ASPER
OTOGENETICS

Asper Otogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Alport Syndrome	3	COL4A3, COL4A4, COL4A5	NGS NGS with CNV	1030 1400
	1	COL4A5	MLPA	590
Aminoglycoside-Induced Deafness	1/1	MT-RNR1	Sanger sequencing	87
Branchiootorenal Syndrome	3	EYA1, SIX1, SIX5	NGS NGS with CNV	1030 1400
	1	EYA1	MLPA	310
Jervell and Lange-Nielson Syndrome	2	KCNE1, KCNQ1	Sanger Sequencing	773
Palmoplantar Keratoderma with Deafness	1	GJB2	Sanger Sequencing	257
Pendred Syndrome	1	SLC26A4	Sanger Sequencing MLPA	773 310
Sensorineural Hearing Loss	76	ACTG1, ADGRV1, ATP2B2, ATP6V1B1, BSND, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLRN1, COCH, COL11A2, CRYM, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, ESPN, ESRRB, EYA4, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, HGF, ILDR1, KCNJ10, KCNQ4, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MIR96, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, OTOA, OTOF, PCDH15, PDZD7, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SERPINB6, SIX1, SLC17A8, SLC26A4, SLC26A5, SMPX, STRC, TECTA, TJP2, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP (excluding exon 7), TRMU, USH2A, USH1C, USH1G, WFS1	NGS NGS with CNV	1030 1400
	1	GJB2	Sanger Sequencing	257
	11	GJB2, GJB3, GJB6, KCNQ4, MYO7A, MYO15A, MT-RNR1, MT-TS1, SLC26A4, SLC26A5, TMC1	Targeted regions sequencing by NGS	450
Stickler Syndrome	6	COL2A1, COL11A1, COL11A2, COL9A1, COL9A2, COL9A3	NGS NGS with CNV	1030 1400
	1	COL11A1	MLPA	590
Treacher-Collins Syndrome	3	POLR1C, POLR1D, TCOF1	NGS NGS with CNV	1030 1400
	1	TCOF1	MLPA	310

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Usher Syndrome	20	ABHD12, ADGRV1 (GPR98), CDH23, CIB2, CLRN1, COL4A6, DSPP (excluding exon 5), GIPC3, HARS, KARS, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G, WHRN (DFNB31)	NGS NGS with CNV	1030 1400
	1	USH2A	MLPA	590
	1	PCDH15	MLPA	310
	9	ADGRV1 (GPR98), CDH23, CLRN1, MYO7A, PCDH15, USH2A, USH1C, USH1G, WHRN (DFNB31)	Targeted regions sequencing by NGS	450
Waardenburg Syndrome	6	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10	NGS NGS with CNV	1030 1400
	3	MITF, PAX3, SOX10	MLPA	590
Zellweger Spectrum Disorders	14	PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH	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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