (AR) 肢帶型肌肉失養症-2E 型	SGCB	全人種	< 1 in 500	99%	1 in 49,901
165	LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2I (AR) 肢帶型肌肉失養症-2I 型	FKRP	全人種	1 in 158	99%	1 in 15,701
166	LIPOAMIDE DEHYDROGENASE DEFICIENCY (DIHYDROLIPOAMIDE DEHYDROGENASE DEFICIENCY) (AR) 二氫硫辛醯胺脫氫酶缺乏症	DLD	全人種	< 1 in 500	99%	1 in 49,901
167	LIPOID ADRENAL HYPERPLASIA (AR) 脂肪性先天性腎上腺皮質增生症	STAR	全人種	< 1 in 500	99%	1 in 49,901
168	LIPOPROTEIN LIPASE DEFICIENCY (AR) 脂蛋白脂肪酶缺乏症	LPL	全人種	< 1 in 500	99%	1 in 49,901
169	LONG CHAIN 3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (AR) 長鏈 3-羥烷基輔酶A脫氫酶缺乏症	HADHA	全人種	1 in 138	99%	1 in 13,701
170	LYSINURIC PROTEIN INTOLERANCE (AR) Lysinuric 蛋白質耐受不良症	SLC7A7	全人種	< 1 in 500	99%	1 in 49,901
171	MAPLE SYRUP URINE DISEASE, TYPE 1A (AR) 楓糖尿症-1A型	BCKDHA	全人種	1 in 321	99%	1 in 32,001
172	MAPLE SYRUP URINE DISEASE, TYPE 1B (AR) 楓糖尿症-1B型	BCKDHB	全人種	1 in 364	99%	1 in 36,301

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
173	MAPLE SYRUP URINE DISEASE, TYPE 2 (AR) 楓糖尿症-2型	DBT	全人種	1 in 321	99%	1 in 32,001
174	MECKEL-GRUBER SYNDROME, TYPE 1 (AR) Meckel-Gruber症候群-1型	MKS1	全人種	1 in 260	99%	1 in 25,901
175	MEDIUM CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (AR) 中鏈醯輔酶 A 去氫酶缺乏症	ACADM	全人種	1 in 64	99%	1 in 6,301
176	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS (AR) 巨腦性腦白質病伴有皮層下囊腫	MLC1	全人種	< 1 in 500	99%	1 in 49,901
177	MEROSIN-DEFICIENT MUSCULAR DYSTROPHY (AR) 肌肉失養症-LAMA2相關	LAMA2	全人種	1 in 87	99%	1 in 8,601
178	METACHROMATIC LEUKODYSTROPHY, ARSA-RELATED (AR) 異染性腦白質退化症-ARSA相關	ARSA	全人種	1 in 100	99%	1 in 9,901
179	METACHROMATIC LEUKODYSTROPHY, PSAP-RELATED (AR) 異染性腦白質退化症-PSAP相關	PSAP	全人種	< 1 in 500	99%	1 in 49,901
180	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE Cb1C (AR) 甲基丙二酸血症併高胱胺酸血症-Cb1C型	MMACHC	全人種	1 in 138	99%	1 in 13,701
181	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CblD (AR) 甲基丙二酸血症併高胱胺酸血症-Cb1D型	MMADHC	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
182	METHYLMALONIC ACIDURIA, MMAA-RELATED (AR) 甲基丙二酸血症-MMAA相關	MMAA	全人種	1 in 316	99%	1 in 31,501
183	METHYLMALONIC ACIDURIA, MMAB-RELATED (AR) 甲基丙二酸血症-MMAB相關	MMAB	全人種	1 in 456	99%	1 in 45,501
184	METHYLMALONIC ACIDURIA, TYPE MUT(0) (AR) 甲基丙二酸血症-MUT型	MUT	全人種	1 in 383	99%	1 in 38,201
185	MICROPHTHALMIA / ANOPHTHALMIA, VSX2-RELATED (AR) 小眼症/無眼症-VSX2相關	VSX2	全人種	< 1 in 500	99%	1 in 49,901
186	MITOCHONDRIAL COMPLEX 1 DEFICIENCY, ACAD9-RELATED (AR) 粒線體複合物 I 缺乏症-ACAD9相關	ACAD9	全人種	< 1 in 500	99%	1 in 49,901
187	MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFAF5-RELATED (AR) 粒線體複合物 I 缺乏症-NDUFAF5相關	NDUFAF5	全人種	< 1 in 500	99%	1 in 49,901
188	MITOCHONDRIAL COMPLEX 1 DEFICIENCY, NDUFS6-RELATED (AR) 粒線體複合物 I 缺乏症-NDUFS6相關	NDUFS6	全人種	< 1 in 500	99%	1 in 49,901
189	MITOCHONDRIAL MYOPATHY AND SIDEROBLASTIC ANEMIA (MLASA1) (AR) 線粒體肌病和鐵粒細胞性貧血	PUS1	全人種	< 1 in 500	99%	1 in 49,901
190	MUCOLIPIDOSIS II/III A (AR) 黏脂質症 -2/3型	GNPTAB	全人種	1 in 158	99%	1 in 15,701
191	MUCOLIPIDOSIS III GAMMA (AR) 黏脂症 III γ 型	GNPTG	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
192	MUCOLIPIDOSIS, TYPE IV (AR) 黏脂質症-4型	MCOLN1	全人種	< 1 in 500	99%	1 in 49,901
193	MUCOPOLYSACCHARIDOSIS, TYPE I (HURLER SYNDROME) (AR) 黏多醣症-1型 (Hurler 症候群)	IDUA	全人種	1 in 158	99%	1 in 15,701
194	MUCOPOLYSACCHARIDOSIS, TYPE III A (SANFILIPPO A) (AR) 黏多醣症-3A型	SGSH	全人種	1 in 415	99%	1 in 41,401
195	MUCOPOLYSACCHARIDOSIS, TYPE III B (SANFILIPPO B) (AR) 黏多醣症-3B型	NAGLU	全人種	< 1 in 500	99%	1 in 49,901
196	MUCOPOLYSACCHARIDOSIS, TYPE III C (SANFILIPPO C) (AR) 黏多醣症-3C型	HGSNAT	全人種	1 in 482	99%	1 in 48,101
197	MUCOPOLYSACCHARIDOSIS, TYPE III D (SANFILIPPO D) (AR) 黏多醣症-3D型	GNS	全人種	< 1 in 500	99%	1 in 49,901
198	MUCOPOLYSACCHARIDOSIS, TYPE IV B/GM1 GANGLIOSIDOSIS (AR) 黏多醣症-4B型	GLB1	全人種	1 in 158	99%	1 in 15,701
199	MUCOPOLYSACCHARIDOSIS, TYPE IX (AR) 黏多醣症-9型	HYAL1	全人種	< 1 in 500	99%	1 in 49,901
200	MUCOPOLYSACCHARIDOSIS, TYPE VI (MAROTEAUX-LAMY) (AR) 黏多醣症-6型	ARSB	全人種	1 in 291	99%	1 in 29,001
201	MULTIPLE SULFATASE DEFICIENCY (AR) 多發性硫酸脂酶缺乏症	SUMF1	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
202	MUSCLE-EYE-BRAIN DISEASE, POMGNT1-RELATED (AR) 肌肉-眼睛-大腦-POMGNT1相關疾病	POMGNT1	全人種	1 in 462	99%	1 in 46,101
203	MYONEUROGASTROINTESTINAL ENCEPHALOPATHY (MNGIE) (AR) 粒線體性神經胃腸腦病變症候群	TYMP	全人種	< 1 in 500	99%	1 in 49,901
204	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY (AR) N-乙醯穀胺酸合成酶缺乏症	NAGS	全人種	< 1 in 500	99%	1 in 49,901
205	NEMALINE MYOPATHY, NEB-RELATED (AR) 桿狀體肌症-NEB相關	NEB	全人種	1 in 224	99%	1 in 22,301
206	NEURONAL CEROID LIPOFUSCINOSIS, CLN5-RELATED (AR) 神經元蠟樣脂褐質沉著症-CLN5相關	CLN5	全人種	1 in 317	99%	1 in 31,601
207	NEURONAL CEROID LIPOFUSCINOSIS, CLN6-RELATED (AR) 神經元蠟樣脂褐質沉著症-CLN6相關	CLN6	全人種	1 in 261	99%	1 in 26,001
208	NEURONAL CEROID LIPOFUSCINOSIS, CLN8-RELATED (AR) 神經元蠟樣脂褐質沉著症-CLN8相關	CLN8	全人種	1 in 349	99%	1 in 34,801
209	NEURONAL CEROID LIPOFUSCINOSIS, MFSD8-RELATED (AR) 神經元蠟樣脂褐質沉著症-MFSD8相關	MFSD8	全人種	< 1 in 500	99%	1 in 49,901
210	NEURONAL CEROID LIPOFUSCINOSIS, PPT1-RELATED (AR) 神經元蠟樣脂褐質沉著症-PPT1相關	PPT1	全人種	1 in 368	99%	1 in 36,701

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
211	NEURONAL CEROID LIPOFUSCINOSIS, TPP1-RELATED (AR) 神經元蠟樣脂褐質沉著症-TPP1相關	TPP1	全人種	1 in 314	99%	1 in 31,301
212	NIEMANN-PICK DISEASE, TYPE C1 / D (AR) 尼曼匹克症-C1/D型	NPC1	全人種	1 in 282	99%	1 in 28,101
213	NIEMANN-PICK DISEASE, TYPE C2 (AR) 尼曼匹克症-C2型	NPC2	全人種	< 1 in 500	99%	1 in 49,901
214	NIEMANN-PICK DISEASE, TYPES A / B (AR) 尼曼匹克症-A/B型	SMPD1	全人種	1 in 196	99%	1 in 19,501
215	NIJMEGEN BREAKAGE SYNDROME (AR) Nijmegen破損症候群	NBN	全人種	< 1 in 500	99%	1 in 49,901
216	NON-SYNDROMIC HEARING LOSS, GJB2-RELATED (AR) 非症候群感覺神經性聽損-GJB2相關	GJB2	全人種	1 in 42	99%	1 in 4,101
217	ODONTO-ONYCHO-DERMAL DYSPLASIA / SCHOPF-SCHULZ-PASSARGE SYNDROME (AR) 指甲-皮膚-外胚層發育不全症/ Schopf-Schulz-Passarge症候群	WNT10A	全人種	1 in 305	99%	1 in 30,401
218	OMENN SYNDROME, RAG2-RELATED (AR) Omenn 症候群-RAG2相關	RAG2	全人種	< 1 in 500	99%	1 in 49,901
219	ORNITHINE AMINOTRANSFERASE DEFICIENCY (AR) 鳥胺酸酮酸轉胺酶缺乏症	OAT	全人種	< 1 in 500	99%	1 in 49,901
220	OSTEOPETROSIS, INFANTILE MALIGNANT, TCIRG1-RELATED (AR) 骨質石化症-TCIRG1相關	TCIRG1	全人種	1 in 316	99%	1 in 31,501

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
221	PENDRED SYNDROME (AR) Pendred 氏症候群	SLC26A4	全人種	1 in 80	99%	1 in 7,901
222	PHENYLKETONURIA (AR) 苯酮尿症	PAH	全人種	1 in 65	99%	1 in 6,401
223	PITUITARY HORMONE DEFICIENCY, COMBINED 3 (AR) 結合性腦下垂體賀爾蒙缺失-3型	LHX3	全人種	< 1 in 500	99%	1 in 49,901
224	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (AR) 隱性多囊腎病	PKHD1	全人種	1 in 144	99%	1 in 14,301
225	PONTOCEREBELLAR HYPOPLASIA, RARS2-RELATED (AR) 橋腦小腦發育不全-RARS2相關	RARS2	全人種	< 1 in 500	99%	1 in 49,901
226	PONTOCEREBELLAR HYPOPLASIA, TYPE 1A (AR) 橋腦小腦發育不全-1A型	VRK1	全人種	< 1 in 500	99%	1 in 49,901
227	PONTOCEREBELLAR HYPOPLASIA, TYPE 2D (AR) 橋腦小腦發育不全-2D型	SEPSECS	全人種	< 1 in 500	99%	1 in 49,901
228	PRIMARY CILIARY DYSKINESIA, DNAH5-RELATED (AR) 原發性纖毛運動障礙-DNAH5相關	DNAH5	全人種	1 in 120	99%	1 in 11,901
229	PRIMARY CILIARY DYSKINESIA, DNAI1-RELATED (AR) 原發性纖毛運動障礙-DNAI1相關	DNAI1	全人種	1 in 182	99%	1 in 18,101
230	PRIMARY CILIARY DYSKINESIA, DNAI2-RELATED (AR) 原發性纖毛運動障礙-DNAI2相關	DNAI2	全人種	< 1 in 500	99%	1 in 49,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
231	PRIMARY HYPEROXALURIA, TYPE 1 (AR) 原發性高草酸尿症-1型	AGXT	全人種	1 in 158	99%	1 in 15,701
232	PRIMARY HYPEROXALURIA, TYPE 2 (AR) 原發性高草酸尿症-2型	GRHPR	全人種	< 1 in 500	99%	1 in 49,901
233	PRIMARY HYPEROXALURIA, TYPE 3 (AR) 原發性高草酸尿症-3型	HOGA1	全人種	1 in 309	99%	1 in 30,801
234	PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 2 (AR) 進行性家族性肝內膽汁滯留症第二型	ABCB11	全人種	1 in 158	99%	1 in 15,701
235	PROPIONIC ACIDEMIA, PCCA-RELATED (AR) 丙酸血症-PCCA相關	PCCA	全人種	1 in 224	99%	1 in 22,301
236	PROPIONIC ACIDEMIA, PCCB-RELATED (AR) 丙酸血症-PCCB相關	PCCB	全人種	1 in 224	99%	1 in 22,301
237	PYCNODYSOSTOSIS (AR) 緻密性成骨不全症	CTSK	全人種	1 in 439	99%	1 in 43,801
238	PYRUVATE CARBOXYLASE DEFICIENCY (AR) 丙酮酸羧化酶缺乏症	PC	全人種	1 in 250	99%	1 in 24,901
239	PYRUVATE DEHYDROGENASE DEFICIENCY, PDHB-RELATED (AR) 丙酮酸鹽脫氫酶缺乏症-PDHB相關	PDHB	全人種	< 1 in 500	99%	1 in 49,901
240	RENAL TUBULAR ACIDOSIS AND DEAFNESS, ATP6V1B1-RELATED (AR) 腎小管酸中毒/耳聾-ATP6V1B1相關	ATP6V1B1	全人種	< 1 in 500	99%	1 in 49,901
241	RETINITIS PIGMENTOSA 25 (AR) 視網膜色素病變25型	EYS	全人種	1 in 129	99%	1 in 12,801

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
242	RETINITIS PIGMENTOSA 26 (AR) 視網膜色素病變26型	CERKL	全人種	1 in 137	99%	1 in 13,601
243	RETINITIS PIGMENTOSA 28 (AR) 視網膜色素病變28型	FAM161A	全人種	1 in 289	99%	1 in 28,801
244	RETINITIS PIGMENTOSA 59 (AR) 視網膜色素病變59型	DHDDS	全人種	< 1 in 500	99%	1 in 49,901
245	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 (AR) 肢近端型點狀軟骨發育不良-1型	PEX7	全人種	< 1 in 500	99%	1 in 49,901
246	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3 (AR) 肢近端型點狀軟骨發育不良-3型	AGPS	全人種	< 1 in 500	99%	1 in 49,901
247	ROBERTS SYNDROME (AR) Roberts症候群	ESCO2	全人種	< 1 in 500	99%	1 in 49,901
248	SALLA DISEASE (AR) 薩拉氏症	SLC17A5	全人種	< 1 in 500	99%	1 in 49,901
249	SANDHOFF DISEASE (AR) Sandoff症	HEXB	全人種	1 in 278	99%	1 in 27,701
250	SCHIMKE IMMUNOOSSEOUS DYSPLASIA (AR) Schimke免疫-骨發育不良	SMARCAL1	全人種	< 1 in 500	99%	1 in 49,901
251	SEGAWA SYNDROME, TH-RELATED (AR) Segawa 症候群-TH相關	TH	全人種	< 1 in 500	99%	1 in 49,901
252	SEVERE COMBINED IMMUNODEFICIENCY, ADA-Related (AR) 嚴重結合免疫缺乏症-ADA相關	ADA	全人種	1 in 224	99%	1 in 22,301

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
253	SEVERE COMBINED IMMUNODEFICIENCY, TYPE ATHABASKAN (AR) 嚴重結合免疫缺乏症-DCLRE1C相關	DCLRE1C	全人種	< 1 in 500	99%	1 in 49,901
254	SJOGREN-LARSSON SYNDROME (AR) Sjogren-Larsson症候群	ALDH3A2	全人種	1 in 223	99%	1 in 22,201
255	SMITH-LEMLI-OPITZ SYNDROME (AR) Smith-Lemli-Opitz 症候群	DHCR7	全人種	1 in 100	99%	1 in 9,901
256	SPASTIC PARAPLEGIA, TYPE 15 (AR) 痙攣性下身麻痺-15型	ZFYVE26	全人種	< 1 in 500	99%	1 in 49,901
257	SPINAL MUSCULAR ATROPHY (AR) {SMN1: Two copies; g.27134T>G: absent; the absence of the g.27134T>G variant decreases the chance to be a silent (2+0) carrier.} 脊髓性肌肉萎縮症	SMN1	全人種	1 in 54	91%	1 in 589 (2 copy)
258	SPONDYLOTHORACIC DYSOSTOSIS, MESP2-Related (AR) 脊椎肋骨發育不全-MESP2相關	MESP2	全人種	1 in 224	99%	1 in 22,301
259	STEEL SYNDROME (AR) Steel 症候群	COL27A1	全人種	< 1 in 500	99%	1 in 49,901
260	STEROID-RESISTANT NEPHROTIC SYNDROME (AR) 類固醇抗性腎病症候群	NPHS2	全人種	1 in 377	99%	1 in 37,601
261	STUVE-WIEDEMANN SYNDROME (AR) Stuve-Wiedemann 症候群	LIFR	全人種	< 1 in 500	99%	1 in 49,901
262	TAY-SACHS DISEASE (AR) 泰-薩克斯症	HEXA	全人種	1 in 250	99%	1 in 24,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
263	TYROSINEMIA, TYPE 1 (AR) 酪胺酸血症-1型	FAH	全人種	1 in 100	99%	1 in 9,901
264	TYROSINEMIA, TYPE 2 (AR) 酪胺酸血症-2型	TAT	全人種	< 1 in 500	99%	1 in 49,901
265	USHER SYNDROME, TYPE 1B (AR) 尤塞氏症候群- 1B 型	MYO7A	全人種	1 in 206	99%	1 in 20,501
266	USHER SYNDROME, TYPE 1C (AR) 尤塞氏症候群- 1C 型	USH1C	全人種	1 in 353	99%	1 in 35,201
267	USHER SYNDROME, TYPE 1D (AR) 尤塞氏症候群- 1D 型	CDH23	全人種	1 in 202	99%	1 in 20,101
268	USHER SYNDROME, TYPE 1F (AR) 尤塞氏症候群- 1F 型	PCDH15	全人種	1 in 395	99%	1 in 39,401
269	USHER SYNDROME, TYPE 2A (AR) 尤塞氏症候群- 2A 型	USH2A	全人種	1 in 126	99%	1 in 12,501
270	USHER SYNDROME, TYPE 3 (AR) 尤塞氏症候群- 3 型	CLRN1	全人種	< 1 in 500	99%	1 in 49,901
271	VERY LONG-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (AR) 特長鏈醯輔酶 A 去氫酶缺乏症	ACADVL	全人種	1 in 86	99%	1 in 8,501
272	WALKER-WARBURG SYNDROME, FKTN-RELATED (AR) Walker-Warburg 症候群	FKTN	全人種	< 1 in 500	99%	1 in 49,901
273	WILSON DISEASE (AR) 威爾森氏症	ATP7B	全人種	1 in 90	99%	1 in 8,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
274	WOLMAN DISEASE (AR) 伍爾曼氏症	LIPA	全人種	< 1 in 500	99%	1 in 49,901
276	XERODERMA PIGMENTOSUM, GROUP C (AR) 著色性乾皮症-C型	XPC	全人種	< 1 in 500	99%	1 in 49,901
277	ZELLWEGER SPECTRUM DISORDERS, PEX10-RELATED (AR) 柴爾維格氏症候群-PEX10相關	PEX10	全人種	< 1 in 500	99%	1 in 49,901
278	ZELLWEGER SPECTRUM DISORDERS, PEX12-RELATED (AR) 柴爾維格氏症候群-PEX12相關	PEX12	全人種	1 in 406	99%	1 in 40,501
279	ZELLWEGER SPECTRUM DISORDERS, PEX1-RELATED (AR) 柴爾維格氏症候群-PEX1相關	PEX1	全人種	< 1 in 500	99%	1 in 49,901
280	ZELLWEGER SPECTRUM DISORDERS, PEX2-RELATED (AR) 柴爾維格氏症候群-PEX2相關	PEX2	全人種	< 1 in 500	99%	1 in 49,901
281	ZELLWEGER SPECTRUM DISORDERS, PEX6-RELATED (AR) 柴爾維格氏症候群-PEX6相關	PEX6	全人種	1 in 280	99%	1 in 27,901

X-Linked

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
282	ADRENOLEUKODYSTROPHY, X-LINKED (XL) 性聯遺傳腎上腺白質退化症	ABCD1	全人種	1 in 10,500	99%	1 in 1,049,901
283	ALPHA-THALASSEMIA INTELLECTUAL DISABILITY SYNDROME (XL) 甲型海洋性貧血-性聯遺傳智力障礙症候群	ATRX	全人種	< 1 in 750,000	99%	1 in 74,999,901
284	ALPORT SYNDROME, X-LINKED (XL) 亞伯氏症候群	COL4A5	全人種	1 in 47,000	99%	1 in 4,699,901
285	ARTS SYNDROME (XL) Arts 症候群	PRPS1	全人種	1 in 500,000	99%	1 in 49,999,901
286	CHARCOT-MARIE-TOOTH DISEASE WITH DEAFNESS, X-LINKED (CMTX1) (XL) 進行性神經性腓骨萎縮症-1型	GJB1	全人種	1 in 3,700	99%	1 in 369,901
287	CHOROIDEREMIA (XL) 脈絡膜缺失症	CHM	全人種	1 in 25,000	99%	1 in 2,499,901
288	CHRONIC GRANULOMATOUS DISEASE, X-LINKED (XL) 性聯遺傳慢性肉芽腫病	CYBB	全人種	1 in 150,000	99%	1 in 15,000,000
289	CREATINE TRANSPORTER DEFECT (Cerebral Creatine Deficiency Syndrome 1, X-Linked) (XL) 性聯遺傳肌酸缺乏症	SLC6A8	全人種	1 in 20,600	99%	1 in 2,060,000
290	DUCHENNE/BECKER MUSCULAR DYSTROPHY (XL) 裘馨氏肌肉萎縮症	DMD	全人種	1 in 4,200	99%	1 in 419,901

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
291	EMERY-DREIFUSS MUSCULAR DYSTROPHY 1, X-LINKED (XL) Emery-Dreifuss 肌失養症-EMD相關	EMD	全人種	1 in 250,000	99%	1 in 25,000,000
292	FABRY DISEASE (XL) 法布瑞氏症	GLA	全人種	1 in 1,500	99%	1 in 149,901
293	FACTOR IX DEFICIENCY (XL) B型血友病	F9	全人種	1 in 10,000	99%	1 in 999,901
294	FRAGILE X SYNDROME (XL) X染色體脆折症	FMR1	全人種	1 in 250	99%	1 in 24,901
295	GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY (XL) 蠶豆症	G6PD	全人種	1 in 30	99%	1 in 2,901
296	HYPOHIDROTIC ECTODERMAL DYSPLASIA, X-LINKED (XL) 少汗性外胚層發育不良症	EDA	全人種	1 in 3,800	99%	1 in 379,901
297	JUVENILE RETINOSCHISIS, X-LINKED (XL) 性聯遺傳視網膜裂損症	RS1	全人種	1 in 2,500	99%	1 in 249,901
298	MENKES SYNDROME (XL) Menkes氏症候群	ATP7A	全人種	1 in 75,000	99%	1 in 7,499,901
299	MUCOPOLYSACCHARIDOSIS, TYPE II (HUNTER SYNDROME) (XL) 黏多醣症-2型 (Hunter 症候群)	IDS	全人種	1 in 60,000	90%	1 in 599,991

序號 No.	疾病 DISORDER	基因 GENE	種族 ETHNICITY	帶因率 CARRIER RATE	檢出率 DETECTION RATE	剩餘風險 RESIDUAL RISK
300	MYOTUBULAR MYOPATHY, X-LINKED (XL) 性聯遺傳肌小管病變	MTM1	全人種	1 in 38,000	99%	1 in 3,799,901
301	ORNITHINE TRANSCARBAMYLASE DEFICIENCY (XL) 鳥胺酸氨甲醯基轉移酶缺乏症	OTC	全人種	< 1 in 30,000	99%	1 in 2,999,901
302	PYRUVATE DEHYDROGENASE DEFICIENCY, X-LINKED (XL) 丙酮酸鹽脫氫酶缺乏症	PDHA1	全人種	< 1 in 750,000	99%	1 in 74,999,901
303	SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED (XL) 性聯遺傳嚴重免疫缺陷	IL2RG	全人種	1 in 69,000	99%	1 in 6,899,901