愛唯美帶因篩檢 Carrier Screening 569+

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，檢測率估計是基於Invitae使用的測試技術。殘留風險值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。

疾病 DISORDER	基因 GENE	種族 ETHNICITY	檢測前帶因率 CARRIER FREQUENCY BEFORE SCREENING	檢出率 DETECTION RATE	陰性剩餘風險 CARRIER RESIDUAL RISK AFTER NEGATIVE RESULT
2-methyl-3-hydroxybutyric aciduria (XL) 17β-羥基類脱氫酶10缺乏症	HSD17B10	全人種	1 in 500	99%	Reduced
3-hydroxy-3-methylglutaryl-CoA lyase deficiency (AR) 3-氫基-3-甲基戊二酸血症	HMGCL	全人種	1 in 500	99%	Reduced
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC1-related) (AR) 三甲基巴豆醯輔酶A梭化酶缺乏症-1型	MCCC1	全人種	1 in 134	99%	1 in 13,300
3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency (MCCC2-related) (AR) 三甲基巴豆醯輔酶A梭化酶缺乏症-2型	MCCC2	全人種	1 in 134	99%	1 in 13,300
17-beta hydroxysteroid dehydrogenase 3 deficiency (AR) 第三型 17-β-羥基類固醇脱氫酶缺乏症	HSD17B3	全人種	1 in 500	99%	Reduced
ABCA3-related conditions (AR) ABCA3相關疾病	ABCA3	全人種	1 in 500	99%	Reduced
ABCA4-related conditions (AR) ABCA4相關疾病	ABCA4	全人種	1 in 45	90%	1 in 441
ABCB11-related conditions (AR) ABCB11相關疾病	ABCB11	全人種	1 in 100	99%	1 in 9,900
ABCC8-related conditions (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型	ABCC8	全人種	1 in 177	99%	1 in 17,600
Abetalipoproteinemia (AR) 無β脂蛋白血症	MTTP	全人種	1 in 500	99%	Reduced
Achromatopsia (CNGB3-related) (AR) 色彩感應失能症	CNGB3	全人種	1 in 93	99%	1 in 9,200
ACOX1-related conditions (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%	Reduced
Acrodermatitis enteropathica (AR) 腸病變性肢端皮膚炎	SLC39A4	全人種	1 in 354	99%	1 in 35,300
Adenosine deaminase deficiency (AR) 腺苷脱氨酶缺乏症	ADA	全人種	1 in 224	92%	1 in 2,788
ADGRV1-related conditions (AR) ADGRV1相關疾病	ADGRV1	全人種	1 in 223	99%	1 in 22200
AHI1-related conditions (AR) Joubert氏症候群-AHI-1型	AHI1	全人種	1 in 447	99%	1 in 44600

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Aicardi-Goutieres syndrome 2 (AR) Aicardi-Goutieres 症候群第二型	RNASEH2B	全人種	1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 3 (AR) Aicardi-Goutieres 症候群第三型	RNASEH2C	全人種	1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 4 (AR) Aicardi-Goutieres 症候群第四型	RNASEH2A	全人種	1 in 500	99%	Reduced
Aicardi-Goutieres syndrome 5 (AR) Aicardi-Goutieres 症候群第五型	SAMHD1	全人種	1 in 500	99%	Reduced
AIPL1-related conditions (AR) 幼兒型嚴重視網膜失養症-AIPL1型	AIPL1 *	全人種	1 in 408	99%	1 in 40700
Aldosterone synthase deficiency (AR) 皮質酮甲基氧化酶缺乏症	CYP11B2	全人種	1 in 500	99%	Reduced
ALG13-related conditions (XL) ALG13相關疾病	ALG13	全人種	1 in 500	99%	Reduced
Alkaptonuria (AR) 黑尿症	HGD	全人種	1 in 250	99%	1 in 24,900
Alpha-1 antitrypsin deficiency (AR) α1-抗胰蛋白酶缺乏症	SERPINA1	全人種	1 in 13	99%	1 in 1200
Alpha-mannosidosis (AR) α 型甘露糖症	MAN2B1	全人種	1 in 354	99%	1 in 35,300
Alpha-N-acetylgalactosaminidase deficiency (AR) α -NAGA缺乏症	NAGA	全人種	1 in 500	99%	Reduced
Alpha-thalassemia (AR) 甲型海洋性貧血	HBA1 * HBA2	全人種	1 in 25	90%	1 in 241
Alpha-thalassemia X-linked 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 500	99%	Reduced
Alport syndrome (COL4A3-related) (AR) 亞伯氏症候群-COL4A3型	COL4A3	全人種	1 in 354	99%	1 in 35,300
Alport syndrome (COL4A4-related) (AR) 亞伯氏症候群-COL4A4型	COL4A4	全人種	1 in 353	99%	1 in 35,200
Alport syndrome (COL4A5-related) (XL) 性聯遺傳亞伯氏症候群	COL4A5	全人種	1 in 500	98%	Reduced
Alström syndrome (AR) Alstrom症候群	ALMS1	全人種	1 in 500	99%	Reduced
Androgen insensitivity syndrome (XL) 雄性激素不敏感症候群	AR *	全人種	1 in 500	99%	Reduced
Arginase deficiency (AR) 精胺酸酶缺乏症	ARG1	全人種	1 in 274	99%	1 in 27,300
Argininosuccinate lyase deficiency (AR) 精胺丁二酸酶缺乏症	ASL	全人種	1 in 133	90%	1 in 1,321
ARL6-related conditions (AR) ARL6相關疾病	ARL6	全人種	1 in 500	99%	Reduced

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Aromatase deficiency (AR) 芳香環轉化酶缺乏症	CYP19A1	全人種	1 in 500	99%	Reduced
ARX-related conditions (XL) ARX相關疾病	ARX *	全人種	1 in 500	99%	Reduced
Asparagine synthetase deficiency (AR) 天門冬醯胺酶合成缺乏症	ASNS	全人種	1 in 500	99%	Reduced
Aspartylglucosaminuria (AR) 天冬氨醯葡萄糖胺尿症	AGA	全人種	1 in 500	99%	Reduced
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	90%	Reduced
Ataxia-telangiectasia-like disorder (AR) 類共濟失調性毛细血管擴張症	MRE11	全人種	1 in 500	99%	Reduced
ATM-related conditions (AR) 共濟失調微血管擴張症候群	ATM *	全人種	1 in 100	99%	1 in 9,900
ATP7A-related conditions (XL) Menkes氏症候群	ATP7A	全人種	1 in 500	99%	Reduced
ATP8B1-related conditions (AR) ATP8B1相關疾病	ATP8B1 *	全人種	1 in 112	99%	1 in 11100
Atransferrinemia (AR) 無運鐵蛋白症	TF	全人種	1 in 500	99%	Reduced
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AR) 自體免疫多腺體症候群I型	AIRE	全人種	1 in 150	99%	1 in 14,900
Autosomal recessive congenital ichthyosis (ABCA12-related) (AR) 魚鱗癬-ABCA12型	ABCA12	全人種	1 in 500	99%	Reduced
Autosomal recessive congenital ichthyosis (TGM1-related) 魚鱗癬-TGM1型	TGM1	全人種	1 in 224	95%	1 in 4460
Autosomal recessive spastic ataxia of Charlevoix-Saguenay (AR) 體染色體隱性遺傳痙攣性共濟失調症	SACS	全人種	1 in 500	99%	Reduced
AVPR2-related conditions (XL) 腎因型尿崩症-AVPR2型	AVPR2	全人種	1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS7-related) (AR) Bardet-Biedl氏症候群-7型	BBS7	全人種	1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS9-related) (AR) Bardet-Biedl氏症候群-7型	BBS9 *	全人種	1 in 500	99%	Reduced
Bardet-Biedl syndrome (BBS7-related) (AR) Bardet-Biedl氏症候群-10型	BBS10	全人種	1 in 354	99%	1 in 35,300
Bardet-Biedl syndrome (BBS7-related) (AR) Bardet-Biedl氏症候群-12型	BBS12	全人種	1 in 500	99%	Reduced
Barth syndrome (XL) 巴氏症候群	TAZ	全人種	1 in 500	99%	Reduced
Bartter syndrome type 1 (AR) 巴特氏症候群-1型	SLC12A1	全人種	1 in 224	99%	1 in 22300

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Bartter syndrome type 2 (AR) 巴特氏症候群-2型	KCNJ1	全人種	1 in 500	99%	Reduced
BBS1-related conditions (AR) Bardet-Biedl氏症候群-1型	BBS1	全人種	1 in 330	99%	1 in 32,900
BBS2-related conditions (AR) Bardet-Biedl氏症候群-2型	BBS2	全人種	1 in 500	99%	Reduced
BBS4-related conditions (AR) Bardet-Biedl氏症候群-4型	BBS4	全人種	1 in 500	99%	Reduced
BBS5-related conditions (AR) Bardet-Biedl氏症候群-5型	BBS5	全人種	1 in 500	99%	Reduced
BCS1L-related conditions (AR) GRACILE 症候群	BCS1L	全人種	1 in 500	99%	Reduced
Bernard-Soulier syndrome (GP9-related) (AR) Bernard-Soulier症候群-GP9型	GP9	全人種	1 in 500	99%	Reduced
Beta-ketothiolase deficiency (AR) β-酮硫解酶缺乏症	ACAT1	全人種	1 in 500	99%	Reduced
Beta-mannosidosis (AR) β-甘露糖苷貯積症	MANBA	全人種	1 in 500	99%	Reduced
Biopterin-deficient hyperphenylalaninemia (PCBD1-related) (AR) 四氫基喋呤缺乏症-PCBD1型	PCBD1	全人種	1 in 500	99%	Reduced
Tetrahydrobiopterin deficiency (AR) NM_000317.2 苯酮尿症-PTS型	PTS	全人種	1 in 433	99%	1 in 43,200
Biopterin-deficient hyperphenylalaninemia (QDPR-related) (AR) 四氫基喋呤缺乏症-QPPR型	QDPR	全人種	1 in 500	99%	Reduced
Biotin-responsive basal ganglia disease (AR) 生物素-硫胺素反應性基底節病	SLC19A3	全人種	1 in 500	99%	Reduced
Biotinidase deficiency (AR) 生物素酶缺乏症	BTD	全人種	1 in 125	99%	1 in 12,400
Bloom syndrome (AR) Bloom症候群	BLM	全人種	1 in 500	99%	Reduced
BRIP1-related conditions (AR) BRIP1相關疾病	BRIP1	全人種	1 in 500	99%	Reduced
Brittle cornea syndrome (PRDM5-related) (AR) 脆性角膜症候群-PRDM5型	PRDM5	全人種	1 in 500	99%	Reduced
Brittle cornea syndrome (ZNF469-related) (AR) 脆性角膜症候群-ZNF469型	ZNF469	全人種	1 in 500	99%	Reduced
BSND-related conditions (AR) Bartter 氏症候群-4型	BSND	全人種	1 in 500	99%	Reduced
Canavan disease (AR) 家族性軸突海綿退化	ASPA	全人種	1 in 159	99%	1 in 15800
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%	Reduced

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Cardioencephalomyopathy (AR) 致死性心腦血管病	SCO2	全人種	1 in 387	99%	1 in 38600
Carnitine palmitoyltransferase I deficiency (AR) 肉鹼結合酶缺乏症-1型	CPT1A	全人種	1 in 500	99%	Reduced
Carnitine palmitoyltransferase II deficiency (AR) 肉鹼結合酶缺乏症-2型	CPT2	全人種	1 in 182	99%	1 in 18,100
Carnitine-acylcarnitine translocase deficiency (AR) 肉鹼轉位酶缺乏症	SLC25A20	全人種	1 in 500	99%	Reduced
Carpenter syndrome (RAB23-related) (AR) Carpenter症候群	RAB23	全人種	1 in 500	99%	Reduced
Cartilage-hair hypoplasia-anauxetic dysplasia spectrum disorders (AR) 骨骼發育異常症候群	RMRP	全人種	1 in 500	99%	Reduced
Catecholaminergic polymorphic ventricular tachycardia (CASQ2-related) (AR) 兒茶酚胺多型性心室頻脈-CASQ2型	CASQ2	全人種	1 in 224	99%	1 in 22300
CC2D2A-related conditions (AR) CC2D2A相關疾病	CC2D2A	全人種	1 in 426	99%	1 in 42500
CDH23-related conditions (AR) 尤塞氏症候群- 1D 型	CDH23	全人種	1 in 202	95%	1 in 4020
CEP290-related conditions (AR) 萊伯氏先天性黑矇症-10型	CEP290	全人種	1 in 185	99%	1 in 18,400
Cerebellar ataxia, intellectual disability, and dysequilibrium syndrome 1 (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%	Reduced
Cerebral dysgenesis, neuropathy, ichthyosis, and keratoderma (AR) 腦發育不全、魚鱗癬及角化症	SNAP29	全人種	1 in 500	99%	Reduced
Cerebrotendinous xanthomatosis (AR) 腦腱性黃瘤症	CYP27A1	全人種	1 in 112	98%	1 in 5550
CERKL-related conditions (AR) 視網膜色素病變-26型	CERKL	全人種	1 in 137	99%	1 in 13,600
CFTR-related conditions (AR) 囊腫纖維症	CFTR *	全人種	1 in 45	99%	1 in 4,400
Charcot-Marie-Tooth disease type 1X (XL) 進行性神經性腓骨萎縮症性聯遺傳-1型	GJB1	全人種	1 in 500	99%	Reduced
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%	Reduced
Chediak-Higashi syndrome (AR) Chediak-Higashi 症候群	LYST	全人種	1 in 500	99%	Reduced
Childhood-onset dystonia with optic atrophy and basal ganglia abnormalities (AR) 幼童期肌張力、視神經異常症	MECR	全人種	1 in 500	99%	Reduced
Chorea-acanthocytosis (AR) 舞蹈棘狀紅血球症	VPS13A *	全人種	1 in 500	97%	Reduced
Choroideremia (XL) 脈絡膜缺失症	CHM	全人種	1 in 500	95%	Reduced

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Chronic granulomatous disease (CYBA-related) (AR) 慢性肉芽腫病-CYBA型	CYBA	全人種	1 in 500	99%	Reduced
Chronic granulomatous disease (XL) NM_000397.3 性聯遺傳慢性肉芽腫病	CYBB	全人種	1 in 500	99%	Reduced
Chronic granulomatous disease (NCF2-related) (AR) 慢性肉芽腫病-NCF2型	NCF2	全人種	1 in 500	99%	Reduced
Citrin deficiency (AR) Citrin缺乏症	SLC25A13	全人種	1 in 313	99%	1 in 31200
Citrullinemia type 1 (AR) 瓜胺酸血症-1型	ASS1	全人種	1 in 120	96%	1 in 2,975
CLN3-related conditions (AR) 神經元蠟樣脂褐質沉著症-CLN3型	CLN3	全人種	1 in 230	99%	1 in 22,900
CLRN1-related conditions (AR) 尤塞氏症候群- 3A 型	CLRN1	全人種	1 in 500	99%	Reduced
Cobalamin C deficiency (AR) 甲基丙二酸血症併高胱胺酸血症-cblC 型	MMACHC	全人種	1 in 123	99%	1 in 12,200
Cobalamin D deficiency (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%	Reduced
Cobalamin F deficiency (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%	Reduced
Cockayne syndrome A (AR) 柯凱因氏症候群-A型	ERCC8	全人種	1 in 500	99%	Reduced
Cockayne syndrome B (AR) 柯凱因氏症候群-B型	ERCC6	全人種	1 in 377	99%	1 in 37,600
Cohen syndrome (AR) NM_017890.4 科恩症候群	VPS13B	全人種	1 in 500	99%	Reduced
COL11A2-related conditions (AR) 史蒂克勒氏症-COL11A2型	COL11A2 *	全人種	1 in 500	99%	Reduced
COL17A1-related conditions (AR) 遺傳性表皮分解性水皰症-COL17A1型	COL17A1	全人種	1 in 500	99%	Reduced
Combined malonic and methylmalonic aciduria (AR) 丙二酸及甲基丙二酸血症	ACSF3	全人種	1 in 87	99%	1 in 8,600
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%	Reduced
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	93%	Reduced
Combined pituitary hormone deficiency (LHX3-related)(AR) 結合性腦下垂體賀爾蒙缺失-LHX3型	LHX3	全人種	1 in 500	99%	Reduced
Combined pituitary hormone deficiency (POU1F1-related) (AR) 結合性腦下垂體賀爾蒙缺失-POU1F1型	POU1F1	全人種	1 in 500	99%	Reduced
Combined pituitary hormone deficiency (PROP1-related)(AR) 結合性腦下垂體賀爾蒙缺失-PROP1型	PROP1	全人種	1 in 45	98%	1 in 2200

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Congenital adrenal hyperplasia due to 3-beta-3-β-hydroxysteroid dehydrogenase deficiency (AR) 羥基類固醇脫氫酶缺乏症-2型	HSD3B2	全人種	1 in 500	99%	Reduced
Congenital adrenal hyperplasia due to 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	99%	Reduced
Congenital adrenal insufficiency (AR) 先天性腎上腺異常症	CYP11A1	全人種	1 in 500	99%	Reduced
Congenital chronic diarrhea (DGAT1-related) (AR) 先天性腹瀉病-DGAT1型	DGAT1	全人種	1 in 500	99%	Reduced
Congenital disorder of glycosylation (SLC35A3-related) SLC35A3相關疾病	SLC35A3	全人種	1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ia (AR) 先天性醣基化疾病-1a型	PMM2	全人種	1 in 190	99%	1 in 18900
Congenital disorder of glycosylation type Ib (AR) 先天性醣基化疾病-1b型	MPI	全人種	1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ic (AR) 先天性醣基化疾病-1c型	ALG6	全人種	1 in 500	99%	Reduced
Congenital disorder of glycosylation type Ik (AR) 先天性醣基化疾病-1k型	ALG1	全人種	1 in 500	99%	Reduced
Congenital disorder of glycosylation type Iv (AR) 先天性醣基化疾病-1v型	NGLY1	全人種	1 in 500	99%	Reduced
Congenital disorder of glycosylation type Iv (AR) 先天性醣基化疾病-2型	SEC23B	全人種	1 in 500	99%	Reduced
Congenital hydrocephalus-1 (AR) 先天性腦積水-1型	CCDC88C	全人種	1 in 500	99%	Reduced
Congenital hypothyroidism (TSHB-related) (AR) 先天性甲狀腺低功能症-TSHB型	TSHB	全人種	1 in 500	99%	Reduced
Congenital insensitivity to pain with anhidrosis (AR) 先天性痛覺不敏感合併無汗症	NTRK1	全人種	1 in 500	99%	Reduced
Congenital myasthenic syndrome (CHAT-related) (AR) 先天性肌無力症候群-CHAT型	CHAT	全人種	1 in 500	99%	Reduced
Congenital myasthenic syndrome (CHRNE-related) (AR) 先天性肌無力症候群-CHRNE型	CHRNE	全人種	1 in 200	99%	1 in 19,900
Congenital nephrotic syndrome type 1 (AR) 先天性腎病症候群-1型	NPHS1	全人種	1 in 500	99%	Reduced
Congenital nephrotic syndrome type 2 (AR) 先天性腎病症候群-2型	NPHS2	全人種	1 in 500	99%	Reduced
Congenital secretory chloride diarrhea (AR) 先天分泌性氯化物腹瀉	SLC26A3	全人種	1 in 500	99%	Reduced
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%	Reduced
CRB1-related conditions (AR) 萊伯氏先天性黑矇症-8型	CRB1	全人種	1 in 112	99%	1 in 11,100

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CTSC-related conditions (AR) CTSC相關疾病	CTSC	全人種	1 in 250	99%	1 in 24900
CYP1B1-related conditions (AR) 原發先天性青光眼-CYP1B1型	CYP1B1	全人種	1 in 79	99%	1 in 7800
CYP7B1-related conditions (AR) CYP7B1相關疾病	CYP7B1	全人種	1 in 500	99%	Reduced
CYP11B1-related conditions (AR) 先天性腎上腺增生症-11b羥化酶缺失症	CYP11B1	全人種	1 in 194	99%	1 in 19,300
CYP17A1-related conditions (AR) 先天性腎上腺增生症-17a羥化酶缺失症	CYP17A1	全人種	1 in 500	99%	Reduced
Cystinosis (AR) 胱胺酸血症	CTNS	全人種	1 in 158	99%	1 in 15,700
Cytochrome P450 oxidoreductase deficiency (AR) 細胞色素 P450 氧化還原酶缺乏症	POR	全人種	1 in 158	99%	1 in 15,700
Desbuquois dysplasia type 1 (AR) Desbuquois發育不全症-1型	CANT1	全人種	1 in 500	99%	Reduced
Developmental and epileptic encephalopathy (CAD-related) (AR) 發育癲癇性腦病變-CAD型	CAD	全人種	1 in 500	99%	Reduced
DGUOK-related conditions (AR) DGUOK相關疾病	DGUOK	全人種	1 in 500	99%	Reduced
DHDDS-related conditions (AR) 視網膜色素病變-59 型	DHDDS	全人種	1 in 500	99%	Reduced
Dihydrolipoamide dehydrogenase deficiency (AR) 二氫硫辛醯胺脫氫酶缺乏症	DLD	全人種	1 in 500	99%	Reduced
Distal renal tubular acidosis with deafness (ATP6V1B1-related) (AR) 腎小管酸中毒/耳聾 -ATP6V1B1型	ATP6V1B1	全人種	1 in 500	99%	Reduced
DMD-related conditions (XL) 裴馨氏肌肉萎縮症	DMD	全人種	1 in 500	99%	Reduced
DOK7-related conditions (AR) 肌無力症候群-DOK7型	DOK7	全人種	1 in 115	99%	1 in 11400
Donnai-Barrow syndrome (AR) Donnai-Barrow症候群	LRP2	全人種	1 in 500	99%	Reduced
Dubin-Johnson syndrome (AR) Dubin-Johnson症候群	ABCC2 *	全人種	1 in 500	99%	Reduced
DUOX2-related conditions (AR) 先天性甲狀腺低功能症-DUOX2型	DUOX2 *	全人種	1 in 58	91%	1 in 634
DYNC2H1-related conditions (AR) DYNC2H1相關疾病	DYNC2H1	全人種	1 in 224	99%	1 in 22300
DYSF-related conditions (AR) 肢帶型肌失養症 2B 型	DYSF	全人種	1 in 311	99%	1 in 31,000
Dyskeratosis congenita spectrum disorders (DKC1-related) (XL) 先天性角化不全症-DKC1型	DKC1	全人種	1 in 500	99%	Reduced

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Dyskeratosis congenita spectrum disorders (RTEL1-related) (AR) 先天性角化不全症-RTEL1型	RTEL1	全人種	1 in 500	99%	Reduced
Dyskeratosis congenita spectrum disorders (TERT-related) (AR) 先天性角化不全症-TERT型	TERT	全人種	1 in 500	99%	Reduced
Dystrophic epidermolysis bullosa (AR) 表皮分解性水皰症	COL7A1	全人種	1 in 370	97%	1 in 12,300
EDA-related conditions (XL) 少汗性外胚層發育不良症	EDA	全人種	1 in 500	99%	Reduced
Ehlers-Danlos syndrome, dermatosparaxis type (AR) 埃勒斯-當洛斯症候群	ADAMTS2	全人種	1 in 500	99%	Reduced
Ehlers-Danlos syndrome, kyphoscoliotic type (AR) Ehlers-Danlos症候群-PLOD1型	PLOD1	全人種	1 in 150	99%	1 in 14900
Ellis-van Creveld syndrome (EVC-related) (AR) 埃利偉氏症候群-EVC型	EVC	全人種	1 in 220	99%	1 in 21,900
Emery-Dreifuss muscular dystrophy (EMD-related) (XL) Emery-Dreifuss 肌失養症	EMD	全人種	1 in 500	99%	Reduced
Epidermolysis bullosa with pyloric atresia (ITGB4-related) (AR) 皰性表皮鬆解症-ITGB4型	ITGB4	全人種	1 in 393	99%	1 in 39200
Epimerase deficiency galactosemia (AR) 半乳糖異構酶缺乏症	GALE *	全人種	1 in 500	99%	Reduced
ERCC2-related conditions (AR) ERCC2相關疾病	ERCC2	全人種	1 in 500	99%	Reduced
Ethylmalonic encephalopathy (AR) 乙基丙二酸腦病變	ETHE1	全人種	1 in 500	99%	Reduced
EVC2-related conditions (AR) 埃利偉氏症候群-2型	EVC2	全人種	1 in 199	99%	1 in 19800
Fabry disease (XL) 法布瑞氏症	GLA	全人種	1 in 500	99%	Reduced
Factor IX deficiency (hemophilia B) (XL) 血友病 B 型	F9	全人種	1 in 500	99%	Reduced
Factor V Leiden thrombophilia (AD) 第五凝血因子突變血栓症	F5	全人種	1 in 26	99%	1 in 2,500
Factor XI deficiency (hemophilia C) (AR) 第11凝血因子缺乏症	F11	全人種	1 in 500	99%	Reduced
Familial chylomicronemia syndrome (AR) 家族性高乳糜微粒血症	LPL	全人種	1 in 500	99%	Reduced
Familial dysautonomia (AR) 家族性自主神經失調症-ELP1型	ELP1	全人種	1 in 500	99%	Reduced
Familial hemophagocytic lymphohistiocytosis type 2 (AR) 家族性噬血細胞性淋巴組織球增生症-2型	PRF1	全人種	1 in 177	99%	1 in 17600
Familial hemophagocytic lymphohistiocytosis type 3 (AR) 家族性噬血細胞性淋巴組織球增生症-3型	UNC13D	全人種	1 in 177	93%	1 in 2515

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Familial hemophagocytic lymphohistiocytosis type 4 (AR) 家族性嗜血細胞性淋巴組織球增生症-4型	STX11	全人種	1 in 224	99%	1 in 22300
Familial hemophagocytic lymphohistiocytosis type 5 (AR) 家族性嗜血細胞性淋巴組織球増生症-5型	STXBP2	全人種	1 in 224	99%	1 in 22300
Familial hypercholesterolemia (LDLR-related) (AD) 家族性高膽固醇血症-LDLR型	LDLR	全人種	1 in 250	99%	1 in 24900
Familial hypercholesterolemia (LDLRAP1-related) (AR) 家族性高膽固醇血症-LDLRAP型	LDLRAP1	全人種	1 in 500	99%	Reduced
Familial Mediterranean fever (AR) 家族性地中海熱	MEFV	全人種	1 in 64	90%	1 in 631
Fanconi anemia type A (AR) Fanconi氏貧血-A型	FANCA	全人種	1 in 345	99%	1 in 34400
Fanconi anemia type B (XL) Fanconi氏貧血-B型	FANCB	全人種	1 in 500	99%	Reduced
Fanconi anemia type C (AR) Fanconi氏貧血-C型	FANCC	全人種	1 in 417	99%	1 in 41600
Fanconi anemia type D2 (AR) Fanconi氏貧血-D2型	FANCD2 *	全人種	1 in 500	94%	Reduced
Fanconi anemia type E (AR) Fanconi氏貧血-E型	FANCE	全人種	1 in 500	99%	Reduced
Fanconi anemia type G (AR) Fanconi氏貧血-G型	FANCG	全人種	1 in 500	99%	Reduced
Fanconi anemia type I (AR) Fanconi氏貧血-I型	FANCI	全人種	1 in 500	99%	Reduced
Fanconi anemia type L (AR) Fanconi氏貧血-L型	FANCL *	全人種	1 in 500	99%	Reduced
FH-related conditions (AR) 延胡索酸酶缺乏症	FH	全人種	1 in 500	99%	Reduced
FHL1-related conditions (XL) Emery-Dreifuss肌失養症-FHL1型	FHL1	全人種	1 in 500	99%	Reduced
FKBP10-related conditions (AR) 先天性成骨不全症-FKBP10型	FKBP10	全人種	1 in 500	99%	Reduced
FMR1-related conditions including fragile X syndrome (XL) X染色體脆折症	FMR1 *	全人種	1 in 259	99%	1 in 25,800
Foveal hypoplasia (SLC38A8-related) (AR) 中心凹發育不全症	SLC38A8	全人種	1 in 500	99%	Reduced
FOXN1-related conditions (AR) FOXN1相關疾病	FOXN1	全人種	1 in 500	99%	Reduced
Fraser syndrome (FRAS1-related) (AR) Fraser症候群	FRAS1	全人種	1 in 316	99%	1 in 31500
Fraser syndrome (FREM2-related) (AR) Fraser氏症候-FREM2型	FREM2	全人種	1 in 500	99%	Reduced

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Fraser syndrome (GRIP1-related) (AR) Fraser氏症候-GRIP1型	GRIP1	全人種	1 in 447	99%	1 in 44600
Fructose-1,6-bisphosphatase deficiency (AR) 果糖-1,6-双磷酸酶缺乏症	FBP1	全人種	1 in 500	99%	Reduced
Fucosidosis (AR) 岩藻糖代謝異常儲積症	FUCA1	全人種	1 in 500	99%	Reduced
Galactokinase deficiency galactosemia (AR) 半乳糖激酶缺乏症	GALK1	全人種	1 in 122	99%	1 in 12100
Galactosemia (GALT-related) (AR) 半乳糖血症	GALT	全人種	1 in 100	99%	1 in 9900
Galactosialidosis (AR) 半乳糖唾液酸儲積症	CTSA	全人種	1 in 500	99%	Reduced
GATM-related conditions (AR) 先天性腦部肌酸缺乏症候群-GATM型	GATM	全人種	1 in 500	99%	Reduced
GBA-related conditions including Gaucher disease (AR) 高雪氏症	GBA *	全人種	1 in 158	72%	1 in 561
GBE1-related conditions (AR) 肝醣儲積症-GBE1型	GBE1	全人種	1 in 387	99%	1 in 38600
GCH1-related conditions (AR) GCH1相關疾病	GCH1	全人種	1 in 500	99%	Reduced
GDF5-related conditions (AR) 杜潘症候群	GDF5	全人種	1 in 500	99%	Reduced
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%	Reduced
GHR-related conditions (AR) GHR相關疾病	GHR *	全人種	1 in 500	98%	Reduced
Gitelman syndrome (AR) Gitelman症候群	SLC12A3	全人種	1 in 100	99%	1 in 9900
GJB2-related conditions (AR) 感覺神經性聽損-GJB2型	GJB2	全人種	1 in 50	99%	1 in 4900
GLB1-related conditions (AR) 黏多醣症-4B型	GLB1	全人種	1 in 158	99%	1 in 15700
GLE1-related conditions (AR) 致死先天性攣縮症候群	GLE1	全人種	1 in 500	99%	Reduced
Glucose-6-phosphate dehydrogenase deficiency (XL) 蠶豆症	G6PD	全人種	1 in 10	99%	1 in 900
Glutaric acidemia type I (AR) 戊二酸血症-1型	GCDH	全人種	1 in 87	99%	1 in 8600
Glutaric acidemia type IIA (AR) 戊二酸血症-2A型	ETFA	全人種	1 in 500	99%	Reduced
Glutaric acidemia type IIB (AR) 戊二酸血症-2B型	ETFB	全人種	1 in 500	99%	Reduced

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Glutaric acidemia type IIC (AR) 戊二酸血症-2C型	ETFDH	全人種	1 in 250	99%	1 in 24900
Glutathione synthetase deficiency (AR) 格斯特曼症候群	GSS	全人種	1 in 500	99%	Reduced
Glycine encephalopathy (AMT-related) (AR) 非酮性高甘胺酸血症-AMT型	AMT	全人種	1 in 325	99%	1 in 32400
Glycine encephalopathy (GLDC-related) (AR) 非酮性高甘胺酸血症-GLDC型	GLDC	全人種	1 in 165	99%	1 in 16400
Glycogen storage disease type Ia (AR) 肝醣儲積症-1A型	G6PC	全人種	1 in 177	95%	1 in 3520
Glycogen storage disease type II (Pompe disease) (AR) 龐貝氏症	GAA	全人種	1 in 100	99%	1 in 9900
Glycogen storage disease type III (AR) 肝醣儲積症-3型	AGL	全人種	1 in 159	95%	1 in 3160
Glycogen storage disease type IXb (AR) 肝醣儲積症-9B型	PHKB	全人種	1 in 500	99%	Reduced
Glycogen storage disease type IXc (AR) 肝醣儲積症-9C型	PHKG2	全人種	1 in 500	99%	Reduced
Glycogen storage disease type V (AR) 肝醣儲積症-5型	PYGM	全人種	1 in 171	99%	1 in 17000
Glycogen storage disease type VII (AR) 肝醣儲積症-7型	PFKM	全人種	1 in 500	99%	Reduced
GM3 synthase deficiency (AR) 阿米什嬰兒癲癇症候群	ST3GAL5	全人種	1 in 500	99%	Reduced
GNE-related conditions (AR) 包涵體肌炎	GNE *	全人種	1 in 179	99%	1 in 17800
GNPTAB-related conditions (AR) 黏脂質症 -2/3型	GNPTAB	全人種	1 in 200	99%	1 in 19900
GP1BA-related conditions (AR) Bernard-Soulier症候群-GP1BA型	GP1BA *	全人種	1 in 500	99%	Reduced
Guanidinoacetate methyltransferase deficiency (AR) 胍基乙酸甲基轉移酶缺乏症	GAMT	全人種	1 in 500	99%	Reduced
GUCY2D-related conditions (AR) GUCY2D相關疾病	GUCY2D	全人種	1 in 204	99%	1 in 20300
Gyrate atrophy of the choroid and retina (AR) 鳥胺酸酮酸轉胺酶缺乏症	OAT *	全人種	1 in 500	99%	Reduced
HADHA-related conditions (AR) 長鏈 3-羥烷基輔酶A脫氫酶缺乏症	HADHA	全人種	1 in 350	99%	1 in 34900
HBB-related hemoglobinopathies (AR) 乙型地中海貧血	HBB *	全人種	1 in 49	99%	1 in 4800
HCFC1-related conditions (XL) HCFC1相關疾病	HCFC1	全人種	1 in 500	99%	Reduced

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Heme oxygenase 1 deficiency (AR) 血鐵質氧化酶-1缺乏症	HMOX1	全人種	1 in 500	99%	Reduced
Hemolytic anemia, CD59-mediated (AR) 溶血性貧血-CD59型	CD59	全人種	1 in 500	99%	Reduced
Hereditary fructose intolerance (AR) 遺傳性果糖不耐症	ALDOB	全人種	1 in 122	99%	1 in 12100
Hereditary hemochromatosis type 1 (AR) 血鐵沉積症-1	HFE	全人種	1 in 4	99%	1 in 300
Hereditary hemochromatosis type 2 (HAMP-related) (AR) 血鐵沉積症-2型(HAMP類)	HAMP	全人種	1 in 500	99%	Reduced
Hereditary hemochromatosis type 2 (HJV-related) (AR) 血鐵沉積症-2型(HJV類)	HJV	全人種	1 in 500	99%	Reduced
Hereditary hemochromatosis type 3 (AR) 血鐵沉積症-3型	TFR2	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 1 (AR) Hermansky-Pudlak 症候群-1型	HPS1	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 3 (AR) Hermansky-Pudlak 症候群-3型	HPS3	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 4 (AR) Hermansky-Pudlak 症候群-4型	HPS4	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 5 (AR) Hermansky-Pudlak 症候群-5型	HPS5	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 6 (AR) Hermansky-Pudlak 症候群-6型	HPS6	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 8 (AR) Hermansky-Pudlak 症候群-8型	BLOC1S3	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome type 9 (AR) Hermansky-Pudlak 症候群-9型	BLOC1S6	全人種	1 in 500	99%	Reduced
HGSNAT-related conditions (AR) 黏多醣症-3C型	HGSNAT	全人種	1 in 500	99%	Reduced
Holocarboxylase synthetase deficiency (AR) 多發性羧化酶缺乏症	HLCS	全人種	1 in 224	99%	1 in 22300
Homocystinuria due to cobalamin E deficiency (AR) 高胱胺酸尿症-cobalamin E型	MTRR	全人種	1 in 500	99%	Reduced
Homocystinuria due to cobalamin G deficiency (AR) 高胱胺酸尿症-cobalamin G型	MTR	全人種	1 in 500	99%	Reduced
Homocystinuria due to cystathionine beta-synthase deficiency (AR) 高胱胺酸尿症	CBS	全人種	1 in 224	99%	1 in 22300
Homocystinuria due to MTHFR deficiency (AR) 高胱胺酸尿症-MTHFR型	MTHFR *	全人種	1 in 500	99%	Reduced
HPRT1-related conditions (XL) HPRT1相關疾病	HPRT1	全人種	1 in 500	99%	Reduced

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HSD17B4-related conditions (AR) D-雙功能蛋白缺乏症	HSD17B4	全人種	1 in 158	99%	1 in 15700
Hydrolethalus syndrome type 1 (AR) Hydrolethalus症候群	HYLS1	全人種	1 in 500	99%	Reduced
Hyper-IgM immunodeficiency (CD40-related) (AR) 高免疫球蛋白M症候群	CD40	全人種	1 in 500	99%	Reduced
Hyperornithinemia-hyperammonemia-homocitrullinuria 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%	Reduced
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%	Reduced
Hypomyelinating leukodystrophy-12 (AR) 低髓鞘腦白質失養症-12	VPS11	全人種	1 in 500	99%	Reduced
Hypophosphatasia (AR) 低磷酸酯酶症	ALPL	全人種	1 in 150	95%	1 in 2980
Ichthyosis prematurity syndrome (AR) 魚鱗癬-SLC27A4型	SLC27A4	全人種	1 in 500	99%	Reduced
IGHMBP2-related conditions (AR) IGHMBP2相關疾病	IGHMBP2	全人種	1 in 500	99%	Reduced
IKBKB-related conditions (AR) IKBKB相關疾病	IKBKB	全人種	1 in 500	99%	Reduced
Imerslund-Grsbeck syndrome (AR) Imerslund-Grsbeck症候群	AMN *	全人種	1 in 500	99%	Reduced
Immunodeficiency-centromeric instability-facial anomalies syndrome 1 (AR) ICF症候群-1 (DNMT3B類)	DNMT3B	全人種	1 in 500	99%	Reduced
Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (AR) ICF症候群-1 (ZBTB24類)	ZBTB24	全人種	1 in 500	99%	Reduced
Isolated ectopia lentis (AR) 孤立性水晶體異味症	ADAMTSL4	全人種	1 in 500	99%	Reduced
Isovaleric acidemia (AR) 異戊酸血症	IVD	全人種	1 in 250	99%	1 in 24900
ITGB3-related conditions (AR) ITGB3相關疾病	ITGB3	全人種	1 in 500	99%	Reduced
Johanson-Blizzard syndrome (AR) Johanson-Blizzard症候群	UBR1	全人種	1 in 250	99%	1 in 24900
Joubert syndrome and related disorders (MKS1-related) (AR) MKS1相關疾病	MKS1	全人種	1 in 260	95%	1 in 5180
Joubert syndrome and related disorders (RPGRIP1L-related) (AR) RPGRIP1L相關疾病	RPGRIP1L	全人種	1 in 259	95%	1 in 5160
Joubert syndrome and related disorders (TMEM216-related) (AR) Joubert 症候群-2型	TMEM216	全人種	1 in 500	99%	Reduced
Junctional epidermolysis bullosa (LAMC2-related) (AR) 接合性表皮溶解水皰症-LAMC2型	LAMC2	全人種	1 in 500	99%	Reduced

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Junctional epidermolysis bullosa with pyloric atresia (ITGA6-related) (AR) 接合性表皮溶解水皰症-ITGA6型	ITGA6	全人種	1 in 500	99%	Reduced
KCNJ11-related conditions (AR) 家族性胰島素過多症-KCNJ11型	KCNJ11	全人種	1 in 500	99%	Reduced
Krabbe disease (AR) Krabbe 症	GALC *	全人種	1 in 158	99%	1 in 15700
L1 syndrome (XL) L1症候群	L1CAM	全人種	1 in 500	99%	Reduced
LAMA2-related muscular dystrophy (AR) 肌肉失養症-LAMA2型	LAMA2	全人種	1 in 87	99%	1 in 8600
LAMA3-related conditions (AR) 接合性表皮溶解水皰症-LAMA3型	LAMA3	全人種	1 in 500	99%	Reduced
LAMB3-related conditions (AR) 接合性表皮溶解水皰症-LAMB3型	LAMB3	全人種	1 in 317	99%	1 in 31600
Leber congenital amaurosis 5 (AR) 萊伯氏先天性黑矇症-5型	LCA5	全人種	1 in 500	97%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B1-related) (AR) 腦白質病伴隨白質消失症-EIF2B1型	EIF2B1	全人種	1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B2-related) (AR) 腦白質病伴隨白質消失症-EIF2B2型	EIF2B2	全人種	1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B3-related) (AR) 腦白質病伴隨白質消失症-EIF2B3型	EIF2B3	全人種	1 in 500	99%	Reduced
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%	Reduced
Leukoencephalopathy with vanishing white matter (EIF2B5-related) (AR) 腦白質病伴隨白質消失症-EIF2B5型	EIF2B5	全人種	1 in 500	99%	Reduced
LIG4 syndrome (AR) LIG4症候群	LIG4	全人種	1 in 500	99%	Reduced
Limb-girdle muscular dystrophy (CAPN3-related) (AR) 肢帶型肌肉失養症-2A型	CAPN3	全人種	1 in 134	99%	1 in 13300
Limb-girdle muscular dystrophy type 2C (AR) 肢帶型肌肉失養症-2C型	SGCG	全人種	1 in 500	99%	Reduced
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%	Reduced
Limb-girdle muscular dystrophy type 2E (AR) 肢帶型肌肉失養症-2E型	SGCB	全人種	1 in 500	92%	Reduced
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%	Reduced
Lipoid congenital 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%	Reduced
LRAT-related conditions (AR) LRAT相關疾病	LRAT	全人種	1 in 296	99%	1 in 29500

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Lysinuric protein intolerance (AR) Lysunyruc 蛋白質耐受不良症	SLC7A7	全人種	1 in 500	99%	Reduced
Lysosomal acid lipase deficiency (AR) 溶酶體酸性脂肪酶缺乏症	LIPA	全人種	1 in 359	94%	1 in 5967
Major histocompatibility complex class II deficiency (CIITA- related) (AR) 裸淋巴球症候症-2型	CIITA	全人種	1 in 500	99%	Reduced
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%	Reduced
Maple syrup urine disease type 1A (AR) 楓糖尿症-1a型	BCKDHA	全人種	1 in 373	99%	1 in 37200
Maple syrup urine disease type 1B (AR) 楓糖尿症-1b型	BCKDHB	全人種	1 in 346	99%	1 in 34500
Maple syrup urine disease type 2 (AR) 楓糖尿症-2型	DBT	全人種	1 in 500	99%	Reduced
MECP2-related conditions (XL) MECP2相關疾病	MECP2	全人種	1 in 500	99%	Reduced
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 66	99%	1 in 6500
Medium/short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (AR) 長鏈 3-羥烷基輔酶A脫氫酶缺乏症	HADH	全人種	1 in 500	99%	Reduced
MEDNIK syndrome (AR) MEDNIK症候群	AP1S1	全人種	1 in 500	99%	Reduced
Megalencephalic leukoencephalopathy with subcortical cysts type 1 (AR) 巨腦性腦白質病伴有皮層下囊腫-1型	MLC1 *	全人種	1 in 500	99%	Reduced
Metabolic crises with rhabdomyolysis, cardiac arrhythmias and neurodegeneration (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%	Reduced
Metachromatic leukodystrophy (ARSA-related) (AR) 異染性腦白質退化症	ARSA	全人種	1 in 100	95%	1 in 1980
Methylmalonic acidemia (MCEE-related) (AR) 甲基丙二酸單醯輔酶A異構酶 缺乏症	MCEE	全人種	1 in 500	99%	Reduced
Methylmalonic acidemia (MMAA-related) (AR) 甲基丙二酸血症-cb1A型	MMAA	全人種	1 in 316	97%	1 in 10500
Methylmalonic acidemia (MMAB-related) (AR) 甲基丙二酸血症-cb1B型	MMAB	全人種	1 in 456	98%	1 in 22750
Methylmalonic acidemia (MUT-related) (AR) 甲基丙二酸血症-MUT型	MUT	全人種	1 in 204	96%	1 in 5075
MFSD8-related conditions (AR) MFSD8相關疾病	MFSD8	全人種	1 in 500	99%	Reduced
Microcephalic osteodysplastic primordial dwarfism type II (AR) 原始侏儒症II型	PCNT	全人種	1 in 500	99%	Reduced
Microcephaly, postnatal progressive, with seizures and brain 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%	Reduced

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Mitochondrial complex I deficiency 1 (AR) 粒線體複合物 I缺乏症-NDUFS4型	NDUFS4	全人種	1 in 500	99%	Reduced
Mitochondrial complex I deficiency 3 (AR) 粒線體複合物 I缺乏症-NDUFS7型	NDUFS7	全人種	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 4 (AR) 粒線體複合物 I缺乏症-NDUFV1型	NDUFV1	全人種	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 9 (AR) 粒線體複合物 I缺乏症-NDUFS6型	NDUFS6	全人種	1 in 500	99%	Reduced
Mitochondrial complex I deficiency 10 (AR) 粒線體複合物 I缺乏症-NDUFAF2型	NDUFAF2	全人種	1 in 387	99%	1 in 38600
Mitochondrial complex I deficiency 16 (AR) 粒線體複合物 I缺乏症-NDUFAF5型	NDUFAF5	全人種	1 in 500	99%	Reduced
Mitochondrial complex I deficiency 19 (AR) 粒線體複合物 I缺乏症-FOXRED1型	FOXRED1	全人種	1 in 376	99%	1 in 37500
Mitochondrial complex I deficiency 20 (AR) 粒線體複合物 I缺乏症-ACAD9型	ACAD9	全人種	1 in 500	99%	Reduced
Mitochondrial complex IV deficiency 6 (AR) 粒線體複合物 IV缺乏症-COX15型	COX15	全人種	1 in 500	99%	Reduced
Mitochondrial complex IV deficiency 12 (AR) 粒線體複合物 IV缺乏症-PET100型	PET100	全人種	1 in 387	99%	1 in 38600
Mitochondrial complex IV deficiency (AR) 粒線體複合物 IV缺乏症-LRPPRC型	LRPPRC	全人種	1 in 500	99%	Reduced
Mitochondrial DNA depletion syndrome-2 (AR) 粒線體DNA減除症候群	TK2	全人種	1 in 500	99%	Reduced
Mitochondrial neurogastrointestinal encephalomyopathy (AR) 粒線體性神經胃腸腦病變症候群	TYMP	全人種	1 in 500	99%	Reduced
Mitochondrial trifunctional protein deficiency (HADHB-related) (AR) 粒線體三功能蛋白缺失症-HADHB型	HADHB	全人種	1 in 500	99%	Reduced
MKKS-related conditions (AR) MKKS相關疾病	MKKS	全人種	1 in 500	99%	Reduced
Molybdenum cofactor deficiency (MOCS1-related) (AR) 鉬輔酶缺乏症-MOCS1型	MOCS1	全人種	1 in 226	99%	1 in 22500
Molybdenum cofactor deficiency (MOCS2B-related) (AR) 鉬輔酶缺乏症-MOCS2B型	MOCS2B	全人種	1 in 500	99%	Reduced
Molybdenum cofactor deficiency (MOCS2A-related) (AR) 鉬輔酶缺乏症-MOCS2A型	MOCS2A	全人種	1 in 500	99%	Reduced
MPL-related conditions (AR) 先天巨核細胞缺乏血小板低下症	MPL	全人種	1 in 500	99%	Reduced
MPV17-related conditions (AR) 肝腦病變型粒線體DNA耗竭症候群-MPV17型	MPV17	全人種	1 in 500	99%	Reduced
Mucolipidosis type III gamma (AR) 黏脂質症 -3型	GNPTG	全人種	1 in 500	99%	Reduced

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Mucolipidosis type IV (AR) 黏脂質症 -4型	MCOLN1	全人種	1 in 500	99%	Reduced
Mucopolysaccharidosis type I (AR) 黏多醣症-1型	IDUA	全人種	1 in 148	97%	1 in 4900
Mucopolysaccharidosis type II (XL) 黏多醣症-2型	IDS *	全人種	1 in 500	90%	Reduced
Mucopolysaccharidosis type IIIA (AR) 黏多醣症-3a型	SGSH	全人種	1 in 215	99%	1 in 21400
Mucopolysaccharidosis type IIIB (AR) 黏多醣症-3b型	NAGLU	全人種	1 in 224	99%	1 in 22300
Mucopolysaccharidosis type IIID (AR) 黏多醣症-3d型	GNS	全人種	1 in 500	99%	Reduced
Mucopolysaccharidosis type IVA (AR) 黏多醣症-4a型	GALNS	全人種	1 in 224	99%	1 in 22300
Mucopolysaccharidosis type IX (AR) 黏多醣症-9型	HYAL1	全人種	1 in 500	99%	Reduced
Mucopolysaccharidosis type VI (AR) 黏多醣症-6型	ARSB	全人種	1 in 250	99%	1 in 24900
Mucopolysaccharidosis type VII (AR) 黏多醣症-7型	GUSB	全人種	1 in 250	99%	1 in 24900
Mulibrey nanism (AR) 侏儒症併肌肉、肝、腦、眼異常	TRIM37	全人種	1 in 500	99%	Reduced
Multiple pterygium syndrome (AR) 多發性翼狀膜症候群	CHRNG	全人種	1 in 500	99%	Reduced
Multiple sulfatase deficiency (AR) 多發性硫酸脂酶缺乏症	SUMF1	全人種	1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (FKRP-related) (AR) Walker-Warburg症候群-FKRP型	FKRP	全人種	1 in 158	99%	1 in 15700
Muscular dystrophy-dystroglycanopathy (FKTN-related)(AR) Walker-Warburg 綜合症-FKTN型	FKTN	全人種	1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (LARGE1-related) (AR) Walker-Warburg 綜合症-LARGE1型	LARGE1	全人種	1 in 500	99%	Reduced
Muscular dystrophy-dystroglycanopathy (POMT1-related) (AR) 肌肉失養症醣基化功能缺陷-POMT1型	POMT1	全人種	1 in 268	99%	1 in 26700
Muscular dystrophy-dystroglycanopathy (POMT2-related) (AR) 肌肉失養症醣基化功能缺陷-POMT2型	POMT2	全人種	1 in 371	99%	1 in 37000
Muscular dystrophy-dystroglycanopathy (RXYLT1-related) (AR) 肌肉失養症醣基化功能缺陷-RXYLT1型	RXYLT1	全人種	1 in 500	99%	Reduced
MUSK-related conditions (AR) MUSK相關疾病	MUSK	全人種	1 in 447	99%	1 in 44600
MVK-related conditions (AR) 高免疫球蛋白血症D症候群	MVK	全人種	1 in 500	99%	Reduced

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MYO7A-related conditions (AR) 尤塞氏症候群- 1B 型	MYO7A	全人種	1 in 200	95%	1 in 3980
Myopathy, lactic acidosis, and sideroblastic anemia 1 (AR) 線粒體肌病和鐵粒細胞性貧血-1	PUS1	全人種	1 in 500	99%	Reduced
Myotonia congenita (AR) 先天性肌強直	CLCN1	全人種	1 in 112	99%	1 in 11100
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%	Reduced
Nemaline myopathy 2 (AR) 桿狀體肌症-2型	NEB *	全人種	1 in 158	95%	1 in 3140
Nephrogenic diabetes insipidus (AQP2-related) (AR) 腎性尿崩症	AQP2	全人種	1 in 500	99%	Reduced
Nephronophthisis (INVS-related) (AR) 腎消耗病-INVS型	INVS	全人種	1 in 500	99%	Reduced
Nephronophthisis (NPHP1-related) (AR) 腎消耗病-NPHP1型	NPHP1	全人種	1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 1 (AR) 神經元蠟樣脂褐質沉著症-PPT1型	PPT1	全人種	1 in 199	98%	1 in 9900
Neuronal ceroid lipofuscinosis type 2 (AR) 神經元蠟樣脂褐質沉著症-TPP1型	TPP1	全人種	1 in 250	97%	1 in 8300
Neuronal ceroid lipofuscinosis type 5 (AR) 神經元蠟樣脂褐質沉著症-CLN5型	CLN5	全人種	1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 6 (AR) 神經元蠟樣脂褐質沉著症-CLN6型	CLN6	全人種	1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 8 (AR) 神經元蠟樣脂褐質沉著症-CLN8型	CLN8	全人種	1 in 500	99%	Reduced
Neuronal ceroid lipofuscinosis type 10 (AR) 神經元蠟樣脂褐質沉著症-CTSD型	CTSD	全人種	1 in 500	99%	Reduced
Niemann-Pick disease type C (NPC1-related) (AR) 尼曼匹克症-C1型	NPC1	全人種	1 in 183	99%	1 in 18200
Niemann-Pick disease type C (NPC2-related) (AR) 尼曼匹克症-C2型	NPC2	全人種	1 in 500	99%	Reduced
Niemann-Pick disease types A and B (AR) 尼曼匹克症-A/B型	SMPD1	全人種	1 in 250	95%	1 in 4980
Nijmegen breakage syndrome (AR) Nijmegen破損症候群	NBN	全人種	1 in 500	99%	Reduced
Nonsyndromic deafness (LOXHD1-related) (AR) 遺傳性聽損-LOXHD1型	LOXHD1	全人種	1 in 500	99%	Reduced
Nonsyndromic deafness (MYO15A-related) (AR) 遺傳性聽損-MYO15A型	MYO15A	全人種	1 in 500	99%	Reduced
Nonsyndromic deafness (OTOA-related) (AR) 遺傳性聽損-OTOA型	OTOA *	全人種	1 in 500	88%	Reduced

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Nonsyndromic deafness (SYNE4-related) (AR) 遺傳性聽損-SYNE4型	SYNE4	全人種	1 in 500	99%	Reduced
Nonsyndromic deafness (TMC1-related) (AR) 遺傳性聽損-TMC1型	TMC1	全人種	1 in 500	99%	Reduced
Nonsyndromic deafness (TMPRSS3-related) (AR) 遺傳性聽損-TMPRSS3型	TMPRSS3	全人種	1 in 500	99%	Reduced
Nonsyndromic intellectual disability (CC2D1A-related) (AR) 無症狀性智力障礙-CC2D1A型	CC2D1A	全人種	1 in 500	99%	Reduced
NR0B1-related conditions (XL) 先天性腎上腺增生症-NR0B1型	NR0B1	全人種	1 in 500	99%	Reduced
NR2E3-related conditions (AR) 視網膜色素病變-37型	NR2E3	全人種	1 in 500	99%	Reduced
NSMCE3 deficiency (AR) NSMCE3缺發症	NSMCE3	全人種	1 in 500	99%	Reduced
OCRL-related conditions (XL) Lowe氏症候群	OCRL	全人種	1 in 500	99%	Reduced
Oculocutaneous albinism type 2 (AR) 眼睛皮膚白化症第2型	OCA2	全人種	1 in 95	99%	1 in 9400
Oculocutaneous albinism type 3 (AR) 眼睛皮膚白化症第3型	TYRP1	全人種	1 in 500	99%	Reduced
Oculocutaneous albinism type 4 (AR) 眼睛皮膚白化症第4型	SLC45A2	全人種	1 in 158	99%	1 in 15700
Oculocutaneous albinism types 1A and 1B (AR) 眼睛皮膚白化症第1型	TYR *	全人種	1 in 100	97%	1 in 3300
OPA3-related conditions (AR) Costeff症候群	OPA3	全人種	1 in 500	99%	Reduced
Opitz GBBB syndrome (MID1-related) (XL) Opitz-GBBB症候群	MID1 *	全人種	1 in 500	98%	Reduced
Ornithine transcarbamylase deficiency (XL) 鳥胺酸氨甲醯基轉移酶缺乏症	OTC	全人種	1 in 500	85%	Reduced
Osteogenesis imperfecta (BMP1-related) (AR) 骨質形成不全症-BMP1型	BMP1	全人種	1 in 500	99%	Reduced
Osteogenesis imperfecta (CRTAP-related) (AR) 骨質形成不全症-CRTAP型	CRTAP	全人種	1 in 500	99%	Reduced
Osteogenesis imperfecta (P3H1-related) (AR) 骨質形成不全症-P3H1型	P3H1	全人種	1 in 500	99%	Reduced
Osteopetrosis (TCIRG1-related) (AR) 骨質石化症-TCIRG1型	TCIRG1	全人種	1 in 317	99%	1 in 31600
OSTM1 deficiency associated osteopetrosis (AR) 骨質石化症-OSTM1型	OSTM1	全人種	1 in 500	99%	Reduced
OTOF-related conditions (AR) 感覺神經性聽損-OTOF型	OTOF	全人種	1 in 500	99%	Reduced

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Pantothenate kinase-associated neurodegeneration (AR) 泛酸鹽激活酵素關聯之神經退化性疾病	PANK2	全人種	1 in 289	99%	1 in 28800
Parkinson disease 15 (AR) 帕金森氏症-FBXO7型	FBXO7	全人種	1 in 500	99%	Reduced
PCDH15-related conditions (AR) 尤塞氏症候群- 1F 型	PCDH15	全人種	1 in 400	99%	1 in 39900
PEX5-related conditions (AR) PEX5相關疾病	PEX5	全人種	1 in 500	99%	Reduced
PEX7-related conditions (AR) 肢近端型點狀軟骨發育不良-1型	PEX7	全人種	1 in 157	99%	1 in 15600
PGM3-congenital disorder of glycosylation (AR) 先天性醣基化障礙-PGM3型	PGM3	全人種	1 in 500	99%	Reduced
Phenylalanine hydroxylase deficiency (AR) 苯酮尿症	PAH	全人種	1 in 58	99%	1 in 5700
Phosphoglycerate dehydrogenase deficiency (AR) 磷酸甘油酸脫氫酶缺乏症	PHGDH	全人種	1 in 500	99%	Reduced
PIGN-congenital disorder of glycosylation (AR) 先天性醣基化障礙-PIGN型	PIGN	全人種	1 in 500	99%	Reduced
PJVK-related conditions (AR) PJVK相關疾病	DFNB59	全人種	1 in 500	99%	Reduced
PLA2G6-related conditions (AR) 新生兒神經軸發育不良-PLA2G6型	PLA2G6	全人種	1 in 500	99%	Reduced
PLEKHG5-related conditions (AR) PLEKHG5相關疾病	PLEKHG5	全人種	1 in 500	99%	Reduced
PLP1-related conditions (XL) 痙攣性下身麻痺-PLP1型	PLP1	全人種	1 in 500	99%	Reduced
POLG-related conditions (AR) 漸進性外眼肌麻痺-POLG型	POLG	全人種	1 in 113	95%	1 in 2240
Polycystic kidney disease (PKHD1-related) (AR) 隱性多囊腎病-PKHD1型	PKHD1 *	全人種	1 in 70	99%	1 in 6900
Polymicrogyria (ADGRG1-related) (AR) 雙側額頂葉多小腦迴畸形症	ADGRG1	全人種	1 in 500	99%	Reduced
POMGNT1-related conditions (AR) POMGNT1相關症	POMGNT1	全人種	1 in 500	99%	Reduced
Pontocerebellar hypoplasia (TSEN54-related) (AR) 橋腦小腦發育不全-TSEN54型	TSEN54	全人種	1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 1B (AR) 橋腦小腦發育不全-1B型	EXOSC3	全人種	1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 2D (AR) 橋腦小腦發育不全-2D型	SEPSECS	全人種	1 in 500	99%	Reduced
Pontocerebellar hypoplasia type 6 (AR) 橋腦小腦發育不全-6型	RARS2	全人種	1 in 500	99%	Reduced

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Primary carnitine deficiency (AR) 原發性肉鹼缺乏症	SLC22A5	全人種	1 in 71	99%	1 in 7000
Primary ciliary dyskinesia (CCDC39-related) (AR) 原發性纖毛運動障礙-CCDC39型	CCDC39	全人種	1 in 211	99%	1 in 21000
Primary ciliary dyskinesia (CCDC103-related) (AR) 原發性纖毛運動障礙-CCDC103型	CCDC103	全人種	1 in 316	99%	1 in 31500
Primary ciliary dyskinesia (DNAH5-related) (AR) 原發性纖毛運動障礙-DNAH5型	DNAH5	全人種	1 in 109	99%	1 in 10800
Primary ciliary dyskinesia (DNAH11-related) (AR) 原發性纖毛運動障礙-DNAH11型	DNAH11	全人種	1 in 211	99%	1 in 21000
Primary ciliary dyskinesia (DNAI1-related) (AR) 原發性纖毛運動障礙-DNAI1型	DNAI1	全人種	1 in 250	99%	1 in 24900
Primary ciliary dyskinesia (DNAI2-related) (AR) 原發性纖毛運動障礙-DNAI2型	DNAI2	全人種	1 in 354	99%	1 in 35300
Primary hyperoxaluria type 1 (AR) 原發性高草酸尿症-1型	AGXT	全人種	1 in 135	99%	1 in 13400
Primary hyperoxaluria type 2 (AR) 原發性高草酸尿症-2型	GRHPR	全人種	1 in 500	99%	Reduced
Primary hyperoxaluria type 3 (AR) 原發性高草酸尿症-3型	HOGA1	全人種	1 in 354	99%	1 in 35300
Primary microcephaly (MCPH1-related) (AR) 原發性小頭畸形症-MCPH1型	MCPH1	全人種	1 in 500	99%	Reduced
Progressive early-onset encepahlopathy with brain atrophy and thin corpus callosum (PEBAT) (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%	Reduced
Progressive familial intrahepatic cholestasis 3 (AR) 家族進行性膽汁淤積症 -3型 (AR)	ABCB4	全人種	1 in 204	99%	1 in 20300
Progressive pseudorheumatoid dysplasia (AR) 進行性假性類風濕發育不良症候群	WISP3	全人種	1 in 500	99%	Reduced
Prolidase deficiency (AR) 脯氨酸肽酶缺乏症	PEPD	全人種	1 in 500	99%	Reduced
Propionic acidemia (PCCA-related) (AR) 丙酸血症-PCCA型	PCCA	全人種	1 in 224	96%	1 in 5575
Propionic acidemia (PCCB-related) (AR) 丙酸血症-PCCB型	PCCB	全人種	1 in 224	99%	1 in 22300
Prothrombin-related thrombophilia (AD) 凝血酶原相關血栓病	F2	全人種	1 in 62	99%	1 in 6100
PRPS1-related conditions (XL) PRPS1相關疾病	PRPS1	全人種	1 in 500	99%	Reduced
PSAP-related conditions (AR) 結合性SAP缺乏症	PSAP	全人種	1 in 500	99%	Reduced
Pycnodysostosis (AR) 緻密性成骨不全症	CTSK	全人種	1 in 438	99%	1 in 43700

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Pyridoxal 5'-phosphate-dependent epilepsy (AR) 磷酸吡哆醇依賴性癲癇	PNPO	全人種	1 in 500	99%	Reduced
Pyridoxine-dependent epilepsy (ALDH7A1-related) (AR) 維生素B6依賴性癲癇-ALDH7A1型	ALDH7A1	全人種	1 in 127	99%	1 in 12600
Pyruvate carboxylase deficiency (AR) 丙酮酸羧化酶缺乏症	PC	全人種	1 in 250	95%	1 in 4980
Pyruvate dehydrogenase complex deficiency (PDHA1-related) (XL) 丙酮酸鹽脫氫酶缺乏症-PDHA1型	PDHA1	全人種	1 in 500	99%	Reduced
Pyruvate dehydrogenase complex deficiency (PDHB-related) (AR) 丙酮酸鹽脫氫酶缺乏症-PDHB型	PDHB	全人種	1 in 500	99%	Reduced
RAPSN-related conditions (AR) 先天性肌無力症候群-RAPSN型	RAPSN	全人種	1 in 283	99%	1 in 28200
RDH12-related conditions (AR) 萊伯氏先天性黑矇症-13型	RDH12	全人種	1 in 460	99%	1 in 45900
Refsum disease (PHYH-related) (AR) 雷弗素姆病-PHYH型	PHYH	全人種	1 in 500	99%	Reduced
Retinitis pigmentosa 2 (XL) 網膜色素變性-RP2型	RP2	全人種	1 in 500	99%	Reduced
Retinitis pigmentosa 25 (AR) 視網膜色素病變-25型	EYS *	全人種	1 in 129	99%	1 in 12800
Retinitis pigmentosa 28 (AR) 視網膜色素病變-28型	FAM161A	全人種	1 in 289	99%	1 in 28800
Retinitis pigmentosa 36 (AR) 視網膜色素病變-36型	PRCD	全人種	1 in 296	99%	1 in 29500
Retinitis pigmentosa 62 (AR) 視網膜色素病變-62型	MAK	全人種	1 in 274	99%	1 in 27300
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%	Reduced
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%	Reduced
RLBP1-related conditions (AR) RLBP1相關疾病	RLBP1	全人種	1 in 296	99%	1 in 29500
Roberts syndrome (AR) Roberts症候群	ESCO2	全人種	1 in 500	99%	Reduced
RPE65-related conditions (AR) 萊伯氏先天性黑矇症-2型	RPE65	全人種	1 in 228	99%	1 in 22700
RYR1-related conditions (AR) RYR1相關疾病	RYR1	全人種	1 in 500	99%	Reduced
SAMD9-related conditions (AR) 腫瘤性石灰沉著症-SAMD9型	SAMD9	全人種	1 in 500	99%	Reduced
Sandhoff disease (AR) Sandoff症	HEXB	全人種	1 in 180	99%	1 in 17900

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Schimke immuno-osseous dysplasia (AR) Schimke免疫-骨發育不良	SMARCAL1	全人種	1 in 500	99%	Reduced
Seckel syndrome (CEP152-related) (AR) Seckel症候群	CEP152	全人種	1 in 500	99%	Reduced
Sepiapterin reductase deficiency (AR) 墨蝶呤還原酶缺乏症	SPR	全人種	1 in 500	99%	Reduced
Severe combined immunodeficiency due to CD3-delta deficiency (AR) 嚴重複合型免疫缺乏症-CD3D型	CD3D	全人種	1 in 500	99%	Reduced
Severe combined immunodeficiency due to CD3-epsilon deficiency (AR) 嚴重複合型免疫缺乏症-CD3E型	CD3E	全人種	1 in 500	99%	Reduced
Severe combined immunodeficiency due to CD45 deficiency (AR) 嚴重複合型免疫缺乏症-PTPRC型	PTPRC *	全人種	1 in 500	99%	Reduced
Severe combined immunodeficiency due to DCLRE1C (Artemis) deficiency (AR) 嚴重複合型免疫缺乏症-DCLRE1C型	DCLRE1C	全人種	1 in 500	99%	Reduced
Severe combined immunodeficiency due to IL7R-alpha deficiency (AR) 嚴重複合型免疫缺乏症-IL7R型	IL7R	全人種	1 in 348	99%	1 in 34700
Severe combined immunodeficiency due to JAK3 deficiency (AR) 嚴重複合型免疫缺乏症-JAK3型	JAK3	全人種	1 in 455	99%	1 in 45400
Severe combined immunodeficiency due to RAG1 deficiency (AR) 嚴重複合性免疫缺乏症-RAG1型	RAG1	全人種	1 in 301	99%	1 in 30000
Severe combined immunodeficiency due to RAG2 deficiency (AR) 嚴重複合性免疫缺乏症-RAG2型	RAG2	全人種	1 in 500	99%	Reduced
Severe congenital neutropenia due to G6PC3 deficiency (AR) 嚴重先天性嗜中性球減少症-G6PC3型	G6PC3	全人種	1 in 500	99%	Reduced
Severe congenital neutropenia due to HAX1 deficiency (AR) 嚴重先天性嗜中性球減少症-HAX1型	HAX1	全人種	1 in 500	99%	Reduced
Severe congenital neutropenia due to VPS45 deficiency (AR) 嚴重先天性嗜中性球減少症-VPS45型	VPS45	全人種	1 in 500	99%	Reduced
Sialic acid storage diseases (AR) 唾液酸貯積症	SLC17A5	全人種	1 in 500	99%	Reduced
Sialidosis (AR) 涎酸酵素缺乏症	NEU1	全人種	1 in 500	99%	Reduced
Sjögren-Larsson syndrome (AR) Sjögren-Larsson症候群	ALDH3A2	全人種	1 in 500	99%	Reduced
SLC12A6-related conditions (AR) Andermann 症候群	SLC12A6	全人種	1 in 500	99%	Reduced
SLC26A2-related conditions (AR) SLC26A2相關疾病	SLC26A2	全人種	1 in 158	95%	1 in 3140

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SLC26A4-related conditions (AR) 感覺神經性聽損-SLC26A4型	SLC26A4	全人種	1 in 80	99%	1 in 7900
SLC37A4-related conditions (AR) 肝醣儲積症-1B型	SLC37A4	全人種	1 in 354	95%	1 in 7060
Smith-Lemli-Opitz syndrome (AR) Smith-Lemli-Opitz 症候群	DHCR7	全人種	1 in 71	99%	1 in 7000
Spastic paraplegia type 15 (AR) 痙攣性下身麻痺-15型	ZFYVE26	全人種	1 in 500	99%	Reduced
Spastic paraplegia type 49 (AR) 痙攣性下身麻痺-49型	TECPR2	全人種	1 in 500	99%	Reduced
Spastic tetraplegia, thin corpus callosum, and progressive microcephaly (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%	Reduced
SPG11-related conditions (AR) 神經肌肉疾病-SPG11型	SPG11	全人種	1 in 141	99%	1 in 14000
Spinal muscular atrophy (AR) 脊髓性肌肉萎縮症	SMN1 *	全人種	1 in 49	94%	1 in 800
Spinocerebellar ataxia (ANO10-related) (AR) 脊髓小腦共濟失調症-ANO10型	ANO10 *	全人種	1 in 500	99%	Reduced
Spondylocostal dysostosis (DLL3-related) (AR) 脊椎肋骨發育不全-DLL3型	DLL3	全人種	1 in 350	99%	1 in 34900
Spondylocostal dysostosis (MESP2-related) (AR) 脊椎肋骨發育不全-MESP2	MESP2	全人種	1 in 224	99%	1 in 22300
Steel syndrome (AR) 鋼鐵症候群	COL27A1	全人種	1 in 500	99%	Reduced
Steroid 5-alpha-reductase deficiency (AR) 類固醇5α-還原酶2缺乏症	SRD5A2	全人種	1 in 500	99%	Reduced
Stüve-Wiedemann syndrome (AR) Stuve-Wiedemann 症候群	LIFR *	全人種	1 in 500	99%	Reduced
Sulfite oxidase deficiency (AR) 亞硫酸鹽氧化酵素缺乏症	SUOX	全人種	1 in 500	99%	Reduced
SURF1-related conditions (AR) SURF1相關疾病	SURF1	全人種	1 in 128	99%	1 in 12700
Tay-Sachs disease (AR) 薩克斯症	HEXA	全人種	1 in 250	99%	1 in 24900
TBCE-related conditions (AR) TBCE相關疾病	TBCE *	全人種	1 in 500	99%	Reduced
Thiamine-responsive megaloblastic anemia (AR) 維生素B1代謝功能障礙症候群	SLC19A2	全人種	1 in 500	99%	Reduced

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Thyroid dyshormonogenesis (SLC5A5-related) (AR) 甲狀腺素合成異常-SLC5A5型	SLC5A5	全人種	1 in 500	99%	Reduced
Thyroid dyshormonogenesis (TG-related) (AR) 甲狀腺素合成異常-TG型	TG *	全人種	1 in 500	99%	Reduced
Thyroid dyshormonogenesis (TPO-related) (AR) 甲狀腺素合成異常-TPO型	TPO	全人種	1 in 129	99%	1 in 12800
TMEM67-related conditions (AR) TMEM67相關疾病	TMEM67	全人種	1 in 316	99%	1 in 31500
Transcobalamin II deficiency (AR) 轉鈷胺素II缺乏症	TCN2	全人種	1 in 500	99%	Reduced
Transient infantile liver failure (AR) 急性新生兒肝衰竭	TRMU	全人種	1 in 500	99%	Reduced
TREX1-related conditions (AR) TREX1相關疾病	TREX1	全人種	1 in 500	99%	Reduced
Trichohepatoenteric syndrome (SKIV2L-related) (AR) 髮-肝-腸症候群-SKIV2L型	SKIV2L	全人種	1 in 500	99%	Reduced
Trichohepatoenteric syndrome (TTC37-related) (AR) 髮-肝-腸症候群-TTC37型	TTC37	全人種	1 in 500	99%	Reduced
TRIM32-related conditions (AR) Bardet-Biedl氏症候群-TRIM32	TRIM32	全人種	1 in 408	99%	1 in 40700
Trimethylaminuria (AR) 臭魚症	FMO3	全人種	1 in 500	99%	Reduced
Triple A syndrome (AR) Triple A 症候群	AAAS	全人種	1 in 500	99%	Reduced
TSHR-related conditions (AR) TSHR相關疾病	TSHR	全人種	1 in 158	99%	1 in 15700
TULP1-related conditions (AR) TULP1相關疾病	TULP1	全人種	1 in 296	99%	1 in 29500
Tyrosine hydroxylase deficiency (AR) 酪胺酸羥化酶缺乏症	TH	全人種	1 in 500	99%	Reduced
Tyrosinemia type I (AR) 酪胺酸血症-1型	FAH *	全人種	1 in 125	95%	1 in 2480
Tyrosinemia type II (AR) 酪胺酸血症-2型	TAT	全人種	1 in 250	99%	1 in 24900
Tyrosinemia type III (AR) 酪胺酸血症-3型	HPD	全人種	1 in 500	99%	Reduced
USH1C-related conditions (AR) 尤塞氏症候群- 1C 型	USH1C *	全人種	1 in 353	90%	1 in 3521

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USH2A-related conditions (AR) 尤塞氏症候群- 2A 型	USH2A	全人種	1 in 112	99%	1 in 11100
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 100	99%	1 in 9900
Vici syndrome (AR) Vici 症候群	EPG5	全人種	1 in 500	99%	Reduced
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 500	99%	Reduced
Vitamin D-dependent rickets type 2A (AR) 維生素 D 依賴型佝僂症 -2A型	VDR	全人種	1 in 500	99%	Reduced
VPS53-related conditions (AR) VPS53相關疾病	VPS53 *	全人種	1 in 500	99%	Reduced
VRK1-related conditions (AR) 橋腦小腦發育不全-1A型	VRK1	全人種	1 in 500	99%	Reduced
VSX2-related conditions (AR) 小眼症-VSX2型	VSX2	全人種	1 in 500	99%	Reduced
Warsaw syndrome (AR) Warsaw症候群	DDX11 *	全人種	1 in 500	15%	Reduced
WAS-related conditions (XL) WAS相關疾病	WAS	全人種	1 in 500	99%	Reduced
Werner syndrome (AR) Werner症候群	WRN *	全人種	1 in 224	99%	1 in 22300
Wilson disease (AR) 威爾森氏症	ATP7B	全人種	1 in 90	98%	1 in 4450
WNT10A-related conditions (AR) WNT10A相關疾病	WNT10A	全人種	1 in 305	99%	1 in 30400
Wolcott-Rallison syndrome (AR) Wolcott-Rallison 症候群	EIF2AK3	全人種	1 in 500	99%	Reduced
Woodhouse-Sakati syndrome (AR) Woodhouse-Sakati 症候群	DCAF17	全人種	1 in 500	99%	Reduced
X-linked adrenoleukodystrophy (XL) 性聯遺傳腎上腺白質退化症	ABCD1	全人種	1 in 500	99%	Reduced
X-linked agammaglobulinemia (XL) 布魯頓氏低免疫球蛋白血症	BTK	全人種	1 in 500	99%	Reduced
X-linked chondrodysplasia punctata type 1 (XL) 性聯遺傳點狀軟骨發育不良-1型	ARSE	全人種	1 in 500	99%	Reduced
X-linked creatine transporter deficiency (XL) 性聯遺傳肌酸缺乏症	SLC6A8	全人種	1 in 500	99%	Reduced

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X-linked hyper-IgM immunodeficiency (XL) 高免疫球蛋白M症候群	CD40LG	全人種	1 in 500	99%	Reduced
X-linked juvenile retinoschisis (XL) 性聯遺傳視網膜裂損症	RS1	全人種	1 in 500	99%	Reduced
X-linked myotubular myopathy (XL) 性聯遺傳肌小管病變	MTM1	全人種	1 in 500	96%	Reduced
X-linked severe combined immunodeficiency (XL) 性聯遺傳嚴重免疫缺陷-IL2RG型	IL2RG	全人種	1 in 500	99%	Reduced
Xeroderma pigmentosum complementation group A (AR) 著色性乾皮症-A型	XPA	全人種	1 in 500	99%	Reduced
Xeroderma pigmentosum complementation group C (AR) 著色性乾皮症-C型	XPC	全人種	1 in 500	99%	Reduced
Xeroderma pigmentosum, variant type (AR) 著色性乾皮症-POLH型	POLH	全人種	1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX1-related) (AR) 柴爾維格氏症候群-PEX1型	PEX1 *	全人種	1 in 144	99%	1 in 14300
Zellweger spectrum disorder (PEX2-related) (AR) 柴爾維格氏症候群-PEX2型	PEX2	全人種	1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX6-related) (AR) 柴爾維格氏症候群-PEX6型	PEX6	全人種	1 in 294	99%	1 in 29300
Zellweger spectrum disorder (PEX10-related) (AR) 柴爾維格氏症候群-PEX10型	PEX10	全人種	1 in 500	94%	Reduced
Zellweger spectrum disorder (PEX12-related) (AR) 柴爾維格氏症候群-PEX12型	PEX12	全人種	1 in 409	99%	1 in 40800
Zellweger spectrum disorder (PEX13-related) (AR) 柴爾維格氏症候群-PEX13型	PEX13	全人種	1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX16-related) (AR) 柴爾維格氏症候群-PEX16型	PEX16	全人種	1 in 500	99%	Reduced
Zellweger spectrum disorder (PEX26-related) (AR) 柴爾維格氏症候群-PEX26型	PEX26	全人種	1 in 500	99%	Reduced