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| 先天性魚鱗癬 4A 型 | Ichthyosis, congenital, autosomal recessive 4A |
| 先天性魚鱗癬 4B 型 | Ichthyosis, congenital, autosomal recessive 4B (harlequin) |
| 肺表面活性劑功能障礙 3 型 | Surfactant metabolism dysfunction, pulmonary, 3 |
| 進行性家族性膽汁鬱積2 | Cholestasis, progressive familial 2 |
| 進行性家族性肝內膽汁滯留症3型 | Progressive familial intrahepatic cholestasis type 3 |
| 家族性胰島素過度分泌低血糖症1 | Hyperinsulinemic hypoglycemia, familial, 1 |
| 永久性新生兒糖尿病3型 | Diabetes, permanent neonatal 3 |
| 腎上腺腦白質營養不良 | Adrenoleukodystrophy |
| 甲基丙二酸尿症症伴高胱胺酸尿症 cblJ 型 | Methylmalonic aciduria and homocystinuria, cblJ type |
| 谷固醇血症 2 型 | Sitosterolemia 2 |
| 谷固醇血症 1 型 | Sitosterolemia 1 |
| 異丁醯輔酶 A 去氫酶缺乏症 | Isobutyryl-CoA dehydrogenase deficiency |
| 醯基輔酶A去氫酵素缺乏症 中鏈 | Acyl-coa dehydrogenase, medium-chain, deficiency of |
| 醯基輔酶A去氫酵素缺乏症 短鏈 | Acyl-CoA dehydrogenase, short-chain, deficiency of |
| 2-甲基丁醯輔酶 A 去氫酶缺乏症 | 2-methylbutyrylglycinuria |
| 醯基輔酶A去氫酵素缺乏症 超長鏈 | Acyl-CoA dehydrogenase, very long-chain, deficiency of |
| 脊椎幹骺端發育不良 | Spondyloepimetaphyseal dysplasia, aggrecan type |
| α - 甲基乙醯乙酸尿症 | Alpha-methylacetoacetic aciduria |
| 腺苷脫氨酶缺乏/部分缺乏引起的嚴重聯合免疫缺陷病 | Severe combined immunodeficiency, due to adenosine deaminase deficiency / partial deficiency |
| 埃勒斯-當洛二氏綜合症第VIIC型 | Ehlers-Danlos syndrome, type VIIC |
| 腺苷激酶缺乏性高甲硫氨酸血症 | Hypermethioninemia due to adenosine kinase deficiency |
| 天冬氨酰葡萄糖胺尿症 | Aspartylglucosaminuria |
| 肝醣儲積症第IIIa, IIIb型 | Glycogen storage disease IIIa, IIIb |
| 肢近端型點狀軟骨發育不良第3型 | Rhizomelic chondrodysplasia punctata type 3 |
| 原發性高草酸尿症第I型 | Hyperoxaluria, primary, type I |
| 高甲硫胺酸血症併S-腺苷高半胱氨酸水解酶缺乏症 | Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency |
| Joubert 綜合症 3 型 | Joubert syndrome 3 |
| 自體免疫性內分泌綜合症I型, 有/無可逆性幹骺端發育異常 | Autoimmune polyendocrine syndrome, type I, with or without reversible metaphyseal dysplasia |
| 舍格倫—拿臣綜合症 | Sjogren-Larsson syndrome |
| 果糖不耐受症 | Fructose intolerance |
| 先天性糖基化障礙Ik型 | Congenital disorder of glycosylation type Ik |
| 先天性糖基化障礙 Ig 型 | Congenital disorder of glycosylation, type Ig |
| 先天性糖基化障礙 Id 型 | Congenital disorder of glycosylation, type Id |
| 先天性糖基化障礙 IC型 | Congenital disorder of glycosylation, type Ic |
| 先天性糖基化障礙 II 型 | Congenital disorder of glycosylation, type II |
| Alstrom氏綜合症 | Alstrom syndrome |
| 先天性魚鱗癬 2 型 | Ichthyosis, congenital, autosomal recessive 2 |
| 先天性魚鱗癬 3 型 | Ichthyosis, congenital, autosomal recessive 3 |
| 兒童低磷酸酶血症 | Hypophosphatasia, childhood |
| 嬰兒低磷酸酶血症 | Hypophosphatasia, infantile |
| 成人低磷酸酶血症 | Hypophosphatasia, adult |
| 高甘胺酸血症, 腦病變 2 | Hyperglycinemia encephalopathy 2 |
| 小腦萎縮症 | Spinocerebellar ataxia |
| 肢帶型肌肉萎縮症第2L型 | Muscular dystrophy, limb-girdle, type 2L |
| 卡爾曼症候群 1 型 | Kallmann syndrome 1 |
| 赫曼斯基-普德拉克氏綜合症2 | Hermansky-Pudlak syndrome 2 |
| 腎源性尿崩症2 | Nephrogenic diabetes insipidus 2 |
| 雄激素不敏感綜合症 | Androgen insensitivity syndrome |
| 部份雄激素不敏感症 | Androgen insensitivity, partial |
| 精胺酸血症 | Argininemia |

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| 朱伯特綜合症8 | Joubert syndrome 8 |
| 異染性腦白質退化症 | Metachromatic leukodystrophy |
| 黏多醣症VI型 | Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome) |
| X-連鎖點狀軟骨發育異常1型 | X-linked chondrodysplasia punctata type 1 |
| 早期嬰兒型癲癇性腦病變 1 型 | Epileptic encephalopathy, early infantile, 1 |
| 精胺丁二酸酵素缺乏症 | Argininosuccinic aciduria |
| 海綿狀腦白質營養不良症 | Canavan disease |
| 瓜氨酸血症 | Citrullinemia |
| 共濟失調微血管擴張症 | Ataxia-telangiectasia |
| 腎小管性酸中毒併耳聾 | Renal tubular acidosis with deafness |
| 緬克斯症 | Menkes disease |
| 威爾森氏症 | Wilson disease |
| 進行性家族性肝內膽汁滯留症1型 | Cholestasis, progressive familial intrahepatic 1 |
| 澤克爾氏綜合症1 | Seckel syndrome 1 |
| X連鎖智力障礙-臉部肌張力失調症候群 | Mental retardation-hypotonic facies syndrome, X-linked |
| 1型3-甲基戊二酸尿症 | 3-methylglutaconic aciduria, type I |
| 腎源性尿崩症1 | Nephrogenic Urogenital Disease 1 |
| 埃勒斯-當洛二氏綜合症-脊椎發育不全2 型 | Ehlers-Danlos syndrome, spondylodysplastic type, 2 |
| 先天性糖基化障礙IId型 | Congenital disorder of glycosylation, type IId |
| 埃勒斯-當洛二氏綜合症-脊椎發育不全1型 | Ehlers-Danlos syndrome, spondylodysplastic type, 1 |
| 巴德-畢德氏綜合症, 1 | Bardet-Biedl syndrome, 1 |
| 巴德-畢德氏綜合症, 10 | Bardet-Biedl syndrome, 10 |
| 巴德-畢德氏綜合症, 12 | Bardet-Biedl syndrome 12 |
| 巴德-畢德氏綜合症, 2 | Bardet-Biedl syndrome, 2 |
| 視網膜色素病變74 | Retinitis pigmentosa 74 |
| 楓糖尿症Ia型 | Maple syrup urine disease, type Ia |
| 楓糖尿症Ib型 | Maple syrup urine disease, type Ib |
| 畢卓斯坦綜合症 | Bjornstad syndrome |
| GRACILE綜合症 | GRACILE syndrome |
| 線粒體複合體III缺乏症，核型1 | Mitochondrial complex III deficiency, nuclear type 1 |
| 布隆氏綜合症 | Bloom syndrome |
| 肢端及中部骨骺發育不良Demirhan 型 | Acromesomelic dysplasia, Demirhan type |
| 巴特綜合症4A型 | Sensorineural deafness with renal dysfunction (Bartter syndrome type 4a) |
| 生物素酶缺乏症 | Biotinidase deficiency |
| X-連鎖無　　γ球蛋白血症 | X-linked agammaglobulinemia |
| 單一生長激素缺乏症 III 型伴隨無　γ球蛋白血症 | Growth hormone deficiency, isolated, type III, with agammaglobulinemia |
| 肢帶型肌肉萎縮症第2A型 | Muscular dystrophy, limb-girdle, type 2A |
| 同型半胱氨酸尿症/高同型半胱氨酸血栓形成 | Homocystinuria / hyperhomocysteinemic thrombosis |
| COACH綜合症2 | COACH syndrome 2 |
| 朱伯特綜合症9 | Joubert syndrome 9 |
| 梅克爾綜合症6 | Meckel syndrome 6 |
| 原發性纖毛運動障礙14 | Primary Ciliary Dyskinesia 14 |
| 原發性纖毛運動障礙15 | Primary Ciliary Dyskinesia 15 |
| 先天性腦積水1 | Congenital hydrocephalus 1 |
| X-連鎖IgM過高免疫缺陷病 | X-linked hyper-IgM immunodeficiency |
| Meier-Gorlin綜合症7 | Meier-Gorlin syndrome 7 |
| 尤塞氏綜合症ID型 | Usher syndrome, type ID |
| Meier-Gorlin綜合症4 | Meier-Gorlin syndrome 4 |
| 澤克爾氏綜合症5 | Seckel syndrome 5 |
| 巴德-畢德氏綜合症, 14 | Bardet-Biedl syndrome 14 |

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| 朱伯特綜合症5 | Joubert syndrome 5 |
| 梅克爾綜合症4 | Meckel syndrome 4 |
| 賴博氏先天性黑矇症10 | Leber congenital amaurosis 10 |
| 囊性纖維化 | Cystic fibrosis |
| 突觸前先天性重症肌無力綜合症6 | Congenital myasthenic syndrome, 6, presynaptic |
| 先天性重症肌無力綜合症4A | Congenital myasthenic syndrome, 4A |
| 先天性重症肌無力綜合症4B | Congenital myasthenic syndrome, 4B |
| 先天性重症肌無力綜合症4C | Congenital myasthenic syndrome, 4C |
| 失天性肌強直 | Myotonia congenita |
| Dent 病 1 型 | Dent disease 1 |
| 低磷性佝僂病 | Hypophosphatemic rickets |
| 巴特綜合症3型 | Bartter syndrome, type 3 |
| 神經元蠟樣脂褐質儲積症3 | Ceroid lipofuscinosis, neuronal 3 |
| 神經元蠟樣脂褐質儲積症5 | Ceroid lipofuscinosis, neuronal 5 |
| 神經元蠟樣脂褐質儲積症6A | Ceroid lipofuscinosis, neuronal 6A |
| 成人型蠟樣脂褐質儲積症6B | Ceroid lipofuscinosis type 6B, Kufs |
| 神經元蠟樣脂褐質儲積症8 | Ceroid lipofuscinosis, neuronal, 8 |
| 神經元蠟樣脂褐質儲積症8, 北方癲癇型 | Ceroid lipofuscinosis, neuronal, 8, northern epilepsy variant |
| 7型3-甲基戊二酸尿症伴隨白內障、神經病變及嗜中性白血球減少 | 3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia |
| 尤塞氏綜合症IIIa型 | Usher syndrome, type IIIa |
| 全色盲3 | Achromatopsia 3 |
| 先天性糖基化障礙IIg型 | Congenital disorder of glycosylation, type IIg |
| 先天性糖基化障礙IIe型 | Congenital disorder of glycosylation, type IIe |
| 非Herlitz型交界性水皰性表皮鬆解症4 | Epidermolysis bullosa, junctional, 4, non-Herlitz type |
| 心臟瓣膜型埃勒斯-當洛二氏綜合症 | Ehlers-Danlos syndrome, cardiac valvular type |
| 阿爾波特綜合症3A,3B,隱性遺傳 | Alport syndrome3A,3B, recessive |
| 阿爾波特綜合症2,隱性遺傳 | Alport syndrome 2, recessive |
| 隱性遺傳失養性水皰性表皮鬆解症 | Epidermolysis bullosa dystrophica, recessive |
| 先天性重症肌無力綜合症 5 | Myasthenic syndrome, congenital, 5 |
| Joubert 綜合症 17 型 | Joubert syndrome 17 |
| 口面指 綜合症6型 | Orofaciodigital syndrome VI |
| 氨甲酰磷酸合成酶I缺乏症 | Carbamoylphosphate synthetase I deficiency |
| 肝臟肉鹼棕櫚醯基轉移酶缺乏症IA型 | Hepatic carnitine palmitoyltransferase deficiency, type IA |
| 肉鹼棕櫚醯基轉移酶II缺乏症 | Carnitine palmitoyltransferase II deficiency |
| 肉鹼棕櫚醯基轉移酶II缺乏症, 新生兒致命型 | Carnitine palmitoyltransferase II deficiency, lethal neonaltal |
| 嬰兒肉鹼棕櫚醯基轉移酶II缺乏症 | Carnitine palmitoyltransferase II deficiency, infantile |
| 遲發性青少年腎病性胱氨酸症 | Cystinosis, late-onset juvenile or adolescent nephropathic type |
| 腎病性胱氨酸症 | Cystinosis, nephropathic |
| 非腎病性胱氨酸症 | Cystinosis, non-nephropathic |
| 神經元蠟樣脂褐質儲積症10 | Neuronal ceroid lipofuscinosis type 10 |
| 慢性肉芽腫病 4 | Chronic granulomatous disease 4 |
| 慢性肉芽腫病 | Granulomatous disease, chronic |
| 先天性腎上腺發育不全 | Congenital adrenal insufficiency |
| 先天性腎上腺皮質增生症, 類固醇11-β-羥化酶缺乏 | Adrenal hyperplasia, congenital, due to steroid 11-beta-hydroxylase deficiency |
| 皮質酮甲基氧化酶 I 缺乏先天性低醛固酮症 | Hypoaldosteronism, congenital, due to cmo I deficiency |
| 皮質酮甲基氧化酶 II 缺乏先天性低醛固酮症 | Hypoaldosteronism, congenital, due to cmo II deficiency |
| 17,20-裂解酶缺乏症 | 17,20-lyase deficiency |
| 青少年及成人先天性原發性開角青光眼 3A 型 | Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset |
| 先天性腎上腺皮質增生症,非典型雄激素過多症,21-羥化酶缺乏 | Adrenal hyperplasia, congenital / hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency |
| 腦腱性黃瘤症 | Cerebrotendinous xanthomatosis |

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| 維他命D依賴性佝僂病1A型 | Vitamin D-dependent rickets type 1A |
| 維他命D依賴性佝僂病1B型 | Vitamin D-dependent rickets, type IB |
| 楓糖尿症II型 | Maple syrup urine disease, type II |
| 歐門氏綜合症 | Omenn syndrome |
| 嚴重聯合免疫缺陷病, 阿薩巴斯卡型 | Severe combined immunodeficiency Athabaskan-type |
| 史密斯-萊姆利-奧皮茲綜合症 | Smith-Lemli-Opitz syndrome |
| 視網膜色素病變59 | Retinitis Pigmentosa 59 |
| 先天性糖基化障礙1bb型 | Congenital disorder of glycosylation, type 1bb |
| X-連鎖先天性角化不全譜系障礙 | Dyskeratosis congenita, X-linked |
| 二氫硫辛醯胺脫氫酶缺乏症 | Dihydrolipoamide dehydrogenase deficiency |
| 貝克型肌萎縮症 | Muscular dystrophy, Becker type |
| 杜興氏肌肉萎縮症 | Muscular dystrophy, Duchenne type |
| 低磷性佝僂病 | Hypophosphatemic rickets |
| 原發性纖毛運動障礙7 | Primary Ciliary Dyskinesia 7 |
| 原發性纖毛運動障礙3 | Primary Ciliary Dyskinesia 3 |
| 原發性纖毛運動障礙1 | Primary Ciliary Dyskinesia 1 |
| 5型3-甲基戊二酸尿症 | 3-methylglutaconic aciduria, type V |
| 免疫缺陷-著粒不穩定-面部異常綜合症1 | Immunodeficiency-centromeric instability-facial anomalies syndrome 1 |
| 先天性重症肌無力綜合症10 | Myasthenic syndrome, congenital, 10 |
| 先天性糖基化障礙Im型 | Congenital disorder of glycosylation, type Im |
| 小頭畸形症-身材矮小-四肢畸形 | Microcephaly, short stature, and limb abnormalities |
| 先天性糖基化障礙Ij型 | Congenital disorder of glycosylation, type Ij |
| 先天性糖基化障礙Ie型 | Congenital disorder of glycosylation, type Ie |
| 短肋胸廓發育不良伴或不伴多指綜合症 3 型 | Short-rib thoracic dysplasia 3 with or without polydactyly |
| 肢帶型肌肉萎縮症第2B型 | Limb-girdle muscular dystrophy, type 2B |
| 無汗外胚層發育不良1 | Ectodermal dysplasia 1, hypohidrotic |
| 家族性自主神經失調 | Familial dysautonomia |
| Emery-Dreifuss氏肌肉萎縮症1 | Emery-Dreifuss muscular dystrophy 1 |
| 低磷性佝僂病2 | Hypophosphatemic rickets 2 |
| 光敏感性毛髮低硫營養不良綜合症 1 型 | Trichothiodystrophy 1, photosensitive |
| 變異型著色性乾皮病,互補群D | Xeroderma pigmentosum, group D |
| 變異型著色性乾皮病,互補群F | Xeroderma pigmentosum, group F |
| 變異型著色性乾皮病,互補群G | Xeroderma pigmentosum, group G |
| 柯凱因氏綜合症B型 | Cockayne syndrome type B |
| 柯凱因氏綜合症A型 | Cockayne syndrome type A |
| 戊二酸血症IIA型 | Glutaric acidemia IIA |
| 戊二酸血症IIB型 | Glutaric acidemia IIB |
| 戊二酸血症IIC型 | Glutaric acidemia IIC |
| 乙基丙二酸腦病變 | Encephalopathy, ethylmalonic |
| 埃利偉氏綜合症 | Ellis-van Creveld syndrome |
| 第10凝血因子缺乏症 | Factor X deficiency |
| 第11凝血因子缺乏症, 隱性遺傳 | Factor XI deficiency, recessive |
| 第13A凝血因子缺乏症 | Factor XIIIA deficiency |
| 第13B凝血因子缺乏症 | Factor XIIIB deficiency |
| 第5凝血因子缺乏症 | Factor V deficiency |
| 血友病A | Hemophilia A |
| 血友病B | Hemophilia B |
| 酪氨酸血症I型 | Tyrosinemia, type I |
| 范干尼氏貧血, 互補群A | Fanconi anemia, complementation group A |
| 范干尼氏貧血, 互補群C | Fanconi anemia, complementation group C |

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| 范干尼氏貧血D2型 | Fanconi anemia type D2 |
| 范干尼氏貧血, 互補群G | Fanconi anemia, complementation group G |
| 范干尼氏貧血I型 | Fanconi anemia type I |
| 延胡索酸酶缺乏症 | Fumarase deficiency |
| 肌肉萎縮症-營養不良糖蛋白疾病A5型 | Muscular dystrophy-dystroglycanopathy, type A5 |
| 肌肉萎縮症-營養不良糖蛋白疾病B5型 | Muscular dystrophy-dystroglycanopathy, type B5 |
| 肌肉萎縮症-營養不良糖蛋白疾病C5型 | Muscular dystrophy-dystroglycanopathy, type C5 |
| 肌肉萎縮症-營養不良糖蛋白疾病A4型 | Muscular dystrophy-dystroglycanopathy, type A4 |
| 肌肉萎縮症-營養不良糖蛋白疾病B4型 | Muscular dystrophy-dystroglycanopathy, type B4 |
| 肌肉萎縮症-營養不良糖蛋白疾病C4型 | Muscular dystrophy-dystroglycanopathy, type C4 |
| 擴張型心肌病, 1X型 | Cardiomyopathy, dilated, 1X |
| 三甲基胺尿症 | Trimethylaminuria |
| 脆性X綜合症 | Fragile X mental retardation syndrome |
| 前庭導水管症候群 | Enlarged vestibular aqueduct |
| 岩藻糖沉積症 | Fucosidosis |
| 肝醣儲積症第Ia型 | Glycogen storage disease Ia |
| G6PC3缺乏嚴重先天性粒⊠胞缺乏症 | Severe congenital neutropenia due to G6PC3 deficiency |
| G6PD缺乏溶血性貧血 | Hemolytic anemia due to g6pd deficiency |
| 肝醣儲積症第II型 | Glycogen storage disease II |
| 克拉畢氏症 | Krabbe disease |
| 半乳糖差向異構酶缺乏症 | Galactose epimerase deficiency |
| 半乳糖激酶缺乏症併白內障 | Galactokinase deficiency with cataracts |
| 黏多醣症IVA型 | Mucopolysaccharidosis type IVA |
| 半乳糖血症 | Galactosemia |
| 腦肌酸缺乏症2 | Cerebral creatine deficiency syndrome 2 |
| 精胺酸:甘氨酸脒基轉移酶缺乏症 | Arginine:glycine amidinotransferase deficiency |
| 周產期致命性高雪氏症 | Gaucher disease, perinatal lethal |
| 高雪氏症第I型 | Gaucher disease, type I |
| 高雪氏症第II型 | Gaucher disease, type II |
| 高雪氏症第III型 | Gaucher disease, type III |
| 高雪氏症第IIIC型 | Gaucher disease, type IIIC |
| 肝醣儲積症第IV型 | Glycogen storage disease IV |
| 成人多葡萄聚糖體神經病變 | Polyglucosan body neuropathy, adult form |
| 戊二酸血症I型 | Glutaric acidemia I |
| BH4 缺乏性高苯丙氨酸血症 C 型 | Hyperphenylalaninemia, BH4-deficient, C |
| 單一生長激素缺乏症 IA 型 | Growth hormone deficiency, isolated, type IA |
| 單一生長激素缺乏症 IB 型 | Growth hormone deficiency, isolated, type IB |
| Laron綜合症 | Laron syndrome |
| 單一生長激素缺乏症 4 型 | Growth hormone deficiency, isolated, type IV |
| 部分單一生長激素缺乏症 | Growth hormone deficiency, isolated partial |
| 1q21.1 復發性微缺失/微重複症候群 | 1q21.1 recurrent microdeletion/microduplication syndrome |
| 耳聾, 隱性遺傳1a | Deafness, recessive 1a |
| 耳聾, 隱性遺傳1B | Deafness, autosomal recessive 1B |
| 法布瑞氏症 | Fabry disease |
| GM1-神經節苷脂儲積症第I型 | GM1-gangliosidosis, type I |
| GM1-神經節苷脂儲積症第II型 | GM1-gangliosidosis, type II |
| GM1-神經節苷脂儲積症第III型 | GM1-gangliosidosis, type III |
| 黏多醣症IVB型 | Mucopolysaccharidosis type IVB (Morquio) |
| 高甘胺酸血症, 腦病變 1 | Hyperglycinemia encephalopathy 1 |
| 黏脂質症 α / β 第II型 | Mucolipidosis alpha/beta II |

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| 黏脂質症 α / β 第III型 | Mucolipidosis alpha/beta III |
| 黏脂質症第III型 | Mucolipidosis type III |
| 黏多醣症IIID型 | Mucopolysaccharidosis, type IIID |
| 伯納德－蘇里爾綜合症 A1 | Bernard-Soulier syndrome, type A1 |
| Bernard-Soulier綜合症B 型 | Bernard-Soulier syndrome, type B |
| 伯納德－蘇里爾綜合症 C | Bernard-Soulier syndrome, type C |
| X-連鎖先天性眼球震顫 6 型 | Nystagmus 6, congenital, X-linked |
| 原發性高草酸尿症第II型 | Hyperoxaluria, primary, type II |
| 弗雷澤綜合症3 | Fraser syndrome 3 |
| 黏多醣症VII型 | Mucopolysaccharidosis type VII |
| 中鏈/短鏈3-羥基醯基輔酶A去氫酵素缺乏症 | Medium/short-chain 3-hydroxyacyl-CoA dehydrogenase deficiency |
| 長鏈3-羥基醯基輔酶A去氫酵素缺乏症 | Long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency |
| 三功能蛋白缺乏症 | Trifunctional protein deficiency |
| 線粒體三功能蛋白缺乏症 | Mitochondrial trifunctional protein deficiency |
| 嚴重先天性中性粒細胞減少症第3型 | Neutropenia, severe congenital, 3 |
| 甲型地中海貧血 | Alpha-Thalassemia |
| 乙型地中海貧血 | Beta-Thalassemia |
| 鐮刀型貧血 | Sickle cell anemia |
| 甲基丙二酸血症伴高胱胺酸血症 cblX 型 | methylmalonic acidemia and homocysteinemia, cblX type |
| 戴薩克斯症 | Tay-Sachs disease |
| 嬰兒、青少年及成人山德霍夫氏症 | Sandhoff disease, infantile, juvenile, and adult forms |
| 血色病 | Hemochromatosis |
| 黑尿症 | Alkaptonuria |
| 黏多醣症IIIC型 | Mucopolysaccharidosis, type IIIC (Sanfilippo C) |
| 遺傳性血色病2A型 | Hereditary hemochromatosis type 2A |
| 全羧化酶合成酶缺乏症 | Holocarboxylase synthetase deficiency |
| 羥甲基戊二酰輔酶A裂解酶缺乏症 | Hydroxymethylglutaryl-coa lyase deficiency |
| 3-羥基-3-甲基戊二醯輔酶A合成酶 2 缺乏症 | HMG-CoA synthase-2 deficiency |
| 17q12 複發性區域 | 17q12 recurrent (RCAD syndrome) region |
| 酪胺酸血症第III型 | Tyrosinemia type III |
| 赫曼斯基-普德拉克氏綜合症1 | Hermansky-Pudlak syndrome 1 |
| 赫曼斯基-普德拉克氏綜合症3 | Hermansky-Pudlak syndrome 3 |
| 男性假雌雄同體,乳房發育症 | Pseudohermaphroditism, male, with gynecomastia |
| 先天性腎上腺皮質增生症, 3- β -羥基類固醇脫氫酶2缺乏 | Adrenal hyperplasia, congenital, due to 3-beta- hydroxysteroid dehydrogenase 2 deficiency |
| 8型3-甲基戊二酸尿症 | 3-methylglutaconic aciduria, type VIII |
| Xp11.22微小重複綜合症 | Xp11.22 micro-repeat syndrome |
| 黏多醣症IX型 | Mucopolysaccharidosis type IX |
| 黏多醣症II型 | Mucopolysaccharidosis, type II |
| 黏多醣症Ih型 | Mucopolysaccharidosis Ih |
| 黏多醣症Ih/Is型 | Mucopolysaccharidosis Ih/s |
| 黏多醣症Is型 | Mucopolysaccharidosis Is |
| 嚴重聯合免疫缺陷病 | Severe combined immunodeficiency |
| IL7R- α 缺乏嚴重聯合免疫缺陷病 | Severe combined immunodeficiency due to IL7R-alpha deficiency |
| 交界性水皰性表皮鬆解症伴隨幽門閉鎖6 | Junctional epidermolysis bullosa with pyloric atresia 6 |
| 非Herlitz型交界性水皰性表皮鬆解症5A | Epidermolysis bullosa, junctional, 5A, non-Herlitz type |
| 交界性水皰性表皮鬆解症伴隨幽門閉鎖5B | Junctional epidermolysis bullosa with pyloric atresia 5B |
| 異戊酸血症 | Isovaleric acidemia |
| JAK3缺乏嚴重聯合免疫缺陷病 | Severe combined immunodeficiency due to JAK3 deficiency |
| 巴特綜合症2型 | Bartter syndrome type 2 |
| 雙基因前庭導水管症候群 | Enlarged vestibular aqueduct, digenic |

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| 家族性胰島素過度分泌低血糖症2 | Hyperinsulinemic hypoglycemia, familial, 2 |
| 單純型良性水皰性表皮鬆解症 1 型 | Epidermolysis bullosa simplex, recessive 1 |
| 單純型良性水皰性表皮鬆解症 2 型 | Epidermolysis bullosa simplex, recessive 2 |
| 中腦導水管狹窄性腦積水 | Hydrocephalus due to aqueductal stenosis |
| 先天性Merosin 蛋白缺乏型肌肉萎縮症 | Muscular dystrophy, congenital, merosin deficient or partially deficient |
| 常染色體隱性肢帶型肌肉萎縮症第23 型 | Muscular dystrophy, limb-girdle, autosomal recessive 23 |
| Herlitz型交界性水皰性表皮鬆解症2B | Epidermolysis bullosa, junctional, 2B, Herlitz type |
| 廣泛性水皰性表皮鬆解症 | Epidermolysis bullosa, generalized |
| 喉-甲-皮綜合症 | Laryngoonychocutaneous syndrome |
| 非Herlitz型交界性水皰性表皮鬆解症1A | Epidermolysis bullosa, junctional, 1A, non-Herlitz type |
| Herlitz型交界性水皰性表皮鬆解症1B | Epidermolysis bullosa, junctional, 1B, Herlitz type |
| 非Herlitz型交界性水皰性表皮鬆解症3A | Epidermolysis bullosa, junctional, 3A, non-Herlitz type |
| Herlitz型交界性水皰性表皮鬆解症3B | Epidermolysis bullosa, junctional, 3B, Herlitz type |
| 肌肉萎縮症-營養不良糖蛋白疾病 | Muscular dystrophy-dystroglycanopathy |
| 肌肉萎縮症-營養不良糖蛋白疾病B6 型（伴隨智力發展遲緩型） | Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 6 |
| 家族性高膽固醇血症 1 | Familial hypercholesterolemia 1 |
| 結合性腦下垂體激素缺乏症 3 | Combined pituitary hormone deficiency 3 |
| 溶酶體酸性脂肪酶缺乏症 | Lysosomal acid lipase deficiency (includes Wolman disease and Cholesterol ester storage disease) |
| Cobalamin F 缺乏症 | Cobalamin F deficiency |
| 眼皮膚白化病7型 | Albinism, oculocutaneous, type VII |
| Donnai-Barrow綜合症 | Donnai-Barrow syndrome |
| 骨質疏鬆症-假性神經膠質瘤綜合症 | Osteoporosis-pseudoglioma syndrome |
| 契東綜合症 | Chediak-Higashi syndrome |
| α-甘露糖苷貯積症B, I及II型 | Mannosidosis, Alpha B, types I, II |
| 3-甲基巴豆醯輔酵素羧化酵素1缺乏症 | 3-methylcrotonyl-CoA carboxylase 1 deficiency |
| 3-甲基巴豆醯輔酵素羧化酵素2缺乏症 | 3-methylcrotonyl-CoA carboxylase 2 deficiency |
| 甲基丙二酸醯輔酶A差向異構酶缺乏症 | Methylmalonyl-CoA epimerase deficiency |
| 黏脂質症第IV型 | Mucolipidosis IV |
| 原發性小頭畸形症 | Primary microcephaly |
| MECP2重複綜合症 | MECP2 Duplication Syndrome (Mental retardation, X-linked syndromic, Lubs type) |
| Xq28(MECP2)微缺失相關Rett 症候群 | Rett syndrome |
| 家族性地中海熱, 隱性遺傳 | Familial mediterranean fever, recessive |
| 神經元蠟樣脂褐質儲積症7 | Ceroid lipofuscinosis, neuronal 7 |
| Opitz-GBBB綜合症 | Opitz GBBB syndrome |
| 巴德-畢德氏綜合症, 13 | Bardet-Biedl syndrome, 13 |
| 梅克爾綜合症1 | Meckel syndrome 1 |
| 巨腦性白質腦病變併皮質下囊腫1 | Megalencephalic leukoencephalopathy with subcortical cysts 1 |
| 丙二醯輔酶A脱羧酶缺乏症 | Malonyl-CoA decarboxylase deficiency |
| 甲基丙二酸尿症, 維他命B12敏感 | Methylmalonic aciduria, vitamin B12-responsive |
| 甲基丙二酸尿症cblB型 | Methylmalonic aciduria, cblB type |
| 甲基丙二酸尿症和高胱氨酸尿症,cblC型 | Methylmalonic aciduria and homocystinuria, cblC type |
| 甲基丙二酸尿症/高胱氨酸尿症,cblD型, 變異1, 2 | Methylmalonic aciduria/homocystinuria, type cblD, variants 1, 2 |
| 鉬輔酶缺乏症A | Molybdenum cofactor deficiency A |
| 先天性糖基化障礙IIB型 | Congenital disorder of glycosylation, type IIb |
| 先天性糖基化障礙If型 | Congenital disorder of glycosylation, type If |
| 先天性糖基化障礙IB型 | Congenital disorder of glycosylation, type Ib |
| 肌管肌肉病變 | Myotubular myopathy |
| cobalamin G缺乏高胱胺酸尿症 | Homocystinuria due to cobalamin G deficiency |
| 同型半胱氨酸尿症, cblE型 | Homocystinuria, cobalamin E type |
| 甲基丙二酸尿症，mut(0)型 | Methylmalonic aciduria, mut(0) type |

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| 高免疫球蛋白D綜合症 | Hyper IgD syndrome |
| 甲羥戊酸尿症 | Mevalonic aciduria |
| 耳聾, 隱性遺傳3 | Deafness, recessive 3 |
| 耳聾, 隱性遺傳2 | Deafness, recessive 2 |
| 尤塞氏綜合症IB型 | Usher syndrome, type IB |
| 2,4-二烯醯輔酶A還原酶缺乏症 | 2,4-dienoyl-CoA reductase deficiency |
| α-N-乙酰半乳糖胺酶缺乏症 | Alpha-N-acetylgalactosaminidase deficiency I/III |
| 黏多醣症IIIB型 | Mucopolysaccharidosis, type IIIB (sanfilippo B) |
| N-乙酰谷氨酸合酶缺乏症 | N-Acetylglutamate synthase deficiency |
| 奈梅亨氏綜合症 | Nijmegen syndrome |
| 慢性肉芽腫病1 | Chronic granulomatous disease 1 |
| 慢性肉芽腫病2 | Chronic granulomatous disease 2 |
| 慢性肉芽腫病3 | Chronic granulomatous disease 3, autosomal recessive |
| Norrie 氏病 | Norrie disease |
| 桿狀體肌肉病變2 | Nemaline myopathy 2 |
| 尼曼匹克症C1,D型 | Niemann-Pick disease, types C1, D |
| 尼曼匹克症C2型 | Niemann-pick disease, type C2 |
| 梅克爾綜合症7 | Meckel syndrome 7 |
| 腎消耗病3 型 | Nephronophthisis 3 |
| 腎-肝-胰臟發育不良 1 型 | Renal-hepatic-pancreatic dysplasia 1 |
| 腎病綜合症第1型 | Nephrotic syndrome, type 1 |
| 腎病綜合症第2型 | Nephrotic syndrome, type 2 |
| 肢端及中部骨骺發育不良Maroteaux 型 | Acromesomelic dysplasia, maroteaux type |
| 先天性腎上腺發育不良 | Adrenal hypoplasia, congenital |
| 進行性家族性肝內膽汁滯留症5型 | Cholestasis, progressive familial intrahepatic 5 |
| Seckel綜合症10 型 | Seckel syndrome 10 |
| 鳥胺酸轉氨酶缺乏症 | Ornithine aminotransferase deficiency |
| 眼皮膚白化病2型 | Oculocutaneous albinism type 2 |
| Dent 病 2 型 | Dent disease 2 |
| 朱伯特綜合症10 | Joubert syndrome 10 |
| 3型3-甲基戊二酸尿症 | 3-methylglutaconic aciduria, type III |
| Meier-Gorlin綜合症1 | Meier-Gorlin syndrome 1 |
| Meier-Gorlin綜合症2 | Meier-Gorlin syndrome 2 |
| Meier-Gorlin綜合症3 | Meier-Gorlin syndrome 3 |
| 鳥氨酸氨甲醯基轉移酶缺乏症 | Ornithine transcarbamylase deficiency |
| 耳聾, 隱性遺傳9 | Deafness, autosomal recessive 9 |
| 苯丙酮尿症 | Phenylketonuria |
| 生物喋呤缺乏高苯丙氨酸血症D | Biopterin-deficient hyperphenylalaninemia D |
| 丙酸血症 | Propionic acidemia |
| 耳聾23 | Deafness 23 |
| 尤塞氏綜合症IF型 | Usher syndrome, type IF |
| 過氧化物酶體生源失調症1A | Peroxisome biogenesis disorder 1A (Zellweger) |
| 過氧化物酶體生源失調症6A | Peroxisome biogenesis disorder 6A (Zellweger) |
| 過氧化物酶體生源失調症3A | Peroxisome biogenesis disorder 3A (Zellweger) |
| 過氧化物酶體生源失調症7A | Peroxisome biogenesis disorder 7A (Zellweger) |
| 過氧化物酶體生源失調症4A | Peroxisome biogenesis disorder 4A (Zellweger) |
| 肢近端型點狀軟骨發育不良第1型 | Rhizomelic chondrodysplasia punctata, type 1 |
| 肝醣儲積症第VII型 | Glycogen storage disease VII |
| 肝醣儲積症第IXb型 | Glycogen storage disease IXb |
| 耳聾, 隱性遺傳59 | Deafness, autosomal recessive 59 |

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| 多囊腎和肝病 | Polycystic kidney and hepatic disease |
| 單純型良性水皰性表皮鬆解症伴隨幽門閉鎖 | Epidermolysis bullosa simplex 5C, with pyloric atresia |
| 單純型良性水皰性表皮鬆解症伴隨肌肉萎縮症 | Epidermolysis bullosa simplex 5B, with muscular dystrophy |
| 脊柱側凸型埃勒斯-當洛二氏綜合症 | Ehlers-Danlos syndrome, kyphoscoliotic type |
| Pelizaeus- Merzbacher 病 | Pelizaeus- Merzbacher's disease |
| 先天性糖基化障礙IA型 | Congenital disorder of glycosylation, type Ia |
| 線粒體DNA耗竭綜合症4A | Mitochondrial DNA depletion syndrome 4A |
| 線粒體DNA耗竭綜合症4B | Mitochondrial DNA depletion syndrome 4B |
| 肌肉萎縮症-營養不良糖蛋白疾病B3型 | Muscular dystrophy-dystroglycanopathy type B3 |
| 肌肉萎縮症-營養不良糖蛋白疾病C3型 | Muscular dystrophy-dystroglycanopathy type C3 |
| 肌肉萎縮症-營養不良糖蛋白疾病A3型 | Muscular dystrophy-dystroglycanopathy type A3 |
| 肌肉萎縮症-營養不良糖蛋白疾病C1型 | Muscular dystrophy-dystroglycanopathy, type C1 |
| 肌肉萎縮症-營養不良糖蛋白疾病C2型 | Muscular dystrophy-dystroglycanopathy (limb-girdle), type C2 |
| 結合性腦下垂體激素缺乏症 1 | Combined pituitary hormone deficiency 1 |
| 神經元蠟樣脂褐質儲積症1 | Ceroid lipofuscinosis, neuronal 1 |
| 甲基丙二酸尿症症伴高胱胺酸尿症 cblC 型 | Methylmalonic aciduria and homocystinuria, cblC type, digenic |
| 先天性重症肌無力綜合症22 | Myasthenic syndrome, congenital, 22 |
| 噬血細胞性淋巴組織細胞增多症2型 | Familial hemophagocytic lymphohistiocytosis type 2 |
| 泛腦垂體機能低下症 | Panhypopituitarism |
| 進行性神經性腓骨萎縮症5 | Charcot-Marie-Tooth disease 5 |
| 磷酸核糖焦磷酸合成酶超活性痛風症 | Gout/ phosphoribosylpyrophosphate synthetase superactivity |
| SAP-b缺陷所致雜合子腦白質營養不良" | Heterozygous cerebral leukodystrophy due to SAP-b deficiency |
| 軟骨發育異常Blomstrand型 | Chondrodysplasia, Blomstrand type |
| CD45缺乏嚴重聯合免疫缺陷病 | Severe combined immunodeficiency due to CD45 deficiency |
| 高苯丙氨酸血症 | Hyperphenylalaninemia |
| 肝醣儲積症第VI型 | Glycogen storage disease VI |
| 肝醣儲積症第V型 | McArdle disease |
| 生物喋呤缺乏高苯丙氨酸血症C | Biopterin-deficient hyperphenylalaninemia C |
| RAG1缺乏嚴重聯合免疫缺陷病 | Severe combined immunodeficiency due to RAG1 deficiency |
| 嚴重聯合免疫缺陷病, B細胞缺乏 | Severe combined immunodeficiency (B cell negative) |
| 先天性重症肌無力綜合症11 | Congenital myasthenic syndrome, 11 |
| 腦橋小腦發育不全6型 | Pontocerebellar hypoplasia type 6 |
| 先天性糖基化障礙In型 | Congenital disorder of glycosylation, type In |
| Aicardi-Goutieres綜合症2 | Aicardi-Goutieres syndrome 2 |
| 朱伯特綜合症7 | Joubert syndrome 7 |
| 梅克爾綜合症5 | Meckel syndrome 5 |
| 視網膜分裂症 | Retinoschisis |
| 痙攣性共濟失調, 沙勒沃伊-薩格奈型 | Spastic ataxia, Charlevoix-Saguenay type |
| Shwachman-Diamond綜合症 | Shwachman-Diamond syndrome |
| 先天性重症肌無力綜合症16 | Myasthenic syndrome, congenital, 16 |
| 心腦肌病變 | Cardioencephalomyopathy |
| 3-甲基戊二酸尿症併耳聾、腦病變及萊氏類綜合症 | 3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome |
| α-1-抗胰蛋白酶缺乏症 | Alpha-1-antitrypsin deficiency |
| 肢帶型肌肉萎縮症第2D型 | Muscular dystrophy, limb-girdle, type 2D |
| 肢帶型肌肉萎縮症第2E型 | Muscular dystrophy, limb-girdle, type 2E |
| 肢帶型肌肉萎縮症第2F型 | Muscular dystrophy, limb-girdle, type 2F |
| 肢帶型肌肉萎縮症第2C型 | Muscular dystrophy, limb-girdle, type 2C |
| 黏多醣症IIIA型 | Mucopolysaccharidosis, type IIIA (Sanfilippo A) |
| 16p11.2 複發性區域 (遠端，BP2-BP3) | 16p11.2 recurrent region (distal, BP2-BP3) |
| X 連鎖淋巴球增生症候群 1 型 | Lymphoproliferative syndrome, X-linked, 1 |

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| 肢中部發育不良 | Langer mesomelic dysplasia |
| 巴特綜合症1型 | Bartter syndrome type 1 |
| 吉特曼氏綜合症 | Gitelman syndrome |
| 胼胝體發育不全併周圍神經病變 | Agenesis of the corpus callosum with peripheral neuropathy |
| 生物素反應性基底節病 | Biotin-responsive basal ganglia disease |
| 肉鹼缺乏症,系統型原發性 | Carnitine deficiency, systemic primary |
| 眼皮膚白化病, VI型 | Albinism, oculocutaneous, type VI |
| 新生兒瓜氨酸血症II, | Citrullinemia, type II, neonatal-onset |
| 成人瓜氨酸血症II, | Citrullinemia, adult-onset type II |
| 高鳥胺酸血症-高氨血症-高瓜胺酸血綜合症 | Hyperornithinemia-hyperammonemia- homocitrullinuria syndrome |
| 肉鹼轉位酶缺乏症 | Carnitine-acylcarnitine translocase deficiency |
| 軟骨發育不全第Ib型 | Achondrogenesis, type Ib |
| 骨發育不全症第二型 | Atelosteogenesis, type II / De la Chapelle dysplasia |
| 畸型發育不良 | Diastrophic dysplasia |
| 多發性骨骺發育不良4 | Epiphyseal dysplasia, multiple, 4 |
| 耳聾, 隱性遺傳4 | Deafness, recessive 4 |
| 甲狀腺腫大併耳聾綜合症 | Pendred syndrome |
| 低磷性佝僂病伴高尿鈣 | Hypophosphatemic rickets with hypercalciuria |
| 先天性糖基化障礙IIf型 | Congenital disorder of glycosylation, type IIf |
| 先天性糖基化障礙IIc型 | Congenital disorder of glycosylation, type IIc |
| 肝醣儲積症第Ib型 | Glycogen storage disease Ib |
| 肝醣儲積症第Ic型 | Glycogen storage disease Ic |
| 埃勒斯-當洛二氏綜合症-脊椎發育不全3 型 | Ehlers-Danlos syndrome, spondylodysplastic type 3 |
| 眼皮膚白化病, IV型 | Albinism, oculocutaneous, type IV |
| 腦肌酸缺乏症1 | Cerebral creatine deficiency syndrome 1 |
| SCHIMKE免疫-骨發育不良 | Schimke immuno-osseous dysplasia |
| 脊髓性肌肉萎縮症1 | Spinal muscular atrophy 1 |
| 脊髓性肌肉萎縮症2 | Spinal muscular atrophy 2 |
| 脊髓性肌肉萎縮症3 | Spinal muscular atrophy 3 |
| 脊髓性肌肉萎縮症4 | Spinal muscular atrophy 4 |
| 尼曼匹克症A型 | Niemann-pick disease, type A |
| 尼曼匹克症B型 | Niemann-pick disease, type B |
| Netherton 綜合症 | Netherton syndrome |
| 墨蝶呤還原酶缺乏症 | Sepiapterin reductase deficiency |
| 脂質先天性腎上腺皮質增生症 | Lipoid congenital adrenal hyperplasia |
| X-連鎖魚鱗癬 | Ichthyosis, X-linked |
| 噬血細胞性淋巴組織細胞增多症4型 | Familial hemophagocytic lymphohistiocytosis type 4 |
| 噬血細胞性淋巴組織細胞增多症5型 | Familial hemophagocytic lymphohistiocytosis type 5 |
| 酪胺酸血症第II型 | Tyrosinemia type II |
| 巴氏綜合症 | Barth syndrome |
| 早期嬰兒型癲癇性腦病變 16 型 | Epileptic encephalopathy, early infantile, 16 |
| 22q11.2 微缺失/微重複症候群 | 22q11.2 microdeletion/microduplication syndrome |
| 16p11.2 複發性區域 (近端，BP4-BP5) | 16p11.2 recurrent region (proximal, BP4-BP5) |
| 骨質石化症 | Osteopetrosis |
| 轉鐵蛋白缺乏症 | Atransferrinemia |
| 血色病第3型 | Hemochromatosis, type 3 |
| 先天性魚鱗癬1 | Ichthyosis, congenital, 1 |
| 酪氨酸羥化酶缺乏症 | Segawa syndrome |
| 9型3-甲基戊二酸尿症 | 3-Methylglutaconic Aciduria type 9 |
| 進行性家族性肝內膽汁滯留症4型 | Cholestasis, progressive familial intrahepatic 4 |

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| 非綜合症型耳聾7 | Nonsyndromic deafness 7 |
| 朱伯特綜合症2 | Joubert syndrome 2 |
| 梅克爾綜合症 | Meckel syndrome |
| COACH綜合症1 | COACH syndrome 1 |
| 朱伯特綜合症6 | Joubert syndrome 6 |
| 梅克爾綜合症3 | Meckel syndrome 3 |
| 腎消耗病11 型 | Nephronophthisis 11 |
| 埃勒斯-當洛二氏綜合症經典1型 | Ehlers-Danlos syndrome, classic-like, 1 |
| 神經元蠟樣脂褐質儲積症2 | Ceroid lipofuscinosis, neuronal 2 |
| 侏儒症併肌肉、肝、腦、眼異常 | Mulibrey nanism |
| 軟骨發育不全第IA型 | Achondrogenesis, type IA |
| 肢帶型肌肉萎縮症第2J型 | Muscular dystrophy, limb-girdle, type 2J |
| 眼皮膚白化病, IA型 | Albinism, oculocutaneous, type IA |
| 眼皮膚白化病, IB型 | Albinism, oculocutaneous, type IB |
| 眼皮膚白化病3型 | Oculocutaneous albinism type 3 |
| 克果納傑氏綜合症I型 | Crigler-Najjar syndrome, types I |
| 克果納傑氏綜合症II型 | Crigler-Najjar syndrome, types II |
| 噬血細胞性淋巴組織細胞增多症3型 | Familial hemophagocytic lymphohistiocytosis type 3 |
| 耳聾18A | Deafness 18A |
| 尤塞氏綜合症IC型 | Usher syndrome, type IC |
| 尤塞氏綜合症IG型 | Usher syndrome, type 1G |
| 尤塞氏綜合症IIa型 | Usher syndrome, type IIa |
| 維他命D依賴性佝僂病1A型 | Vitamin D-dependent rickets type 2A |
| 科恩綜合症 | Cohen syndrome |
| X連鎖嚴重先天性中性粒細胞減少症 | Neutropenia, severe congenital, X-linked |
| X連鎖血小板減少症 | Thrombocytopenia, X-linked |
| Wiskott-Aldrich綜合症 | Wiskott-Aldrich syndrome |
| Wolfram 綜合症 1 型 | Wolfram syndrome 1 |
| 尤塞氏綜合症2D型 | Usher syndrome, type 2D |
| X 連鎖淋巴球增生症候群2型 | Lymphoproliferative syndrome, X-linked, 2 |
| 着色性乾皮症,互補群A | Xeroderma pigmentosum complemetation group A |
| 着色性乾皮症,互補群C | Xeroderma pigmentosum complemetation group C |
| Int22h1/Int22h2類Xq28重複綜合症 | Int22h1/Int22h2-mediated Xq28 duplication syndrome |
| 身材矮小-小頭畸形症-內分泌功能障礙 | Short stature - microcephaly - endocrine dysfunction |
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