ASPER
DERMATOLOGY

Asper Dermatology

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Cutis Laxa	NGS	9	ALDH18A1, ATP6V0A2, ATP7A, EFEMP2, ELN, FBLN5, GORAB, LTBP4, PYCR1	1070
Ehlers-Danlos Syndrome	NGS	37	ADAMTS2, AEBP1, ALDH18A1, ATP7A, ATP6V0A2, B3GALT6, B3GAT3, B4GALT7, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, C1R, C1S, GORAB, DSE, EFEMP2, ELN, FBLN5, FBN1, FKBP14, FLNA, LTBP4, PLOD1, PRDM5, PYCR1, SLC39A13, RIN2, SMAD3, TGFB2, TGFBR1, TGFBR2, TNXB, ZNF469	1070
Epidermolysis Bullosa	NGS	24	CD151, CDSN, CHST8, COL7A1, COL17A1, DSG1, DSP, DST, EXPH5, FERMT1, ITGA3, ITGB4, ITGA6, JUP, KRT1, KRT5, KRT14, LAMA3, LAMB3, LAMC2, MMP1, PLEC, PKP1, TGM5	1070
Hermansky-Pudlak Syndrome	NGS	9	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	1070
Hypotrichosis	NGS	10	APCDD1, CDSN, DSG4, HR, KRT71, KRT74, LIPH, LPAR6, RPL21, SNRPE	1070
Ichthyosis	NGS	38	ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, CDSN, CERS3, CLDN1, CYP4F22, EBP, ERCC2, ERCC3, FLG, GJB2, GJB3, GJB4, GTF2H5, KRT1, KRT2, KRT9, KRT10, LIPN, LOR, MPLKIP, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SLC27A4, SNAP29, SPINK5, ST14, STS, SUMF1, TGM1, TGM5	1070
Melanoma	NGS	3	CDK4, CDKN2A, MITF	1070
	MLPA	4	CDK4, CDKN2A, CDKN2B, MITF	590
Neurofibromatosis	NGS	4	NF1, NF2, SMARCB1, SPRED1	1070
Oculocutaneous Albinism	NGS	17	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	1051
	MLPA	3	GPR143, OCA2, TYR	710
Palmoplantar Keratoderma	NGS	11	AAGAB, AQP5, DSG1, KRT1, KRT9, KRT10, KRT16, KRT6C, SERPINB7, SNAP29, TRPV3	1070
Tuberous Sclerosis	NGS	2	TSC1, TSC2	803
	MLPA	2	TSC1, TSC2	590
Waardenburg Syndrome	NGS	6	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10	1030
	MLPA	3	MITF, PAX3, SOX10	590
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

ASPER BIOGENE
Vaksali 17A, 50410 Tartu, Estonia
tel +372 7307 295 • fax +372 7307 298
info@asperbio.com • www.asperbio.com

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Service includes
- DNA extraction
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- Clinical and biological interpretation
- Results delivery by registered mail (on request)
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Vaksali 17A, 50410 Tartu, Estonia
tel +372 7307 295 • fax +372 7307 298
info@asperbio.com • www.asperbio.com