CENTOGENE
THE RARE DISEASE COMPANY

CentoMetabolic®

COMBINING GENETIC AND BIOCHEMICAL TESTING FOR THE FAST AND COMPREHENSIVE DIAGNOSTIC OF METABOLIC DISORDERS

GENE	ASSOCIATED DISEASE(S)
ABCA1	HDL deficiency, familial, 1; Tangier disease
ABCB4	Cholestasis, progressive familial intrahepatic 3
ABCC2	Dubin-Johnson syndrome
ABCD1	Adrenoleukodystrophy; Adrenomyeloneuropathy, adult
ABCD4	Methylmalonic aciduria and homocystinuria, cblJ type
ABCG5	Sitosterolemia 2
ABCG8	Sitosterolemia 1
ACAT1	Alpha-methylacetoacetic aciduria
ADA	Adenosine deaminase deficiency
AGA	Aspartylglucosaminuria
AGL	Glycogen storage disease IIIa; Glycogen storage disease IIIb
AGPS	Rhizomelic chondrodysplasia punctata, type 3
AGXT	Hyperoxaluria, primary, type 1
ALAD	Porphyria, acute hepatic
ALAS2	Anemia, sideroblastic, 1; Protoporphyria, erythropoietic, X-linked
ALDH4A1	Hyperprolinemia, type II
ALDOA	Glycogen storage disease XII
ALDOB	Fructose intolerance, hereditary
ALG3	Congenital disorder of glycosylation, type Id
ALPL	Hypophosphatasia, adult; Hypophosphatasia, childhood; Hypophosphatasia, infantile; Odontohypophosphatasia
ANTXR2	Hyaline fibromatosis syndrome
APOA2	Hypercholesterolemia, familial, modifier of
APOA5	Hyperchylomicronemia, late-onset
APOB	Hypercholesterolemia, familial, 2
APOC2	Hyperlipoproteinemia, type Ib
APOE	Sea-blue histiocyte disease; Hyperlipoproteinemia, type III
ARG1	Argininemia
ARSA	Metachromatic leukodystrophy
ARSB	Mucopolysaccharidosis type VI (Maroteaux-Lamy)

GENE	ASSOCIATED DISEASE(S)
ASAH1	Farber lipogranulomatosis; Spinal muscular atrophy with progressive myoclonic epilepsy
ASL	Argininosuccinic aciduria
ASS1	Citrullinemia
ATP7A	Menkes disease; Occipital horn syndrome
ATP7B	Wilson disease
BCKDHA	Maple syrup urine disease, type Ia
BCKDHB	Maple syrup urine disease, type Ib
BTD	Biotinidase deficiency
CBS	Homocystinuria, B6-responsive and nonresponsive types
CD320	Methylmalonic aciduria, transient, due to transcobalamin receptor defect
CETP	Hyperalphalipoproteinemia
CLN3	Ceroid lipofuscinosis, neuronal, 3
CLN5	Ceroid lipofuscinosis, neuronal, 5
CLN6	Ceroid lipofuscinosis, neuronal, 6; Ceroid lipofuscinosis, neuronal, Kufs type, adult onset
CLN8	Ceroid lipofuscinosis, neuronal, 8; Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant
CPOX	Coproporphyria
CPS1	Carbamoylphosphate synthetase I deficiency
CPT1A	CPT deficiency, hepatic, type IA
CTNS	Cystinosis
CTSA	Galactosialidosis
CTSD	Ceroid lipofuscinosis, neuronal, 10
CTSK	Pycnodysostosis
CYP11B1	Congenital adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
CYP17A1	Congenital adrenal hyperplasia, due to 17-alpha-hydroxylase deficiency
CYP19A1	Aromatase deficiency
CYP21A2	Congenital adrenal hyperplasia, due to 21-hydroxylase deficiency
DBT	Maple syrup urine disease, type II
DDC	Aromatic L-amino acid decarboxylase deficiency
DHCR7	Smith-Lemli-Opitz syndrome
DIABLO	Deafness, autosomal dominant 64
DLX4	Orofacial cleft 15
DNAJC5	Ceroid lipofuscinosis, neuronal, 4, Parry type
DPYD	Dihydropyrimidine dehydrogenase deficiency

GENE	ASSOCIATED DISEASE(S)
ENO3	Beta-enolase deficiency
ENPP1	Arterial calcification, generalized, of infancy, 1; Cole disease; Hypophosphatemic rickets, autosomal recessive, 2
EPHX2	Hypercholesterolemia, familial, due to LDLR defect, modifier of
ETHE1	Ethylmalonic encephalopathy
FAH	Tyrosinemia, type I
FBP1	Fructose-1,6-bisphosphatase deficiency
FECH	Protoporphyria, erythropoietic, 1
FGF23	Hypophosphatemic rickets, autosomal dominant; Tumoral calcinosis, hyperphosphatemic, familial, 2
FUCA1	Fucosidosis
G6PC	Glycogen storage disease Ia
G6PD	Hemolytic anemia, G6PD deficient (favism)
GAA	Glycogen storage disease II
GALC	Krabbe disease
GALE	Galactose epimerase deficiency
GALK1	Galactokinase deficiency with cataracts
GALNS	Mucopolysaccharidosis IVA
GALT	Galactosemia
GAMT	Cerebral creatine deficiency syndrome 2
GATM	Cerebral creatine deficiency syndrome 3
GBA	Gaucher disease
GBE1	Glycogen storage disease IV; Polyglucosan body disease, adult form
GHR	Laron dwarfism; Hypercholesterolemia, familial, modifier of
GK	Glycerol kinase deficiency
GLA	Fabry disease
GLB1	GM1-gangliosidosis; Mucopolysaccharidosis type IVB (Morquio)
GM2A	GM2-gangliosidosis, AB variant
GNPAT	Rhizomelic chondrodysplasia punctata, type 2
GNPTAB	Mucolipidosis II alpha/beta; Mucolipidosis III alpha/beta
GNPTG	Mucolipidosis III gamma
GNS	Mucopolysaccharidosis type IIID
GUSB	Mucopolysaccharidosis VII
GYG1	Polyglucosan body myopathy 2
GYS1	Glycogen storage disease 0, muscle

GENE	ASSOCIATED DISEASE(S)
GYS2	Glycogen storage disease 0, liver
HCFC1	methylmalonic acidemia and homocysteinemia, cblX type
HEXA	Tay-Sachs disease; GM2-gangliosidosis
HEXB	Sandhoff disease
HFE	Hemochromatosis
HGD	Alkaptonuria
HGSNAT	Mucopolysaccharidosis type IIIC (Sanfilippo C)
HJV	Hemochromatosis, type 2A
HLCS	Holocarboxylase synthetase deficiency
HMBS	Porphyria, acute intermittent
HPD	Hawkinsinuria; Tyrosinemia, type III
HPRT1	Lesch-Nyhan syndrome
HSD3B2	Congenital adrenal hyperplasia, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency
HYAL1	Mucopolysaccharidosis type IX
IDS	Mucopolysaccharidosis II
IDUA	Mucopolysaccharidosis I
ITIH4	Hypercholesterolemia, susceptibility to
IVD	Isovaleric acidemia
KHK	Fructosuria
LAMP2	Danon disease
LCAT	Fish-eye disease; Norum disease
LDHA	Glycogen storage disease XI
LDLR	Hypercholesterolemia, familial, 1
LDLRAP1	Hypercholesterolemia, familial, 4
LIPA	Cholesteryl ester storage disease; Wolman disease
LIPC	Hepatic lipase deficiency
LIPI	Hypertriglyceridemia, susceptibility to
LMBRD1	Methylmalonic aciduria and homocystinuria, cblF type
LPA	LPA deficiency, congenital
LPL	Lipoprotein lipase deficiency
MAN2B1	Mannosidosis, alpha-, types I and II
MANBA	Mannosidosis, beta
MCOLN1	Mucolipidosis IV

GENE	ASSOCIATED DISEASE(S)
MFSD8	Ceroid lipofuscinosis, neuronal, 7; Macular dystrophy with central cone involvement
MMAA	Methylmalonic aciduria, vitamin B12-responsive
MMAB	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type
MMACHC	Methylmalonic aciduria and homocystinuria, cblC type
MMADHC	Homocystinuria, cblD type, variant 1; Methylmalonic aciduria and homocystinuria, cblD type; Methylmalonic aciduria, cblD type, variant 2
MMUT	Methylmalonic aciduria, mut(0) type
NAGA	Kanzaki disease; Schindler disease, type I; Schindler disease, type III
NAGLU	Mucopolysaccharidosis type IIIB (Sanfilippo B)
NAGS	N-acetylglutamate synthase deficiency
NEU1	Sialidosis, type I; Sialidosis, type II
NPC1	Niemann-Pick disease, type C1; Niemann-Pick disease, type D
NPC2	Niemann-pick disease, type C2
OTC	Ornithine transcarbamylase deficiency
PAH	Phenylketonuria
PCSK9	Hypercholesterolemia, familial, 3
PDHB	Pyruvate dehydrogenase E1-beta deficiency
PEX1	Heimler syndrome 1; Peroxisome biogenesis disorder 1A (Zellweger); Peroxisome biogenesis disorder 1B (NALD/IRD)
PEX10	Peroxisome biogenesis disorder 6A (Zellweger); Peroxisome biogenesis disorder 6B
PEX12	Peroxisome biogenesis disorder 3A (Zellweger); Peroxisome biogenesis disorder 3B
PEX13	Peroxisome biogenesis disorder 11A (Zellweger); Peroxisome biogenesis disorder 11B
PEX14	Peroxisome biogenesis disorder 13A (Zellweger)
PEX16	Peroxisome biogenesis disorder 8A (Zellweger); Peroxisome biogenesis disorder 8B
PEX19	Peroxisome biogenesis disorder 12A (Zellweger)
PEX2	Peroxisome biogenesis disorder 5A (Zellweger); Peroxisome biogenesis disorder 5B
PEX26	Peroxisome biogenesis disorder 7A (Zellweger); Peroxisome biogenesis disorder 7B
PEX3	Peroxisome biogenesis disorder 10A (Zellweger)
PEX5	Peroxisome biogenesis disorder 2A (Zellweger); Peroxisome biogenesis disorder 2B; Rhizomelic chondrodysplasia punctata, type 5
PEX6	Heimler syndrome 2; Peroxisome biogenesis disorder 4A (Zellweger); Peroxisome biogenesis disorder 4B
PEX7	Peroxisome biogenesis disorder 9B; Rhizomelic chondrodysplasia punctata, type 1
PFKM	Glycogen storage disease VII
PGAM2	Glycogen storage disease X

GENE	ASSOCIATED DISEASE(S)
PGK1	Phosphoglycerate kinase 1 deficiency
PGM1	Congenital disorder of glycosylation, type It
PHKA1	Muscle glycogenosis
PHKA2	Glycogen storage disease, type IXa1; Glycogen storage disease, type IXa2
PHKB	Glycogen storage disease type IX (Phosphorylase kinase deficiency of liver and muscle)
PHKG2	Glycogen storage disease IXc
PKLR	Pyruvate kinase deficiency
PNPO	Pyridoxamine 5'-phosphate oxidase deficiency
POR	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis; Disordered steroidogenesis due to cytochrome P450 oxidoreductase
PPOX	Porphyria variegata
PPP1R17	Hypercholesterolemia, susceptibility to
PPT1	Ceroid lipofuscinosis, neuronal, 1
PRKAG2	Glycogen storage disease type IX (Cardiac muscle phosphorylase kinase defi ciency)
PSAP	Combined SAP deficiency; Gaucher disease, atypical; Krabbe disease, atypical; Metachromatic leukodystrophy due to SAP-b deficiency
PYGL	Glycogen storage disease VI
PYGM	McArdle disease
RBCK1	Polyglucosan body myopathy 1 with or without immunodeficiency
SGSH	Mucopolysaccharidosis type IIIA (Sanfilippo A)
SI	Sucrase-isomaltase deficiency, congenital
SLC17A5	Salla disease; Sialic acid storage disorder, infantile
SLC22A5	Carnitine deficiency, systemic primary
SLC25A13	Citrullinemia type II
SLC25A15	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome
SLC25A20	Carnitine-acylcarnitine translocase deficiency
SLC25A36	Pyrimidine Nucleotide Carrier 2
SLC2A1	Glucose transporter 1 deficiency
SLC2A2	Fanconi-Bickel syndrome
SLC2A3	Glucose transporter 3; GLUT3 Deficiency
SLC37A4	Glycogen storage disease Ib; Glycogen storage disease Ic
SLC3A1	Cystinuria
SLC3A2	Solute Carrier Family 3 (Activator of dibasic and neutral amino acid transport), Member 2
SLC40A1	Hemochromatosis, type 4
SLC6A19	Hartnup disorder

GENE	ASSOCIATED DISEASE(S)
SLC6A8	Cerebral creatine deficiency syndrome 1
SLC7A7	Lysinuric protein intolerance
SLC7A9	Cystinuria
SLCO1B1	Hyperbilirubinemia, Rotor type, digenic
SLCO1B3	Hyperbilirubinemia, Rotor type, digenic
SMPD1	Niemann-Pick disease, type A; Niemann-Pick disease, type B
SUMF1	Multiple sulfatase deficiency
TAT	Tyrosinemia, type II
TFR2	Hemochromatosis, type 3
TPP1	Ceroid lipofuscinosis, neuronal, 2; Spinocerebellar ataxia, autosomal recessive 7
UGT1A1	Crigler-Najjar syndrome, type I; Crigler-Najjar syndrome, type II; Hyperbilirubinemia, familial transient neonatal
UMPS	Orotic aciduria
UROD	Porphyria cutanea tarda
UROS	Porphyria, congenital erythropoietic

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