Test Menu

GENE	ASSOCIATED CONDITIONS
ABCC8	Familial Hyperinsulinism (ABCC8-Related)
ABCD1	Adrenoleukodystrophy, X-Linked
ACADM	Medium Chain Acyl-CoA Dehydrogenase Deficiency
ACADVL	Very Long Chain Acyl-CoA Dehydrogenase Deficiency
ACAT1	Beta-Ketothiolase Deficiency
ACSF3	Combined Malonic Methylmalonic Aciduria
ADA	Adenosine Deaminase Deficiency
AGA	Aspartylglycosaminuria
AGL	Glycogen Storage Disease, Type III
AGXT	Primary Hyperoxaluria, Type I
AIRE	Polyglandular Autoimmune Syndrome Type I
ALDH3A2	Sjogren-Larsson Syndrome
ALDOB	Hereditary Fructose Intolerance
ALG6	Congenital Disorder of Glycosylation Type Ic
ALMS1	Alstrom Syndrome
ALPL	Hypophosphatasia
ARSA	Metachromatic Leukodystrophy
ARSB	Mucopolysaccharidosis type VI
ASL	Argininosuccinic Aciduria
ASPA	Canavan Disease
ASS1	Citrullinemia, Type I
ATM	Ataxia-Telangiectasia
ATP7A	Menkes Disease
ATP7B	Wilson Disease
BBS1	Bardet-Biedl Syndrome (BBS1-Related)
BCKDHA	Maple Syrup Urine Disease, Type 1a
BCKDHB	Maple Syrup Urine Disease, Type 1b
BCS1L	GRACILE Syndrome Other BCS1L-Related Disorders
BLM	Bloom Syndrome
BTD	Biotinidase Deficiency
CAPN3	Limb-Girdle Muscular Dystrophy Type 2A
CBS	Homocystinuria (CBS-Related)
CFTR	Cystic Fibrosis
CLN3	Neuronal Ceroid-Lipofuscinosis (CLN3-Related)
CLN5	Neuronal Ceroid-Lipofuscinosis (CLN5-Related)
CLN6	Neuronal Ceroid-Lipofuscinosis (CLN6-Related)
CLN8	Neuronal Ceroid-Lipofuscinosis (CLN8-Related)
CLRN1	Usher Syndrome, Type III
COL4A3	Alport Syndrome (COL4A3-Related)
COL4A4	Alport Syndrome (COL4A4-Related)
COL4A5	Alport Syndrome (COL4A5-Related)
CPT1A	Carnitine Palmitoyltransferase IA Deficiency

GENE	ASSOCIATED CONDITIONS
CPT2	Carnitine Palmitoyltransferase II Deficiency
CTNS	Cystinosis
CTSK	Pycnodysostosis
CYP27A1	Cerebrotendinous Xanthomatosis
DHCR7	Smith-Lemli-Opitz Syndrome
DLD	Lipoamide Dehydrogenase Deficiency
DMD	Duchenne Muscular Dystrophy Becker Muscular Dystrophy
DYSF	Dysferlinopathy
ETFA	Glutaric Acidemia, Type IIa
ETFDH	Glutaric Acidemia, Type IIc
F11	Factor XI Deficiency
F9	Factor IX Deficiency
FAH	Tyrosinemia, Type I
FANCA	Fanconi Anemia, Group A
FANCC	Fanconi Anemia, Group C
FKTN	FKTN-Related Disorders (including Walker-Warburg Syndrome)
FMR1	Fragile X Syndrome
G6PC	Glycogen Storage Disease, Type Ia
GAA	Glycogen Storage Disease, Type II
GALC	Krabbe Disease
GALK1	Galactokinase Deficiency
GALT	Galactosemia
GAMT	Cerebral Creatine Deficiency Syndrome 2
GBA	Gaucher Disease
GCDH	Glutaric Acidemia, Type I
GJB2	Non-Syndromic Hearing Loss (GJB2-Related)
GLA	Fabry Disease
GLB1	GLB1-Related Disorders
GNE	Inclusion Body Myopathy 2
GNPTAB	Mucolipidosis II / IIIA
GNPTG	Mucolipidosis III Gamma
GP9	Bernard-Soulier Syndrome, Type C
GRHPR	Primary Hyperoxaluria, Type 2
HADHA	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency
HBB	Beta-Globin-Related Hemoglobinopathies
HEXA	Hexosaminidase A Deficiency (including Tay-Sachs Disease)
HEXB	Sandhoff Disease
HGSNAT	Mucopolysaccharidosis Type IIIC
HLCS	Holocarboxylase Synthetase Deficiency
HMGCL	HMG-CoA Lyase Deficiency
HOGA1	Primary Hyperoxaluria, Type 3
HSD17B4	D-Bifunctional Protein Deficiency

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GENE	ASSOCIATED CONDITIONS
HYLS1	Hydrolethalus Syndrome
IDS	Mucopolysaccharidosis Type II
IDUA	Mucopolysaccharidosis Type I
IKBKAP	Familial Dysautonomia
IL2RG	X-Linked Severe Combined Immunodeficiency
IVD	Isovaleric Acidemia
KCNJ11	Familial Hyperinsulinism (KCNJ11-Related)
LAMA3	Junctional Epidermolysis Bullosa (LAMA3-Related)
LAMB3	Junctional Epidermolysis Bullosa (LAMB3-Related)
LAMC2	Junctional Epidermolysis Bullosa (LAMC2-Related)
LIPA	Lysosomal Acid Lipase Deficiency
LRPPRC	Leigh Syndrome, French-Canadian Type
MAN2B1	Alpha-Mannosidosis
MCCC1	3-Methylcrotonyl-CoA Carboxylase Deficiency
MCCC2	3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC2-Related)
MCOLN1	Mucolipidosis IV
MEFV	Familial Mediterranean Fever
MLC1	Megalencephalic Leukoencephalopathy with Subcortical Cysts
MMAA	Methylmalonic Acidemia (MMAA-Related)
MMAB	Methylmalonic Acidemia (MMAB-Related)
MMACHC	Methylmalonic Aciduria and Homocystinuria Cobalamin C Type
MMADHC	Methylmalonic Aciduria and Homocystinuria Cobalamin D Type
MPI	Congenital Disorder of Glycosylation, Type Ib
MTRR	Homocystinuria, cblE Type
MUT	Methylmalonic Acidemia (MUT-Related)
NAGLU	Mucopolysaccharidosis Type IIIB
NBN	Nijmegen Breakage Syndrome
NEB	Nemaline Myopathy 2
NPC1	Niemann-Pick Disease, Type C (NPC1-Related)
NPC2	Niemann-Pick Disease, Type C (NPC2-Related)
NPHS1	Nephrotic Syndrome (NPHS1-Related) Congenital Finnish Nephrosis
NPHS2	Nephrotic Syndrome (NPHS2-Related) Steroid-Resistant Nephrotic Syndrome
OAT	Ornithine Aminotransferase Deficiency
OPA3	3-Methylglutaconic Aciduria, Type III
OTC	Ornithine Transcarbomylase Deficiency
PAH	Phenylalanine Hydroxylase Deficiency
PCCA	Propionic Acidemia (PCCA-Related)
PCCB	Propionic Acidemia (PCCB-Related)
PEX1	Zellweger Syndrome Spectrum (PEX1-Related)
PEX2	Zellweger Syndrome Spectrum (PEX2-Related)
PEX6	Zellweger Syndrome Spectrum (PEX6-Related)
PEX7	Rhizomelic Chondrodysplasia Punctata, Type 1
PKHD1	Polycystic Kidney Disease, Autosomal Recessive
PMM2	Congenital Disorder of Glycosylation, Type Ia
POMGNT1	Muscle-Eye-Brain Disease and Other POMGNT1-Related Congenital Muscular Dystrophy-Dystroglycanopathies
PPT1	Neuronal Ceroid-Lipofuscinosis (PPT1-Related)
PROP1	Combined Pituitary Hormone Deficiency 2
PTS	6-Pyruvoyl-Tetrahydropterin Synthase Deficiency
PYGM	Glycogen Storage Disease, Type V
RMRP	Cartilage-Hair Hypoplasia
RS1	X-Linked Juvenile Retinoschisis
RTEL1	Dyskeratosis Congenita (RTEL1-Related)
SACS	Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay
SGCA	Limb-Girdle Muscular Dystrophy, Type 2D
SGCB	Limb-Girdle Muscular Dystrophy, Type 2E
SGCG	Limb-Girdle Muscular Dystrophy, Type 2C
SGSH	Mucopolysaccharidosis Type IIIA
SLC12A6	Andermann Syndrome
SLC17A5	Salla Disease
SLC22A5	Primary Carnitine Deficiency
SLC25A13	Citrin Deficiency
SLC25A15	Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome
SLC26A2	Sulfate Transporter-Related Osteochondrodysplasia
SLC26A4	Pendred Syndrome
SLC35A3	Arthrogryposis, Mental Retardation, and Seizures
SLC37A4	Glycogen Storage Disease, Type Ib
SMN1	Spinal Muscular Atrophy
SMPD1	Niemann-Pick Disease (SMPD1-Related)
STAR	Lipoid Adrenal Hyperplasia
TCIRG1	Osteopetrosis 1
TH	Segawa Syndrome
TMEM216	Joubert Syndrome 2
TPP1	Neuronal Ceroid-Lipofuscinosis (TPP1-Related)
ACADS	Short Chain Acyl-CoA Dehydrogenase Deficiency
ARG1	Argininemia
BBS10	Bardet-Biedl Syndrome (BBS10-Related)
CYP21A2	Congenital Adrenal Hyperplasia (CAH)
DBT	Maple Syrup Urine Disease, Type II
TAT	Tyrosinemia type II
G6PD	Glucose-6-phosphate dehydrogenase deficiency
GJB6	Non-Syndromic Hearing Loss (GJB6-Related)
HOGA1	Primary Hyperoxaluria, Type 3