	GENE TEST記載疾患名	GENE TEST記載遺伝子名	申請診療科	申請疾患名(英名)	申請疾患名(和名)
1	**Aarskog-Scott syndrome**	*FDGL*	小児科	Aarskog-Scott syndrome	アースコグ－スコット症候群
2	**Achondrogenesis**	*COL2A1*	小児科	achondrogenesis	軟骨無発生症
	Achondrogenesis Type 1A				
	Type II Collagenopathies				
	Achondrogenesis Type 2				
	Kniest Dysplasia				
	Spondyloepiphyseal Dysplasia				
	Spondyloepiphyseal Dysplasia, Congenita				
	Stickler Syndrome Type I				
3	**Achondroplasia**		小児科	achondroplasia	軟骨無形成症
	Achondroplasia	*FGFR3*			
	Pseudoachondroplasia	*COMP*			
	Severe Achondroplasia with Developmental Delay and Acanthosis Nigricans (SADDAN)	*FGFR3*			
4	**Adenosine Deaminase Deficiency**	*ADA*	小児科	ADENOSINE DEAMINASE DEFICIENCY	アデノシンデアミナーゼ欠損症
5	**Adenylosuccinase Deficiency**	*ADSL*	小児科	ADENYLOSUCCINASE DEFICIENCY	アデニロスクシナーゼ欠損症
6	**Adrenoleukodystrophy**		小児科	ADRENOLEUKODYSTROPHY	副腎白質ジストロフィー
	Peroxisome Biogenesis Disorders (PBD)	(別項参照)	小児科	ADRENOLEUKODYSTROPHY, AUTOSOMAL NEONATAL FORM	副腎白質ジストロフィー, 常染色体性新生児型
	Zellweger Syndrome Spectrum		小児科	PSEUDONEONATAL ADRENOLEUKODYSTROPHY	副腎白質ジストロフィー,偽新生児型
	Neonatal Adrenoleukodystrophy				
	Adrenoleukodystrophy, X-Linked	*ABCD1*			
	Adrenomyeloneuropathy				
	Pseudoneonatal Adrenoleukodystrophy	*ACOX1*			
7	**Aldolase A Deficiency**	*ALDOA*	小児科	aldolase deficiency	アルドラーゼ欠損症
8	**Alkaptonuria**	*HGD*	小児科	ALKAPTONURIA	アルカプトン尿症
9	**Alport Syndrome**		眼科	Alport syndrome	アルポート症候群
	Alport Syndrome		小児科	Epstein syndrome	エプスタイン 症候群
	Alport Syndrome, Autosomal Recessive	*COL4A3,COL4A4*	小児科	Fechtner syndrome	フェクトナー症候群
	Alport Syndrome, X-Linked	*COL4A5*			
	Alport Syndrome with Leukocyte Inclusions and Macrothrombocytopenia [**Fechtner Syndrome**]	*MYH9*			
	Alport Syndrome with Macrothrombocytopenia [**Epstein Syndrome**]	*MYH9*			
10	**Alzheimer Disease**		神経内科 精神科神経科	familial Alzheimer disease	家族性アルツハイマー病
	Alzheimer Disease				
	Early-Onset Familial Alzheimer Disease				
	Alzheimer Disease Type 1	*APP*			
	Alzheimer Disease Type 3	*PSEN1*			
	Alzheimer Disease Type 4	*PSEN2*			
	Late-Onset Familial Alzheimer Disease				
	Alzheimer Disease Type 5	*AD5*			
	Alzheimer Disease Type 2 [Apolipoprotein E Genotyping]	*APOE*			
11	**Amegakaryocytic Thrombocytopenia, Congenital**	*MPL*	小児科	congenital amegakaryocytic thrombocytopenia	先天性無巨核芽球性血小板減少症
12	**Amyloidosis**		小児科	AMYLOID POLYNEUROPATHY,FAMILIAL	家族性アミロイドポリニューロパチー
	Transthyretin Amyloidosis	*TTR*	小児科	AMYLOIDOSIS, FAMILIAL VISCERAL	家族性アミロイドーシス
	Familial Amyloid Cardiomyopathy		神経内科	familial amyloid polyneuropathy	家族性アミロイドポリニューロパチー
	Familial Amyloid Polyneuropathy Type 1(Portuguese-Swedish-Japanese type)				
	Familial Amyloid Polyneuropathy Type II (Indiana/Swiss or Maryland/German type)				
	Familial Oculoleptomeningeal Amyloidosis				
	Leptomeningeal Amyloidosis				
	Urticaria-Deafness-Amyloidosis Syndrome [Muckle-Wells Syndrome]	*CIAS1*			
	Amyloidosis V	*GSN*			
13	**Amyotrophic Lateral Sclerosis**		小児科　神経内科	familial amyotrophic lateral sclerosis	家族性筋萎縮性側索硬化症
	Amyotrophic Lateral Sclerosis with Frontotemporal Dementia	*ALS2,ALS3,ALS4,ALS5,ALS6,ALS7*			
	SOD1-Related Amyotrophic Lateral Sclerosis	*SOD1(=ALS1)*			
14	**Angelman syndrome**	*UBE3A*	小児科	Angelman syndrome	アンジェルマン症候群
15	**Aniridia**		眼科	aniridia	無虹彩
	Isolated Aniridia	*PAX6*	小児科　眼科	WAGR syndrome	WAGR症候群
	WAGR Syndrome [Wilms Tumor-Aniridia-Genital Anomalies-Retardation Syndrome]	*PAX6,WT1*			
16	**Anterior Segment Mesenchymal Dysgenesis**	*FOXE3,PITX2,PITX3*	眼科	anterior segment mesenchymal dysgenesis	前眼部間葉組織異形成
17	**ARX-Related Disorders**	*ARX*	小児科	Infantile spasms, West syndrome	点頭てんかん、ウエスト症候群
	Nonspecific X-Linked Mental Retardation-36				
	Partington X-Linked Mental Retardation Syndrome				
	X-Linked Infantile Spasm Syndrome				
	X-Linked Lissencephaly with Ambiguous Genitalia				

18	**Ataxia-Telangiectasia**		小児科	Ataxia-telangiectasia	毛細血管拡張性失調症
	Ataxia-Telangiectasia	*ATM*			
	Ataxia-Telangiectasia Variant 1 (ATV1) [Nijmegen Breakage Syndrome]	*NBS1*			
19	**Atelosteogenesis**		小児科	Atelosteogenesis	骨発生不全症
	Sulfate Transporter-Related Osteochondrodysplasia	*SLC26A2(=DTDST)*			
	Achondrogenesis Type 1B				
	Atelosteogenesis Type 2				
	Diastrophic Dysplasia				
	Multiple Epiphyseal Dysplasia, Recessive				
20	**ATP7A-Related Copper Transport Disorders**	*ATP7A*	小児科	MENKES SYNDROME	メンケス症候群
	Menkes Disease				
21	**Azoospermia [Y Chromosome Infertility]**	*DAZ,DDX3Y,USP9Y,RBMY1A1*	泌尿器科	azoospermia,oligozoospermia	無精子症,乏精子症
22	**Basal Cell Nevus Syndrome (BCNS) [Nevoid Basal Cell Carcinoma Syndrome]**	*PTCH*	小児科	BASAL CELL NEVUS SYNDROME, Gorlin syndrome	母斑性基底細胞癌症候群、Gorlin症候群
23	**Beckwith-Wiedemann Syndrome**	*BWS*	小児科	Beckwith-Wiedemann syndrome	ベックウィズ−ウィードマン症候群
24	**Best Vitelliform Macular Dystrophy**	*VMD2*	眼科	Best disease	ベスト病
25	**Biotin-Responsive Multiple Carboxylase Deficiencies**		小児科	BIOTINIDASE DEFICIENCY	ビオチナーゼ欠損症
	Juvenile Multiple Carboxylase Deficiency [Biotinidase Deficiency]	*BTD*	小児科	MULTIPLE CARBOXYLASE DEFICIENCY	ビオチン反応性マルチプルカルボキシラーゼ欠損症
	Holocarboxylase Synthetase Deficiency	*HLCS*			
256	**Blau syndrome**		皮膚科	SYNOVITIS, GRANULOMATOUS, WITH UVEITIS AND CRANIAL NEUR	ブラウ症候群
	Early-Onset Sarcoidosis	*CARD15/NOD2*		Early-onset sarcoidosis	若年発症サルコイドーシス
26	**Bloom Syndrome**	*BLM*	小児科	Bloom syndrome	ブルーム症候群
27	**BRCA1 and BRCA2 Hereditary Breast/Ovarian Cance**		第一外科	familial breast cancer	家族性乳癌
	Breast cancer [BRCA1 Hereditary Breast/Ovarian Cancer]	*BRCA1*			
	Breast cancer [BRCA2 Hereditary Breast/Ovarian Cancer]	*BRCA2*			
28	**Bruton's Agammaglobulinemia [X-Linked Agammaglobulinemia]**	*BTK*	小児科	Bruton agammaglobulinemia	ブルトン型無ガンマグロブリン血症
29	**Campomelic Dysplasia**	*SOX9*	小児科	campomelic dysplasia	屈曲肢異形成症
30	**Carnitine Deficiency, Systemic**	*SLC22A5(=OCTN2)*	小児科	CARNITINE DEFICIENCY, SYSTEMIC PRIMARY	原発性カルニチン欠損症
31	**Carnitine Palmitoyltransferase Deficiency**		小児科	CARNITINE PALMITOYLTRANSFERASE I DEFICIENCY	カルニチンパルミトイルトランスフェラーゼ I 欠損症
	Carnitine Palmitoyltransferase IA (liver) Deficiency	*CPT1A*	小児科	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY	カルニチンパルミトイルトランスフェラーゼ II 欠損症
	Carnitine Palmitoyltransferase IB (muscle) Deficiency	*CPY1B*			
	Carnitine Palmitoyltransferase II Deficiency	*CPT2*			
32	**Carnitine-Acylcarnitine Translocase Deficiency**	*SLC25A20*	小児科	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY	カルニチン−アシルカルニチントランスロカーゼ 欠損症
33	**Cataract**		眼科	anteriorpolar cataract	先天極白内障
	Cataract, Anterior Polar	*CTAA1,CTAA2*	眼科	congenital cataract	先天白内障
	Cataract, Crystalline Aculeiform	*CRYGD*	眼科	pulverulent zonular cataract	先天層間白内障
	Cataracts, Autosomal Dominant	*FOXE3,PITX3*			
	Peters Anomaly with Cataract	*FOXE3,PITX2,PITX3,PAX6*			
	Poikiloderma Atrophicans and Cataract [Rothmund-Thomson Syndrome]	*RECQL4*			
	Zonular Pulverulent Cataract	*GJA8*			
34	**Cerebrotendinous Xanthomatosis**	*CYP27A1*	小児科	Cerebrotendinous Xanthomatosis	脳腱黄色腫症
35	**CFTR-Related Disorders**	*CFTR*	泌尿器科	absent vas deferens	精管欠損症
	Congenital Bilateral Absence of the Vas Deferens		小児科	Cystic fibrosis	嚢胞性線維症
	Cystic Fibrosis				
36	**Charcot-Marie-Tooth Disease**		神経内科 小児科	Charcot-Marie-Tooth disease	シャルコ-マリ-トゥース病
	Charcot-Marie-Tooth Neuropathy Type 1		小児科	Dejerine-Sottas disease(Charcot-Marie-Tooth disease, 1A/1B/4)	デジャリン−ソタス病
	Charcot-Marie-Tooth Disease Type 1A	*PMP22*			
	Charcot-Marie-Tooth Disease Type 1B	*MPZ*			
	Charcot-Marie-Tooth Disease Type 1C	*LITAF*			
	Charcot-Marie-Tooth Disease Type 1D	*EGR2*			
	Charcot-Marie-Tooth Disease Type 1E	*PMP22*			
	Charcot-Marie-Tooth Disease, Axonal Type [Charcot-Marie-Tooth Neuropathy Type 2]				
	Charcot-Marie-Tooth Neuropathy Type 2A	*KIF1B,MFN2*			
	Charcot-Marie-Tooth Neuropathy Type 2B	*RAB7*			
	Charcot-Marie-Tooth Neuropathy Type 2B1	*LMNA*			
	Charcot-Marie-Tooth Neuropathy Type 2D	*GARS*			
	Charcot-Marie-Tooth Neuropathy Type 2E/1F	*NEFL*			
	Charcot-Marie-Tooth Neuropathy Type 2F	*HSPB1*			
	Charcot-Marie-Tooth Disease, X-Linked [Charcot-Marie-Tooth Neuropathy Type X]	*GJB1*			
	Charcot-Marie-Tooth Neuropathy Type 4				
	Charcot-Marie-Tooth Neuropathy Type 4A	*GDAP1*			
	Charcot-Marie-Tooth Neuropathy Type 4B1	*MTMR2*			
	Charcot-Marie-Tooth Neuropathy Type 4B2	*MTMR13*			
	Charcot-Marie-Tooth Neuropathy Type 4C	*KIAA1985*			

	Charcot-Marie-Tooth Neuropathy Type 4D	*NDRG1*			
	Charcot-Marie-Tooth Neuropathy Type 4E	*EGR2*			
	Charcot-Marie-Tooth Neuropathy Type 4F	*PRX*			
37	**Chediak-Higashi Syndrome**	*CHS1*	小児科	Chediak-Higashi syndrome	チェディアク-東 症候群
38	**Chondrodysplasia**		小児科	Blomstrand chondrodysplasia	ブロムストランド軟骨異形成症
	Rhizomelic Chondrodysplasia Punctata Spectrum		小児科	metaphyseal chondrodysplasia	骨幹端軟骨異形成症
	Rhizomelic Chondrodysplasia Punctata Type 1	*PEX7*	小児科	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1	肢根型点状軟骨異形成症 1型
	Rhizomelic Chondrodysplasia Punctata Type 2	*GNPAT*	小児科	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2	肢根型点状軟骨異形成症 2型
	Rhizomelic Chondrodysplasia Punctata Type 3	*AGPS*	小児科	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3	肢根型点状軟骨異形成症 3型
	Chondrodysplasia Punctata, X-Linked Dominant	*EBP*	小児科	Cartilage-Hair hypoplasia	軟骨毛髪低形成症
	Chondrodysplasia, Blomstrand Type	*PTHR1*			
	Metaphyseal Chondrodysplasia, McKusick Type [**Cartilage-Hair Hypoplasia**]	*RMRP*			
	Metaphyseal Chondrodysplasia, Schmid Type	*COL10A1*			
	Pierre Robin Syndrome with Fetal Chondrodysplasia [Weissenbacher-Zweymuller Syndrome]	*COL11A2*			
39	**Chorea**		神経内科	familial chorea	家族性舞踏病
	Benign Hereditary Chorea	*TITF1*			
	Mitochondrial Disorders				
	Chorea and Dementia	*MTTW*			
	Huntington Chorea [Huntington Disease]	*HD*			
	Chorea-Acanthocytosis [Choreoacanthocytosis]	*VPS13A*			
40	**Choroideremia**	*CHM(=REP1)*	眼科	choroideremia	脈絡膜欠如症
41	**Chronic Granulomatous Disease**	*CYBA,CYBB,NCF1,NCF2*	小児科	chronic granulomatous disease	慢性肉芽腫症
254	**Chronic Infantile Neurological Cutaneous and Articular Syndrome**	*CIAS1*	小児科	Chronic Infantile Neurological Cutaneous and Articular (CINCA) syndrome	CINCA症候群
42	**Cockayne Syndrome**	*CKN1,ERCC6*	小児科	Cockayne syndrome	コケイン症候群
43	**Coffin-Lowry Syndrome**	*RPS6KA3(=RSK2)*	小児科	Coffin-Lowry syndrome	コフィン―ローウリィ症候群
44	**Cohen Syndrome**	*COH1*	小児科	Cohen syndrome	コーヘン症候群
45	**Congenital Adrenal Hyperplasia**		第二内科	11beta-hydroxylase deficiency	11β-ヒドロキシラーゼ欠損症
	11 beta Hydroxylase Deficiency	*CYP11B1*	第二内科	21-hydroxylase deficiency	21-ヒドロキシラーゼ欠損症
	Congenital Adrenal Hyperplasia, 21-Hydroxylase Deficient [**21-Hydroxylase Deficiency**]	*CYP21A2*	小児科	congenital adrenal hyperplasia	先天性副腎皮質過形成症
	Lipid Congenital Adrenal Hyperplasia [Cholesterol Desmolase Deficiency]	*STAR*			
46	**Congenital Adrenal Hypoplasia [X-Linked Adrenal Hypoplasia Congenita]**	*NROB1*	小児科	congenital adrenal hypoplasia	先天性副腎低形成症
	Complex Glycerol Kinase Deficiency				
	Isolated X-Linked Adrenal Hypoplasia Congenita				
47	**Congenital Disorders of Glycosylation**		小児科	CARBOHYDRATE-DEFICIENT GLYCOPROTEIN SYNDROME, TYPE Ib	先天性糖化異常症 Ib型
	Carbohydrate-Deficient Glycoprotein Syndrome, Type Ia [Congenital Disorder of Glycosylation Ia]	*PMM2*	小児科	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ia	先天性糖化異常症 Ia型
	Carbohydrate-Deficient Glycoprotein Syndrome, Type Ib [Congenital Disorder of Glycosylation Ib]	*MPI*	小児科	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Ic	先天性糖化異常症 Ic型
	Carbohydrate-Deficient Glycoprotein Syndrome, Type V [Congenital Disorder of Glycosylation Ic]	*ALG6*	小児科	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE Id	先天性糖化異常症 Id型
	Congenital Disorder of Glycosylation Id	*ALG3*			
	Congenital Disorder of Glycosylation Ie	*DPM1*			
	Congenital Disorder of Glycosylation If	*MPDU1*			
	Congenital Disorder of Glycosylation Ig	*ALG12*			
	Congenital Disorder of Glycosylation Ih	*ALG8*			
	Congenital Disorder of Glycosylation Ii	*ALG2*			
48	**Congenital Muscular Dystrophy**		神経内科	congenital muscular dystrophy	先天性筋ジストロフィー
	Congenital Muscular Dystrophy Type 1C	*FRKP*			
	Congenital Muscular Dystrophy with Early Spine Rigidity	*SEPN1*			
	Congenital Muscular Dystrophy with Merosin Deficiency	*LAMA2*			
	Fukuyama Congenital Muscular Dystrophy	*FCMD*			
	Muscle-Eye-Brain Disease	*MEB*			
	Walker-Warburg Syndrome	*POMT1*			
49	**Congenital Neutropenia**		小児科	congenital neutropenia	先天性好中球減少症
	ELA2-Related Neutropenia	*ELA2*	小児科	Wiskott-Aldrich syndrome	ウィスコット-オルドリッヒ症候群
	Cyclic Neutropenia				
	Severe Congenital Neutropenia				
	WAS-Related Disorders	*WAS*			
	Wiskott-Aldrich Syndrome				
	X-Linked Severe Congenital Neutropenia				
	X-Linked Thrombocytopenia				
50	**Corneal Dystrophy**		眼科	latticedystrophy（GroenouwtypeIII）	格子状角膜変性
	Lattice Corneal Dystrophy Type I	*TGFBI*			
	Granular Corneal Dystrophy	*TGFBI*			
	Keratosis Palmoplantaris with Corneal Dystrophy [Tyrosinemia Type II]	*TAT*			
	Avellino Corneal Dystroph	*TGFBI*			

	Corneal Dystrophy, Gelatinous Drop-Like	*M1S1*			
51	**Cystathioninuria**	*CTH*	小児科	CYSTATHIONINURIA	シスタチオニン尿症
52	**Cystinosis**	*CTNS*	小児科	CYSTINOSIS	シスチノーシス
	Intermediate Cystinosis				
	Nephropathic Cystinosis				
	Non-Nephropathic Cystinosis				
53	**Cystinuria**	*SLC3A1, SLC7A9*	泌尿器科 小児科	Cystinuria	シスチン尿症
54	**Deafness**		耳鼻咽喉科	congenital deafness(autosomal dominant)	先天性難聴(優性遺伝性)
	Autosomal Dominant Nonsyndromic Sensorineural Deafness [Nonsyndromic Low-Frequency Sensorineural Hearing Lo	*WFS1*	耳鼻咽喉科	congenital deafness(autosomal recessibe)	先天性難聴(劣性遺伝性)
	Autosomal Dominant Nonsyndromic Sensorineural Deafness 17 [DFNA 17]	*MYH9*	耳鼻咽喉科	congenital deafness(X linked)	先天性難聴(X連鎖性)
	Chondrodystrophy with Sensorineural Deafness [Otospondylomegaepiphyseal Dysplasia]	*COL11A2*	神経内科	hereditary deafness	遺伝性難聴
	Deafness and Mutilating Keratoderma [Vohwinkel Syndrome]	*GJB2*	小児科　耳鼻咽喉科	nonsyndromic sensorineural deafness	非症候性感音性難聴
	Deafness-Dystonia-Optic Neuronopathy Syndrome	*TIMM8A*			
	Diabetes, Optic Atrophy, Deafness [Wolfram Syndrome]	*WFS1*			
	Hereditary Hearing Loss and Deafness				
	Nonsyndromic Hearing Loss and Deafness				
	Nonsyndromic Hearing Loss and Deafness, Autosomal Dominant				
	DFNA 3 Nonsyndromic Hearing Loss and Deafness				
	GJB2-Related DFNA 3 Nonsyndromic Hearing Loss and Deafness	*GJB2*			
	GJB6-Related DFNA 3 Nonsyndromic Hearing Loss and Deafness	*GJB6*			
	DFNA 9 Nonsyndromic Hearing Loss and Deafness (COCH) [DFNA 9 (COCH)]	*COCH*			
	Nonsyndromic Hearing Loss and Deafness, Autosomal Recessive				
	DFNB 1 Nonsyndromic Hearing Loss and Deafness				
	GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness	*GJB2*			
	GJB6-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness	*GJB6*			
	DFNB 4 Nonsyndromic Hearing Loss and Deafness [DFNB 4]	*SLC26A4*			
	Nonsyndromic Hearing Loss and Deafness, Mitochondrial				
	MTRNR1-Related Hearing Loss and Deafness	*MTRNR1*			
	MTTS1-Related Hearing Loss and Deafness	*MTTS1*			
	Nonsyndromic Hearing Loss and Deafness, X-Linked				
	DFN 3 Nonsyndromic Hearing Loss and Deafness [DFN 3]	*POU3F4*			
	Keratitis-Ichthyosis-Deafness Syndrome, Autosomal Dominant	*GJB2*			
	Macrothrombocytopathy, Nephritis, and Deafness [Epstein Syndrome]	*MYH9*			
	Macrothrombocytopathy, Nephritis, Deafness, and Leukocyte Inclusions [Fechtner Syndrome]	*MYH9*			
	Renal Tubular Acidosis with Progressive Nerve Deafness	*ATP6V1B1*			
	Gitelman Syndrome	*SCL12A3*			
	Urticaria-Deafness-Amyloidosis Syndrome [Muckle-Wells Syndrome]	*CIAS1*			
55	**Dentatorubral-Pallidoluysian Atrophy [DRPLA]**	*DRPLA*	小児科	DENTATORUBRAL-PALLIDOLUYSIAN ATROPHY; DRPLA	歯状核赤核淡蒼球ルイ体萎縮症
56	**Diabetes Insipidus**		小児科　第二内科	DIABETES INSIPIDUS, NEPHROGENIC, AUTOSOMAL DOMINANT	腎性尿崩症、常染色体優性(2型)
	Nephrogenic Diabetes Insipidus		小児科　第二内科	DIABETES INSIPIDUS, NEPHROGENIC, X-LINKED	腎性尿崩症、X連鎖性(1型)
	Nephrogenic Diabetes Insipidus, Autosomal	*AQP2*	小児科　第二内科	DIABETES INSIPIDUS, NEUROHYPOPHYSEAL TYPE	中枢性尿崩症(神経下垂体型)、家族性中枢性尿崩症
	Nephrogenic Diabetes Insipidus, X-Linked	*AVPR2*			
	Neurohypophyseal Diabetes Insipidus	*AVP*			
57	**Diabetes Mellitus**		第二内科	IABETES MELLITUS, INSULIN-RESISTANT, WITH ACANTHOSIS NIGRIC	黒色表皮症合併インスリン抵抗性糖尿病,妖精症
	Diabetes Mellitus with Acanthosis Nigricans and Hypertension	*PPARG*			
	Diabetes Mellitus, MODY Type 1 [Maturity-Onset Diabetes of the Young Type I]	*HNF4A*			
	Diabetes Mellitus, MODY Type 2 [Maturity-Onset Diabetes of the Young Type II]	*GCK*			
	Diabetes Mellitus, MODY Type 3 [Maturity-Onset Diabetes of the Young Type III]	*TCF1*			
	Diabetes Mellitus, MODY Type 4 [Maturity-Onset Diabetes of the Young Type IV]	*IPF1*			
	Diabetes Mellitus, MODY Type 5 [Maturity-Onset Diabetes of the Young Type V]	*TCF2*			
	Diabetes Mellitus, MODY Type 6 [Maturity-Onset Diabetes of the Young Type VI]	*NEUROD1*			
58	**Diamond-Blackfan Anemia**	*RPS19*	小児科	Diamond-Blackfan syndrome	ダイアモンドーブラックファン症候群
59	**DiGeorge Syndrome**		小児科	DiGeorge syndrome	ディジョージ症候群
	22q11.2 Deletion Syndrome	DGCR (CRITICAL REGION)			
	10p13-p14 Deletion	DGS2,VS2 (CRITICAL REGION)			
60	**2,4-Dienoyl-CoA Reductase Deficiency**	*DECR*	小児科	2,4-Dienoyl-CoA reductase	2,3-ジエノイル-CoAレダクターゼ欠損症
61	**Dilated Cardiomyopathy**		第三内科	Dilated Cardiomyopathy	家族性拡張型心筋症
	Dilated Cardiomyopathy				
	DMD-Related Dilated Cardiomyopathy	*DMD*			
	LMNA-Related Dilated Cardiomyopathy	*LMNA*			
	TAZ-Related Dilated Cardiomyopathy	*TAZ*			
	Dilated Cardiomyopathy with Quadriceps Myopathy	*LMNA*			
62	**Down Syndrome Critical Region**	*DSCR1*	小児科	Down syndrome	ダウン症候群

63	**Duchenne/Becker Muscular Dystrophy**	*DMD*	神経内科 小児科	Duchenne / Becker muscular dystrophy	デュシェンヌ／ベッカー型筋ジストロフィー
	Becker Muscular Dystrophy				
	Duchenne Muscular Dystrophy				
64	**Dystonia**		小児科	DYSTONIA, PROGRESSIVE	遺伝性進行性ジストニー（瀬川病）
	Early-Onset Primary Dystonia (DYT1)	*DYT1*	神経内科	familial dystonia syndrome	家族性ジストニア症候群
	Dystonia 4	DTY4 (LOCUS)			
	Dystonia 6	*DTY6*			
	Dystonia 7	DTY7 (LOCUS)			
	Dystonia11	*SGCE*			
	Dopa-Responsive Dystonia [Dystonia 5][Segawa Disease]				
	GTP Cyclohydrolase 1-Deficient DRD	*GCH1*			
	Tyrosine Hydroxylase-Deficient DRD	*TH*			
65	**Ehlers-Danlos Syndrome**		第一外科 小児科	Ehlers-Danlos syndrome	エラス-ダンロス症候群
	Ehlers-Danlos Syndrome, Arthrochalasia Type	*COL1A1,COL1A2*			
	Ehlers-Danlos Syndrome, Classic Type	*COL5A1,COL5A2*			
	Ehlers-Danlos Syndrome Type I				
	Ehlers-Danlos Syndrome Type II				
	Ehlers-Danlos Syndrome, Kyphoscoliotic Form	*PLOD*			
	Ehlers-Danlos Syndrome, Vascular Type	*COL3A1*			
66	**Emery-Dreifuss Muscular Dystrophy**		小児科	Emery-Dreifess type muscular dystrophy	エメリーードレイフェス型筋ジストロフィー症
	Emery-Dreifuss Muscular Dystrophy, Autosomal		神経内科	Emery-Dreifuss muscular dystrophy	エメリー型筋ジストロフィー
	Emery-Dreifuss Muscular Dystrophy, Autosomal Dominant	*LMNA*			
	Emery-Dreifuss Muscular Dystrophy, X-Linked	*EMD*			
67	**Epidermolysis Bullosa**		皮膚科	epidermolysis bullosa	表皮水疱症
	Epidermolysis Bullosa Dystrophica, Bart Type	*COL7A1*			
	Epidermolysis Bullosa Dystrophica, Cockayne-Touraine Type	*COL7A1*			
	Epidermolysis Bullosa Dystrophica, Hallopeau-Siemens Type	*COL7A1*			
	Epidermolysis Bullosa Dystrophica, Pasini Type	*COL7A1*			
	Epidermolysis Bullosa Junctional, Disentis Type	*COL7A1,LAMB3*			
	Epidermolysis Bullosa Junctional, Herlitz-Pearson Type	*LAMB3,LAMA3,LAMC2*			
	Epidermolysis Bullosa Letalis with Pyloric Atresia	*ITGA6,ITGB4*			
	Epidermolysis Bullosa Simplex	*KR14,KRT5*			
	Epidermolysis Bullosa Simplex with Mottled Pigmentation				
	Epidermolysis Bullosa Simplex, Dowling-Meara Type				
	Epidermolysis Bullosa Simplex, Koebner Type				
	Epidermolysis Bullosa Simplex, Weber-Cockayne Type				
	Epidermolysis Bullosa with Muscular Dystrophy	*PLEC1*			
	Epidermolysis Bullosa, Pretibial	*COL7A1*			
251	**Epilepsy**		神経内科	Epilepsy	てんかん
	Generalized Epilepsy with Febrile Seizures Plus	*SCN1A, SCN1B, GABRG2, SCN2A*		Generalized Epilepsy with Febrile Seizures Plus	熱性痙攣プラス
68	**Episodic Ataxia**		神経内科	Episodic Ataxia	周期性失調症
	Episodic Ataxia Type 1	*KCNA1*	小児科	Familial episodic ataxia	発作性運動失調症
	Episodic Ataxia Type 2	*CACNA1A,CACNB4*			