NIPD GENETICS

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 acidemia

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: Methylmalonyl-CoA mutase deficiency; 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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