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	17-BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY (AR) 第三型 17-β-羥基類固醇脫氫酶缺乏症	HSD17B3	全人種	< 1 in 500	99%	1 in 49,901
2	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 49,901
3	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 49,901
4	3-HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (AR) 3-羥基醯輔酶A去氫酶缺乏症	HADHA	全人種	1 in 138	99%	1 in 13,701
5	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 14,601

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
6	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
7	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
8	5-ALPHA-REDUCTASE DEFICIENCY (AR) 類固醇5α-還原酶2缺乏症	SRD5A2	全人種	< 1 in 500	99%	1 in 49,901
9	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
10	ABCA4-RELATED CONDITIONS (AR) ABCA4相關疾病	ABCA4	全人種	1 in 45	99%	1 in 4,401
11	ABETALIPOPROTEINEMIA (AR) 無β脂蛋白血症	MTTP	全人種	< 1 in 500	99%	1 in 49,901
12	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
13	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
14	ACRODERMATITIS ENTEROPATHICA (AR) 腸病變性肢端皮膚炎	SLC39A4	全人種	1 in 354	99%	1 in 35,301
15	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
16	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

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	AICARDI-GOUTIERES SYNDROME (AR) Aicardi-Goutieres 症候群	SAMHD1	全人種	< 1 in 500	99%	1 in 49,901
18	AICARDI-GOUTIERES SYNDROME, RNASEH2A-RELATED (AR) Aicardi-Goutieres 症候群-RNASEH2A相關	RNASEH2A	全人種	< 1 in 500	99%	1 in 49,901
19	AICARDI-GOUTIERES SYNDROME, RNASEH2B-RELATED (AR) Aicardi-Goutieres 症候群-RNASEH2B相關	RNASEH2B	全人種	< 1 in 500	99%	1 in 49,901
20	AICARDI-GOUTIERES SYNDROME, RNASEH2C-RELATED (AR) Aicardi-Goutieres 症候群-RNASEH2C相關	RNASEH2C	全人種	< 1 in 500	99%	1 in 49,901
21	AICARDI-GOUTIERES SYNDROME, TREX1-RELATED (AR) Aicardi-Goutieres 症候群-TREX1相關	TREX1	全人種	< 1 in 500	99%	1 in 49,901
22	ALKAPTONURIA (AR) 黑尿症	HGD	全人種	1 in 250	99%	1 in 24,900
23	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
24	ALPHA-MANNOSIDOSIS (AR) α-甘露糖貯積症	MAN2B1	全人種	1 in 354	99%	1 in 35,300
25	ALPHA-THALASSEMIA (AR) 甲型海洋性貧血	HBA1/ HBA2	全人種	< 1 in 500	99%	1 in 49,901
26	ALPORT SYNDROME, COL4A3-RELATED (AR) 亞伯氏症候群-COL4A3相關	COL4A3	全人種	1 in 323	99%	1 in 32,201

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	ALPORT SYNDROME, COL4A4-RELATED (AR) 亞伯氏症候群-COL4A4相關	COL4A4	全人種	1 in 353	99%	1 in 35,201
28	ALSTROM SYNDROME (AR) Alstrom症候群	ALMS1	全人種	< 1 in 500	99%	1 in 49,901
29	AMISH INFANTILE EPILEPSY SYNDROME (AR) 阿米什嬰兒癲癇症候群	ST3GAL5	全人種	< 1 in 500	99%	1 in 49,901
30	ANDERMANN SYNDROME (AR) Andermann 症候群	SLC12A6	全人種	< 1 in 500	99%	1 in 49,901
31	ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY (AGAT DEFICIENCY) (AR) 精胺酸：甘胺酸脒基轉移酶(AGAT缺乏症)	GATM	全人種	≤1 in 500	99%	1 in 49,901
32	ARGININEMIA (AR) 精胺酸血症	ARG1	全人種	< 1 in 500	99%	1 in 49,901
33	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
34	AROMATASE DEFICIENCY (AR) 芳香環轉化酶缺乏症	CYP19A1	全人種	< 1 in 500	99%	1 in 49,901
35	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
36	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
37	ASPARTYLGLYCOSAMINURIA (AR) 天冬氨醯葡萄糖胺尿症	AGA	全人種	< 1 in 500	99%	1 in 49,901
38	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
39	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
40	ATAXIA-TELANGIECTASIA-LIKE DISORDER 1 (AR) 類共濟失調微血管擴張症 1 型	MRE11	全人種	< 1 in 500	99%	1 in 49,901
41	ATRANSFERRINEMIA (AR) 無運鐵蛋白症	TF	全人種	1 in 116	99%	1 in 11,501
42	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
43	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
44	AUTOSOMAL RECESSIVE CONGENITAL ICHTHYOSIS (ARCI), SLC27A4-RELATED (AR) 魚鱗癬-SLC27A4相關	SLC27A4	全人種	≤1 in 500	99%	1 in 49,901
45	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
46	BARDET-BIEDL SYNDROME, ARL6-RELATED (AR) Bardet-Biedl 症候群-ARL6相關	ARL6	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
47	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
48	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
49	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
50	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
51	BARDET-BIEDL SYNDROME, BBS4-RELATED (AR) Bardet-Biedl氏症候群-BBS4相關	BBS4	全人種	< 1 in 500	99%	1 in 49,901
52	BARDET-BIEDL SYNDROME, BBS5-RELATED (AR) Bardet-Biedl氏症候群-BBS5相關	BBS5	全人種	≤1 in 500	99%	1 in 49,901
53	BARDET-BIEDL SYNDROME, BBS7-RELATED (AR) Bardet-Biedl氏症候群-BBS7相關	BBS7	全人種	< 1 in 500	99%	1 in 49,901
54	BARDET-BIEDL SYNDROME, BBS9-RELATED (AR) Bardet-Biedl氏症候群-BBS9相關	BBS9	全人種	< 1 in 500	99%	1 in 49,901
55	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

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	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
57	BARTTER SYNDROME, KCNJ1-RELATED (AR) 巴特氏症候群-KCNJ1相關	KCNJ1	全人種	≤1 in 500	99%	1 in 49,901
58	BARTTER SYNDROME, SLC12A1-RELATED (AR) 巴特氏症候群-SLC12A1相關	SLC12A1	全人種	1 in 360	99%	1 in 35,901
59	BATTEN DISEASE, CLN3-RELATED (AR) 巴頓氏症-CLN3相關	CLN3	全人種	1 in 145	99%	1 in 14,401
60	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
61	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
62	BETA-HEMOGLOBINOPATHIES (AR) β-血紅素疾病	HBB	全人種	1 in 129	99%	1 in 12,801
63	BETA-KETOTHIOLASE DEFICIENCY (AR) β-酮硫解酶缺乏症	ACAT1	全人種	1 in 347	99%	1 in 34,601
64	BETA-MANNOSIDOSIS (AR) β-甘露糖苷貯積症	MANBA	全人種	≤1 in 500	99%	1 in 49,901
65	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

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	BIOTINIDASE DEFICIENCY (AR) 生物素酶缺乏症	BTD	全人種	1 in 120	99%	1 in 11,901
67	BIOTIN-THIAMINE-RESPONSIVE BASAL GANGLIA DISEASE (BTBGD) (AR) 生物素-硫胺素反應性基底核疾病	SLC19A3	全人種	1 in 109	99%	1 in 10,801
68	BLOOM SYNDROME (AR) Bloom症候群	BLM	全人種	< 1 in 500	99%	1 in 49,901
69	BRITTLE CORNEA SYNDROME 1 (AR) 脆性角膜症候群-1型	ZNF469	全人種	≤1 in 500	99%	1 in 49,901
70	BRITTLE CORNEA SYNDROME 2 (AR) 脆性角膜症候群-2型	PRDM5	全人種	≤1 in 500	99%	1 in 49,901
71	CANAVAN DISEASE (AR) 家族性軸突海綿退化	ASPA	全人種	< 1 in 500	99%	1 in 49,901
72	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
73	CARNITINE DEFICIENCY (AR) 原發性肉鹼缺乏症	SLC22A5	全人種	1 in 200	99%	1 in 19,901
74	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
75	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

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
76	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY (AR) 肉鹼轉位酶缺乏症	SLC25A20	全人種	< 1 in 500	99%	1 in 49,901
77	CARPENTER SYNDROME (AR) Carpenter症候群	RAB23	全人種	< 1 in 500	99%	1 in 49,901
78	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
79	CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (AR) 兒茶酚胺多型性心室頻脈-CASQ2相關	CASQ2	全人種	1 in 224	99%	1 in 22,300
80	CD59-MEDIATED HEMOLYTIC ANEMIA (AR) 溶血性貧血-CD59型	CD59	全人種	≤1 in 500	99%	1 in 49,901
81	CEP152-RELATED MICROCEPHALY (AR) 小頭畸形-CEP152相關	CEP152	全人種	≤1 in 500	99%	1 in 49,901
82	CEREBRAL DYSGENESIS, NEUROPATHY, ICHTHYOSIS, AND PALMOPLANTAR KERATODERMA (CEDNIK) SYNDROME (AR) 腦發育不全、魚鱗癬及角化症	SNAP29	全人種	≤1 in 500	99%	1 in 49,901
83	CEREBROTENDINOUS XANTHOMATOSIS (AR) 腦腱性黃瘤症	CYP27A1	全人種	1 in 115	99%	1 in 11,401
84	CHARCOT-MARIE-TOOTH DISEASE, RECESSIVE INTERMEDIATE C (AR) Charcot-marie-tooth氏症,隱性中間型	PLEKHG5	全人種	≤1 in 500	99%	1 in 49,901
85	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
86	CHEDIAK-HIGASHI SYNDROME (AR) Chediak-Higashi 症候	LYST	全人種	< 1 in 500	99%	1 in 49,901
87	CHOREOACANTHOCYTOSIS (AR) 舞蹈棘狀紅血球症	VPS13A	全人種	< 1 in 500	99%	1 in 49,901
88	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
89	CHRONIC GRANULOMATOUS DISEASE, NCF2-RELATED (AR) 慢性肉芽腫病-NCF2相關	NCF2	全人種	≤1 in 500	99%	1 in 49,901
90	CILIOPATHIES, RPGRIP1L-RELATED (AR) RPGRIP1L相關疾病	RPGRIP1L	全人種	1 in 259	99%	1 in 25,801
91	CITRIN DEFICIENCY (AR) Citrin缺乏症	SLC25A13	全人種	< 1 in 500	99%	1 in 49,901
92	CITRULLINEMIA, TYPE 1 (AR) 瓜胺酸血症-1型	ASS1	全人種	1 in 119	99%	1 in 11,801
93	CLN10 DISEASE (AR) 神經元蠟樣脂褐質沉著症-CTSD型	CTSD	全人種	< 1 in 500	99%	1 in 49,901
94	COHEN SYNDROME (AR) 科恩症候群	VPS13B	全人種	< 1 in 500	99%	1 in 49,901
95	COL11A2-RELATED CONDITIONS (AR) COL11A2相關疾病	COL11A2	全人種	≤1 in 500	99%	1 in 49,901
96	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

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
97	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
98	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
99	COMBINED PITUITARY HORMONE DEFICIENCY 1 (AR) 結合性腦下垂體賀爾蒙缺失-1型	POU1F1	全人種	≤1 in 500	99%	1 in 49,901
100	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
101	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
102	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
103	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
104	CONGENITAL ADRENAL INSUFFICIENCY, CYP11A1-RELATED (AR) 先天性腎上腺異常症	CYP11A1	全人種	1 in 114	99%	1 in 11,301
105	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

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
106	CONGENITAL CHRONIC DIARRHEA (AR) 先天性腹瀉病-DGAT1相關	DGAT1	全人種	≤1 in 500	99%	1 in 49,901
107	CONGENITAL DISORDER OF GLYCOSYLATION TYPE 1, ALG1-RELATED (AR) 先天性醣基化疾病-ALG1相關	ALG1	全人種	≤1 in 500	99%	1 in 49,901
108	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
109	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
110	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
111	CONGENITAL DYSERYTHROPOIETIC ANEMIA TYPE 2 (AR) 先天性紅血球生成異常性貧血-2型	SEC23B	全人種	≤1 in 500	99%	1 in 49,901
112	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
113	CONGENITAL HYDROCEPHALUS 1 (AR) 先天性腦積水-1型	CCDC88C	全人種	1 in 137	99%	1 in 13,601
114	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
115	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
116	CONGENITAL MYASTHENIC SYNDROME, CHAT-RELATED (AR) 先天性肌無力症候群-CHAT相關	CHAT	全人種	< 1 in 500	99%	1 in 49,901
117	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
118	CONGENITAL MYASTHENIC SYNDROME, DOK7-RELATED (AR) 先天性肌無力症候群-DOK7相關	DOK7	全人種	1 in 454	99%	1 in 45,301
119	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
120	CONGENITAL NEUTROPENIA, G6PC3-RELATED (AR) 先天性嗜中性白血球減少症-G6PC3相關	G6PC3	全人種	≤1 in 500	99%	1 in 49,901
121	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%	1 in 49,901
122	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%	1 in 49,901
123	CONGENITAL SECRETORY CHLORIDE DIARRHEA 1 (AR) 先天性氯化物腹瀉	SLC26A3	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
124	COQ4-RELATED CONDITIONS (AR) COQ4相關疾病	COQ4	全人種	< 1 in 500	99%	1 in 49,901
125	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
126	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
127	COSTEFF SYNDROME (3-METHYLGLUTACONIC ACIDURIA, TYPE 3) (AR) Costeff症候群	OPA3	全人種	< 1 in 500	99%	1 in 49,901
128	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
129	CYSTIC FIBROSIS (AR) 囊腫纖維症	CFTR	全人種	1 in 45	99%	1 in 4,401
130	CYSTINOSIS (AR) 胱胺酸血症	CTNS	全人種	1 in 158	99%	1 in 15,701
131	CYTOCHROME C OXIDASE DEFICIENCY, PET100-RELATED (AR) 細胞色素c氧化酶缺乏症-PET100相關	PET100	全人種	1 in 452	99%	1 in 45,101
132	CYTOCHROME P450 OXIDOREDUCTASE DEFICIENCY (AR) 細胞色素 P450 氧化還原酶缺乏	POR	全人種	1 in 323	99%	1 in 32,201
133	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

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	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
135	DIHYDROPTERIDINE REDUCTASE (DHPR) DEFICIENCY (AR) 雙氫喋啶還原酶缺乏症-QDPR相關	QDPR	全人種	≤1 in 500	99%	1 in 49,901
136	DONNAI-BARROW SYNDROME (AR) Donnai-Barrow症候群	LRP2	全人種	1 in 213	99%	1 in 21,201
137	DUBIN-JOHNSON SYNDROME (AR) Dubin-Johnson症候群	ABCC2	全人種	1 in 158	99%	1 in 15,701
138	DYSKERATOSIS CONGENITA SPECTRUM DISORDERS (AR) 先天性角化不全症-TERT相關	TERT	全人種	≤1 in 500	99%	1 in 49,901
139	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
140	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
141	EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY, CAD-RELATED (AR) 早期嬰兒癲癇性腦病變	CAD	全人種	≤1 in 500	99%	1 in 49,901
142	EHLERS-DANLOS SYNDROME TYPE VI (AR) Ehlers-Danlos症候群-6型	PLOD1	全人種	1 in 158	99%	1 in 15,701
143	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

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	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
145	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
146	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
147	EPIMERASE DEFICIENCY (GALACTOSEMIA TYPE III) (AR) 半乳糖異構酶缺乏症	GALE	全人種	1 in 132	99%	1 in 13,101
148	EPIPHYSEAL DYSPLASIA, MULTIPLE, 7/DESBUQUOIS DYSPLASIA 1 (AR) 第7型多發性骨骺發育不全 / Desbuquois發育不全症-1型	CANT1	全人種	< 1 in 500	99%	1 in 49,901
149	ERCC6-RELATED DISORDERS (AR) ERCC6相關疾病	ERCC6	全人種	< 1 in 500	99%	1 in 49,901
150	ERCC8-RELATED DISORDERS (AR) ERCC8相關疾病	ERCC8	全人種	< 1 in 500	99%	1 in 49,901
151	ETHYLMALONIC ENCEPHALOPATHY (AR) 乙基丙二酸腦病變	ETHE1	全人種	< 1 in 500	99%	1 in 49,901
152	F2-RELATED CONDITIONS (AR) F2相關疾病	F2	全人種	1 in 33	99%	1 in 3,201
153	F5-RELATED CONDITIONS (AR) F5相關疾病	F5	全人種	1 in 12	99%	1 in 1,101

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
154	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
155	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
156	FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, PRF1-RELATED (AR) 家族性噬血細胞性淋巴組織球增生症-PRF1相關	PRF1	全人種	< 1 in 500	99%	1 in 49,901
157	FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STX11-RELATED (AR) 家族性噬血細胞性淋巴組織球增生症-STX11相關	STX11	全人種	1 in 354	99%	1 in 35,301
158	FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, STXBP2-RELATED (AR) 家族性噬血細胞性淋巴組織球增生症-STXBP2相關	STXBP2	全人種	1 in 296	99%	1 in 29,501
159	FAMILIAL HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, UNC13D-RELATED (AR) 家族性噬血細胞性淋巴組織球增生症-UNC13D相關	UNC13D	全人種	< 1 in 500	99%	1 in 49,901
160	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
161	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
162	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
163	FAMILIAL MEDITERRANEAN FEVER (AR) 家族性地中海熱	MEFV	全人種	1 in 115	99%	1 in 11,401
164	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
165	FANCONI ANEMIA, GROUP A (AR) Fanconi氏貧血-A型	FANCA	全人種	1 in 345	99%	1 in 34,401
166	FANCONI ANEMIA, GROUP C (AR) Fanconi氏貧血-C型	FANCC	全人種	1 in 1,053	99%	1 in 105,201
167	FANCONI ANEMIA, GROUP D2 (AR) Fanconi氏貧血-D2型	FANCD2	全人種	< 1 in 500	99%	1 in 49,901
168	FANCONI ANEMIA, GROUP E (AR) Fanconi氏貧血-E型	FANCE	全人種	< 1 in 500	99%	1 in 49,901
169	FANCONI ANEMIA, GROUP G (AR) Fanconi氏貧血-G型	FANCG	全人種	< 1 in 500	99%	1 in 49,901
170	FANCONI ANEMIA, GROUP I (AR) Fanconi氏貧血-I型	FANCI	全人種	< 1 in 500	99%	1 in 49,901
171	FANCONI ANEMIA, GROUP J (AR) Fanconi氏貧血-J型	BRIP1	全人種	≤1 in 500	99%	1 in 49,901
172	FANCONI ANEMIA, GROUP L (AR) Fanconi氏貧血-L型	FANCL	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
173	FOVEAL HYPOPLASIA (AR) 中心凹發育不全症	SLC38A8	全人種	≤1 in 500	99%	1 in 49,901
174	FRASER SYNDROME 3, GRIP1-RELATED (AR) Fraser氏症候-GRIP1型	GRIP1	全人種	1 in 83	99%	1 in 8,201
175	FRASER SYNDROME, FRAS1-RELATED (AR) Fraser症候群-FRAS1型	FRAS1	全人種	1 in 316	99%	1 in 31,500
176	FRASER SYNDROME, FREM2-RELATED (AR) Fraser氏症候-FREM2型	FREM2	全人種	≤1 in 500	99%	1 in 49,901
177	FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY (AR) 果糖-1,6-二磷酸酶缺乏症	FBP1	全人種	≤1 in 500	99%	1 in 49,901
178	FUCOSIDOSIS, FUCA1-RELATED (AR) 岩藻糖代謝異常儲積症	FUCA1	全人種	≤1 in 500	99%	1 in 49,901
179	FUMARASE DEFICIENCY (AR) 延胡索酸酶缺乏症	FH	全人種	< 1 in 500	99%	1 in 49,901
180	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
181	GALACTOSEMIA (AR) 半乳糖血症	GALT	全人種	1 in 110	99%	1 in 10,901
182	GALACTOSIALIDOSIS (AR) 半乳糖唾液酸儲積症	CTSA	全人種	< 1 in 500	99%	1 in 49,901
183	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
184	GCH1-RELATED CONDITIONS (AR) GCH1相關疾病	GCH1	全人種	≤1 in 500	99%	1 in 49,901
185	GDF5-RELATED CONDITIONS (AR) GDF5相關疾病	GDF5	全人種	≤1 in 500	99%	1 in 49,901
186	GERODERMA OSTEODYSPLASTICA (AR) 老年樣皮膚營養不良及骨結構不良	GORAB	全人種	≤1 in 500	99%	1 in 49,901
187	GITELMAN SYNDROME (AR) Gitelman症候群	SLC12A3	全人種	1 in 100	99%	1 in 9,901
188	GLANZMANN THROMBASTHENIA (AR) 血小板無力症	ITGB3	全人種	≤1 in 500	99%	1 in 49,901
189	GLUTARIC ACIDEMIA, TYPE 1 (AR) 戊二酸血症-1型	GCDH	全人種	1 in 112	99%	1 in 11,101
190	GLUTARIC ACIDEMIA, TYPE 2A (AR) 戊二酸血症-2A型	ETFA	全人種	< 1 in 500	99%	1 in 49,901
191	GLUTARIC ACIDEMIA, TYPE 2B (AR) 戊二酸血症-2B型	ETFB	全人種	1 in 408	99%	1 in 40,701
192	GLUTARIC ACIDEMIA, TYPE 2C (AR) 戊二酸血症-2C型	ETFDH	全人種	1 in 250	99%	1 in 24,901
193	GLUTATHIONE SYNTHETASE DEFICIENCY (AR) 谷胱甘肽合成酶缺乏症	GSS	全人種	≤1 in 500	99%	1 in 49,901
194	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

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
195	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
196	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
197	GLYCOGEN STORAGE DISEASE TYPE IXB(AR) 肝醣儲積症-9B型	PHKB	全人種	≤1 in 500	99%	1 in 49,901
198	GLYCOGEN STORAGE DISEASE TYPE IXC(AR) 肝醣儲積症-9C型	PHKG2	全人種	≤1 in 500	99%	1 in 49,901
199	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
200	GLYCOGEN STORAGE DISEASE, TYPE 1b(AR) 肝醣儲積症-1B型	SLC37A4	全人種	1 in 354	99%	1 in 35301
201	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
202	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
203	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
204	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
205	GRACILE SYNDROME (AR) Gracll 症候群	BCS1L	全人種	1 in 111	99%	1 in 11,001

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
206	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
207	HARLEQUIN ICHTHYOSIS (AR) 斑色魚鱗癬	ABCA12	全人種	< 1 in 500	99%	1 in 49,901
208	HEME OXYGENASE 1 DEFICIENCY (AR) 血鐵質氧化酶-1缺乏症	HMOX1	全人種	≤1 in 500	99%	1 in 49,901
209	HEMOCHROMATOSIS TYPE 2A (AR) 血鐵沉積症-2A型	HFE2	全人種	< 1 in 500	99%	1 in 49,901
210	HEMOCHROMATOSIS, TYPE 3, TFR2-Related (AR) 血鐵沉積症-TFR2相關	TFR2	全人種	< 1 in 500	99%	1 in 49,901
211	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
212	HEREDITARY FRUCTOSE INTOLERANCE (AR) 遺傳性果糖不耐症	ALDOB	全人種	1 in 55	99%	1 in 5,401
213	HEREDITARY HEMOCHROMATOSIS TYPE 1 (AR) 血鐵沉積症-1型	HFE	全人種	1 in 10	99%*	1 in 901*
214	HEREDITARY HEMOCHROMATOSIS TYPE 2B (AR) 血鐵沉積症-2B型	HAMP	全人種	≤1 in 500	99%	1 in 49,901
215	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

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
216	HEREDITARY SPASTIC PARAPLEGIA, CYP7B1-RELATED (AR) 痙攣性下身麻痺-CYP7B1相關	CYP7B1	全人種	1 in 338	99%	1 in 33,701
217	HERMANSKY-PUDLAK SYNDROME, BLOC1S3-RELATED (AR) Hermansky-Pudlak 症候群-BLOC1S3相關	BLOC1S3	全人種	≤1 in 500	99%	1 in 49,901
218	HERMANSKY-PUDLAK SYNDROME, BLOC1S6-RELATED (AR) Hermansky-Pudlak 症候群-BLOC1S6相關	BLOC1S6	全人種	≤1 in 500	99%	1 in 49,901
219	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
220	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
221	HERMANSKY-PUDLAK SYNDROME, HPS4-RELATED (AR) Hermansky-Pudlak 症候群-HPS4相關	HPS4	全人種	< 1 in 500	99%	1 in 49,901
222	HERMANSKY-PUDLAK SYNDROME, HPS5-RELATED (AR) Hermansky-Pudlak 症候群-HPS5相關	HPS5	全人種	≤1 in 500	99%	1 in 49,901
223	HERMANSKY-PUDLAK SYNDROME, HPS6-RELATED (AR) Hermansky-Pudlak 症候群-HPS6相關	HPS6	全人種	≤1 in 500	99%	1 in 49,901
224	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

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
225	HOMOCYSTINURIA AND MEGALOBLASTIC ANEMIA TYPE CBLG (AR) 甲基鈷胺素缺乏症cblG型	MTR	全人種	≤1 in 500	99%	1 in 49,901
226	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
227	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
228	HOMOCYSTINURIA, Type cblE (AR) 高胱胺酸尿症- cblE型	MTRR	全人種	< 1 in 500	99%	1 in 49,901
229	HYDROLETHALUS SYNDROME (AR) Hydrolethalus症候群	HYLS1	全人種	1 in 455	99%	1 in 45,401
230	HYPER-IGM IMMUNODEFICIENCY (AR) 高免疫球蛋白M症候群	CD40	全人種	≤1 in 500	99%	1 in 49,901
231	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
232	HYPERPHOSPHATEMIC FAMILIAL TUMORAL CALCINOSIS, GALNT3-RELATED (AR) 高磷血症家族性腫瘤性鈣質沉著症	GALNT3	全人種	< 1 in 500	99%	1 in 49,901
233	HYPOMYELINATING LEUKODYSTROPHY 12 低髓鞘腦白質失養症-12型	VPS11	全人種	≤1 in 500	99%	1 in 49,901
234	HYPOPHOSPHATASIA,ALPL-RELATED (AR) 低磷酸酯酶症-ALPL相關	ALPL	全人種	1 in 158	99%	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
235	IMERSLUND-GRASBECK SYNDROME 2 (AR) Imerslund-Grsbeck症候群 -2型	AMN	全人種	≤1 in 500	99%	1 in 49,901
236	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES (ICF) SYNDROME, DNMT3B-RELATED (AR) ICF症候群-DNMT3B相關	DNMT3B	全人種	≤1 in 500	99%	1 in 49,901
237	IMMUNODEFICIENCY-CENTROMERIC INSTABILITY-FACIAL ANOMALIES (ICF) SYNDROME, ZBTB24-RELATED (AR) ICF症候群-ZBTB24相關	ZBTB24	全人種	≤1 in 500	99%	1 in 49,901
238	INCLUSION BODY MYOPATHY 2 (AR) 包涵體肌炎	GNE	全人種	< 1 in 500	99%	1 in 49,901
239	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
240	INFANTILE NEPHRONOPHTHISIS (AR) 腎消耗病-INVS相關	INVS	全人種	1 in 373	99%	1 in 37,201
241	INFANTILE NEUROAXONAL DYSTROPHY (AR) 新生兒神經軸發育不良-PLA2G6相關	PLA2G6	全人種	< 1 in 500	99%	1 in 49,901
242	ISOLATED ECTOPIA LENTIS (AR) 孤立性水晶體異味症	ADAMTSL4	全人種	≤1 in 500	99%	1 in 49,901
243	ISOLATED SULFITE OXIDASE DEFICIENCY (AR) 亞硫酸鹽氧化酵素缺乏症	SUOX	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
244	ISOLATED THYROID-STIMULATING HORMONE DEFICIENCY (AR) 先天性甲狀腺低功能症-TSHB相關	TSHB	全人種	≤1 in 500	99%	1 in 49,901
245	ISOVALERIC ACIDEMIA (AR) 異戊酸血症	IVD	全人種	1 in 250	99%	1 in 24,901
246	JOHANSON-BLIZZARD SYNDROME (AR) Johanson-Blizzard症候群	UBR1	全人種	1 in 250	99%	1 in 24,901
247	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
248	JOUBERT SYNDROME AND RELATED DISORDERS (JSRD), TMEM67-RELATED (AR) Joubert 症候群-TMEM67相關	TMEM67	全人種	1 in 216	99%	1 in 21,501
249	JOUBERT SYNDROME, AHI1-RELATED (AR) Joubert氏症候群-AHI-相關	AHI1	全人種	1 in 176	99%	1 in 17,501
250	JOUBERT SYNDROME, CC2D2A-RELATED/COACH SYNDROME (AR) Joubert 症候群-CC2D2A相關/Coach症候群	CC2D2A	全人種	< 1 in 500	99%	1 in 49,901
251	JUNCTIONAL EPIDERMOLYSIS BULLOSA, COL17A1-RELATED (AR) 接合性表皮溶解水皰症-COL17A1相關	COL17A1	全人種	≤1 in 500	99%	1 in 49,901
252	JUNCTIONAL EPIDERMOLYSIS BULLOSA, ITGA6-RELATED (AR) 接合性表皮溶解水皰症-ITGA6相關	ITGA6	全人種	1 in 159	99%	1 in 15,801
253	JUNCTIONAL EPIDERMOLYSIS BULLOSA, ITGB4-RELATED (AR) 接合性表皮溶解水皰症-ITGB4相關	ITGB4	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
254	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
255	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
256	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
257	KRABBE DISEASE (AR) Krabbe氏症	GALC	全人種	1 in 500	99%	1 in 49,901
258	LAMELLAR ICHTHYOSIS, TYPE 1 (AR) 層狀魚鱗癬-1型	TGM1	全人種	1 in 301	99%	1 in 30,001
259	LARON SYNDROME (AR) Laron 症候群	GHR	全人種	1 in 167	99%	1 in 16,601
260	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
261	LEBER CONGENITAL AMAUROSIS TYPE AIPL1 (AR) 萊伯氏先天性黑矇症-AIPL1型	AIPL1	全人種	1 in 387	99%	1 in 38,601
262	LEBER CONGENITAL AMAUROSIS TYPE GUCY2D (AR) 萊伯氏先天性黑矇症-GUCY2D型	GUCY2D	全人種	1 in 200	99%	1 in 19,901
263	LEBER CONGENITAL AMAUROSIS TYPE TULP1 (AR) 萊伯氏先天性黑矇症-TULP1型	TULP1	全人種	1 in 186	99%	1 in 18,501

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
264	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
265	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
266	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
267	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
268	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
269	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
270	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B1-RELATED (AR) 腦白質病伴隨白質消失症-EIF2B1相關	EIF2B1	全人種	≤1 in 500	99%	1 in 49,901
271	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B2-RELATED (AR) 腦白質病伴隨白質消失症-EIF2B2相關	EIF2B2	全人種	≤1 in 500	99%	1 in 49,901
272	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B3-RELATED (AR) 腦白質病伴隨白質消失症-EIF2B3相關	EIF2B3	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
273	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER, EIF2B4-RELATED (AR) 腦白質病伴隨白質消失症-EIF2B4型	EIF2B4	全人種	≤1 in 500	99%	1 in 49,901
274	LIG4 SYNDROME (AR) LIG4症候群	LIG4	全人種	≤1 in 500	99%	1 in 49,901
275	LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 8 (AR) 肢帶型肌肉失養症-8型	TRIM32	全人種	<1 in 500	99%	1 in 49,901
276	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
277	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
278	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
279	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
280	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
281	LIMB-GIRDLE MUSCULAR DYSTROPHY, TYPE 2F (AR) 肢帶型肌肉失養症-2F 型	SGCD	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
282	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
283	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
284	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
285	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
286	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
287	LRAT-RELATED CONDITIONS (AR) LRAT相關疾病	LRAT	全人種	<1 in 500	99%	1 in 49,901
288	LUNG DISEASE, IMMUNODEFICIENCY, AND CHROMOSOME BREAKAGE SYNDROME (LICS) (AR) LICS 症候群	NSMCE3	全人種	≤1 in 500	99%	1 in 49,901
289	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
290	MALONYL-COA DECARBOXYLASE DEFICIENCY (AR) 丙二酰輔酶 A 脫羧酶缺乏症	MLYCD	全人種	<1 in 500	99%	1 in 49,901
291	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

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
292	MAPLE SYRUP URINE DISEASE, TYPE 1B (AR) 楓糖尿症-1B型	BCKDHB	全人種	1 in 346	99%	1 in 36,301
293	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
294	MCKUSICK-KAUFMAN SYNDROME (AR) McKusick-Kaufman症候群	MKKS	全人種	1 in 219	99%	1 in 21,801
295	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
296	MECR-RELATED NEUROLOGIC DISORDER (AR) MECR 基因相關神經系統疾病	MECR	全人種	≤1 in 500	99%	1 in 49,901
297	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
298	MEDNIK SYNDROME (AR) MEDNIK 症候群	AP1S1	全人種	<1 in 500	99%	1 in 49,901
299	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
300	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

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
301	METABOLIC ENCEPHALOPATHY AND ARRHYTHMIAS, TANGO2-RELATED (AR) 橫紋肌溶解、心律不整、神經病變代謝異常	TANGO2	全人種	<1 in 500	99%	1 in 49,901
302	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
303	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
304	METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLF (AR) 甲基丙二酸血症併高胱胺酸血 症-cblF型	LMBRD1	全人種	≤1 in 500	99%	1 in 49,901
305	METHYLMALONIC ACIDEMIA, MCEE-RELATED (AR) 甲基丙二酸血症-MCEE相關	MCEE	全人種	≤1 in 500	99%	1 in 49,901
306	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, TYPE CBLC (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
307	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
308	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
309	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

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
310	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
311	MEVALONIC KINASE DEFICIENCY (AR) 高免疫球蛋白血症D症候群	MVK	全人種	1 in 167	99%	1 in 16,601
312	MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II (AR) 原始侏儒症II型	PCNT	全人種	≤1 in 500	99%	1 in 49,901
313	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
314	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
315	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
316	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
317	MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 1 (AR) 粒線體複合物 I缺乏症-NDUFS4相關	NDUFS4	全人種	1 in 423	99%	1 in 42,201
318	MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 10 (AR) 粒線體複合物 I缺乏症-NDUFAF2相關	NDUFAF2	全人種	<1 in 500	99%	1 in 49,901

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319	MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 19 (AR) 粒線體複合物 I缺乏症-FOXRED1相關	FOXRED1	全人種	<1 in 500	99%	1 in 49,901
320	MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 3 (AR) 粒線體複合物 I缺乏症-NDUFS7相關	NDUFS7	全人種	<1 in 500	99%	1 in 49,901
321	MITOCHONDRIAL COMPLEX I DEFICIENCY, NUCLEAR TYPE 4 (AR) 粒線體複合物 I缺乏症-NDUFV1相關	NDUFV1	全人種	<1 in 500	99%	1 in 49,901
322	MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 2, SCO2-RELATED (AR) 粒線體複合物IV缺乏症-SCO2相關	SCO2	全人種	1 in 149	99%	1 in 14,801
323	MITOCHONDRIAL COMPLEX IV DEFICIENCY, NUCLEAR TYPE 6 (AR) 粒線體複合物 IV缺乏症-COX15相關	COX15	全人種	≤1 in 500	99%	1 in 49,901
324	MITOCHONDRIAL DNA DEPLETION SYNDROME 2 (AR) 粒線體DNA耗竭症候群-2型	TK2	全人種	≤1 in 500	99%	1 in 49,901
325	MITOCHONDRIAL DNA DEPLETION SYNDROME 3 (AR) 粒線體DNA耗竭症候群-3型	DGUOK	全人種	≤1 in 500	99%	1 in 49,901
326	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
327	MITOCHONDRIAL TRIFUNCTIONAL PROTEIN DEFICIENCY, HADHB-RELATED (AR) 粒線體三功能蛋白缺失症-HADHB相關	HADHB	全人種	1 in 146	99%	1 in 14,501

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
328	MOLYBDENUM COFACTOR DEFICIENCY TYPE B (AR) 鉬輔酶缺乏症 B 型	MOCS2	全人種	1 in 236	99%	1 in 23,501
329	MOLYBDENUM COFACTOR DEFICIENCY, TYPE A 鉬輔酶缺乏症 A 型	MOCS1	全人種	<1 in 500	99%	1 in 49,901
330	MUCOLIPIDOSIS II/III A (AR) 黏脂質症 -2/3型	GNPTAB	全人種	1 in 158	99%	1 in 15,701
331	MUCOLIPIDOSIS III GAMMA (AR) 黏脂症 III γ 型	GNPTG	全人種	<1 in 500	99%	1 in 49,901
332	MUCOLIPIDOSIS, TYPE IV (AR) 黏脂質症-4型	MCOLN1	全人種	<1 in 500	99%	1 in 49,901
333	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
334	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
335	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
336	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
337	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

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
338	MUCOPOLYSACCHARIDOSIS, TYPE IV A (MORQUIO SYNDROME) (AR) 黏多醣症-4A型 (Morquio 症候群)	GALNS	全人種	1 in 307	99%	1 in 30,601
339	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
340	MUCOPOLYSACCHARIDOSIS, TYPE IX (AR) 黏多醣症-9型	HYAL1	全人種	<1 in 500	99%	1 in 49,901
341	MUCOPOLYSACCHARIDOSIS, TYPE VI (MAROTEAUX-LAMY) (AR) 黏多醣症-6型	ARSB	全人種	1 in 291	99%	1 in 29,001
342	MUCOPOLYSACCHARIDOSIS, TYPE VII (AR) 黏多醣症-7型	GUSB	全人種	<1 in 500	99%	1 in 49,901
343	MULIBREY NANISM (AR) MULIBREY侏儒症	TRIM37	全人種	<1 in 500	99%	1 in 49,901
344	MULTIPLE PTERYGIUM SYNDROME, CHRNG-RELATED/ESCOBAR SYNDROME(AR) 多發性翼狀膜症候群	CHRNG	全人種	1 in 50	99%	1 in 4,901
345	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
346	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
347	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (AR) 肌肉失養症醣基化功能缺陷-RXYLT1相關	RXYLT1	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
348	MUSK-RELATED CONGENITAL MYASTHENIC SYNDROME (AR) 先天性肌無力症候群-MUSK型	MUSK	全人種	<1 in 500	99%	1 in 49,901
349	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
350	MYOTONIA CONGENITA (AR) 先天性肌強直症	CLCN1	全人種	1 in 158	99%	1 in 15,701
351	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
352	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
353	NEPHRONOPHTHISIS 1 (AR) 腎消耗病-NPHP1型	NPHP1	全人種	1 in 202	99%	1 in 20,101
354	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
355	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
356	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
357	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

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
358	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
359	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
360	NGLY1-CONGENITAL DISORDER OF GLYCOSYLATION (AR) 先天性醣基化疾病-NGLY1型	NGLY1	全人種	1 in 274	99%	1 in 27,301
361	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
362	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
363	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
364	NIJMEGEN BREAKAGE SYNDROME (AR) Nijmegen破損症候群	NBN	全人種	<1 in 500	99%	1 in 49,901
365	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
366	NON-SYNDROMIC HEARING LOSS, MYO15A-RELATED (AR) 非症候群感覺神經性聽損-MYO15A相關	MYO15A	全人種	1 in 117	99%	1 in 11,601
367	NONSYNDROMIC HEARING LOSS, OTOA-RELATED (AR) 非症候群感覺神經性聽損-OTOA相關	OTOA	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
368	NONSYNDROMIC HEARING LOSS, OTOF-RELATED (AR) 非症候群感覺神經性聽損-OTOF相關	OTOF	全人種	1 in 96	99%	1 in 9,501
369	NONSYNDROMIC HEARING LOSS, PJVK-RELATED (AR) 非症候群感覺神經性聽損-PJVK相關	PJVK	全人種	≤1 in 500	99%	1 in 49,901
370	NONSYNDROMIC HEARING LOSS, SYNE4-RELATED (AR) 非症候群感覺神經性聽損-SYNE4相關	SYNE4	全人種	≤1 in 500	99%	1 in 49,901
371	NONSYNDROMIC HEARING LOSS, TMC1-RELATED (AR) 非症候群感覺神經性聽損-TMC1相關	TMC1	全人種	1 in 96	99%	1 in 9,501
372	NONSYNDROMIC HEARING LOSS, TMPRSS3-RELATED (AR) 非症候群感覺神經性聽損-TMPRSS3相關	TMPRSS3	全人種	1 in 96	99%	1 in 9,501
373	NONSYNDROMIC INTELLECTUAL DISABILITY (AR) 非綜合徵型智力障礙-CC2D1A型	CC2D1A	全人種	≤1 in 500	99%	1 in 49,901
374	NORMOPHOSPHATEMIC TUMORAL CALCINOSIS (AR) 正常血磷性家族性腫瘤性鈣化症	SAMD9	全人種	≤1 in 500	99%	1 in 49,901
375	OCULOCUTANEOUS ALBINISM TYPE III (AR) 眼睛皮膚白化症第3型	TYRP1	全人種	1 in 488	99%	1 in 48,701
376	OCULOCUTANEOUS ALBINISM TYPE IV (AR) 眼睛皮膚白化症第4型	SLC45A2	全人種	1 in 158	99%	1 in 15,700

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
377	OCULOCUTANEOUS ALBINISM, OCA2-RELATED (AR) 眼睛皮膚白化症-OCA2相關	OCA2	全人種	1 in 76	99%	1 in 7,501
378	OCULOCUTANEOUS ALBINISM, TYPES 1A AND 1B (AR) 眼睛皮膚白化症-TYR相關	TYR	全人種	1 in 20	97%	1 in 1,901
379	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
380	OMENN SYNDROME, RAG2-RELATED (AR) Omenn 症候群-RAG2相關	RAG2	全人種	≤1 in 500	99%	1 in 49,901
381	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
382	OSTEOGENESIS IMPERFECTA TYPE VII (AR) 骨質形成不全症-7型	CRTAP	全人種	≤1 in 500	99%	1 in 49,901
383	OSTEOGENESIS IMPERFECTA TYPE VIII (AR) 骨質形成不全症-8型	P3H1	全人種	≤1 in 500	99%	1 in 49,901
384	OSTEOGENESIS IMPERFECTA TYPE XI (AR) 骨質形成不全症-11型	FKBP10	全人種	≤1 in 500	99%	1 in 49,901
385	OSTEOGENESIS IMPERFECTA TYPE XIII (AR) 骨質形成不全症-13型	BMP1	全人種	≤1 in 500	99%	1 in 49,901
386	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
387	OSTEOPETROSIS, OSTM1-RELATED (AR) 骨質石化症-OSTM1相關	OSTM1	全人種	≤1 in 500	99%	1 in 49,901
388	PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION (AR) 泛酸鹽激活酵素關聯之神經退化性疾	PANK2	全人種	1 in 289	99%	1 in 28,800
389	PAPILLON LEFEVRE SYNDROME (AR) CTSC相關疾病	CTSC	全人種	1 in 250	99%	1 in 24,900
390	PARKINSON DISEASE 15 (AR) 早發性巴金森氏症 15 型	FBXO7	全人種	≤1 in 500	99%	1 in 49,901
391	PENDRED SYNDROME (AR) Pendred 氏症候群	SLC26A4	全人種	1 in 80	99%	1 in 7,901
392	PGM3-CONGENITAL DISORDER OF GLYCOSYLATION (AR) PGM3-先天性醣基化障礙	PGM3	全人種	≤1 in 500	99%	1 in 49,901
393	PHENYLKETONURIA (AR) 苯酮尿症	PAH	全人種	1 in 65	99%	1 in 6,401
394	PIGN-CONGENITAL DISORDER OF GLYCOSYLATION (AR) PIGN-先天性醣基化障礙	PIGN	全人種	≤1 in 500	99%	1 in 49,901
395	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
396	POLG-RELATED DISORDERS (AR) POLG相關疾病	POLG	全人種	1 in 50	99%	1 in 4,901

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
397	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE (AR) 隱性多囊腎病	PKHD1	全人種	1 in 144	99%	1 in 14,301
398	PONTOCEREBELLAR HYPOPLASIA, EXOSC3-RELATED (AR) 橋腦小腦發育不全-1B型	EXOSC3	全人種	< 1 in 500	99%	1 in 49,901
399	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
400	PONTOCEREBELLAR HYPOPLASIA, TSEN54-RELATED (AR) 橋腦小腦發育不全-TSEN54相關	TSEN54	全人種	< 1 in 500	99%	1 in 49,901
401	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
402	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
403	PONTOCEREBELLAR HYPOPLASIA, VPS53-RELATED (AR) 橋腦小腦發育不全-VPS53相關	VPS53	全人種	< 1 in 500	99%	1 in 49,901
404	PRIMARY CILIARY DYSKINESIA, CCDC103-RELATED (AR) 原發性纖毛運動障礙-CCDC103相關	CCDC103	全人種	1 in 217	99%	1 in 21,601
405	PRIMARY CILIARY DYSKINESIA, CCDC39-RELATED (AR) 原發性纖毛運動障礙-CCDC39相關	CCDC39	全人種	1 in 144	99%	1 in 14,301

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
406	PRIMARY CILIARY DYSKINESIA, DNAH11-RELATED (AR) 原發性纖毛運動障礙-DNAH11相關	DNAH11	全人種	1 in 144	99%	1 in 14,301
407	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
408	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
409	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
410	PRIMARY CONGENITAL GLAUCOMA/PETERS ANOMALY (AR) 原發先天性青光眼-CYP1B1型	CYP1B1	全人種	1 in 74	99%	1 in 7,301
411	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
412	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
413	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
414	PRIMARY MICROCEPHALY 1, AUTOSOMAL RECESSIVE (AR) 原發性小頭畸形症第1型	MCPH1	全人種	1 in 146	99%	1 in 14,501
415	PROGRESSIVE EARLY-ONSET ENCEPHALOPATHY WITH BRAIN ATROPHY AND THIN CORPUS CALLOSUM (AR) 早發進行性腦病變伴隨腦萎縮與薄胼胝體	TBCD	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
416	PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, ABCB4-RELATED (AR) 進行性家族性肝內膽汁滯留症-ABCB4相關	ABCB4	全人種	1 in 194	99%	1 in 19,301
417	PROGRESSIVE FAMILIAL INTRAHEPATIC CHOLESTASIS, TYPE 1 (PFIC1) (AR) 進行性家族性肝內膽汁滯留症第一型	ATP8B1	全人種	1 in 53	99%	1 in 5,201
418	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
419	PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA (AR) 進行性假性類風濕發育不良症候群	CCN6	全人種	≤1 in 500	99%	1 in 49,901
420	PROLIDASE DEFICIENCY (AR) 脯氨酸肽酶缺乏症	PEPD	全人種	1 in 242	99%	1 in 24,101
421	PROPIONIC ACIDEMIA, PCCA-RELATED (AR) 丙酸血症-PCCA相關	PCCA	全人種	1 in 224	99%	1 in 22,301
422	PROPIONIC ACIDEMIA, PCCB-RELATED (AR) 丙酸血症-PCCB相關	PCCB	全人種	1 in 224	99%	1 in 22,301
423	PTERIN-4 ALPHA-CARBINOLAMINE DEHYDRATASE (PCD) DEFICIENCY (AR) 四氫基喋呤缺乏症-PCBD1型	PCBD1	全人種	≤1 in 500	99%	1 in 49,901
424	PYCNODYSOSTOSIS (AR) 緻密性成骨不全症	CTSK	全人種	1 in 439	99%	1 in 43,801
425	PYRIDOXAL 5'-PHOSPHATE-DEPENDENT EPILEPSY (AR) 磷酸吡哆醇依賴性癲癇	PNPO	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
426	PYRIDOXINE-DEPENDENT EPILEPSY (AR) 維生素B6依賴性癲癇	ALDH7A1	全人種	1 in 158	99%	1 in 15,701
427	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
428	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
429	REFSUM DISEASE, PHYH-RELATED (AR) 雷弗素姆病-PHYH相關	PHYH	全人種	< 1 in 500	99%	1 in 49,901
430	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
431	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
432	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
433	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
434	RETINITIS PIGMENTOSA 36 (AR) 視網膜色素病變36型	PRCD	全人種	< 1 in 500	99%	1 in 49,901
435	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
436	RETINITIS PIGMENTOSA 62 (AR) 視網膜色素病變62型	MAK	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
437	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
438	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2 (AR) 肢近端型點狀軟骨發育不良-2型	GNPAT	全人種	< 1 in 500	99%	1 in 49,901
439	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
440	RLBP1-RELATED RETINOPATHY (AR) RLBP1相關疾病	RLBP1	全人種	1 in 296	99%	1 in 29,500
441	ROBERTS SYNDROME (AR) Roberts症候群	ESCO2	全人種	< 1 in 500	99%	1 in 49,901
442	RYR1-RELATED CONDITIONS (AR) RYR1相關疾病	RYR1	全人種	1 in 150	99%	1 in 14,901
443	SALLA DISEASE (AR) 薩拉氏症	SLC17A5	全人種	< 1 in 500	99%	1 in 49,901
444	SANDHOFF DISEASE (AR) Sandoff症	HEXB	全人種	1 in 278	99%	1 in 27,701
445	SCHIMKE IMMUNOOSSEOUS DYSPLASIA (AR) Schimke免疫-骨發育不良	SMARCAL1	全人種	< 1 in 500	99%	1 in 49,901
446	SCHINDLER DISEASE (AR) Schindler症	NAGA	全人種	1 in 94	99%	1 in 9,301
447	SEGAWA SYNDROME, TH-RELATED (AR) Segawa 症候群-TH相關	TH	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
448	SEPIAPTERIN REDUCTASE DEFICIENCY (AR) 墨蝶呤還原酶缺乏症	SPR	全人種	≤1 in 500	99%	1 in 49,901
449	SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3D-RELATED (AR) 嚴重複合型免疫缺乏症-CD3D相關	CD3D	全人種	≤1 in 500	99%	1 in 49,901
450	SEVERE COMBINED IMMUNODEFICIENCY (SCID), CD3E-RELATED (AR) 嚴重複合型免疫缺乏症-CD3E相關	CD3E	全人種	≤1 in 500	99%	1 in 49,901
451	SEVERE COMBINED IMMUNODEFICIENCY (SCID), FOXN1-RELATED (AR) 嚴重複合型免疫缺乏症-FOXN1相關	FOXN1	全人種	≤1 in 500	99%	1 in 49,901
452	SEVERE COMBINED IMMUNODEFICIENCY (SCID), IKBKB-RELATED (AR) 嚴重複合型免疫缺乏症-IKBKB相關	IKBKB	全人種	≤1 in 500	99%	1 in 49,901
453	SEVERE COMBINED IMMUNODEFICIENCY (SCID), IL7R-RELATED (AR) 嚴重複合型免疫缺乏症-IL7R相關	IL7R	全人種	1 in 388	99%	1 in 38,701
454	SEVERE COMBINED IMMUNODEFICIENCY (SCID), JAK3-RELATED (AR) 嚴重複合型免疫缺乏症-JAK3相關	JAK3	全人種	1 in 299	99%	1 in 29,801
455	SEVERE COMBINED IMMUNODEFICIENCY (SCID), PTPRC-RELATED (AR) 嚴重複合型免疫缺乏症-PTPRC相關	PTPRC	全人種	≤1 in 500	99%	1 in 49,901
456	SEVERE COMBINED IMMUNODEFICIENCY (SCID), RAG1-RELATED (AR) 嚴重複合性免疫缺乏症-RAG1相關	RAG1	全人種	1 in 245	99%	1 in 24,401
457	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
458	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
459	SHORT-RIB THORACIC DYSPLASIA 3 WITH OR WITHOUT POLYDACTYLY (AR) 短肋胸廓發育不良-3型	DYNC2H1	全人種	1 in 67	99%	1 in 6,601
460	SIALIDOSIS (AR) 涎酸酵素缺乏症	NEU1	全人種	< 1 in 500	99%	1 in 49,901
461	SJOGREN-LARSSON SYNDROME (AR) Sjogren-Larsson症候群	ALDH3A2	全人種	1 in 223	99%	1 in 22,201
462	SMITH-LEMLI-OPITZ SYNDROME (AR) Smith-Lemli-Opitz 症候群	DHCR7	全人種	1 in 100	99%	1 in 9,901
463	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
464	SPASTIC TETRAPLEGIA, THIN CORPUS CALLOSUM, AND PROGRESSIVE MICROCEPHALY (SPATCCM) (AR) 進行性小頭畸形與薄胼胝體伴隨痙攣性四肢癱瘓	SLC1A4	全人種	< 1 in 500	99%	1 in 49,901
465	SPG11-RELATED CONDITIONS (AR) SPG11相關疾病	SPG11	全人種	1 in 141	99%	1 in 14,000
466	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)

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
467	SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS TYPE 1 (AR) 脊髓性肌肉萎縮症伴呼吸窘迫第一型	IGHMBP2	全人種	≤1 in 500	99%	1 in 49,901
468	SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE 10 (AR) 脊髓小腦共濟失調症-ANO10型	ANO10	全人種	1 in 93	99%	1 in 9,201
469	SPONDYLOCOSTAL DYSOSTOSIS 1 (AR) 脊椎肋骨發育不全-DLL3相關	DLL3	全人種	1 in 289	99%	1 in 28,801
470	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
471	STEEL SYNDROME (AR) Steel 症候群	COL27A1	全人種	<1 in 500	99%	1 in 49,901
472	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
473	STUVE-WIEDEMANN SYNDROME (AR) Stuve-Wiedemann 症候群	LIFR	全人種	<1 in 500	99%	1 in 49,901
474	SURF1-RELATED CONDITIONS (AR) SURF1相關疾病	SURF1	全人種	1 in 316	99%	1 in 31,501
475	SURFACTANT DYSFUNCTION, ABCA3 - RELATED (AR) ABCA3相關疾病	ABCA3	全人種	1 in 116	99%	1 in 11,501
476	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
477	TBCE-RELATED CONDITIONS (AR) TBCE相關疾病	TBCE	全人種	≤1 in 500	99%	1 in 49,901
478	THIAMINE-RESPONSIVE MEGALOBLASTIC ANEMIA SYNDROME (AR) 維生素B1代謝功能障礙症候群	SLC19A2	全人種	≤1 in 500	99%	1 in 49,901
479	THYROID DYSHORMONOGENESIS 1 (AR) 甲狀腺素合成異常-SLC5A5相關	SLC5A5	全人種	≤1 in 500	99%	1 in 49,901
480	THYROID DYSHORMONOGENESIS 2A (AR) 甲狀腺素合成異常-TPO相關	TPO	全人種	1 in 227	99%	1 in 22,601
481	THYROID DYSHORMONOGENESIS 3 (AR) 甲狀腺素合成異常-TG相關	TG	全人種	1 in 217	99%	1 in 21,601
482	THYROID DYSHORMONOGENESIS 6 (AR) 甲狀腺素合成異常-DUOX2相關	DUOX2	全人種	1 in 55	95%	1 in 1,081
483	TRANSCOBALAMIN II DEFICIENCY (AR) 轉鈷胺素II缺乏症	TCN2	全人種	≤1 in 500	99%	1 in 49,901
484	TRICHOHEPATOENTERIC SYNDROME, SKIC2-RELATED (AR) 髮-肝-腸症候群-SKIC2相關	SKIC2	全人種	≤1 in 500	99%	1 in 49,901
485	TRICHOHEPATOENTERIC SYNDROME, TTC37-RELATED (AR) 髮-肝-腸症候群-TTC37相關	TTC37	全人種	1 in 381	99%	1 in 38,001
486	TRICHOTHIODYSTROPHY 1/ XERODERMA PIGMENTOSUM, GROUP D (AR) 毛髮低硫營養不良症第-1型/ 著色性乾皮症-D型	ERCC2	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
487	TRIMETHYLAMINURIA (AR) 臭魚症	FMO3	全人種	1 in 139	99%	1 in 13,801
488	TRIPLE A SYNDROME (AR) Triple A 症候群	AAAS	全人種	<1 in 500	99%	1 in 49,901
489	TSHR-RELATED CONDITIONS (AR) TSHR相關疾病	TSHR	全人種	1 in 322	99%	1 in 32,101
490	TYROSINEMIA TYPE III (AR) 酪胺酸血症-3型	HPD	全人種	≤1 in 500	99%	1 in 49,901
491	TYROSINEMIA, TYPE 1 (AR) 酪胺酸血症-1型	FAH	全人種	1 in 100	99%	1 in 9,901
492	TYROSINEMIA, TYPE 2 (AR) 酪胺酸血症-2型	TAT	全人種	<1 in 500	99%	1 in 49,901
493	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
494	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
495	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
496	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
497	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

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
498	USHER SYNDROME, TYPE 2C (AR) 尤塞氏症候群- 2C 型	ADGRV1	全人種	1 in 176	99%	1 in 17,501
499	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
500	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
501	VICI SYNDROME (AR) Vici 症候群	EPG5	全人種	≤1 in 500	99%	1 in 49,901
502	VITAMIN D-DEPENDENT RICKETS, TYPE 1A (AR) 維生素 D 依賴型佝僂症 -1A型	CYP27B1	全人種	1 in 22	99%	1 in 2,101
503	VITAMIN D-RESISTANT RICKETS TYPE 2A (AR) 維生素 D 依賴型佝僂症 -2A型	VDR	全人種	≤1 in 500	99%	1 in 49,901
504	VLDLR-ASSOCIATED CEREBELLAR HYPOPLASIA (AR) 小腦性共濟失調、智力低下、平衡不良症候群	VLDLR	全人種	≤1 in 500	99%	1 in 49,901
505	WALKER-WARBURG SYNDROME, FKTN-RELATED (AR) Walker-Warburg 綜合症-FKTNV相關	FKTN	全人種	≤1 in 500	99%	1 in 49,901
506	WALKER-WARBURG SYNDROME, LARGE1-RELATED (AR) Walker-Warburg 綜合症-LARGE1相關	LARGE1	全人種	< 1 in 500	99%	1 in 49,901
507	WALKER-WARBURG SYNDROME, POMT1-RELATED (AR) Walker-Warburg 綜合症-POMT1相關	POMT1	全人種	1 in 275	99%	1 in 27,401

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
508	WALKER-WARBURG SYNDROME, POMT2-RELATED (AR) Walker-Warburg 綜合症-POMT2相關	POMT2	全人種	1 in 465	99%	1 in 46,401
509	WARSAW BREAKAGE SYNDROME (AR) Warsaw症候群	DDX11	全人種	≤1 in 500	99%	1 in 49,901
510	WERNER SYNDROME (AR) Werner症候群	WRN	全人種	1 in 224	99%	1 in 22,301
511	WILSON DISEASE (AR) 威爾森氏症	ATP7B	全人種	1 in 90	99%	1 in 8,901
512	WOLCOTT-RALLISON SYNDROME (AR) Wolcott-Rallison 症候群	EIF2AK3	全人種	<1 in 500	99%	1 in 49,901
513	WOLMAN DISEASE (AR) 伍爾曼氏症	LIPA	全人種	<1 in 500	99%	1 in 49,901
514	WOODHOUSE-SAKATI SYNDROME (AR) Woodhouse-Sakati 症候群	DCAF17	全人種	≤1 in 500	99%	1 in 49,901
515	XERODERMA PIGMENTOSUM VARIANT TYPE (AR) 著色性乾皮症-POLH型	POLH	全人種	≤1 in 500	99%	1 in 49,901
516	XERODERMA PIGMENTOSUM, GROUP A (AR) 著色性乾皮症-A型	XPA	全人種	<1 in 500	99%	1 in 49,901
517	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

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
518	ZELLWEGER SPECTRUM DISORDER, PEX13-RELATED (AR) 柴爾維格氏症候群-PEX13相關	PEX13	全人種	≤1 in 500	99%	1 in 49,901
519	ZELLWEGER SPECTRUM DISORDER, PEX16-RELATED (AR) 柴爾維格氏症候群-PEX16相關	PEX16	全人種	≤1 in 500	99%	1 in 49,901
520	ZELLWEGER SPECTRUM DISORDER, PEX5-RELATED (AR) 柴爾維格氏症候群-PEX5相關	PEX5	PEX5	1 in 408	99%	1 in 40,701
521	ZELLWEGER SPECTRUM DISORDERS, PEX10-RELATED (AR) 柴爾維格氏症候群-PEX10相關	PEX10	全人種	< 1 in 500	94%	1 in 49,901
522	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
523	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
524	ZELLWEGER SPECTRUM DISORDERS, PEX26-RELATED (AR) 柴爾維格氏症候群-PEX26相關	PEX26	全人種	< 1 in 500	99%	1 in 49,901
525	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
526	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
527	ADRENAL HYPOPLASIA CONGENITA, X - LINKED (XL) 先天性腎上腺增生症-NR0B1型	NR0B1	全人種	1 in 52,500	99%	1 in 5,249,901
528	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
529	AGAMMAGLOBULINEMIA, X - LINKED (XL) 布魯頓氏低免疫球蛋白血症	BTK	全人種	1 in 250,000	99%	1 in 250,000
530	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 in74,999,901
531	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
532	ANDROGEN INSENSITIVITY SYNDROME (XL) 雄性激素不敏感症候群	AR	全人種	1 in 5,000	99%	1 in 499,901
533	ARTS SYNDROME (XL) Arts 症候群	PRPS1	全人種	1 in 500,000	99%	1 in 49,999,901
534	BARTH SYNDROME (XL) 巴氏症候群	TAZ	全人種	1 in 225,000	99%	1 in 225,000
535	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

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
536	CHOROIDEREMIA (XL) 脈絡膜缺失症	CHM	全人種	1 in 25,000	99%	1 in 2,499,901
537	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
538	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
539	DENT DISEASE, TYPE 2/LOWE SYNDROME (XL) Lowe氏症候群	OCRL	全人種	1 in 375,000	99%	1 in 37,499,901
540	DEVELOPMENTAL AND EPILEPTIC ENCEPHALOPATHY 36 (XL) 發育性和癲癇性腦病變 36	ALG13	全人種	<1 in 750,000	99%	1 in 74,999,901
541	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
542	DYSKERATOSIS CONGENITA, DKC1-RELATED (XL) 先天性角化不全症-DKC1相關	DKC1	全人種	<1 in750,000	99%	1 in 74,999,901
543	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

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
544	EMERY-DREIFUSS MUSCULAR DYSTROPHY 6. X-LINKED (XL) Emery-Dreifuss肌失養症-FHL1相關	FHL1	全人種	<1 in 750,000	99%	1 in 74,999,901
545	FABRY DISEASE (XL) 法布瑞氏症	GLA	全人種	1 in 1,500	99%	1 in 149,901
546	FACTOR IX DEFICIENCY (XL) B型血友病	F9	全人種	1 in10,000	99%	1 in 999,901
547	FANCONI ANEMIA, GROUP B (XL) Fanconi氏貧血-B型	FANCB	全人種	1 in 750,000	99%	1 in 74,999,901
548	FRAGILE X SYNDROME (XL) X染色體脆折症	FMR1	全人種	1 in 250	99%	1 in 24,901
549	GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY (XL) 蠶豆症	G6PD	全人種	1 in 30	99%	1 in 2,901
550	HSD10 DISEASE (XL) HSD10 疾病	HSD17B10	全人種	<1 in 750,000	99%	1 in 74,999,901
551	HYPER IGM SYNDROME, X-LINKED (XL) 高免疫球蛋白M症候群	CD40LG	全人種	1 in 500,000	99%	1 in 50,000,000
552	HYPOHIDROTIC ECTODERMAL DYSPLASIA, X-LINKED (XL) 少汗性外胚層發育不良症	EDA	全人種	1 in 10,500	99%	1 in 1,049,901
553	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

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
554	L1 SYNDROME (XL) L1症候群	L1CAM	全人種	1 in 22,500	99%	1 in 2,249,901
555	LESCH-NYHAN SYNDROME (XL) Lesch-Nynan 症候群	HPRT1	全人種	1 in 285,000	99%	1 in 28,499,901
556	MECP2-RELATED CONDITIONS (XL) MECP2相關疾病	MECP2	全人種	≤1 in 500	99%	Reduced
557	MENKES SYNDROME (XL) Menkes氏症候群	ATP7A	全人種	1 in 75,000	99%	1 in 7,499,901
558	METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA TYPE CBLX (XL) 甲基丙二酸血症併高胱胺酸血症	HCFC1	全人種	<1 in 750,000	99%	1 in 74,999,901
559	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
560	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
561	NEPHROGENIC DIABETES INSIPIDUS, AVPR2-RELATED (XL) 腎因型尿崩症-AVPR2相關	AVPR2	全人種	1 in 63,000	99%	1 in 6,299,901
562	OPITZ G/BBB SYNDROME, X-LINKED (XL) Opitz-GBBB症候群	MID1	全人種	1 in 37,500	99%	1 in 3,749,901
563	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

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
564	PLP1 DISORDERS (XL) PLP1相關疾病	PLP1	全人種	1 in 37,500	99%	1 in 3,749,901
565	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
566	RETINITIS PIGMENTOSA 2 (XL) 視網膜色素變性 2 型	RP2	全人種	1 in 58,000	99%	1 in 5,799,901
567	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
568	WISKOTT-ALDRICH SYNDROME (XL) Wiskott-Aidrich症候群	WAS	全人種	1 in 187,500	99%	1 in 18,749,901
569	X-LINKED CHONDRODYSPLASIA PUNCTATA 1 (XL) 性聯遺傳點狀軟骨發育不良-1型	ARSL	全人種	1 in 375,000	99%	1 in 37,499,901
570	X-LINKED LISSENCEPHALY WITH ABNORMAL GENITALIA (XL) 性聯遺傳平腦症合併生殖器異常	ARX	全人種	1 in 750,000	50%	1 in 1,499,999