ASPER
OTOGENETICS

# Asper Otogenetics

| Disease/Condition name | Method | No of detectable genes/markers | Genes | Price (EUR) |
|---|---|---|---|---|
| Alport Syndrome | NGS | 3 | COL4A3, COL4A4, COL4A5 | 1030 |
| | MLPA | 1 | COL4A5 | 590 |
| Aminoglycoside-Induced Deafness | Sanger sequencing | 1/1 | MT-RNR1 | 87 |
| Branchiootorenal Syndrome | NGS | 3 | EYA1, SIX1, SIX5 | 1288 |
| | MLPA | 1 | EYA1 | 310 |
| Jervell and Lange-Nielson Syndrome | Sanger Sequencing | 2 | KCNE1, KCNQ1 | 773 |
| Palmoplantar Keratoderma with Deafness | Sanger Sequencing | 1 | GJB2 | 257 |
| Pendred Syndrome | Sanger Sequencing | 1 | SLC26A4 | 773 |
| | MLPA | 1 | SLC26A4 | 310 |
| Sensorineural Hearing Loss | Targeted regions sequencing by NGS | 11 | GJB2, GJB3, GJB6, KCNQ4, MYO7A, MYO15A, MT-RNR1, MT-TS1, SLC26A4, SLC26A5, TMC1 | 450 |
| | NGS | 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 | 1314 |
| | Sanger Sequencing | 1 | GJB2 | 257 |
| Stickler Syndrome | NGS | 6 | COL2A1, COL11A1, COL11A2, COL9A1, COL9A2, COL9A3 | 1030 |
| | MLPA | 1 | COL11A1 | 590 |
| Treacher-Collins Syndrome | NGS | 3 | POLR1C, POLR1D, TCOF1 | 1030 |
| | MLPA | 1 | TCOF1 | 310 |

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|---|---|---|---|---|
| Usher Syndrome | Targeted regions sequencing by NGS | 9 | ADGRV1 (GPR98), CDH23, CLRN1, MYO7A, PCDH15, USH2A, USH1C, USH1G, WHRN (DFNB31) | 450 |
| | NGS | 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) | 1051 |
| | MLPA | 1 | USH2A | 590 |
| | MLPA | 1 | PCDH15 | 310 |
| Waardenburg Syndrome | NGS | 6 | EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10 | 1030 |
| | MLPA | 3 | MITF, PAX3, SOX10 | 590 |
| Zellweger Spectrum Disorders | NGS | 14 | PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH | 1051 |
| Whole Exome Sequencing (WES) | NGS | | Solo sample<br>Trio samples | 1567<br>3100 |
| Whole Genome Sequencing (WGS) | NGS | | Solo sample<br>Trio samples | 3635<br>7449 |

**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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