FirmaLab
Bio-Diagnostics

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The following pages are informational, and do not have to be returned with specimen.

GENETIC TEST MENU

Tests are organized in four tables (Carrier, Specific Exon, Diagnostic Panels, and Single Gene).

Carrier Screening Tests

Carrier screening tests are used for asymptomatic individuals with or without family history of the disease.

Test No.	Test Name	Test Description
10	CYP450 Panel	Sequence of CYP2C9, CYP2C19, and CYP2D6 genes
15	ME Panel	Middle-East Carrier Screening Panel (Wolman, HIBM, Usher, Dubin Johnson, G6PD deficiency, founder and common mutations)
16	ME Panel w/ MTHFR	Middle-East Carrier Screening Panel (Wolman, HIBM, Usher, Dubin Johnson, G6PD deficiency, MTHFR, founder and common mutations)
25	AIRE p.Y85C	Polyglandular Syndrome, Autoimmune (APS I)
28	BCHE p.D70G	Pseudocholinesterase Deficiency
30	CYP11B2 p.R181W, p.V386A	Congenital hypoaldosteronism, Corticosterone methyl oxidase II (CMO II) deficiency
35	mtDNA analysis	Sequence of hypervariable regions I, II, and III. These are linked to ancestry.
40	GNE p.M712T	Hereditary Inclusion Body Myopathy type 2 (HIBM, IBM2), Distal Myopathy with Rimmed Vacuoles (DMRV), GNE Myopathy, Middle-East founder mutation
41	GNE p.V572L	Hereditary Inclusion Body Myopathy type 2 (HIBM, IBM2), Distal Myopathy with Rimmed Vacuoles (DMRV), GNE Myopathy, Asian founder mutation
50	G6PD p.S188F	G6PD deficiency (G6PD) Middle-East common mutation
65	LAL p.G87V	Wolman Disease (LAL/LIPA), Iranian-Jewish founder mutation, LAL p.G87V (G66V based on Anderson, et.al. 1991).
70	MRP2 p.I1173F	Dubin Johnson Syndrome, DJS (MRP2/CMOAT/ABCC2), Middle-East founder mutation
80	MTHFR p.A222V	MTHFR thermolabile variant (MTHFR), Worldwide, MTHFR p.A222V (677C>T)
85	Oxidation Panel	Oxidation Panel (SOD1, CBS, COMT, MAOA)
90	USH2A p.T80fsX28	Usher Syndrome (USH2A), Middle-East founder mutation

On a case by case basis, reserved for patients in significant financial hardship, we are able to provide discounted or possibly pro-bono testing services. Pro-bono services are limited to fewer than 10% of specimens received per month.

Specific Exon Sequencing

Specific Exon sequencing is used to confirm a previously recorded mutation, or test only for specific founder/common mutations in a patient of high risk (e.g. with positive family history and known mutation, or member of high risk population).

Test No.	Test Name	Test Description
1	Single Exon	Sequencing of any single exon in the gene of interest
2	Two Exons	Sequencing of any two exons in the gene of interest
3	Three Exons	Sequencing of any three exons in the gene of interest

Diagnostic Sequencing Tests (Panel table are followed by single gene table)

Diagnostic testing panels and Single Gene sequencing are requested to confirm diagnosis of symptomatic patients. To discover common mutations rapidly and cost effectively, tests are performed in sequential, tiered, or stepwise bases when possible (e.g. the genes or exons known to be responsible for common mutations are sequenced first). Testing is stopped once the likely responsible mutation is discovered.

Test No.	Test Name	Disease Description
100	ALS Panel	Amyotrophic Laterial Sclerosis (ALS) Sequencing Panel (SOD1, FUS/TLS, TARDBP, ANG, VAPB)

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110	ARVD/C Panel	Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia, Sequential Panel (DSG2, DSP, PKP2, DSC2, RYR2, TMEM43, TGFB3, JUP)
520	BBS Panel	Bardet-Biedl Syndrome Sequential Panel (BBS1, BBS10, BBS2, MKKS/BBS6, BBS12, BBS4, BBS7, TTC8/BBS8, BBS5, ARL6/BBS3, BBS9, TRIM32/BBS11)
530	Bernard-Soulier Panel	Bernard-Soulier Sequential Panel (GP1BA, GP1BB, GP9)
445	Bethlem Panel	Bethlem Myopathy (COL6A1, COL6A2, COL6A3)
540	CFC Panel	Cardio-Facio-Cutaneous (CFC) Syndrome Sequential Panel (BRAF, MEK1, MEK2, KRAS)
490	CCM Panel	Cerebral Cavernous Malformations (CCM) Sequential Panel (KRIT1, CCM2/MGC4607, CCM3/PDCD10)
470	CMT1 Panel	Charcot-Marie-Tooth Type 1 (CMT1) Sequential Panel (PMP22, MPZ, LITAF, EGR2, NEFL)
471	CMT2 Panel	Charcot-Marie-Tooth Type 2 (CMT2) Sequential Panel (MFN2, GDAP1, NEFL, GARS, LMNA, YARS, KIF1B, RAB7A, MED25, TRPV4, HSPB1, MPZ, HSPB8, AARS, DNM2)
474	CMT4 Panel	Charcot-Marie-Tooth Type 4 (CMT4), recessive, Sequential Panel (GDAP1, MTMR2, SBF2, SH3TC2, NDRG1, EGR2, PRX, FGD4, FIG4)
477	CMTx Panel	Charcot-Marie-Tooth Other, Sequential Panel (GARS, HSPB1, HSPB8, LMNA, DNM2, YARS, MPZ)
201	CDG Panel 1	Congenital Disorders of Glycosylation (CDG), Sequential Panel (PMM2, ALG6, MPI, SRD5A3)
202	CDG Panel 2	Congenital Disorders of Glycosylation (CDG), Sequential Panel (RFT1, ALG3, ALG12, ALG8, ALG1/HMT-1, MAGT1, MGAT2, ALG2, ALG9)
203	CDG Panel 3	Congenital Disorders of Glycosylation (CDG), Sequential Panel (DOLK/DK1, DPM3, ALG11, MOGS/GCS1, SLC35C1, B4GALT1, SLC35A1, COG1, COG4, COG5, COG6, COG7, COG8)
430	CMS Panel	Congenital Myasthenic Syndrome (CMS) Panel (CHAT, CHRNE, COLQ, RAPSN, CHRNA1, CHRNB1, MUSK, CHRND, DOK7)
220	CGL Panel	Congenital Generalized Lipodystrophy Sequential Panel (AGPAT2, BSCL20)
10	CYP450 Panel	Pharmacogenomic Panel (CYP2C9, CYP2C19, CYP2D6)
310	DCM Panel	Dilated Cardiomyopathy Sequential Panel (LMNA, MYH7, TNNT2, SCN5A, MYH7, ACTC1, TTN,
240	Distal Arth. Panel	Distal Arthrogryposis Syndrome Sequential Panel (MYH3, TPM2, TNNI2, TNNT3)
440	Distal Myopathy Panel	Distal Myopathy Sequential Panel (DYSF, GNE, TTN, CAV3. MYH7, MATR3, MYOT, NEB, LDB3, ANO5, KLHL9, DNM2)
420	DG Panel	Dystroglycanopathy Sequential Panel (DAG1, FKRP, FKTN, POMT1, POMT2, POMGNT1, LARGE)
270	FHL Panel	Familial Hemophagocytic Lymphohistiocytosis (FHL) Panel (PRF1, UNC13D//MUNC13-4, STX11, STXBP2, RAB27A)
280	Fanconi Panel	Fanconi Anemia Sequential Panel (FANCA, FANCC, FANCG, FANCE, FANCF, FANCJ/BRIP1, FANCL, FANCM, FANCN/PALB2, RAD51C, SLX4)
290	HPE Panel	Holoprosencephaly (HPE), Autosomal Dominant, Nonsyndromic, Sequential Testing (SHH, ZIC2, SIX3, TGIF1, PTCH1, GLI)
310	HCM Panel	Hypertrophic Cardiomyopathy (HCM) Sequential Panel (MYH7, MYBPC3, TNNT2, TNNI3, TPM1, MYL2, MYL3, ACTC1, CSRP3, TTN, ACTN2, MYH6, TCAP, TNNC1)
410	LGMD Panel	Limb Girdle Muscular Dystrophy Autosomal Recessive Panel (CAPN3, DYSF, SGCG, SGCA, SGCB, SGCE, TCAP, TRIM32, FKRP, TTN, TMEM16E/ANO5)
220	Joubert Panel	Joubert Syndrome Sequential Panel (AHI1, CEP290, TMEM67, RPGRIP1L, ARL13B, CC2D2A, CXORF5, TMEM67)
225	MKS Panel	Meckel-Gruber Syndrome (MKS) Sequencing Panel (MKS1, TMEM67/MKS3, CEP290, RPGRIP1L)
230	ARSA, PSAP	Metachromatic Leukodystrophy Sequential Panel (ARSA, PSAP)
235	MMA Panel	Methylmalonic Acidemia IMMA) Sequential Panel (MMAA, MMAB, MUT, MMACHC, MCEE)

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240	MED Panel	Multiple Epiphyseal Dysplasia (MED) Sequential Panel (COMP, MATN3, SLC26A2, COL9A1, COL9A2, COL9A3)
242	Myofibrillar Panel	Myofibrillar Myopathy (MFM) Sequential Panel (BAG3, CRYAB, DES, FLNC, LDB3, MYOT)
245	Nemaline Panel	Nemaline Myopathy Sequential Panel (ACTA1, NEB, NEBx55 Del, TPM2, TPM3, TNNT1, CFL2)
250	Noonan Panel	Noonan Syndrome Sequential Panel (PTPN11, SOS1, RAF1, KRAS)
85	Oxidation Panel	Oxidation Panel (SOD1, CBS, COMT, MAOA)
255	PCD Panel	Primary Ciliary Dyskinesia (PCD) Sequential Panel (DNAH5/PCD3, DNAI1/PCD1, TXNDC3/PCD6, DNAI2/PCD9, C14orf104, DNAH11, RSPH4A, RSPH9)
260	SCN Panel	Severe Congenital Neutropenia (SCN) Sequential Panel (ELANE, HAX1, G6PC3, GFI1, WAS)
265	SCDO Panel	Spondylocostal Dysostosis (SCDO) Sequential Panel (DLL3, MESP2, LFNG, HES7)
270	Stuttering Panel	Stuttering Sequential Panel (GNPTAB, GNPTG, NAGPA)
300	Usher 1 Panel	Usher Syndrome type 1 Sequential Sequencing (MYO7A, CDH23, PCDH15, USH1C)
301	Usher 2 Panel	Usher Syndrome type 2 Sequential Sequencing (USH2A, GPR98, DFNB31)

Single gene sequencing

Test No.	Test Name	Disease Description
1000	ABCA4	Stargardt Disease, Autosomal Recessive
1003	ABCC9	Dilated Cardiomyopathy
1006	ACADM	Medium Chain Acyl-CoA Dehydrogenase Deficiency
1009	ACADS	Short Chain Acyl-CoA Dehydrogenase Deficiency
1012	ACADVL	Very Long Chain Acyl-CoA Dehydrogenase Deficiency
1015	ACTA1	alpha-Actin (Skeletal Muscle Form)-Related Myopathies
1018	ACTC1	Hypertrophic Cardiomyopathy and Related Disorders
1021	ACTN2	Hypertrophic Cardiomyopathy
1024	ADAMTS13	Thrombotic Thrombocytopenia Purpura (TTP)
1027	ADSL	Adenylosuccinase Gene Sequencing
1030	AGL	Glycogen Storage Disease, Type III
1033	AGPAT2	Congenital Generalized Lipodystrophy, Type 1
1036	AGRN	Familial Limb Girdle Myasthenic Syndrome
1039	AGRN	Familial Limb Girdle Myasthenic Syndrome Panel
1042	AHI1	Joubert Syndrome
1045	ALG12	Congenital Disorders of Glycosylation, Type Ig
1048	ALG2	Congenital Disorders of Glycosylation, Type Ii
1048	ALG2	Panel 2: ALG3, DPM1, MPDU1, ALG12, ALG8, ALG2
1051	ALG3	Congenital Disorders of Glycosylation, Type Id
1051	ALG3	Panel 2: ALG3, DPM1, MPDU1, ALG12, ALG8, ALG2
1054	ALG6	Congenital Disorders of Glycosylation, Type Ic
1054	ALG6	Panel 1: PMM2, MPI, ALG6
1057	ALG8	Congenital Disorders of Glycosylation, Type Ih
1060	ALMS1	Alstrom Syndrome
1063	ALS2	Juvenile Amyotrophic Lateral Sclerosis
1066	AN05/TMEM16E	Limb Girdle Muscular Dystrophy, Type 2L (LGMD2L) and Distal Miyoshi Myopathy (MMD3)
1069	ANG	Amyotrophic Lateral Sclerosis, Autosomal Dominant or Sporadic
1072	AP3B1	Hermansky-Pudlak Syndrome Type 2 (HPS2)
1075	APC	Familial Adenomatous Polyposis
1078	AR	Androgen Insensitivity Syndrome
1081	ARG1	Argininemia
1084	ARL13B	Joubert Syndrome

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1087	ARL6/BBS3	Bardet-Biedl Syndrome
1090	ARSA	Metachromatic Leukodystrophy
1093	ARSB	Mucopolysaccharidosis Type VI / Maroteaux-Lamy Syndrome
1096	ARX	X-Linked Lissencephaly-2
1099	ASAH1	Farber Lipogranulomatosis
1102	ASS1	Citrullinemia, Type I
1105	ATP7A	Menkes Disease and Hereditary Motor Neuropathy, X-Linked
1108	ATP7B	Wilson Disease/Hepatolenticular Degeneration
1111	AUH	3-Methylglutaconic Aciduria Type I
1114	AURKC	Male Infertility with Large-Headed Spermatozoa
1117	AXIN2	Oligodontia-Colorectal Cancer
1120	BAG3	Childhood Onset Muscular Dystrophy with Myofibrillar Myopathy
1123	BBS1	Bardet-Biedl Syndrome
1126	BBS10	Bardet-Biedl Syndrome
1129	BBS11/TRIM32	Bardet-Biedl Syndrome
1132	BBS12	Bardet-Biedl Syndrome
1135	BBS2	Bardet-Biedl Syndrome
1138	BBS3/ARL6	Bardet-Biedl Syndrome
1141	BBS4	Bardet-Biedl Syndrome
1144	BBS5	Bardet-Biedl Syndrome
1147	BBS6/MKKS	Bardet-Biedl Syndrome
1150	BBS7	Bardet-Biedl Syndrome
1153	BBS8/TTC8	Bardet-Biedl Syndrome
1156	BBS9	Bardet-Biedl Syndrome
1159	BCKDHA	Maple Syrup Urine Disease Type IA
1162	BCKDHB	Maple Syrup Urine Disease Type IB
1165	BLM	Bloom's Syndrome
1168	BLOC1S3	Hermansky-Pudlak Syndrome Type 8 (HPS8)
1171	BMP15	Ovarian Dysgenesis 2
1174	BMPR1A	Juvenile Polyposis Syndrome (JPS)
1177	BMPR2	Primary Pulmonary Hypertension (PPH1)/Pulmonary Arterial Hypertension (PAH)
1180	BRAF	Cardio-Facio-Cutaneous (CFC) Syndrome
1183	BRIP1/FANCJ	Fanconi Anemia
1186	BSCL2	Seipin-Related Disorders
1189	BTD	Multiple Carboxylase Deficiency (Juvenile Onset)
1192	C14orf104/KTU	Primary Ciliary Dyskinesia (PCD)
1195	CAPN3	Limb Girdle Muscular Dystrophy, Type 2A
1195	CAPN3	Myositis, eosinophilic
1198	CAV3	Caveolinopathy Testing
1201	CC2D2A	Joubert and Meckel-Gruber Syndromes
1201	CC2D2A	Meckel-Gruber Syndrome Sequencing Panel
1204	CCBE1	Hennekam Lymphangiectasia-Lymphedema Syndrome
1207	KRIT1/CCM1	Cerebral Cavernous Malformations "Common Hispanic" Mexican Mutation (Exon 10)
1207	KRIT1/CCM1	Cerebral Cavernous Malformations
1208	CBS	Cystathionine Bata-Synthase
1210	CCM2	Cerebral Cavernous Malformations 2 (CCM2)
1211	CCM2-Exon2-10 deletion	Cerebral Cavernous Malformations 2 (CCM2). Exon 2-10 deletion
1213	CCM3/PDCD10	Cerebral Cavernous Malformations 3 (CCM3)
1216	CDH23	Usher Syndrome Type 1
1219	CDKN1B	Multiple Endocrine Neoplasia Type 1
1222	CEP290	Joubert Syndrome
1222	CEP290	Meckel-Gruber Syndrome
1225	CFL2	Nemaline Myopathy 7 Skeletal Muscle Cofilin-2 Sequencing

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1228	CFTR	Cystic Fibrosis and CBAVD Testing
1231	CHAT	Congenital Myasthenic Syndrome Panel
1231	CHAT	Congenital Myasthenic Syndrome with Episodic Apnea
1234	CHD7	CHARGE Syndrome
1237	CHEK2	Hereditary Breast Cancer
1240	CHRNA1	Congenital Myasthenic Syndromes and Lethal Multiple Pterygium Syndrome
1243	CHRNA1, CHRND, CHRNG	Multiple Pterygium Syndromes Panel (CHRNA1, CHRND, CHRNG)
1246	CHRNB1	Congenital Myasthenic Syndromes
1252	CHRND	Congenital Myasthenic Syndromes and Lethal Multiple Pterygium Syndromes
1252	CHRND	Multiple Pterygium Syndromes
1253	CHRNE	Congenital Myasthenic Syndrome
1254	CHRNG	Multiple Pterygium Syndromes
1257	CLRN1	Usher Syndrome Type 3
1260	CNGA1	Retinitis Pigmentosa, Autosomal Recessive
1262	CNGA3	Achromatopsia
1265	CNGB1	Retinitis Pigmentosa, Autosomal Recessive
1267	CNGB3	Achromatopsia
1270	COL18A1	Knobloch Syndrome, Type I
1273	COL2A1	Achondrogenesis Type II (ACG2)-Hypochondrogenesis
1273	COL2A1	Kniest Dysplasia
1273	COL2A1	Osteoarthritis with Mild Chondrodysplasia
1273	COL2A1	Spondyloepiphyseal Dysplasia, Congenita (SEDc) and Spondylometaepiphyseal Dysplasia, Strudwick Type (SEMD)
1273	COL2A1	Spondyloperipheral Dysplasia (SPD) and Platyspondylic Lethal Skeletal Dysplasia, Torrance Type (PLSDT)
1273	COL2A1	Stickler Syndrome, Type 1 (STL1)
1275	COL9A1	Stickler Syndrome, Autosomal Recessive
1286	COL9A2	Multiple Epiphyseal Dysplasia
1287	COL9A3	Multiple Epiphyseal Dysplasia
1280	COLQ	Congenital Myasthenic Syndrome
1290	COMP	Pseudoachondroplasia (PSACH) and Multiple Epiphyseal Dysplasia (MED)
1292	COMT	Metabolism of catechol drugs
1294	CPS1	Hyperammonemia
1297	CRB1	Leber Congenital Amaurosis and Retinitis Pigmentosa
1300	CRX	Leber Congenital Amaurosis, Type VII
1303	CRYAB	Myofibrillar Myopathy
1306	CSRP3	Hypertrophic Cardiomyopathy
1309	CTSC	Papillon-Lefevre Syndrome (PLS)
1312	CUL4B	Cabezas X-Linked Mental Retardation Syndrome
1315	CUL7	3-M Syndrome
1316	CYP2C9	Drug metabolism
1317	CYP2C19	Drug metabolism
1318	CYP2D6	Drug metabolism
1319	DAG1	Dystroglycan Testing
1321	DBT	Maple Syrup Urine Disease Type II
1324	DCTN1	Distal Hereditary Motor Neuronopathy, Type VIIB
1324	DCTN1	Dynactin-Related Disorders
1324	DCTN1	Perry Syndrome
1327	DCX	Classic lissencephaly
1330	DDR2	Spondylo-Meta-Epiphyseal Dysplasia, Short Limb-Hand Type (SMED-SL)
1333	DES	Myofibrillar Myopathy
1336	DHCR7	Smith-Lemli-Opitz Syndrome
1339	DLD	Maple Syrup Urine Disease Type III

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1342	DLL3	Spondylocostal Dysostosis
1345	DNAH11	Primary Ciliary Dyskinesia (PCD)
1348	DNAH5	Primary Ciliary Dyskinesia (PCD)
1351	DNAI1	Primary Ciliary Dyskinesia (PCD)
1354	DNAI2	Primary Ciliary Dyskinesia (PCD)
1357	DOK7	Familial Limb Girdle Myasthenia Syndrome
1360	DOK7, RAPSN	Pena-Shokeir Syndrome, Type 1 (Fetal Akinesia Deformation Sequence)
1363	DOK7, AGRN	Familial Limb Girdle Myasthenic Syndrome
1366	DPM1	Congenital Disorders of Glycosylation, Type Ie (CDG Ie)
1369	DPM3	Congenital Disorders of Glycosylation, Type Io Plus Secondary Dystroglycanopathy
1372	DSC2	Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia
1375	DSG2	Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia
1378	DSP	Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia and Related Disorders
1381	DSPP	Dentinogenesis Imperfecta (DGI) and Dentin Dysplasia (DD)
1384	DTNBP1	Hermansky-Pudlak Syndrome Type 7 (HPS7)
1387	DYSF	Limb Girdle Muscular Dystrophy, Type 2B
1390	EBP	Conradi-Hunermann Syndrome / Chondrodysplasia Punctata, X-Linked Dominant
1393	EIF2B1-EIF2B5	Leukoencephalopathy with Vanishing White Matter and Ovarian Failure / Ovarioleukodystrophy Panel (EIF2B5,2,4,3,1)
1396	ELANE	Autosomal Dominant Severe Congenital Neutropenia and Cyclic Neutropenia
1399	EMD	Emery-Dreifuss Muscular Dystrophy-1 (EDMD1)
1402	EVC, EVC2	Ellis-van Creveld Syndrome (EVC)
1405	EXT1, EXT2	Hereditary Multiple Osteochondromas (HMO)
1408	FAH	Tyrosinemia, Type I
1411	FANCA	Fanconi Anemia
1414	FANCC	Fanconi Anemia
1417	FANCE	Fanconi Anemia
1420	FANCF	Fanconi Anemia
1423	FANCG	Fanconi Anemia
1426	FANCJ/BRIP1	Fanconi Anemia
1429	FANCL	Fanconi Anemia
1432	FANCM	Fanconi Anemia
1435	FANCN/PALB2	Fanconi Anemia
1438	FBN1	Marfan Syndrome
1441	FBN2	Congenital Contractural Arachnodactyly (Beals Syndrome)
1444	FBP1	Fructose-1,6-Bisphosphatase Deficiency
1447	FGFR3	Achondroplasia
1450	FGFR3	Hypochondroplasia
1453	FGFR3	Thanatophoric Dysplasia
1456	FIG4	Amyotrophic Lateral Sclerosis, Autosomal Dominant and Sporadic
1459	FKRP	Limb Girdle Muscular Dystrophy, Type 21
1459	FKRP	Congenital Muscular Dystrophy, Type 1C
1462	FKTN	Fukuyama Congenital Muscular Dystrophy, Walker-Warburg Syndrome, Limb Girdle Muscular Dystrophy
1462	FKTN	Japanese Founder Mutation PCR
1462	FKTN	Limb Girdle Muscular Dystrophy
1462	FKTN	Walker-Warburg Syndrome
1465	FLNC	Myofibrillar Myopathy
1468	FLT4	Hereditary Lymphedema, Type I (Milroy Disease)
1471	FMO3	Trimethylaminuria via FMO3 Gene Sequencing
1474	FOXC2	Lymphedema/Distichiasis Syndrome, Hereditary Lymphedema, Type II
1477	FSHB	Male and Female Infertility
1480	FSHR	Ovarian Dysgenesis 1
1480	FSHR	Ovarian Hyperstimulation Syndrome

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1483	FUS	Amyotrophic Lateral Sclerosis / Motor Neuron Disease
1486	G6PC	Glycogen Storage Disease, Type Ia
1489	G6PC3	Severe Congenital Neutropenia
1492	GAA	Glycogen Storage Disease, Type II
1495	GALC	Krabbe Disease
1498	GALC-del	Krabbe Disease - GALC Deletion Test
1501	GALNS	Mucopolysaccharidosis Type IVA / Morquio Type A Disease
1504	GALT	Galactosemia
1507	GAMT	Creatine Deficiency Syndrome
1510	GARS	Charcot-Marie-Tooth Disease, Type 2D
1510	GARS	Distal Hereditary Motor Neuronopathy, Type V
1510	GARS	Glycyl tRNA Synthetase-Related Disorders
1513	GATA1	Thrombocytopenia
1516	GATM	Creatine Deficiency Syndrome
1519	GBA	Gaucher Disease
1522	GBE1	Glycogen Storage Disease, Type IV
1525	GCDH	Glutaric Acidemia
1528	GCH1	DOPA Responsive Dystonia
1531	GDF6	Klippel-Feil Syndrome
1534	GFI1	Severe Congenital Neutropenia
1537	GHR	Laron Syndrome / Pituitary Dwarfism II (Growth Hormone Insensitivity)
1540	GHSR	Short Stature with or without Partial Isolated Growth Hormone Deficiency
1543	GLA	Fabry Disease
1546	GLB1	GM1 Gangliosidosis and Morquio Syndrome Type B via GLB1 Gene Sequencing
1549	GLI2	Holoprosencephaly-9 (Autosomal Dominant, Nonsyndromic)
1552	GLI3	Pallister-Hall Syndrome, Greig Cephalopolysyndactyly Syndrome, Postaxial Polydactyly A, Preaxial Polydactyly IV
1555	GLIS2	Nephronophthisis
1558	GM2A	Tay-Sachs Disease AB Variant/GM2-Gangliosidosis Variant AB
1561	GNAT2	Achromatopsia
1564	GNE-Myopathy	Hereditary Inclusion Body Myopathy 2 (HIBM), Autosomal Recessive, Distal Myopathy with Rimmed Vacuoles (DMRV), Nonaka Myopathy
1567	GNE-Sialuria	Sialuria, Exon 5 Sequencing
1570	GNPTAB	Mucolipidosis II & Mucolipidosis III Alpha/Beta
1573	GNPTAB	Stuttering
1579	GNPTG	Mucolipidosis III Gamma
1579	GNPTG	Stuttering
1582	GNS	Mucopolysaccharidosis Type IIID/Sanfilippo Syndrome D
1585	GP1BA	Bernard-Soulier Syndrome
1588	GP1BB	Bernard-Soulier Syndrome
1591	GP9	Bernard-Soulier Syndrome
1594	GPC3	Simpson-Golabi-Behmel Syndrome
1597	GPR56	Bilateral Frontoparietal Polymicrogyria (BFPP)
1600	GUSB	Mucopolysaccharidosis Type VII / Sly Syndrome
1603	GYS2	Glycogen Storage Disease, Type 0
1606	HAX1	Autosomal Recessive Severe Congenital Neutropenia (Kostmann Disease)
1609	HES7	Spondylocostal Dysostosis 4 (SCDO4)
1612	HEXA	Tay-Sachs Disease
1615	HEXB	Sandhoff Disease
1618	HGSNAT	Mucopolysaccharidosis Type IIIC/Sanfilippo Syndrome C
1621	HLCS	Multiple Carboxylase Deficiency
1624	HMGCL	HMG-CoA Lyase Deficiency
1627	HPS1	Hermansky-Pudlak Syndrome Type 1
1630	HPS3	Hermansky-Pudlak Syndrome Type 3 (HPS3)

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1633	HPS4	Hermansky-Pudlak Syndrome Type 4 (HPS4)
1636	HPS5	Hermansky-Pudlak Syndrome Type 5 (HPS5)
1639	HPS6	Hermansky-Pudlak Syndrome Type 6 (HPS6)
1642	HRAS	Costello Syndrome
1645	HSD17B3	46,XY Disorder of Sex Development (DSD)
1648	HSPB1	Heat Shock 27 kDa Protein-Related Disorders
1651	HSPB8	Heat Shock 22 kDa Protein-Related Disorders
1654	HYAL1	Mucopolysaccharidosis Type IX via HYAL1 Gene Sequencing
1657	IDS	Mucopolysaccharidosis Type 2
1660	IDUA	Mucopolysaccharidosis Type 1
1663	IFT122	Cranioectodermal Dysplasia 1
1666	IGHMBP2	Spinal Muscular Atrophy with Respiratory Distress Type 1
1669	IMPDH1	Retinitis Pigmentosa (Autosomal Dominant, Nonsyndromic)
1672	INPP5E	Ciliopathy
1675	INVS	Nephronophthisis
1678	IQCB1/NPHP5	Nephronophthisis and Senior-Loken Syndrome
1681	ITGA7	Integrin Alpha 7-Related Congenital Myopathy Testing
1684	IVD	Isovaleric Acidemia
1687	JAG1	Alagille Syndrome
1690	JUP	Palmoplantar Keratoderma with Arrhythmogenic Right Ventricular Cardiomyopathy and Woolly Hair/Naxos Disease
1693	KIT	Piebaldism and Familial Gastrointestinal Stromal Tumors (GISTs)
1696	KRAS	Cardio-Facio-Cutaneous Syndrome
1696	KRAS	Noonan, CFC and Costello Syndromes
1700	KRIT1	Cerebral Cavernous Malformations (CMM)
1701	KRIT1-Exon10 Hispanic	Cerebral Cavernous Malformations (CMM), Common Hispanic Mutations
1705	KTU/C14orf104	Primary Ciliary Dyskinesia (PCD)
1708	L1CAM	L1 Syndrome
1711	LAMA2	Merosin-Deficient Congenital Muscular Dystrophy
1714	LAMA2-Exon55 Mexico	Merosin-Deficient Congenital Muscular Dystrophy, Mexican Exon 55 Mutation
1717	LAMP2	Danon Disease/Glycogen Storage Disease IIb
1720	LARGE	Congenital Muscular Dystrophy, Type 1D
1720	LARGE	Walker-Warburg Syndrome
1723	LDB3	Myofibrillar Myopathy
1726	LFNG	Spondylocostal Dysostosis
1729	LHX4	Combined Pituitary Hormone Deficiency-4 (CPHD-4)
1732	LIPA	Wolman Disease and Cholesteryl Ester Storage Disease
1735	LMNA	Atypical Werner Syndrome
1735	LMNA	Charcot-Marie-Tooth Neuropathy Type 2B1
1735	LMNA	Dilated Cardiomyopathy
1735	LMNA	Emery-Dreifuss Muscular Dystrophy
1735	LMNA	Familial Partial Lipodystrophy
1735	LMNA	Hutchinson-Gilford Progeria Syndrome
1735	LMNA	Hutchinson-Gilford Progeria Syndrome
1735	LMNA	Lethal Tight Skin Contracture Syndrome (Restrictive Dermopathy)
1735	LMNA	Limb-Girdle Muscular Dystrophy Type 1B
1735	LMNA	Lipoatrophy
1735	LMNA	LMNA-Related Dilated Cardiomyopathy
1735	LMNA	Mandibuloacral Dysplasia
1738	LPIN1	Childhood Recurrent Acute Myoglobinuria, Autosomal Recessive
1741	LPIN1 Exon18-19 deletion	Paroxysmal Paralytic Rhabdomyolysis, Autosomal Recessive, Exons 18-19 Deletion Test

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1744	LRRC50	Primary Ciliary Dyskinesia (PCD)
1747	LYST	Chediak-Higashi Syndrome (CHS)
1748	MAG	Spastic Paraplegia
1749	MAOA	Brunner's Syndrome
1750	MAP2K1	Cardio-Facio-Cutaneous Syndrome
1753	MAP2K2	Cardio-Facio-Cutaneous Syndrome
1756	MASTL	Thrombocytopenia
1759	MATN3	Multiple Epiphyseal Dysplasia
1762	MATR3	Distal Myopathy 2
1765	MCCC1	3-Methylcrotonyl-CoA Carboxylase Deficiency
1768	MCCC2	3-Methylcrotonyl-CoA Carboxylase Deficiency
1771	MCEE	Methylmalonyl-CoA Epimerase Deficiency
1774	MED12	Lujan Syndrome / FG Syndrome Type 1
1777	MEK1	Cardio-Facio-Cutaneous Syndrome
1780	MEK2	Cardio-Facio-Cutaneous Syndrome
1783	MEN1	Multiple Endocrine Neoplasia Type 1
1786	MESP2	Jarcho-Levin Syndrome
1786	MESP2	Spondylocostal Dysostosis
1786	MESP2	Spondylothoracic Dysostosis
1789	MKKS/BBS6	Bardet-Biedl Syndrome
1792	MKS1	Meckel-Gruber Syndrome Testing
1795	MKS3	Joubert and Meckel-Gruber Syndrome
1798	MLH1	Lynch Syndrome
1801	MLH3	Lynch Syndrome
1804	MMAA	Methylmalonic Acidemia (cblA type)
1807	MMAB	Methylmalonic Acidemia (cblB type)
1810	MMACHC	Methylmalonic Aciduria and Homocystinuria, cblC type
1813	MPDU1	Congenital Disorders of Glycosylation, Type If (CDG If)
1816	MPI	Congenital Disorders of Glycosylation, Type Ib
1822	MPL	Congenital Amegakaryocytic Thrombocytopenia
1825	MSH2	Lynch Syndrome
1828	MSH6	Lynch Syndrome
1831	MUSK	Congenital Myasthenic Syndrome
1837	MUT	Methylmalonic Acidemia
1840	MUTYH	MUTYH Associated Polyposis (MAP) Syndrome
1843	MYBPC3	Hypertrophic Cardiomyopathy
1846	MYH2	Inclusion Body Myopathy-3, Autosomal Dominant
1849	MYH3	MYH3-Related Distal Arthrogryposis Syndromes
1852	MYH6	Hypertrophic Cardiomyopathy
1855	MYH7	Hypertrophic Cardiomyopathy and other MYH7-Related Disorders
1858	MYH9	MYH9-Related Disorders
1861	MYL2	Hypertrophic Cardiomyopathy
1864	MYL3	Hypertrophic Cardiomyopathy
1867	MYO7A	Usher Syndrome Type 1
1870	MYOT/TTID	Myotilinopathy Testing, Limb Girdle Muscular Dystrophy, Type 1A (LGMD1A), Myotilin-Related Myofibrillar Myopathy
1873	NAGLU	Mucopolysaccharidosis Type IIIB/Sanfilippo Syndrome B
1876	NAGPA	Stuttering
1879	NAGS	Hyperammonemia
1882	NEB	Nemaline Myopathy
1885	NEB-Exon55 del	Nemaline Myopathy, Ashkenazi Jewish Mutation Exon 55 Deletion Test
1888	NEK8/NPHP9	Nephronophthisis
1891	NF1	Neurofibromatosis Type 1 and Related Disorders
1891	NF1	Neurofibromatosis-Noonan Syndrome

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1894	NF2	Neurofibromatosis Type 2
1897	NPC1	Niemann-Pick Disease Type C1
1900	NPC2	Niemann-Pick Disease Type C2
1903	NPHP1	Nephronophthisis and Joubert Syndrome
1906	NPHP1	NPHP1 Homozygous Deletion Testing for Joubert Syndrome and Nephronophthisis
1909	NPHP2/INVS	Nephronophthisis
1912	NPHP3	Nephronophthisis
1915	NPHP4	Nephronophthisis
1918	NPHP9/NEK8	Nephronophthisis
1921	NR2E3	Retinitis Pigmentosa (Autosomal Dominant or Recessive, Nonsyndromic)
1924	NRAS	Noonan Syndrome
1927	NSD1	Sotos Syndrome
1930	OFD1	Oral-Facial-Digital Syndrome Type 1, Simpson-Golabi-Behmel Syndrome Type 2 and X-Linked Recessive Joubert Syndrome-10
1933	OPHN1	X-Linked Mental Retardation with Cerebellar Hypoplasia and Distinctive Facial Appearance
1936	OPTN	Amyotrophic Lateral Sclerosis and Primary Open-Angle Glaucoma
1939	OTC	Hyperammonemia
1942	PAFAH1B1/LIS1	Classic lissencephaly
1945	PAH	Phenylalanine Hydroxylase Deficiency
1948	PALB2/FANCN	Fanconi Anemia
1951	PCCA	Propionic Acidemia
1954	PCCB	Propionic Acidemia
1957	PCDH15	Usher Syndrome Type 1
1960	PDCD10 (CCM3)	Cerebral Cavernous Malformations
1963	PDE6A	Retinitis Pigmentosa, Autosomal Recessive
1966	PDE6B	Retinitis Pigmentosa, Autosomal Recessive and Congenital Stationary Night Blindness
1969	PDE6C	Achromatopsia
1972	PFKM	Glycogen Storage Disease, Type VII (Tarui Disease)
1975	PHKA1	Glycogen Storage Disease, Type IX
1978	PHKA2	Glycogen Storage Disease, Type IX
1981	PHKB	Glycogen Storage Disease, Type IX
1984	PHKG2	Glycogen Storage Disease, Type IX
1987	PKHD1	Autosomal Recessive Polycystic Kidney Disease
1990	PKP2	Arrhythmogenic Right Ventricular Cardiomyopathy/Dysplasia
1993	PLP1	PLP1-Related Disorders (Pelizaeus-Merzbacher Disease and Spastic Paraplegia 2)
1996	PMM2	Congenital Disorders of Glycosylation, Type Ia
1999	PMM2	Panel 1: PMM2, MPI, ALG6
2002	PMS1	Lynch Syndrome
2005	PMS2	Lynch Syndrome
2008	PNKD	Paroxysmal Nonkinesigenic Dyskinesia
2011	POMGNT1	Muscle-Eye-Brain Disease
2014	POMT1	Limb-Girdle Muscular Dystrophy, Type 2K
2017	POMT1	Walker-Warburg Syndrome
2020	POMT2	Muscle-Eye-Brain Disease
2023	POMT2	Walker-Warburg Syndrome
2026	PRF1	Familial Hemophagocytic Lymphohistiocytosis-Type 2 (FHL2)
2029	PRKAG2	PRKAG2-Related Disorders; Wolff-Parkinson-White Syndrome; Hypertrophic Cardiomyopathy with Wolff-Parkinson-White Syndrome; Glycogen Storage Disease of Heart, Lethal Congenital
2032	PRPF3	Retinitis Pigmentosa (Autosomal Dominant or Sporadic, Nonsyndromic)
2035	PRPF31	Retinitis Pigmentosa (Autosomal Dominant and Sporadic, Nonsyndromic)
2038	PRPF8	Retinitis Pigmentosa (Autosomal Dominant, Nonsyndromic)
2041	PRPH2	Retinitis Pigmentosa (Autosomal Dominant, Nonsyndromic)

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2044	PSAP	Metachromatic Leukodystrophy
2047	PTCH1	Holoprosencephaly-7 (Autosomal Dominant, Nonsyndromic)
2050	PTEN	PTEN Hamartoma Tumor Syndrome
2053	PTPN11	LEOPARD Syndrome
2056	PTPN11	Noonan Syndrome
2059	PYGL	Glycogen Storage Disease, Type VI
2062	PYGM	Glycogen Storage Disease, Type V (McArdle Disease)
2065	RAB27A	Griscelli Syndrome-Type 2 (GS2)
2068	RAF1	Noonan and Leopard Syndromes
2071	RAPSN	Rapsyn-Related Disorders
1360	RAPSN, DOK7	Pena-Shokeir Syndrome, Type 1 (Fetal Akinesia Deformation Sequence)
2074	RDH12	Leber Congenital Amaurosis, Progressive
2077	RELN	Lissencephaly with Cerebellar Hypoplasia
2080	RET-all Exons	Hirschsprung Disease
2083	RET-Exons 8-15	Multiple Endocrine Neoplasia Type 2A (MEN2A) and Familial Medullary Thyroid Carcinoma (FMTC)
2086	RET-Exons 15-16	Multiple Endocrine Neoplasia Type 2B (MEN2B)
2089	RHO	Retinitis Pigmentosa (Autosomal Dominant, Nonsyndromic)
2092	RMRP	Cartilage-Hair Hypoplasia (CHH) and Related Disorders
2095	ROR2	Robinow Syndrome, Autosomal Recessive; Brachydactyly, Type B1
2098	RP1	Retinitis Pigmentosa (Autosomal Dominant, Nonsyndromic)
2101	RPE65	Leber Congenital Amaurosis or Retinitis Pigmentosa
2104	RPGR-VariantC	Retinitis Pigmentosa 3 (RP3), X-Linked (XLRP)
2107	RPGR	Cone-Rod Dystrophy, X-Linked (CORDX1)
2208	RPGRIP1L	Joubert and Meckel-Gruber Syndromes
2110	RSPH4A	Primary Ciliary Dyskinesia (PCD)
2113	RSPH9	Primary Ciliary Dyskinesia (PCD)
2116	RUNX1	Familial Thrombocytopenia with Predisposition to Acute Myelogenous Leukemia
2119	RUNX2	Cleidocranial Dysplasia (CCD)
2122	RYR1	Central Core Disease
2125	RYR1	Malignant Hyperthermia Susceptibility
2125	RYR1	Multiminicore Disease
2125	RYR2	Catecholaminergic Polymorphic Ventricular Tachycardia
2128	SCN5A	Brugada Syndrome 1
2131	SEPN1	Multiminicore Disease (MmD)
2134	SEPN1	Rigid Spine Muscular Dystrophy-1
2134	SETX	Autosomal Recessive Spinocerebellar Ataxia and Amyotrophic Lateral Sclerosis Type-4
2137	SGCA	Limb Girdle Muscular Dystrophy, Type 2D
2140	SGCB	Limb Girdle Muscular Dystrophy, Type 2E
2143	SGCD	Dilated Cardiomyopathy and Limb Girdle Muscular Dystrophy, Type 2F
2146	SGCE	Myoclonus-Dystonia Syndrome
2149	SGCG	Limb Girdle Muscular Dystrophy, Type 2C
2152	SGSH	Mucopolysaccharidosis Type IIIA/Sanfilippo Syndrome A
2155	SHH	Holoprosencephaly-3 (Autosomal Dominant, Nonsyndromic)
2158	SHOC2	Noonan-Like Syndrome with Loose Anagen Hair
2161	SHOX	Short Stature Homeobox (SHOX)-related Haploinsufficiency Disorder
2164	SIX3	Holoprosencephaly-2 (Autosomal Dominant, Nonsyndromic)
2167	SLC26A2/DTDST	Achondrogenesis Type 1B (ACG1B)
2170	SLC26A2/DTDST	Atelosteogenesis Type 2 (AO2)
2170	SLC26A2/DTDST	Diastrophic Dysplasia (DTD)
2170	SLC26A2/DTDST	Multiple Epiphyseal Dysplasia, Recessive (EDM4/rMED)
2170	SLC2A2/GLUT2	Fanconi-Bickel Syndrome (Glycogen Storage Disease, Type XI)
2173	SLC35C1	Leukocyte Adhesion Deficiency Type 2 (LADII)
2176	SLC37A4	Glycogen Storage Disease, Type Ib

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2179	SLC9A6	Christianson Type X-Linked Mental Retardation
2182	SMAD4	Juvenile Polyposis Syndrome (JPS)
2185	SOD1	Amyotrophic Lateral Sclerosis / Motor Neuron Disease (Autosomal Dominant)
2188	SOS1	Noonan Syndrome
2191	SOX18	Lymphedema with Hypotrichosis and Telangiectasia
2194	SPRED1	Neurofibromatosis Type 1-Like Syndrome
2197	STK11	Peutz-Jeghers Syndrome
2200	STX11	Familial Hemophagocytic Lymphohistiocytosis-Type 4 (FHL4)
2203	STXBP2	Familial Hemophagocytic Lymphohistiocytosis-Type 5 (FHL5)
2206	SUMF1	Multiple Sulfatase Deficiency / Mucosulfatidosis
2209	SYNE1	Synaptic Nuclear Envelope Protein-1 Related Disorders
2212	TARDBP	Amyotrophic Lateral Sclerosis / Motor Neuron Disease
2215	TBX5	Holt-Oram Syndrome (HOS)
2218	TCAP	Telethoninopathy Testing
2219	TCF7L2	Susceptibility to Diabetes mellitus, type 2
2221	TGFB1	Camurati-Engelmann Disease (CED)
2224	TGFBR1	Loeys-Dietz Syndrome
2227	TGFBR2	Loeys-Dietz Syndrome
2230	TGIF1	Holoprosencephaly-4 (Autosomal Dominant, Nonsyndromic)
2233	TH	Tyrosine Hydroxylase Deficiency and Related Disorders
2236	TMEM216	Joubert Syndrome
2239	TMEM67	Joubert and Meckel-Gruber Syndromes
2242	TNNI2	Distal Arthrogryposis 2B (Sheldon-Hall Syndrome)
2245	TNNI3	Hypertrophic Cardiomyopathy and Related Disorders
2248	TNNT1	Nemaline Myopathy 5 (Amish Nemaline Myopathy)
2251	TNNT2	Hypertrophic Cardiomyopathy and Related Disorders
2254	TNNT3	Distal Arthrogryposis 2B (Sheldon-Hall Syndrome)
2257	TOR1A	Early-Onset Primary Dystonia
2260	TP53	Li-Fraumeni Syndrome
2263	TPM1	Hypertrophic Cardiomyopathy and Dilated Cardiomyopathy
2266	TPM2	Tropomyosin 2-Related Disorders
2269	TPM3	Tropomyosin 3-Related Myopathies
2272	TSEN2	Pontocerebellar Hypoplasias Subtype 2
2275	TSEN34	Pontocerebellar Hypoplasias Subtype 2
2278	TSEN54	Pontocerebellar Hypoplasias Subtypes 2 and 4
2281	TTC8/BBS8	Bardet-Biedl Syndrome
2284	TTID/MYOT	Myotilinopathy Testing, Limb Girdle Muscular Dystrophy, Type 1A (LGMD1A), Myotilin-Related Myofibrillar Myopathy
2287	TTN	Limb Girdle Muscular Dystrophy, Type 2J and Tibial Muscular Dystrophy
2290	TUBA1A	Lissencephaly 3
2293	TULP1	Retinitis Pigmentosa and Leber Congenital Amaurosis
2296	TXNDC3	Primary Ciliary Dyskinesia (PCD)
2299	UNC13D	Familial Hemophagocytic Lymphohistiocytosis-Type 3 (FHL3)
2302	USH1C	Usher Syndrome Type 1
2305	USH2A	Usher Syndrome Type 2
2308	VAPB	Amyotrophic Lateral Sclerosis-8 / Spinal Muscular Atrophy, Autosomal Dominant, Adult-Onset
2311	VCL	Hypertrophic Cardiomyopathy
2314	VIPAR	Arthrogryposis-Renal Dysfunction-Cholestasis (ARC) Syndrome
2317	VPS13B (COH1)	Cohen Syndrome
2320	VPS33B	Arthrogryposis-Renal Dysfunction-Cholestasis (ARC) Syndrome
2323	WAS	Wiskott-Aldrich Syndrome and Related Disorders
2326	XPNPEP3	Nephronophthisis-Like Nephropathy-1
2329	YWHAE	Miller-Dieker Lissencephaly Syndrome

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2332	ZIC2	Holoprosencephaly-5
2335	ZMPSTE24	Mandibuloacral Dysplasia
2338	ZMPSTE24	Restrictive Dermopathy