ASPER HEMATOLOGY

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Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Alpha-Thalassemia	2/7	HBA1, HBA2	PCR	91
	2	HBA1, HBA2	MLPA	310
Beta-Thalassemia, Sickle Cell Disease	1	HBB	Sanger Sequencing	257
			MLPA	310
Coagulation Disorders	17	F2, F5, F8, F9, F10, F11, F12, F13A1, FGA, FGB, FGG, GGCX, LMAN1, MCFD2, SERPINC1, VKORC1, VWF	NGS NGS with CNV	1030 1400
Fanconi Anemia	17	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, XRCC2	NGS NGS with CNV	1030 1400
	4	FANCA, FANCB, FANCD2, PALB2	MLPA	1070
Hereditary Sideroblastic Anemia	9	ABCB7, ALAS2, GLRX5, HSPA9, PUS1, SLC19A2, SLC25A38, TRNT1, YARS2	NGS NGS with CNV	1030 1400
Thrombocytopenia	14	ADAMTS13, ANKRD26, CYCS, GATA1, GP1BA, GP1BB, GP9, ITGA2B, ITGB3, MASTL, MPL, MYH9, RUNX1, WAS	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

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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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