ASPER
METABOLIC DISORDERS

Asper Metabolic Disorders

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Citrin Deficiency	NGS	1	SLC25A13	803
Citrullinemia, type 1	NGS	21	ASS1	803
Fatty Acid Oxidation Disorder	NGS	21	ACAD9, ACADM, ACADS, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, PPARG, SLC22A5, SLC25A20, TAZ	1070
	MLPA	1	SLC22A5	310
Glutaric Aciduria, type 1	Sanger Sequencing	1	GCDH	535
Glutaric Aciduria, type 2	Sanger Sequencing	1	EFTA	535
Glycogen Storage Disease	NGS	25	AGL, ALDOA, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4	1070
Hemochromatosis	NGS	5	HAMP, HFE, HJV, SLC40A1, TFR2	1070
Lysosomal Storage Disease	NGS	50	AGA, ARSA, ARSB, ASAH1, CLN3, CLN5. CLN6, CLN8, CTNS, CTSA, CTSC, CTSD, CTSK, DNAJC5, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, TPP1	1338
	MLPA	1	GLA	310
	MLPA	1	IDS	310
	MLPA	2	NPC1, NPC2	310
	MLPA	1	HEXA	310
Metabolic Myopathy and Rhabdomyolysis	NGS	44	ABHD5, ACAD9, ACADM, ACADVL, AGL, ALDOA, AMPD1, CAV3, CPT2, ENO3, ETFA, ETFB, ETFDH, GAA, GBE1, GYG1, GYS1, HADHA, HADHB, ISCU, LDHA, LPIN1, OPA1, OPA3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, PRKAG2, POLG, POLG2, PYGM, RRM2B, RYR1, SCN4A, SLC22A5, SLC25A20, SUCLA2, TAZ, TK2, TWNK, TYMP	1070
Methylmalonic Aciduria and Homocystinuria	NGS	23	ABCD4, ACSF3, AMN, CBS, CD320, CUBN, GIF, IVD, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, SUCLA2, SUCLG1, TCN1, TCN2	1070
	MLPA	1	MLYCD	310

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Porphyria	NGS	9	ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	1070
	MLPA	7	ALAD, CPOX, FECH, HMBS, PPOX, UROD, UROS	710
Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	Sanger Sequencing	1	ACADS	535
Smith Lemli Opitz Syndrome	Sanger Sequencing	1	DHCR7	525
Urea Cycle Disorder	NGS	10	ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC7A7, SLC25A13, SLC25A15	1070
	MLPA	1	OTC	310
Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency	NGS	1	ACADVL	803
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

Service includes
- DNA extraction
- Genotyping
- Bioinformatic analysis of detected variants
- Detected variants available in Excel format (on request)
- Validation of phenotype/diagnosis associated known or possibly pathogenic variants
- Clinical and biological interpretation
- Results delivery by registered mail (on request)
- TAT 2-9 weeks, excl. WES and WGS 15 weeks

Online ordering, specimen requirements and shipping instructions are available at www.asperbio.com.

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