ASPER
REPROGENETICS

Asper Reprogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Ashkenazi Jewish Diseases	32	ABCC8, AGL, ASPA, BCKHDB, BLM, BRCA1, BRCA2, CFTR, CLRN1, DLD, F11, FANCC, FKTN, GBA, GJB2, G6PC, HEXA, IKBKAP, LCA5, LDLR, LRRK2, MCOLN1, MEFV, MSH2, MSH6, NEB, PCDH15, SERPINA1, SMN1, SMPD1, TMEM216, TOR1A	NGS NGS with CNV	1030 1400
	31	ABCC8, AGL, ASPA, BBS2, BCKDHB, BLM, BRCA1, BRCA2, CFTR, CLRN1, DHDDS, DLD, FAM161A, F11, FANCC, GBA, GJB2, G6PC, HEXA, IKBKAP, LCA5, LDLR, MAK, MCOLN1, MEFV, NEB, PCDH15, SERPINA1, SMPD1, TMEM216, TOR1A	Targeted regions sequencing by NGS	450
Carriership	550	550 genes associated with inherited diseases	NGS NGS with CNV	952 1400
Cystic Fibrosis	1	CFTR	NGS MLPA	360 310
Folate-Dependent Neural Tube Defects	1	MTHFR	Targeted mutation analysis	93
Fragile X Syndrome	1	FMR1	Repeat Expansion/ Fragment Length Analysis	262
Male Factor Infertility	19	CFTR, DDX25, DNAH5, DNAH11, DNAI1, ESR2, FSHB, GNRHR, INSL3, NLRP14, PRDM9, PRM1, PRM2, PRM3, RBMXL2, RXFP2, TEKT2, USP26, UTP14C Klinefelter Syndrome (47XXY) AZF deletions	Targeted regions sequencing by NGS, PCR	624
	19	CFTR, DDX25, DNAH5, DNAH11, DNAI1, ESR2, FSHB, GNRHR, INSL3, NLRP14, PRDM9, PRM1, PRM2, PRM3, RBMXL2, RXFP2, TEKT2, USP26, UTP14C	Targeted regions sequencing by NGS	450
	N/A	Klinefelter Syndrome (47XXY)	PCR	87
	3/8 (+9 for extended analysis)	AZF deletions	PCR	87
Maternal Cell Contamination	NA/6		Fragment Analysis	87
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

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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

* Clinical interpretation is not available

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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