ASPER
OPHTHALMICS

Asper Ophthalmics

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Achromatopsia	NGS	6	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H	1051
Age-related Macular Degeneration	NGS	21	ABCA4, ARMS2, C2, C3, C9, CCR3, CFB, CFH, CFI, CST3, CXCL8, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, IL6, IL1A, NLRP3, RAX2, TLR4	995*
	Sanger Sequencing	2/3	ARMS2, CFH	87
Aniridia	Sanger Sequencing	1	PAX6	773
Anophthalmia/ Microphthalmia/ Coloboma/Anterior Segment Dysgenesis	NGS	35	ABCB6, ALDH1A3, ASPH, BCOR, B3GLCT, BMP4, CHD7, COL4A1, CYP1B1, EYA1, FOXC1, FOXE3, GDF3, GDF6, HCCS, HESX1, HMGB3, LTBP2, MAB21L2, MFRP, OTX2, PAX6, PITX2, PRSS56, RARB, RAX, SIX6, SLC38A8, SMOC1, SOX2, STRA6, TENM3, VAX1, VSX1, VSX2	1314
Autosomal Dominant Optic Atrophy	Targeted regions sequencing by NGS	1	OPA1	450
	NGS	3	OPA1, OPA3, TMEM126A (autosomal recessive optic atrophy)	1030
Autosomal Dominant Retinitis Pigmentosa	Targeted regions sequencing by NGS	16	CA4, CRX, FSCN2, IMPDH1, KLHL7, NR2E3, NRL, PRPF3, PRPF8, PRPF31, PRPH2, RHO, ROM1, RP1, RP9, TOPORS	450
	NGS	26	AIPL1, BEST1, CA4, CRX, FSCN2, GUCA1B, IMPDH1, KLHL7, NR2E3, NRL, PRKCG, PRPF3, PRPF6, PRPF8, PRPF31, PRPH2, RDH12, RGR, RHO, ROM1, RP1, RP9, RPE65, SEMA4A, SNRNP200, TOPORS	1314
Autosomal Recessive Retinitis Pigmentosa	Targeted regions sequencing by NGS	28	ABCA4, AIPL1, CERKL, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CRB1, EYS, GRK1, IMPG2, LRAT, MERTK, NR2E3, PDE6A, PDE6B, PROM1, RBP3, RDH12, RGR, RHO, RLBP1, RP1, RPE65, SAG, TULP1, USH2A	450
	NGS	58	ABCA4, AIPL1, ARL6, BEST1, C2orf71, C8ORF37, CA4, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, IDH3B, IMPDH1, IMPG2, KLHL7, LRAT, MAK, MERTK, NR2E3, NRL, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF6, PRPF8, PRPF31, PRPH2, RBP3, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR (ORF15 excluded), SAG, SEMA4A, SPATA7, TOPORS, TTC8, TULP1, USH2A, ZNF513	1314
	Sanger Sequencing	1	RPE65	515

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Vaksali 17A, 50410 Tartu, Estonia
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ASPER OPHTHALMICS

Asper Ophthalmics

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Bardet-Biedl Syndrome, McKusick-Kaufman Syndrome, Borjeson-Forssman-Lehmann Syndrome,	Targeted regions sequencing by NGS	16	ALMS1 (excluding exon 8), ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, GNAS, MKKS, MKS1, PHF6, TRIM32, TTC8	450
	NGS	22	ALMS1, ARL6, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CCDC28B, CEP290, GNAS, LZTFL1, MKS1, MKKS, PHF6, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP	1314
Alstrom Syndrome, Albright Hereditary Osteodystrophy				
Cataract	NGS	44	AGK, BCOR, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, EPHA2, EYA1, FTL, FYCO1, GALK1, GCNT2, GJA1, GJA3, GJA8, HSF4, LIM2, LSS, MAF, MIP, NHS, P3H2, PAX6, PITX3, PXDN, SIPA1L3, SIL1, SIX6, SLC16A12, TDRD7, UNC45B, VIM, VSX2	1314
Choroideremia	Sanger Sequencing	1	CHM	773
Cone-Rod Dystrophy	NGS	32	ABCA4, ADAM9, AIPL1, BEST1, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNNM4, C8ORF37, CRX, GNAT2, GUCA1A, GUCY2D, KCNV2, PDE6C, PDE6H, PITPNM3, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR (ORF15 excluded), RPGRIP1, SEMA4A, UNC119	1314
Congenital Stationary Night Blindness	NGS	13	CABP4, CACNA1F, CHM, GNAT1, GRK1, GRM6, NYX, PDE6B, RDH5, RHO, SAG, SLC24A1, TRPM1	1051
Corneal Dystrophy	NGS	21	CHST6, COL5A1, COL17A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, PIKFYVE, PRDM5, SLC4A11, SOD1, ZEB1, ZNF469, TACSTD2, TCF4, TGFBI, UBIAD1, VSX1	1051
Glaucoma	NGS	19	ACVR1, ASB10, BEST1, CANT1, COL18A1, CYP1B1, FOXC1, LMX1B, LOXL1, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, PITX3, SBF2, SLC4A4, WDR36	1051
Leber Congenital Amaurosis	Targeted regions sequencing by NGS	15	AIPL1, CEP290, CRB1, CRX, GUCY2D, IQCB1, LCA5, LRAT, MERTK, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1	450
	NGS	20	AIPL1, CABP4, CEP290 (intronic position c.2991+1655A>G included), CRB1, CRX, GDF6, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, NMNAT1, OTX2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1	1051
Leber Hereditary Optic Neuropathy	RFLP	3/3		176
Norrie Disease	Sanger Sequencing	1	NDP	262

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ASPER OPHTHALMICS

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Norrie Disease	Sanger Sequencing	1	NDP	262
Oculocutaneous Albinism, Ocular Albinism, Hermansky-Pudlak Syndrome, Chediak-Higashi Syndrome	NGS	17	AP3B1, BLOC1S3, BLOC1S6, C10orf11, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	1051
Papillorenal Syndrome	Sanger Sequencing	1	PAX2	525
Retinoblastoma	NGS	1	RB1	960
Stargardt Disease	NGS	1	ABCA4	450
	NGS	4	ABCA4, CNGB3, ELOVL4, PROM1	1030
Usher Syndrome	Targeted regions sequencing by NGS	9	CDH23, CLRN1, DFNB31, GPR98, MYO7A, PCDH15, USH2A, USH1C, USH1G	450
	NGS	20*	ABHD12, CDH23, CIB2, CLRN1, COL4A6, DFNB31, DSPP (excluding exon 5), GIPC3, GPR98, HARS, KARS, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G	1051
Vitelliform Macular Dystrophy	Sanger Sequencing	1	BEST1	395
	NGS	2	BEST1, PRPH2	1030
X-Linked Retinitis Pigmentosa	Targeted regions sequencing by NGS, Sanger Sequencing	2	RP2, RPGR (ORF15 included)	640
	Sanger Sequencing	1	RPGR (ORF15 region only)	257
	NGS, Sanger Sequencing	3	OFD1, RP2, RPGR (ORF15 included)	1051
X-Linked Retinoschisis	Sanger Sequencing	1	RS1	395
Eye Diseases	NGS	277	277 genes associated with different eye diseases	1567
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

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ASPER
REPROGENETICS

Asper Reprogenetics

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Ashkenazi Jewish Diseases	Targeted mutation analysis by APEX	31/117	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	316
	NGS	33	ABCC8, AGL, ASPA, BCKHDB, BLM, BRCA1, BRCA2, CFTR, CLRN1, CYP21A2, DLD, F11, FANCC, FKTN, GBA, GJB2, G6PC, HEXA, IKBKAP, LCA5, LDLR, LRRK2, MCOLN1, MEFV, MSH2, MSH6, NEB, PCDH15, SERPINA1, SMN1, SMPD1, TMEM216, TOR1A	1051
Carriership	Targeted mutation analysis by APEX	20/96	ACADM, ATP7B, CFTR, CHRNE, DHCR7, F2, F5, FAH, FSHR, HBB, HEXA, HFE, GALT, GJB2, MEFV, MTHFR, PAH, SERPINA1, SLC26A4, TCN2	359
	NGS TruSight™ Inherited Disease	550	550 genes associated with inherited diseases	952
Cystic Fibrosis	Targeted mutation analysis by APEX	1/289	CFTR	316
	Targeted regions sequencing by NGS	1	CFTR	450
Fragile X Syndrome	Repeat Expansion/ Fragment Length Analysis	1	FMR1	262
Folate-Dependent Neural Tube Defects	Targeted mutation analysis	1	MTHFR	93
Hutterite Genetic Diseases	Targeted mutation analysis by APEX	28/31	ABCC8, ABCG8, ALPL, BBS2, BCHE, CFTR, CPT1A, DNAJC19, DPH1, EMG1, FKRP, GJB2, MUT, MYO7A, NDUFS4, NPHP1, PCDH15, PROP1, SLC5A5, SLC39A8, TECR, TH, TMEM237, TRIM32, TYR, VLDLR, ZMPSTE24	235*
Male Factor Infertility	Targeted mutation analysis by APEX, PCR	19/291	CFTR, DDX25, DNAH5, DNAH11, DNAI1, ESR2, FSHB, GNRHR, INSL3, NLRP14, PRDM9, PRM1, PRM2, PRM3, RBMXL2, RXFP2, TEKT2, USP26, UTP14C 47XXY AZF deletions	492
	PCR	N/A	47XXY	87
	PCR	3/8 (+9 for extended analysis)	AZF deletions	87

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ASPER
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Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Maternal Cell Contamination	Fragment Analysis	NA/6		87
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

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ASPER ONCOGENETICS

Asper Oncogenetics

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Breast and Ovarian/ Endometrial Cancer	NGS	2	BRCA1, BRCA2	535
	NGS	33	ATM, BARD1, BRCA1, BRCA2, BRIP1, CASP8, CDH1, CHEK2, FANCA, FANCC, FANCD2 (excluding exon 15), FANCE, FANCF, FANCG, KRAS, MAP3K1, MEN1, MLH1, MSH2, MSH6, MRE11A, MUTYH, NBN, PALB2, PTCH1, PTEN, RAD50, RAD51C, RAD51D, STK11, TGFB1, TP53, XRCC2	1051
Cancer Predisposition	NGS	92	AIP, ALK, APC, ATM, BAP1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CYLD, DDB2, DICER1, DIS3L2, EGFR, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2 (excluding exon 15, 16), FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GATA2, GPC3, HNF1A, HRAS, KIT, MAX, MEN1, MET, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NSD1, PALB2, PHOX2B, PRF1, PRKAR1A, PTCH1, PTEN, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RUNX1, SBDS, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCB1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XPC	1051
Familial Adenomatous Polyposis	Sanger Sequencing	1	APC	773
Fanconi Anemia	NGS	17	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, XRCC2	1051
Lynch Syndrome	Sanger Sequencing	1 1 1	MLH1 MSH2 MSH6	515 515 515
	NGS	3	MLH1, MSH2, MSH6	1030
	MLPA	2/NA	MLH1, MSH2	257
Melanoma	NGS	3	CDK4, CDKN2A, MITF	1070
Microsatellite instability	Fragment Analysis	NA/6		175
MUTYH-associated Polyposis	Sanger Sequencing	1	MUTYH	257
	RFLP	1/2	MUTYH	170
Nijmegen Breakage Syndrome	Sanger Sequencing	1 1/1	NBN NBN	788 90

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

ASPER
ONCOGENETICS

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Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Polyposis Syndromes	NGS	6	APC, BMPR1A, MUTYH, PTEN, SMAD4, STK11	1030
Thyroid Cancer	NGS	10	APC, CDC73, DICER1, MEN1, PRKAR1A, PTEN, SDHB, SDHD, RET, TP53	1051
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

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ASPER
CARDIOGENETICS

Asper Cardiogenetics

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy	NGS	14	CTNNA3, DES, DSG2, DSC2, DSP, JUP, LDB3, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN	1070
Brugada Syndrome	NGS	15	CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNE1L, KCNJ8, RANGRF, SCN5A, SCN1B, SCN2B, SCN3B, TRPM4	1314
Familial Hypercholesterolemia	NGS	4	APOB, LDLR, LDLRAP1, PCSK9	1030
Familial Thoracic Aortic Aneurysm and Dissection and Related Syndromes	NGS	12	ACTA2, COL3A1, COL5A1, FBN1, MYH11, MYLK, SLC2A10, SMAD3, TGFB2, TGFBR1, TGFBR2, TGFBR3	1288
Hyperlipidemia	Sanger Sequencing	1/2	APOE	87
Hypertrophic Cardiomyopathy	NGS	28	ACTC1, ACTN2, CALR3, CAV3, CSRP3, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PLN, PRKAG2, SLC25A4, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL	1314
Long QT Syndrome	NGS	14	AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN5A, SCN4B, SNTA1	1051
Noonan Syndrome	NGS	13	BRAF, CBL, HRAS, KAT6B, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1, SPRED11	1051
Pulmonary Arterial Hypertension	NGS, Sanger Sequencing	7	ACVRL1, BMPR2, BMPR1B, CAV1, ENG, KCNK3, SMAD9	1051
Statin-Induced Myopathy	Sanger Sequencing	1/1	SLCO1B1	87
Venous Thrombosis	RFLP	3/4	F2, F5, MTHFR	87
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

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ASPER
OTOGENETICS

Asper Otogenetics

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Alport Syndrome	NGS	3	COL4A3, COL4A4, COL4A5	1030
Aminoglycoside-Induced Deafness	Sanger sequencing	1/1	MT-RNR1	87
Branchiootorenal Syndrome	NGS	3	EYA1, SIX1, SIX5	1288
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
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, 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, GPR98, 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
Treacher-Collins Syndrome	NGS	3	POLR1C, POLR1D, TCOF1	1030
Usher Syndrome	Targeted regions sequencing by NGS	9	CDH23, CLRN1, DFNB31, GPR98, MYO7A, PCDH15, USH2A, USH1C, USH1G	450
	NGS	20	ABHD12, CDH23, CIB2, CLRN1, COL4A6, DFNB31, DSPP, GIPC3, GPR98, HARS, KARS, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G	1051
Waardenburg Syndrome	NGS	6	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10	1030

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ASPER
OTOGENETICS

Asper Otogenetics

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
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 Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

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ASPER
HEMATOLOGY

Asper Hematology

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Alpha-Thalassemia	PCR	2/7	HBA1, HBA2	91
Beta-Thalassemia, Sickle Cell Disease	Sanger Sequencing	1	HBB	257
Fanconi Anemia	NGS	17	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, XRCC2	1051
Thrombocytopenia	NGS	14	ADAMTS13, ANKRD26, CYCS, GATA1, GP1BA, GP1BB, GP9, ITGA2B, ITGB3, MASTL, MPL, MYH9, RUNX1, WAS	1070
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

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Vaksali 17A, 50410 Tartu, Estonia
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ASPER
DYSMORPHOLOGY

Asper Dysmorphology

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Brain malformations	NGS	147	ACTB, ACTG1, ADGRG1, AHI1, AKT3, AMPD2, AMT, AP4M1, ARFGEF2, ARL13B, ARX, ASPM, ATP6V0A2, ATR, ATRX, B9D1, B3GALNT2, B4GAT1, CASK, CC2D2A, CCND2, CDK5RAP2, CENPJ, CEP135, CEP290, CEP152, CEP63, CEP41, CHMP1A, CLP1, C5orf42, CREBBP, CUL4B, DCX, DHCR7, DHCR24, DLAT, DLD, DYNC1H1, ETFA, EFTUD2, ERMARD, ETFB, ETFDH, EXOSC3, FAT4, FKRP, FKTN, FLNA, GCSH, GLDC, GMPPB, GPSM2, IER3IP1, INPP5E, ISPD, KIF11, KIF7, KIF2A, KIF1BP, KIF5C, KNL1, LAMA2, LAMB1, LAMC3, LARGE1, MCPH1, MECP2, MKS1, NBN, NDE1, NHEJ1, NPHP1*, OCLN, OFD1, OPHN1, PAFAH1B1, PCNT, PDHA1, PDHB, PDHX, PDP1, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PIEZO2, PIK3R2, PNKP, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PQBP1, RAB18, RAB3GAP1, RAB3GAP2, RARS2, RELN, RPGRIP1L, RTTN, SEPSECS, SLC12A6, SLC25A19, SNAP29, SRD5A3, SRPX2, STIL, TBC1D20, TCF4, TCTN1, TCTN2, TCTN3, TMEM231, TMEM237, TMEM216, TMEM138, TMEM67, TMEM5, TSEN2, TSEN34, TSEN54, TTC21B, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, VLDLR, VRK1, WASHC5, WDR62, ZEB2, ZNF423 * - 250kb deletion in the heterozygous state is not detectable with the test	1314
Craniosynostosis	NGS, Sanger Sequencing	7	FGFR1, FGFR2, FGFR3, IL11RA, MSX2, RECQL4, TWIST1	1051
Microcephaly	NGS	24	AP4M1, ASPM, CASC5, CASK, CDK5RAP2, CENPJ, CEP63, CEP135, CEP152, EFTUD2, IER3IP1, KIF11, MCPH1, NDE1, NHEJ1, PAFAH1B1, PCNT, PNKP, POMT1, SLC25A19, STIL, TUBB2B, TUBGCP6, WDR62	1314
Noonan Syndrome	NGS	13	BRAF, CBL, HRAS, KAT6B, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, SHOC2, SOS1, SPRED1	1051
Skeletal Ciliopathies	NGS	23	CEP120, COMP, CSPP1, DYNC2H1, DYNC2LI1, EVC, EVC2, FGFR3, IFT43, IFT122, IFT52, IFT80, IFT140, IFT172, KIAA0586, NEK1, TCTEX1D2, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60	1546
Skeletal Dysplasia	NGS	15	ALPL, COL2A1, ESCO2, FGFR1, FGFR2, FGFR3, IL11RA, MSX2, RECQL4, ROR2, SLC26A2, SOX9, TRIP11, TWIST1, WNT5A	1051
Smith-Lemli-Opitz Syndrome	Sanger Sequencing	1	DHCR7	525

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ASPER
DYSMORPHOLOGY

Asper Dysmorphology

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
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)
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ASPER
ENDOCRINOLOGY

Asper Endocrinology

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Androgen Insensitivity Syndrome	Sanger Sequencing	1	AR	535
Combined pituitary hormone deficiency	NGS	6	HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1	1070
Familial Hypocalciuric Hypercalcemia	Sanger Sequencing	1	CASR	535
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	
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
Thyroid Dyshormonogenesis	NGS	6	DUOX2, DUOXA2, IYD, SLC5A5, TG, TPO	1070
Whole Exome Sequencing (WES)	NGS		Solo sample Trio samples	1567 3100
Whole Genome Sequencing (WGS)	NGS		Solo sample Trio samples	4500 8800

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