ASPER
ENDOCRINOLOGY

Asper Endocrinology

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Androgen Insensitivity Syndrome	Sanger Sequencing	1	AR	535
	MLPA	1	AR	310
Combined Pituitary Hormone Deficiency	NGS	6	HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1	1070
	MLPA	7	GH1, GHRHR, HESX1, LHX3, LHX4, POU1F1, PROP1	590
Familial Hypocalciuric Hypercalcemia	Sanger Sequencing	1	CASR	535
	MLPA	1	CASR	310
Hypothyroidism and Thyroid Hormone Resistance	NGS	22	DUOX2, DUOXA2, GNAS, HESX1, IYD, NKX2-1, NKX2-5, PAX8, POU1F1, PROP1, SECISBP2, SLC5A5, SLC16A2, SLC26A4, TG, THRA, THRB, TPO, TRHR, TSHB, TSHR	1070
	MLPA	1	GNAS	310
Maturity Onset Diabetes of the Young (MODY)	NGS	15	ABCC8, BLK, CEL, GCK, HNF1A, HNF4A, HNF1B, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1, RFX6, ZFP57	1338
	MLPA	10	CEL, GCK, HNF1A, HNF4A, HNF1B, INS, KLF11, NEUROD1, PAX4, PDX1	590
Thyroid Dyshormonogenesis	NGS	6	DUOX2, DUOXA2, IYD, SLC5A5, TG, TPO	1070
	MLPA	5	FOXE1, NKX2-1, PAX8, TPO, TSHR	590
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