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，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
Cystic fibrosis (AR) NM_000492.3 囊腫纖維症	CFTR	全人種	1 in 45	99%	1 in 4,400
Fragile X syndrome (XL) NM_002024.5 X染色體脆折症	FMR1＊	全人種	1 in 259	99%	1 in 25,800
Spinal muscular atrophy (AR) NM_000344.3 (SMN1: 2 copies g.27134T>G not detected") Carrier residual risks listed are for 2 copy SMN1 results. Carrier residual risks for > 2 copies are 5-to 10-fold lower. 脊髓性肌肉萎縮症	SMN1＊	亞洲	1 in 53	93%	1 in 743
3-beta-hydroxysteroid dehydrogenase type II deficiency (congenital adrenal hyperplasia) (AR) NM_000198.3 3-β-羥基類固醇脫氫酶缺乏症-2型	HSD3B2	全人種	1 in 500	99%	Reduced
3-hydroxy-3-methylglutarayl-CoA (HMG-CoA) lyase deficiency (AR) NM_000191.2 3-氫基-3-甲基戊二酸血症	HMGCL	全人種	1 in 500	99%	Reduced
		葡萄牙	1 in 160	99%	1 in 15,900
3-methylcrotonyl-CoA carboxylase deficiency (AR) NM_020166.4 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 deficiency (AR) NM_022132.4 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
3-methylglutaconic aciduria type III (Costeff optic atrophy) (AR) NM_025136.3 Costeff症候群	OPA3	全人種	1 in 500	99%	Reduced
		猶太	1 in 10	99%	1 in 900

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11-beta-hydroxylase-deficient congenital adrenal hyperplasia (AR) NM_000497.3 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
		猶太	1 in 40	99%	1 in 3,900
3-beta-hydroxysteroid dehydrogenase type II deficiency (congenital adrenal hyperplasia) (AR) NM_000198.3 3-β-羥基類固醇脫氫酶缺乏症-2型	HSD3B2	全人種	1 in 500	99%	Reduced
17-alpha-hydroxylase-deficient congenital adrenal hyperplasia (AR) NM_000102.3 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
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency NM_000500.7 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
Abetalipoproteinemia (AR) NM_000253.3 無β脂蛋白血症	MTTP	全人種	1 in 500	99%	Reduced
		猶太	1 in 131	99%	1 in 13,000
ACAD9 deficiency (AR) NM_014049.4 粒線體複合物 I 缺乏症第 20 型	ACAD9	全人種	1 in 500	99%	Reduced
Achromatopsia (AR) NM_019098.4 色彩感應失能症	CNGB3	全人種	1 in 93	99%	1 in 9,200
Acrodermatitis enteropathica (AR) NM_130849.3 腸病變性肢端皮膚炎	SLC39A4	全人種	1 in 354	99%	1 in 35,300
ADA-related conditions (AR) NM_000022.2 腺苷脫氨酶缺乏症	ADA	全人種	1 in 224	92%	1 in 2,788
Aicardi-Gouti è res syndrome (AR) NM_015474.3 Aicardi-Goutieres 症候群	SAMHD1	全人種	1 in 500	99%	Reduced
Alkaptonuria (AR) NM_000187.3 黑尿症	HGD	全人種	1 in 250	99%	1 in 24,900
		斯洛伐克	1 in 69	99%	1 in 6,800

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Alpha-1 antitrypsin deficiency (AR) NM_000295.4 α 1-抗胰蛋白酶缺乏症	SERPINA1	全人種	1 in 13	95%	1 in 240
		亞洲	1 in 249	95%	1 in 4,960
Alpha-mannosidosis (AR) NM_000528.3 α 型甘露糖症	MAN2B1	全人種	1 in 354	99%	1 in 35,300
Alpha-thalassemia (AR) NM_000558.4, NM_000517.4 甲型海洋性貧血	HBA1*	全人種	1 in 25	90%	1 in 241
Alpha-thalassemia (AR) NM_000558.4, NM_000517.4 甲型海洋性貧血	HBA2*	全人種	1 in 25	90%	1 in 241
Alpha-thalassemia X-linked intellectual disability syndrome (XL) NM_000489.4 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 (AR) NM_000091.4 亞伯氏症候群-COL4A3型	COL4A3	全人種	1 in 354	99%	1 in 35,300
Alport syndrome (AR) NM_000092.4 亞伯氏症候群-COL4A4型	COL4A5*	全人種	1 in 500	98%	Reduced
Alstrom syndrome (AR) NM_015120.4 Alstrom症候群	ALMS1	全人種	1 in 500	99%	Reduced
Andermann syndrome (AR) NM_133647.1 Andermann 症候群	SLC12A6	全人種	1 in 500	99%	Reduced
		法裔加拿大	1 in 23	99%	1 in 2,200
Arginase deficiency (AR) NM_000045.3 精胺酸酶缺乏症	ARG1	全人種	1 in 274	99%	1 in 27,300
Argininosuccinic aciduria (AR) NM_000048.3 精胺丁二酸酶缺乏症	ASL	全人種	1 in 133	90%	1 in 1,321

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Aromatase deficiency (AR) NM_031226.2 芳香環轉化酶缺乏症	CYP19A1	全人種	1 in 500	99%	Reduced
Asparagine synthetase deficiency (AR) NM_133436.3 天門冬醯胺酶合成缺乏症	ASNS	全人種	1 in 500	99%	Reduced
		猶太	1 in 80	99%	1 in 7,900
Aspartylglucosaminuria (AR) NM_000027.3 天冬氨醯葡萄糖胺尿症	AGA	全人種	1 in 500	99%	Reduced
		芬蘭	1 in 69	99%	1 in 6,800
Ataxia telangiectasia (AR) NM_000051.3 共濟失調微血管擴張症候群	ATM	全人種	1 in 100	99%	1 in 9,900
		猶太	1 in 69	99%	1 in 6,800
Ataxia with vitamin E deficiency (AR) NM_000370.3 共濟失調與維生素 E 缺乏症	TTPA	全人種	1 in 500	90%	Reduced
Autoimmune polyendocrinopathy with candidiasis and ectodermal dysplasia (AR) NM_000383.3 自體免疫多腺體症候群I型	AIRE	全人種	1 in 150	99%	1 in 14,900
		猶太	1 in 18	99%	1 in 1,700
Autosomal recessive deafness 77 (AR) NM_144612.6 遺傳性聽力障礙77型	LOXHD1	全人種	1 in 500	99%	Reduced
		猶太	1 in 180	99%	1 in 17,900
Autosomal recessive spastic ataxia of Charlevoix-Saguenay(ARSACS) (AR) NM_014363.5 遺傳性痙攣性共濟失調症	SACS	全人種	1 in 500	99%	Reduced
		法裔加拿大	1 in 21	99%	1 in 2,000
Bardet-Biedl syndrome (AR) NM_024649.4 Bardet-Biedl氏症候群-1型	BBS1	全人種	1 in 330	99%	1 in 32,900
Bardet-Biedl syndrome (AR) NM_031885.3 Bardet-Biedl氏症候群-2型	BBS2	全人種	1 in 560	99%	Reduced
Bardet-Biedl syndrome (AR) NM_024685.3 Bardet-Biedl氏症候群-10型	BBS10	全人種	1 in 354	99%	1 in 35,300

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Bardet-Biedl syndrome (AR) NM_152618.2 Bardet-Biedl氏症候群-12型	BBS12	全人種	1 in 708	99%	Reduced
Bartter syndrome type IV (AR) NM_057176.2 Bartter 氏症候群-4型	BSND	全人種	1 in 500	99%	Reduced
Bernard-Soulier syndrome (AR) NM_000174.4 Bernard-Soulier症候群-GP9型	GP9	全人種	1 in 500	99%	Reduced
Bernard-Soulier syndrome (GP1BA-related) (AR) NM_000173.6 Bernard-Soulier症候群-GP1BA型	GP1BA*	全人種	1 in 500	99%	Reduced
Beta-ketothiolase deficiency (AR) NM_000019.3 β-酮硫解酶缺乏症	ACAT1	全人種	1 in 500	99%	1 in 35,300
Biotinidase deficiency (AR) NM_000060.3 生物素酶缺乏症	BTD	全人種	1 in 125	99%	1 in 12,400
Bloom syndrome (AR) NM_000057.3 Bloom症候群	BLM	全人種	1 in 500	99%	Reduced
limb-girdle muscular dystrophy type 2A (AR) NM_000070.2s 肢帶型肌肉失養症-2A型	CAPN3	全人種	1 in 144	99%	1 in 14,300
Canavan disease (AR) NM_000049.2 家族性軸突海綿退化	ASPA	全人種	1 in 500	99%	Reduced
Carbamoylphosphate synthetase I deficiency (AR) NM_001875.4 氨甲醯磷酸合成酶缺失症-1型	CPS1	全人種	1 in 500	99%	Reduced
Carnitine palmitoyltransferase I deficiency (AR) NM_001876.3 肉鹼結合酶缺乏-1型	CPT1A	全人種	1 in 500	99%	Reduced

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Carnitine palmitoyltransferase I deficiency (AR) NM_000098.2 肉鹼結合酶缺乏-2型	CPT2	全人種	1 in 182	99%	1 in 18,100
Carpenter syndrome (AR) NM_183227.2 Carpenter症候群	RAB23	全人種	1 in 500	99%	Reduced
Cartilage-hair hypoplasia - anauxetic dysplasia spectrum disorders (AR) NR 003051.3 骨骼發育異常症候群	RMRP	全人種	1 in 500	99%	Reduced
Cerebrotendinous xanthomatosis (AR) NM_000784.3 腦腱性黃瘤症	CYP27A1	全人種	1 in 112	98%	1 in 5,550
Charcot-Marie-Tooth disease (AR) NM_006096.3 進行性神經性腓骨萎縮症-4D型	NDRG1	羅馬	1 in 22	99%	1 in 2,100
Charcot-Marie-Tooth disease, X-linked (XL) NM_000166.5 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
Chorea-acanthocytosis (AR) NM_033305.2 舞蹈棘狀紅血球症	VPS13A*	全人種	1 in 500	97%	Reduced
Choroideremia (XL) NM_000390.2 脈絡膜缺失症	CHM	全人種	1 in 500	95%	Reduced
Chronic granulomatous disease (AR) NM_000101.3 慢性肉芽腫病	CYBA	全人種	1 in 500	99%	Reduced
		猶太	1 in 13	99%	1 in 1,200
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
Citrin deficiency (AR) NM_014251.2 Citrin缺乏症	SLC25A13	亞洲	1 in 65	99%	1 in 6,400

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Citrullinemia type 1 (AR) NM_000050.4 瓜胺酸血症-1型	ASS1	全人種	1 in 120	96%	1 in 2,975
Cockayne syndrome type A (AR) NM_000082.3 柯凱因氏症候群-A型	ERCC8	全人種	1 in 514	99%	Reduced
Cockayne syndrome type B (AR) NM_000124.3 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
Combined malonic and methylmalonic aciduria (AR) NM_174917.4 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 (AR) NM_024996.5 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 (AR) NM_001172696.1 結合性氧化磷酸化缺乏症-3型	TSFM*	全人種	1 in 500	93%	Reduced
		芬蘭	1 in 80	93%	1 in 1,129
Combined pituitary hormone deficiency (AR) NM_014564.4 結合性腦下垂體賀爾蒙缺失-3型	LHX3	全人種	1 in 500	99%	Reduced
Combined pituitary hormone deficiency (AR) NM_006261.4 結合性腦下垂體賀爾蒙缺失-2型	PROP1	全人種	1 in 45	98%	1 in 2,200
Combined SAP deficiency (AR) NM_002778.3 結合性SAP缺乏症	PSAP	全人種	1 in 500	99%	Reduced
Congenital amegakaryocytic thrombocytopenia (AR) NM_005373.2 先天巨核細胞缺乏血小板低下症	MPL	全人種	1 in 500	99%	Reduced
		猶太	1 in 57	99%	1 in 5,600

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Congenital disorder of glycosylation (AR) NM_013339.3 先天性醣基化疾病-1c型	ALG6*	全人種	1 in 500	99%	Reduced
Congenital disorder of glycosylation (AR) NM_002435.2 先天性醣基化疾病-1b型	MPI	全人種	1 in 500	99%	Reduced
Congenital disorders of glycosylation (AR) NM_000303.2 先天性醣基化疾病-1a型	PMM2	全人種	1 in 190	99%	1 in 18,900
		高加索	1 in 60	99%	1 in 5,900
Congenital ichthyosis (AR) NM_000359.2 先天性魚鱗癬	TGM1	全人種	1 in 224	95%	1 in 4,460
Congenital insensitivity to pain with anhidrosis (AR) NM_001012331.1 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 (AR) NM_000080.3 先天性肌無力症候群-CHRNE型	CHRNE	全人種	1 in 200	99%	1 in 19,900
		羅馬	1 in 25	99%	1 in 2,400
Congenital myasthenic syndrome (AR) NM_005055.4 先天性肌無力症候群-RAPSN型	RAPSN	全人種	1 in 283	99%	1 in 28,200
Congenital neutropenia (AR) NM_006118.3 嚴重先天性嗜中性白血球減少症-HAX1型	HAX1	全人種	1 in 500	99%	Reduced
Corneal dystrophy and perceptive deafness (AR) NM_032034.3 角膜失養和感音性失聰症	SLC4A11	全人種	1 in 500	99%	Reduced
Corticosterone methyloxidase deficiency (AR) NM_000498.3 皮質酮甲基氧化酶缺乏症	CYP11B2	全人種	1 in 500	99%	Reduced
		猶太	1 in 30	99%	1 in 2,900
Cystinosis (AR) NM_004937.2 胱胺酸血症	CTNS	全人種	1 in 158	99%	1 in 15,700

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D-bifunctional protein deficiency (AR) NM_000414.3 D-雙功能蛋白缺乏症	HSD17B4	全人種	1 in 158	99%	1 in 15,700
DHDDS-related disorders (AR) NM_024887.3 視網膜色素病變-59 型	DHDDS	猶太	1 in 117	99%	1 in 11,600
Dihydrolipoamide dehydrogenase deficiency (AR) NM_000108.4 二氫硫辛醯胺脫氫酶缺乏症	DLD	全人種	1 in 500	99%	Reduced
DMD-related dystrophinopathy (XL) NM_004006.2 裘馨氏肌肉萎縮症	DMD	全人種	1 in 667	99%	Reduced
Dysferlinopathy (AR) NM_003494.3 肢帶型肌失養症 2B 型	DYSF	全人種	1 in 311	99%	1 in 31,000
Dystrophic epidermolysis bullosa (AR) NM_000094.3 表皮分解性水皰症	COL7A1	全人種	1 in 370	97%	1 in 12,300
Ehlers-Danlos syndrome VIIC (AR) NM_014244.4 埃勒斯-當洛斯症候群	ADAMTS2	全人種	1 in 500	99%	Reduced
Ellis-Van Creveld syndrome (AR) NM_147127.4 埃利偉氏症候群-2型	EVC2	全人種	1 in 152	99%	1 in 15,100
Ellis-Van Creveld syndrome (AR) NM_153717.2 埃利偉氏症候群-1型	EVC	全人種	1 in 220	99%	1 in 21,900
Emery-Dreifuss muscular dystrophy (XL) NM_000117.2 Emery-Dreifuss 肌失養症	EMD	全人種	1 in 200	99%	1 in 19,900
Retinitis pigmentosa 37 (AR) NM_014249.3 視網膜色素病變-37型	NR2E3	全人種	1 in 500	99%	Reduced

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Ethylmalonic encephalopathy (AR) NM_014297.3" 乙基丙二酸腦病變	ETHE1	全人種	1 in 500	99%	Reduced
Fabry disease (XL) NM_000169.2 法布瑞氏症	GLA	全人種	1 in 500	99%	Reduced
Factor IX deficiency/ Hemophilia B (XL) NM_000133.3 血友病 B 型	F9	全人種	1 in 500	99%	Reduced
Factor V Leiden (AD) NM_000130.4 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) NM_000128.3 第11凝血因子缺乏症	F11	全人種	1 in 500	99%	Reduced
		猶太	1 in 11	99%	1 in 1,000
Familial dysautonomia (AR) NM_003640.3 家族性自主神經失調症-IKBKAP型	IKBKAP	全人種	1 in 500	99%	Reduced
Familial dysautonomia (AR) NM_003640.3 家族性自主神經失調症-ELP1型	ELP1	全人種	1 in 500	99%	Reduced
		猶太	1 in 36	99%	1 in 3,500
Familial hypercholesterolemia (AR/AD) NM_000527.4 家族性高膽固醇血症-LDLR型	LDLR	全人種	1 in 250	99%	1 in 24,900
Familial hypercholesterolemia (AR) NM_015627.2 家族性高膽固醇血症-LDLRAP型	LDLRAP1	全人種	1 in 500	99%	Reduced
Familial hyperinsulinism (AR) NM_000352.4 (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
Familial hyperinsulinism (AR) NM_000525.3 家族性胰島素過多症-KCNJ11型	KCNJ11	全人種	1 in 500	99%	Reduced

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Familial Mediterranean fever (AR) NM_000243.2 家族性地中海熱	MEFV	全人種	1 in 64	90%	1 in 631
Fanconi anemia type A (AR) NM_000135.2 Fanconi氏貧血-A型	FANCA	全人種	1 in 345	99%	1 in 34,400
Fanconi anemia type C (AR) NM_000136.2 Fanconi氏貧血-C型	FANCC	全人種	1 in 417	99%	1 in 41,600
Fanconi anemia type G (AR) NM_004629.1 Fanconi氏貧血-G型	FANCG	全人種	1 in 500	99%	Reduced
Fumarate hydratase deficiency (AR) NM_000143.3 延胡索酸酶缺乏症	FH	全人種	1 in 500	99%	Reduced
Galactokinase deficiency galactosemia (AR) NM_000154.1 半乳糖激酶缺乏症	GALK1	全人種	1 in 122	99%	1 in 12,100
Galactosemia (AR) NM_000155.3 半乳糖血症	GALT	全人種	1 in 100	99%	1 in 9,900
Gaucher disease (AR) NM_001005741.2 高雪氏症	GBA*	全人種	1 in 158	72%	1 in 561
Gitelman syndrome (AR) NM_000339.2 Gitelman症候群	SLC12A3	全人種	1 in 100	99%	1 in 9,900
GJB2-related DFNB1 nonsyndromic hearing loss and deafness (AR) NM_004004.5 感覺神經性聽損-GJB2型	GJB2	全人種	1 in 50	99%	1 in 4,900
Glucose-6-phosphate dehydrogenase deficiency (XL) NM_001042351.2 蠶豆症	G6PD	全人種	1 in 10	99%	1 in 900

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Glutaric acidemia type I (AR) NM_000159.3 戊二酸血症-1型	GCDH	全人種	1 in 87	99%	1 in 8,600
Glutaric acidemia type II (AR) NM_000126.3 戊二酸血症-2A型	ETFA	全人種	1 in 500	99%	Reduced
Glutaric acidemia type II (AR) NM_004453.3 戊二酸血症-2C型	ETFDH	全人種	1 in 250	99%	1 in 24,900
Glycine encephalopathy (AR) NM_000481.3 非酮性高甘胺酸血症-AMT型	AMT	全人種	1 in 325	99%	1 in 32,400
Glycine encephalopathy (AR) NM_000170.2 非酮性高甘胺酸血症-GLDC型	GLDC	全人種	1 in 165	99%	1 in 16,400
Glycogen storage disease type Ia (AR) NM_000151.3 肝醣儲積症-1A型	G6PC	全人種	1 in 177	95%	1 in 3,520
Glycogen storage disease type Ib (AR) NM_001164277.1 肝醣儲積症-1B型	SLC37A4	全人種	1 in 354	95%	1 in 7,060
Glycogen storage disease type II (Pompe disease) (AR) NM_000152.3 龐貝氏症	GAA	全人種	1 in 100	99%	1 in 9,900
Glycogen storage disease type III (AR) NM_000642.2 肝醣儲積症-3型	AGL	全人種	1 in 159	95%	1 in 3,160
Glycogen storage disease type IV (AR) NM_000158.3 肝醣儲積症-4型	GBE1	全人種	1 in 387	99%	1 in 38,600
Glycogen storage disease type V (AR) NM_005609.3 肝醣儲積症-5型	PYGM	高加索	1 in 158	99%	1 in 15,700

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Glycogen storage disease type VII (AR) NM_000289.5 肝醣儲積症-7型	PFKM	全人種	1 in 500	99%	Reduced
GRACILE syndrome/BCS1L-related disorders (AR) NM_004328.4 GRACILE 症候群	BCS1L	全人種	1 in 500	99%	Reduced
Guanidinoacetate methyltransferase deficiency (AR) NM_000156.5 胍基乙酸甲基轉移酶缺乏症	GAMT	全人種	1 in 500	99%	Reduced
HBB-related hemoglobinopathies (AR) NM_000518.4 乙型地中海貧血	HBB	全人種	1 in 49	99%	1 in 4,800
Hereditary fructose intolerance (AR) NM_000035.3 遺傳性果糖不耐症	ALDOB	全人種	1 in 122	99%	1 in 12,100
Hereditary hemochromatosis (AR) NM_000410.3 血鐵沉積症-1型	HFE	北歐	1 in 9	99%	1 in 800
Hereditary hemochromatosis (AR) NM_213653.3 血鐵沉積症-2型	HFE2	全人種	1 in 500	99%	Reduced
Hereditary hemochromatosis type 2 (HJV-related) (AR) NM_213653.3 血鐵沉積症-2A型	HJV	全人種	1 in 500	99%	Reduced
Hereditary hemochromatosis (AR) NM_003227.3 血鐵沉積症-3型	TFR2	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome (AR) NM_000195.4 Hermansky-Pudlak 症候群-1型	HPS1	全人種	1 in 500	99%	Reduced
Hermansky-Pudlak syndrome (AR) NM_032383.4 Hermansky-Pudlak 症候群-3型	HPS3	全人種	1 in 500	99%	Reduced

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Holocarboxylase synthetase deficiency (AR) NM_000411.6 多發性羧化酶缺乏症	HLCS	全人種	1 in 224	99%	1 in 22,300
Homocystinuria (AR) NM_000071.2 高胱胺酸尿症	CBS	全人種	1 in 224	99%	1 in 22,300
Homocystinuria due to MTHFR deficiency (AR) NM_005957.4 高胱胺酸尿症-MTHFR缺乏型	MTHFR*	猶太	1 in 39	99%	1 in 38,00
Homocystinuria, cobalamin E type (AR) NM_002454.2 高胱胺酸尿症-cobalamin E型	MTRR	全人種	1 in 500	99%	Reduced
Hydrolethalus syndrome type 1 (AR) NM_145014.2 Hydrolethalus症候群	HYLS1	全人種	1 in 500	99%	Reduced
Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (AR) NM_014252.3 鳥氨酸-高血氨-高瓜胺酸綜合症候群	SLC25A15	全人種	1 in 500	99%	Reduced
Hypohidrotic ectodermal dysplasia (XL) NM_001399.4 少汗性外胚層發育不良症3型	EDA	全人種	1 in 112	99%	1 in 11,100
Hypophosphatasia (AR) NM_000478.5 低磷酸酯酶症	ALPL	全人種	1 in 150	95%	1 in 2,980
Inclusion body myopathy 2 (AR) NM_001128227.2 包涵體肌炎	GNE	全人種	1 in 179	99%	1 in 17,800
Isovaleric acidemia (AR) NM_002225.3 異戊酸血症	IVD	全人種	1 in 250	99%	1 in 24,900
Joubert syndrome 2 (AR) NM_001173990.2 Joubert 症候群-2型	TMEM216	全人種	1 in 500	99%	Reduced
		猶太	1 in 92	99%	1 in 9,100

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Junctional epidermolysis bullosa (AR) NM_000227.4 接合性表皮溶解水皰症-LAMA3型	LAMA3	全人種	1 in 500	99%	Reduced
Junctional epidermolysis bullosa (AR) NM_000228.2 接合性表皮溶解水皰症-LAMB3型	LAMB3	全人種	1 in 317	99%	1 in 31,600
Junctional epidermolysis bullosa (AR) NM_005562.2 接合性表皮溶解水皰症-LAMC2型	LAMC2	全人種	1 in 500	99%	Reduced
Krabbe disease (AR) NM_000153.3 Krabbe 症	GALC*	全人種	1 in 158	99%	1 in 15,700
LAMA2-related muscular dystrophy (AR) NM_000426.3 肌肉失養症-LAMA2型	LAMA2	全人種	1 in 87	99%	1 in 8,600
Leber congenital amaurosis 2 (AR) NM_000329.2 萊伯氏先天性黑矇症-2型	RPE65	Pan-ethnic	1 in 228	99%	1 in 22,700
Leber congenital amaurosis 5 (AR) NM_181714.3 萊伯氏先天性黑矇症-5型	LCA5	Pan-ethnic	1 in 645	97%	Reduced
Leber congenital amaurosis 8 (AR) NM_201253.2 萊伯氏先天性黑矇症-8型	CRB1	Pan-ethnic	1 in 112	99%	1 in 11,100
Leber congenital amaurosis 10 (AR) NM_025114.3 萊伯氏先天性黑矇症-10型	CEP290	Pan-ethnic	1 in 185	99%	1 in 18,400
Leber congenital amaurosis 13 (AR) NM_152443.2 萊伯氏先天性黑矇症-13型	RDH12	Pan-ethnic	1 in 460	99%	1 in 45,900
Leigh syndrome, French Canadian type (AR) NM_133259.3 Leigh症候群-法國/加拿大型	LRPPRC	全人種	1 in 500	99%	Reduced

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Lethal congenital contracture syndrome 1 (AR)NM_001003722.1 致死先天性攣縮症候群	GLE1	全人種	1 in 500	99%	Reduced
Leukoencephalopathy with vanishing white matter (AR) NM_003907.2 腦白質病伴隨白質消失症	EIF2B5	全人種	1 in 500	99%	Reduced
Limb-girdle muscular dystrophy type 2C (AR) NM_000231.2 肢帶型肌肉失養症-2C型	SGCG	全人種	1 in 500	99%	Reduced
		羅馬	1 in 59	95%	1 in 5,800
Limb-girdle muscular dystrophy type 2D (AR) NM_000023.2 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) NM_000232.4 肢帶型肌肉失養症-2E 型	SGCB	全人種	1 in 500	92%	Reduced
Lipoid congenital adrenal hyperplasia (AR) NM_000349.2 脂肪性先天性腎上腺皮質增生症	STAR	全人種	1 in 500	99%	Reduced
Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (AR) NM_000182.4 長鏈 3-羥烷基輔酶A脱氫酶缺乏症	HADHA	全人種	1 in 350	99%	1 in 34,900
Lysinuric protein intolerance (AR) NM_001126106.2 Lysunyruc 蛋白質耐受不良症	SLC7A7	全人種	1 in 500	99%	Reduced
Lysosomal acid lipase deficiency (AR) NM_000235.3 溶酶體酸性脂肪酶缺乏症	LIPA	高加索	1 in 112	94%	1 in 1,850
Major histocompatibility complex class II deficiency (AR) NM_000246.3 裸淋巴球症候症-2型	CIITA	全人種	1 in 500	99%	Reduced
Maple syrup urine disease type 1a (AR) NM_000709.3 楓糖尿症-1a型	BCKDHA	全人種	1 in 373	99%	1 in 37,200

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Maple syrup urine disease type 1b (AR) NM_183050.2 楓糖尿症-1b型	BCKDHB	全人種	1 in 346	99%	1 in 34,500
Maple syrup urine disease type 2 (AR) NM_001918.3 楓糖尿症-2型	DBT	全人種	1 in 500	99%	Reduced
Medium chain acyl-coa dehydrogenase deficiency (AR) NM_000016.5 中鏈醯輔酶 A 去氫酶缺乏症	ACADM	全人種	1 in 66	99%	1 in 6,500
Megalencephalic leukoencephalopathy with subcortical cysts type 1 (AR) NM_015166.3 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
Menkes disease/ATP7A-related disorders (XL) NM_000052.6 Menkes氏症候群	ATP7A	全人種	1 in 500	99%	Reduced
Metachromatic leukodystrophy (AR) NM_000487.5 異染性腦白質退化症	ARSA	全人種	1 in 100	95%	1 in 1,980
Methylmalonic acidemia (AR) NM_172250.2 甲基丙二酸血症-cb1A型	MMAA	全人種	1 in 316	97%	1 in 10,500
Methylmalonic acidemia (AR) NM_052845.3 甲基丙二酸血症-cb1B型	MMAB	全人種	1 in 456	98%	1 in 22,750
Methylmalonic acidemia (AR) NM_000255.3 甲基丙二酸血症-MUT型	MUT	全人種	1 in 204	96%	1 in 5,075
Methylmalonic acidemia with homocystinuria, cobalamin C type (AR) NM_015506.2 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
Methylmalonic acidemia with homocystinuria, cobalamin D type (AR) NM_015702.2 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

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Microphthalmia/clinical anophthalmia (AR) NM_182894.2 小眼症	VSX2	全人種	1 in 500	99%	Reduced
Mitochondrial complex I deficiency/Leigh syndrome (AR) NM_024120.4 Leigh症候群-NDUFAF5型	NDUFAF5	全人種	1 in 500	99%	Reduced
Mitochondrial complex I deficiency/Leigh syndrome (AR) NM_004553.4 Leigh症候群-NDUFS6型	NDUFS6	全人種	1 in 500	99%	Reduced
Mitochondrial DNA depletion syndrome (AR) NM_002437.4 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
Mitochondrial myopathy and sideroblastic anemia 1 (AR) NM_025215.5 線粒體肌病和鐵粒細胞性貧血	PUS1	全人種	1 in 500	99%	Reduced
Mitochondrial neurogastrointestinal encephalopathy disease (AR) NM_001953.4 粒線體性神經胃腸腦病變症候群	TYMP	全人種	1 in 500	99%	Reduced
MKS1-related disorders (AR) NM_017777.3 MKS1相關疾病	MKS1	全人種	1 in 260	95%	1 in 5,180
Mucolipidosis type II/III (AR) NM_024312.4 黏脂質症 -2/3型	GNPTAB	全人種	1 in 200	99%	1 in 19,900
Mucolipidosis type III (AR) NM_032520.4 黏脂質症 -3型	GNPTG	全人種	1 in 500	99%	Reduced
Mucolipidosis type IV (AR) NM_020533.2 黏脂質症 -4型	MCOLN1	全人種	1 in 500	99%	Reduced
Mucopolysaccharidosis type I (AR) NM_000203.4 黏多醣症-1型	IDUA	全人種	1 in 148	97%	1 in 4,900

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Mucopolysaccharidosis type II (Hunter syndrome) (XL) NM_000202.6 黏多醣症-2型	IDS*	全人種	1 in 500	90%	Reduced
Mucopolysaccharidosis type IIIA (Sanfilippo A syndrome) (AR) NM_000199.3 黏多醣症-3A型	SGSH	全人種	1 in 215	99%	1 in 21,400
Mucopolysaccharidosis type IIIB (AR) NM_000263.3 黏多醣症-3B型	NAGLU	全人種	1 in 224	99%	1 in 22,300
Mucopolysaccharidosis type IIIC / Retinitis pigmentosa 73 (AR) NM_152419.2 黏多醣症-3C型	HGSNAT	全人種	1 in 500	99%	Reduced
Mucopolysaccharidosis type IIID (Sanfilippo syndrome) (AR) NM_002076.3 黏多醣症-3D型	GNS	全人種	1 in 500	99%	Reduced
Mucopolysaccharidosis type IVB / GM1 gangliosidosis (AR) NM_000404.2 黏多醣症-4B型	GLB1	全人種	1 in 158	99%	1 in 15,700
Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome) (AR) NM_000046.3 黏多醣症-6型	ARSB	全人種	1 in 250	99%	1 in 24,900
Mucopolysaccharidosis type IX (AR) NM_153281.1 黏多醣症-9型	HYAL1	全人種	1 in 500	99%	Reduced
Multiple sulfatase deficiency (AR) NM_182760.3 多發性硫酸脂酶缺乏症	SUMF1	全人種	1 in 500	99%	Reduced
N-acetylglutamate synthase deficiency (AR) NM_153006.2 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) NM_001271208.1 桿狀體肌症-2型	NEB*	全人種	1 in 158	95%	1 in 3,140

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Nephrogenic diabetes insipidus (AR) NM_000486.5 腎性尿崩症	AQP2	全人種	1 in 1118	99%	Reduced
Nephrotic syndrome/congenital Finnish nephrosis (AR) NM_004646.3 先天性腎病症候群-1型	NPHS1	全人種	1 in 500	99%	Reduced
Nephrotic syndrome/steroid-resistant nephrotic syndrome (AR) NM_014625.3 先天性腎病症候群-2型	NPHS2	全人種	1 in 500	99%	Reduced
Neuronal ceroid-lipofuscinosis (AR) NM_001042432.1 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
Neuronal ceroid-lipofuscinosis (AR) NM_006493.2 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 (AR) NM_017882.2 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 (AR) NM_152778.2 神經元蠟樣脂褐質沉著症-MFSD8型	MFSD8	全人種	1 in 500	99%	Reduced
Neuronal ceroid-lipofuscinosis (AR) NM_000310.3 神經元蠟樣脂褐質沉著症-PPT1型	PPT1	全人種	1 in 119	98%	1 in 9,900
Neuronal ceroid-lipofuscinosis (AR) NM_000391.3 神經元蠟樣脂褐質沉著症-TPP1型	TPP1	全人種	1 in 250	97%	1 in 8,300
Neuronal ceroid-lipofuscinosis (AR) NM_018941.3 神經元蠟樣脂褐質沉著症-CLN8型	CLN8	全人種	1 in 500	99%	Reduced
Niemann-pick disease type A/B (AR) NM_000543.4 尼曼匹克症-A/B型	SMPD1	全人種	1 in 250	95%	1 in 4,980

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Niemann-pick disease type C (AR) NM_000271.4 尼曼匹克症-C1型	NPC1	全人種	1 in 183	99%	1 in 18,200
Niemann-pick disease type C (AR) NM_006432.3 尼曼匹克症-C2型	NPC2	全人種	1 in 871	99%	Reduced
Nijmegen breakage syndrome (AR) NM_002485.4 Nijmegen破損症候群	NBN*	全人種	1 in 500	99%	Reduced
Ornithine aminotransferase deficiency (AR) NM_000274.3 鳥胺酸酮酸轉胺酶缺乏症	OAT*	全人種	1 in 500	99%	Reduced
Ornithine transcarbamylase deficiency (XL) NM_000531.5 鳥胺酸氨甲醯基轉移酶缺乏症	OTC	全人種	1 in 250	85%	1 in 8,300
Osteopetrosis (AR) NM_006019.3 骨質石化症	TCIRG1	全人種	1 in 317	99%	1 in 31,600
Pendred syndrome (AR) NM_000441.1 感覺神經性聽損-SLC26A4型	SLC26A4	全人種	1 in 80	99%	1 in 7,900
Peroxisomal acyl-coa oxidase deficiency (AR) NM_004035.6 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
Phenylalanine hydroxylase deficiency (AR) NM_000277.1 苯酮尿症	PAH	全人種	1 in 58	99%	1 in 5,700
Phosphoglycerate dehydrogenase deficiency/ Neu-Laxova syndrome (AR) NM_006623.3 磷酸甘油酸脱氫酶缺乏症	PHGDH	全人種	1 in 500	99%	Reduced
Polycystic kidney disease (AR) NM_138694.3 隱性多囊腎病-PKHD1型	PKHD1	全人種	1 in 70	99%	1 in 6,900

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Polymicrogyria (AR) NM_005682.6 雙側額頂葉多小腦迴畸形症	ADGRG1	全人種	1 in 500	99%	Reduced
POMGNT1-related disorders (AR) NM_017739.3 POMGNT1相關症	POMGNT1	全人種	1 in 500	99%	Reduced
Pontocerebellar hypoplasia (AR) NM_020320.3 橋腦小腦發育不全-6型	RARS2	全人種	1 in 500	99%	Reduced
Pontocerebellar hypoplasia (AR) NM_016955.3 橋腦小腦發育不全-2D型	SEPSECS	全人種	1 in 500	99%	Reduced
Pontocerebellar hypoplasia (AR) NM_003384.2 橋腦小腦發育不全-1A型	VRK1	全人種	1 in 500	99%	Reduced
Postnatal progressive microcephaly 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
Primary carnitine deficiency (AR) NM_003060.3 原發性肉鹼缺乏症	SLC22A5	全人種	1 in 500	99%	Reduced
Primary ciliary dyskinesia (AR) NM_001369.2 原發性纖毛運動障礙-DNAH5型	DNAH5	全人種	1 in 109	99%	1 in 10,800
Primary ciliary dyskinesia (AR) NM_012144.3 原發性纖毛運動障礙-DNAI1型	DNAI1	全人種	1 in 250	99%	1 in 24,900
Primary ciliary dyskinesia (AR) NM_023036.4 原發性纖毛運動障礙-DNAI2型	DNAI2	全人種	1 in 354	99%	1 in 35,300
Primary hyperoxaluria type 1 (AR) NM_000030.2 原發性高草酸尿症 1型	AGXT	全人種	1 in 135	99%	1 in 13,400

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Primary hyperoxaluria type 2 (AR) NM_012203.1 原發性高草酸尿症 2型	GRHPR	全人種	1 in 500	99%	Reduced
Primary hyperoxaluria type 3 (AR) NM_138413.3 原發性高草酸尿症 3 型	HOGA1	全人種	1 in 354	99%	1 in 35,300
Progressive familial intrahepatic cholestasis type 2 (AR) NM_003742.2 進行性家族性肝內膽汁滯留症第二型	ABCB11	全人種	1 in 100	99%	1 in 9,900
Propionic acidemia (AR) NM_000282.3 丙酸血症-PCCA型	PCCA	全人種	1 in 224	96%	1 in 22,300
Propionic acidemia (AR) NM_000532.4 丙酸血症-PCCB型	PCCB	全人種	1 in 224	96%	1 in 22,300
Prothrombin-related thrombophilia (AD) NM_000506.3 凝血酶原相關血栓病	F2*	全人種	1 in 62	99%	1 in 6,100
PRPS1-related disorders (XL) NM_002764.3 PRPS1相關疾病	PRPS1	全人種	1 in 500	99%	Reduced
Pycnodysostosis (AR) NM_000396.3 緻密性成骨不全症	CTSK	全人種	1 in 438	99%	1 in 43,700
Pyruvate carboxylase deficiency (AR) NM_000920.3 丙酮酸羧化酶缺乏症	PC	全人種	1 in 250	95%	1 in 4,980
Pyruvate dehydrogenase deficiency (AR) NM_000925.3 乙型丙酮酸鹽脱氫酶缺乏症	PDHB	全人種	1 in 500	99%	Reduced
Pyruvate dehydrogenase deficiency (XL) NM_000284.3 甲型丙酮酸鹽脱氫酶缺乏症	PDHA1	全人種	1 in 500	99%	Reduced

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Renal tubular acidosis with deafness (AR) NM_001692.3 腎小管酸中毒/耳聾	ATP6V1B1	全人種	1 in 500	99%	Reduced
		猶太	1 in 140	99%	1 in 13,900
Retinitis pigmentosa 25 (AR) NM_001142800.1 視網膜色素病變-25型	EYS	全人種	1 in 129	99%	1 in 12,800
Retinitis pigmentosa 26 (AR) NM_001030311.2 視網膜色素病變-26型	CERKL	全人種	1 in 137	99%	1 in 13,600
Retinitis pigmentosa 28 (AR) NM_001201543.1 視網膜色素病變-28型	FAM161A	全人種	1 in 289	99%	1 in 28,800
Rhizomelic chondrodysplasia punctata type 1 (AR) NM_000288.3 肢近端型點狀軟骨發育不良-1型	PEX7	全人種	1 in 157	95%	1 in 15,600
Rhizomelic chondrodysplasia punctata type 3 (AR) NM_003659.3 肢近端型點狀軟骨發育不良-3型	AGPS	全人種	1 in 500	99%	Reduced
Roberts syndrome (AR) NM_001017420.2 Roberts症候群	ESCO2	全人種	1 in 500	99%	Reduced
RPGRIP1L-related disorders (AR) NM_015272.2 RPGRIP1L相關疾病	RPGRIP1L*	全人種	1 in 259	95%	1 in 5,160
RTEL1-related disorders (AR) NM_001283009.1 RTEL1相關疾病	RTEL1	全人種	1 in 500	99%	Reduced
Sandhoff disease (AR) NM_000521.3 Sandoff症	HEXB	全人種	1 in 180	99%	1 in 17,900
Schimke immuno-osseous dysplasia (AR) NM_014140.3 Schimke免疫-骨發育不良	SMARCAL1	全人種	1 in 500	99%	Reduced

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Severe combined immunodeficiency (AR) NM_001033855.2 嚴重結合免疫缺乏症-DCLRE1C型	DCLRE1C	全人種	1 in 500	99%	Reduced
Severe combined immunodeficiency/ Omenn syndrome (AR) 嚴重結合免疫缺乏症-RAG2型	RAG2	全人種	1 in 500	99%	Reduced
Severe congenital neutropenia (AR) NM_007259.4 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 disorders (AR) NM_012434.4 唾液酸貯積症	SLC17A5	全人種	1 in 500	99%	Reduced
Sjögren-Larsson syndrome (AR) NM_000382.2 Sjögren-Larsson症候群	ALDH3A2	全人種	1 in 500	99%	Reduced
SLC26A2-related disorders (AR) NM_000112.3 SLC26A2相關疾病	SLC26A2	全人種	1 in 158	95%	1 in 15,700
SLC35A3-related disorder (AR) NM_012243.2 SLC35A3相關疾病	SLC35A3	全人種	1 in 500	99%	Reduced
Smith-Lemli-Opitz syndrome (AR) NM_001360.2 Smith-Lemli-Opitz 症候群	DHCR7	全人種	1 in 71	96%	1 in 1,750
Spastic paraplegia type 15 (AR) NM_015346.3 痙攣性下身麻痺-15型	ZFYVE26	全人種	1 in 500	99%	Reduced
Spastic paraplegia type 49 (AR) NM_014844.3 痙攣性下身麻痺-49型	TECPR2	猶太	1 in 38	99%	1 in 3,700
Spondylothoracic dysostosis (AR) NM_001039958.1 脊椎肋骨發育不全	MESP2	全人種	1 in 224	99%	1 in 22,300

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Steel syndrome (AR) NM_032888.3 鋼鐵症候群	COL27A1*	全人種	1 in 500	99%	Reduced
Stuve-Wiedemann syndrome (AR) NM_002310.5 Stuve-Wiedemann 症候群	LIFR	全人種	1 in 500	99%	Reduced
Tay-Sachs disease/hexosaminidase A deficiency (AR) NM_000520.4 薩克斯症	HEXA	全人種	1 in 250	99%	1 in 24,900
		法裔加拿大	1 in 27	99%	1 in 2,600
Tetrahydrobiopterin deficiency (AR) NM_000317.2 苯酮尿症-PTS型	PTS	全人種	1 in 433	99%	1 in 43,200
Transient infantile liver failure (AR) NM_018006.4 急性新生兒肝衰竭	TRMU	全人種	1 in 500	99%	Reduced
		猶太	1 in 34	99%	1 in 3,300
Tyrosine hydroxylase deficiency (AR) NM_199292.2 酪胺酸羥化酶缺乏症	TH	全人種	1 in 500	99%	Reduced
Tyrosinemia type I (AR) NM_000137.2 酪胺酸血症-1型	FAH*	全人種	1 in 125	95%	1 in 2,480
Tyrosinemia type II (AR) NM_000353.2 酪胺酸血症-2型	TAT	全人種	1 in 250	99%	1 in 24,900
Usher syndrome type IB/ MYO7A-related disorders (AR) NM_000260.3 尤塞氏症候群- 1B 型	MYO7A	全人種	1 in 200	95%	1 in 3,980
Usher syndrome type IC/USH1C-related disorders (AR) NM_005709.3 尤塞氏症候群- 1C 型	USH1C*	全人種	1 in 353	90%	1 in 3,521
Usher syndrome type ID (AR) NM_022124.5 尤塞氏症候群- 1D 型	CDH23*	全人種	1 in 202	95%	1 in 20,100

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Usher syndrome type IF/PCDH15-related disorders (AR) NM_033056.3 尤塞氏症候群- 1F 型	PCDH15	全人種	1 in 400	99%	1 in 39,900
Usher syndrome type IIA/USH2A-related disorders (AR) NM_206933.2 尤塞氏症候群- 2A 型	USH2A	全人種	1 in 158	99%	1 in 15,700
Usher syndrome type IIIA (AR) NM_174878.2 尤塞氏症候群- 3A 型	CLRN1	全人種	1 in 533	99%	Reduced
Very long-chain acyl-coa dehydrogenase deficiency (AR) NM_000018.3 特長鏈醯輔酶 A 去氫酶缺乏症	ACADVL	全人種	1 in 100	99%	1 in 9,900
Walker-Warburg syndrome/FKRP-related disorders (AR) NM_024301.4 Walker-Warburg症候群	FKRP	全人種	1 in 158	99%	1 in 15,700
Walker-Warburg syndrome/ FKTN-related disorders (AR) NM_001079802.1 Walker-Warburg 綜合症	FKTN	全人種	1 in 500	99%	Reduced
		日本	1 in 188	90%	1 in 18,700
Wilson disease (AR) NM_000053.3 威爾森氏症	ATP7B	全人種	1 in 90	98%	1 in 4,450
WNT10A-related disorders (AR) NM_025216.2 WNT10A相關疾病	WNT10A	全人種	1 in 305	99%	1 in 30,400
X-linked adrenoleukodystrophy (XL) NM_000033.3 性聯遺傳腎上腺白質退化症	ABCD1	猶太	1 in 500	99%	Reduced
X-linked creatine transporter deficiency (XL) NM_005629.3 性聯遺傳肌酸缺乏症	SLC6A8	全人種	1 in 500	99%	Reduced
X-linked juvenile retinoschisis (XL) NM_000330.3 性聯遺傳視網膜裂損症	RS1	全人種	1 in 500	99%	Reduced

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X-linked myotubular myopathy (XL) NM_000252.2 性聯遺傳肌小管病變	MTM1	全人種	1 in 500	96%	Reduced
X-linked severe combined immunodeficiency (XL) NM_000206.2 性聯遺傳嚴重免疫缺陷	IL2RG	全人種	1 in 500	99%	Reduced
Xeroderma pigmentosum complemetation group A (AR) NM_000380.3 著色性乾皮症-A型	XPA	日本	1 in 100	99%	1 in 9,900
Xeroderma pigmentosum complemetation group C (AR) NM_004628.4 著色性乾皮症-C型	XPC	全人種	1 in 763	99%	Reduced
Zellweger spectrum disorder (AR) NM_000466.2 柴爾維格氏症候群-PEX1型	PEX1	全人種	1 in 144	98%	1 in 7,150
Zellweger spectrum disorder (AR) NM_000318.2 柴爾維格氏症候群-PEX2型	PEX2	全人種	1 in 500	99%	Reduced
Zellweger spectrum disorder (AR) NM_000287.3 柴爾維格氏症候群-PEX6型	PEX6	全人種	1 in 294	99%	1 in 29,300
		法裔加拿大	1 in 55	99%	1 in 5,400
Zellweger spectrum disorder (AR) NM_153818.1 柴爾維格氏症候群-PEX10型	PEX10	全人種	1 in 606	94%	Reduced
Zellweger spectrum disorder (AR) NM_000286.2 柴爾維格氏症候群-PEX12型	PEX12	全人種	1 in 409	99%	1 in 40,800