Evartia METABOLIC PANEL

Evartia 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	●	●	
TAFAZZIN			●

Disorders tested include: Barth syndrome; 3-Methylglutaconic aciduria type III; 3-Methylgutaconic 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, TAFAZZIN			●

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, BTD, 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

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