SOFIVA GENOMICS

疾病及基因列表
List of Genes & Disorders

慧智新生兒基因檢測
SOFIVA Baby Scan

序號	類別	v1.0	v2.0	v3.0
1	Drug Hypersensitivity Reaction 藥物過敏反應	●	●	●
2	Hearing Loss 聽損	●	●	●
3	Central Nervous System Disease 中樞神經系統疾病	●	●	●
4	Metabolic Disorders 代謝疾病		●	●
5	Blood Disorders 血液疾病		●	●
6	Multi Symptom Disorders 多症狀疾病		●	●
7	Immunodeficiency Disorders 免疫缺陷疾病			●
8	Pediatric Cancers 小兒癌症			●
9	Epilepsy 癲癇			●
10	Muscular Diseases 肌肉疾病			●
11	Vision Loss 視力喪失			●
12	Congenital Heart Defect 先天性心臟缺損			●

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慧智新生兒基因檢測
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序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
1	Aminoglycoside 氨基糖苷類抗生素	MTRNR1	Drug Hypersensitivity Reaction 藥物過敏反應	●	●	●
2	Sensorineural hearing loss 感覺神經性聽損	CMV GJB2 GJB3 OTOF SLC26A4	Hearing Loss 聽損	●	●	●
3	Congenital Central Hypoventilation Syndrome,CCHS 先天中樞性換氣不足症	PHOX2B	Central Nervous System Disease 中樞神經系統疾病	●	●	●
4	Allopurinol 安樂普利諾	HLA-B*5801	Drug Hypersensitivity Reaction 藥物過敏反應		●	●
5	Carbamazepine 卡馬西平	HLA-A*3101 HLA-B*1502			●	●
6	Dapsone 達普頌	HLA-B*13:01			●	●
7	Halothane 氟烷	CACNA1S RYR1			●	●
8	Isoflurane 異氟醚				●	●
9	Suxamethonium 琥珀膽鹼				●	●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
10	Warfarin 華法林	CYP2C9 VKORC1	Drug Hypersensitivity Reaction 藥物過敏反應		●	●
11	ABCD syndrome ABCD 症候群	EDNRB	Hearing Loss 聽損		●	●
12	Waardenburg syndrome type 4A 瓦登伯革氏症候群第 4A 型				●	●
13	Branchiootic syndrome type 1 Branchiootic 症候群第 1 型	EYA1			●	●
14	Branchiootorenal syndrome type 1 Branchiootorenal 症候群第 1 型				●	●
15	Branchiootic syndrome type 3 Branchiootic 症候群第 3 型	SIX1			●	●
2	Sensorineural hearing loss 感覺神經性聽損				●	●
16	Branchiootorenal syndrome type 2 Branchiootorenal 症候群第 2 型	SIX5			●	●
17	COMMAD syndrome COMMAD 症候群	MITF			●	●
18	Tietz syndrome Tietz 症候群				●	●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
19	Waardenburg syndrome type 2A 瓦登伯革氏症候群第 2A 型	MITF	Hearing Loss 聽損		●	●
20	Waardenburg syndrome/ocular albinism, digenic 雙基因型之瓦登伯革氏症候群/眼睛白化症				●	●
21	Craniofacial-deafness-hand syndrome 顱面聾手綜合症	PAX3			●	●
22	Waardenburg syndrome type 1 瓦登伯革氏症候群第 1 型				●	●
23	Waardenburg syndrome type 3 瓦登伯革氏症候群第 3 型				●	●
24	Jervell and Lange-Nielsen syndrome type 2 Jervell & Lange-Nielsen 氏症候群第 2 型	KCNE1			●	●
25	Oculodentodigital dysplasia 眼齒指發育不全	GJA1			●	●
26	PCWH syndrome PCWH 症候群	SOX10			●	●
27	Waardenburg syndrome type 2E 瓦登伯革氏症候群第 2E 型				●	●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
28	Waardenburg syndrome type 4C 瓦登伯革氏症候群第 4C 型	SOX10	Hearing Loss 聽損		●	●
2	Sensorineural hearing loss 感覺神經性聽損	AIFM1				
		DIAPH3				
		FOXI1				
		GJB4				
		GJB6			●	●
		MYO15A				
		PJVK				
		POU3F4				
		POU4F3				
2	Sensorineural hearing loss 感覺神經性聽損	KCNJ10			●	●
29	SeSAME syndrome SeSAME 症候群				●	●
30	Waardenburg syndrome type 2D 瓦登伯革氏症候群第 2D 型	SNAI2			●	●
31	Waardenburg syndrome type 4B 瓦登伯革氏症候群第 4B 型	EDN3			●	●
32	X-linked Charcot-Marie-Tooth disease 性聯遺傳進行性腓骨肌萎縮症	GJB1			●	●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
33	3-Hydroxy-3-methylglutaryl CoA lyase deficiency, HMG 3-羥基-3-甲基戊二酸尿症	HMGCL	Metabolic Disorders 代謝疾病		●	●
34	3-Methylcrotonyl-CoA carboxylase 1 deficiency 3-甲基巴豆醯輔酵素羧化酵素缺乏症第 1 型	MCCC1			●	●
35	3-Methylcrotonyl-CoA carboxylase 2 deficiency 3-甲基巴豆醯輔酵素羧化酵素缺乏症第 2 型	MCCC2			●	●
36	Anemia, nonspherocytic hemolytic, due to G6PD deficiency G6PD 缺乏症 (蠶豆症)	G6PD			●	●
37	Biotinidase deficiency, BD 生物素酶缺乏症	BTD			●	●
38	Carnitine palmitoyltransferase deficiency type 1, CPT I 肉鹼棕櫚醯基轉移酶缺乏症第 1 型	CPT1A			●	●
39	Carnitine palmitoyltransferase deficiency type 2, CPT II 肉鹼棕櫚醯基轉移酶缺乏症第 2 型	CPT2			●	●
40	Citrullinemia type 1, CIT I 瓜胺酸血症第 1 型	ASS1			●	●
41	Citrullinemia type 2, CIT II 瓜胺酸血症第 2 型	SLC25A13			●	●
42	Congenital adrenal hyperplasia due to 21 hydroxylase deficiency, CAH 21-羥化酶缺乏之先天性腎上腺增生症	CYP21A2			●	●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
43	Congenital nongoitrous hypothyroidism-1 先天性甲狀腺機能低下症第 1 型	TSHR	Metabolic Disorders 代謝疾病		●	●
44	Congenital nongoitrous hypothyroidism-2 先天性甲狀腺機能低下症第 2 型	PAX8			●	●
45	Congenital nongoitrous hypothyroidism-4 先天性甲狀腺機能低下症第 4 型	TSHB			●	●
46	Congenital nongoitrous hypothyroidism-5 先天性甲狀腺機能低下症第 5 型	NKX2-5			●	●
47	Congenital nongoitrous hypothyroidism-6 先天性甲狀腺機能低下症第 6 型	THRA			●	●
48	Dihydrolipoamide dehydrogenase deficiency 二氫硫辛醯胺脫氫酶缺乏症	DLD			●	●
49	Galactosemia, GAL 半乳糖血症	GALT			●	●
50	Glutaricaciduria type 1, GA I 戊二酸血症第 1 型	GCDH			●	●
51	Holocarboxylase synthetase deficiency, HCSD 全羧化酶合成酶缺乏	HLCS			●	●
52	Homocystinuria due to cystathionine beta-synthase deficiency, HCU 高胱胺酸尿症	CBS			●	●
53	Isovaleric academia, IVA 異戊酸血症	IVD			●	●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
54	Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency, LCHAD 長鏈 3-羥基醯基輔酶 A 脫氫酶缺乏症	HADHA	Metabolic Disorders 代謝疾病		●	●
55	Maple syrup urine disease, MSUD 楓糖尿症	BCKDHA				
		BCKDHB			●	●
		DBT				
56	Medium-chain acyl-CoA dehydrogenase deficiency, MCAD 中鏈醯輔酶A去氫酶缺乏症	ACADM			●	●
57	Methylmalonic academia, MMA 甲基丙二酸血症	MMAA				
		MMAB			●	●
		MUT				
58	Multiple acyl-CoA dehydrogenase deficiency, MADD 戊二酸血症第 2 型	ETFA				
		ETFB			●	●
		ETFDH				
59	Phenylketonuria, PKU 苯酮尿症	PAH			●	●
60	Primary carnitine deficiency, PCD 原發性肉鹼缺乏症	SLC22A5			●	●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
61	Propionic academia, PA 丙酸血症	PCCA	Metabolic Disorders 代謝疾病		●	●
		PCCB				
62	Thyroid dyshormonogenesis type 6 甲狀腺激素生成障礙第 6 型	DUOX2			●	●
63	Trifunctional protein deficiency, MTPD 三功能蛋白缺乏症	HADHB			●	●
64	Very long chain acyl-CoA dehydrogenase deficiency, VLCAD 極長鏈醯輔酶 A 去氫酶缺乏症	ACADVL			●	●
65	Beta-thalassemia 乙型海洋性貧血	HBB	Blood Disorders 血液疾病		●	●
66	Sickle cell anemia 鐮刀型貧血					
67	Familial transient neonatal hyperbilirubinemia 家族性暫時性新生兒高膽紅素血症	UGT1A1	Multi Symptom Disorders 多症狀疾病		●	●
2	Sensorineural hearing loss 感覺神經性聽損	CDH23	Hearing Loss 聽損			●
		TECTA				
		TMIE				
		TMPRSS3				
		TPRN				
		TRIOBP				

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
2	Sensorineural hearing loss 感覺神經性聽損	USH1C	Hearing Loss 聽損			●
68	Usher syndrome type 1C 尤塞氏綜合症第 1C 型					●
2	Sensorineural hearing loss 感覺神經性聽損	WHRN				●
69	Usher syndrome type 2D 尤塞氏綜合症第 2D 型					●
70	Usher syndrome type 1G 尤塞氏綜合症第 1G 型	USH1G				●
71	Usher syndrome type 2A 尤塞氏綜合症第 2A 型	USH2A				●
72	17-Beta hydroxysteroid dehydrogenase 3 deficiency 17-β-羥基類固醇脫氫酶缺乏症第 3 型	HSD17B3	Metabolic Disorders 代謝疾病			●
73	2-Methylbutyryl-CoA dehydrogenase deficiency 2-甲基丁醯輔酶 A 去氫酶缺乏症	ACADSB				●
74	3-Methylglutaconyl-CoA hydratase deficiency 3-甲基戊烯二醯輔酶A水和酶缺乏症	AUH				●
75	Adrenoleukodystrophy, ALD 腎上腺腦白質失養症	ABCD1				●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
76	Alpha-methylacetoacetic aciduria (Mitochondrial acetoacetyl-CoA thiolase deficiency) 粒線體乙醯輔酶 A 硫解酶缺乏症	ACAT1	Metabolic Disorders 代謝疾病			●
77	Argininemia, ARG 精胺酸血症	ARG1				●
78	Argininosuccinic aciduria 精胺丁二酸酵素缺乏症	ASL				●
79	Carbamoylphosphate synthetase 1 deficiency 氨甲醯磷酸合成酶缺乏症第 1 型	CPS1				●
80	Carnitine acylcarnitine translocase deficiency, CACTD 肉鹼穿透障礙	SLC25A20				●
81	Congenital adrenal hyperplasia due to 17-alpha-hydroxylase deficiency 17-α-羥化酶缺乏引起之先天性腎上腺增生	CYP17A1				●
82	Congenital adrenal hyperplasia due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency 3-β-羥基類固醇脫氫酶缺乏引起之先天性腎上腺增生第 2 型	HSD3B2				●
83	Congenital amegakaryocytic thrombocytopenia 先天巨核細胞缺乏血小板低下症	MPL				●
84	Ethylmalonic encephalopathy, EE 乙基丙二酸腦病變	ETHE1				●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
85	Fabry disease 法布瑞氏症	GLA	Metabolic Disorders 代謝疾病			●
86	Familial hypercholesterolemia 家族性高膽固醇血症	LDLR				●
87	Familial hyperinsulinism 家族性胰島素過多症	ABCC8				●
88	Neonatal diabetes 新生兒糖尿病					●
89	Familial lipoprotein lipase deficiency, LPL 家族性脂蛋白酵素功能不良症	LPL				●
90	Galactokinase deficiency 半乳糖激酶缺乏症	GALK1				●
91	Gaucher disease 高雪氏症	GBA				●
92	Glycine encephalopathy, GCE (Nonketotic Hyperglycemia, NKHG) 甘氨酸腦病(非酮性高甘胺酸血症)	AMT GCSH GLDC				●
93	Glycine N-methyltransferase deficiency 甘氨酸 N-甲基轉移酶缺乏症	GNMT				●
94	Glycogen storage disease type 1A, GSD1A 肝醣儲積症第 1 型	G6PC				●
95	Glycogen storage disease type 2, GSD II (Pompe Disease) 肝醣儲積症第 2 型 (龐貝氏症)	GAA				●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
96	Gyrate atrophy of choroid and retina, HOGA (Hyperornithinemia with gyrate atrophy) 高鳥胺酸血症伴隨脈絡膜及視網膜渦旋狀萎縮	OAT	Metabolic Disorders 代謝疾病			●
97	Hyperammonemia, Hyperornithinemia, Homocitrullinuria Syndrome, HHHS 高鳥氨酸-高血氨-高瓜胺酸綜合症候群	SLC25A15				●
98	Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency 高甲硫胺酸血症伴隨 S-腺苷-L-高半胺酸水解酵素缺乏症	AHCY				●
99	Hyperphenylalaninemia, BH4-deficient, Type A A 型四氫基喋呤缺乏之高苯丙胺酸血症	PTS				●
100	Hyperphenylalaninemia, BH4-deficient, Type B B 型四氫基喋呤缺乏之高苯丙胺酸血症	GCH1				●
101	Hyperphenylalaninemia, BH4-deficient, Type C C 型四氫基喋呤缺乏之高苯丙胺酸血症	QDPR				●
102	Hyperphenylalaninemia, BH4-deficient, Type D D 型四氫基喋呤缺乏之高苯丙胺酸血症	PCBD1				●
103	Hypophosphatasia 低磷酸酯酶症	ALPL				●
104	Isobutyryl-CoA dehydrogenase deficiency, IDBB 異丁醯輔酶 A 去氫酶缺乏症	ACAD8				●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
105	Malonyl-CoA decarboxylase deficiency 丙二醯輔酶 A 脫羧酶缺乏症	MLYCD	Metabolic Disorders 代謝疾病			●
106	Maternal vitamin B12 deficiency (Methylmalonic aciduria and homocystinuria cblC type) 甲基丙二酸血症缺乏維生素 B12	MMACHC				●
		PRDX1				
107	Menkes syndrome 緬克斯症候群	ATP7A				●
108	Methionine adenosyltransferase I/III deficiency 甲硫胺酸腺苷轉移酶 I/III 缺乏症	MAT1A				●
109	Mucopolysaccharidosis type 1, MPS I (Hurler syndrome) 黏多醣症第 1 型(賀勒氏症)	IDUA				●
110	Mucopolysaccharidosis type 2 (Hunter syndrome), MPS II 黏多醣症第 2 型(韓特氏症)	IDS				●
111	Mucopolysaccharidosis type 3C (Sanfilippo Syndrome), MPS 3C 黏多醣症第 3C 型(聖菲利柏氏症)	HGSNAT				●
112	Mucopolysaccharidosis type 6 (Maroteaux-Lamy syndrome), MPS VI 黏多醣症第 6 型(馬洛托-拉米氏症)	ARSB				●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
113	Niemann-Pick disease type A 尼曼匹克症 A 型	SMPD1	Metabolic Disorders 代謝疾病			●
114	Niemann-Pick disease type B 尼曼匹克症 B 型					●
115	Niemann-Pick disease type C1 尼曼匹克氏症 CI 型	NPC1				●
116	Ornithine transcarbamylase deficiency, OTC 鳥胺酸氨甲醯基轉移酶缺乏症	OTC				●
117	Permanent neonatal diabetes mellitus 永久性新生兒糖尿病	KCNJ11				●
118	Pyruvate carboxylase deficiency, PC 丙酮酸羧化酶缺乏症	PC				●
119	Short chain hydroxy acyl-CoA dehydrogenase deficiency, SCHAD 短鏈羥基醯基輔酶 A 脫氫酶缺乏症	HADH				●
120	Short-chain acyl-CoA dehydrogenase deficiency, SCAD 短鏈醯輔酶A去氫酶缺乏症	ACADS				●
121	Tyrosine hydroxylase deficiency (Segawa syndrome) 酪胺酸羥化酶缺乏症 (瀨川氏症)	TH				●
122	Tyrosinemia Type 1, TYRSN1 酪胺酸血症第 1 型	FAH				●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
123	Tyrosinemia Type 2, TYRSN2 酪胺酸血症第 2 型	TAT	Metabolic Disorders 代謝疾病			●
124	Tyrosinemia Type 3, TYRSN3 酪胺酸血症第 3 型	HPD				●
125	Vitamin D-dependent rickets 維生素 D 依賴型佝僂病	VDR				●
126	Wilson disease 威爾森氏症	ATP7B				●
127	Cyclic neutropenia 週期性嗜中性白血球減少症	ELANE	Blood Disorders 血液疾病			●
128	Severe congenital neutropenia (Neutropenia, severe congenital) 嚴重型先天性白血球減少症	HAX1				●
129	Hereditary hemorrhagic telangiectasia 遺傳性出血性之血管擴張症	ACVRL1 SMAD4				●
130	Hereditary spherocytosis type 1 遺傳性球形紅血球增多症第 1 型	ANK1				●
131	Hereditary spherocytosis type 2 遺傳性球形紅血球增多症第 2 型	SPTB				●
132	Hereditary spherocytosis type 3 遺傳性球形紅血球增多症第 3 型	SPTA1				●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
133	Hereditary spherocytosis type 4 遺傳性球形紅血球增多症第 4 型	SLC4A1	Blood Disorders 血液疾病			●
134	Hereditary spherocytosis type 5 遺傳性球形紅血球增多症第 5 型	EPB42				●
135	Achondroplasia 軟骨發育不全症	FGFR3	Multi Symptom Disorders 多症狀疾病			●
136	Hypochondroplasia 次軟骨發育不全症					●
137	Alagille syndrome 阿拉吉歐症	JAG1				●
138	Alport syndrome 亞伯氏症候群	COL4A3 COL4A4 COL4A5				●
139	Apert syndrome 愛伯特氏症	FGFR2				●
140	Congenital insensitivity to pain with anhidrosis 先天性痛不敏感症合併無汗症	NTRK1				●
141	Cystic fibrosis 囊腫纖維症	CFTR				●
142	Familial Mediterranean fever 家族性地中海熱	MEFV				●
143	Protein C deficiency 蛋白質 C 缺乏症	PROC				●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
144	Thrombophilia due to protein S deficiency 蛋白質 S 缺乏症	PROS1	Multi Symptom Disorders 多症狀疾病			●
145	Ataxia telangiectasia 共濟失調微血管擴張症候群	ATM	Immunodeficiency Disorders 免疫缺陷疾病			●
146	Bare lymphocyte syndrome type 2 裸淋巴症候群第 2 型	CIITA				●
147	Bruton's Agammaglobulinemia (X-linked hypogammaglobulinemia) 布魯頓氏低免疫球蛋白症	BTK				●
148	Cartilage-hair hypoplasia 軟骨毛髮發育不全	RMRP				●
149	Hyper IgE syndrome type 1 高免疫球蛋白 E 症候群第 1 型	STAT3				●
150	Nijmegen breakage syndrome 奈梅亨破損症候群	NBN				●
151	Severe combined immunodefciency, SCID 嚴重複合型免疫缺乏症	ADA				●
		IL2RG				
		IL7R				
		JAK3				
		PTPRC				
		ZAP70				

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
152	X-linked hyper IgM syndrome 性聯遺傳高免疫球蛋白 M 症候群	CD40LG	Immunodeficiency Disorders 免疫缺陷疾病			●
153	Fraiser syndrome Fraiser 症候群	WT1	Pediatric Cancers 小兒癌症			●
154	Li-Fraumeni syndrome 李-佛美尼症候群	TP53				●
155	Multiple endocrine neoplasia type 1 多發性內分泌腫瘤第 1 型	MEN1				●
156	Neurofibromatosis type 1 神經纖維瘤第 1 型	NF1				●
157	Nevoid basal cell carcinoma syndrome (Gorlin syndrome) 基底細胞痣癌症候群(戈林症候群)	PTCH1				●
158	Peutz-Jeghers syndrome 珀茨-傑格斯症候群(黑斑息肉症候群)	STK11				●
159	Retinoblastoma 視網膜母細胞瘤	RB1				●
160	Von Hippel-Lindau syndrome 逢希伯-林道症候群(視網膜小腦脊髓血管瘤症)	VHL				●
161	Benign familial infantile epilepsy 良性家族性嬰兒癲癇症	PRRT2	Epilepsy 癲癇			●
162	Familial paroxysmal kinesigenic dyskinesia 陣發性動作手足舞蹈徐動症					●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
161	Benign familial infantile epilepsy 良性家族性嬰兒癲癇症	SCN2A SCN8A KCNQ2 KCNQ3	Epilepsy 癲癇			●
163	Early infantile epileptic encephalopathy 早期嬰兒癲癇性腦病	KCNT1				●
164	Nocturnal frontal lobe epilepsy 夜間額葉癲癇					●
165	Glucose transport type 1 deficiency syndrome 腦血管屏障葡萄糖輸送缺陷	SLC2A1				●
166	Pyridoxine-dependent epilepsy 吡哆醇依賴性癲癇	ALDH7A1				●
167	Pyridoxamine 5'-phosphate oxidase deficiency 吡哆胺五端磷酸氧化酶缺乏	PNPO				●
168	Severe myoclonic epilepsy in infancy 嚴重嬰兒肌跳躍癲癇	SCN1A				●
169	Tuberous sclerosis complex 結節性硬化症	TSC1 TSC2				●
170	Becker Muscular Dystrophy, BMD 貝克氏肌肉萎縮症	DMD	Muscular Diseases 肌肉疾病			●
171	Duchenne muscular dystrophy, DMD 裘馨氏肌肉萎縮症					●

序號	疾病/藥物 Disorder/Medicine	基因 Gene	類別 Category	v1.0	v2.0	v3.0
172	Spinal muscular atrophy, SMA 脊髓性肌肉萎縮症	SMN1	Muscular Diseases 肌肉疾病			●
173	Aniridia 無虹膜症	PAX6	Vision Loss 視力喪失			●
174	Ocular albinism type 1 眼睛白化症第 1 型	GPR143				●
175	Oculocutaneous albinism type 1, OCA1 眼睛皮膚白化症第 1 型	TYR				●
176	Oculocutaneous albinism type 2, OCA2 眼睛皮膚白化症第 2 型	OCA2				●
177	Oculocutaneous albinism type 4, OCA4 眼睛皮膚白化症第 4 型	SLC45A2				●
178	Optic atrophy type 1 視神經萎縮症第 1 型	OPA1				●
179	Arrhythmogenic right ventricular dysplasia 心律失常性右心室發育不全	TMEM43	Congenital Heart Defect 先天性心臟缺損			●
180	Barth syndrome 巴氏症候群	TAZ				●
181	Danon disease 溶酶體儲積症	LAMP2				●
182	Heterotaxy 臟器異位症候群	ZIC3				●
183	Marfan syndrome 馬凡氏症候群	FBN1				●