Evartia®
metabolic test

# 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 disorders

| Genes (8) | AR | AD | XL |
|---|---|---|---|
| AUH, CLPB, DNAJC19, SERAC1, TIMM50, TMEM70 | ● | | |
| OPA3 | ● | ● | |
| TAZ | | | ● |

**Disorders tested include:** Barth syndrome; Costeff syndrome; DCMA syndrome; Megdel syndrome

## 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, ALG9, ALG11, ALG12, B4GALT1, CAD, CCDC115, COG1, COG2, COG5, COG7, COG8, DDOST, DOLK, DPAGT1, DPM1, DPM2, DPM3, FUT8, GMPPA, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SLC35A1, SLC35C1, SLC39A8, SRD5A3, STT3A, STT3B, TMEM165, TMEM199, TUSC3 | ● | | |
| ALG8, COG4, COG6, DHDDS, NUS1, GNE | ● | ● | |
| ALG13, SLC35A2, SSR4 | | | ● |

**Disorders tested include:** ALG6-Congenital disorder of glycosylation (CDG-Ic); PMM2-Congenital disorder of glycosylation (CDG-Ia)

## Glycine encephalopathy

| Genes (5) | AR | AD | XL |
|---|---|---|---|
| AMT, GCSH, GLDC, LIAS, SLC6A9 | ● | | |

**Disorders tested include:** Glycine encephalopathy (Non-ketonic hyperglycinemia); Hyperglycinemia, lactic acidosis, and seizures [Pyruvate dehydrogenase lipoic acid synthetase deficiency (PDHLD)]

## 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, SLC37A4 | ● | | |
| PRKAG2 | | ● | |
| CPT2 | ● | ● | |
| LAMP2, PHKA1, PHKA2 | | | ● |

**Disorders tested include:** Glycogen storage disease Ia (Von Gierke disease); Glycogen storage disease II (Pompe disease)

## 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, TAZ | | | ● |

**Disorders tested include:** Medium chain acyl-CoA dehydrogenase (MCHAD) deficiency; Short chain acyl-CoA dehydrogenase (SCHAD) deficiency; Systemic primary carnitine deficiency; Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency

## Hyperinsulinemic hypoglycemia

| Genes (7) | AR | AD | XL |
|---|---|---|---|
| HADH | ● | | |
| GLUD1, SLC16A1 | | ● | |
| ABCC8, GCK, INSR, KCNJ11 | ● | ● | |

**Disorders tested include:** Hyperinsulinemic hypoglycemia, familial, types 1, 2, 3, 4, 5, 6, 7

## Hyperphenylalaninemia

| Genes (6) | AR | AD | XL |
|---|---|---|---|
| DNAJC12, PAH, PCBD1, PTS, QDPR | ● | | |
| GCH1 | ● | ● | |

**Disorders tested include:** Hyperphenylalaninemia, BH4-deficient, A (6-pyruvoyltetrahydropterin synthase deficiency), Phenylketonuria

## 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, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SLC25A15, SMPD1, SUMF1, TPP1, VPS33A | ● | | |
| HRAS | | ● | |
| GNE, HPD, NAGLU | ● | ● | |
| GLA, IDS, LAMP2 | | | ● |

**Disorders tested include:** Aspartylglucosaminuria; Fabry disease; Gaucher disease; Metachromatic leukodystrophy; 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 disorders

| Genes (5) | AR | AD | XL |
|---|---|---|---|
| BCKDHA, BCKDHB, DBT, DLD, PPM1K | ● | | |

**Disorders tested include:** Maple syrup urine disease types Ia, Ib, II, III

## Methylmalonic Aciduria, Type mut(0)

| Genes (17) | AR | AD | XL |
|---|---|---|---|
| ABCD4, ACSF3, ALDH6A1, CD320, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, SUCLA2, SUCLG1, MTR, MTRR | ● | | |
| HCFC1 | | | ● |

**Disorders tested include:** Methylmalonic aciduria, Type mut(0); Methylmalonic acidemia due to cobalamin A deficiency

## Peroxisomal disorders

| Genes (21) | AR | AD | XL |
|---|---|---|---|
| AGPS, AMACR, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH, SCP2 | ● | | |
| ACOX1, PEX6 | ● | ● | |
| ABCD1 | | | ● |

**Disorders tested include:** Adrenoleukodystrophy; Zellweger syndrome

## Urea cycle disorders

| Genes (8) | AR | AD | XL |
|---|---|---|---|
| ARG1, ASL, ASS1, CPS1, NAGS, SLC25A13, SLC25A15 | ● | | |
| OTC | | | ● |

**Disorders tested include:** Argininemia, Citrin deficiency

● **AR: AUTOSOMAL RECESSIVE** ● **AD: AUTOSOMAL DOMINANT** ● **XL: X-LINKED**

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