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Carrier Screening Disease & Gene List

BASIC 5					
HBA1/HBA2	Alpha-Thalassemia	CFTR	Cystic Fibrosis	SMN1	Spinal Muscular Atrophy
HBB	Beta Hemoglobinopathies (Beta-Thalassemia and Sickle Cell)	DMD	Duchenne/Becker Muscular Dystrophy		
ACOG/ACMG 13					
HBA1/HBA2	Alpha-Thalassemia	DMD	Duchenne/Becker Muscular Dystrophy	MCOLN1	Mucolipidosis IV
HBB	Beta Hemoglobinopathies (Beta-thalassemia and Sickle Cell)	IKBKAP	Familial Dysautonomia	SMPD1	Niemann-Pick Disease, Types A and B
BLM	Bloom Syndrome	FANCC	Fanconi Anaemia	SMN1	Spinal Muscular Atrophy
ASPA	Canavan Disease	GBA	Gaucher Disease	HEXA	Tay-Sachs Disease
CFTR	Cystic Fibrosis				
ASHKANAZI JEWISH 38					
PHGDH	3-Phosphoglycerate Dehydrogenase Deficiency	RTEL1	Dyskeratosis Congenita	SUMF1	Multiple Sulphatase Deficiency
MTTP	Abetalipoproteinaemia	ADAMTS2	Ehlers-Danlos Syndrome VIIc	NEB	Nemaline Myopathy: NEB Related
COL4A3	Alport Syndrome	IKBKAP	Familial Dysautonomia	SMPD1	Niemann-Pick Disease, Type A&B
SLC35A3	Arthrogryposis, Mental Retardataion and Seizures	ABCC8	Familial Hyperinsulinism	PKHD1	Polycystic Kidney Disease, Autosomal Recessive
BBS2	Bardet-Biedl Syndrome	FANCC	Fanconi Anaemia	DHDDS	Retinitis Pigmentosa, Autosomal Recessive
BLM	Bloom Syndrome	FKTN	Fukuyama Congenital Muscular Dystrophy	DHCR7	Smith-Lemli-Opitz Syndrome
ASPA	Canavan Disease	GALT	Galactosemia Type I	SMN1	Spinal Muscular Atrophy
CPT2	Carnitine Palmitoyltransferase II Deficiency	GBA	Gaucher Disease Types I, II, III	HEXA	Tay-Sachs Disease
MPL	Congenital Amegakaryocytic Thrombocytopenia	G6PC	Glycogen Storage Disease: Type IA	FAH	Tyrosinemia: Type I, II, III
PMM2	Congenital Disorder of Glycosylation: Type IA	TMEM216	Joubert Syndrome	PCDH15	Usher Syndrome: Type IF
CFTR	Cystic Fibrosis	BCKDHB	Maple Syrup Urine Disease, Type IB	CLRN1	Usher Syndrome: Type IIIA
DLD	Dihydrolipoamide Dehydrogenase Deficiency	MCOLN1	Mucolipidosis IV	ATP7B	Wilson Disease
DMD	Duchenne/Becker Muscular Dystrophy			PEX2	Zellweger Spectrum Disorders
PAN-ETHNIC 167					
ABCC8	Familial hyperinsulinism	G6PC	Glycogen storage disease typeIa (GSDIa)	MYL3	Hypertrophic cardiomyopathy, dilated cardiomyopathy
ABCD1	X-linked adrenoleukodystrophy Adrenomyeloneuropathy Addison disease (X-ALD)	G6PD	Glucose-6-phosphate dehydrogenase deficiency	NEB	Nemaline Myopathy: NEB related
ABCD4	Methylmalonic acidemia with	GAA	Pompe disease (GAA deficiency)	NPC1	Niemann-Pick disease (Type C1)
ACAD8	homocystinuria	GALC	Krabbe Disease	NPC2	Niemann-Pick disease (Type C2)
ACADM	Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency	GALE	Galactosemia Type III	NPHS1	Nephrotic syndrome
ACADS	Short-chain acyl-CoA dehydrogenase (SCAD) deficiency	GALK1	Galactosemia Type II	NPHS2	Nephrotic syndrome
ACADSB	2-methylbutyryl-CoA dehydrogenase deficiency	GALT	Galactosemia Type I	OPA3	3-methylglutaconic aciduria (3MGA) Type I

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ACADVL	Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency	GBA	Gaucher disease (Types I, II, & III)	OTC	Ornithine Transcarbamylase Deficiency (OTC)
ACAT1	Beta-ketothiolase deficiency (βKT)	GCDH	Glutaric acidemia Type I (GA1)	PAH	Phenylketonuria (PKU)
ACSF3	Combined malonic and methylmalonic aciduria	GCH1	Disorders of biopterin regeneration	PAX8	Congenital hypothyroidism (CH)
ACTA2	Marfan syndrome	GJB2	Hearing Loss	PC	Pyruvate Carboxylase deficiency
ACTC1	hypertrophic cardiomyopathy, dilated cardiomyopathy	GJB3	hearing loss	PCBD1	Disorders of biopterin regeneration
ADA	Severe combined immunodeficiency (SCID)	GJB6	Hearing Loss	PCCA	Propionic acidemia (PROP)
ADAMTS2	Ehlers-Danlos Syndrome	GLA	Fabry disease	PCCB	Propionic acidemia (PROP)
AGXT	Primary hyperoxaluria	GNMT	Hypermethioninemia (MET)	PCDH15	Usher syndrome 1F
AHCY	Hypermethioninemia (MET)	GRHPR	Primary hyperoxaluria	PEX1	Zellweger Spectrum Disorders
APC	Familial Adenomatous Polyposis	HADH	3-hydroxyacyl-CoA dehydrogenase deficiency (M/SCHAD)	PEX2	Zellweger Spectrum Disorders
APOB	Familiar hypercholesterolemia	HADHA	Trifunctional protein deficiency (TFP)	PHGDH	3-Phosphoglycerate Dehydrogenase Deficiency (PHGDH)
ARG1	Arginase deficiency (ARG)	HADHB	Trifunctional protein deficiency (TFP)	PKHD1	Autosomal recessive polycystic kidney disease
ASL	Argininosuccinic aciduria (ASA)	HBA1	Alpha thalassemia (Hemoglobin Disorder-Var-Hb)	PKP2	Arrhythmogenic right ventricular cardiomyopathy
ASPA	Canavan disease	HBA2	Alpha thalassemia (Hemoglobin Disorder-Var-Hb)	PMM2	Congenital disorder of glycosylation, type 1A, PMM2 related
ASS1	Citrullinemia (CIT) Type I	HBB	(I) Sickle cell disease	POMGNT1	Muscle eye brain disease
ATP7B	Wilson Disease	HCFC1	Methylmalonic acidemia with homocystinuria	PRKAG2	Wolff-Parkinson-White syndrome
AUH	3-methylglutaconic aciduria (3MGA) Type I	HEXA	Tay-Sachs disease	RTEL1	Dyskeratosis Congenita
BCKDHA	Maple Syrup Urine Disease (MSUD) Type 1A	HEXB	Sandhoff disease	RYR1	Malignant hyperthermia
BBS2	Bardet-Biedl Syndrome BBS2 related	HLCS	Multiple Carboxylase Deficiency (MCD)	RYR2	Catecholaminergic polymorphic ventricular tachycardia
BCKDHB	Maple Syrup Urine Disease (MSUD) Type 1B	HMGCL	3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)	PTS	Disorders of biopterin regeneration
BLM	Bloom syndrome	HPD	Tyrosinemia (TYR I, II, III)	QDPR	Disorders of biopterin regeneration
BTD	Biotinidase deficiency (BIOT)	HSD17B10	3-hydroxy-2-methylbutyryl-CoA dehydrogenase deficiency (2M3HBA)	SCN5A	Cardiomyopathy, Brugada syndrome, and cardiac conditions
CBS	Homocystinuria (HCY)	IDUA	Mucopolysaccharidosis Type I (MPS I)	SLC22A5	Primary carnitine deficiency (CUD)
col3A1	Ehlers-Danlos syndrome, vascular type	IKBKAP	Familial dysautonomia	SLC25A13	Citrullinemia Type II (CIT II)
COL4A3	Alport Syndrome	IL2RG	X-linked severe combined immunodeficiency (SCID)	SLC25A20	Carnitine-acylcarnitine translocase (CACT)
CD320	Methylmalonic acidemia due to transcobalamin receptor defect	IVD	Isovaleric acidemia (IVA)	SLC26A4	Pendred syndrome

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CFTR	Cystic fibrosis (CF)	KCNH2	Romano-Ward long-QT syndrome types 1,2, 3, Brugada syndrome	SLC35A3	Arthrogryposis, Mental retardation and Seizures
CLRN1	Usher syndrome 3A	KCNQ1	Romano-Ward long-QT syndrome types 1,2, 3, Brugada syndrome	SLC5A5	Congenital hypothyroidism (CH)
CPT1A	Carnitine palmitoyltransferase I deficiency (CPT IA)	MAT1A	Hypermethioninemia (MET)	SMN1	Spinal Muscular Atrophy
CPT2	Carnitine palmitoyltransferase II deficiency (CPT II)	MCCC1	3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)	SMPD1	Niemann-Pick disease (Type A & B)
CYP1B1	primary congenital glaucoma	MCCC2	3-methylcrotonyl-CoA carboxylase deficiency (3-MCC)	SUMF1	Multiple sulpjhatase deficiency
CYP21A2	21-hydroxylase deficiency (CAH)	MCEE	Methylmalonic acidemia	TAT	Tyrosinemia (TYR I, II, III)
DBT	Maple Syrup Urine Disease (MSUD)	MCOLN1	Mucolipidosis type IV	TAZ	3-methylglutaconic aciduria (3MGA) Type I
DHCR7	Smith-Lemli-Opitz Syndrome	MLYCD	Malonyl-CoA decarboxylase deficiency (MAL)	TCN2	Transcobalamin deficiency
DHDDS	Retinitis pigmentosa, autosomal recessive	MMAA	Methylmalonic acidemia (cblA)	TG	Congenital hypothyroidism (CH)
DLD	Maple Syrup Urine Disease (MSUD) Type 111	MMAB	Methylmalonic acidemia (cblB)	TGFBR1	Familial thoracic aordic aneurysm, Loeys-Dietz Syndrome, etc.
DMD	Duchenne/Becker Muscular Dystrophy	MMACHC	(I) Methylmalonic acidemia (cblC)	TGFBR2	Familial thoracic aordic aneurysm, Loeys-Dietz Syndrome
DNAJC19	3-methylglutaconic aciduria (3MGA) Type V	MMADHC	(I) Methylmalonic acidemia (cblC)	THRA	Congenital hypothyroidism (CH)
DSC2	Arthythmogenic right ventricular cardiomyopathy	MPL	Congenital amegakaryocytic thrombocytopenia	THRB	Congenital hypothyroidism (CH)
DSG2	Arthythmogenic right ventricular cardiomyopathy	MTHFR	Homocystinuria (HCY)	TMEM216	Joubert syndrome
DSP	Arthythmogenic right ventricular cardiomyopathy	MTR	Homocystinuria (HCY)	TMEM43	Arrhythmogenic right ventricular cardiomyopathy, muscular dystrophy
DUOX2	Congenital hypothyroidism (CH)	MTRR	Homocystinuria (HCY)	TNNI3	Familial cardiomyopathy
ETFA	Glutaric acidemia type II (GA2)	MTTP	Abetalipoproteinase Deficiency (ADA)	TNNT2	Familial cardiomyopathy
ETFB	Glutaric acidemia type II (GA2)	MUT	Methylmalonic acidemia (MUT)	TPM1	Familial cardiomyopathy
ETFDH	Glutaric acidemia type II (GA2)	MyBPC2	Hypertrophic cardiomyopathy, dilated cardiomyopathy	TPO	Congenital hypothyroidism (CH)
FAH	Tyrosinemia (TYR I, II, III)	MYH11	Warfin syndrome	TSHB	Congenital hypothyroidism (CH)
FANCC	Fanconi Anemia	MYH7	Hypertrophic cardiomyopathy, dilated cardiomyopathy	TSHR	Congenital hypothyroidism (CH)
FBN1	Marfan Syndrome, etc.	MYL2	Hypertrophic cardiomyopathy, dilated cardiomyopathy	USH1b (Myo7a)	Usher syndrome and deafness
FKTN	Fukuyama congenital muscular dystrophy			USH1C	Usher Syndrome