孕知因 全球版-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	3-氫基-3-甲基戊二酸血症	3-Hydroxy-3-methylglutaryl-CoA lyase deficiency	HMGCL	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
2	Beta硫解酶缺乏症	3-Ketothiolase deficiency	ACAT1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
3	三甲基巴豆醯輔酶A梭化酶缺乏症	3-Methylcrotonyl-CoA carboxylase 1 deficiency (3-MCC deficiency)	MCCC1	AR	General	1 in 95	98%	1 in 4,701	1 in 1,786,380
4	三甲基巴豆醯輔酶A梭化酶缺乏症	3-Methylcrotonyl-CoA carboxylase 2 deficiency (3-MCC deficiency)	MCCC2	AR	General	1 in 95	98%	1 in 4,701	1 in 1,786,380
5	無β脂蛋白血症	Abetalipoproteinemia	MTTP	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
6	軟骨生成不全症候群-1B型; 畸型發育不良-II型; 畸型發育不良; 多發性骨骺發育不全症	Achondrogenesis, type IB; Atelosteogenesis II; Diastrophic dysplasia; Multiple epiphyseal dysplasia	SLC26A2	AR	General	1 in 158	90%	1 in 1,571	1 in 992,872
7	色彩感應失能症	Achromatopsia	CNGB3	AR	General	1 in 87	99%	1 in 8,601	1 in 2,993,148
8	腸道內鋅吸收不良症	Acrodermatitis enteropathica	SLC39A4	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
9	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
10	腺嘌呤脫氨酶缺乏症	Adenosine deaminase deficiency	ADA	AR	General	1 in 224	93%	1 in 3,187	1 in 2,855,552
11	性聯遺傳腎上腺腦白質失養症	Adrenoleukodystrophy, X-linked†	ABCD1	XL	General	1 in 21,000	99%	1 in 2,099,901	1 in 8,399,804
12	Aicardi-Goutieres 症候群	Aicardi-Goutieres syndrome	SAMHD1	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
13	黑尿症	Alkaptonuria	HGD	AR	General	1 in 250	90%	1 in 2,491	1 in 2,491,000
14	Allan-Herndon-Dudley 症候群	Allan-Herndon-Dudley syndrome	SLC16A2	XL	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
15	甘露糖症	Alpha-mannosidosis	MAN2B1	AR	General	1 in 354	99%	1 in 35,301	<1 in 10 million
16	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
17	甲型海洋性貧血-性聯遺傳智力障礙症候群	Alpha thalassemia X-linked intellectual disability syndrome†	ATRX	XL	General	<1 in 250,000	99%	1 in 24,999,901	<1 in 10 million
18	艾柏症候群-COL4A3型	Alport syndrome, COL4A3-related	COL4A3	AR	General	1 in 267	98%	1 in 13,301	<1 in 10 million
19	艾柏症候群-COL4A4型	Alport syndrome, COL4A4-related	COL4A4	AR	General	1 in 267	98%	1 in 13,301	<1 in 10 million
20	艾柏症候群-COL4A5型	Alport syndrome, COL4A5-related†	COL4A5	XL	General	1 in 139	98%	1 in 6,901	1 in 27,604
21	Alstrom氏症候群	Alstrom syndrome	ALMS1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
22	軟骨頭髮發育不全; 幹骨后端的軟骨生成; 骨骼發育異常症候群	Anauxetic dysplasia; Cartilage-hair hypoplasia; Metaphyseal dysplasia without hypotrichosis	RMRP	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
23	安德曼综合症	Andermann syndrome	SLC12A6	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
24	精胺酸酶缺乏症	Arginase deficiency	ARG1	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
25	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
26	芳香環轉化酶缺乏症	Aromatase deficiency	CYP19A1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
27	遠端關節攣縮, 智能障礙及癲癇症候群	Arthrogryposis, mental retardation, and seizures	SLC35A3	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
28	Arts症候群; Rosenberg-Chutorian症候群; PRPS1基因相關症候群	Arts syndrome; Rosenberg-Chutorian syndrome; Phosphoribosylpyrophosphate synthetase superactivity; Non-syndromic hearing loss, PRPS1-related†	PRPS1	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
29	天門冬醯胺酶合成缺乏症	Asparagine synthetase deficiency	ASNS	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
30	天門冬醯胺葡萄糖胺尿症	Aspartylglucosaminuria	AGA	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
31	維他命E缺乏共濟失調症	Ataxia with isolated vitamin E deficiency	TTPA	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million

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32	共濟失調微血管擴張性症候群	Ataxia-telangiectasia	ATM	AR	General	1 in 100	92%	1 in 1,239	1 in 495,600
33	自體免疫多腺體症候群I型	Autoimmune polyendocrinopathy syndrome type I	AIRE	AR	General	1 in 150	98%	1 in 7,451	1 in 4,470,600
34	體染色體隱性遺傳痙攣性共濟失調症	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	SACS	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
35	Bardet-Biedl氏症候群-14型	Bardet-Biedl syndrome 14; Joubert syndrome 5; Leber congenital amaurosis 10; Meckel syndrome 4; Senior-Løken syndrome 6 ; CEP290-related disorders	CEP290	AR	General	1 in 190	98%	1 in 9,451	1 in 7,182,760
36	Bardet-Biedl氏症候群-2型	Bardet-Biedl syndrome 2; Retinitis Pigmentosa 74	BBS2	AR	General	1 in 621	99%	1 in 62,001	<1 in 10 million
37	Bardet-Biedl氏症候群-1型	Bardet-Biedl syndrome type 1	BBS1	AR	General	1 in 367	99%	1 in 36,601	<1 in 10 million
38	Bardet-Biedl氏症候群-10型	Bardet-Biedl syndrome type 10	BBS10	AR	General	1 in 395	99%	1 in 39,401	<1 in 10 million
39	Bardet-Biedl氏症候群-12型	Bardet-Biedl syndrome type 12	BBS12	AR	General	1 in 791	99%	1 in 79,001	<1 in 10 million
40	第二型裸淋巴細胞綜合症	Bare lymphocyte syndrome, type II	CIITA	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
41	Bartter氏症候群	Bartter syndrome	BSND	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
42	巨大血小板症候群-A1型	Bernard-Soulier syndrome type A1	GP1BA	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
43	巨大血小板症候群-C型	Bernard-Soulier syndrome type C	GP9	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
44	雙側額頂多小腦迴畸形症	Bilateral frontoparietal polymicrogyria	ADGRG1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
45	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
46	Björnstad症候群; Gracile症候群; 粒線體Complex III缺乏症	Björnstad syndrome; GRACILE syndrome; Mitochondrial complex III deficiency	BCS1L	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
47	布隆氏症候群	Bloom syndrome	BLM	AR	General	1 in 800	87%	1 in 6,147	<1 in 10 million
48	卡那凡氏症	Canavan disease	ASPA	AR	General	1 in 300	97%	1 in 9,968	<1 in 10 million
49	甲酉先磷酸合成酶缺乏症	Carbamoylphosphate synthetase I deficiency	CPS1	AR	General	1 in 570	98%	1 in 28,451	<1 in 10 million
50	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
51	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
52	肉鹼轉位酶缺乏症	Carnitine-acylcarnitine translocase deficiency	SLC25A20	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
53	Carpenter症候群	Carpenter syndrome	RAB23	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
54	兒茶酚胺敏感性多形性室性心搏過速(CPVT)-CASQ2型	Catecholaminergic polymorphic ventricular tachycardia, CASQ2-related	CASQ2	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
55	兒茶酚胺敏感性多形性室性心搏過速(CPVT)-TRDN型	Catecholaminergic polymorphic ventricular tachycardia, TRDN-related	TRDN	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
56	腦腱性黃瘤症	Cerebrotendinous xanthomatosis	CYP27A1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
57	進行性神經性腓骨萎縮症-4B1型	Charcot-Marie-Tooth disease, type 4B1	MTMR2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
58	進行性神經性腓骨萎縮症-4D型	Charcot-Marie-Tooth disease, type 4D	NDRG1	AR	General	1 in 22	98%	1 in 1,051	1 in 92,488
59	進行性神經性腓骨萎縮症-GDAP1型	Charcot-Marie-Tooth disease, GDAP1-related	GDAP1	AR	General	1 in 152	99%	1 in 15,101	1 in 9,181,408
60	進行性神經性腓骨萎縮症-SH3TC2型	Charcot-Marie-Tooth disease, SH3TC2-related	SH3TC2	AR	General	1 in 69	99%	1 in 6,801	1 in 1,877,076
61	性聯遺傳進行性神經性腓骨萎縮症-1型	Charcot-Marie-Tooth disease, X-linked type 1†	GJB1	XL	General	1 in 667	90%	1 in 6,661	1 in 26,644
62	Chediak-Higashi 症候群	Chediak-Higashi syndrome	LYST	AR	General	<1 in 500	90%	<1 in 4,991	1 in 9,982,000
63	幼兒型嚴重視網膜失養症-AIPL1型	Childhood-onset severe retinal dystrophy, AIPL1-related	AIPL1	AR	General	1 in 409	99%	1 in 40,801	<1 in 10 million
64	性聯遺傳點狀軟骨發育不良一型	Chondrodysplasia punctata type 1, X-linked†	ARSE	XL	General	1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
65	舞蹈棘狀紅血球症	Choreoacanthocytosis	VPS13A	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
66	脈絡膜缺失症	Choroideremia†	CHM	XL	General	1 in 25,000	95%	1 in 499,981	1 in 1,999,964

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67	慢性肉芽腫病	Chronic granulomatous disease	CYBA	AR	General	1 in 224	99%	1 in 22,301	<1 in 10 million
68	性聯遺傳慢性肉芽腫病	Chronic granulomatous disease, X-linked†	CYBB	XL	General	1 in 149,254	99%	1 in 14,925,301	<1 in 10 million
69	Citrin缺乏症	Citrin deficiency	SLC25A13	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
					East Asian	1 in 65	95%	1 in 1,281	1 in 333,060
70	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
71	A型柯凱因氏症候群	Cockayne syndrome type A	ERCC8	AR	General	1 in 822	98%	1 in 41,051	<1 in 10 million
72	B型柯凱因氏症候群	Cockayne syndrome type B; DeSanctis-Cacchione syndrome	ERCC6	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
					Japanese	1 in 74	99%	1 in 7,301	1 in 2,161,096
73	柯恩綜合症	Cohen syndrome	VPS13B	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
74	丙二酸及甲基丙二酸血症	Combined malonic and methylmalonic aciduria	ACSF3	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
75	氧化磷酸化缺失症-GFM1型	Combined oxidative phosphorylation deficiency, GFM1-related	GFM1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
76	氧化磷酸化缺失症-TSFM型	Combined oxidative phosphorylation deficiency, TSFM-related	TSFM	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
77	下垂體激素缺失症-2型	Combined pituitary hormone deficiency 2	PROP1	AR	General	1 in 45	98%	1 in 2,201	1 in 396,180
78	下垂體激素缺失症-3型	Combined pituitary hormone deficiency 3	LHX3	AR	General	1 in 45	98%	1 in 2,201	1 in 396,180
79	先天性腎上腺增生症-11b羥化酶缺失症	Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency	CYP11B1	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
80	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
81	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
82	先天性腎上腺增生症-3b羥化酶缺失症	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	HSD3B2	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
83	先天性腎上腺增生症-X染色體遺傳	Congenital adrenal hypoplasia,X-linked†	NR0B1	XL	General	1 in 6,250	99%	1 in 624,901	1 in 2,499,804
84	先天性無巨型細胞性血小板缺乏症	Congenital amegakaryocytic thrombocytopenia	MPL	AR	General	1 in 102	98%	1 in 5,051	1 in 2,060,808
85	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
86	先天性醣基化疾病-1b型	Congenital disorder of glycosylation type Ib	MPI	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
87	先天性醣基化疾病-1c型	Congenital disorder of glycosylation type Ic	ALG6	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
88	永久性新生兒糖尿病	Congenital hyperinsulinism; Permanent neonatal diabetes mellitus	KCNJ11	AR	General	1 in 423	99%	1 in 42,201	<1 in 10 million
89	先天性甲狀腺低功能症-DUOX2型	Congenital hypothyroidism, DUOX2-related	DUOX2	AR	General	1 in 366	91%	1 in 4,057	1 in 5,938,797
90	先天性甲狀腺低功能症-DUOXA2型	Congenital hypothyroidism, DUOXA2-related	DUOXA2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
91	先天性甲狀腺低功能症-TSHB型	Congenital hypothyroidism, TSHB-related	TSHB	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
92	先天性魚鱗癬狀紅皮症	Congenital ichthyosis	TGM1	AR	General	1 in 224	95%	1 in 4,461	1 in 3,997,056
93	先天性痛覺不敏感合併無汗症	Congenital insensitivity to pain with anhidrosis	NTRK1	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
94	先天性肌無力症候群-CHRNE型	Congenital myasthenic syndrome, CHRNE-related	CHRNE	AR	General	1 in 408	99%	1 in 40,701	<1 in 10 million
95	先天性肌無力症候群-RAPSN型	Congenital myasthenic syndrome, RAPSN-related; Fetal akinesia deformation sequence	RAPSN	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
96	先天性腎病症候群-1型	Congenital nephrotic syndrome, type 1	NPHS1	AR	General	1 in 289	98%	1 in 14,401	<1 in 10 million
97	先天性腎病症候群-2型	Congenital nephrotic syndrome, type 2	NPHS2	AR	General	1 in 289	98%	1 in 14,401	<1 in 10 million
98	先天性分泌性氯化物腹瀉	Congenital secretory chloride diarrhea	SLC26A3	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million

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99	眼角膜內皮細胞失養症	Corneal endothelial dystrophy	SLC4A11	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
100	皮質酮甲基氧化酶缺乏症	Corticosterone methyloxidase deficiency	CYP11B2	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
101	Costeff症候群	Costeff syndrome	OPA3	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
102	肌酸缺乏症候群	Creatine deficiency syndrome†	SLC6A8	XL	General	1 in 3,434	98%	1 in 171,651	1 in 686,716
103	克果納傑氏症候群	Crigler-Najjar syndrome	UGT1A1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
104	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
105	胱胺酸血症	Cystinosis	CTNS	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
106	D-雙功能蛋白缺乏症	D-bifunctional protein deficiency	HSD17B4	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
107	Lowe氏症候群	Dent disease 2; Lowe syndrome†	OCRL	XL	General	1 in 250,000	95%	1 in 4,999,981	<1 in 10 million
108	二氫硫辛醯胺脫氫酶(E3)缺乏症	Dihydrolipoamide dehydrogenase deficiency	DLD	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
109	二氫嘧啶脫氫酶缺乏症	Dihydropyrimidine dehydrogenase deficiency	DPYD	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
110	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
111	先天性角化不全症-5型	Dyskeratosis congenita type 5	RTEL1	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
112	醣基化功能缺陷肌肉失養症-POMT1型	Dystroglycanopathy, POMT1-related	POMT1	AR	General	1 in 290	99%	1 in 28,901	<1 in 10 million
113	表皮分解性水皰症	Dystrophic epidermolysis bullosa	COL7A1	AR	General	1 in 196	97%	1 in 6,501	1 in 5,096,784
114	膠原蛋白髮育異常埃勒斯-當洛二氏症候群	Ehlers-Danlos syndrome, Dermatosparaxis type VIIC	ADAMTS2	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
115	埃勒斯-當洛二氏症候群伴隨脊柱後凸- PLOD1型	Ehlers-Danlos syndrome with kyphoscoliosis, PLOD1-related	PLOD1	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million
116	埃利偉氏症候群-EVC型	Ellis-van Creveld syndrome, EVC-related; Weyers acrofacial dysostosis, EVC-related	EVC	AR	General	1 in 142	98%	1 in 7,051	1 in 4,004,968
117	埃利偉氏症候群-EVC2型	Ellis-van Creveld syndrome, EVC2-related; Weyers acrodental dysostosis, EVC2-related	EVC2	AR	General	1 in 240	98%	1 in 11,951	<1 in 10 million
118	肌失養症	Emery-Dreifuss muscular dystrophy†	EMD	XL	General	1 in 81,967	99%	1 in 8,196,601	<1 in 10 million
119	網膜色素變性-37型	Enhanced S-cone syndrome; Retinitis pigmentosa 37	NR2E3	AR	General	1 in 209	98%	1 in 10,401	1 in 8,695,236
120	乙基丙二酸腦病變	Ethylmalonic encephalopathy	ETHE1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
121	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
122	第十一凝血因子缺乏症	Factor XI deficiency	F11	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
123	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
124	家族性胰島素過多症-ABCC8型	Familial hyperinsulinism, ABCC8-related	ABCC8	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
125	家族性地中海熱病	Familial Mediterranean fever	MEFV	AR	General	1 in 20	99%	1 in 1,901	1 in 152,080
126	Fanconi氏貧血A型	Fanconi anemia group A	FANCA	AR	General	1 in 239	98%	1 in 11,901	<1 in 10 million
127	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
128	Fanconi氏貧血G型	Fanconi anemia group G	FANCG	AR	General	1 in 632	90%	1 in 6,311	<1 in 10 million
129	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
130	延胡索酸缺乏症	Fumarase deficiency	FH	AR	General	<1 in 500	90%	<1 in 4,991	1 in 9,982,000
131	半乳糖激酶缺乏症	Galactokinase deficiency	GALK1	AR	General	1 in 110	95%	1 in 2,181	1 in 959,640
132	半乳糖異構酶缺乏症	Galactose epimerase deficiency	GALE	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million

孕知因 全球版-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
133	半乳糖血症	Galactosemia	GALT	AR	General	1 in 110	95%	1 in 2,181	1 in 959,640
134	高雪氏症	Gaucher disease	GBA	AR	General	1 in 77	99%	1 in 7,601	1 in 2,341,108
135	Gitelman氏症候群	Gitelman syndrome	SLC12A3	AR	General	1 in 100	98%	1 in 4,951	1 in 1,980,400
136	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
137	谷氨酸甲酰亞胺基轉移酶缺乏症	Glutamate formiminotransferase deficiency	FTCD	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
138	戊二酸尿症-IIA型	Glutaric aciduria IIA	ETFA	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
139	戊二酸尿症-IIB型	Glutaric aciduria IIB	ETFB	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
140	戊二酸尿症-IIC型	Glutaric aciduria IIC	ETFDH	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
					East Asian	1 in 74	98%	1 in 3,651	1 in 1,080,696
141	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
142	非酮性高甘胺酸血症-AMT型	Glycine encephalopathy, AMT-related	AMT	AR	General	1 in 373	98%	1 in 18,601	<1 in 10 million
143	非酮性高甘胺酸血症-GLDC型	Glycine encephalopathy, GLDC-related	GLDC	AR	General	1 in 193	98%	1 in 9,601	1 in 7,411,972
144	肝醣儲積症第四型	Glycogen storage disease IV	GBE1	AR	General	1 in 387	99%	1 in 38,601	<1 in 10 million
145	肝醣儲積症第三型	Glycogen storage disease type III	AGL	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
146	肝醣儲積症第五型	Glycogen storage disease type V	PYGM	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
147	肝醣儲積症第七型	Glycogen storage disease VII	PFKM	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
148	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
149	肝醣儲積症第1b型	Glycogen storage disease, type Ib	SLC37A4	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
150	胍基乙酸鹽甲基轉移酶缺乏症	Guanidinoacetate methyltransferase deficiency	GAMT	AR	General	1 in 371	99%	1 in 37,001	<1 in 10 million
151	鳥胺酸酮酸轉胺酶缺乏症	Gyrate atrophy of choroid and retina	OAT	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
152	哈納氏病	Hartnup disorder	SLC6A19	AR	General	1 in 87	99%	1 in 8,601	1 in 2,993,148
153	血鐵沉積症-2A型	Hemochromatosis, type 2A	HJV	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
154	血鐵沉積症-3型	Hemochromatosis, type 3	TFR2	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
155	血友病A型	Hemophilia A†	F8	XL	General	1 in 3,250	48%	1 in 6,249	1 in 25,000
156	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
157	肝腦病變型粒線體DNA耗竭症候群-MPV17型	Hepatocerebral mitochondrial DNA depletion syndrome, MPV17-related	MPV17	AR	General	<1 in 500	96%	<1 in 12,476	<1 in 10 million
158	遺傳性葉酸吸收不良	Hereditary folate malabsorption	SLC46A1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
					Puerto Rican	1 in 500	99%	1 in 49,901	<1 in 10 million
159	遺傳性果糖不耐症	Hereditary fructose intolerance	ALDOB	AR	General	1 in 122	99%	1 in 12,101	1 in 5,905,288
160	Hermansky-Pudlak症候群-1型	Hermansky-Pudlak syndrome 1	HPS1	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
161	Hermansky-Pudlak症候群-3型	Hermansky-Pudlak syndrome 3	HPS3	AR	General	1 in 354	98%	1 in 17,651	<1 in 10 million
162	多發性羧化酶缺乏症	Holocarboxylase synthetase deficiency	HLCS	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
163	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
164	高胱胺酸尿症-巨母紅血球性貧血-cobalamin E型	Homocystinuria-megaloblastic anemia, cobalamin E type	MTRR	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
165	Hydrolethalus症候群	Hydrolethalus syndrome	HYLS1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million

孕知因 全球版-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
166	XHIM syndrome	Hyper IgM syndrome, X-linked†	CD40LG	XL	General	1 in 50,000	98%	1 in 2,499,951	1 in 9,999,904
167	高免疫球蛋白血症D症候群; 甲羥戊酸激酶缺乏症	Hyperimmunoglobulinemia D syndrome; Mevalonate kinase deficiency	MVK	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
168	腺苷激酶缺乏引起高甲硫胺酸血症	Hypermethioninemia due to adenosine kinase deficiency	ADK	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
169	高甲硫胺酸伴隨S-腺苷半胱氨酸水解酶缺乏症	Hypermethioninemia due to deficiency of S-adenosylhomocysteine hydrolase	AHCY	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
170	高鳥氨酸-高血氨-高瓜胺酸綜合症候群(HHH症候群)	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome (Triple H syndrome)	SLC25A15	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
171	高脯胺酸血症第二型	Hyperprolinemia type II	ALDH4A1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
172	性腺功能低下症-GNRHR型	Hypogonadotropic hypogonadism, GNRHR-related	GNRHR	AR	General	1 in 347	99%	1 in 34,601	<1 in 10 million
173	汗性外胚層增生不良症	Hypohidrotic ectodermal dysplasia†	EDA	XL	General	1 in 14,167	99%	1 in 1,416,601	1 in 5,666,472
174	Treacher Collins氏症候群-POLR1C型	Hypomyelinating leukodystrophy, POLR1C-related ; Treacher Collins syndrome-POLR1C-related	POLR1C	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
175	低磷酸酯酶症	Hypophosphatasia	ALPL	AR	General	1 in 158	95%	1 in 3,141	1 in 1,985,112
176	包涵體肌肉病變	Inclusion body myopathy type 2 (Nonaka myopathy)	GNE	AR	General	<1 in 500	80%	<1 in 2,496	1 in 4,992,000
177	新生兒神經軸發育不良	Infantile neuroaxonal dystrophy	PLA2G6	AR	General	1 in 500	97%	1 in 16,634	<1 in 10 million
178	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
179	Joubert氏症候群-2型	Joubert syndrome 2; Meckel syndrome 2	TMEM216	AR	General	1 in 141	98%	1 in 7,001	1 in 3,948,564
180	Joubert氏症候群-28型	Joubert syndrome 28; Meckel syndrome 1; Bardet-Biedl syndrome 13	MKS1	AR	General	1 in 260	98%	1 in 12,951	<1 in 10 million
181	Joubert氏症候群-4型	Joubert syndrome 4; Senior-Løken syndrome 1; Nephronophthisis	NPHP1	AR	General	1 in 480	98%	1 in 23,951	<1 in 10 million
182	Joubert氏症候群-AHI-1型	Joubert syndrome, AHI-1 related	AHI1	AR	General	1 in 448	99%	1 in 44,701	<1 in 10 million
183	Joubert氏症候群-ARL13B型	Joubert syndrome, ARL13B- related	ARL13B	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
184	接合型表皮溶解水皰症-LAMA3型	Junctional epidermolysis bullosa, LAMA3-related; Laryngo-onycho-cutaneous syndrome	LAMA3	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
185	接合型表皮溶解水皰症-LAMB3型	Junctional epidermolysis bullosa, LAMB3-related	LAMB3	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
186	接合型表皮溶解水皰症-LAMC2型	Junctional epidermolysis bullosa, LAMC2-related	LAMC2	AR	General	1 in 781	98%	1 in 39,001	<1 in 10 million
187	性聯遺傳視網膜裂損症/視覺黃斑症	Juvenile retinoschisis, X-linked†	RS1	XL	General	1 in 2,500	96%	1 in 62,476	1 in 249,956
188	Krabbe氏症(球細胞腦白質失養症)	Krabbe disease	GALC	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
189	L1症候群	L1 syndrome†	L1CAM	XL	General	1 in 15,000	99%	1 in 1,499,901	1 in 5,999,804
190	萊伯氏先天性黑矇症-2型; 色素性視網膜炎-20型	Leber congenital amaurosis 2; Retinitis pigmentosa 20	RPE65	AR	General	1 in 228	98%	1 in 11,351	<1 in 10 million
191	萊伯氏先天性黑矇症-5型	Leber congenital amaurosis 5	LCA5	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
192	萊伯氏先天性黑矇症-8型; 色素性視網膜炎-12型	Leber congenital amaurosis 8; Retinitis pigmentosa 12	CRB1	AR	General	1 in 104	98%	1 in 5,151	1 in 2,142,816
193	萊伯氏先天性黑矇症-13型	Leber congenital amaurosis type 13	RDH12	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
194	Leigh症候群-SURF1型; 進行性神經性腓骨萎縮症-SURF1型	Leigh syndrome, SURF1-related ; Charcot-Marie-Tooth disease, SURF1-related	SURF1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
195	Leigh症候群-complex IV (COX)酶缺乏	Leigh syndrome with Complex IV deficiency	LRPPRC	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
196	致死先天性攣縮綜合症	Lethal congenital contracture syndrome 1	GLE1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million

孕知因 全球版-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
197	腦白質病伴隨白質消失症	Leukoencephalopathy with vanishing white matter	EIF2B5	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
198	肢帶型肌肉失養症-2A型	Limb-girdle muscular dystrophy type 2A	CAPN3	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
199	肢帶型肌肉失養症-2B型	Limb-girdle muscular dystrophy type 2B	DYSF	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
					Japanese	1 in 332	95%	1 in 6,621	1 in 8,792,688
200	肢帶型肌肉失養症-2C型	Limb-girdle muscular dystrophy, type 2C	SGCG	AR	General	1 in 381	98%	1 in 19,001	<1 in 10 million
201	肢帶型肌肉失養症-2D型	Limb-girdle muscular dystrophy, type 2D	SGCA	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
202	肢帶型肌肉失養症-2E型	Limb-girdle muscular dystrophy, type 2E	SGCB	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
203	肢帶型肌肉失養症-2F型	Limb-girdle muscular dystrophy, type 2F	SGCD	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
204	肢帶型肌肉失養症-2H型; Bardet-Biedl氏症候群-11型	Limb-girdle muscular dystrophy, type 2H; Bardet-Biedl syndrome 11	TRIM32	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
205	脂質先天性腎上腺發育不全	Lipoid congenital adrenal hyperplasia	STAR	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
206	性聯遺傳平腦症	Lissencephaly, X-linked†	DCX	XL	General	1 in 42,500	98%	1 in 2,124,951	1 in 8,499,904
207	急性新生兒肝衰竭	Liver failure, acute infantile	TRMU	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
208	三功能蛋白缺乏症及長鏈3-羥醯輔酶A脫氫酶缺乏症	Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency; Trifunctional protein deficiency	HADHA	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
209	Lujan-Fryns症候群-UPF3B型	Lujan-Fryns syndrome, UPF3B-related	UPF3B	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
210	Lujan-Fryns症候群-ZDHHC9型	Lujan-Fryns syndrome, ZDHHC9-related	ZDHHC9	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
211	Lysunyruc 蛋白質耐受不良症	Lysinuric protein intolerance	SLC7A7	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
					Japanese	1 in 119	95%	1 in 2,361	1 in 1,123,836
212	溶酶體酸性脂肪酶缺乏症	Lysosomal acid lipase deficiency	LIPA	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
213	黃斑角膜營養不良症-CHST6型	Macular corneal dystrophy, CHST6-related	CHST6	AR	General	1 in 79	99%	1 in 7,801	1 in 2,465,116
214	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
215	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
216	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
217	Meckel症候群; Joubert氏症候群	Meckel syndrome 5; Joubert syndrome 7; COACH syndrome	RPGRIP1L	AR	General	1 in 259	98%	1 in 12,901	<1 in 10 million
218	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
219	巨腦性腦白質病伴有皮層下囊腫第一型	Megalencephalic leukoencephalopathy with subcortical cysts	MLC1	AR	General	<1 in 500	97%	<1 in 16,634	<1 in 10 million
220	Menkes氏症候群	Menkes disease†	ATP7A	XL	General	1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
221	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
222	saposin-b缺乏引發異染性白質退化症	Metachromatic leukodystrophy due to saposin-b deficiency	PSAP	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
223	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
224	甲基丙二酸血症併高胱胺酸血症-cb1C型	Methylmalonic aciduria and homocystinuria, cblC type	MMACHC	AR	General	1 in 134	90%	1 in 1,331	1 in 713,416
225	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
226	甲基丙二酸血症併高胱胺酸血症-cblF型	Methylmalonic aciduria and homocystinuria, cblF type	LMBRD1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
227	甲基丙二酸血症併高胱胺酸血症-cb1J型	Methylmalonic aciduria and homocystinuria, cblJ type	ABCD4	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million

孕知因 全球版-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
228	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
229	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
230	甲基丙二酸單醯輔酶A異構酶缺乏症	Methylmalonyl-CoA epimerase deficiency	MCEE	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
231	小眼症	Microphthalmia, isolated 3	RAX	AR	General	1 in 289	99%	1 in 28,801	<1 in 10 million
232	小眼症	Microphthalmia with or without coloboma	VSX2	AR	General	1 in 91	98%	1 in 4,501	1 in 1,638,364
233	粒線體Complex I缺乏症(Leigh症候群)-NDUFAF5型	Mitochondrial complex I deficiency (Leigh syndrome), NDUFAF5-related	NDUFAF5	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
234	粒線體Complex I缺乏症(Leigh症候群)-NDUFAF6型	Mitochondrial complex I deficiency (Leigh syndrome), NDUFS6-related	NDUFS6	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
235	粒線體病變合併鐵粒幼紅細胞性貧血	Mitochondrial myopathy and sideroblastic anemia 1	PUS1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
236	MNGIE症候群粒線體性神經胃腸腦病變症候群	Mitochondrial neurogastrointestinal encephalopathy (MNGIE) disease	TYMP	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
237	黏脂症第二型和第三型-alpha/beta	Mucolipidosis III alpha/beta; Mucolipidosis II alpha/beta	GNPTAB	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
238	黏脂症第三型-gamma	Mucolipidosis III gamma	GNPTG	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
239	黏脂症第四型	Mucolipidosis IV	MCOLN1	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
240	黏多醣症-3A型(聖菲利柏氏症)	Mucopolysaccharidosis IIIA (Sanfilippo syndrome A)	SGSH	AR	General	1 in 454	98%	1 in 22,651	<1 in 10 million
241	黏多醣症-3D型(聖菲利柏氏症)	Mucopolysaccharidosis IIID (Sanfilippo syndrome D)	GNS	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
242	黏多醣症-4A型(莫奎歐氏症)	Mucopolysaccharidosis IVA (Morquio syndrome A)	GALNS	AR	General	1 in 224	97%	1 in 7,434	1 in 6,660,864
243	黏多醣症-2型(韓特氏症)	Mucopolysaccharidosis type II (Hunter syndrome)†	IDS	XL	General	1 in 50,000	91%	1 in 555,545	1 in 2,222,204
244	黏多醣症-3B型(聖菲利柏氏症)	Mucopolysaccharidosis type IIIB (Sanfilippo syndrome B)	NAGLU	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
					East Asian	1 in 298	99%	1 in 29,701	<1 in 10 million
245	黏多醣症-3C型(聖菲利柏氏症)	Mucopolysaccharidosis type IIIC (Sanfilippo syndrome C)	HGSNAT	AR	General	1 in 434	98%	1 in 21,651	<1 in 10 million
246	黏多醣症-4B型(莫奎歐氏症)	Mucopolysaccharidosis type IVB (Morquio syndrome B); GM1-gangliosidosis	GLB1	AR	General	1 in 134	99%	1 in 13,301	1 in 7,129,336
247	黏多醣症-9型	Mucopolysaccharidosis type IX	HYAL1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
248	黏多醣症-6型(馬洛托－拉米氏症)	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome)	ARSB	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
249	黏多醣症-7型	Mucopolysaccharidosis type VII	GUSB	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
250	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
251	多發性翼狀膜症候群	Multiple pterygium syndrome	CHRNG	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
252	多發性硫酸脂酶缺乏症	Multiple sulfatase deficiency	SUMF1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
253	肌肉失養症醣基化功能缺陷-FKRP型	Muscular dystrophy-dystroglycanopathy, FKRP-related	FKRP	AR	General	1 in 158	98%	1 in 7,851	1 in 4,961,832
254	肌肉失養症醣基化功能缺陷-FKTN型	Muscular dystrophy-dystroglycanopathy, FKTN-related; Fukuyama congenital muscular dystrophy	FKTN	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
					Japanese	1 in 82	99%	1 in 8,101	1 in 2,657,128
255	肌肉失養症醣基化功能缺陷-POMT2型	Muscular dystrophy-dystroglycanopathy, POMT2-related	POMT2	AR	General	1 in 371	99%	1 in 37,001	<1 in 10 million
256	肌肉失養症醣基化功能缺陷-網膜色素變性76型	Muscular dystrophy-dystroglycanopathy; Retinitis pigmentosa 76	POMGNT1	AR	General	1 in 462	98%	1 in 23,051	<1 in 10 million
257	肌肉失養症-LAMA2型	Muscular dystrophy, LAMA2-related	LAMA2	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
258	性聯遺傳肌小管病變	Myotubular myopathy, X-linked†	MTM1	XL	General	1 in 25,000	98%	1 in 1,249,951	1 in 4,999,904

孕知因 全球版-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
259	N-乙醯穀胺酸合成酶缺乏症	N-acetylglutamate synthase deficiency	NAGS	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
260	線狀體肌肉病變	Nemaline myopathy	NEB	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
261	腎性尿崩症	Nephrogenic diabetes insipidus	AQP2	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
262	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
263	神經元蠟樣脂褐質沉著疾病-CLN5型	Neuronal ceroid lipofuscinosis, CLN5-related	CLN5	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
264	神經元蠟樣脂褐質沉著疾病-CLN6型	Neuronal ceroid lipofuscinosis, CLN6-related	CLN6	AR	General	<1 in 500	92%	<1 in 6,239	<1 in 10 million
265	神經元蠟樣脂褐質沉著疾病-CLN8型	Neuronal ceroid lipofuscinosis, CLN8-related	CLN8	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
266	神經元蠟樣脂褐質沉著疾病-MFSD8型	Neuronal ceroid lipofuscinosis, MFSD8-related	MFSD8	AR	General	<1 in 500	95%	<1 in 9,981	<1 in 10 million
267	神經元蠟樣脂褐質沉著疾病-PPT1型	Neuronal ceroid lipofuscinosis, PPT1-related	PPT1	AR	General	1 in 368	98%	1 in 18,351	<1 in 10 million
268	神經元蠟樣脂褐質沉著疾病-TPP1型	Neuronal ceroid lipofuscinosis, TPP1-related	TPP1	AR	General	1 in 252	97%	1 in 8,368	1 in 8,434,944
269	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
270	尼曼匹克症-C1型	Niemann-Pick disease, type C1	NPC1	AR	General	1 in 194	90%	1 in 1,931	1 in 1,498,456
271	尼曼匹克症-C2型	Niemann-Pick disease, type C2	NPC2	AR	General	1 in 194	99%	1 in 19,301	<1 in 10 million
272	Nijmegen斷裂症候群	Nijmegen breakage syndrome	NBN	AR	General	1 in 158	99%	1 in 15,701	1 in 9,923,032
273	非症候群型遺傳性聽障-MYO7A型; 尤塞氏綜合症1B型	Non-syndromic hearing loss, MYO7A-related; Usher syndrome, type 1B	MYO7A	AR/Digenic	General	1 in 206	98%	1 in 10,251	1 in 8,446,824
					East Asian	1 in 62	98%	1 in 3,051	1 in 756,648
274	非症候群型遺傳性聽障-OTOF型	Nonsyndromic hearing loss, OTOF-related	OTOF	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
					Spanish	1 in 106	99%	1 in 10,501	1 in 4,452,424
275	非症候群型遺傳性聽障-PCDH15型; 尤塞氏綜合症1F型	Non-syndromic hearing loss, PCDH15-related; Usher syndrome, type 1F	PCDH15	AR/Digenic	General	1 in 395	98%	1 in 19,701	<1 in 10 million
276	非症候群型遺傳性聽障-USH1C型; 尤塞氏綜合症1C型	Non-syndromic hearing loss, USH1C-related; Usher syndrome, type IC	USH1C	AR/Digenic	General	1 in 353	90%	1 in 3,521	1 in 4,971,652
277	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
278	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
279	非症候群型遺傳性聽障-LOXHD1型	Nonsyndromic hearing loss, LOXHD1-related	LOXHD1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
280	諾里氏病	Norrie disease	NDP	XL	General	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
281	歐門氏症候群-RAG1型	Omenn syndrome, RAG1-related	RAG1	AR	General	1 in 137	98%	1 in 6,801	1 in 3,726,948
282	歐門氏症候群-RAG2型	Omenn syndrome, RAG2-related	RAG2	AR	General	1 in 137	98%	1 in 6,801	1 in 3,726,948
283	鳥胺酸氨甲醯基轉移酶缺乏症	Ornithine transcarbamylase deficiency†	OTC	XL	General	1 in 7,000	90%	1 in 69,991	1 in 279,984
284	成骨不全症第八型	Osteogenesis imperfecta, type VIII	P3H1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
					West African	1 in 67	99%	1 in 6,601	1 in 1,769,068
					African American	1 in 250	99%	1 in 24,901	<1 in 10,000,000
285	骨質石化症-TCIRG1型	Osteopetrosis, TCIRG1-related	TCIRG1	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
286	泛酸鹽激活酵素關聯之神經退化性疾病	Pantothenate kinase-associated neurodegeneration	PANK2	AR	General	1 in 289	99%	1 in 28,801	<1 in 10 million
287	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

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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
288	過氧化物酶醯基輔酶A氧化酶缺乏症	Peroxisomal acyl-CoA oxidase deficiency	ACOX1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
289	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
290	磷酸脫氫酶缺乏症	Phosphoglycerate dehydrogenase deficiency	PHGDH	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
291	磷酸甘油酸激酶1缺乏症	Phosphoglycerate kinase 1 deficiency	PGK1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
292	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
293	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
294	橋腦小腦發育不全-1A型	Pontocerebellar hypoplasia type 1A	VRK1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
295	橋腦小腦發育不全-1B型	Pontocerebellar hypoplasia type 1B	EXOSC3	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
296	橋腦小腦發育不全-6型	Pontocerebellar hypoplasia type 6	RARS2	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
297	橋腦小腦發育不全-2D型	Pontocerebellar hypoplasia, type 2D	SEPSECS	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
298	產後進展性小頭畸形伴隨癲癇及腦萎縮	Postnatal progressive microcephaly with seizures and brain atrophy	MED17	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
299	原發性纖毛運動障礙-DNAH5型	Primary ciliary dyskinesia, DNAH5-related	DNAH5	AR	General	1 in 142	98%	1 in 7,051	1 in 4,004,968
300	原發性纖毛運動障礙-DNAI1型	Primary ciliary dyskinesia, DNAI1-related	DNAI1	AR	General	1 in 230	98%	1 in 11,451	<1 in 10 million
301	原發性纖毛運動障礙-DNAI2型	Primary ciliary dyskinesia, DNAI2-related	DNAI2	AR	General	1 in 447	98%	1 in 22,301	<1 in 10 million
302	原發性纖毛運動障礙-DNAL1型	Primary ciliary dyskinesia, DNAL1-related	DNAL1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
303	原發性纖毛運動障礙-14型	Primary ciliary dyskinesia, type 14	CCDC39	AR	General	1 in 211	98%	1 in 10,501	1 in 8,862,844
304	原發性纖毛運動障礙-17型	Primary ciliary dyskinesia, type 17	CCDC103	AR	General	1 in 316	98%	1 in 15,751	<1 in 10 million
305	原發性纖毛運動障礙-30型	Primary ciliary dyskinesia, type 30	CCDC151	AR	General	1 in 365	98%	1 in 18,201	<1 in 10 million
306	原發先天性青光眼	Primary congenital glaucoma	CYP1B1	AR	General	1 in 50	99%	1 in 4,901	1 in 980,200
307	原發性高草酸鹽尿症-1型	Primary hyperoxaluria type 1	AGXT	AR	General	1 in 120	99%	1 in 11,901	1 in 5,712,480
308	原發性高草酸鹽尿症-2型	Primary Hyperoxaluria type II	GRHPR	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
309	原發性高草酸鹽尿症-3型	Primary hyperoxaluria type III	HOGA1	AR	General	1 in 184	99%	1 in 18,301	<1 in 10 million
310	漸進性外眼肌麻痺	Progressive external ophthalmoplegia; Alpers-Huttenlocher syndrome; Ataxia neuropathy spectrum; Myocerebrohepatopathy syndrome ; POLG-related disorder	POLG	AR	General	1 in 113	95%	1 in 2,241	1 in 1,012,932
311	進行性家族性肝內膽汁滯留症	Progressive Familial Intrahepatic Cholestasis	ABCB11	AR	General	1 in 112	98%	1 in 5,551	1 in 2,486,848
312	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
313	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
314	緻密性成骨不全症	Pycnodysostosis	CTSK	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
315	丙酮酸羧化酶缺乏症	Pyruvate carboxylase deficiency	PC	AR	General	1 in 250	95%	1 in 4,981	1 in 4,981,000
316	丙酮酸鹽脫氫酶E1-alpha缺乏症	Pyruvate dehydrogenase E1-alpha deficiency†	PDHA1	XL	General	<1 in 250,000	98%	1 in 12,499,951	<1 in 10 million
317	丙酮酸鹽脫氫酶E1-beta缺乏症	Pyruvate dehydrogenase E1-beta deficiency	PDHB	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
318	腎小管酸中毒伴隨耳聾	Renal tubular acidosis with deafness	ATP6V1B1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million

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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
319	Renpenning症候群	Renpenning syndrome	PQBP1	XL	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
320	視網膜色素變性-25型	Retinitis pigmentosa 25	EYS	AR	General	1 in 66	98%	1 in 3,251	1 in 858,264
321	視網膜色素變性-26型	Retinitis pigmentosa 26	CERKL	AR	General	1 in 148	98%	1 in 7,351	1 in 4,351,792
322	視網膜色素變性-28型	Retinitis pigmentosa 28	FAM161A	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
323	視網膜色素變性-59型	Retinitis pigmentosa 59	DHDDS	AR	General	1 in 296	98%	1 in 14,751	<1 in 10 million
324	網膜色素變性-CNGA1型	Retinitis pigmentosa, CNGA1-related	CNGA1	AR	General	1 in 210	99%	1 in 20,901	<1 in 10 million
325	網膜色素變性-CNGB1型	Retinitis pigmentosa, CNGB1-related	CNGB1	AR	General	1 in 296	99%	1 in 29,501	<1 in 10 million
326	網膜色素變性-IDH3B型	Retinitis pigmentosa, IDH3B-related	IDH3B	AR	General	1 in 296	99%	1 in 29,501	<1 in 10 million
327	網膜色素變性-PDE6A型	Retinitis pigmentosa, PDE6A-related	PDE6A	AR	General	1 in 133	99%	1 in 13,201	1 in 7,022,932
328	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
329	肢近端型點狀軟骨發育不良第三型	Rhizomelic chondrodysplasia punctata, type 3	AGPS	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
330	Roberts症候群	Roberts syndrome	ESCO2	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
331	Sandoff症 (成年型GM2神經節甘脂儲積症)	Sandhoff disease	HEXB	AR	General	1 in 600	98%	1 in 29,951	<1 in 10 million
332	Schimke免疫-骨發育不良	Schimke immunoosseous dysplasia	SMARCAL1	AR	General	1 in 500	90%	1 in 4,991	1 in 9,982,000
333	Schopf-Schulz-Passarge症候群	Schopf-Schulz-Passarge syndrome; Odontoonychodermal dysplasia	WNT10A	AR	General	<1 in 500	99%	<1 in 49,901	<1 in 10 million
334	Segawa症候群(瀨川氏病)	Segawa syndrome	TH	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296
335	嚴重複合型免疫缺乏症-JAK3型	Severe combined immunodeficiency, JAK3- related	JAK3	AR	General	1 in 299	99%	1 in 29,801	<1 in 10 million
336	嚴重複合型免疫缺乏症伴隨離子輻射	Severe combined immunodeficiency with sensitivity to ionizing radiation	DCLRE1C	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
337	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
338	嚴重先天性中性粒細胞減少症-HAX1型	Severe Congenital Neutropenia, HAX1-related	HAX1	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296
339	嚴重先天性中性粒細胞減少症-VPS45型	Severe congenital neutropenia, VPS45-related	VPS45	AR	General	1 in 224	98%	1 in 11,151	1 in 9,991,296
340	短支鏈醯輔酶A去氫酶缺乏症	Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency	ACADSB	AR	General	1 in 368	99%	1 in 36,701	<1 in 10 million
					Hmong	1 in 6	99%	1 in 501	<1 in 10 million
341	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
342	唾液酸儲存症	Sialic acid storage disorder	SLC17A5	AR	General	<1 in 500	91%	<1 in 5,545	<1 in 10 million
343	鐮刀型紅血球症; 乙型海洋性貧血; 血紅素C疾病	Sickle cell disease; Beta thalassemia; hemoglobin C disease	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
344	Sjögren-Larsson症候群	Sjögren-Larsson syndrome	ALDH3A2	AR	General	1 in 250	98%	1 in 12,451	<1 in 10 million
345	Smith-Lemli-Opitz症候群	Smith-Lemli-Opitz syndrome	DHCR7	AR	General	1 in 30	96%	1 in 726	1 in 87,120
346	痙攣性下身麻痺-15型	Spastic paraplegia 15	ZFYVE26	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
347	痙攣性下身麻痺-49型	Spastic paraplegia 49	TECPR2	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
348	痙攣性下身麻痺第七型	Spastic paraplegia type7	SPG7	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million

孕知因 全球版-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
349	SPG11型神經肌肉疾病	SPG11-related Neuromuscular Disorders	SPG11	AR	General	1 in 159	99%	1 in 15,801	<1 in 10 million
350	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
351	脊椎肋骨發育不全	Spondylocostal dysostosis	MESP2	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
352	鋼鐵症候群	Steel syndrome	COL27A1	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
353	Stuve-Wiedemann症候群	Stuve-Wiedemann syndrome	LIFR	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
354	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
355	戴薩克斯症	Tay-Sachs disease	HEXA	AR	General	1 in 300	99%	1 in 29,901	<1 in 10 million
356	四氫基喋呤缺乏症	Tetrahydrobiopterin deficiency	PTS	AR	General	1 in 354	96%	1 in 8,826	<1 in 10 million
357	四氫基喋呤缺乏症-PCBD1型	Tetrahydrobiopterin deficiency, PCBD1-related	PCBD1	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
358	四氫基喋呤缺乏症-QPPR型	Tetrahydrobiopterin deficiency, QDPR-related	QDPR	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
359	甲狀腺素合成不足-IYD型	Thyroid dyshormonogenesis, IYD-related	IYD	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
360	甲狀腺素合成異常-SLC5A5型	Thyroid dyshormonogenesis, SLC5A5-related	SLC5A5	AR	General	<1 in 500	99%	1 in 49,901	<1 in 10 million
361	甲狀腺素合成異常-TG型	Thyroid dyshormonogenesis, TG-related	TG	AR	General	1 in 241	99%	1 in 24,001	<1 in 10 million
362	甲狀腺素合成異常-TPO型	Thyroid dyshormonogenesis, TPO-related	TPO	AR	General	1 in 373	99%	1 in 37,201	<1 in 10 million
363	髮-肝-腸症候群	Trichohepatoenteric syndrome	TTC37	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
364	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
365	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
366	尤塞氏綜合症-1D型	Usher syndrome, type 1D	CDH23	AR/Digenic	General	1 in 285	90%	1 in 2,841	1 in 3,238,740
367	尤塞氏綜合症-1G型	Usher syndrome, type 1G	USH1G	AR	General	1 in 434	99%	1 in 43,301	<1 in 10 million
368	尤塞氏綜合症-2A型	Usher syndrome, type 2A	USH2A	AR	General	1 in 126	96%	1 in 3,126	1 in 1,575,504
369	尤塞氏綜合症-2D型	Usher syndrome, type 2D	WHRN	AR	General	1 in 282	99%	1 in 28,101	<1 in 10 million
370	尤塞氏綜合症-3A型	Usher syndrome, type 3A	CLRN1	AR	General	1 in 500	98%	1 in 24,951	<1 in 10 million
371	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
372	威爾森氏症	Wilson disease	ATP7B	AR	General	1 in 87	98%	1 in 4,301	1 in 1,496,748
373	Wiskott-Aldrich氏症候群; 性聯遺傳血小板缺乏症; 低嗜嚴重先天性中性球症-WAS型	Wiskott-Aldrich syndrome; Thrombocytopenia, X-linked; Severe Congenital Neutropenia, WAS-related†	WAS	XL	General	1 in 125,000	99%	1 in 12,499,901	<1 in 10 million
374	Wolcott-Rallison症候群	Wolcott-Rallison syndrome	EIF2AK3	AR	General	<1 in 500	98%	<1 in 24,951	<1 in 10 million
375	著色性乾皮症-A型	Xeroderma pigmentosum, group A	XPA	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million
					Japanese	1 in 74	99%	1 in 7,301	1 in 2,161,096
376	著色性乾皮症-C型	Xeroderma pigmentosum, group C	XPC	AR	General	1 in 500	99%	1 in 49,901	<1 in 10 million

孕知因 全球版-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
377	性聯遺傳性Aarskog Scott 症候群	X-linked Aarskog-Scott syndrome	FGD1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
378	性聯遺傳癲癇伴隨多樣性學習障礙	X-linked epilepsy with variable learning disabilites	SYN1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
379	性聯遺傳性聽障-POU3F4型	X-linked hearing loss, POU3F4-related	POU3F4	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
380	性聯遺傳智能障礙伴隨小腦發育不全及面部異常	X-linked intellectual disability with cerebellar hypoplasia and distinctive facial appearance	OPHN1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
381	性聯遺傳智能障礙-AP1S2型	X-linked intellectual disability, AP1S2-related	AP1S2	XL	General	<1 in 500	99%	1 in 4,999,901	<1 in 10 million
382	性聯遺傳智能障礙-ARX型	X-linked intellectual disability, ARX-related	ARX	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
383	性聯遺傳智能障礙-BRWD3型	X-linked intellectual disability, BRWD3-related	BRWD3	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
384	性聯遺傳智能障礙-CUL4B型	X-linked intellectual disability, CUL4B-related	CUL4B	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
385	性聯遺傳性智能障礙-DLG3型	X-linked intellectual disability, DLG3-related	DLG3	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
386	性聯遺傳智能障礙-FTSJ1型	X-linked intellectual disability, FTSJ1-related	FTSJ1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
387	性聯遺傳智能障礙-IL1RAPL1型	X-linked intellectual disability, IL1RAPL1- related	IL1RAPL1	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
388	性聯遺傳智能障礙-KDM5C型	X-linked intellectual disability, KDM5C-related	KDM5C	XL	General	<1 in 50,000	98%	1 in 2,499,951	<1 in 10 million
389	性聯遺傳智能障礙-PAK3型	X-linked intellectual disability, PAK3-related	PAK3	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
390	性聯遺傳智能障礙-Siderius型	X-linked intellectual disability, Siderius type	PHF8	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
391	性聯遺傳智能障礙-THOC2型	X-linked intellectual disability, THOC2-related	THOC2	XL	General	<1 in 50,000	99%	1 in 4,999,901	<1 in 10 million
392	性聯遺傳智能障礙-ZNF711型	X-linked intellectual disability, ZNF711-related	ZNF711	XL	General	<1 in 50,000	93%	1 in 714,272	1 in 2,857,143
393	性聯遺傳眼睛白化症第一型-GPR143型	X-linked Ocular albinism, GPR143-related	GPR143	XL	General	1 in 25,000	99%	1 in 2,499,901	<1 in 10 million
394	性聯遺傳性網膜色素變性-RP2型	X-linked Retinitis pigmentosa, RP2-related	RP2	XL	General	1 in 4,000	99%	1 in 399,901	1 in 1,600,000
395	性聯遺傳性網膜色素變性-RPGR型	X-linked Retinitis pigmentosa, RPGR-related	RPGR	XL	General	1 in 3,000	75%	1 in 11,997	1 in 48,000
396	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
397	柴爾維格氏症候群-PEX10型	Zellweger syndrome, PEX10-related	PEX10	AR	General	1 in 500	95%	1 in 9,981	<1 in 10 million
					Japanese	1 in 354	95%	1 in 7,061	1 in 9,998,376
398	柴爾維格氏症候群-PEX12型	Zellweger syndrome, PEX12-related	PEX12	AR	General	1 in 373	95%	1 in 7,441	<1 in 10 million
399	柴爾維格氏症候群-PEX2型	Zellweger syndrome, PEX2-related	PEX2	AR	General	1 in 500	95%	1 in 9,981	<1 in 10 million
400	柴爾維格氏症候群-PEX6型	Zellweger syndrome, PEX6-related	PEX6	AR	General	1 in 280	95%	1 in 5,581	1 in 6,250,720
	†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								