疾病名稱	帶因率	偵測率	發生率
01.胺基酸/有機酸代謝異常			
藥物型苯酮尿症 (Phenylketonuria, PKU) 6-pyruvoyl-tetrahydropterin Synthase Deficiency (*PTS*) NM_000317:1-6 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 350 Middle East: 1 in 45 Other Populations: < 1 in 500	Eastern Asia: 99% Middle East: 99% Other Populations: 99%	Eastern Asia: < 1 in 35,000 Middle East: < 1 in 4,400 Other Populations: < 1 in 50,000
苯酮尿症-苯丙胺酸去氫酶缺乏症 Phenylalanine Hydroxylase Deficiency (*PAH*) NM_000277:1-13 Inheritance: Autosomal Recessive	African American: 1 in 160 Ashkenazi Jewish: 1 in 220 Finland: 1 in 200 Middle East: 1 in 26 Native American: 1 in 220 Other Populations: 1 in 51	African American: 99% Ashkenazi Jewish: 99% Finland: 99% Middle East: 99% Native American: 99% Other Populations: 99%	African American: < 1 in 16,000 Ashkenazi Jewish: < 1 in 22,000 Finland: < 1 in 20,000 Middle East: < 1 in 2,500 Native American: < 1 in 22,000 Other Populations: < 1 in 5,000
非酮性高甘胺酸血症 (Nonketotic hyperglycinemia, NKH) AMT-related Glycine Encephalopathy (*AMT*) NM_000481:1-9 Inheritance: Autosomal Recessive	Finland: 1 in 120 Other Populations: 1 in 220	Finland: 99% Other Populations: 99%	Finland: < 1 in 12,000 Other Populations: < 1 in 22,000
生物素酶缺乏症 (BTD deficiency) Biotinidase Deficiency (*BTD*) NM_000060:1-4 Inheritance: Autosomal Recessive	African American: 1 in 310 Ashkenazi Jewish: 1 in 440 Eastern Asia: 1 in 460 Hispanic: 1 in 160 Northwestern Europe: 1 in 130 Southeast Asia: 1 in 160 Other Populations: 1 in 160	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Hispanic: 99% Northwestern Europe: 99% Southeast Asia: 99% Other Populations: 99%	African American: < 1 in 38,000 Ashkenazi Jewish: < 1 in 60,000 Eastern Asia: < 1 in 67,000 Hispanic: < 1 in 17,000 Northwestern Europe: < 1 in 13,000 Southeast Asia: < 1 in 18,000 Other Populations: < 1 in 17,000
非酮性高甘胺酸血症(Nonketotic hyperglycinemia)/甘氨酸腦病 GLDC-related Glycine Encephalopathy (*GLDC*) NM_000170:1-25 Inheritance: Autosomal Recessive	Finland: 1 in 120 Other Populations: 1 in 160	Finland: 94% Other Populations: 94%	Finland: < 1 in 2,100 Other Populations: < 1 in 2,800
戊二酸血症 第一型 Glutaric Acidemia, GCDH-related (*GCDH*) NM_000159:2-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 140 French Canadian/Cajun: 1 in 160 Middle East: 1 in 66 Northwestern Europe: 1 in 160 Southern Europe: 1 in 140 Other Populations: 1 in 160	Eastern Asia: 99% French Canadian/Cajun: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 13,000 French Canadian/Cajun: < 1 in 16,000 Middle East: < 1 in 6,500 Northwestern Europe: < 1 in 16,000 Southern Europe: < 1 in 14,000 Other Populations: < 1 in 16,000

3羥基3甲基戊二酸尿症 (HMG尿症、白胺酸代謝異常) HMG-CoA Lyase Deficiency (*HMGCL*) NM_000191:1-9 Inheritance: Autosomal Recessive	Middle East: 1 in 160 Southern Europe: 1 in 160 Other Populations: < 1 in 500	Middle East: 99% Southern Europe: 99% Other Populations: 99%	Middle East: < 1 in 10,000 Southern Europe: < 1 in 10,000 Other Populations: < 1 in 33,000
多發性羧化酶缺乏症 (Multiple carboxylase deficiency)-羧化全酶合成酶缺乏症 Holocarboxylase Synthetase Deficiency (*HLCS*) NM_000411:4-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 160 Other Populations: 1 in 150	Eastern Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 16,000 Other Populations: < 1 in 15,000
高胱胺酸血症-胱硫醚β合成酶缺乏型 Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (*CBS*) NM_000071:3-17 Inheritance: Autosomal Recessive	Worldwide: 1 in 250	Worldwide: 99%	Worldwide: < 1 in 25,000
異戊酸血症 (IVA) Isovaleric Acidemia (*IVD*) NM_002225:1-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 250	Worldwide: 99%	Worldwide: < 1 in 25,000
甲基丙二酸血症(MMA) MUT-related Methylmalonic Acidemia (*MUT*) NM_000255:2-13 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 110 French Canadian/Cajun: 1 in 380 Hispanic: 1 in 170 Middle East: 1 in 53 Northwestern Europe: 1 in 260 South Asia: 1 in 72 Southeast Asia: 1 in 54 Other Populations: 1 in 180	Eastern Asia: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 11,000 French Canadian/Cajun: < 1 in 38,000 Hispanic: < 1 in 17,000 Middle East: < 1 in 5,200 Northwestern Europe: < 1 in 26,000 South Asia: < 1 in 7,100 Southeast Asia: < 1 in 5,300 Other Populations: < 1 in 18,000
甲基丙二酸血症 (MMA) cblA型 Methylmalonic Acidemia, cblA Type (*MMAA*) NM_172250:2-7 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 470 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 47,000 Other Populations: < 1 in 50,000
甲基丙二酸血症 (MMA) cblB型 Methylmalonic Acidemia, cblB Type (*MMAB*) NM_052845:1-9 Inheritance: Autosomal Recessive	French Canadian/Cajun: < 1 in 660 Northwestern Europe: 1 in 480 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Northwestern Europe: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 66,000 Northwestern Europe: < 1 in 48,000 Other Populations: < 1 in 50,000
甲基丙二酸血症 (MMA) cblC型 (伴隨高胱胺酸尿症) Methylmalonic Aciduria and Homocystinuria, cblC Type (*MMACHC*) NM_015506:1-4 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 330 French Canadian/Cajun: 1 in 230 Other Populations: 1 in 160	Eastern Asia: 99% French Canadian/Cajun: 99% Other Populations: 99%	Eastern Asia: < 1 in 33,000 French Canadian/Cajun: < 1 in 23,000 Other Populations: < 1 in 16,000

楓糖尿症 (第一B型) Maple Syrup Urine Disease Type 1B (*BCKDHB*) NM_183050:1-10 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 97 Other Populations: 1 in 250	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 9,600 Other Populations: < 1 in 25,000
楓糖尿症(第二型) Maple Syrup Urine Disease Type II (*DBT*) NM_001918:1-11 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 French Canadian/Cajun: 1 in 480 Middle East: 1 in 120 Southeast Asia: 1 in 280 Southern Europe: 1 in 410 Other Populations: 1 in 480	Eastern Asia: 96% French Canadian/Cajun: 95% Middle East: 96% Southeast Asia: 96% Southern Europe: 96% Other Populations: 96%	Eastern Asia: < 1 in 13,000 French Canadian/Cajun: < 1 in 9,600 Middle East: < 1 in 3,300 Southeast Asia: < 1 in 7,600 Southern Europe: < 1 in 11,000 Other Populations: < 1 in 13,000
楓糖尿症 (第一A型) Maple Syrup Urine Disease Type Ia (*BCKDHA*) NM_000709:1-9 Inheritance: Autosomal Recessive	African American: 1 in 260 Ashkenazi Jewish: 1 in 320 Eastern Asia: 1 in 490 Finland: 1 in 320 French Canadian/Cajun: 1 in 290 Hispanic: 1 in 150 Middle East: 1 in 110 Native American: 1 in 320 Northwestern Europe: 1 in 420 South Asia: 1 in 95 Southeast Asia: 1 in 190 Southern Europe: 1 in 160	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99%	African American: < 1 in 26,000 Ashkenazi Jewish: < 1 in 32,000 Eastern Asia: < 1 in 49,000 Finland: < 1 in 32,000 French Canadian/Cajun: < 1 in 29,000 Hispanic: < 1 in 14,000 Middle East: < 1 in 11,000 Native American: < 1 in 32,000 Northwestern Europe: < 1 in 42,000 South Asia: < 1 in 9,400 Southeast Asia: < 1 in 19,000 Southern Europe: < 1 in 16,000
丙酸血症（PA）_PCCA變異造成 PCCA-related Propionic Acidemia (*PCCA*) NM_000282:1-24 Inheritance: Autosomal Recessive	Middle East: 1 in 91 Other Populations: 1 in 220	Middle East: 95% Other Populations: 95%	Middle East: < 1 in 1,700 Other Populations: < 1 in 4,200
丙酸血症（PA）_PCCB變異造成 PCCB-related Propionic Acidemia (*PCCB*) NM_000532:1-15 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 66 Middle East: 1 in 100 Other Populations: 1 in 220	Eastern Asia: 1 in 66 Middle East: 1 in 100 Other Populations: 1 in 220	Eastern Asia: < 1 in 6,500 Middle East: < 1 in 10,000 Other Populations: < 1 in 22,000
遺傳性酪胺酸血症第一型 Tyrosinemia Type I (*FAH*) NM_000137:1-14 Inheritance: Autosomal Recessive	Finland: 1 in 120 French Canadian/Cajun: 1 in 64 Other Populations: 1 in 160	Finland: 99% French Canadian/Cajun: 99% Other Populations: 99%	Finland: < 1 in 12,000 French Canadian/Cajun: < 1 in 6,300 Other Populations: < 1 in 16,000
酪胺酸血症第二型 (Richner-Hanhart 症候群) Tyrosinemia Type II (*TAT*) NM_000353:2-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 250	Worldwide: 99%	Worldwide: < 1 in 25,000

02.尿素循環代謝異常			
精胺酸血症 Argininemia (*ARG1*) NM_000045:1-8 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 97%	Worldwide: < 1 in 17,000
精胺丁二酸酵素缺乏症 Argininosuccinic Aciduria (*ASL*) NM_001024943:1-16 Inheritance: Autosomal Recessive	Finland: 1 in 190 Hispanic: 1 in 290 Other Populations: 1 in 130	Finland: 99% Hispanic: 99% Other Populations: 99%	Finland: < 1 in 19,000 Hispanic: < 1 in 29,000 Other Populations: < 1 in 13,000
瓜胺酸血症第一型 Citrullinemia Type 1 (*ASS1*) NM_000050:3-16 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 97 Northwestern Europe: 1 in 140 Other Populations: 1 in 120	Eastern Asia: 86% Northwestern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 700 Northwestern Europe: < 1 in 14,000 Other Populations: < 1 in 12,000
鳥胺酸氨甲醯基轉移酶缺乏症(OTC) Ornithine Transcarbamylase Deficiency (*OTC*) NM_000531:1-10 Inheritance: X-linked Recessive	Finland: 1 in 31,000 Other Populations: 1 in 34,000	Finland: 97% Other Populations: 97%	Finland: < 1 in 1,000,000 Other Populations: < 1 in 1,000,000
03.其他代謝異常			
緬克斯症候群 (Menkes disease) ATP7A-related Disorders (*ATP7A*) NM_000052:2-23 Inheritance: X-linked Recessive	Eastern Asia: 1 in 180,000 Northwestern Europe: 1 in 150,000 Southern Europe: 1 in 150,000 Other Populations: 1 in 150,000	Eastern Asia: 92% Northwestern Europe: 96% Southern Europe: 96% Other Populations: 92%	Eastern Asia: < 1 in 1,000,000 Northwestern Europe: < 1 in 1,000,000 Southern Europe: < 1 in 1,000,000 Other Populations: < 1 in 1,000,000
α-甘露糖苷儲積症 Alpha-mannosidosis (*MAN2B1*) NM_000528:1-23 Inheritance: Autosomal Recessive	Worldwide: 1 in 350	Worldwide: 99%	Worldwide: < 1 in 35,000
天冬氨醯葡萄糖胺尿症 Aspartylglucosaminuria (*AGA*) NM_000027:1-9 Inheritance: Autosomal Recessive	Finland: 1 in 71 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 7,000 Other Populations: < 1 in 50,000
肉鹼棕櫚醯基轉移酶缺乏症第1型 Carbamoylphosphate Synthetase I Deficiency (*CPS1*) NM_001875:1-38 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 450 Finland: 1 in 370 Other Populations: < 1 in 570	Eastern Asia: 99% Finland: 99% Other Populations: 99%	Eastern Asia: < 1 in 45,000 Finland: < 1 in 37,000 Other Populations: < 1 in 57,000

肉鹼棕櫚醯基轉移酶缺乏症第1A型 Carnitine Palmitoyltransferase IA Deficiency (*CPT1A*) NM_001876:2-19 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
肉鹼棕櫚醯基轉移酶缺乏症第2型 Carnitine Palmitoyltransferase II Deficiency (*CPT2*) NM_000098:1-5 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 47 Eastern Asia: 1 in 320 Middle East: 1 in 110 Northwestern Europe: 1 in 250 Southern Europe: 1 in 200 Other Populations: 1 in 180	Ashkenazi Jewish: 99% Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 4,600 Eastern Asia: < 1 in 31,000 Middle East: < 1 in 11,000 Northwestern Europe: < 1 in 25,000 Southern Europe: < 1 in 20,000 Other Populations: < 1 in 18,000
腦腱性黃瘤症 Cerebrotendinous Xanthomatosis (*CYP27A1*) NM_000784:1-9 Inheritance: Autosomal Recessive	Worldwide: 1 in 110	Worldwide: 99%	Worldwide: < 1 in 11,000
先天性醣基化障礙(CDG)1a型 Congenital Disorder of Glycosylation Type Ia (*PMM2*) NM_000303:1-8 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
先天性醣基化障礙(CDG)1b型 Congenital Disorder of Glycosylation Type Ib (*MPI*) NM_002435:1-8 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 500
先天性醣基化障礙(CDG)1c型 Congenital Disorder of Glycosylation Type Ic (*ALG6*) NM_013339:2-15 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 500
胱胺酸血症 Cystinosis (*CTNS*) NM_004937:3-12 Inheritance: Autosomal Recessive	Worldwide: 1 in 220	Worldwide: 99%	Worldwide: < 1 in 22,000
丙酮酸鹽脫氫酵素缺乏症 (Pyruvate dehydrogenase deficiency)-E3缺乏症 Dihydrolipoamide Dehydrogenase Deficiency (*DLD*) NM_000108:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 94 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 9,300 Other Populations: < 1 in 50,000
法布瑞氏症 Fabry Disease (*GLA*) NM_000169:1-7 Inheritance: X-linked Recessive	Worldwide: 1 in 20,000	Worldwide: 98%	Worldwide: < 1 in 1,000,000

黏脂質症 (Mucolipidosis) GNPTAB-related Disorders (*GNPTAB*) NM_024312:1-21 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 320 French Canadian/Cajun: 1 in 40 Middle East: 1 in 140 Other Populations: 1 in 320	Eastern Asia: 98% French Canadian/Cajun: 99% Middle East: 99% Other Populations: 99%	Eastern Asia: < 1 in 17,000 French Canadian/Cajun: < 1 in 3,900 Middle East: < 1 in 14,000 Other Populations: < 1 in 32,000
GRACILE 症候群 GRACILE Syndrome (*BCS1L*) NM_004328:3-9 Inheritance: Autosomal Recessive	Finland: 1 in 110 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 11,000 Other Populations: < 1 in 50,000
半乳糖血症-半乳糖-1-磷酸尿甘醯轉移酵素異常 Galactosemia (*GALT*) NM_000155:1-11 Inheritance: Autosomal Recessive	African American: 1 in 71 Ashkenazi Jewish: 1 in 160 Eastern Asia: 1 in 320 Northwestern Europe: 1 in 87 Other Populations: 1 in 110	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Northwestern Europe: 99% Other Populations: 99%	African American: < 1 in 7,000 Ashkenazi Jewish: < 1 in 16,000 Eastern Asia: < 1 in 32,000 Northwestern Europe: < 1 in 8,600 Other Populations: < 1 in 11,000
半乳糖血症-半乳糖激酶缺乏症 Galactokinase Deficiency (*GALK1*) NM_000154:1-8 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Northwestern Europe: 1 in 100 Southern Europe: 1 in 310 Other Populations: 1 in 350	Eastern Asia: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 50,000 Northwestern Europe: < 1 in 10,000 Southern Europe: < 1 in 31,000 Other Populations: < 1 in 35,000
高雪氏症 Gaucher Disease (*GBA*) N409S, V433L, D448H, D448V, L483P, R502C, R502H, R535H, c.84dupG, c.115+1G>A Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 14 Eastern Asia: 1 in 220 Finland: 1 in 110 French Canadian/Cajun: 1 in 110 Northwestern Europe: 1 in 110 Southern Europe: 1 in 110 Other Populations: 1 in 120	Ashkenazi Jewish: 95% Eastern Asia: 60% Finland: 60% French Canadian/Cajun: 60% Northwestern Europe: 60% Southern Europe: 60% Other Populations: 60%	Ashkenazi Jewish: 1 in 270 Eastern Asia: < 1 in 560 Finland: 1 in 280 French Canadian/Cajun: 1 in 280 Northwestern Europe: 1 in 280 Southern Europe: 1 in 280 Other Populations: 1 in 310
遺傳性果糖不耐症, 果酸尿症 Hereditary Fructose Intolerance (*ALDOB*) NM_000035:2-9 Inheritance: Autosomal Recessive	African American: 1 in 230 French Canadian/Cajun: 1 in 81 Middle East: 1 in 98 Other Populations: 1 in 80	African American: 99% French Canadian/Cajun: 99% Middle East: 99% Other Populations: 99%	African American: < 1 in 23,000 French Canadian/Cajun: < 1 in 8,000 Middle East: < 1 in 9,700 Other Populations: < 1 in 7,900
低磷酸酯酶症 Hypophosphatasia (*ALPL*) NM_000478:2-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 190 Northwestern Europe: 1 in 270 Southern Europe: 1 in 270 Other Populations: 1 in 220	Eastern Asia: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 19,000 Northwestern Europe: < 1 in 27,000 Southern Europe: < 1 in 27,000 Other Populations: < 1 in 22,000
嬰兒型溶酶體酸性脂肪酶缺乏症(伍爾曼氏症, Wolman Disease) Lysosomal Acid Lipase Deficiency (*LIPA*) NM_000235:2-10 Inheritance: Autosomal Recessive	Hispanic: 1 in 180 Northwestern Europe: 1 in 180 Southern Europe: 1 in 180 Other Populations: 1 in 300	Hispanic: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Hispanic: < 1 in 18,000 Northwestern Europe: < 1 in 18,000 Southern Europe: < 1 in 18,000 Other Populations: < 1 in 30,000

黏多醣症第一型 Mucopolysaccharidosis Type I (*IDUA*) NM_000203:1-14 Inheritance: Autosomal Recessive	Middle East: 1 in 80 Other Populations: 1 in 160	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 7,900 Other Populations: < 1 in 16,000
黏多醣症第二型 Mucopolysaccharidosis Type II (*IDS*) NM_000202:1-9 Inheritance: X-linked Recessive	Eastern Asia: 1 in 24,000 Northwestern Europe: 1 in 38,000 Other Populations: 1 in 75,000	Eastern Asia: 88% Northwestern Europe: 88% Other Populations: 88%	Eastern Asia: 1 in 200,000 Northwestern Europe: 1 in 300,000 Other Populations: 1 in 600,000
黏多醣症第三A型 Mucopolysaccharidosis Type IIIA (*SGSH*) NM_000199:1-8 Inheritance: Autosomal Recessive	Middle East: 1 in 150 Northwestern Europe: 1 in 120 Other Populations: 1 in 160	Middle East: 99% Northwestern Europe: 99% Other Populations: 99%	Middle East: < 1 in 14,000 Northwestern Europe: < 1 in 12,000 Other Populations: < 1 in 16,000
黏多醣症第三B型 Mucopolysaccharidosis Type IIIB (*NAGLU*) NM_000263:1-6 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 300 Middle East: < 1 in 500 Northwestern Europe: 1 in 250 Southern Europe: 1 in 180 Other Populations: 1 in 310	Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 30,000 Middle East: < 1 in 50,000 Northwestern Europe: < 1 in 25,000 Southern Europe: < 1 in 18,000 Other Populations: < 1 in 31,000
黏多醣症第三C型 Mucopolysaccharidosis Type IIIC (*HGSNAT*) NM_152419:1-18 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Middle East: < 1 in 500 Northwestern Europe: 1 in 370	Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99%	Eastern Asia: < 1 in 50,000 Middle East: < 1 in 50,000 Northwestern Europe: < 1 in 37,000
黏脂質症第三型γ Mucolipidosis III Gamma (*GNPTG*) NM_032520:1-11 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
黏脂質症第四型 Mucolipidosis IV (*MCOLN1*) NM_020533:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 90 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 8,900 Other Populations: < 1 in 50,000
MLD症候群 Metachromatic Leukodystrophy (*ARSA*) NM_000487:1-8 Inheritance: Autosomal Recessive	Native American: 1 in 41 Other Populations: 1 in 160	Native American: 99% Other Populations: 99%	Native American: < 1 in 4,000 Other Populations: < 1 in 16,000
尼曼匹克症C1型 Niemann-Pick Disease Type C (*NPC1*) NM_000271:1-25 Inheritance: Autosomal Recessive	Worldwide: 1 in 190	Worldwide: 99%	Worldwide: < 1 in 19,000

尼曼匹克症C2型 Niemann-Pick Disease Type C2 (*NPC2*) NM_006432:1-5 Inheritance: Autosomal Recessive	Worldwide: 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
尼曼匹克症A(B)型(差別在於酵素活性的高低) Niemann-Pick Disease, SMPD1-associated (*SMPD1*) NM_000543:1-6 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 100 Other Populations: 1 in 250	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 10,000 Other Populations: < 1 in 25,000
肝醣儲積症第一a型 Glycogen Storage Disease Type Ia (*G6PC*) NM_000151:1-5 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 71 Other Populations: 1 in 180	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 7,000 Other Populations: < 1 in 18,000
肝醣儲積症第一b型 Glycogen Storage Disease Type Ib (*SLC37A4*) NM_001164277:3-11 Inheritance: Autosomal Recessive	Worldwide: 1 in 350	Worldwide: 99%	Worldwide: < 1 in 35,000
龐貝氏症(肝醣儲積症第二型glycogen storage disorder, GSD; glycogenosis II) Pompe Disease (*GAA*) NM_000152:2-20 Inheritance: Autosomal Recessive	African American: 1 in 60 Eastern Asia: 1 in 110 Hispanic: 1 in 160 Northwestern Europe: 1 in 160 Southeast Asia: 1 in 110 Southern Europe: 1 in 160 Other Populations: 1 in 160	African American: 99% Eastern Asia: 99% Hispanic: 99% Northwestern Europe: 98% Southeast Asia: 99% Southern Europe: 98% Other Populations: 99%	African American: < 1 in 5,900 Eastern Asia: < 1 in 11,000 Hispanic: < 1 in 10,000 Northwestern Europe: < 1 in 6,300 Southeast Asia: < 1 in 11,000 Southern Europe: < 1 in 6,300 Other Populations: < 1 in 16,000
肝醣儲積症第三型 Glycogen Storage Disease Type III (*AGL*) NM_000642:2-34 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
原發性肉鹼缺乏症 Primary Carnitine Deficiency (*SLC22A5*) NM_003060:1-10 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 100 Northwestern Europe: 1 in 110 Other Populations: 1 in 160	Eastern Asia: 99% Northwestern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 10,000 Northwestern Europe: < 1 in 11,000 Other Populations: < 1 in 16,000
先天性高乳酸血症 (Congenital Hyperlactic Acidemia)-丙酮酸去氫酶缺乏 Pyruvate Carboxylase Deficiency (*PC*) NM_000920:3-22 Inheritance: Autosomal Recessive	Worldwide: 1 in 250	Worldwide: 99%	Worldwide: < 1 in 25,000

肢近端型點狀軟骨發育不良第一型 Rhizomelic Chondrodysplasia Punctata Type 1 (*PEX7*) NM_000288:1-10 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
短鏈脂肪酸去氫酶缺乏症 Short-chain Acyl-CoA Dehydrogenase Deficiency (*ACADS*) NM_000017:1-10 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 110 Other Populations: 1 in 98	Northwestern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 11,000 Other Populations: < 1 in 9,700
中鏈脂肪酸去氫酵素缺乏症 (MCAD) Medium Chain Acyl-CoA Dehydrogenase Deficiency (*ACADM*) NM_000016:1-12 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 110 Middle East: 1 in 68 Northwestern Europe: 1 in 45 Southern Europe: 1 in 62 Other Populations: 1 in 61	Eastern Asia: 99% Middle East: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 11,000 Middle East: < 1 in 6,700 Northwestern Europe: < 1 in 4,400 Southern Europe: < 1 in 6,100 Other Populations: < 1 in 6,000
長鏈酰基輔酶A脫氫酶缺乏症 (Long-Chain-AcylCoA Dehydrogenase) HADHA-related Disorders (*HADHA*) NM_000182:1-20 Inheritance: Autosomal Recessive	Finland: 1 in 130 Northwestern Europe: 1 in 200 Other Populations: 1 in 250	Finland: 99% Northwestern Europe: 99% Other Populations: 99%	Finland: < 1 in 12,000 Northwestern Europe: < 1 in 20,000 Other Populations: < 1 in 25,000
極長鏈醯輔酶A去氫酶缺乏症 (VLCAD) Very-long-chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*) NM_000018:1-20 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 130 Northwestern Europe: 1 in 180 Southern Europe: 1 in 200 Other Populations: 1 in 140	Eastern Asia: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 12,000 Northwestern Europe: < 1 in 18,000 Southern Europe: < 1 in 20,000 Other Populations: < 1 in 14,000
威爾森氏症 Wilson Disease (*ATP7B*) NM_000053:1-21 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 51 Southeast Asia: 1 in 51 Other Populations: 1 in 87	Eastern Asia: 99% Southeast Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 5,000 Southeast Asia: < 1 in 5,000 Other Populations: < 1 in 8,600
腎上腺腦白質失養症(ALD) X-linked Adrenoleukodystrophy (*ABCD1*) NM_000033:1-6 Inheritance: X-linked Recessive	Eastern Asia: 1 in 20,000 Southern Europe: 1 in 14,000 Other Populations: 1 in 11,000	Eastern Asia: 77% Southern Europe: 77% Other Populations: 77%	Eastern Asia: 1 in 86,000 Southern Europe: 1 in 60,000 Other Populations: 1 in 45,000
04.皮膚病變			
Herlitz交界型表皮溶解水皰症 (泡泡龍)-LAMA3變異 Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (*LAMA3*) NM_000227:1-38 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000

Herlitz交界型表皮溶解水皰症 (泡泡龍)-LAMB3變異 Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (*LAMB3*) NM_000228:2-23 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
Herlitz交界型表皮溶解水皰症 (泡泡龍)-LAMC2變異 Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (*LAMC2*) NM_005562:1-23 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
先天性角化不全症(DC)_RTEL1變異 RTEL1-related Disorders (*RTEL1*) NM_032957:2-35 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 100 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 10,000 Other Populations: < 1 in 50,000
Sjogren-Larsson氏症候群 Sjogren-Larsson Syndrome (*ALDH3A2*) NM_000382:1-10 Inheritance: Autosomal Recessive	Worldwide: 1 in 250	Worldwide: 97%	Worldwide: < 1 in 9,100
層狀魚鱗癬（自體隱性遺傳型） TGM1-related Autosomal Recessive Congenital Ichthyosis (*TGM1*) NM_000359:2-15 Inheritance: Autosomal Recessive	Worldwide: 1 in 220	Worldwide: 99%	Worldwide: < 1 in 22,000
著色性乾皮症(XP)A型 Xeroderma Pigmentosum Group A (*XPA*) NM_000380:1-6 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 100 Middle East: 1 in 280 South Asia: 1 in 280 Other Populations: < 1 in 500	Eastern Asia: 99% Middle East: 99% South Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 10,000 Middle East: < 1 in 28,000 South Asia: < 1 in 28,000 Other Populations: < 1 in 50,000
著色性乾皮症(XP)C型 Xeroderma Pigmentosum Group C (*XPC*) NM_004628:1-16 Inheritance: Autosomal Recessive	Worldwide: 1 in 240	Worldwide: 97%	Worldwide: < 1 in 7,300
05.肌肉病變			
裘馨氏肌肉萎縮症/貝克氏肌肉萎縮症 Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (*DMD*) NM_004006:1-79 Inheritance: X-linked Recessive		Worldwide: 99%	Worldwide: < 1 in 3,500
肢帶型肌肉失養症2A型 Calpainopathy (*CAPN3*) NM_000070:1-24 Inheritance: Autosomal Recessive	Worldwide: 1 in 130	Worldwide: 99%	Worldwide: < 1 in 13,000

肢帶型肌肉失養症2B型-修肌蛋白缺乏症/三好氏肌肉病變 (Miyoshi myopathy) Dysferlinopathy (*DYSF*) NM_003494:1-55 Inheritance: Autosomal Recessive	Worldwide: 1 in 190	Worldwide: 98%	Worldwide: < 1 in 11,000
肢帶型肌肉失養症2C型-γ-肌聚糖病 Gamma-sarcoglycanoCpathy (*SGCG*) NM_000231:2-8 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 380 Other Populations: 1 in 350	Eastern Asia: 88% Other Populations: 88%	Eastern Asia: < 1 in 3,200 Other Populations: < 1 in 3,000
肢帶型肌肉失養症2D型-α-肌聚糖病 Alpha-sarcoglycanopathy (*SGCA*) NM_000023:1-9 Inheritance: Autosomal Recessive	Worldwide: 1 in 450	Worldwide: 99%	Worldwide: < 1 in 45,000
肢帶型肌肉失養症2E型-β-肌聚糖病 Beta-sarcoglycanopathy (*SGCB*) NM_000232:1-6 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
肢帶型肌肉失養症2F型-δ-肌聚糖病 Delta-sarcoglycanopathy (*SGCD*) NM_000337:2-9 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 40,000
肢帶型肌肉失養症2I型 FKRP-related Disorders (*FKRP*) NM_024301:4 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Northwestern Europe: 1 in 160 South Asia: < 1 in 500 Southeast Asia: < 1 in 500 Other Populations: 1 in 190	Eastern Asia: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 50,000 Northwestern Europe: < 1 in 16,000 South Asia: < 1 in 50,000 Southeast Asia: < 1 in 50,000 Other Populations: < 1 in 19,000
肢帶型肌肉失養症2M型 FKTN-related Disorders (*FKTN*) NM_001079802:3-11 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 150 Eastern Asia: 1 in 190 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Eastern Asia: 10% Other Populations: 99%	Ashkenazi Jewish: < 1 in 15,000 Eastern Asia: 1 in 210 Other Populations: < 1 in 50,000
遺傳性包涵體肌炎 (IBM) Inclusion Body Myopathy 2 (*GNE*) NM_001128227:1-12 Inheritance: Autosomal Recessive	Middle East: 1 in 130 Other Populations: < 1 in 500	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 12,000 Other Populations: < 1 in 50,000

先天性肌失養症 (Congenital Muscular Dystrophy)-層粘連蛋白α-2缺乏症(非福山型) LAMA2-related Muscular Dystrophy (*LAMA2*) NM_000426:1-65 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 610 Northwestern Europe: 1 in 340 Southern Europe: 1 in 340 Other Populations: 1 in 170	Eastern Asia: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 61,000 Northwestern Europe: < 1 in 34,000 Southern Europe: < 1 in 34,000 Other Populations: < 1 in 17,000
先天性肌失養症 (Congenital Muscular Dystrophy)-肌-眼-腦病(非福山型) Muscle-eye-brain Disease (*POMGNT1*) NM_017739:2-22 Inheritance: Autosomal Recessive	Finland: < 1 in 500 Other Populations: < 1 in 500	Finland: 98% Other Populations: 96%	Finland: < 1 in 25,000 Other Populations: < 1 in 12,000
線狀體肌肉病變第二型(NEM2) NEB-related Nemaline Myopathy (*NEB*) NM_001271208:3-80,117-183 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 110 Other Populations: 1 in 87	Ashkenazi Jewish: 99% Other Populations: 93%	Ashkenazi Jewish: < 1 in 11,000 Other Populations: < 1 in 1,200
性聯肌小管病變 X-linked Myotubular Myopathy (*MTM1*) NM_000252:2-15 Inheritance: X-linked Recessive	Worldwide: 1 in 500	Worldwide: 98%	Worldwide: <1 in 50,000
06.骨頭病變			
體染色體隱性骨質石化症 (大理石寶寶) Autosomal Recessive Osteopetrosis Type 1 (*TCIRG1*) NM_006019:2-20 Inheritance: Autosomal Recessive	Middle East: 1 in 97 Other Populations: 1 in 350	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 9,600 Other Populations: < 1 in 35,000
軟骨頭髮發育不全 Cartilage-hair Hypoplasia (*RMRP*) NR_003051:1 Inheritance: Autosomal Recessive	Finland: 1 in 76 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 7,500 Other Populations: < 1 in 50,000
緻密性骨發育不全症 Pycnodysostosis (*CTSK*) NM_000396:2-8 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
脊椎肋骨發育不全 Spondylothoracic Dysostosis (*MESP2*) NM_001039958:1-2 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
硫酸鹽轉運蛋白相關軟骨成長不全 Sulfate Transporter-related Osteochondrodysplasia (*SLC26A2*) NM_000112:2-3 Inheritance: Autosomal Recessive	Finland: 1 in 75 Other Populations: 1 in 110	Finland: 99% Other Populations: 99%	Finland: < 1 in 7,400 Other Populations: < 1 in 11,000

07.血液疾病			
再生不良性貧血(Aplastic anemia)-Fanconi氏貧血 Fanconi Anemia Complementation Group A (*FANCA*) NM_000135:1-43 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 260 Hispanic: 1 in 250 Northwestern Europe: 1 in 240 Southern Europe: 1 in 240 Other Populations: 1 in 260	Eastern Asia: 92% Hispanic: 92% Northwestern Europe: 92% Southern Europe: 92% Other Populations: 92%	Eastern Asia: < 1 in 3,100 Hispanic: < 1 in 2,900 Northwestern Europe: < 1 in 2,800 Southern Europe: < 1 in 2,800 Other Populations: < 1 in 3,100
再生不良性貧血(Aplastic anemia)-Fanconi氏貧血 Fanconi Anemia, FANCC-related (*FANCC*) NM_000136:2-15 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 94 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 9,300 Other Populations: < 1 in 50,000
α-地中海型貧血 Alpha Thalassemia (*HBA1/HBA2*) Variants Genotyped (13): Hb Constant Spring, -alpha4.2, anti4.2, -alpha3.7, anti3.7, --MEDII, --BRIT, -(alpha)20.5, --MEDI, --SEA, del HS-40, --THAI/--FIL, HBA1+HBA2 deletion. Inheritance: Autosomal Recessive	Worldwide: 1/270	Worldwide: 99%	Worldwide: < 1 in 27,000
β鏈相關血紅素病變 (β-地中海型貧血與鐮刀型貧血) Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (*HBB*) NM_000518:1-3 Inheritance: Autosomal Recessive	African American: 1 in 11 Ashkenazi Jewish: 1 in 67 Eastern Asia: 1 in 51 Finland: 1 in 80 French Canadian/Cajun: 1 in 67 Hispanic: 1 in 25 Middle East: 1 in 23 Native American: 1 in 67 Northwestern Europe: 1 in 32 South Asia: 1 in 23 Southeast Asia: 1 in 23 Southern Europe: 1 in 14	African American: 99% Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Middle East: 99% Native American: 99% Northwestern Europe: 99% South Asia: 99% Southeast Asia: 99% Southern Europe: 99%	African American: < 1 in 950 Ashkenazi Jewish: < 1 in 6,600 Eastern Asia: < 1 in 5,000 Finland: < 1 in 7,900 French Canadian/Cajun: < 1 in 6,600 Hispanic: < 1 in 2,400 Middle East: < 1 in 2,200 Native American: < 1 in 6,600 Northwestern Europe: < 1 in 3,100 South Asia: < 1 in 2,200 Southeast Asia: < 1 in 2,200 Southern Europe: < 1 in 1,300
08.腦部及神經病變			
複雜型體染色體隱性遺傳痙攣性下身麻痺(Hereditary Spastic Paraplegia, HSP) ARSACS (*SACS*) NM_014363:2-10 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 22 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 1,900 Other Populations: < 1 in 44,000
Andermann症候群 Andermann Syndrome (*SLC12A6*) NM_133647:1-25 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 23 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 2,200 Other Populations: < 1 in 50,000

共濟失調微血管擴張症候群(AT) Ataxia-telangiectasia (*ATM*) NM_000051:2-63 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 200 Finland: 1 in 200 French Canadian/Cajun: 1 in 200 Hispanic: 1 in 120 Northwestern Europe: 1 in 200 Southern Europe: 1 in 200 Other Populations: 1 in 120	Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 97% Northwestern Europe: 98% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 20,000 Finland: < 1 in 20,000 French Canadian/Cajun: < 1 in 20,000 Hispanic: < 1 in 3,700 Northwestern Europe: < 1 in 11,000 Southern Europe: < 1 in 18,000 Other Populations: < 1 in 12,000
神經元蠟樣脂褐質儲積症(NCLs)-嬰兒晚期型(芬蘭變異型) CLN5-related Neuronal Ceroid Lipofuscinosis (*CLN5*) NM_006493:1-4 Inheritance: Autosomal Recessive	Finland: 1 in 24 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 2,300 Other Populations: < 1 in 50,000
神經元蠟樣脂褐質儲積症(NCLs)-嬰兒晚期型(吉普賽/印度變異型) CLN6-related Neuronal Ceroid Lipofuscinosis (*CLN6*) NM_017882:1-7 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 430 Northwestern Europe: 1 in 430 Southern Europe: 1 in 430 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 43,000 Northwestern Europe: < 1 in 43,000 Southern Europe: < 1 in 43,000 Other Populations: < 1 in 50,000
神經元蠟樣脂褐質儲積症(NCLs)-青春型 CLN3-related Neuronal Ceroid Lipofuscinosis (*CLN3*) NM_001042432:2-16 Inheritance: Autosomal Recessive	Finland: 1 in 71 Northwestern Europe: 1 in 87 Southern Europe: 1 in 280 Other Populations: 1 in 130	Finland: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Finland: < 1 in 7,000 Northwestern Europe: < 1 in 8,600 Southern Europe: < 1 in 28,000 Other Populations: < 1 in 13,000
家族性軸突海綿退化 Canavan Disease (*ASPA*) NM_000049:1-6 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 55 Other Populations: 1 in 160	Ashkenazi Jewish: 98% Other Populations: 98%	Ashkenazi Jewish: < 1 in 3,300 Other Populations: < 1 in 9,700
家族性廣泛型自主神經病變 Familial Dysautonomia (*IKBKAP*) NM_003640:2-37 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 31 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 3,000 Other Populations: < 1 in 50,000
Joubert氏症候群 (家族性小腦蚓部發育不全) Joubert Syndrome 2 (*TMEM216*) NM_001173990:1-5 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 97 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 9,600 Other Populations: < 1 in 50,000
Krabbe氏症(球細胞腦白質失養症) Krabbe Disease (*GALC*) NM_000153:1-17 Inheritance: Autosomal Recessive	Ashkenazi Jewish: < 1 in 500 Other Populations: 1 in 150	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 50,000 Other Populations: < 1 in 15,000

Leigh症候群 (LS)-亞急性壞死性腦脊髓病 Leigh Syndrome, French-Canadian Type (*LRPPRC*) NM_133259:1-38 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 23 Other Populations: < 1 in 500	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 2,200 Other Populations: < 1 in 50,000
GM1神經節苷脂儲積症 (GM1 Gangliosidosis) GLB1-related Disorders (*GLB1*) NM_000404:1-16 Inheritance: Autosomal Recessive	Worldwide: 1 in 190	Worldwide: 99%	Worldwide: < 1 in 19,000
成年型GM2神經節甘脂儲積症 (包含戴薩克斯症) Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (*HEXA*) NM_000520:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 31 French Canadian/Cajun: 1 in 51 Other Populations: 1 in 300	Ashkenazi Jewish: 99% French Canadian/Cajun: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 3,000 French Canadian/Cajun: < 1 in 5,000 Other Populations: < 1 in 30,000
GM2 神經節苷脂儲積症-山德霍夫氏病 Sandhoff Disease (*HEXB*) NM_000521:1-14 Inheritance: Autosomal Recessive	Ashkenazi Jewish: < 1 in 500 French Canadian/Cajun: 1 in 120 Northwestern Europe: 1 in 320 Southern Europe: 1 in 320 Other Populations: 1 in 320	Ashkenazi Jewish: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 47,000 French Canadian/Cajun: < 1 in 11,000 Northwestern Europe: < 1 in 32,000 Southern Europe: < 1 in 32,000 Other Populations: < 1 in 30,000
神經元蠟樣脂褐質儲積症-北方癲癇型(NE) Northern Epilepsy (*CLN8*) NM_018941:2-3 Inheritance: Autosomal Recessive	Finland: 1 in 140 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 13,000 Other Populations: < 1 in 50,000
神經元蠟樣脂褐質儲積症-PPT1變異 PPT1-related Neuronal Ceroid Lipofuscinosis (*PPT1*) NM_000310:1-9 Inheritance: Autosomal Recessive	Finland: 1 in 71 Other Populations: 1 in 78	Finland: 99% Other Populations: 99%	Finland: < 1 in 7,000 Other Populations: < 1 in 7,700
神經元蠟樣脂褐質儲積症-TPP1變異 TPP1-related Neuronal Ceroid Lipofuscinosis (*TPP1*) NM_000391:1-13 Inheritance: Autosomal Recessive	Worldwide: 1 in 300	Worldwide: 99%	Worldwide: < 1 in 30,000
Zellweger氏症候群_PEX1變異 PEX1-related Zellweger Syndrome Spectrum (*PEX1*) NM_000466:1-24 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 350 Native American: 1 in 350 South Asia: 1 in 350 Southeast Asia: 1 in 350 Other Populations: 1 in 110	Eastern Asia: 99% Native American: 99% South Asia: 99% Southeast Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 35,000 Native American: < 1 in 35,000 South Asia: < 1 in 35,000 Southeast Asia: < 1 in 35,000 Other Populations: < 1 in 11,000
過氧化物酶體生源失調症第三型-*Zellweger氏症候群_PEX12變異* Peroxisome Biogenesis Disorder Type 3 (*PEX12*) NM_000286:1-3 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 500 Other Populations: 1 in 440	Eastern Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 50,000 Other Populations: < 1 in 44,000

過氧化物酶體生源失調症第四型-*Zellweger氏症候群_PEX6變異* Peroxisome Biogenesis Disorder Type 4 (***PEX6***) NM_000287:1-17 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 56 Other Populations: 1 in 310	French Canadian/Cajun: 97% Other Populations: 97%	French Canadian/Cajun: < 1 in 1,600 Other Populations: < 1 in 9,300
過氧化物酶體生源失調症第五型 Peroxisome Biogenesis Disorder Type 5 (***PEX2***) NM_000318:4 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 120 Other Populations: < 1 in 710	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 12,000 Other Populations: < 1 in 71,000
過氧化物酶體生源失調症第六型-*Zellweger氏症候群_PEX10變異* Peroxisome Biogenesis Disorder Type 6 (***PEX10***) NM_153818:1-6 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
瀨川氏症(SS)/多巴胺反應性肌張力不全症(DRD) Segawa Syndrome (***TH***) NM_199292:1-14 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
遺傳性痙攣性下身麻痺(HSP)第15型 (SPG15) Spastic Paraplegia Type 15 (***ZFYVE26***) NM_015346:2-42 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
脊髓性肌肉萎縮症(SMA) Spinal Muscular Atrophy (***SMN1***) SMN1 copy number Inheritance: Autosomal Recessive	African American: 1 in 66 Ashkenazi Jewish: 1 in 41 Eastern Asia: 1 in 53 Finland: 1 in 35 French Canadian/Cajun: 1 in 35 Hispanic: 1 in 120 Middle East: 1 in 50 Native American: 1 in 50 Northwestern Europe: 1 in 35 South Asia: 1 in 50 Southeast Asia: 1 in 53 Southern Europe: 1 in 57	African American: 71% Ashkenazi Jewish: 94% Eastern Asia: 93% Finland: 94% French Canadian/Cajun: 95% Hispanic: 91% Middle East: 92% Native American: 93% Northwestern Europe: 95% South Asia: 93% Southeast Asia: 93% Southern Europe: 94%	African American: 1 in 120 Ashkenazi Jewish: 1 in 350 Eastern Asia: < 1 in 630 Finland: < 1 in 560 French Canadian/Cajun: < 1 in 570 Hispanic: < 1 in 1,100 Middle East: < 1 in 560 Native American: < 1 in 690 Northwestern Europe: < 1 in 630 South Asia: < 1 in 630 Southeast Asia: < 1 in 630 Southern Europe: < 1 in 890
09.外觀異常			

科恩綜合症 Cohen Syndrome (*VPS13B*) NM_017890:2-62 Inheritance: Autosomal Recessive	Finland: 1 in 160 Other Populations: < 1 in 500	Finland: 97% Other Populations: 97%	Finland: < 1 in 4,800 Other Populations: < 1 in 15,000
Hydrolethalus 綜合症 Hydrolethalus Syndrome (*HYLS1*) NM_145014:4 Inheritance: Autosomal Recessive	Finland: 1 in 71 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 7,000 Other Populations: < 1 in 50,000
Meckel-Gruber症候群-MKSI變異 MKS1-related Disorders (*MKS1*) NM_017777:1-18 Inheritance: Autosomal Recessive	Finland: 1 in 48 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 4,700 Other Populations: < 1 in 50,000
Smith-Lemli-Opitz症候群 Smith-Lemli-Opitz Syndrome (*DHCR7*) NM_001360:3-9 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 100 Finland: 1 in 100 French Canadian/Cajun: 1 in 100 Hispanic: 1 in 140 Northwestern Europe: 1 in 50 Southern Europe: 1 in 83 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 10,000 Finland: < 1 in 10,000 French Canadian/Cajun: < 1 in 10,000 Hispanic: < 1 in 13,000 Northwestern Europe: < 1 in 4,900 Southern Europe: < 1 in 8,200 Other Populations: < 1 in 50,000
10.感覺器官失能(聽覺/視覺)			
Costeff症候群 Costeff Optic Atrophy Syndrome (*OPA3*) NM_025136:1-2 Inheritance: Autosomal Recessive	Middle East: 1 in 51 Other Populations: < 1 in 500	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 5,000 Other Populations: < 1 in 50,000
遺傳性耳聾-GJB2變異 GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (*GJB2*) NM_004004:1-2 Inheritance: Autosomal Recessive	African American: 1 in 48 Ashkenazi Jewish: 1 in 21 Finland: 1 in 42 French Canadian/Cajun: 1 in 42 Northwestern Europe: 1 in 33 Southern Europe: 1 in 42 Other Populations: 1 in 100	African American: 99% Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	African American: < 1 in 4,700 Ashkenazi Jewish: < 1 in 2,000 Finland: < 1 in 4,100 French Canadian/Cajun: < 1 in 4,100 Northwestern Europe: < 1 in 3,200 Southern Europe: < 1 in 4,100 Other Populations: < 1 in 10,000
遺傳性耳聾-MYO7A變異 MYO7A-related Disorders (*MYO7A*) NM_000260:2-49 Inheritance: Autosomal Recessive	Worldwide: 1 in 150	Worldwide: 99%	Worldwide: < 1 in 15,000

Pendred氏症候群 Pendred Syndrome (*SLC26A4*) NM_000441:2-21 Inheritance: Autosomal Recessive	Worldwide: 1 in 71	Worldwide: 99%	Worldwide: < 1 in 7,000
尤塞氏综合症 PCDH15-related Disorders (*PCDH15*) NM_033056:2-33 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 78 Other Populations: 1 in 220	Ashkenazi Jewish: 93% Other Populations: 93%	Ashkenazi Jewish: < 1 in 1,200 Other Populations: < 1 in 3,300
尤塞氏综合症1C型 USH1C-related Disorders (*USH1C*) NM_005709:1-21 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 230 Other Populations: 1 in 350	French Canadian/Cajun: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 23,000 Other Populations: < 1 in 35,000
尤塞氏综合症第二型 USH2A-related Disorders (*USH2A*) NM_206933:2-72 Inheritance: Autosomal Recessive	Worldwide: 1 in 130	Worldwide: 94%	Worldwide: < 1 in 2,200
尤塞氏综合症第三型 Usher Syndrome Type 3 (*CLRN1*) NM_174878:1-3 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 120 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 12,000 Other Populations: < 1 in 50,000
X染色體串聯視網膜裂損症 X-linked Juvenile Retinoschisis (*RS1*) NM_000330:1-6 Inheritance: X-linked Recessive	Ashkenazi Jewish: 1 in 10,000 Finland: 1 in 8,500 French Canadian/Cajun: 1 in 10,000 Northwestern Europe: 1 in 10,000 Southern Europe: 1 in 10,000 Other Populations: 1 in 13,000	Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: 1 in 670,000 Finland: 1 in 570,000 French Canadian/Cajun: 1 in 670,000 Northwestern Europe: 1 in 670,000 Southern Europe: 1 in 670,000 Other Populations: 1 in 840,000
11.免疫疾病			
嚴重複合型免疫缺乏症(severe combined immunodeficiency, SCID)-腺苷脱氨酶缺乏症 Adenosine Deaminase Deficiency (*ADA*) NM_000022:1-12 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 220 Other Populations: 1 in 390	Northwestern Europe: 99% Other Populations: 99%	Northwestern Europe: < 1 in 22,000 Other Populations: < 1 in 39,000
自體免疫多腺體症候群第一型(APS) Autoimmune Polyglandular Syndrome Type 1 (*AIRE*) NM_000383:1-14 Inheritance: Autosomal Recessive	Finland: 1 in 80 Northwestern Europe: 1 in 150 Other Populations: 1 in 180	Finland: 99% Northwestern Europe: 99% Other Populations: 99%	Finland: < 1 in 7,900 Northwestern Europe: < 1 in 15,000 Other Populations: < 1 in 18,000

嚴重複合型免疫缺乏症 (SCID) X-linked Severe Combined Immunodeficiency (*IL2RG*) NM_000206:1-8 Inheritance: X-linked Recessive	Worldwide: 1 in 50,000	Worldwide: 99%	Worldwide: < 1 in 1,000,000
12.心肺功能失調			
囊狀纖維化 (CF) Cystic Fibrosis (*CFTR*) NM_000492:1-27 Inheritance: Autosomal Recessive	African American: 1 in 66 Ashkenazi Jewish: 1 in 28 Finland: 1 in 80 French Canadian/Cajun: 1 in 16 Hispanic: 1 in 46 Northwestern Europe: 1 in 28 Southern Europe: 1 in 28 Other Populations: 1 in 87	African American: 99% Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Hispanic: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	African American: < 1 in 6,500 Ashkenazi Jewish: < 1 in 2,700 Finland: < 1 in 7,900 French Canadian/Cajun: < 1 in 1,500 Hispanic: < 1 in 4,500 Northwestern Europe: < 1 in 2,700 Southern Europe: < 1 in 2,700 Other Populations: < 1 in 8,600
埃利偉氏症候群-EVC變異 EVC-related Ellis-van Creveld Syndrome (*EVC*) NM_153717:1-21 Inheritance: Autosomal Recessive	Worldwide: 1 in 330	Worldwide: 96%	Worldwide: < 1 in 7,500
埃利偉氏症候群-EVC2 EVC2-related Ellis-van Creveld Syndrome (*EVC2*) NM_147127:1-22 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
13.泌尿系統問題			
體染色體隱性多囊性腎臟疾病 (ARPKD) Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (*PKHD1*) NM_138694:2-67 Inheritance: Autosomal Recessive	Finland: 1 in 52 Other Populations: 1 in 82	Finland: 99% Other Populations: 99%	Finland: < 1 in 5,100 Other Populations: < 1 in 8,100
亞伯氏症候群-COL4A3變異 COL4A3-related Alport Syndrome (*COL4A3*) NM_000091:1-52 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 180 Finland: 1 in 370 Northwestern Europe: 1 in 210 Southern Europe: 1 in 210 Other Populations: 1 in 350	Ashkenazi Jewish: 97% Finland: 97% Northwestern Europe: 97% Southern Europe: 97% Other Populations: 97%	Ashkenazi Jewish: < 1 in 5,500 Finland: < 1 in 11,000 Northwestern Europe: < 1 in 6,200 Southern Europe: < 1 in 6,200 Other Populations: < 1 in 11,000
亞伯氏症候群-COL4A4變異 COL4A4-related Alport Syndrome (*COL4A4*) NM_000092:2-48 Inheritance: Autosomal Recessive	Finland: 1 in 370 Northwestern Europe: 1 in 210 Southern Europe: 1 in 210 Other Populations: 1 in 350	Finland: 98% Northwestern Europe: 98% Southern Europe: 98% Other Populations: 98%	Finland: < 1 in 22,000 Northwestern Europe: < 1 in 12,000 Southern Europe: < 1 in 13,000 Other Populations: < 1 in 21,000

亞伯氏症候群-COL4A5變異 COL4A5-related Alport Syndrome (*COL4A5*) NM_000495:1-51 Inheritance: Inheritance: X-linked Recessive	Worldwide: < 1 in 500	Worldwide: 95%	Worldwide: <1 in 50,000
芬蘭型先天性腎病症候群 Congenital Finnish Nephrosis (*NPHS1*) NM_004646:1-29 Inheritance: Autosomal Recessive	Finland: 1 in 45 Other Populations: < 1 in 500	Finland: 99% Other Populations: 99%	Finland: < 1 in 4,400 Other Populations: < 1 in 50,000
NPHS2相關腎病症候群 Nephrotic Syndrome, NPHS2-related (*NPHS2*) NM_014625:1-8 Inheritance: Autosomal Recessive	French Canadian/Cajun: 1 in 360 Middle East: 1 in 360 Native American: 1 in 360 Southeast Asia: 1 in 360 Southern Europe: 1 in 360 Other Populations: 1 in 360	French Canadian/Cajun: 99% Middle East: 99% Native American: 99% Southeast Asia: 99% Southern Europe: 99% Other Populations: 99%	French Canadian/Cajun: < 1 in 35,000 Middle East: < 1 in 35,000 Native American: < 1 in 35,000 Southeast Asia: < 1 in 35,000 Southern Europe: < 1 in 35,000 Other Populations: < 1 in 35,000
原發型高草酸鹽尿症第一型(PH1) Primary Hyperoxaluria Type 1 (*AGXT*) NM_000030:1-11 Inheritance: Autosomal Recessive	Worldwide: 1 in 350	Worldwide: 99%	Worldwide: < 1 in 35,000
原發型高草酸鹽尿症第二型(PH2) Primary Hyperoxaluria Type 2 (*GRHPR*) NM_012203:1-9 Inheritance: Autosomal Recessive	Worldwide: 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
原發型高草酸鹽尿症第三型(PH3) Primary Hyperoxaluria Type 3 (*HOGA1*) NM_138413:1-7 Inheritance: Autosomal Recessive	African American: < 1 in 500 Ashkenazi Jewish: 1 in 87 Finland: 1 in 130 French Canadian/Cajun: 1 in 130 Northwestern Europe: 1 in 130 Southern Europe: 1 in 130 Other Populations: 1 in 200	African American: 99% Ashkenazi Jewish: 99% Finland: 99% French Canadian/Cajun: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	African American: < 1 in 50,000 Ashkenazi Jewish: < 1 in 8,600 Finland: < 1 in 13,000 French Canadian/Cajun: < 1 in 13,000 Northwestern Europe: < 1 in 13,000 Southern Europe: < 1 in 13,000 Other Populations: < 1 in 20,000
14.內分泌疾病			
11β-羥化酶缺乏-先天性腎上腺皮質增生(congenital adrenal hyperplasia, CAH) 11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (*CYP11B1*) NM_000497:1-9 Inheritance: Autosomal Recessive	Northwestern Europe: 1 in 220 Southern Europe: 1 in 220 Other Populations: 1 in 190	Northwestern Europe: 94% Southern Europe: 94% Other Populations: 94%	Northwestern Europe: < 1 in 3,800 Southern Europe: < 1 in 3,800 Other Populations: < 1 in 3,300

21-羥化酶缺乏-先天性腎上腺皮質增生(congenital adrenal hyperplasia, CAH) 21-hydroxylase-deficient Congenital Adrenal Hyperplasia (*CYP21A2*) I173N, V282L, R357W, P31L, c.293-13C>G, G111VfsX21, Q319*, L308FfsX6, CYP21A2 deletion, CYP21A2 duplication, Q319*+CYP21A2dup, [I237N;V238E;M240K], CYP21A2 triplication Inheritance: Autosomal Recessive	African American: 1 in 120 Ashkenazi Jewish: 1 in 58 Eastern Asia: 1 in 72 Finland: 1 in 58 French Canadian/Cajun: 1 in 58 Hispanic: 1 in 56 Middle East: 1 in 42 Native American: 1 in 56 Northwestern Europe: 1 in 58 South Asia: 1 in 42 Southeast Asia: 1 in 59 Southern Europe: 1 in 58	African American: 92% Ashkenazi Jewish: 99% Eastern Asia: 88% Finland: 89% French Canadian/Cajun: 96% Hispanic: 95% Middle East: 97% Native American: 90% Northwestern Europe: 96% South Asia: 89% Southeast Asia: 88% Southern Europe: 96%	African American: < 1 in 1,400 Ashkenazi Jewish: < 1 in 5,700 Eastern Asia: < 1 in 590 Finland: < 1 in 530 French Canadian/Cajun: < 1 in 1,400 Hispanic: < 1 in 1,100 Middle East: < 1 in 1,200 Native American: < 1 in 550 Northwestern Europe: < 1 in 1,400 South Asia: 1 in 360 Southeast Asia: 1 in 480 Southern Europe: < 1 in 1,300
Alstrom 氏症候群 Alstrom Syndrome (*ALMS1*) NM_015120:1-23 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
巴德-畢德氏症候群 (BBS)-BBS1變異 Bardet-Biedl Syndrome, BBS1-related (*BBS1*) NM_024649:1-17 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
巴德-畢德氏症候群 (BBS)-BBS2變異 Bardet-Biedl Syndrome, BBS2-related (*BBS2*) NM_031885:1-17 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 140 Middle East: 1 in 180 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Middle East: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 14,000 Middle East: < 1 in 18,000 Other Populations: < 1 in 50,000
巴德-畢德氏症候群 (BBS)-BBS10變異 Bardet-Biedl Syndrome, BBS10-related (*BBS10*) NM_024685:1-2 Inheritance: Autosomal Recessive	African American: 1 in 440 Eastern Asia: < 1 in 500 French Canadian/Cajun: 1 in 150 Middle East: 1 in 110 Northwestern Europe: 1 in 420 South Asia: < 1 in 500 Southern Europe: < 1 in 500 Other Populations: 1 in 420	African American: 99% Eastern Asia: 99% French Canadian/Cajun: 99% Middle East: 99% Northwestern Europe: 99% South Asia: 99% Southern Europe: 99% Other Populations: 99%	African American: < 1 in 44,000 Eastern Asia: < 1 in 50,000 French Canadian/Cajun: < 1 in 15,000 Middle East: < 1 in 11,000 Northwestern Europe: < 1 in 42,000 South Asia: < 1 in 50,000 Southern Europe: < 1 in 50,000 Other Populations: < 1 in 42,000
巴德-畢德氏症候群 (BBS)-BBS2變異 Bardet-Biedl Syndrome, BBS12-related (*BBS12*) NM_152618:2 Inheritance: Autosomal Recessive	Middle East: 1 in 210 Other Populations: < 1 in 500	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 20,000 Other Populations: < 1 in 50,000

聯合腦垂體激素缺乏 (CPHD)第二型-PROP1變異 Combined Pituitary Hormone Deficiency, PROP1-related (*PROP1*) NM_006261:1-3 Inheritance: Autosomal Recessive	Worldwide: 1 in 62	Worldwide: 99%	Worldwide: < 1 in 6,100
家族性持續性幼兒型胰島素過度分泌低血糖症 (PHHI)-ABCC8變異 ABCC8-related Familial Hyperinsulinism (*ABCC8*) NM_000352:1-39 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 45 Eastern Asia: 1 in 140 Finland: 1 in 100 Middle East: 1 in 140 Other Populations: 1 in 170	Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% Middle East: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 4,400 Eastern Asia: < 1 in 14,000 Finland: < 1 in 10,000 Middle East: < 1 in 14,000 Other Populations: < 1 in 17,000
家族性持續性幼兒型胰島素過度分泌低血糖症 (PHHI)-KCNJ11變異 KCNJ11-related Familial Hyperinsulinism (*KCNJ11*) NM_000525:1 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 200 Eastern Asia: 1 in 420 Finland: 1 in 450 Middle East: 1 in 410 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Eastern Asia: 99% Finland: 99% Middle East: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 20,000 Eastern Asia: < 1 in 42,000 Finland: < 1 in 45,000 Middle East: < 1 in 41,000 Other Populations: < 1 in 50,000
先天性類脂質性腎上腺增生症 (CLAH) Lipoid Congenital Adrenal Hyperplasia (*STAR*) NM_000349:1-7 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 400 Other Populations: < 1 in 500	Eastern Asia: 99% Other Populations: 99%	Eastern Asia: < 1 in 40,000 Other Populations: < 1 in 50,000
先天性腎上腺發育不全-性染色體隱性遺傳型 X-linked Congenital Adrenal Hypoplasia (*NR0B1*) NM_000475:1-2 Inheritance: X-linked Recessive	Worldwide: 1 in 300,000	Worldwide: 99%	Worldwide: < 1 in 1,000,000
15.染色體異常			
布盧姆症候群 Bloom Syndrome (*BLM*) NM_000057:2-22 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 110 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 11,000 Other Populations: < 1 in 50,000
X染色體脆折症 Fragile X Syndrome (*FMR1*) FMR1 CGG repeat number Inheritance: X-linked	Not Calculated	Worldwide: 99%	Not Calculated
Nijmegen斷裂症候群 Nijmegen Breakage Syndrome (*NBN*) NM_002485:1-16 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 99%	Worldwide: < 1 in 16,000
16.其他分類			

共濟失調伴維生素E缺乏症 (AVED) Ataxia with Vitamin E Deficiency (*TTPA*) NM_000370:1-5 Inheritance: Autosomal Recessive	Middle East: 1 in 160 Other Populations: < 1 in 500	Middle East: 99% Other Populations: 99%	Middle East: < 1 in 16,000 Other Populations: < 1 in 50,000
丁型雙功能蛋白缺乏症 D-bifunctional Protein Deficiency (*HSD17B4*) NM_000414:1-24 Inheritance: Autosomal Recessive	Worldwide: 1 in 160	Worldwide: 98%	Worldwide: < 1 in 9,000
柯凱因氏症候群 (Cockayne syndrome)-ERCC6變異 ERCC6-related Disorders (*ERCC6*) NM_000124:2-21 Inheritance: Autosomal Recessive	Eastern Asia: 1 in 370 Northwestern Europe: 1 in 380 Southern Europe: 1 in 380 Other Populations: 1 in 280	Eastern Asia: 99% Northwestern Europe: 99% Southern Europe: 99% Other Populations: 99%	Eastern Asia: < 1 in 26,000 Northwestern Europe: < 1 in 26,000 Southern Europe: < 1 in 26,000 Other Populations: < 1 in 19,000
柯凱因氏症候群 (Cockayne syndrome)-ERCC8變異 ERCC8-related Disorders (*ERCC8*) NM_000082:1-12 Inheritance: Autosomal Recessive	Eastern Asia: < 1 in 510 Northwestern Europe: < 1 in 520 Southern Europe: < 1 in 520 Other Populations: 1 in 380	Eastern Asia: 95% Northwestern Europe: 95% Southern Europe: 95% Other Populations: 95%	Eastern Asia: < 1 in 9,800 Northwestern Europe: < 1 in 9,900 Southern Europe: < 1 in 9,900 Other Populations: < 1 in 7,300
家族性地中海熱 (FMF) Familial Mediterranean Fever (*MEFV*) NM_000243:1-10 Inheritance: Autosomal Recessive	Ashkenazi Jewish: 1 in 11 Middle East: 1 in 16 Southern Europe: 1 in 110 Other Populations: < 1 in 500	Ashkenazi Jewish: 99% Middle East: 99% Southern Europe: 99% Other Populations: 99%	Ashkenazi Jewish: < 1 in 1,000 Middle East: < 1 in 1,500 Southern Europe: < 1 in 10,000 Other Populations: < 1 in 50,000
巨腦性腦白質病伴有皮層下囊腫第一型 Megalencephalic Leukoencephalopathy with Subcortical Cysts (*MLC1*) NM_015166:2-12 Inheritance: Autosomal Recessive	Worldwide: < 1 in 500	Worldwide: 99%	Worldwide: < 1 in 50,000
唾液酸儲積病 Salla Disease (*SLC17A5*) NM_012434:1-11 Inheritance: Autosomal Recessive	Finland: 1 in 100 Other Populations: < 1 in 500	Finland: 99% Other Populations: 98%	Finland: < 1 in 10,000 Other Populations: < 1 in 30,000