Uni-ECS gene panel

代謝系統							
疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
11β-羥化酶缺乏症	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	AR	*CYP11B1*	1/158	1/15800	1/998560000	1/63200
17α-羥化酶缺乏所致先天性腎上腺皮質增生症	17-alpha-hydroxylase	AR	*CYP17A1*	1/500	1/50000	1/10000000000	1/200000
17β-羥類固醇去氫酶 3 型缺乏症	Pseudohermaphroditism, male, with gynecomastia	AR	*HSD17B3*	1/135	1/13500	1/729000000	1/54000
過氧化物酶體生物發生障礙1A 型	Peroxisome biogenesis disorder 1A (Zellweger)	AR	*PEX1*	1/500	1/50000	1/10000000000	1/200000
過氧化物酶體生物合成障礙3A 型	Peroxisome biogenesis disorder 3A (Zellweger)	AR	*PEX12*	1/373	1/37300	1/5565160000	1/149200
過氧化物酶體生物發生障礙4A 型	Peroxisome biogenesis disorder 4A (Zellweger)	AR	*PEX6*	1/280	1/28000	1/3136000000	1/112000
過氧化物酶體生物合成障礙5A 型	Peroxisome biogenesis disorder 5A (Zellweger)	AR	*PEX2*	1/500	1/50000	1/10000000000	1/200000
過氧化物酶體生物合成障礙6A 型	Peroxisome biogenesis disorder 6A (Zellweger)	AR	*PEX10*	1/500	1/3571	1/51020408	1/14286
2,4-二烯醯輔酶A 還原酵素缺乏症	2,4-dienoyl-CoA reductase deficiency	AR	*NADK2*	<1/500	1/50000	1/10000000000	1/200000
21-羥化酶缺陷型先天性腎上腺增生	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency	AR	*CYP21A2*	1/61	1/165	1/108722	1/659
2-甲基丁醯甘氨酸尿症	2-methylbutyrylglycinuria	AR	*ACADSB*	<1/500	1/50000	1/10000000000	1/200000
3β-羥基類固醇去氫酶 2 缺乏所致先天性腎上腺皮質增生症	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR	*HSD3B2*	<1/500	1/50000	1/10000000000	1/200000
3-甲基巴豆醯輔酶A 羧化酶1 缺乏症	3-Methylcrotonyl-CoA carboxylase 1 deficiency	AR	*MCCC1*	1/95	1/4750	1/90250000	1/19000
3-甲基巴豆醯輔酶A 羧化酶2 缺乏症	3-Methylcrotonyl-CoA carboxylase 2 deficiency	AR	*MCCC2*	1/95	1/1583	1/10027778	1/6333
3-甲基戊烯二酸尿症I 型	3-methylglutaconic aciduria, type I	AR	*AUH*	3/500	1/16667	1/1111111111	1/66667
3-甲基戊烯二酸尿症III 型	3-methylglutaconic aciduria, type III	AR	*OPA3*	<1/500	1/50000	1/10000000000	1/200000
3-甲基戊烯二酸尿症 V 型	3-methylglutaconic aciduria, type V	AR	*DNAJC19*	3/500	1/16667	1/1111111111	1/66667
3-甲基戊烯二酸尿症VII 型	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia	AR	*CLPB*	3/500	1/16667	1/1111111111	1/66667
3-甲基戊烯二酸尿症VIII 型	3-methylglutaconic aciduria, type VIII	AR	*HTRA2*	3/500	1/16667	1/1111111111	1/66667
3-甲基戊烯二酸尿症IX 型	3-methylglutaconic aciduria, type IX	AR	*TIMM50*	3/500	1/1515	1/9182736	1/6061
3-甲基戊烯二酸尿症伴耳聾、腦病及Leigh 樣綜合徵	3-methylglutaconic aciduria with deafness,	AR	*SERAC1*	3/500	1/2381	1/22675737	1/9524

	encephalopathy, and Leigh-like syndrome						
3-羥基-3-甲基戊二醯輔酵素A 合成酶2 缺乏症	HMG-CoA synthase-2 deficiency	AR	*HMGCS2*	<1/250	1/25000	1/2500000000	1/100000
3-羥基醯基輔酶A 去氫酶缺乏症	3-hydroxyacyl-CoA dehydrogenase deficiency	AR	*HADH*	1/240	1/6250	1/156250000	1/25000
甘胺酸腦病，AMT 相關	Glycine encephalopathy	AR	*AMT*	1/373	1/37300	1/5565160000	1/149200
甘胺酸腦病，GLDC 相關	Glycine encephalopathy	AR	*GLDC*	1/193	1/19300	1/1489960000	1/77200
BH4 缺乏性高苯丙胺酸血症A 型	Hyperphenylalaninemia, BH4-deficient, A	AR	*PTS*	<1/500	1/25000	1/2500000000	1/100000
BH4 缺乏性高苯丙胺酸血症B 型	Hyperphenylalaninemia, BH4-deficient, B	AR	*GCH1*	<1/500	1/2941	1/34602076	1/11765
BH4 缺乏性高苯丙胺酸血症C 型	Hyperphenylalaninemia, BH4-deficient, C	AR	*QDPR*	<1/500	1/50000	1/10000000000	1/200000
BH4 缺乏性高苯丙胺酸血症D 型	Hyperphenylalaninemia, BH4-deficient, D	AR	*PCBD1*	1/200	1/20000	1/1600000000	1/80000
D-雙功能蛋白缺乏症	D-bifunctional protein deficiency	AR	*HSD17B4*	1/158	1/2257	1/20378776	1/9029
HMG-CoA 裂解酶缺乏症	HMG-CoA lyase deficiency	AR	*HMGCL*	<1/500	1/50000	1/10000000000	1/200000
肝醣累積病Ia 型	Glycogen storage disease Ia	AR	*G6PC*	1/177	1/17700	1/1253160000	1/70800
肝醣累積病Ib 型	Glycogen storage disease Ib	AR	*SLC37A4*	1/158	1/5267	1/110951111	1/21067
肝醣累積病II 型	Glycogen storage disease II	AR	*GAA*	1/100	1/159	1/100781	1/635
肝醣累積病IIIa 型	Glycogen storage disease IIIa	AR	*AGL*	1/158	1/15800	1/998560000	1/63200
肝醣累積病VII 型	Glycogen storage disease VII	AR	*PFKM*	<1/500	1/50000	1/10000000000	1/200000
肝醣累積病IV 型	Glycogen storage disease IV	AR	*GBE1*	1/387	1/4838	1/93605625	1/19350
肝醣累積病V 型	McArdle disease	AR	*PYGM*	<1/500	1/50000	1/10000000000	1/200000
N-乙醯谷氨酸合成酶缺疲倦症	N-acetylglutamate synthase deficiency	AR	*NAGS*	<1/500	1/50000	1/10000000000	1/200000
合併氧化磷酸化缺乏症 3 型	Combined oxidative phosphorylation deficiency 3	AR	*TSFM*	<1/500	1/50000	1/10000000000	1/200000
X 連鎖先天性腎上腺發育不全	Adrenal hypoplasia, congenital	XL	*NR0B1*	1/12500	1/1250000	1/6250000000000	1/5000000
α-1 抗胰蛋白酶缺乏症	Emphysema due to AAT deficiency	AR	*SERPINA1*	1/33	1/3300	1/43560000	1/13200
α-甘露糖苷貯積症	Mannosidosis, alpha-, types I and II	AR	*MAN2B1*	1/354	1/35400	1/5012640000	1/141600
β-酮硫解酶缺乏症	Alpha-methylacetoacetic aciduria	AR	*ACAT1*	<1/500	1/4545	1/82644628	1/18182

氨基甲醯磷酸合成酶I 缺疲倦症	Carbamoylphosphate synthetase I deficiency	AR	*CPS1*	1/570	1/57000	1/12996000000	1/228000
半乳糖-1-磷酸尿苷轉移酵素缺乏型半乳糖血症	Galactosemia	AR	*GALT*	1/110	1/11000	1/484000000	1/44000
半乳糖差向異構酶缺乏症	Galactose epimerase deficiency	AR	*GALE*	1/100	1/10000	1/400000000	1/40000
半乳糖激酶缺乏型半乳糖血症	Galactokinase deficiency with cataracts	AR	*GALK1*	1/110	1/11000	1/484000000	1/44000
半乳糖血症4 型	Galactosemia IV	AR	*GALM*	1/250	1/25000	1/2500000000	1/100000
丙酮酸羧化酶缺乏症	Pyruvate carboxylase deficiency	AR	*PC*	1/250	1/25000	1/2500000000	1/100000
苯酮尿症	Phenylketonuria	AR	*PAH*	1/99	1/9900	1/392040000	1/39600
丙酸血症，PCCA 相關	Propionicacidemia	AR	*PCCA*	1/224	1/1244	1/6194568	1/4978
丙酸血症，PCCB 相關	Propionicacidemia	AR	*PCCB*	1/224	1/5600	1/125440000	1/22400
丙酮酸脫氫酶E1-β 亞單位元缺陷	Pyruvate dehydrogenase E1-beta deficiency	AR	*PDHB*	<1/500	1/12500	1/625000000	1/50000
大腦肌酸缺陷症候群	Cerebral creatine deficiency syndrome 1	XL	*SLC6A8*	<1/500	1/8333	1/277777778	1/33333
膽固醇酯貯積病	Cholesteryl ester storage disease	AR	*LIPA*	<1/500	1/50000	1/10000000000	1/200000
低磷酸酯酶症	Hypophosphatasia, infantile	AR	*ALPL*	1/158	1/15800	1/998560000	1/63200
丁醯膽鹼酯酶缺乏症	Butyrylcholinesterase deficiency	AR	*BCHE*	1/28	1/2800	1/31360000	1/11200
多發性硫酸酯酶缺乏症	Multiple sulfatase deficiency	AR	*SUMF1*	1/500	1/50000	1/10000000000	1/200000
二氫嘧啶脫氫酶缺乏症	Dihydropyrimidine dehydrogenase deficiency	AR	*DPYD*	<1/500	1/50000	1/10000000000	1/200000
法布瑞氏症	Fabry disease	XL	*GLA*	<1/500	1/7143	1/204081633	1/28571
芳香化酶缺乏症	Aromatase deficiency	AR	*CYP19A1*	<1/500	1/50000	1/10000000000	1/200000
非BH4 缺乏性高苯丙胺酸酸血症C 型	Hyperphenylalaninemia, mild, non-BH4-deficient	AR	*DNAJC12*	<1/200	1/3333	1/44444444	1/13333
楓糖尿症Ia 型	Maple syrup urine disease, type Ia	AR	*BCKDHA*	1/321	1/32100	1/4121640000	1/128400
楓糖尿症Ib 型	Maple syrup urine disease, type Ib	AR	*BCKDHB*	1/364	1/3640	1/52998400	1/14560
楓糖尿症II 型	Maple syrup urine disease, type II	AR	*DBT*	1/481	1/1552	1/9630010	1/6206
楓糖尿症III 型	Maple syrup urine disease, type III	AR	*DLD*	1/500	1/50000	1/10000000000	1/200000
肝豆狀核變性	Wilson disease	AR	*ATP7B*	1/90	1/9000	1/324000000	1/36000

肝腦型粒線體DNA 耗竭症候群6 型	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type)	AR	*MPV17*	<1/500	1/50000	1/10000000000	1/200000
高蛋氨酸血症	Methionine adenosyltransferase deficiency, autosomal recessive	AR/AD	*MAT1A*	3/500	1/16667	1/1111111111	1/66667
高胱氨酸尿-巨成紅血球性貧血症cblE 型	Homocystinuria-megaloblastic anemia, cbl E type	AR	*MTRR*	<1/500	1/7143	1/204081633	1/28571
高胱氨酸尿-巨成紅血球性貧血症cblG 型	Homocystinuria-megaloblastic anemia, cblG complementation type	AR	*MTR*	<1/2000	1/33333	1/4444444444	1/133333
高胱氨酸尿症，MTHFR 相關	Homocystinuria due to MTHFR deficiency	AR	*MTHFR*	1/2	1/200	1/160000	1/800
高鳥氨酸血症-高氨血症-高同型瓜氨酸尿症	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	AR	*SLC25A15*	<1/500	1/6250	1/156250000	1/25000
高同型半胱氨酸血症	Thrombosis, hyperhomocysteinemic	AR	*CBS*	1/224	1/249	1/247783	1/996
戈謝氏症	Gaucher disease	AR	*GBA*	3/500	1/463	1/857339	1/1852
谷固醇血症1 型	Sitosterolemia 1	AR	*ABCG8*	9/1000	1/11111	1/493827160	1/44444
谷固醇血症2 型	Sitosterolemia 2	AR	*ABCG5*	9/1000	1/1587	1/10078105	1/6349
瓜氨酸血症I 型	Citrullinemia	AR	*ASS1*	1/119	1/202	1/162723	1/807
瓜氨酸血症II 型	Citrullinemia, 型 II, neonatal-onset	AR	*SLC25A13*	1/65	1/6500	1/169000000	1/26000
胍基乙酸甲基轉移酶缺乏症	Cerebral creatine deficiency syndrome 2	AR	*GAMT*	1/371	1/1325	1/7022500	1/5300
胱胺酸病	Cystinosis, ocular nonnephropathic	AR	*CTNS*	1/158	1/2257	1/20378776	1/9029
過氧化物酶醯基輔酶 A 氧化酶缺乏症	Peroxisomal acyl-CoA oxidase deficiency	AR	*ACOX1*	<1/500	1/50000	1/10000000000	1/200000
極長鏈醯基輔酶A 去氫酵素缺乏症	VLCAD deficiency	AR	*ACADVL*	1/200	1/20000	1/1600000000	1/80000
家族性脂蛋白脂肪酶缺乏症	Lipoprotein lipase deficiency	AR	*LPL*	1/500	1/50000	1/10000000000	1/200000
甲基丙二酸單醯輔酶 A 差向異構酶缺乏症	Methylmalonyl-CoA epimerase deficiency	AR	*MCEE*	3/500	1/16667	1/1111111111	1/66667
甲基丙二酸尿症cblA 型	Methylmalonic aciduria, vitamin B12-responsive, cblA type	AR	*MMAA*	1/301	1/30100	1/3624040000	1/120400
甲基丙二酸尿症cblB 型	Methylmalonic aciduria, vitamin B12-responsive, cblB type	AR	*MMAB*	1/435	1/43500	1/7569000000	1/174000
甲基丙二酸尿症合併同型半胱氨酸血症cblC 型	Methylmalonic aciduria and homocystinuria, cblC type	AR	*MMACHC*	1/134	1/13400	1/718240000	1/53600
甲基丙二酸尿症合併同型半胱氨酸血症，PRDX1 相關	Methylmalonic aciduria and homocystinuria, cblC type, digenic	AR	*PRDX1*	1/25	1/2500	1/25000000	1/10000
甲基丙二酸尿症合併同型半胱氨酸血症cblD 型	Methylmalonic aciduria and homocystinuria, cblD type	AR	*MMADHC*	<1/500	1/2941	1/34602076	1/11765

甲基丙二酸尿症cblF 型	Methylmalonic aciduria and homocystinuria, cblF type	AR	*LMBRD1*	1/25	1/250	1/250000	1/1000
甲基丙二酸尿症cblX 型	Mental retardation, X-linked 3 (methylmalonic acidemia and homocysteinemia, cblX type)	XL	*HCFC1*	1/25	1/179	1/127551	1/714
甲基丙二酸尿症mut 型	Methylmalonic aciduria, mut(0) type	AR	*MMUT*	<9/1000	1/2778	1/30864198	1/11111
精氨酸琥珀酸尿症	Argininosuccinic aciduria	AR	*ASL*	1/200	1/6667	1/177777778	1/26667
精胺酸酶缺乏症	Argininemia	AR	*ARG1*	<4/1000	1/12500	1/625000000	1/50000
賴氨酸尿蛋白不耐症	Lysinuric protein intolerance	AR	*SLC7A7*	<1/125	1/12500	1/625000000	1/50000
酪胺酸血症I 型	Tyrosinemia, type I	AR	*FAH*	1/99	1/9900	1/392040000	1/39600
酪胺酸血症II 型	Tyrosinemia, type II	AR	*TAT*	1/200	1/20000	1/1600000000	1/80000
酪胺酸血症III 型	Tyrosinemia, type III	AR	*HPD*	<1/500	1/50000	1/10000000000	1/200000
類脂質性先天性腎上腺增生症	Lipoid adrenal hyperplasia	AR	*STAR*	<1/500	1/50000	1/10000000000	1/200000
聯合丙二酸和甲基丙二酸尿症	Combined malonic and methylmalonic aciduria	AR	*ACSF3*	<1/500	1/50000	1/10000000000	1/200000
合併氧化磷酸化缺乏症 1 型	Combined oxidative phosphorylation deficiency 1	AR	*GFM1*	<1/500	1/12500	1/625000000	1/50000
磷酸甘油酸脫氫酶缺乏症	Phosphoglycerate dehydrogenase deficiency	AR	*PHGDH*	<1/500	1/50000	1/10000000000	1/200000
門克斯病	Menkes disease	XL	*ATP7A*	<1/500	1/7143	1/204081633	1/28571
鉬輔因子缺乏症A 型	Molybdenum cofactor deficiency A	AR	*MOCS1*	<3/500	1/1389	1/7716049	1/5556
腦腱黃瘤病	Cerebrotendinous xanthomatosis	AR	*CYP27A1*	1/500	1/50000	1/10000000000	1/200000
尼曼匹克病B 型	Niemann-Pick disease, type B	AR	*SMPD1*	<9/1000	1/5556	1/123456790	1/22222
尼曼匹克病C1 型	Niemann-Pick disease, type C1	AR	*NPC1*	<3/500	1/16667	1/1111111111	1/66667
尼曼匹克病C2 型	Niemann-pick disease, type C2	AR	*NPC2*	<3/500	1/16667	1/1111111111	1/66667
黏多醣貯積症I 型	Mucopolysaccharidosis Ih	AR	*IDUA*	<1/500	1/1515	1/9182736	1/6061
黏多醣貯積症II 型	Mucopolysaccharidosis II	XL	*IDS*	<1/500	1/3846	1/59171598	1/15385
黏多醣貯積症IIIA 型	Mucopolysaccharidosis type IIIA (Sanfilippo A)	AR	*SGSH*	1/454	1/22700	1/2061160000	1/90800
黏多醣貯積症IIIB 型	Mucopolysaccharidosis type IIIB (Sanfilippo B)	AR	*NAGLU*	<1/500	1/4545	1/82644628	1/18182
黏多醣貯積症IIIC 型	Mucopolysaccharidosis type IIIC (Sanfilippo C)	AR	*HGSNAT*	1/434	1/10850	1/470890000	1/43400

黏多醣貯積症IVA 型	Mucopolysaccharidosis IVA	AR	*GALNS*	1/224	1/5600	1/125440000	1/22400
黏多醣貯積症IVB 型	Mucopolysaccharidosis type IVB (Morquio)	AR	*GLB1*	1/134	1/13400	1/718240000	1/53600
黏多醣貯積症VI 型	Mucopolysaccharidosis type VI (Maroteaux-Lamy)	AR	*ARSB*	1/250	1/3125	1/39062500	1/12500
黏多醣貯積症VII 型	Mucopolysaccharidosis VII	AR	*GUSB*	1/250	1/5000	1/100000000	1/20000
黏多醣貯積症IX 型	Mucopolysaccharidosis type IX	AR	*HYAL1*	<1/500	1/50000	1/10000000000	1/200000
黏脂貯積症IIα/β 型	Mucolipidosis II alpha/beta	AR	*GNPTAB*	<1/500	1/25000	1/2500000000	1/100000
黏脂貯積症IV 型	Mucolipidosis IV	AR	*MCOLN1*	1/300	1/5000	1/100000000	1/20000
鳥氨酸氨甲醯轉移酶缺乏症	Ornithine transcarbamylase deficiency	XL	*OTC*	<1/500	1/50000	1/10000000000	1/200000
皮質酮甲基氧化酶缺乏症	Hypoaldosteronism, congenital, due to CMO I deficiency	AR	*CYP11B2*	<1/500	1/50000	1/10000000000	1/200000
全羧化酶合成酶缺乏症	Holocarboxylase synthetase deficiency	AR	*HLCS*	1/500	1/50000	1/10000000000	1/200000
肉鹼脂醯轉移酶缺乏症	Carnitine-acylcarnitine translocase deficiency	AR	*SLC25A20*	<1/500	1/25000	1/2500000000	1/100000
肉鹼棕櫚醯轉移酶缺乏症 IA 型	CPT deficiency, hepatic, type IA	AR	*CPT1A*	1/354	1/17700	1/1253160000	1/70800
肉鹼棕櫚醯轉移酶II 缺 疲倦症	CPT II deficiency, myopathic, stress-induced	AR/AD	*CPT2*	<1/500	1/50000	1/10000000000	1/200000
三功能蛋白質缺乏症	Trifunctional protein deficiency	AR	*HADHB*	1/500	1/1923	1/14792899	1/7692
生物素酶缺乏症	Biotinidase deficiency	AR	*BTD*	1/125	1/12500	1/625000000	1/50000
天冬胺酸葡糖胺尿症	Aspartylglucosaminuria	AR	*AGA*	<1/500	1/50000	1/10000000000	1/200000
天冬胺酸合成酶缺乏症	Asparagine synthetase deficiency	AR	*ASNS*	<1/500	1/1064	1/4526935	1/4255
戊二酸血症I 型	Glutaricaciduria, type I	AR	*GCDH*	1/87	1/8700	1/302760000	1/34800
戊二酸血症IIA 型	Glutaric acidemia IIA	AR	*ETFA*	1/500	1/50000	1/10000000000	1/200000
戊二酸血症IIB 型	Glutaric acidemia IIB	AR	*ETFB*	1/500	1/50000	1/10000000000	1/200000
戊二酸血症IIC 型	Glutaric acidemia IIC	AR	*ETFDH*	1/500	1/50000	1/10000000000	1/200000
先天性膽汁酸合成障礙 1 型	Bile acid synthesis defect, congenital, 1	AR	*HSD3B7*	1/500	1/1282	1/6574622	1/5128
先天性膽汁酸合成障礙 2 型	Bile acid synthesis defect, congenital, 2	AR	*AKR1D1*	<3/500	1/16667	1/1111111111	1/66667
先天性膽汁酸合成障礙 3 型	Bile acid synthesis defect, congenital, 3	AR	*CYP7B1*	<1/200	1/3333	1/44444444	1/13333

先天性膽汁酸合成障礙 4 型	Bile acid synthesis defect, congenital, 4	AR	*AMACR*	<1/200	1/20000	1/1600000000	1/80000
先天性膽汁酸合成障礙 5 型	Bile acid synthesis defect, congenital, 5	AR	*ABCD3*	<1/200	1/20000	1/1600000000	1/80000
先天性膽汁酸合成障礙 6 型	Bile acid synthesis defect, congenital, 6	AR	*ACOX2*	<1/200	1/20000	1/1600000000	1/80000
先天性葡萄醣醛酸轉移酶缺乏症	Hyperbilirubinemia, familial transient neonatal	AR	*UGT1A1*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Ia 型	Congenital disorder of glycosylation, type Ia	AR	*PMM2*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Ib 型	Congenital disorder of glycosylation, type Ib	AR	*MPI*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Ic 型	Congenital disorder of glycosylation, type Ic	AR	*ALG6*	<1/500	1/1351	1/7304602	1/5405
先天性糖基化障礙Id 型	Congenital disorder of glycosylation, type Id	AR	*ALG3*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Ie 型	Congenital disorder of glycosylation, type Ie	AR	*DPM1*	<1/500	1/6250	1/156250000	1/25000
先天性糖基化障礙If 型	Congenital disorder of glycosylation, type If	AR	*MPDU1*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Ig 型	Congenital disorder of glycosylation, type Ig	AR	*ALG12*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Ih 型	Congenital disorder of glycosylation, type Ih	AR	*ALG8*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Ij 型	Congenital disorder of glycosylation, type Ij	AR	*DPAGT1*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Ik 型	Congenital disorder of glycosylation, type Ik	AR	*ALG1*	<1/500	1/667	1/1777778	1/2667
先天性糖基化障礙Im 型	Congenital disorder of glycosylation, type Im	AR	*DOLK*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙In 型	Congenital disorder of glycosylation, type In	AR	*RFT1*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙Iq 型	Congenital disorder of glycosylation, type Iq	AR	*SRD5A3*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙IIb 型	Congenital disorder of glycosylation, type IIb	AR	*MOGS*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙IIc 型	Congenital disorder of glycosylation, type IIc	AR	*SLC35C1*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙IId 型	Congenital disorder of glycosylation, type IId	AR	*B4GALT1*	<1/500	1/50000	1/10000000000	1/200000
先天性糖基化障礙IIf 型	Congenital disorder of glycosylation, type IIf	AR	*SLC35A1*	<1/500	1/50000	1/10000000000	1/200000
粒線體呼吸鏈複合體I 缺陷9 型	Mitochondrial complex I deficiency, nuclear type 9	AR	*NDUFS6*	<1/500	1/50000	1/10000000000	1/200000
粒線體呼吸鏈複合體I 缺陷16 型	Mitochondrial complex I deficiency, nuclear type 16	AR	*NDUFAF5*	1/447	1/1355	1/7339174	1/5418
粒線體呼吸鏈複合體I 缺陷20 型	Mitochondrial complex I deficiency, nuclear type 20	AR	*ACAD9*	<1/500	1/50000	1/10000000000	1/200000

粒線體三功能蛋白質缺乏症	Mitochondrial trifunctional protein deficiency	AR	*HADHA*	3/200	1/952	1/3628118	1/3810
腺苷脫氨酶缺乏症	Adenosine deaminase deficiency, partial	AR	*ADA*	1/224	1/11200	1/501760000	1/44800
延胡索酸酶缺乏症	Fumarase deficiency	AR	*FH*	<1/500	1/4167	1/69444444	1/16667
遺傳性果糖不耐症	Fructose intolerance, hereditary	AR	*ALDOB*	1/122	1/12200	1/595360000	1/48800
乙基丙二酸腦病	Ethylmalonic encephalopathy	AR	*ETHE1*	<1/500	1/50000	1/10000000000	1/200000
異丁醯輔酶A 去氫酶缺疲倦症	Isobutyryl-CoA dehydrogenase deficiency	AR	*ACAD8*	1/125	1/12500	1/625000000	1/50000
異戊酸血症	Isovaleric acidemia	AR	*IVD*	1/167	1/16700	1/1115560000	1/66800
原發性高草酸尿症I 型	Hyperoxaluria, primary, type 1	AR	*AGXT*	1/120	1/12000	1/576000000	1/48000
原發性高草酸尿症II 型	Hyperoxaluria, primary, type II	AR	*GRHPR*	<1/500	1/10000	1/400000000	1/40000
原發性高草酸尿症III 型	Hyperoxaluria, primary, type III	AR	*HOGA1*	1/184	1/18400	1/1354240000	1/73600
原發性肉鹼缺乏症	Carnitine deficiency, systemic primary	AR	*SLC22A5*	1/129	1/12900	1/665640000	1/51600
黏多醣症IIIγ 型	Mucolipidosis III gamma	AR	*GNPTG*	<1/500	1/8333	1/277777778	1/33333
黏多醣貯積症IIID 型（聖菲利波症候群IIID）	Mucopolysaccharidosis type IIID	AR	*GNS*	1/500	1/7143	1/204081633	1/28571
中鏈醯基輔酶A 去氫酶缺乏症	Acyl-CoA dehydrogenase, medium chain, deficiency of	AR	*ACADM*	<3/500	1/16667	1/1111111111	1/66667
短鏈醯輔酶A 去氫酶缺疲倦症	Acyl-CoA dehydrogenase, short-chain, deficiency of	AR	*ACADS*	1/85	1/8500	1/289000000	1/34000

骨骼肌肉系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
1B 型軟骨病	Achondrogenesis Ib	AR	*SLC26A2*	1/158	1/15800	1/998560000	1/63200
先天性肌無力症候群，CHRNE 相關	Myasthenic syndrome, congenital	AR	*CHRNE*	1/408	1/40800	1/6658560000	1/163200
Emery-Dreifuss 型肌營養不良1 型	Emery-Dreifuss muscular dystrophy 1, X-linked	XL	*EMD*	<1/500	1/3846	1/59171598	1/15385
Emery-Dreifuss 型肌營養不良3 型	Emery-Dreifuss muscular dystrophy 3, autosomal recessive	AR	*LMNA*	<1/500	1/7143	1/204081633	1/28571
Emery-Dreifuss 型肌營養不良6 型	Emery-Dreifuss muscular dystrophy 6, X-linked	XL	*FHL1*	<1/500	1/50000	1/10000000000	1/200000
肌肉營養不良症，FKTN 相關	Muscular dystrophy-dystroglycanopathy	AR	*FKTN*	<1/500	1/50000	1/10000000000	1/200000

肌肉營養不良症，LAMA2 相關	Muscular dystrophy, limbgirdle, autosomal recessive 23	AR	*LAMA2*	<1/500	1/50000	1/10000000000	1/200000
Schimke 免疫-骨發育不良	Schimke immunoosseous dysplasia	AR	*SMARCAL 1*	1/500	1/50000	1/10000000000	1/200000
包涵體肌病變2 型（Nonaka 肌病）	Nonaka myopathy	AR	*GNE*	<1/500	1/50000	1/10000000000	1/200000
常染色體隱性骨硬化症 1 型	Osteopetrosis, autosomal recessive 1	AR	*TCIRG1*	1/250	1/1250	1/6250000	1/5000
常染色體隱性骨硬化症 4 型	Osteopetrosis, autosomal recessive 4	AR	*CLCN7*	1/250	1/3571	1/51020408	1/14286
杜氏肌肉營養不良症	Duchenne muscular dystrophy	XL	*DMD*	<1/500	1/2500	1/25000000	1/10000
桿狀體肌病變	Nemaline myopathy 2, autosomal recessive	AR	*NEB*	1/112	1/1867	1/13937778	1/7467
肌肉營養不良A6 型	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6	AR	*LARGE1*	1/125	1/12500	1/625000000	1/50000
脊髓性肌肉萎縮症	Spinal muscular atrophy-2	AR	*SMN1*	1/50	1/56	1/12346	1/222
脊髓延髓肌肉萎縮症（肯尼迪病）	Spinal and bulbar muscular atrophy of Kennedy	XL	*AR*	<1/500	1/1087	1/4725898	1/4348
脊椎肋骨發育不全	Spondylocostal dysostosis 2, autosomal recessive	AR	*MESP2*	<1/500	1/50000	1/10000000000	1/200000
尖頭多指並指綜合症	Carpenter syndrome	AR	*RAB23*	<1/500	1/10000	1/400000000	1/40000
先天性多發性關節攣縮症 1 型	Congenital arthrogryposis with anterior horn cell disease	AR	*GLE1*	<1/500	1/50000	1/10000000000	1/200000
先天性肌無力11 型	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency	AR	*RAPSN*	<1/500	1/50000	1/10000000000	1/200000
先天性肌無力症候群1B 型	Myasthenic syndrome, congenital, 1B, fast-channel	AR/AD	*CHRNA1*	1/577	1/57700	1/13317160000	1/230800
先天性肌無力症候群5 型	Myasthenic syndrome, congenital, 5	AR	*COLQ*	1/163	1/16300	1/1062760000	1/65200
先天性肌無力症候群6 型	Myasthenic syndrome, congenital, 6, presynaptic	AR	*CHAT*	1/272	1/27200	1/2959360000	1/108800
先天性肌無力症候群10 型	Myasthenic syndrome, congenital, 10	AR	*DOK7*	1/163	1/441	1/776304	1/1762
遺傳性運動感覺性神經病變和腓骨肌萎縮症4D 型	Charcot-Marie-Tooth disease, type 4D	AR	*NDRG1*	1/22	1/2200	1/19360000	1/8800
肢帶型肌肉營養不良2H 型	Muscular dystrophy, limbgirdle, autosomal recessive 8	AR	*TRIM32*	<1/500	1/50000	1/10000000000	1/200000
肢帶型肌肉營養不良R1 型	Muscular dystrophy, limbgirdle, autosomal recessive 1	AR	*CAPN3*	<1/500	1/50000	1/10000000000	1/200000
肢帶型肌肉營養不良R2 型	Muscular dystrophy, limbgirdle, autosomal recessive 2	AR	*DYSF*	<1/500	1/50000	1/10000000000	1/200000
肢帶型肌肉營養不良R3 型	Muscular dystrophy, limbgirdle, autosomal recessive 3	AR	*SGCA*	<1/500	1/25000	1/2500000000	1/100000

肢帶型肌肉營養不良R4 型	Muscular dystrophy, limbgirdle, autosomal recessive 4	AR	*SGCB*	1/500	1/2273	1/20661157	1/9091
肢帶型肌肉營養不良R5 型	Muscular dystrophy, limbgirdle, autosomal recessive 5	AR	*SGCG*	1/381	1/38100	1/5806440000	1/152400
肢帶型肌肉營養不良R6 型	Muscular dystrophy, limbgirdle, autosomal recessive 6	AR	*SGCD*	<1/500	1/1923	1/14792899	1/7692
肢帶型肌肉營養不良R9 型	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5	AR	*FKRP*	<1/500	1/10000	1/400000000	1/40000
肢帶型肌肉營養不良R11 型	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1	AR	*POMT1*	1/843	1/84300	1/28425960000	1/337200
肢帶型肌肉營養不良R12 型	Muscular dystrophy, limbgirdle, autosomal recessive 12	AR	*ANO5*	<1/500	1/16667	1/1111111111	1/66667
肢帶型肌肉營養不良R14 型	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2	AR	*POMT2*	<1/500	1/50000	1/10000000000	1/200000
肢帶型肌肉營養不良R15 型	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3	AR	*POMGNT1*	1/462	1/46200	1/8537760000	1/184800
肢帶型肌肉營養不良R16 型	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9	AR	*DAG1*	<1/500	1/50000	1/10000000000	1/200000
肢帶型肌肉營養不良R19 型	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14	AR	*GMPPB*	<1/500	1/50000	1/10000000000	1/200000
肢根點狀軟骨發育不良 1 型	Rhizomelic chondrodysplasia punctata, type 1	AR	*PEX7*	1/158	1/465	1/863806	1/1859
肢根點狀軟骨發育不良 3 型	Rhizomelic chondrodysplasia punctata, type 3	AR	*AGPS*	<1/500	1/50000	1/10000000000	1/200000
X 連鎖點狀軟骨發育不良 1 型	Chondrodysplasia punctata, X-linked recessive	XL	*ARSL*	<1/500	1/50000	1/10000000000	1/200000
X 連鎖肌小管肌病變	Myotubular myopathy, X-linked	XL	*MTM1*	<1/500	1/2941	1/34602076	1/11765
緻密性成骨不全症	Pycnodysostosis	AR	*CTSK*	<1/500	1/50000	1/10000000000	1/200000

呼吸系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
原發性纖毛運動障礙1 型	Ciliary dyskinesia, primary, 1, with or without situs inversus	AR	*DNAI1*	1/230	1/23000	1/2116000000	1/92000
原發性纖毛運動障礙3 型	Ciliary dyskinesia, primary, 3, with or without situs inversus	AR	*DNAH5*	1/142	1/14200	1/806560000	1/56800
原發性纖毛運動障礙9 型	Ciliary dyskinesia, primary, 9, with or without situs inversus	AR	*DNAI2*	1/447	1/44700	1/7992360000	1/178800
原發性纖毛運動障礙14 型	Ciliary dyskinesia, primary, 14	AR	*CCDC39*	1/211	1/555	1/1233269	1/2221
原發性纖毛運動障礙16 型	Ciliary dyskinesia, primary, 16	AR	*DNAL1*	<1/500	1/50000	1/10000000000	1/200000

原發性纖毛運動障礙17型	Ciliary dyskinesia, primary, 17	AR	*CCDC103*	1/316	1/31600	1/3994240000	1/126400
原發性纖毛運動障礙30型	Ciliary dyskinesia, primary, 30	AR	*CCDC151*	1/365	1/36500	1/5329000000	1/146000

泌尿系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
黑尿症	Alkaptonuria	AR	*HGD*	<1/500	1/50000	1/10000000000	1/200000
多囊性腎病變4型	Polycystic kidney disease 4, with or without hepatic disease	AR	*PKHD1*	<1/100	1/10000	1/400000000	1/40000
多囊性腎病變5型	Polycystic kidney 區 5	AR	*DZIP1L*	<1/100	1/10000	1/400000000	1/40000
腎病症候群1型	Nephrotic syndrome, type 1	AR	*NPHS1*	1/289	1/14450	1/835210000	1/57800
腎病症候群2型	Nephrotic syndrome, type 2	AR	*NPHS2*	3/500	1/16667	1/1111111111	1/66667
腎病症候群3型	Nephrotic syndrome, type 3	AR	*PLCE1*	3/500	1/16667	1/1111111111	1/66667
腎病症候群6型	Nephrotic syndrome, type 6	AR	*PTPRO*	3/500	1/16667	1/1111111111	1/66667
遺傳性腎炎（奧爾波綜合徵），COL4A3相關	Alport syndrome 2, autosomal recessive	AR	*COL4A3*	1/267	1/26700	1/2851560000	1/106800
遺傳性腎炎（奧爾波綜合徵），COL4A4相關	Alport syndrome 2, autosomal recessive	AR	*COL4A4*	1/267	1/742	1/2200278	1/2967
Dent病2型（遺傳性腎小管病）	Dent disease 2	XL	*OCRL*	<1/500	1/10000	1/400000000	1/40000
腎性尿崩症	Diabetes insipidus, nephrogenic	AR/AD	*AQP2*	<1/500	1/4545	1/82644628	1/18182
腎單位腎癆3型	Nephronophthisis 3	AR	*NPHP3*	<3/1000	1/1149	1/5284714	1/4598

免疫系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
體染色體隱性遺傳慢性肉芽腫病2型	Chronic granulomatous disease 2, autosomal recessive	AR	*NCF2*	1/224	1/22400	1/2007040000	1/89600
體染色體隱性遺傳慢性肉芽腫病4型	Chronic granulomatous disease 4, autosomal recessive	AR	*CYBA*	1/224	1/2036	1/16587107	1/8145
體染色體隱性遺傳慢性肉芽腫病5型	Chronic granulomatous disease 5, autosomal recessive	AR	*CYBC1*	1/224	1/22400	1/2007040000	1/89600

X 連鎖慢性肉芽腫病	Chronic granulomatous disease, X-linked	XL	*CYBB*	<1/500	1/3125	1/39062500	1/12500
家族性地中海熱	Familial Mediterranean fever, AR	AR	*MEFV*	1/20	1/2000	1/16000000	1/8000
濕疹-血小板減少-免疫缺陷症候群	Wiskott-Aldrich syndrome	XL	*WAS*	<1/500	1/685	1/1876525	1/2740
X 連鎖無丙種球蛋白血症 1 型	Agammaglobulinemia, X-linked 1	XL	*BTK*	<1/500	1/50000	1/10000000000	1/200000
X 連鎖淋巴增生症1 型	Lymphoproliferative syndrome, X-linked, 1	XL	*SH2D1A*	<1/500	1/7143	1/204081633	1/28571
X 連鎖淋巴增生症2 型	Lymphoproliferative syndrome, X-linked, 2	XL	*XIAP*	<1/500	1/2500	1/25000000	1/10000
聯合免疫缺陷症（Omenn 綜合症），RAG1 相關	Combined cellular and humoral immune defects with granulomas	AR	*RAG1*	1/137	1/13700	1/750760000	1/54800
聯合免疫缺陷症（Omenn 綜合症），RAG2 相關	Combined cellular and humoral immune defects with granulomas	AR	*RAG2*	1/137	1/13700	1/750760000	1/54800
X 連鎖重症聯合免疫缺陷	Severe combined immunodeficiency, X-linked	XL	*IL2RG*	<1/500	1/50000	1/10000000000	1/200000
對電離輻射敏感的重症聯合免疫缺陷	Severe combined immunodeficiency, Athabascan type	AR	*DCLRE1C*	<1/500	1/50000	1/10000000000	1/200000
X 連鎖高IgM 免疫缺陷	Immunodeficiency, X-linked, with hyper-IgM	XL	*CD40LG*	<1/500	1/16667	1/1111111111	1/66667
裸淋巴球症候群II 型	Bare lymphocyte syndrome, type II, complementation group A	AR	*CIITA*	<1/500	1/50000	1/10000000000	1/200000
家族性噬血細胞性淋巴組織細胞增多症2 型	Hemophagocytic lymphohistiocytosis, familial, 2	AR	*PRF1*	3/500	1/16667	1/1111111111	1/66667
家族性噬血細胞性淋巴組織細胞增多症3 型	Hemophagocytic lymphohistiocytosis, familial, 3	AR	*UNC13D*	3/500	1/16667	1/1111111111	1/66667
家族性噬血細胞性淋巴組織細胞增多症4 型	Hemophagocytic lymphohistiocytosis, familial, 4	AR	*STX11*	3/500	1/16667	1/1111111111	1/66667
家族性噬血細胞性淋巴組織細胞增多症5 型	Hemophagocytic lymphohistiocytosis, familial, 5	AR	*STXBP2*	3/500	1/16667	1/1111111111	1/66667

內分泌系統							
疾病中文名稱	**疾病英文名稱**	**遺傳模式**	**基因**	**攜帶率（總人口）**	**殘餘風險**	**夫妻雙方檢測為陰性的生育風險**	**夫妻一方檢測為陽性另一方為陰性的生育風險**
腦下垂體激素合併缺乏症2 型	Pituitary hormone deficiency, combined, 2	AR	*PROP1*	1/45	1/4500	1/81000000	1/18000
家族性高膽固醇血症	Hypercholesterolemia, familial, 4	AR	*LDLRAP1*	1/8	1/800	1/2560000	1/3200
聯合性腦下垂體激素缺乏症 1 型	Pituitary hormone deficiency, combined, 1	AR/AD	*POU1F1*	1/267	1/4450	1/79210000	1/17800

聯合性腦下垂體激素缺乏症 3 型	Pituitary hormone deficiency, combined, 3	AR	*LHX3*	1/45	1/4500	1/81000000	1/18000
先天性高胰島素性低血糖血症1 型	Hyperinsulinemic hypoglycemia, familial, 1	AR/AD	*ABCC8*	1/112	1/11200	1/501760000	1/44800
先天性高胰島素性低血糖血症2 型	Hyperinsulinemic hypoglycemia, familial, 2	AR/AD	*KCNJ11*	1/423	1/42300	1/7157160000	1/169200
先天性高胰島素性低血糖血症3 型	Hyperinsulinemic hypoglycemia, familial, 4	AR	*HADH*	<1/500	1/6250	1/156250000	1/25000
先天性甲狀腺功能低下症 1 型	Hypothyroidism, congenital, nongoitrous, 1	AR	*TSHR*	1/50	1/5000	1/100000000	1/20000
先天性甲狀腺功能低下症 4 型	Hypothyroidism, congenital, nongoitrous 4	AR	*TSHB*	1/50	1/5000	1/100000000	1/20000
自體免疫性多內分泌腺病症候群I 型	Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia	AR/AD	*AIRE*	1/150	1/15000	1/900000000	1/60000

皮膚系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
Ehlers-Danlos 症候群	Ehlers-Danlos syndrome, dermatosparaxis type	AR	*ADAMTS2*	<1/500	1/5000	1/100000000	1/20000
X 連鎖眼皮膚白化症	Ocular albinism, type I, Nettleship-Falls type	XL	*GPR143*	1/25000	1/147059	1/86505190311	1/588235
腸炎性肢端皮膚炎	Acrodermatitis enteropathica	AR	*SLC39A4*	<1/500	1/50000	1/10000000000	1/200000
體染色體隱性遺傳先天性魚鱗病4 型	Ichthyosis, congenital, autosomal recessive 4A	AR	*ABCA12*	1/284	1/28400	1/3226240000	1/113600
非Herlitz 型交界型大皰性表皮鬆解	Epidermolysis bullosa, junctional, localisata variant	AR	*COL17A1*	<1/2000	1/50000	1/10000000000	1/200000
共濟失調毛細血管擴張	Ataxia-telangiectasia	AR	*ATM*	1/100	1/909	1/3305785	1/3636
交界型大皰性表皮鬆解症	Epidermolysis bullosa	AR	*LAMA3*	1/781	1/78100	1/24398440000	1/312400
交界型大皰性表皮鬆解症，LAMB3 相關	Epidermolysis bullosa, junctional, Herlitz type	AR	*LAMB3*	1/781	1/78100	1/24398440000	1/312400
交界型大皰性表皮鬆解症，LAMC2 相關	Epidermolysis bullosa, junctional, Herlitz type	AR	*LAMC2*	1/781	1/78100	1/24398440000	1/312400
營養不良性大皰性表皮鬆解症	Epidermolysis bullosa dystrophica	AR	*COL7A1*	1/196	1/19600	1/1536640000	1/78400
先天性角化不良5 型	Dyskeratosis congenita, autosomal recessive 5	AR/AD	*RTEL1*	1/500	1/25000	1/2500000000	1/100000
先天性魚鱗癬	Ichthyosis, congenital, autosomal recessive 1	AR	*TGM1*	1/224	1/22400	1/2007040000	1/89600
眼皮膚白化症1 型	Albinism, oculocutaneous, type IB	AR	*TYR*	3/200	1/6667	1/177777778	1/26667

眼皮膚白化症2 型	Albinism, oculocutaneous, type II, modifier of	AR	*MC1R*	3/200	1/6667	1/177777778	1/26667
眼皮膚白化症3 型	Albinism, oculocutaneous, type III	AR	*TYRP1*	3/200	1/6667	1/177777778	1/26667
眼皮膚白化症4 型	Albinism, oculocutaneous, type IV	AR	*SLC45A2*	3/200	1/6667	1/177777778	1/26667
眼皮膚白化症6 型	Albinism, oculocutaneous, type VI	AR	*SLC24A5*	3/200	1/108	1/46248	1/430
眼皮膚白化症7 型	Albinism, oculocutaneous, type VII	AR	*LRMDA*	3/200	1/6667	1/177777778	1/26667
眼皮膚白化症8 型	Oculocutaneous albinism, type VIII	AR	*DCT*	3/200	1/6667	1/177777778	1/26667
色素性乾皮病A 型	Xeroderma pigmentosum, group A	AR	*XPA*	1/500	1/1613	1/10405827	1/6452
色素性乾皮症C 型	Xeroderma pigmentosum, group C	AR	*XPC*	1/500	1/50000	1/10000000000	1/200000
棕色皮膚白化症	Albinism, brown oculocutaneous	AR	*OCA2*	3/200	1/6667	1/177777778	1/26667

神經系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
Amish 嬰兒癲癇綜合症	Salt and pepper developmental regression syndrome	AR	*ST3GAL5*	9/50	1/69	1/19290	1/278
CRASH 症候群	CRASH syndrome/MASA syndrome	XL	*L1CAM*	<1/500	1/50000	1/10000000000	1/200000
SAP-b 蛋白缺乏型異染性腦白質營養不良症	Metachromatic leukodystrophy due to SAP-b deficiency	AR	*PSAP*	<1/500	1/50000	1/10000000000	1/200000
Segawa 症候群（多巴胺反應性肌張力失調）	Segawa syndrome, recessive	AR	*TH*	1/224	1/7467	1/223004444	1/29867
Tay-Sachs 病	Tay-Sachs disease	AR	*HEXA*	1/300	1/30000	1/3600000000	1/120000
X 連鎖無腦迴畸形	Lissencephaly, X-linked	XL	*DCX*	<1/500	1/50000	1/10000000000	1/200000
白質消融性白質腦病變	Leukoencephalopathy with vanishing white matter	AR	*EIF2B5*	<1/500	1/50000	1/10000000000	1/200000
常染色體隱性痙攣性麻痺共濟失調Charlevoix-Saguenay 型	Spastic ataxia, Charlevoix-Saguenay type	AR	*SACS*	<1/500	1/50000	1/10000000000	1/200000
共濟失調伴隨選擇性維生素E 缺乏症	Ataxia with isolated vitamin E deficiency	AR	*TTPA*	<1/500	1/2941	1/34602076	1/11765
關節攣縮，智力障礙和癲癇發作	Arthrogryposis, mental retardation, and seizures	AR	*SLC35A3*	<1/500	1/847	1/2872738	1/3390
家族性自主神經功能失調	Dysautonomia, familial	AR	*ELP1*	1/300	1/30000	1/3600000000	1/120000

進行性小頭畸形，伴隨癲癇和腦萎縮	Microcephaly, postnatal progressive, with seizures and brain atrophy	AR	*MED17*	<1/500	1/50000	1/10000000000	1/200000
痙攣性截癱15 型	Spastic paraplegia 15, autosomal recessive	AR	*ZFYVE26*	<1/500	1/50000	1/10000000000	1/200000
痙攣性截癱49 型	Spastic paraplegia 49, autosomal recessive	AR	*TECPR2*	<1/500	1/50000	1/10000000000	1/200000
巨腦性白質腦病變伴隨皮質下囊腫	Megalencephalic leukoencephalopathy with subcortical cysts	AR	*MLC1*	<1/500	1/50000	1/10000000000	1/200000
卡納萬病	Canavan disease	AR	*ASPA*	1/57	1/5700	1/129960000	1/22800
腦積水症候群	Hydrolethalus syndrome	AR	*HYLS1*	7/500	1/7143	1/204081633	1/28571
腦橋小腦發育不全1A 型	Pontocerebellar hypoplasia type 1A	AR	*VRK1*	<1/500	1/50000	1/10000000000	1/200000
腦橋小腦發育不全1B 型	Pontocerebellar hypoplasia, type 1B	AR	*EXOSC3*	<1/500	1/2174	1/18903592	1/8696
腦橋小腦發育不全2D 型	Pontocerebellar hypoplasia type 2D	AR	*SEPSECS*	<1/500	1/4545	1/82644628	1/18182
腦橋小腦發育不全6 型	Pontocerebellar hypoplasia, type 6	AR	*RARS2*	<1/500	1/16667	1/1111111111	1/66667
胼胝體發育不全伴周圍神經病變	Agenesis of the corpus callosum with peripheral neuropathy	AR	*SLC12A6*	<1/500	1/50000	1/10000000000	1/200000
桑霍夫病	Sandhoff disease, infantile, juvenile, and adult forms	AR	*HEXB*	1/600	1/20000	1/1600000000	1/80000
神經元蠟樣質脂褐質沉積症1 型	Ceroid lipofuscinosis, neuronal, 1	AR	*PPT1*	1/368	1/36800	1/5416960000	1/147200
神經元蠟樣質脂褐質沉積症2 型	Ceroid lipofuscinosis, neuronal, 2	AR	*TPP1*	1/252	1/25200	1/2540160000	1/100800
神經元蠟樣脂褐質沉積症3 型	Ceroid lipofuscinosis, neuronal, 3	AR	*CLN3*	1/230	1/3286	1/43183673	1/13143
神經元蠟樣質脂褐質沉積症5 型	Ceroid lipofuscinosis, neuronal, 5	AR	*CLN5*	<1/500	1/1471	1/8650519	1/5882
神經元蠟樣質脂褐質沉積症6 型	Ceroid lipofuscinosis, neuronal, 6	AR	*CLN6*	<1/500	1/3571	1/51020408	1/14286
神經元蠟樣質脂褐質沉積症7 型	Ceroid lipofuscinosis, neuronal, 7	AR	*MFSD8*	<1/500	1/10000	1/400000000	1/40000
神經元蠟樣質脂褐質沉積症8 型	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant	AR	*CLN8*	<1/500	1/50000	1/10000000000	1/200000
神經元蠟樣質脂褐質沉積症10 型	Ceroid lipofuscinosis, neuronal, 10	AR	*CTSD*	1/1761	1/176100	1/124044840000	1/704400
腎上腺腦白質營養不良	Adrenoleukodystrophy	XL	*ABCD1*	1/100	1/714	1/2040816	1/2857
雙側額頂葉多小腦迴畸形	Polymicrogyria, bilateral frontoparietal	AR	*ADGRG1*	<1/500	1/25000	1/2500000000	1/100000
舞蹈症-棘紅血球增多症	Choreoacanthocytosis	AR	*VPS13A*	<1/500	1/2500	1/25000000	1/10000

亞急性壞死性腦脊髓病（萊氏症候群）伴隨複合物 IV 缺乏症	Mitochondrial complex IV deficiency, nuclear type 5, (French-Canadian)	AR	*LRPPRC*	1/447	1/4967	1/98671111	1/19867
異染性腦白質營養不良	Metachromatic leukodystrophy	AR	*ARSA*	1/100	1/10000	1/400000000	1/40000
嬰兒神經軸索性營養不良	Infantile neuroaxonal dystrophy 1	AR	*PLA2G6*	1/500	1/16667	1/1111111111	1/66667
嬰兒遺傳性腦白質萎縮（克拉伯氏症）	Krabbe disease	AR	*GALC*	1/158	1/5267	1/110951111	1/21067

視力系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
迴旋狀脈絡膜視網膜萎縮	Gyrate atrophy of choroid and retina with or without ornithinemia	AR	*OAT*	<1/500	1/3846	1/59171598	1/15385
角膜內皮營養不良	Corneal endothelial dystrophy, autosomal recessive	AR	*SLC4A11*	<1/500	1/50000	1/10000000000	1/200000
萊伯氏先天性黑朦膿5 型	Leber congenital amaurosis 5	AR	*LCA5*	1/500	1/7143	1/204081633	1/28571
萊伯氏先天性黑蒙症13 型	Leber congenital amaurosis 13	AR/AD	*RDH12*	<1/500	1/1250	1/6250000	1/5000
進行性外眼肌麻痺1 型	Progressive external ophthalmoplegia, autosomal recessive 1	AR	*POLG*	1/113	1/11300	1/510760000	1/45200
色盲	Achromatopsia 3	AR	*CNGB3*	1/87	1/8700	1/302760000	1/34800
視網膜色素變性2 型	Retinitis pigmentosa 2	XL	*RP2*	3/100	1/3333	1/44444444	1/13333
視網膜色素變性3 型	Retinitis pigmentosa 3	XL	*RPGR*	1/259	1/332	1/441032	1/1328
視網膜色素變性12 型	Retinitis pigmentosa-12	AR	*CRB1*	1/104	1/10400	1/432640000	1/41600
視網膜色素變性19 型	Retinitis pigmentosa 19	AR	*ABCA4*	3/100	1/3333	1/44444444	1/13333
視網膜色素變性20 型	Retinitis pigmentosa 20	AR	*RPE65*	1/228	1/1425	1/8122500	1/5700
視網膜色素變性25 型	Retinitis pigmentosa 25	AR	*EYS*	1/66	1/244	1/239012	1/978
視網膜色素變性26 型	Retinitis pigmentosa 26	AR	*CERKL*	1/148	1/4933	1/97351111	1/19733
視網膜色素變性28 型	Retinitis pigmentosa 28	AR	*FAM161A*	1/296	1/29600	1/3504640000	1/118400
視網膜色素變性37 型	Retinitis pigmentosa 37	AR/AD	*NR2E3*	1/209	1/20900	1/1747240000	1/83600
視網膜色素變性40 型	Retinitis pigmentosa 40	AR	*PDE6B*	3/100	1/3333	1/44444444	1/13333

視網膜色素變性43 型	Retinitis pigmentosa 43	AR	*PDE6A*	3/100	1/3333	1/44444444	1/13333
視網膜色素變性47 型	Retinitis pigmentosa 47	AR	*SAG*	3/100	1/3333	1/44444444	1/13333
視網膜色素變性59 型	Retinitis pigmentosa 59	AR	*DHDDS*	1/296	1/29600	1/3504640000	1/118400
X 連鎖青少年型視網膜劈裂症	Retinoschisis	XL	*RS1*	<1/500	1/3333	1/44444444	1/13333
小眼畸形或伴隨眼缺損	Microphthalmia with coloboma 3	AR	*VSX2*	1/91	1/9100	1/331240000	1/36400
原發性先天性青光眼	Glaucoma 3A, primary open angle, congenital, juvenile, 或 adult onset	AR	*CYP1B1*	1/50	1/5000	1/100000000	1/20000

聽力系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
常染色體隱性耳聾1A 型	Deafness, autosomal recessive 1A	AR	*GJB2*	1/80	1/8000	1/256000000	1/32000
常染色體隱性耳聾1B 型	Deafness, autosomal recessive 1B	AR	*GJB6*	1/423	1/42300	1/7157160000	1/169200
常染色體隱性耳聾2 型	Deafness, autosomal recessive 2	AR	*MYO7A*	1/206	1/20600	1/1697440000	1/82400
常染色體隱性耳聾4 型	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR	*SLC26A4*	1/80	1/8000	1/256000000	1/32000
常染色體隱性耳聾77 型	Deafness, autosomal recessive 77	AR	*LOXHD1*	1/500	1/50000	1/10000000000	1/200000

消化系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
進行性家族性肝內膽汁淤積症1 型	Cholestasis, progressive familial intrahepatic 1	AR	*ATP8B1*	<1/500	1/50000	1/10000000000	1/200000
進行性家族性肝內膽汁淤積症2 型	Cholestasis, progressive familial intrahepatic 2	AR	*ABCB11*	<1/500	1/25000	1/2500000000	1/100000
進行性家族性肝內膽汁淤積症3 型	Cholestasis, progressive familial intrahepatic 3	AR/AD	*ABCB4*	<1/500	1/50000	1/10000000000	1/200000
進行性家族性肝內膽汁淤積症4 型	Cholestasis, progressive familial intrahepatic 4	AR	*TJP2*	<1/502	1/50200	1/10080160000	1/200800
進行性家族性肝內膽汁淤積症5 型	Cholestasis, progressive familial intrahepatic, 5	AR	*NR1H4*	<1/500	1/50000	1/10000000000	1/200000
囊性纖維化	Cystic fibrosis	AR	*CFTR*	<1/20	1/400	1/640000	1/1600

嬰兒急性肝衰竭	Liver failure, transient infantile	AR	*TRMU*	<1/500	1/50000	1/10000000000	1/200000
先天性氯化物腹瀉	Diarrhea 1, secretory chloride, congenital	AR	*SLC26A3*	<1/500	1/50000	1/10000000000	1/200000

血液系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
α-地中海貧血	Thalassemia, alpha-	AR	*HBA1/HBA2*	1/20	1/133	1/35556	1/178
β-地中海型貧血	Thalassemia, beta	AR	*HBB*	1/158	1/15800	1/998560000	1/63200
A 型血友病	Hemophilia A	XL	*F8*	<1/500	1/25000	1/2500000000	1/100000
B 型血友病	Hemophilia B	XL	*F9*	<1/500	1/8333	1/277777778	1/33333
C 型血友病	Factor XI deficiency, autosomal recessive	AR/AD	*F11*	1/500	1/50000	1/10000000000	1/200000
體染色體隱性遺傳重症先天性中性白血球減少症	Neutropenia, severe congenital 4, autosomal recessive	AR	*G6PC3*	1/341	1/34100	1/4651240000	1/136400
範可尼貧血互補群A 型	Fanconi anemia, complementation group A	AR	*FANCA*	1/239	1/3414	1/46629388	1/13657
範可尼貧血互補群C 型	Fanconi anemia, complementation group C	AR	*FANCC*	1/535	1/53500	1/11449000000	1/214000
範可尼貧血互補群D2	Fanconi anemia, complementation group D2	AR	*FANCD2*	<1/200	1/4000	1/64000000	1/16000
範可尼貧血互補群G 型	Fanconi anemia, complementation group G	AR	*FANCG*	1/239	1/23900	1/2284840000	1/95600
範可尼貧血互補群 I	Fanconi anemia, complementation group I	AR	*FANCI*	<1/200	1/6667	1/177777778	1/26667
巨血小板症候群A1 型	Bernard-Soulier syndrome, type A1 (recessive)	AR	*GP1BA*	1/500	1/50000	1/10000000000	1/200000
巨血小板症候群C 型	Bernard-Soulier syndrome, type C	AR	*GP9*	1/500	1/50000	1/10000000000	1/200000
低凝血酶原血症	Hypoprothrombinemia	AR	*F2*	1/33	1/3300	1/43560000	1/13200
凝血酶原因子 V 缺乏症	Factor V deficiency	AR	*F5*	1/36	1/3600	1/51840000	1/14400
無β 脂蛋白血症，棘紅細胞增多症	Abetalipoproteinemia	AR	*MTTP*	<1/500	1/50000	1/10000000000	1/200000
先天性低巨核細胞性血小板減少症	Thrombocytopenia, congenital amegakaryocytic	AR	*MPL*	1/102	1/10200	1/416160000	1/40800
粒線體肌病變和鐵粒幼紅細胞性貧血1 型	Myopathy, lactic acidosis, and sideroblastic anemia 1	AR	*PUS1*	<1/500	1/7143	1/204081633	1/28571

重型先天性嗜中性白血球減少症3 型	Neutropenia, severe congenital 3, autosomal recessive	AR	*HAX1*	1/224	1/22400	1/2007040000	1/89600
重症先天性中性粒細胞減少症	Neutropenia, severe congenital, 5, autosomal recessive	AR	*VPS45*	1/224	1/22400	1/2007040000	1/89600

多重系統

疾病中文名稱	疾病英文名稱	遺傳模式	基因	攜帶率（總人口）	殘餘風險	夫妻雙方檢測為陰性的生育風險	夫妻一方檢測為陽性另一方為陰性的生育風險
Usher 症候群1B 型	Usher syndrome, type 1B	AR	*MYO7A*	1/206	1/20600	1/1697440000	1/82400
Usher 症候群1C 型	Usher syndrome, type 1C	AR	*USH1C*	1/353	1/35300	1/4984360000	1/141200
Usher 症候群1D 型	Usher syndrome, type 1D	AR	*CDH23*	1/285	1/28500	1/3249000000	1/114000
Usher 症候群1F 型	Usher syndrome, type 1F	AR	*PCDH15*	1/395	1/13167	1/693444444	1/52667
Usher 症候群1G 型	Usher syndrome, type 1G	AR	*USH1G*	1/600	1/60000	1/14400000000	1/240000
Usher 症候群2A 型	Usher syndrome, type 2A	AR	*USH2A*	1/126	1/12600	1/635040000	1/50400
Usher 症候群2C 型	Usher syndrome, type 2C	AR	*ADGRV1*	3/250	1/8333	1/277777778	1/33333
Usher 症候群3A 型	Usher syndrome, type 3A	AR	*CLRN1*	1/500	1/50000	1/10000000000	1/200000
Aicardi-Goutieres 綜合徵	Aicardi-Goutieres syndrome 5	AR	*SAMHD1*	<1/500	1/10000	1/400000000	1/40000
ARTS 症候群	Arts syndrome	XL	*PRPS1*	<1/500	1/50000	1/10000000000	1/200000
Bardet-Biedl 症候群 1 型	Bardet-Biedl syndrome 1	AR	*BBS1*	1/367	1/36700	1/5387560000	1/146800
Bardet-Biedl 症候群 2 型	Bardet-Biedl syndrome 2	AR	*BBS2*	1/621	1/20700	1/1713960000	1/82800
Bardet-Biedl 症候群 3 型	Bardet-Biedl syndrome 3	AR	*ARL6*	<1/500	1/7143	1/204081633	1/28571
Bardet-Biedl 症候群 4 型	Bardet-Biedl syndrome 4	AR	*BBS4*	<1/500	1/5556	1/123456790	1/22222
Bardet-Biedl 症候群 6 型	Bardet-Biedl syndrome 6	AR	*MKKS*	<1/500	1/50000	1/10000000000	1/200000
Bardet-Biedl 症候群10 型	Bardet-Biedl syndrome 10	AR	*BBS10*	1/395	1/39500	1/6241000000	1/158000
Bardet-Biedl 症候群12 型	Bardet-Biedl syndrome 12	AR	*BBS12*	1/791	1/79100	1/25027240000	1/316400
Bardet-Biedl 症候群14 型	Bardet-Biedl syndrome 14	AR	*CEP290*	1/190	1/629	1/1584242	1/2517

Barth 綜合症	Barth syndrome	XL	*TAZ*	<3/1000	1/33333	1/4444444444	1/133333
Cohen 症候群（肥胖－腦－眼骨骼異常綜合徵）	Cohen syndrome	AR	*VPS13B*	<1/500	1/12500	1/625000000	1/50000
Ehlers-Danlos 症候群 VI 型	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	AR	*PLOD1*	1/158	1/15800	1/998560000	1/63200
Ellis-van Creveld 綜合症，EVC2 相關	Ellis-van Creveld syndrome	AR	*EVC2*	1/240	1/12000	1/576000000	1/48000
Ellis-van Creveld 綜合症，EVC 相關	Ellis-van Creveld syndrome	AR	*EVC*	1/142	1/2029	1/16460408	1/8114
Gitelman 症候群	Gitelman syndrome	AR	*SLC12A3*	1/100	1/10000	1/400000000	1/40000
Hermansky-Pudlak 綜合徵1 型	Hermansky-Pudlak syndrome 1	AR	*HPS1*	<1/500	1/5556	1/123456790	1/22222
Hermansky-Pudlak 綜合徵3 型	Hermansky-Pudlak syndrome 3	AR	*HPS3*	<1/500	1/8333	1/277777778	1/33333
Hermansky-Pudlak 綜合徵4 型	Hermansky-Pudlak syndrome 4	AR	*HPS4*	<1/500	1/50000	1/10000000000	1/200000
Hermansky-Pudlak 綜合徵5 型	Hermansky-Pudlak syndrome 5	AR	*HPS5*	<1/500	1/50000	1/10000000000	1/200000
Hermansky-Pudlak 綜合徵6 型	Hermansky-Pudlak syndrome 6	AR	*HPS6*	<1/500	1/3333	1/44444444	1/13333
Joubert 綜合症2 型	Joubert syndrome 2	AR	*TMEM216*	1/472	1/47200	1/8911360000	1/188800
Joubert 綜合症3 型	Joubert syndrome 3	AR	*AHI1*	1/472	1/1098	1/4819557	1/4391
Joubert 症候群4 型	Joubert syndrome 4	AR	*NPHP1*	1/480	1/16000	1/1024000000	1/64000
Joubert 綜合症5 型	Joubert syndrome 5	AR	*CEP290*	1/472	1/629	1/1584242	1/2517
Joubert 綜合症6 型	Joubert syndrome 6	AR	*TMEM67*	1/472	1/1573	1/9901511	1/6293
Joubert 症候群7 型	Joubert syndrome 7	AR	*RPGRIP1L*	1/259	1/1992	1/15877160	1/7969
Joubert 綜合症9 型	Joubert syndrome 9	AR	*CC2D2A*	1/472	1/9440	1/356454400	1/37760
Joubert 綜合症17 型	Joubert syndrome 17	AR	*CPLANE1*	1/472	1/2484	1/24685208	1/9937
Laron 症候群	Laron dwarfism	AR	*GHR*	1/158	1/15800	1/998560000	1/63200
Meckel 症候群1 型	Meckel syndrome 1	AR	*MKS1*	1/260	1/26000	1/2704000000	1/104000
Netherton 症候群	Netherton syndrome	AR	*SPINK5*	<2/100	1/5000	1/100000000	1/20000
Pendred 症候群	Pendred syndrome	AR	*SLC26A4*	1/80	1/8000	1/256000000	1/32000
Shwachman-Diamond 症候群	Shwachman-Diamond syndrome	AR	*SBDS*	1/146	1/14600	1/852640000	1/58400

Sjogren-Larsson 綜合徵	Sjogren-Larsson syndrome	AR	*ALDH3A2*	1/250	1/25000	1/2500000000	1/100000
Smith-Lemli-Opitz 綜合徵	Smith-Lemli-Opitz syndrome	AR	*DHCR7*	1/30	1/3000	1/36000000	1/12000
Stuve-Wiedemann 綜合徵	Stuve-Wiedemann syndrome	AR	*LIFR*	<1/500	1/2500	1/25000000	1/10000
Wolcott-Rallison 綜合徵	Wolcott-Rallison syndrome	AR	*EIF2AK3*	<1/500	1/5000	1/100000000	1/20000
Wolfram 症候群1 型	Wolfram syndrome 1	AR	*WFS1*	<1/200	1/20000	1/1600000000	1/80000
α-地中海型貧血X 連鎖型智障症候群	Mental retardation-hypotonic facies syndrome, X-linked	XL	*ATRX*	<1/500	1/1190	1/5668934	1/4762
巴特氏症候群	Bartter syndrome, type 4a	AR	*BSND*	1/500	1/50000	1/10000000000	1/200000
布魯姆症候群	Bloom syndrome	AR	*BLM*	1/800	1/3478	1/48393195	1/13913
脆性X 症候群	Fragile X syndrome	XL	*FMR1*	1/250	1/25000	1/1250000000	1/33333
多發性翼狀腎肉症候群	Multiple pterygium syndrome, lethal type	AR	*CHRNG*	<1/500	1/50000	1/10000000000	1/200000
發-肝-腸症候群	Trichohepatoenteric syndrome 1	AR	*TTC37*	1/500	1/50000	1/10000000000	1/200000
骨質疏鬆症-假神經膠質瘤症候群	Osteoporosis-pseudoglioma syndrome	AR	*LRP5*	7/5000	1/71429	1/20408163265	1/285714
肌肝腦眼侏儒症	Mulibrey nanism	AR	*TRIM37*	1/100	1/500	1/1000000	1/2000
科克因綜合症A 型	Cockayne syndrome, type A	AR	*ERCC8*	1/822	1/11743	1/551578776	1/46971
科克因綜合症B 型	Cockayne syndrome, type B	AR	*ERCC6*	<1/500	1/25000	1/2500000000	1/100000
羅伯茲症候群	Roberts-SC phocomelia syndrome	AR	*ESCO2*	<1/500	1/50000	1/10000000000	1/200000
免疫缺陷-著絲粒不穩定-面部異常綜合症1 型	Immunodeficiency-centromeric instability-facial anomalies syndrome 1	AR	*DNMT3B*	<1/500	1/50000	1/10000000000	1/200000
奈梅亨斷裂症候群	Nijmegen breakage syndrome	AR	*NBN*	1/158	1/1129	1/5094694	1/4514
薩拉疾病	Salla disease	AR	*SLC17A5*	<1/500	1/10000	1/400000000	1/40000
少汗性外胚層發育不良	Ectodermal dysplasia 1, hypohidrotic, X-linked	XL	*EDA*	<1/500	1/4167	1/69444444	1/16667
腎小管酸中毒伴隨耳聾	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	AR	*ATP6V1B1*	<1/500	1/50000	1/10000000000	1/200000
視網膜色素變性糖尿病耳聾氏症候群（Alstrom 綜合徵）	Alstrom syndrome	AR	*ALMS1*	<1/500	1/50000	1/10000000000	1/200000
斯蒂爾症候群	Steel syndrome	AR	*COL27A1*	<1/500	1/50000	1/10000000000	1/200000

先天性無痛無汗症	Insensitivity to pain, congenital, with anhidrosis	AR	*NTRK1*	<1/500	1/781	1/2441406	1/3125
粒線體複合物III 缺陷	Mitochondrial complex III deficiency, nuclear type 1	AR	*BCS1L*	<1/500	1/50000	1/10000000000	1/200000
粒線體神經胃腸型腦肌病變（MNGIE）	Mitochondrial DNA depletion syndrome 1 (MNGIE type)	AR	*TYMP*	<1/500	1/50000	1/10000000000	1/200000
謝迪亞克—東氏症候群	Chediak-Higashi syndrome	AR	*LYST*	<1/500	1/3125	1/39062500	1/12500
心臟瓣膜型Ehlers-Danlos 症候群	Ehlers-Danlos syndrome, cardiac valvular type	AR	*COL1A2*	1/500	1/4545	1/82644628	1/18182
眼瞼囊腫-掌跖角化症-牙發育不全和少毛綜合症	Schopf-Schulz-Passarge syndrome	AR	*WNT10A*	<1/500	1/50000	1/10000000000	1/200000