孕知因 精選版-多疾病帶因篩檢

項目	疾病名稱(中文)*	疾病名稱(英文)	基因	遺傳模式	種族	帶因率	檢出率	檢測後 帶因風險	檢測後 胎兒患病風險
1	醯輔酶A去氫酶9缺乏症	Acyl-CoA dehydrogenase-9 (ACAD9) Deficiency	ACAD9	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
2	甲型海洋性貧血	Alpha thalassemia	HBA1/ HBA2	AR	General	1 in 20	90%	1 in 191	1 in 15,280
					East Asian	1 in 8	90%	1 in 71	1 in 2,272
					South Asian/Indian	1 in 5	90%	1 in 41	1 in 820
3	精胺酸酶缺乏症	Arginase deficiency	ARG1	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
4	精胺丁二酸酶缺乏症	Argininosuccinate lyase deficiency	ASL	AR	General	1 in 132	90%	1 in 1,311	1 in 692,208
5	生物素酵素缺乏症	Biotinidase deficiency	BTD	AR	General	1 in 124	99%	1 in 12,301	1 in 6,101,296
6	布隆氏症候群	Bloom syndrome	BLM	AR	General	1 in 800	87%	1 in 6,147	<1 in 10 million
7	卡那凡氏症	Canavan disease	ASPA	AR	General	1 in 300	97%	1 in 9,968	<1 in 10 million
8	肉鹼結合酶缺乏症第一型	Carnitine palmitoyltransferase IA deficiency	CPT1A	AR	General	1 in 354	90%	1 in 3,531	1 in 4,999,896
9	肉鹼結合酶缺乏症第二型	Carnitine palmitoyltransferase II deficiency	CPT2	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
10	瓜胺酸血症	Citrullinemia	ASS1	AR	General	1 in 119	96%	1 in 2,951	1 in 1,404,676
					East Asian	1 in 132	96%	1 in 3,276	1 in 1,729,728
11	先天性腎上腺增生症-17a羥化酶缺失症	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency	CYP17A1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
12	先天性腎上腺增生症-21羥化酶缺失症	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency	CYP21A2	AR	General	1 in 61	99%	1 in 6,001	1 in 1,464,244
13	先天性醣基化疾病-1a型	Congenital disorder of glycosylation type 1a	PMM2	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
14	囊狀纖維化	Cystic fibrosis	CFTR	AR	General	1 in 32	99%	1 in 3101	1 in 396,928
					East Asian	1 in 94	99%	1 in 9301	1 in 3,497,176
15	裘馨氏肌肉失養症	Duchenne muscular dystrophy†	DMD	XL	General	1 in 2,350	93%	1 in 33,558	1 in 134,260
16	法布瑞氏症	Fabry disease†	GLA	XL	General	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
17	Fanconi氏貧血C型	Fanconi anemia group C	FANCC	AR	General	1 in 535	99%	1 in 53,401	<1 in 10 million
18	家族性自主神經功能異常	Familial dysautonomia	ELP1	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
19	X染色體脆折症	Fragile X syndrome†	FMR1	XL	General	1 in 151	99%	1 in 15,001	1 in 60,004
20	半乳糖血症	Galactosemia	GALT	AR	General	1 in 110	95%	1 in 2,181	1 in 959,640
21	高雪氏症	Gaucher disease	GBA	AR	General	1 in 77	99%	1 in 7,601	1 in 2,341,108
22	葡萄糖-六-磷酸鹽去氫酶缺乏症(蠶豆症)	Glucose-6-phosphate dehydrogenase deficiency†	G6PD	XL	General	1 in 7	98%	1 in 301	1 in 1,204
23	戊二酸尿症-I型	Glutaric aciduria, type I	GCDH	AR	General	1 in 87	98%	1 in 4,301	1 in 1,496,748
24	肝醣儲積症第1A型	Glycogen Storage disease, type 1a	G6PC	AR	General	1 in 177	95%	1 in 3,521	1 in 2,492,868
25	血友病A型	Hemophilia A†	F8	XL	General	1 in 3,250	48%	1 in 6,249	1 in 25,000
26	血友病B型	Hemophilia B†	F9	XL	General	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
27	高胱胺酸尿症	Homocystinuria due to cystathionine beta-synthase deficiency	CBS	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
28	異戊酸血症	Isovaleric acidemia	IVD	AR	General	1 in 167	90%	1 in 1,661	1 in 1,109,548
					East Asian	1 in 407	90%	1 in 4,061	1 in 6,611,308
29	楓糖尿症-Ia型	Maple syrup urine disease type Ia	BCKDHA	AR	General	1 in 321	98%	1 in 16,001	<1 in 10 million
30	楓糖尿症-Ib型	Maple syrup urine disease type Ib	BCKDHB	AR	General	1 in 364	98%	1 in 18,151	<1 in 10 million
31	楓糖尿症-II型	Maple syrup urine disease, type II	DBT	AR	General	1 in 481	98%	1 in 24,001	<1 in 10 million
32	中鏈脂肪酸去氫酶缺乏症	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	ACADM	AR	General	1 in 69	98%	1 in 3,401	1 in 938,676
					East Asian	1 in 198	99%	1 in 19,701	<1 in 10 million
33	異染性白質退化症	Metachromatic leukodystrophy	ARSA	AR	General	1 in 100	95%	1 in 1,981	1 in 792,400

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34	甲基丙二酸血症併高胱胺酸血症-cb1D型	Methylmalonic aciduria and homocystinuria, cblD type	MMADHC	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
35	甲基丙二酸血症-MUT型	Methylmalonic acidemia, MUT-related	MUT	AR	General	1 in 195	96%	1 in 4,851	1 in 3,783,780
					East Asian	1 in 53	96%	1 in 1,301	1 in 275,812
36	甲基丙二酸血症-cb1A型	Methylmalonic aciduria, cblA type	MMAA	AR	General	1 in 301	97%	1 in 10,001	<1 in 10 million
37	甲基丙二酸血症-cb1B型	Methylmalonic aciduria, cblB type	MMAB	AR	General	1 in 435	98%	1 in 21,701	<1 in 10 million
38	黏多醣症-1型(賀勒氏症)	Mucopolysaccharidosis, type I (Hurler syndrome)	IDUA	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
39	神經元蠟樣脂褐質沉著疾病-CLN3型	Neuronal ceroid lipofuscinosis, CLN3-related	CLN3	AR	General	1 in 230	98%	1 in 11,451	<1 in 10 million
40	尼曼匹克症-A/B型	Niemann-Pick disease, type A/B	SMPD1	AR	General	1 in 250	95%	1 in 4,981	1 in 4,981,000
41	非症候群型遺傳性聽障-GJB2型	Nonsyndromic hearing loss, GJB2-related	GJB2	AR/Digenic	General	1 in 42	99%	1 in 4,101	1 in 688,968
					South Asian/Indian	1 in 148	99%	1 in 14,701	1 in 8,702,992
42	非症候群型遺傳性聽障-GJB6型	Nonsyndromic hearing loss, GJB6-related	GJB6	AR/Digenic	General	1 in 423	99%	1 in 42,201	<1 in 10 million
43	Pendred 氏症候群	Pendred syndrome	SLC26A4	AR	General	1 in 80	98%	1 in 3,951	1 in 1,264,320
					East Asian	1 in 74	98%	1 in 3,651	1 in 1,080,696
44	苯酮尿症(苯丙胺酸羥基化酶缺乏症)	Phenylalanine hydroxylase deficiency (Phenylketonuria)	PAH	AR	General	1 in 93	99%	1 in 9,201	1 in 3,422,772
					South East Asian	1 in 59	99%	1 in 5,801	1 in 1,369,036
45	隱性多囊性腎疾病-PKHD1型	Polycystic kidney disease, PKHD1-related	PKHD1	AR	General	1 in 70	98%	1 in 3,451	1 in 966,280
46	龐貝氏症(肝糖儲積症第二型)	Pompe disease	GAA	AR	General	1 in 100	98%	1 in 4,951	1 in 1,980,400
					East Asian	1 in 112	98%	1 in 5,551	1 in 2,486,848
47	丙酸血症-PCCA型	Propionic acidemia, PCCA-related	PCCA	AR	General	1 in 224	96%	1 in 5,576	1 in 4,996,096
48	丙酸血症-PCCB型	Propionic acidemia, PCCB-related	PCCB	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
49	肢近端型點狀軟骨發育不良第一型	Rhizomelic chondrodysplasia punctata, type 1	PEX7	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
50	性聯遺傳嚴重複合型免疫缺乏症	Severe combined immunodeficiency, X-linked†	IL2RG	XL	General	1 in 25,000	99%	1 in 2,499,901	1 in 9,999,804
51	短鏈脂肪酸去氫酶缺乏症	Short-chain acyl-coA dehydrogenase (SCAD) Deficiency	ACADS	AR	General	1 in 85	99%	1 in 8,401	1 in 2,856,340
					South Asian/Indian	1 in 51	99%	1 in 5,001	1 in 1,020,204
52	鐮刀型紅血球症; 乙型海洋性貧血	Sickle cell disease; Beta thalassemia	HBB	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
					East Asian	1 in 50	95%	1 in 981	1 in 196,200
					South Asian/Indian	1 in 25	95%	1 in 481	1 in 48,100
53	Smith-Lemli-Opitz症候群	Smith-Lemli-Opitz syndrome	DHCR7	AR	General	1 in 30	96%	1 in 726	1 in 87,120
54	脊髓性肌肉萎縮症	Spinal muscular atrophy	SMN1	AR	General	1 in 54	91%	1 in 590	1 in 127,440
					East Asian	1 in 59	93%	1 in 830	1 in 195,880
55	原發性肉鹼缺乏症	Systemic primary carnitine deficiency	SLC22A5	AR	General	1 in 129	76%	1 in 534	1 in 275,544
					East Asian	1 in 77	76%	1 in 318	1 in 97,944
					South Asian/India	1 in 51	76%	1 in 209	1 in 42,636
56	戴薩克斯症	Tay-Sachs disease	HEXA	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
57	高酪胺酸血症-1型	Tyrosinemia, type 1	FAH	AR	General	1 in 99	95%	1 in 1,961	1 in 776,556
					South Asian/Indian	1 in 172	95%	1 in 3,421	1 in 2,353,648
58	高酪胺酸血症-2型	Tyrosinemia, type II	TAT	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
59	極長鏈醯輔酶A去氫酶缺乏症	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	ACADVL	AR	General	1 in 118	93%	1 in 1,672	1 in 789,184
					South Asian/Indian	1 in 73	93%	1 in 1,030	1 in 300,760
60	威爾森氏症	Wilson disease	ATP7B	AR	General	1 in 87	98%	1 in 4,301	1 in 1,496,748
61	柴爾維格氏症候群-PEX1型	Zellweger syndrome, PEX1-related	PEX1	AR	General	1 in 147	95%	1 in 2,921	1 in 1,717,548

†男性帶因篩檢不會檢測性聯遺傳疾病

*中文疾病名稱以衛生福利部國民健康署遺傳疾病諮詢網頁、財團法人罕病基金會及罕見疾病一點通命名為主

AR: 體染色體隱性遺傳；XL: X染色體隱性遺傳；Digenic: 雙基因遺傳，同時由兩個不同的基因互相影響而導致同一疾病發病。(不列入附件剩餘風險計算)

此份資料參考原廠2019.07 Beacon Expanded Carrier Supplemental Table