ASPER NEPHROLOGY

Asper Nephrology

Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Bardet-Biedl Syndrome	23	ALMS1 (excluding exon 8), ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CCDC28B, CEP290, GNAS, IFT27, IFT172, LZTFL1, MKKS, MKS1, PHF6, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP	NGS NGS with CNV	1030 1400
Bartter syndrome	25	ATP6V1B1, BSND, CA2, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, FXYD2, GNA11, HSD11B2, KCNJ1, KCNJ10, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A2, SLC12A3, SLC4A1, SLC4A4, WNK1, WNK4	NGS NGS with CNV	1030 1400
Branchiootorenal Syndrome	3	EYA1, SIX1, SIX5	NGS NGS with CNV	1030 1400
	1	EYA1	MLPA	310
Ciliopathy	123	ACVR2B, ADGRV1, AHI1, AIPL1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, ATXN10, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C2CD3, C2ORF71, C5ORF42, C8ORF37, C21ORF2, C21ORF59, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CDH23, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFTR, CLRN1, CRB1, CSPP1, DCDC2, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYNC2H1, EVC, EVC2, FOXH1, GAS8, GDF1, GLIS2, IFT43, IFT80, IFT122, IFT140, IFT172, INPP5E, INVS, IQCB1, KIAA0586, KIF7, LEFTY2, LRRC6, MCIDAS, MKKS, MKS1, NEK1, NEK8, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PKD2, PKHD1, RPGR, RPGRIP1, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SDCCAG8, SPAG1, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TTC21B, TTC8, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZIC3, ZMYND10, ZNF423	NGS NGS with CNV	1300 1700
Hemolytic Uremic Syndrome	13	ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, THBD	NGS NGS with CNV	1030 1400
Nephronophthisis	20	ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, INVS, IFT172, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	NGS NGS with CNV	1030 1400
Nephrotic Syndrome	15	ACTN4, ARHGDIA, COQ2, COQ8B, DGKE, EMP2, ITGA3, LAMB2, NPHS1, NPHS2, PLCE1, PTPRO, SMARCAL1, WDR73, WT1	NGS NGS with CNV	1030 1400
Polycystic Kidney Disease	23	ALG8, ANKS6, BICC1, COL4A1, DNAJB11, DZIP1L, GANAB, HNF1B, LRP5, MUC1, NOTCH2, OFD1, PKD1, PKD2, PKHD1, PRKCSH, SEC63, SEC61A1, TSC1, TSC2, UMOD, VHL, ZNF423	NGS NGS with CNV	1300 1700

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Primary Ciliary Dyskinesia	35	ARMC4, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFAP298, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, GAS8, LRRC6, MCIDAS, NME8, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, ZMYND10	NGS NGS with CNV	1300 1700
Senior-Loken Syndrome	9	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	NGS NGS with CNV	1030 1400
Whole Exome Sequencing (WES)		Solo sample	NGS NGS with CNV	1300 1700
		Trio samples	NGS NGS with CNV	2600 3400
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS	3635 7449

Version: 2019-06-28

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 10 weeks and WGS 15 weeks

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

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