Monogenic Screening
(170+ conditions)

Price： HK$8,800

screening for
173 inherited genetic disorders
across **160+** genes

- Suitable for pregnant women, couples planning pregnancy, or prospective parents
- Recommended for individuals or couples with a family history of genetic disorders
- Ideal for couples preparing for assisted reproductive treatments (ART / IVF)
- Recommended for consanguineous couples (close-relative marriage)
- Only a blood sample is required
  (Pregnant women only require 5 mL of peripheral blood)
- Report turnaround time: approximately 4–5 weeks
- High accuracy rate of over 99%
- Utilizes Next-Generation Sequencing (NGS) combined with conventional validation methods

What are Monogenic Genetic Disorders?

Monogenic genetic disorders are inherited conditions caused by mutations in a single gene. To date, **more than 10,000 types of monogenic disorders** have been identified worldwide. Many of these conditions **may lead to disability, severe health complications, or even death.**

Individuals carrying recessive genetic mutations often show no symptoms and routine health examinations usually cannot detect any abnormalities. As a result, the condition may only be discovered after a child is born with the disease.

Studies indicate that an **average person carries approximately 2.8 recessive disease-causing genes**. Although each individual genetic disorder may have a relatively low incidence, the overall combined risk of inherited genetic diseases exceeds 1%, which is higher than the incidence of Down syndrome **(approximately 1 in 700 in Hong Kong)**.

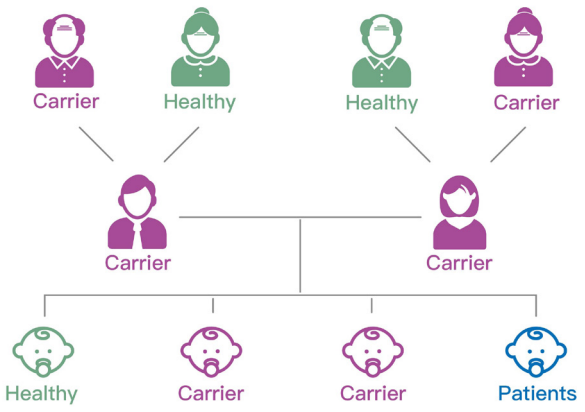
Because **carriers of recessive genetic conditions usually have no symptoms**, if **both parents carry the same recessive genetic mutation, there is a 75% chance that the mutation will be passed on to their child**. If **both partners are carriers of the same genetic disorder, there is a 25% risk that their child may develop the disease.**

Through **early genetic carrier screening, couples planning a pregnancy or expectant parents can identify potential genetic risks in advance, allowing them to make informed**

Test Implications

✓ Detect whether both spouses are carriers of a monogenic hereditary disease and provide information about reproductive risks.

✓ Combining genetic counseling with prenatal screening effectively reduces the occurrence of severe hereditary diseases.



A **POSITIVE** result means one or more mutations exist in the person. In this case, the test should be carried out on the other partner as well.

If both partners carry a mutation in the same gene, there is a high risk their child might have hereditary disorders. Genetic Counselling Services and gene therapy can be options if tested positive.

A **NEGATIVE** result indicates the person does not carry any of the mutation studied.

However, there is still a low possibility that the individual might carry other mutations, such as the De Novo.

Why Choose Health Gene Monogenic Disease Screening?

The Health Gene Monogenic Disease Screening (170+ conditions) can detect **173 inherited disorders** caused by more than **11,000 genetic mutations.**

Compared with testing for a single genetic condition, **the Health Gene Monogenic Disease Screening (170+ conditions)** provides a more comprehensive and cost-effective approach, enabling broader detection of potential inherited genetic risks.

Why Early Testing is Crucially Important?

For individuals with a family history of genetic diseases:

1. **Risk Assessment and Management:**
   For individuals with a family history of genetic diseases, undergoing "Monogenic Screening" helps in early detection of genetic disorders and assessing the risk of developing certain hereditary conditions. This enables them to make lifestyle adjustments as needed or undergo preventive screening.

2. **Family Planning:**
   For individuals planning to expand their families, understanding their genetic risk can influence their reproductive decisions and prompt them to consider other options, such as surrogacy or genetic assisted technologies.

For couples planning to have a baby:

1. **Genetic Risk Assistance:**
   For couples planning pregnancy, early testing can screen for any potential genetic disease risks, enabling them to make informed choices.

2. **Reproductive Decision Support:**
   The test results can provide couples with information about pregnancy risks and options, allowing them to jointly decide on the most suitable reproductive plan, thereby reducing the risk of potential hereditary diseases in the future.

This test includes a panel of common inherited genetic disorders, with a brief overview of each condition:			
1	Alpha-thalassemia	HBA1, HBA2	• Caused by mutations in the **HBA1 / HBA2 genes**. • A **hereditary hemoglobin disorder.** • Reduced α-globin production affects hemoglobin formation. • Leads to decreased oxygen-carrying capacity and anemia. • Mild cases may be asymptomatic. • Severe cases may present with serious complications during fetal development.
2	Beta-thalassemia	HBB	• Caused by mutations in the **HBB gene.** • **A hereditary hemoglobin disorder.** • Reduced β-globin production leads to anemia. Severity ranges from mild to severe. • Common symptoms include **pallor, fatigue, and delayed growth.** • Severe cases may require regular blood transfusions
3	Glucose-6-phosphate dehydrogenase deficiency, G6PD	G6PD	• Caused by mutations in the **G6PD gene (X-linked inheritance).** • A deficiency of an enzyme that protects red blood cells. • Most individuals are asymptomatic under normal conditions. • Certain triggers (e.g., medications, foods, infections) may cause **acute hemolysis.** • Symptoms include **jaundice, dark urine, fatigue, and anemia.** • Common in Hong Kong (affects ~4–5% of males)
4	Spinal Muscular Atrophy, SMA	SMN1	• Caused by mutations in the **SMN1 gene (autosomal recessive inheritance).** • A **hereditary neuromuscular disorder.** • Progressive degeneration of motor neurons leads to **muscle weakness and atrophy.** • **Delayed motor development** (e.g., sitting and walking). • Severe cases **may affect swallowing and respiratory function.** • Can present from infancy to adulthood.
5	Duchenne Muscular Dystrophy, DMD	DMD	• Caused by mutations in the **DMD gene (X-linked inheritance).** • **A severe and progressive muscle disorder** primarily affecting boys. • **Muscle degeneration leads to progressive weakness.** • Early signs include difficulty walking, frequent falls, and poor motor performance. • Later stages may affect cardiac and respiratory function.

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6	Fragile X Sydrome	FMR1	• Caused by mutations in the **FMR1 gene (X-linked inheritance).** • One of the most common inherited causes of **intellectual disability.** • Affects brain development. • Symptoms include **learning difficulties, delayed speech, and attention deficits.** • Some individuals may **exhibit autism spectrum features.** • Males are typically more severely affected.
7	Cystic Fibrosis	CFTR	• Caused by mutations in the **CFTR gene (autosomal recessive inheritance).** • A hereditary disorder affecting multiple organs. • Abnormal salt and water transport leads to thick, sticky secretions. • Primarily **affects the lungs, pancreas, and digestive system.** • Symptoms include **recurrent lung infections, chronic cough, and malabsorption.** • Requires long-term medical management.
8	Phenylketonuria, PKU	PAH	• Caused by mutations in the **PAH gene (autosomal recessive inheritance).** • A **metabolic disorder** affecting phenylalanine breakdown. • **Accumulation of phenylalanine can damage the brain.** • Newborns may initially appear normal. • Untreated cases may result in **intellectual impairment.** • Requires **strict dietary management**.
9	Polycystic Kidney Disease	PKHD1	• Caused by mutations in the **PKHD1 gene (autosomal recessive inheritance).** • A hereditary disorder **affecting the kidneys and liver.** • Characterized by multiple cysts in the kidneys. • May impair kidney function and lead to **kidney failure.** • Often associated with liver involvement. • Typically presents in infancy or childhood
10	Hemophilia B	F9	• Caused by mutations in the **F9 gene (X-linked inheritance).** • Deficiency of clotting factor IX leads to impaired blood clotting. Symptoms include **prolonged bleeding, easy bruising, and joint bleeding.** • Severe cases may experience **spontaneous bleeding.** • Primarily affects males.

This test includes a panel of common inherited genetic disorders, with a brief overview of each condition:			
11	Hereditary Hearing Loss and Deafness	GJB2 SLC26A4	• Commonly associated with mutations in the **GJB2 / SLC26A4 genes (autosomal recessive inheritance)**. • One of the most common causes of inherited hearing loss. • Affects **inner ear function, leading to sensorineural hearing loss**. • May present at birth or early childhood. • Severity ranges from mild to profound. • May **impact speech and language development.**
12	Maple Syrup Urine Disease, MSUD	BCKDHA, BCKDHB, DBT	• Caused by mutations in the **BCKDHA / BCKDHB / DBT genes (autosomal recessive inheritance).** • A metabolic disorder affecting the breakdown of branched-chain amino acids. • Toxic accumulation can occur in the body. • Symptoms in newborns include **feeding difficulties, lethargy, vomiting, and seizures**. • Urine may have a characteristic sweet (maple syrup) odor. • Severe cases **may lead to brain damage or death**. • Requires strict dietary control.
13	Glycogen Storage Disease (Pompe Disease	GAA	• Caused by mutations in the **GAA gene (autosomal recessive inheritance).** • A **lysosomal storage disorder.** • Glycogen accumulates in cells due to enzyme deficiency. • Primarily **affects muscles and the heart.** • Symptoms include **muscle weakness, developmental delay, and breathing difficulties.** • **Infantile form is typically more severe.**
14	Wilson Disease	ATP7B	• Caused by mutations in the **ATP7B gene (autosomal recessive inheritance).** • A **disorder of copper metabolism.** • **Copper accumulates** in organs such as the **liver, brain, and eyes.** • May **cause liver dysfunction, neurological symptoms, and psychiatric issues.** • Severe cases can lead to **organ damage if untreated**

173 Type Monogenic Diseases List			
	遺傳疾病	Diseases	Gene
1	威爾遜氏病 (肝豆狀核變性)	Wilson Disease	ATP7B
2	原發性肉碱缺乏症	Primary Carnitine Deficiency	SLC22A5
3	苯丙酮尿症	Phenylketonuria	PAH
4	四氫生物蝶呤缺乏症 A 型	Hyperphenylalaninemia, BH4-deficient, A	PTS
5	甲基丙二酸血症 Mut 型	MUT-Related Methylmalonic Acidemia	MMUT
6	甲基丙二酸血症 cblA 型	MMAA-Related Methylmalonic Acidemia	MMAA
7	甲基丙二酸血症 cblB 型	MMAB-Related Methylmalonic Acidemia	MMAB
8	甲基丙二酸血症伴同型半胱氨酸血症 cblC 型	Methylmalonic Aciduria and Homocystinuria, cblC Type	MMACHC
9	甲基丙二酸血症伴同型半胱氨酸血症 cblD 型	Methylmalonic Aciduria and Homocystinuria, cblD Type	MMADHC
10	同型半胱氨酸尿症伴巨幼細胞貧血 cblE 型	Homocystinuria-megaloblastic anemia, cblE Type	MTRR
11	高胱氨酸尿症伴巨幼紅細胞貧血 cblG 型	Homocystinuria-Megaloblastic Anemia cblG Type	MTR
12	甲基丙二酸單醯輔酶 A 差向異構酶缺乏症	MCEE-Related Methylmalonic Acidemia	MCEE
13	胱硫醚 β 合成酶缺乏性高胱氨酸尿症	Homocystinuria Due to Cystathionine Beta-Synthase Deficiency	CBS
14	戊二酸血症 I 型	Glutaric Acidemia I	GCDH
15	戊二酸血症 IIA 型	Glutaric Acidemia IIA	ETFA
16	戊二酸血症 IIB 型	Glutaric Acidemia IIB	ETFB
17	戊二酸血症 IIC 型	Glutaric Acidemia IIC	ETFDH
18	中鏈醯基輔酶 A 脫氫酶缺乏症	Acyl-CoA Dehydrogenase Deficiency, Medium-Chain	ACADM
19	短鏈醯基輔酶 A 脫氫酶缺乏症	Acyl-CoA Dehydrogenase Deficiency, Short-Chain	ACADS
20	極長鏈醯輔酶 A 去氫酶缺乏症	Acyl-CoA Dehydrogenase Deficiency, Very Long-Chain	ACADVL
21	3- 甲基巴豆醯輔酵素羧化酵素缺乏症第 1 型	3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	MCCC1
22	3- 甲基巴豆醯輔酵素羧化酵素缺乏症第 2 型	3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	MCCC2
23	瓜氨酸血症 1 型	Citrullinemia	ASS1
24	異戊酸血症	Isovaleric Acidemia	IVD
25	丙酸血症	Propionicacidemia	PCCA, PCCB
26	糖原累積病 Ia 型	Glycogen Storage Disease Type Ia	G6PC
27	糖原累積病 Ib 型	Glycogen Storage Disease Type Ib	SLC37A4

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28	糖原累積病 Ic 型	Glycogen Storage Disease Type Ic	SLC37A4
29	糖原累積病 II 型	Glycogen Storage Disease Type II	GAA
30	糖原累積病 IV 型	Glycogen Storage Disease Type IV	GBE1
31	Niemann-Pick 病 A 型	Niemann-Pick Disease Type A	SMPD1
32	Niemann-Pick 病 B 型	Niemann-Pick Disease Type B	SMPD1
33	Niemann-Pick 病 C1 型	Niemann-Pick Disease Type C1	NPC1
34	Niemann-Pick 病 C2 型	Niemann-Pick Disease Type C2	NPC2
35	楓糖尿病 1A 型	Maple Syrup Urine Disease Type 1A	BCKDHA
36	楓糖尿病 1B 型	Maple Syrup Urine Disease Type 1B	BCKDHB
37	楓糖尿病 2 型	Maple Syrup Urine Disease Type 2	DBT
38	楓糖尿病 3 型	Maple Syrup Urine Disease Type 3	DLD
39	黏多醣貯積症第一型	Hurler Syndrome	IDUA
40	賀勒 - 施艾氏症	Hurler-Scheie Syndrome	IDUA
41	黏多醣貯積症第 V 型	Mucopolysaccharidosis Type V	IDUA
42	黏多醣貯積症 II 型	Mucopolysaccharidosis II	IDS
43	黏多醣貯積症 IIIA 型	Mucopolysaccharidosis Type IIIA	SGSH
44	黏多醣貯積症 IIIB 型	Mucopolysaccharidosis Type IIIB	NAGLU
45	黏多醣貯積症 IIIC 型	Mucopolysaccharidosis Type IIIC	HGSNAT
46	黏多醣貯積症 IIID 型	Mucopolysaccharidosis Type IIID	GNS
47	黏多醣貯積症 IVA 型	Mucopolysaccharidosis Type IVA	GALNS
48	黏多醣貯積症 IVB 型	Mucopolysaccharidosis Type IVB	GLB1
49	黏多醣貯積症 VI 型	Mucopolysaccharidosis Type VI	ARSB
50	酪氨酸血症 1 型	Tyrosinemia Type 1	FAH
51	法布瑞氏症	Fabry Disease	GLA
52	生物素酶缺乏症	Biotinidase Deficiency	BTD
53	全羧化酶合成酶缺乏症	Holocarboxylase Synthetase Deficiency	HLCS
54	高鳥胺酸血症 - 高氨血症 - 高瓜胺酸血症症候群	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome	SLC25A15
55	氨甲酰磷酸合成酶 1 缺乏症	Carbamoylphosphate Synthetase I Deficiency	CPS1
56	鳥氨酸氨甲醯轉移酶缺乏症	Ornithine Transcarbamylase Deficiency	OTC
57	精氨酸琥珀酸尿症	Argininosuccinic Aciduria	ASL
58	甘氨酸腦病	Glycine Encephalopathy	AMT,GLDC
59	3- 羥基 -3- 甲基戊二醯輔酶 A 合成酶 2 缺乏症	3-Hydroxy-3-Methylglutaryl-CoA Synthase 2 Deficiency	HMGCS2
60	先天性糖基化病 1a 型	Congenital Disorders of Glycosylation Ia	PMM2

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61	過氧化物酶體生物合成障礙 1A 型	Peroxisome Biogenesis Disorder 1A (Zellweger)	PEX1
62	球細胞腦白質失養症	Krabbe Disease	GALC
63	家族性高胰島素血症 2 型	Familial Hyperinsulinemic Hypoglycemia 2	KCNJ11
64	家族性高胰島素血症 4 型	Familial Hyperinsulinemic Hypoglycemia 4	HADH
65	嬰兒型低磷酸酯酶症	Hypophosphatasia, infantile	ALPL
66	兒童型低磷酸酯酶症	Hypophosphatasia, childhood	ALPL
67	異染性腦白質營養不良	Metachromatic Leukodystrophy due to Arylsulfatase A	ARSA
68	半乳糖血症	Galactosemia	GALT
69	α- 甘露糖苷貯積症	Alpha-Mannosidosis	MAN2B1
70	β- 酮硫酶缺乏症	Beta-Ketothiolase Deficiency	ACAT1
71	腺苷脫氨酶缺乏症	Adenosine Deaminase Deficiency	ADA
72	穀固醇血症	Sitosterolemia	ABCG5, ABCG8
73	鉬輔因子缺乏症 A 型	Molybdenum Cofactor Deficiency A	MOCS1
74	遺傳性果糖不耐受症	Hereditary Fructose Intolerance	ALDOB
75	戴薩克斯症	Tay-Sachs Disease	HEXA
76	Smith-Lemli-Opitz 症候群	Smith-Lemli-Opitz syndrome	DHCR7
77	杜興氏肌肉萎縮症	Duchenne Muscular Dystrophy	DMD
78	脊髓性肌肉萎縮症	Spinal Muscular Atrophy	SMN1
79	茹貝爾症候群 2 型	Joubert Syndrome 2	TMEM216
80	茹貝爾症候群 3 型	Joubert Syndrome 3	AHI1
81	茹貝爾症候群 5 型	Joubert Syndrome 5	CEP290
82	茹貝爾症候群 6 型	Joubert Syndrome 6	TMEM67
83	茹貝爾症候群 9 型	Joubert Syndrome 9	CC2D2A
84	茹貝爾症候群 17 型	Joubert Syndrome 17	CPLANE1
85	X 連鎖中央核肌病	X-Linked Centronuclear Myopathy	MTM1
86	神經元蠟樣脂褐質沉積症 1 型	Neuronal Ceroid-Lipofuscinoses 1	PPT1
87	神經元蠟樣脂褐質沉積症 2 型	Neuronal Ceroid-Lipofuscinoses 2	TPP1
88	神經元蠟樣脂褐質沉積症 3 型	Neuronal Ceroid-Lipofuscinoses 3	CLN3
89	神經元蠟樣脂褐質沉積症 4A 型	Neuronal Ceroid-Lipofuscinoses 4A	CLN6
90	神經元蠟樣脂褐質沉積症 5 型	Neuronal Ceroid-Lipofuscinoses 5	CLN5
91	神經元蠟樣脂褐質沉積症 6 型	Neuronal Ceroid-Lipofuscinoses 6	CLN6
92	神經元蠟樣脂褐質沉積症 7 型	Neuronal Ceroid-Lipofuscinoses 7	MFSD8
93	肢帶型肌營養不良 2A 型	Limb-Girdle Muscular Dystrophy Type 2A	CAPN3
94	肢帶型肌營養不良 2B 型	Limb-Girdle Muscular Dystrophy Type 2B	DYSF

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	遺傳疾病	Diseases	Gene
95	肢帶型肌營養不良 2C 型	Limb-Girdle Muscular Dystrophy Type 2C	SGCG
96	肢帶型肌營養不良 2D 型	Limb-Girdle Muscular Dystrophy Type 2D	SGCA
97	巨腦性腦白質營養不良伴皮質下囊腫 1 型	Megalencephalic Leukoencephalopathy with Subcortical Cysts 1	MLC1
98	卡納萬病	Canavan Disease	ASPA
99	常染色體隱性骨硬化症 1 型	Autosomal Recessive Osteopetrosis 1	TCIRG1
100	眼睛皮膚白化症 1 型	Oculocutaneous Albinism Type 1	TYR
101	眼睛皮膚白化症 2 型	Oculocutaneous Albinism Type 2	OCA2
102	眼睛皮膚白化症 3 型	Oculocutaneous Albinism Type 3	TYRP1
103	眼睛皮膚白化症 4 型	Oculocutaneous Albinism Type 4	SLC45A2
104	眼睛皮膚白化症 6 型	Oculocutaneous Albinism Type 6	SLC24A5
105	眼睛皮膚白化症 7 型	Oculocutaneous Albinism Type 7	LRMDA
106	X 連鎖遺傳眼白化病	X-Linked Ocular Albinism	GPR143
107	Hermansky-Pudlak 綜合症 1 型	Hermansky-Pudlak Syndrome 1	HPS1
108	Hermansky-Pudlak 綜合症 3 型	Hermansky-Pudlak Syndrome 3	HPS3
109	常染色體隱性遺傳性魚鱗病 1 型	Autosomal Recessive Congenital Ichthyosis 1	TGM1
110	常染色體隱性遺傳性魚鱗病 4A 型	Autosomal Recessive Congenital Ichthyosis 4A	ABCA12
111	常染色體隱性遺傳性魚鱗病 4B 型	Autosomal Recessive Congenital Ichthyosis 4B	ABCA12
112	迂迴性線狀魚鱗病	Netherton Syndrome	SPINK5
113	Sjögren-Larsson 綜合症	Sjögren-Larsson Syndrome	ALDH3A2
114	接合性表皮溶解水皰症 (LAMA3 相關)	LAMA3-Related Junctional Epidermolysis Bullosa	LAMA3
115	接合性表皮溶解水皰症 (LAMB3 相關)	LAMB3-Related Junctional Epidermolysis Bullosa	LAMB3
116	接合性表皮溶解水皰症 (LAMC2 相關)	LAMC2-Related Junctional Epidermolysis Bullosa	LAMC2
117	非 Herlitz 型交界型大疱性表皮鬆解症	Non-Herlitz type Junctional Epidermolysis Bullosa	COL17A1
118	營養不良性大皰性表皮鬆解症 (COL7A1 相關)	Autosomal Recessive Epidermolysis Bullosa	COL7A1
119	凝血因子 IX 缺乏症 (乙型血友病)	Dystrophica	F9
120	甲型地中海貧血	Hemophilia B	HBA1,HBA2
121	β- 地中海貧血	Alpha-thalassemia	HBB
122	鐮狀細胞性貧血症	Beta-thalassemia	HBB
123	範可尼貧血互補群 A	Sickle Cell Anemia	FANCA
124	範可尼貧血互補群 C	Fanconi anemia, Complementation group A	FANCA

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	遺傳疾病	Diseases	Gene
125	範可尼貧血互補群 D2	Fanconi anemia, Complementation group D2	FANCD2
126	範可尼貧血互補群 G	Fanconi anemia, Complementation group G	FANCG
127	範可尼貧血互補群 I	Fanconi Anemia, Complementation Group I	FANCI
128	範可尼貧血互補群 2	Hemophagocytic lymphohistiocytosis, Familial, 2	PRF1
129	範可尼貧血互補群 3	Hemophagocytic lymphohistiocytosis, Familial, 3	UNC13D
130	範可尼貧血互補群 4	Hemophagocytic lymphohistiocytosis, Familial, 4	STX11
131	範可尼貧血互補群 5	Hemophagocytic lymphohistiocytosis, Familial, 5	STXBP2
132	Omenn 綜合症	Omenn Syndrome	RAG1, RAG2
133	常染色體隱性重型聯合免疫缺陷症，B 細胞陰性	Severe combined Immunodeficiency, B cell-negative	RAG1, RAG2
134	X 連鎖重症聯合免疫缺陷	X-Linked Severe Combined Immunodeficiency	IL2RG
135	X 連鎖先天性腎上腺發育不全	X-Linked Adrenal Hypoplasia Congenita	NR0B1
136	進行性家族性肝內膽汁滯留症 2 型	Progressive Familial Intrahepatic Cholestasis 2	ABCB11
137	進行性家族性肝內膽汁滯留症 3 型	Progressive Familial Intrahepatic Cholestasis 3	ABCB4
138	進行性家族性肝內膽汁滯留症 4 型	Progressive Familial Intrahepatic Cholestasis 4	TJP2
139	亞伯氏綜合症 (COL4A3 相關)	Alport Syndrome 2, Autosomal Recessive	COL4A3, COL4A4
140	腎病綜合症 (NPHS1 相關)	Nephrotic Syndrome, Type 1	NPHS1
141	腎單位腎癆 3 型	Nephronophthisis 3	NPHP3
142	腎單位腎癆 11 型	Nephronophthisis 11	TMEM67
143	腎病型胱氨酸症	Nephropathic Cystinosis	CTNS
144	囊性纖維化	Cystic Fibrosis	CFTR
145	常染色體隱性耳聾 1A 型	Autosomal Recessive Deafness 1A	GJB2
146	常染色體隱性耳聾 4 型	Autosomal Recessive Deafness 4, with Enlarged Vestibular Aqueduct	SLC26A4
147	沃夫然症候群	Wolfram Syndrome 1	WFS1
148	Ellis-van Creveld 綜合症	Ellis-van Creveld Syndrome	EVC,EVC2
149	骨質疏鬆症 - 假神經膠質瘤綜合症	Osteoporosis-pseudoglioma Syndrome	LRP5
150	免疫缺陷 - 著絲粒不穩定 - 面部異常綜合症 1 型	Immunodeficiency-centromeric Instability-facial Anomalies Syndrome 1	DNMT3B

173 Type Monogenic Diseases List			
	遺傳疾病	Diseases	Gene
151	Meckel 綜合症 2 型	Meckel Syndrome 2	TMEM216
152	Meckel 綜合症 3 型	Meckel Syndrome 3	TMEM67
153	Meckel 綜合症 4 型	Meckel Syndrome 4	CEP290
154	X 連鎖少汗性外胚層發育不良	X-Linked Hypohidrotic Ectodermal Dysplasia	EDA
155	COACH 綜合症	COACH Syndrome	TMEM67, CC2D2A
156	杆狀體肌病 2 型	Nemaline Myopathy 2	NEB
157	天冬氨醯葡糖胺尿症	Aspartylglucosaminuria	AGA
158	多囊性腎病變 (PKHD1 相關)	Polycystic Kidney Disease	PKHD1
159	家族性自律神經失調症候群	Familial Dysautonomia	ELP1
160	酪胺酸羥化酶缺乏症	Tyrosine Hydroxylase Deficiency	TH
161	共濟失調性毛細血管擴張症	Ataxia-telangiectasia	ATM
162	α1 抗胰蛋白酶缺乏症	Alpha-1 Antitrypsin Deficiency	SERPINA1
163	遺傳性痙攣癱瘓 11 型	Spastic Paraplegia 11, Autosomal Recessive	SPG11
164	布盧姆綜合症	Bloom Syndrome	BLM
165	家族性地中海熱	Familial Mediterranean Fever	MEFV
166	吉特曼症候群	Gitelman Syndrome	SLC12A3
167	半乳糖激酶缺乏症	Galactokinase Deficiency	GALK1
168	粘脂質貯積症 4 型	Mucolipidosis IV	MCOLN1
169	葡萄糖六磷酸去氫酵素缺乏症（蠶豆症)	Glucose-6-phosphate dehydrogenase deficiency	G6PD
170	精氨酸酶缺乏症	Argininemia	ARG1
171	長鏈 3- 羥醯輔酶 A 脫氫酶缺乏症	Long-Chain 3-Hydroxyacyl-Coa Dehydrogenase Deficiency	HADHA
172	三功能蛋白缺乏症	Trifunctional Protein Deficiency	HADHA, HADHB
173	脆性 X 綜合症	Fragile X Sydrome	FMR1