

Evertia METABOLIC PANEL

Evertia metabolic panel tests for 13 main categories of metabolic diseases in a **single, detailed panel of 223 genes**. The clinical phenotype is similar between many metabolic diseases, and may include neurological, motor, behavioral, learning, or gastrointestinal symptoms. Treatment is usually symptomatic and involves dietary management, nutritional supplements and enzyme replacement therapies. Investigational therapies are available for several disorders.

3-Methylglutaconic aciduria			
Genes (8)	AR	AD	XL
AUH, DNAJC19, SERAC1, TIMM50, TMEM70	●		
CLPB, OPA3	●	●	
TFAZZIN			●
Disorders tested include: Barth syndrome; 3-Methylglutaconic aciduria type III; 3-Methylglutaconic aciduria type V			

Cerebral creatine deficiency			
Genes (3)	AR	AD	XL
GAMT, GATM	●		
SLC6A8			●
Disorders tested include: Cerebral creatine deficiency syndrome 1; Cerebral creatine deficiency syndrome 2			

Congenital disorders of glycosylation			
Genes (50)	AR	AD	XL
ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, B4GALT1, CAD, CCDC115, COG1, COG2, COG5, COG6, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, FUT8, GMPPA, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35C1, SLC39A8, SRD5A3, STT3B, TMEM165, TMEM199, TUSC3	●		
COG4, DHDDS, GNE, NUS1, STT3A	●	●	
ALG13, SLC35A2, SSR4			●
Disorders tested include: Congenital disorder of glycosylation type Ic; Congenital disorder of glycosylation type Ia			

Fatty acid oxidation disorders			
Genes (21)	AR	AD	XL
ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, CPT1A, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, SLC22A5, SLC25A20	●		
CPT2	●	●	
HSD17B10, TFAZZIN			●
Disorders tested include: Medium chain acyl-CoA dehydrogenase (MCAD) deficiency; Short chain acyl-CoA dehydrogenase (SCHAD) deficiency; Systemic primary carnitine deficiency; Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency			

Glycine encephalopathy			
Genes (5)	AR	AD	XL
AMT, GCSH, GLDC, LIAS, SLC6A9	●		
Disorders tested include: Glycine encephalopathy; Hyperglycinemia (Lactic acidosis and seizures)			

Glycogen storage diseases			
Genes (25)	AR	AD	XL
AGL, ALDOA, ALDOB, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LDHA, PFKM, PGAM2, PHKB, PHKG2, PYGL, PYGM, SLC2A2	●		
PRKAG2		●	
CPT2, SLC37A4	●	●	
LAMP2, PHKA1, PHKA2			●
Disorders tested include: Glycogen storage disease Ia (Von Gierke disease); Glycogen storage disease II (Pompe disease)			

Hyperinsulinemic hypoglycemia			
Genes (7)	AR	AD	XL
HADH	●		
GCK, GLUD1, INSR, SLC16A1		●	
ABCC8, KCNJ11	●	●	
Disorders tested include: Hyperinsulinemic hypoglycemia, familial, types 1, 2, 3, 4, 5, 7; Hyperinsulinism-Hyperammonemia syndrome			

Hyperphenylalaninemia			
Genes (6)	AR	AD	XL
DNAJC12, PAH, PCBD1, PTS, QDPR	●		
GCH1	●	●	
Disorders tested include: Hyperphenylalaninemia A BH4-Deficient; Phenylalanine hydroxylase deficiency			

Lysosomal storage disorders			
Genes (56)	AR	AD	XL
AGA, ARSA, ARSB, ASPA, BTB, CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSK, DHCR7, FUCA1, GAA, GALC, GALNS, GBA, GCDH, GLB1, GM2A, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDUA, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SLC25A15, SMPD1, SUMF1, TPP1, VPS33A	●		
HRAS		●	
GNE, HPD	●	●	
GLA, IDS, LAMP2			●
Disorders tested include: Aspartylglucosaminuria; Fabry disease; Gaucher disease (atypical); Metachromatic leukodystrophy due to SAP-B deficiency; Mucopolysaccharidosis type II (Hunter syndrome); Mucopolysaccharidosis Type III (Sanfilippo A, B, C and D), Niemann-Pick types A, B, C and D; Sandhoff disease; Tay-Sachs disease			

Maple syrup urine disease and DLD deficiency			
Genes (5)	AR	AD	XL
BCKDHA, BCKDHB, DBT, DLD, PPM1K	●		
Disorders tested include: Maple syrup urine disease types Ia, Ib, II, III			

Methylmalonic Acidemia			
Genes (17)	AR	AD	XL
ABCD4, ACSF3, ALDH6A1, CD320, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MTR, MTRR, SUCLA2, SUCLG1	●		
HCFC1			●
Disorders tested include: Methylmalonic acidemia due to methylmalonic-CoA mutase deficiency; Methylmalonic acidemia, cblA type			

Peroxisomal disorders			
Genes (21)	AR	AD	XL
ACOX1, AGPS, AMACR, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH, SCP2	●		
PEX6	●	●	
ABCD1			●
Disorders tested include: Adrenoleukodystrophy; Peroxisome biogenesis disorder 1A, 2A, 2B (Zellweger syndrome)			

Urea cycle disorders			
Genes (8)	AR	AD	XL
ARG1, ASL, ASS1, CPS1, NAGS, SLC25A13, SLC25A15	●		
OTC			●
Disorders tested include: Argininemia, Citrullinemia			

● AR: AUTOSOMAL RECESSIVE

● AD: AUTOSOMAL DOMINANT

● XL: X-LINKED