ASPER
DYSMORPHOLOGY

Asper Dysmorphology

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Brain malformations	NGS	147	ACTB, ACTG1, ADGRG1, AHI1, AKT3, AMPD2, AMT, AP4M1, ARFGEF2, ARL13B, ARX, ASPM, ATP6V0A2, ATR, ATRX, B9D1, B3GALNT2, B4GAT1, CASK, CC2D2A, CCND2, CDK5RAP2, CENPJ, CEP135, CEP290, CEP152, CEP63, CEP41, CHMP1A, CLP1, C5orf42, CREBBP, CUL4B, DCX, DHCR7, DHCR24, DLAT, DLD, DYNC1H1, ETFA, EFTUD2, ERMARD, ETFB, ETFDH, EXOSC3, FAT4, FKRP, FKTN, FLNA, GCSH, GLDC, GMPPB, GPSM2, IER3IP1, INPP5E, ISPD, KIF11, KIF7, KIF2A, KIF1BP, KIF5C, KNL1, LAMA2, LAMB1, LAMC3, LARGE1, MCPH1, MECP2, MKS1, NBN, NDE1, NHEJ1, NPHP1*, OCLN, OFD1, OPHN1, PAFAH1B1, PCNT, PDHA1, PDHB, PDHX, PDP1, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PIEZO2, PIK3R2, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PQBP1, RAB18, RAB3GAP1, RAB3GAP2, RARS2, RELN, RPGRIP1L, RTTN, SEPSECS, SLC12A6, SLC25A19, SNAP29, SRD5A3, SRPX2, STIL, TBC1D20, TCF4, TCTN1, TCTN2, TCTN3, TMEM231, TMEM237, TMEM216, TMEM138, TMEM67, TMEM5, TSEN2, TSEN34, TSEN54, TTC21B, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, VLDLR, VRK1, WASHC5, WDR62, ZEB2, ZNF423 * - 250kb deletion in the heterozygous state is not detectable with the test	1314
Craniosynostosis	NGS, Sanger Sequencing	7	FGFR1, FGFR2, FGFR3, IL11RA, MSX2, RECQL4, TWIST1	1051
	MLPA	1	TWIST1	310
Frazer Syndrome	NGS	3	GRIP1, FREM2, FRAS1	1070
Microcephaly	NGS	24	AP4M1, ASPM, CASK, CDK5RAP2, CENPJ, CEP63, CEP135, CEP152, EFTUD2, IER3IP1, KIF11, KNL1 (CASC5), MCPH1, NDE1, NHEJ1, PAFAH1B1, PCNT, PNKP, POMT1, SLC25A19, STIL, TUBB2B, TUBGCP6, WDR62	1314
Noonan Syndrome	NGS	14	BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1, SPRED1	1051
Osteogenesis Imperfecta	NGS	20	ALPL, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, P3H1 (LEPRE1), PLOD2, PLS3, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, WNT1	1338
Skeletal Ciliopathies	NGS	23	CEP120, COMP, CSPP1, DYNC2H1, DYNC2LI1, EVC, EVC2, FGFR3, IFT43, IFT122, IFT52, IFT80, IFT140, IFT172, KIAA0586, NEK1, TCTEX1D2, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60	1546

ASPER BIOGENE
Vaksali 17A, 50410 Tartu, Estonia
tel +372 7307 295 • fax +372 7307 298
info@asperbio.com • www.asperbio.com

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Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Skeletal Dysplasia	NGS, Sanger Sequencing	15	ALPL, COL2A1, ESCO2, FGFR1, FGFR2, FGFR3, IL11RA, MSX2, RECQL4, ROR2, SLC26A2, SOX9, TRIP11, TWIST1, WNT5A	1051
	MLPA	1	COL2A1	310
	MLPA	1	TWIST1	310
Smith-Lemli-Opitz Syndrome	Sanger Sequencing	1	DHCR7	525
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.

ASPER BIOGENE
Vaksali 17A, 50410 Tartu, Estonia
tel +372 7307 295 • fax +372 7307 298
info@asperbio.com • www.asperbio.com