ASPER
CARDIOGENETICS

Asper Cardiogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Method
Apolipoprotein C-II Deficiency	1	APOC2	Sanger Sequencing
Arrhythmia	40	ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2, CASQ2, CAV3, DES, DSC2, DSG2, DSP, GJA5, GPD1L, HCN4, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ8, KCNQ1, LMNA, NKX2-5, NPPA, PKP2, PLN, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SNTA1, TGFB3, TMEM43	NGS with CNV analysis
Arrhythmogenic Right Ventricular Dysplasia/ Cardiomyopathy	14	CTNNA3, DES, DSC2, DSG2, DSP, JUP, LDB3, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN	NGS with CNV analysis
	7	DSC2, DSG2, DSP, JUP, PKP2, RYR2, TGFB3	MLPA
Brugada Syndrome	15	CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCND3, KCNE3, KCNE5, KCNJ8, RANGRF, SCN5A, SCN1B, SCN2B, SCN3B, TRPM4	NGS with CNV analysis
	1	SCN5A	MLPA
Catecholaminergic Polymorphic Ventricular Tachycardia	6	ANK2, CALM1, CASQ2, KCNJ2, RYR2, TRDN	NGS with CNV analysis
Dilated Cardiomyopathy	44	ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CSRP3, CRYAB, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, EMD, EYA4, GATAD1, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYPN, NEXN, PKP2, PLN, RAF1, RBM20, SCN5A, SGCD, TAZ, TBX20, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL	NGS with CNV analysis
	2	BAG3, TNNT2	MLPA
Ehlers-Danlos Syndrome	38	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, RIN2, SLC39A13, SMAD2, SMAD3, TGFB2, TGFBR1, TGFBR2, TNXB, ZNF469	NGS with CNV analysis
Familial Hypercholesterolemia	4	APOB, LDLR, LDLRAP1, PCSK9	NGS with CNV analysis
	1	LDLR	MLPA
Familial Lipoprotein Lipase Deficiency	1	LPL	NGS
			MLPA
Familial Thoracic Aortic Aneurysm and Dissection and Related Syndromes	19	ACTA2, BGN, COL3A1, COL5A1, FBN1, LOX, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PRKG1, SLC2A10, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, TGFBR3	NGS with CNV analysis
	2	FBN1, TGFBR2	MLPA

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Hereditary Hemorrhagic Telangiectasia	5	ACVRL1, ENG, GDF2, RASA1, SMAD4	NGS with CNV analysis
Hyperlipoproteinemia, type 3	1/2	APOE	Sanger Sequencing
Hyperlipoproteinemia, type 5	1	APOEA5	Sanger Sequencing
Hypertriglyceridemia	6	APOA5, APOC2, GPD1, GPIHBP1, LMF1, LPL	NGS with CNV analysis
Hypertrophic Cardiomyopathy	36	ACTC1, ACTN2, AGK, ANKRD1, CALR3, CAV3, CRYAB, CSRP3, FLNC, GLA, JPH2, LAMP2, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, PDLIM3, PLN, PRKAG2, RAF1, SLC25A4, SOS1, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL	NGS with CNV analysis
	4	BAG3, MYBPC3, MYH7, TNNT2	MLPA
Lecithin Cholesterol Acyltransferase Deficiency	1	LCAT	Sanger Sequencing
Left Ventricular Noncompaction Cardiomyopathy	13	ACTC1, CASQ2, DTNA, LDB3, LMNA, MIB1, MYBPC3, MYH7, PRDM16, TAZ, TNNT2, TPM1, VCL	NGS with CNV analysis
Long QT Syndrome	14	AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN5A, SCN4B, SNTA1	NGS with CNV analysis
	5	KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1	MLPA
Noonan Spectrum Disorders/Rasopathies	37	A2ML1, ACTB, ACTG1, BRAF, CBL, CHD7, DHCR7, ELN, EPHB4, FGD1, HRAS, JAG1, KAT6B, KDM6A, KMT2D, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NCF1, NF1, NOTCH2, NRAS, PPP1CB, PTPN11, RAF1, RAI1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1, TBX1	NGS with CNV analysis
		Deletion/duplication analysis of selected regions	Chromosomal Microarray Analysis
Pulmonary Arterial Hypertension	12	ACVRL1, BMPR2, BMPR1B, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, SMAD4, SMAD9	NGS with CNV analysis
	3	ACVRL1, BMPR2, ENG	MLPA
Short QT Syndrome	5	CACNA1C, CACNB2, KCNH2, KCNJ2, KCNQ1	NGS with CNV analysis
Statin-Induced Myopathy	1/1	SLCO1B1	Sanger Sequencing
Tangier Disease	1	ABCA1	NGS

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Thrombophilia	3/4	F2, F5, MTHFR	RFLP
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

Version: 2021-06-03

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

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ASPER
DERMATOLOGY

Asper Dermatology

Disease/Condition name	No of detectable genes/markers	Genes	Method
Cutis Laxa	10	ALDH18A1, ATP6V0A2, ATP7A, EFEMP2, ELN, FBLN5, GORAB, LTBP4, PYCR1, RIN2	NGS with CNV analysis
Ehlers-Danlos Syndrome	38	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, RIN2, SLC39A13, SMAD2, SMAD3, TGFB2, TGFBR1, TGFBR2, TNXB, ZNF469	NGS with CNV analysis
Epidermolysis Bullosa	26	CD151, CDSN, CHST8, COL7A1, COL17A1, DSG1, DSP, DST, EXPH5, FERMT1, ITGA3, ITGB4, ITGA6, JUP, KRT1, KRT10, KRT14, KRT5, KRT9, LAMA3, LAMB3, LAMC2, MMP1, PLEC, PKP1, TGM5	NGS with CNV analysis
Hermansky-Pudlak Syndrome	11	AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	NGS with CNV analysis
Hypotrichosis	11	APCDD1, CDSN, DSC3, DSG4, HR, KRT71, KRT74, LIPH, LPAR6, RPL21, SNRPE	NGS with CNV analysis
Ichthyosis	47	ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1S1, ATP2A2, ATP2C1, CARD14, CASP14, CDSN, CERS3, CLDN1, CYP4F22, EBP, ELOVL4, ERCC2, ERCC3, FLG, GJB2, GJB3, GJB4, GJB6, GTF2H5, KRT1, KRT2, KRT9, KRT10, LIPN, LORICRIN, MBTPS2, MPLKIP, NIPAL4, NSDHL, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SLC27A4, SNAP29, SPINK5, ST14, STS, SUMF1, TGM1, TGM5	NGS with CNV analysis
Melanoma	11	BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, RB1, TERT, XRCC3	NGS with CNV analysis
	4	CDK4, CDKN2A, CDKN2B, MITF	MLPA
Neurofibromatosis	9	CCND1, LZTR1, NF1, NF2, SMARCB1, SPRED1, TSC1, TSC2, VHL	NGS with CNV analysis
Oculocutaneous Albinism	22	AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CACNA1F, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, MC1R, OCA2, RAB27A, SLC24A5, SLC45A2, TYR, TYRP1	NGS with CNV analysis
	3	GPR143, OCA2, TYR	MLPA
Palmoplantar Keratoderma	11	AAGAB, AQP5, DSG1, KRT1, KRT9, KRT10, KRT16, KRT6C, SERPINB7, SNAP29, TRPV3	NGS with CNV analysis
Tuberous Sclerosis	2	TSC1, TSC2	NGS (incl. selected non-coding variants)
			MLPA

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Waardenburg Syndrome	7	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR	NGS with CNV analysis
	3	MITF, PAX3, SOX10	MLPA
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

Version: 2021-09-20

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
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Disease/Condition name	No of detectable genes/markers	Genes	Method
Brain malformations	261	ACTB, ACTG1, ADGRG1, AHI1, AKT1, AKT3, AMPD2, AMT, ANKLE2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, ARFGEF2, ARL13B, ARX, ASNS, ASPM, ATP6V0A2, ATR, ATRX, B3GALNT2, B4GAT1, B9D1, B9D2, BICD2, C2CD3, C5orf42, CASK, CC2D2A, CCDC22, CCND2, CDC45, CDC6, CDK5RAP2, CDK6, CDON, CDT1, CENPE, CENPF, CENPJ, CEP120, CEP135, CEP152, CEP290, CEP41, CEP63, CHD7, CHMP1A, CIT, CLP1, COL18A1, COL3A1, COPB1, CREBBP, CSPP1, CTNNA2, CUL4B, DAG1, DCX, DDX3X, DHCR24, DHCR7, DISP1, DLAT, DLD, DLL1, DNA2, DYNC1H1, DYRK1A, EFTUD2, EML1, EOMES, EP300, ERCC6, ERMARD, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, EZH2, FAT4, FBXO11, FGF8, FGFR1, FIG4, FKRP, FKTN, FLNA, FOXP2, GCDH, GCSH, GLDC, GMNN, GMPPB, GPSM2, GRIN1, GRIN2B, GSX2, HDAC8, IER3IP1, IFT172, INPP5E, ISPD, KIF11, KIF14, KIF1BP, KIF2A, KIF2A, KIF5C, KIF5C, KIF7, KIFBP, KMT2A, KMT2D, KNL1, LAGE3, LAMA2, LAMB1, LAMC3, LARGE1, LGI1, MCM5, MCPH1, MECP2, MFSD2A, MKS1, MTOR, NACC1, NBN, NCAPD2, NCAPD3, NCAPH, NDE1, NEDD4L, NHEJ1, NIN, NPHP1*, NSMCE2, NUP107, NUP133, NUP37, OCLN, OFD1, OPHN1, ORC1, ORC4, ORC6, OSGEP, PAFAH1B1, PCDH12, PCNT, PDE6D, PDHA1, PDHB, PDHX, PDP1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PHC1, PHGDH, PI4KA, PIEZO2, PIK3CA, PIK3R2, PLK4, PMM2, PNKP, POC1B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP1R12A, PQBP1, PSAT1, PSPH, PTEN, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RARS2, RBBP8, RELN, RPGRIP1L, RTTN, RXYLT1, SASS6, SEC23B, SEPSECS, SHH, SIX3, SLC12A6, SLC25A19, SLC25A46, SLC9A6, SMC1A, SMC3, SNAP29, SRD5A3, SRPX2, STAG2, STAMBP, STIL, STIL, TBC1D20, TCF4, TCTN1, TCTN2, TCTN3, TGIF1, TMEM138, TMEM216, TMEM231, TMEM237, TMEM5, TMEM67, TMX2, TP53RK, TPRKB, TRAIP, TSEN2, TSEN34, TSEN54, TTC21B, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, VLDLR, VRK1, WASHC5, WDFY3, WDR4, WDR62, WDR73, ZEB2, ZIC2, ZNF335, ZNF423 * - 250kb deletion in the heterozygous state is not detectable with the test	NGS with CNV analysis
Cornelia de Lange Syndrome	9	FF4, ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC3, SMC1A, TAF6	NGS with CNV analysis (incl. selected non-coding variants)
	1	NIPBL	MLPA

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Craniosynostosis	24	ALX4, CD96, EFNB1, ERF, FGFR1, FGFR2, FGFR3, FLNA, GLI3, IHH, IL11RA, MSX2, POR, RAB23, RECQL4, SEC24D, SKI, SMAD6, SOX9, TCF12, TGFBR1, TGFBR2, TWIST1, ZIC1	NGS with CNV analysis
	10	ALX1, ALX3, ALX4, EFNB1, FGFR1, FGFR2, FGFR3, MSX2, RUNX2, TWIST1	MLPA
Frazer Syndrome	7	EYA1, GRIP1, FREM1, FREM2, FRAS1, SIX1, SIX5	NGS with CNV analysis
Microcephaly	98	ANKLE2, AP4M1, ASNS, ASPM, ATR, CASK, CDC45, CDC6, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP135, CEP152, CEP63, CHD7, CIT, COPB1, CREBBP, CTNNA2, DHCR7, DHTKD1, DNA2, DONSON, DYRK1A, EFTUD2, EP300, ERCC6, EXOSC3. FBXO11, GMNN, GSX2, HDAC8, IER3IP1, KIF11, KIF14, KIF2A, KIF5C, KIFBP, KMT2A, KNL1, LAGE3, MCM5, MCPH1, MFSD2A, NACC1, NCAPD2, NCAPD3, NCAPH, NDE1, NHEJ1, NIN, NSMCE2, NUP107, NUP133, NUP37, ORC1, ORC4, ORC6, OSGEP, PAFAH1B1, PCDH12, PCNT, PHC1, PHGDH, PLK4, PNKP, POMT1, PSAT1, PSPH, RBBP8, RTTN, SASS6, SLC25A19, SLC9A6, STAMBP, STIL, TP53RK, TPRKB, TRAIP, TRAPPC9, TRMT10A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBG-CP4, TUBGCP6, WDFY3, WDR4, WDR62, WDR73, XRCC4, ZEB2, ZNF335	NGS with CNV analysis (incl. selected non-coding variants)
Noonan Spectrum Disorders/Rasopathies	37	A2ML1, ACTB, ACTG1, BRAF, CBL, CHD7, DHCR7, ELN, EPHB4, FGD1, HRAS, JAG1, KAT6B, KDM6A, KMT2D, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NCF1, NF1, NOTCH2, NRAS, PPP1CB, PTPN11, RAF1, RAI1, RASA1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1, TBX1	NGS with CNV analysis
		Deletion/duplication analysis of selected regions	Chromosomal Microarray Analysis
Osteogenesis Imperfecta	41	ALPL, ANO5, BMP1, CA5A, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CTSK, DSPP, FGFR3, FKBP10, GORAB, IFITM5, KDELR2, LRP5, MBTPS2, MESD, NOTCH2, OSTM1, P3H1, P4HB, PLEKHM1, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SLC26A2, SNX10, SOX9, SP7, SPARC, TCIRG1, TENT5A, TMEM38B, TNFRSF11A, TNFSF11, WNT1	NGS with CNV analysis
Skeletal Ciliopathies	23	CEP120, COMP, CSPP1, DYNC2H1, DYNC2LI1, EVC, EVC2, FGFR3, IFT43, IFT122, IFT52, IFT80, IFT140, IFT172, KIAA0586, NEK1, TCTEX1D2, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60	NGS with CNV analysis

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Skeletal Dysplasia	79	AGPS, ALPL, ARSL, CANT1, CCN6, CHST3, CLCN7, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CRTAP, DDR2, DHCR24, DVL1, DYM, DYNC2H1, DYNC2I2, EBP, ESCO2, EVC, EVC2, FAM111A, FAM20C, FGFR1, FGFR2, FGFR3, FKBP10, FLNA, FLNB, GNPAT, GPC6, HSPG2, IDUA, IFITM5, IFT80, IL11RA, INPPL1, LBR, LIFR, LRP5, MATN3, MMP13, MMP9, MSX2, NEK1, NOTCH2, NPR2, NSDHL, P3H1, PEX7, PLOD2, PPIB, PTDSS1, PTH1R, RECQL4, ROR2, SERPINH1, SHOX, SLC26A2, SLC29A3, SLC35D1, SOX9, TBX5, TCIRG1, TMEM38B, TRIP11, TRPV4, TWIST1, WDR19, WDR35, WNT1, WNT5A, XYLT1	NGS with CNV analysis
		Deletion/duplication analysis of selected regions	Chromosomal Microarray Analysis
	11	ALX1, ALX3, ALX4, COL2A1, EFNB1, FGFR1, FGFR2, FGFR3, MSX2, RUNX2, TWIST1	MLPA
Smith-Lemli-Opitz Syndrome	1	DHCR7	Sanger Sequencing
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

Version: 2021-06-09

Service includes
- DNA extraction
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ASPER ENDOCRINOLOGY

Asper Endocrinology

Disease/Condition name	No of detectable genes/markers	Genes	Method
Androgen Insensitivity Syndrome	1	AR	Sanger Sequencing MLPA
Combined Pituitary Hormone Deficiency	6	HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1	NGS with CNV analysis
	7	GH1, GHRHR, HESX1, LHX3, LHX4, POU1F1, PROP1	MLPA
Congenital Adrenal Hyperplasia	1	CYP21A2	NGS MLPA
Familial Hypocalciuric Hypercalcemia	1	CASR	NGS MLPA
Hypothyroidism and Thyroid Hormone Resistance	23	DUOX2, DUOXA2, GNAS, HESX1, IYD, NKX2-1, NKX2-5, PAX8, POU1F1, PROP1, SECISBP2, SLC5A5, SLC16A2, SLC26A4, STX16, TG, THRA, THRB, TPO, TRHR, TSHB, TSHR	NGS with CNV analysis
	2	GNAS, STX16	MLPA
Kallmann Syndrome	25	ANOS1, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NSMF, POLR3B, PROK2, PROKR2, SEMA3A, SOX10, SPRY4, TAC3, TACR3, WDR11	NGS with CNV analysis
Maturity Onset Diabetes of the Young (MODY)	15	ABCC8, BLK, CEL, GCK, HNF1A, HNF4A, HNF1B, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1, RFX6, ZFP57	NGS with CNV analysis
	10	CEL, GCK, HNF1A, HNF4A, HNF1B, INS, KLF11, NEUROD1, PAX4, PDX1	MLPA
Thyroid Dyshormonogenesis	6	DUOX2, DUOXA2, IYD, SLC5A5, TG, TPO	NGS with CNV analysis
	5	FOXE1, NKX2-1, PAX8, TPO, TSHR	MLPA
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

Version: 2021-01-28

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- Detected variants available in Excel format (on request)
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ASPER HEMATOLOGY

Asper Hematology

Disease/Condition name	No of detectable genes/markers	Genes	Method
Alpha-Thalassemia	2	HBA1, HBA2	MLPA
Beta-Thalassemia, Sickle Cell Disease	1	HBB	Sanger Sequencing/ NGS
			MLPA
Coagulation Disorders	17	F2, F5, F8, F9, F10, F11, F12, F13A1, FGA, FGB, FGG, GGCX, LMAN1, MCFD2, SERPINC1, VKORC1, VWF	NGS with CNV analysis
Fanconi Anemia	17	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, XRCC2	NGS with CNV analysis
	4	FANCA, FANCB, FANCD2, PALB2	MLPA
Hereditary Sideroblastic Anemia	9	ABCB7, ALAS2, GLRX5, HSPA9, PUS1, SLC19A2, SLC25A38, TRNT1, YARS2	NGS with CNV analysis
Neutropenia	26	AP3B1, CSF3R, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GFI1, G6PC3, HAX1, JAGN1, LAMTOR2, LYST, RAB27A, RAC2, SBDS, SLC37A4, SMARCD2, SRP54, TAFAZZIN, USB1, VPS13B, VPS45, WAS, WIPF1	NGS with CNV analysis
Severe Combined Immunodeficiency	44	ADA, AK2, CARD11, CD247, CD40, CD8A, CD3D, CD3E, CD3G, CD40LG, CIITA, CORO1A, DCLRE1C, DOCK8, FOXN1, IKBKB, IL7R, IL2RA, IL2RG, JAK3, LCK, LIG4, MALT1, MTHFD1, NHEJ1, ORAI1, PGM3, PNP, PRKDC, PTPRC, RAC2, RAG1, RAG2, RFX5, RFXANK, RFXAP, RMRP, SLC46A1, STAT5B, STIM1, TBX1, TTC7A, UNC119, ZAP70	NGS with CNV analysis
Thrombocytopenia	14	ADAMTS13, ANKRD26, CYCS, GATA1, GP1BA, GP1BB, GP9, ITGA2B, ITGB3, MASTL, MPL, MYH9, RUNX1, WAS	NGS with CNV analysis
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

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ASPER
METABOLIC DISORDERS

Asper Metabolic Disorders

Disease/Condition name	No of detectable genes/markers	Genes	Method
Citrin Deficiency	1	SLC25A13	NGS
Citrullinemia, type 1	21	ASS1	NGS
Congenital Disorders of Glycolysation	49	ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B4GALT1, B3GLCT, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, GMPPA, GNE, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PGM3, PMM2, RFT1, SEC23B, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TUSC3	NGS with CNV analysis
Fatty Acid Oxidation Disorder	21	ACAD9, ACADM, ACADS, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, PPARG, SLC22A5, SLC25A20, TAZ	NGS with CNV analysis
	2	ACADVL, SLC22A5	MLPA
Glutaric Aciduria, type 1	1	GCDH	Sanger Sequencing
Glutaric Aciduria, type 2	1	EFTA	Sanger Sequencing
Glycogen Storage Disease	25	AGL, ALDOA, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4	NGS with CNV analysis
Hemochromatosis	5	HAMP, HFE, HJV, SLC40A1, TFR2	NGS with CNV analysis
Lysosomal Storage Disease	50	AGA, ARSA, ARSB, ASAH1, CLN3, CLN5. CLN6, CLN8, CTNS, CTSA, CTSC, CTSD, CTSK, DNAJC5, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, TPP1	NGS with CNV analysis
	4	GLA, NPC1, NPC2, SMPD1	MLPA
	2	IDS, HEXA	MLPA

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ASPER
METABOLIC DISORDERS

Asper Metabolic Disorders

Disease/Condition name	No of detectable genes/markers	Genes	Method
Metabolic Myopathy and Rhabdomyolysis	44	ABHD5, ACAD9, ACADM, ACADVL, AGL, ALDOA, AMPD1, CAV3, CPT2, ENO3, ETFA, ETFB, ETFDH, GAA, GBE1, GYG1, GYS1, HADHA, HADHB, ISCU, LDHA, LPIN1, OPA1, OPA3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PNPLA2, PRKAG2, POLG, POLG2, PYGM, RRM2B, RYR1, SCN4A, SLC22A5, SLC25A20, SUCLA2, TAZ, TK2, TWNK, TYMP	NGS with CNV analysis
Methylmalonic Aciduria and Homocystinuria	23	ABCD4, ACSF3, AMN, CBLIF, CBS, CD320, CUBN, IVD, LMBRD1, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, SUCLA2, SUCLG1, TCN1, TCN2	NGS with CNV analysis
	4	MLC1, MLYCD, D2HGDH, L2HGDH	MLPA
Porphyria	9	ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	NGS with CNV analysis
	7	ALAD, CPOX, FECH, HMBS, PPOX, UROD, UROS	MLPA
Short-Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	1	ACADS	Sanger Sequencing
Smith Lemli Opitz Syndrome	1	DHCR7	Sanger Sequencing
Urea Cycle Disorder	10	ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC7A7, SLC25A13, SLC25A15	NGS with CNV analysis
	1	OTC	MLPA
Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency	1	ACADVL	NGS
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

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ASPER NEPHROLOGY

Asper Nephrology

Disease/Condition name	No of detectable genes/markers	Genes	Method
Bardet-Biedl Syndrome	23	ALMS1 (excluding exon 8), ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CCDC28B, CEP290, GNAS, IFT27, IFT172, LZTFL1, MKKS, MKS1, PHF6, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP	NGS NGS with CNV
Bartter syndrome	25	ATP6V1B1, BSND, CA2, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, FXYD2, GNA11, HSD11B2, KCNJ1, KCNJ10, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A2, SLC12A3, SLC4A1, SLC4A4, WNK1, WNK4	NGS NGS with CNV
Branchiootorenal Syndrome	3	EYA1, SIX1, SIX5	NGS NGS with CNV
	1	EYA1	MLPA
Ciliopathy	123	ACVR2B, ADGRV1, AHI1, AIPL1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, ATXN10, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C2CD3, C2ORF71, C5ORF42, C8ORF37, C21ORF2, C21ORF59, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC28B, CCDC39, CCDC40, CCDC65, CCNO, CDH23, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFTR, CLRN1, CRB1, CSPP1, DCDC2, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYNC2H1, EVC, EVC2, FOXH1, GAS8, GDF1, GLIS2, IFT43, IFT80, IFT122, IFT140, IFT172, INPP5E, INVS, IQCB1, KIAA0586, KIF7, LEFTY2, LRRC6, MCIDAS, MKKS, MKS1, NEK1, NEK8, NME8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PKD2, PKHD1, RPGR, RPGRIP1, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SDCCAG8, SPAG1, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TTC21B, TTC8, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZIC3, ZMYND10, ZNF423	NGS NGS with CNV
Hemolytic Uremic Syndrome	13	ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, THBD	NGS NGS with CNV
Nephronophthisis	20	ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, INVS, IFT172, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	NGS NGS with CNV
Nephrotic Syndrome	15	ACTN4, ARHGDIA, COQ2, COQ8B, DGKE, EMP2, ITGA3, LAMB2, NPHS1, NPHS2, PLCE1, PTPRO, SMARCAL1, WDR73, WT1	NGS NGS with CNV
Polycystic Kidney Disease	23	ALG8, ANKS6, BICC1, COL4A1, DNAJB11, DZIP1L, GANAB, HNF1B, LRP5, MUC1, NOTCH2, OFD1, PKD1, PKD2, PKHD1, PRKCSH, SEC63, SEC61A1, TSC1, TSC2, UMOD, VHL, ZNF423	NGS NGS with CNV

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Disease/Condition name	No of detectable genes/markers	Genes	Method
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
Senior-Loken Syndrome	9	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	NGS NGS with CNV
Whole Exome Sequencing (WES)		Solo sample	NGS NGS with CNV
		Trio samples	NGS NGS with CNV
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS

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ASPER
NEUROGENETICS

Asper Neurogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Method
Alzheimer Disease	1/2	APOE	Sanger Sequencing
Amyotrophic Lateral Sclerosis	22	ALS2, ANG, CHCHD10, CHMP2B, ERBB4, FIG4, FUS, MATR3, OPTN, PFN1, SETX, SIGMAR1, SOD1, SPART, SPG11, SQSTM1, TARDBP, TBK1, TUBA4A, UBQLN2, VAPB, VCP	NGS with CNV analysis
Autism Spectrum Disorders	77	ADNP, ADSL, ANKRD11, ARX, AVPR1A, BCL11A, BRAF, CACNA1C, CC2D1A, CDKL5, CHD2, CHD7, CNOT3, CNTN6, CNTNAP2, COL4A3BP , C12ORF4, CREBBP, CSNK2A1, DHCR7, EHMT1, EN2, FBXO11, FOXG1, FOXP1, FOXP2, GABRB3, HDAC8, HOXA1, HPRT1, KMT5B, MAGEL2, MBOAT7, MECP2, MED12, MID1, NHS, NIPBL, NLGN3, NLGN4X, NRXN1, NSD1, NTNG1, OXTR, PCDH19, PDE8B, POGZ, PTCHD1, PTEN, PTPN11, PQBP1, RAD21, RAI1, RELN, RPL10, SCN1A, SCN2A, SETD2, SHANK2, SHANK3, SLC6A4, SLC6A8, SLC9A9, SMC3, SMC1A, SNAP25, TBL1XR1, TBR1, TCF4, TCF20, TRIP12, TSC1, TSC2, UBE3A, VPS13B, WASF1, ZEB2	NGS with CNV analysis (incl. selected non-coding variants)
Brunner Syndrome	1	MAOA	NGS
Charcot-Marie-Tooth Disease	78	AARS1, ABHD12, AIFM1, ARHGEF10, ARSA, ATP1A1, BSCL2, C12orf65, COX6A1, CTDP1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, KARS1, KIF1A, KIF5A, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NGF, NTRK1, PDK3, PLEKHG5, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SCN9A, SEPTIN9, SETX, SH3TC2, SLC5A7, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TYMP, VCP, WARS1, WNK1, YARS1	NGS with CNV analysis (incl. selected non-coding variants)
	4	GJB1, KIF1B, MPZ, PMP22	MLPA
	2	MFN2, MPZ	MLPA
	12	EGR2, GARS1, GDAP1, HSPB1, HSBP8, MTMR2, NEFL, PRX, RAB7A, SBF2, SH3TC2, SPTLC1	MLPA
Congenital Muscular Dystrophy	29	B3GALNT2, B4GAT1, CHKB, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DMD, DPM1, DPM2, DPM3, EMD, FKRP, FKTN, GMPPB, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1, SELENON, TCAP	NGS with CNV analysis (incl. selected non-coding variants)
Congenital Myasthenic Syndrome	23	AGRN, ALG2, ALG14, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMB2, LRP4, MUSK, PLEC, PREPL, RAPSN, SCN4A, SLC5A7, SNAP25, SYT2	NGS with CNV analysis

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Congenital Myopathy and Distal Myopathy	42	ACTA1, ANO5, BAG3, BIN1, CAV3, CCDC78, CFL2, CNTN1, COL6A1, COL6A3, COL12A1, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FLNC, GNE, KLHL40, KLHL41, LDB3, LMOD3, MATR3, MEGF10, MICU1, MTM1, MTMR14, MYF6, MYH7, MYOT, NEB, RYR1, SELENON, STAC3, SQSTM1, TIA1, TNNT1, TPM2, TPM3, TTN, VCP	NGS with CNV analysis
	2	MTM1, MTMR1	MLPA
Cornelia de Lange Syndrome	9	AFF4, ANKRD11, HDAC8, KMT2A, NIPBL, RAD21, SMC3, SMC1A, TAF6	NGS with CNV analysis (incl. selected non-coding variants)
	1	NIPBL	MLPA
Craniosynostosis	24	ALX4, CD96, EFNB1, ERF, FGFR1, FGFR2, FGFR3, FLNA, GLI3, IHH, IL11RA, MSX2, POR, RAB23, RECQL4, SEC24D, SKI, SMAD6, SOX9, TCF12, TGFBR1, TGFBR2, TWIST1, ZIC1	NGS with CNV analysis
	10	ALX1, ALX3, ALX4, EFNB1, FGFR1, FGFR2, FGFR3, MSX2, RUNX2, TWIST1	MLPA
Dystonia	41	ACTB, ADCY5, ANO3, ARSA, ATM, ATP1A3, ATP7B, CACNA1B, CIZ1, COL6A3, DRD2, GCDH, GCH1, GNAL, GNAO1, HPCA, KCNMA1, KCTD17, KMT2B, MECR, PANK2, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, RELN, SGCE, SLC2A1, SLC6A3, SLC25A1, SLC30A10, SLC39A14, SPR, TAF1, TBCE, TH, THAP1, TIMM8A, TOR1A, TUBB4A	NGS with CNV analysis (incl. selected non-coding variants)
	8	ATP1A3, GCH1, PRKRA, PRRT2, SGCE, TH, THAP1, TOR1A	MLPA
Epilepsy	213	AARS1, ABAT, ACTL6B, ACY1, ADAM22, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG3, ALG13, AMT, AP3B2, ARHGEF9, ARHGEF15, ARX, ASAH1, ATP1A2, ATP1A3, ATP6AP2, ATP6V1A, ATRX, BRAT1, CACNA1A, CACNA1D, CACNA2D2, CACNA1E, CACNA1H, CACNB4, CASK, CDC42, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLN3, CLN8, CNPY3, CNTN2, CNTNAP2, C12orf57, CPA6, CRH, CSTB, CTSF, CYFIP2, DENND5A, DEPDC5, DHFR, D2HGDH, DNAJC5, DNM1, DNM1L, DOCK7, DYRK1A, EEF1A2, EFHC1, EPM2A, ETHE1, FGF12, FLNA, FOLR1, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GLDC, GNAO1, GOSR2, GPHN, GRIN1, GRIN2A, GRIN2B, GRIN2D, HCN1, HNRNPU, HUWE1, IER3IP1, ITPA, IQSEC2, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, KIF1A, KIF5C, LGI1, LIAS, MBD5, MCCC1, MDH2, MECP2,	NGS with CNV analysis (incl. selected non-coding variants)

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Epilepsy	212	MEF2C, MFSD8, MOCS1, MOCS2, MTHFR, MTOR, NACC1, NECAP1, NEUROD2, NEXMIF, NGLY1, NHLRC1, NOL3, NPRL2, NR2F1, NRXN1, PCDH19, PHACTR1, PIK3R2, PIGA, PIGB, PIGN, PIGO, PIGP, PIGQ, PIGT, PLCB1, PLPBP, PNKP, PNPO, POLG, PPP3CA, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRRT2, PURA, QARS, RBFOX1, RBFOX3, RELN, RNASEH2B, ROGDI, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SERAC1, SERPINI1, SIK1, SLC1A2, SLC12A5, SLC13A5, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC35A3, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMC1A, SNAP25, SNIP1, SNX27, SPATA5, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SYP, SZT2, TBCD, TBC1D24, TBCE, TBCK, TCF4, TPP1, TRAK1, TSC1, TSC2, TTC19, TUBB3, UBA5, UBE3A, WASF1, WDR45, WWOX, ZDHHC9, ZEB2	NGS with CNV analysis (incl. selected non-coding variants)
	10	CHRNA4, CHRNB2, EPM2A, KCNQ1, KCNQ3, NHLRC1, PCDH19, SCN1A, SLC2A1, STXBP1	MLPA
Familial Hemiplegic Migraine	15	ALPK1, ATP1A2, ATP1A3, CACNA1A, CSNK1D, KCNK6, KCNK18, NOTCH3, PNKD, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1, SLC4A4	NGS with CNV analysis
Fragile X Syndrome	1	FMR1	Repeat Expansion/ Fragment Length Analysis
Frontotemporal Dementia	22	ABCA7, APOE, APP, CHMP2B, CSF1R, FUS, GRN, ITM2B, MAPT, PRNP, PSEN1, PSEN2, SIGMAR1, SNCA, SORL1, TARDBP, TBK1, TREM2, TUBA4A, UBE3A, UBQLN2, VCP	NGS with CNV analysis (incl. selected non-coding variants)
	3	CRHR1, GRN, MAPT	MLPA
Hereditary Ataxia	148	ABCB7, ABHD12, ACO2, AFG3L2, AHI1, ANO10, APTX, ATCAY, ATG5, ATM, ATP1A2, ATP1A3, ATP8A2, ATP2B3, CA8, CACNA1A, CACNA1G, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CHP1, CHRNA4, CLCN2, CLN5, COA7, COQ2, COQ4, COQ6, COQ7, COQ9, COQ8A, C9orf72, CPLANE1, CWF19L1, CYP27A1, DARS2, DEPDC5, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, FLVCR1, FXN, GDAP2, GOSR2, GRID2, GRM1, GSS, GSX2, IFRD1, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KCNMA1, KCNQ2, KIF1C, LAMA1, MAN2B1, MARS2, MECR, MME, MTCL1, MTPAP, NKX6-2, NOP56, NPC1, NPC2, OFD1, OPHN1, PAX6, PCDH12, PDSS1, PDSS2, PDYN, PEX7, PHYH, PNKD, PNKP, PNPLA6, POLG, PRKCG, RUBCN, PIK3R5, PLD3, PMPCA, POLR3A, POLR3B,	NGS with CNV analysis (incl. selected non-coding variants)

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Hereditary Ataxia	148	PRRT2, PTF1A, PUM1, RNF216, SACS, SAMD9L, SCN1A, SCN2A, SCN8A, SCYL1, SERAC1, SETX,-SIL1, SLC1A3, SLC16A2, SLC2A1, SLC25A46, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG7, SPTBN2, SQSTM1, STUB1, SYNE1, SYT14, TDP2, TRPC3, TSFM, TTBK2, TDP1, TGM6, THG1L, TMEM231, TMEM240, TPP1, TTC19, TTPA, TUBB4A, TWNK, TXN2, UBA5, UBR4, VAMP1, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1	NGS with CNV analysis (incl. selected non-coding variants)
	4	ATXN1, ATXN2, ATXN3, CACNA1A	Repeat Expansion Analysis
	13	ATXN1, ATXN2, ATXN3, ATXN7, ATXN8, ATXN10, ATN1, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP	Repeat Expansion Analysis
	37	Mitochondrial Genome	NGS
		Deletion/duplication analysis of selected regions	Chromosomal Microarray Analysis
	1	FMR1	Repeat Expansion/ Fragment Length Analysis
Hereditary Spastic Paraplegia	38	ATL1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, B4GALNT1, BSCL2, BTD, CYP7B1, CYP2U1, DDHD2, ERLIN2, FA2H, GBA2, GBE1, GJC2, HSPD1, KIF1A, KIF5A, L1CAM, NIPA1, PAH, PLP1, PNPLA6, REEP1, RTN2, SLC16A2, SPART, SPAST, SPG7, SPG11, SPG21, TECPR2, TH, VPS37A, WASHC5, ZFYVE26	NGS with CNV analysis (incl. selected non-coding variants)
	5	ATL1, NIPA1, SPAST, SPG7, REEP1	MLPA
	1/1	MT-ATP6	Sanger Sequencing
Joubert Syndrome	29	AHI1, ARL13B, B9D1, B9D2, C5orf42, CC2D2A, CEP290, CEP41, CEP104, CSPP1, INPP5E, KIF7, KIAA0556, KIAA0586, MKS1, NPHP1, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM67, TMEM237, TTC21B, ZNF423	NGS with CNV analysis
Leukodystrophy and Leukoencephalopathy	40	ABCD1, ADAR, AIMP1, ARSA, ASPA, CLCN2, CSF1R, DARS2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FAM126A, FOLR1, GALC, GFAP, GJC2, HEPACAM, HSPD1, HTRA1, L2HGDH, LMNB1, MLC1, NOTCH3, PLP1, POLR3A, POLR3B, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SOX10, SUMF1, TREX1, TUBB4A	NGS with CNV analysis

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Disease/Condition name	No of detectable genes/markers	Genes	Method	Price (EUR)
Leukodystrophy and Leukoencephalopathy	10	ABCD1, ASPA, D2HGDH, L2HGDH, LMNB1, MLC1, MLYCD, NOTCH3, PLP1, SLC6A8	MLPA	
Limb-Girdle Muscular Dystrophy	40	ANO5, BVES, CAPN3, CAV3, CRPPA, DAG1, DES, DMD, DNAJB6, DOK7, DYSF, FKRP, FKTN, GAA, GMPPB, HNRNPDL, LAMA2, LIMS2, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SELENON, SGCA, SGCB, SGCD, SGCG, SMCHD1, SYNE1, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	NGS with CNV analysis (incl. selected non-coding variants)	
	3	ANO5, CAPN3, DYSF	MLPA	
	9	FKRP, LCAV3, MNA, MYOT, SGCA, SGCB, SGCD, SGCG, ZMPSTE24	MLPA	
Malignant Hyperthermia	3	CACNA1S, RYR1, STAC3	NGS with CNV analysis	
Menkes Disease	1	ATP7A	NGS	
	1	ATP7A	MLPA	
Microcephaly	98	ANKLE2, AP4M1, ASNS, ASPM, ATR, CASK, CDC45, CDC6, CDK5RAP2, CDK6, CDT1, CENPE, CENPJ, CEP135, CEP152, CEP63, CHD7, CIT, COPB1, CREBBP, CTNNA2, DHCR7, DHTKD1, DNA2, DONSON, DYRK1A, EFTUD2, EP300, ERCC6, EXOSC3. FBXO11, GMNN, GSX2, HDAC8, IER3IP1, KIF11, KIF14, KIF2A, KIF5C, KIFBP, KMT2A, KNL1, LAGE3, MCM5, MCPH1, MFSD2A, NACC1, NCAPD2, NCAPD3, NCAPH, NDE1, NHEJ1, NIN, NSMCE2, NUP107, NUP133, NUP37, ORC1, ORC4, ORC6, OSGEP, PAFAH1B1, PCDH12, PCNT, PHC1, PHGDH, PLK4, PNKP, POMT1, PSAT1, PSPH, RBBP8, RTTN, SASS6, SLC25A19, SLC9A6, STAMBP, STIL, TP53RK, TPRKB, TRAIP, TRAPPC9, TRMT10A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP4, TUBGCP6, WDFY3, WDR4, WDR62, WDR73, XRCC4, ZEB2, ZNF335	NGS with CNV analysis (incl. selected non-coding variants)	
Mitochondrial Diseases	37	Mitochondrial Genome	NGS	
	217	AARS2, ABCB7, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACO2, AFG3L2, AGK, AIFM1, ALAS2, APTX, ATPAF2, ATP7B, ATP5F1A, ATP5F1E, AUH, BCS1L, BOLA3, CARS2, C12orf65, C19orf12, CISD2, COA3, COA5, COA6, COA8 (APOPT1), COQ2, COQ6, COQ9, COQ8A, COX10, COX14, COX15, COX20, COX8A, COX6A1, COX6A2, COX6B1, COX4I1, CPT1A, CPT2, CYC1, DARS2, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBP1,	NGS with CNV analysis	

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Mitochondrial Diseases	217	FBXL4,FDX2 (FDX1L), FH, FLAD1, FOXRED1, G6PC, GAMT, GARS1 (GARS), GATM, GCDH, GFER, GFM1, GFM2, GLRX5, GTPBP3, GYS2, HARS2, HLCS, HADH, HADHA, HMGCL, HSPD1, HTRA2, IARS2, IBA57, ISCA2, ISCU, LAMP2, LARS1, LARS2, LIAS, LIPT1, LRPPRC, LYRM4, LYRM7, MARS2, MFF, MFN2, MGME1, MICU1, MPC1, MPV17, MRPL12, MRPL3, MRPL44, MRPS7, MRPS16, MRPS22, MTFMT, MTO1, MTPAP, NARS2, NDUFA1, NDUFA4, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB3, NDUFB9, NDUFB11, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NR2F1, NUBPL, OPA1, OPA3, OTC, PARS2, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PET100, PNPT1, POLG,POLG2, PUS1, RARS2, REEP1, RMND1, RNASEH1, RRM2B, SARS2, SCO1, SCO2, SDHA, SDHAF1,SERAC1, SETX, SFXN4, SLC6A8, SLC19A3, SLC25A3, SLC25A20, SLC25A4, SLC25A26, SLC25A38, SLC25A46, SLC37A4, SOD1, SPAST, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TAZ, TFAM, TIMM8A, TK2, TMEM126A, TMEM126B, TMEM70, TPK1, TRIT1, TRMT10C, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS2, WFS1, YARS2	NGS with CNV analysis
	10	DGUOK, MPV17, POLG, POLG2, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TWNK	MLPA
Mitochondrial Encephalomyopathy Lactic Acidosis and Stroke-Like Episodes (MELAS Syndrome)	1/1	MT-TL1	Sanger Sequencing
Neurodegeneration with Brain Iron Accumulation	10	ATP13A2, COASY, C19orf12, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45	NGS with CNV analysis
	2	PANK2, PLA2G6	MLPA
Parkinson's Disease	40	ADH1C, ATP1A3, ATP13A2, ATP6AP2, ATXN2, CHCHD2, DCTN1, DNAJC6, DNAJC13, EIF4G1, FBXO7, FTL, GBA, GCH1, GIGYF2, HTRA2, LRRK2, MAPT, PARK7, PINK1, PLA2G6, PODXL, PRKN, PRKRA, PTRHD1, RAB39B, SLC6A3, SLC30A10, SNCA, SNCB, SPG11, SPR, SYNJ1, TAF1, TBP (excluding exon 3), TH, TMEM230, UCHL1, VPS35, VPS13C	NGS with CNV analysis
	8	ATP13A2, GCH1, LRRK2, PARK7, PINK1, PRKN, SNCA, UCHL1	MLPA

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Paroxysmal Dyskinesia	5	ADCY5, KCNMA1, PNKD, PRRT2, SLC2A1	NGS with CNV analysis
Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	1	ACADS	Sanger Sequencing
Smith-Lemli-Opitz Syndrome	1	DHCR7	Sanger Sequencing
Tuberous Sclerosis	2	TSC1, TSC2	NGS (incl. selected non-coding variants)
	2	TSC1, TSC2	MLPA
Very Long Chain Acyl-Coenzyme A Dehydrogenase (VLCAD) Deficiency	1	ACADVL	NGS
Wilson Disease	1	ATP7B	NGS
	1	ATP7B	MLPA
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

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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

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ASPER ONCOGENETICS

Asper Oncogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Method
Breast and Ovarian/ Endometrial Cancer	23	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, MEN1, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, PALB2, PTCH1, PTEN, RAD50, RAD51C, RAD51D, STK11, TP53, XRCC2	NGS with CNV analysis
	2	BRCA1, BRCA2	NGS
	3	BRCA1, BRCA2, CHEK2	MLPA
Cancer Predisposition	102	AIP, ALK, APC, ATM, BAP1, BARD1, 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, HOXB13, HRAS, KIT, MAX, MEN1, MET, MITF, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NSD1, NTHL1, PALB2, PHOX2B, POLD1, PRF1, PRKAR1A, PRSS1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RUNX1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCB1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XPC, XRCC2	NGS with CNV analysis
Familial Adenomatous Polyposis	1	APC	NGS MLPA
Fanconi Anemia	17	BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2 (excluding exons 15, 16), FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2, RAD51C, SLX4, XRCC2	NGS with CNV analysis
	4	FANCA, FANCB, FANCD2, PALB2	MLPA
Lynch Syndrome	3	MLH1, MSH2, MSH6	NGS with CNV analysis
	1 1 1	MLH1 MSH2 MSH6	Sanger Sequencing
	2	MLH1, MSH2	MLPA
	1	MSH6	MLPA
Melanoma	11	BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, RB1, TERT, XRCC3	NGS with CNV analysis
	4	CDK4, CDKN2A, CDKN2B, MITF	MLPA
Microsatellite instability	NA/7		Fragment Analysis
MUTYH-associated Polyposis	1	MUTYH	Sanger Sequencing
	3	GREM1, MUTYH, SCG5	MLPA
	1/2	MUTYH	RFLP

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Nijmegen Breakage Syndrome	1	NBN	NGS
	1/1	NBN	Sanger Sequencing
Polyposis Syndromes	6	APC, BMPR1A, MUTYH, PTEN, SMAD4, STK11	NGS with CNV analysis
	4	BMPR1A, PTEN, SMAD4, STK11	MLPA
Prostate Cancer	13	ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PALB2, RNASEL, TP53	NGS with CNV analysis
Renal Cancer	25	BAP1, CDC73, CDKN1C, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, HNF1A, MET, MLH1, MSH2, MSH6, PTEN, REST, SDHB, SDHC, SDHD, SMARCB1, TP53, TSC1, TSC2, VHL, WT1	NGS with CNV analysis
Thyroid Cancer	10	APC, CDC73, DICER1, MEN1, PRKAR1A, PTEN, RET, SDHB, SDHD, TP53	NGS with CNV analysis
	4	MEN1, SDHB, SDHC, SDHD	MLPA
Von Hippel-Lindau Disease	1	VHL	Sanger Sequencing
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

Version: 2021-09-20

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)
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ASPER OPHTHALMICS

Asper Ophthalmics

Disease/Condition name	No of detectable genes/markers	Genes	Method
Achromatopsia	6	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H	NGS with CNV analysis
Age-related Macular Degeneration	2/3	ARMS2, CFH	Sanger Sequencing
Aniridia	1	PAX6	Sanger Sequencing
	3	PAX6, SOX2, WT1	MLPA
Anophthalmia/ Microphthalmia/ Coloboma/Anterior Segment Dysgenesis	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	NGS with CNV analysis
Autosomal Dominant Retinitis Pigmentosa	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	NGS with CNV analysis
	4	IMPDH1, PRPF31, RHO, RP1	MLPA
Autosomal Recessive Retinitis Pigmentosa	57	ABCA4, AIPL1, ARL6, BEST1, C2orf71, C8ORF37, CA4, CERKL, CLRN1, CNGA1, CNGB1, CNGB3, CRB1, CRX, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, IDH3B, IMPDH1, IMPG2, 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	NGS with CNV analysis
	7	EYS, IMPDH1, PRPF31, RHO, RP1, RP2, RPGR	MLPA
	1	RPE65	Sanger Sequencing
Bardet-Biedl Syndrome, McKusick-Kaufman Syndrome, Borjeson-Forssman-Lehmann Syndrome, Alstrom Syndrome, Albright Hereditary Osteodystrophy	25	ALMS1 (excluding exon 8), ARL6, BBIP1, BBS1, BBS2, BBS4, BBS5, BBS7, BBS9, BBS10, BBS12, CCDC28B, CEP290, GNAS, IFT27, IFT172, LZTFL1, MKKS, MKS1, PHF6, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP	NGS with CNV analysis

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Cataract	58	AGK, BCOR, BEST1, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGA, CRYGB, CRYGC, CRYGD, CRYGS, CTDP1, EPHA2, EYA1, FBN1, FTL, FYCO1, GALK1, GCNT2, GJA1, GJA3, GJA8, GUCY2D, HSF4, LEMD2, LIM2, LONP1, LSS, MAF, MIP, NHS, NR2E3, P3H2, PAX6, PITX3, PRX, PXDN, RRAGA, SIPA1L3, SIL1, SIX6, SLC16A12, TDRD7, TMEM114, TRPM3, UNC45B, VIM, VSX2, WDR36, WDR87, WFS1	NGS with CNV analysis
Choroideremia	1	CHM	Sanger Sequencing
	3	CHM, RP2, RPGR	MLPA
Cone-Rod Dystrophy	35	ABCA4, ADAM9, AIPL1, BEST1, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CNGB3, CNNM4, C8ORF37, CRX, GNAT2, GUCA1A, GUCY2D, KCNV2, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR (ORF15 excluded), RPGRIP1, SEMA4A, TTLL5, UNC119	NGS with CNV analysis
Congenital Fibrosis of Extraocular Muscles	4	KIF21A, TUBB3, TUBB2B, PHOX2A	NGS with CNV analysis
Congenital Stationary Night Blindness		CABP4, CACNA1F, CHM, GNAT1, GRK1, GRM6, NYX, PDE6B, RDH5, RHO, SAG, SLC24A1, TRPM1	NGS with CNV analysis
Corneal Dystrophy	20	CHST6, COL5A1, COL17A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, PIKFYVE, PRDM5, SLC4A11, SOD1, ZEB1, ZNF469, TACSTD2, TGFBI, UBIAD1, VSX1	NGS with CNV analysis
Ectopia Lentis	18	AASS, ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, BCOR, CBS, COL8A2, COL18A1, CYP1B1, FBN1, FOXC1, LTBP2, PAX6, P3H2, PORCN, SUOX, VSX2	NGS with CNV analysis
Glaucoma	19	ACVR1, ASB10, BEST1, CANT1, COL18A1, CYP1B1, FOXC1, LMX1B, LOXL1, LTBP2, MYOC, NTF4, OPTN, PAX6, PITX2, PITX3, SBF2, SLC4A4, WDR36	NGS with CNV analysis
Leber Congenital Amaurosis	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	NGS with CNV analysis
	9	AIPL1, CEP290, CRB1, CRX, GUCY2D, LCA5, RDH12, RPE65, RPGRIP1	MLPA

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Leber Hereditary Optic Neuropathy	3/3	MT-ND1, MT-ND4, MT-ND6	Sanger Sequencing
	37	Mitochondrial Genome	NGS
Norrie Disease	1	NDP	Sanger Sequencing
Oculocutaneous Albinism, Ocular Albinism, Hermansky-Pudlak Syndrome, Chediak-Higashi Syndrome	22	AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CACNA1F, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, MC1R, OCA2, RAB27A, SLC24A5, SLC45A2, TYR, TYRP1	NGS with CNV analysis
	7	ATR, FOXL2, FOXC1, FOXC2, GPR143, OCA2, TYR	MLPA
Optic Atrophy	3	OPA1, OPA3, TMEM126A	NGS with CNV analysis
	1	OPA1	MLPA
			Targeted regions sequencing by NGS
Papillorenal Syndrome	1	PAX2	NGS
Retinoblastoma	1	RB1	NGS MLPA
Senior-Loken Syndrome	9	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	NGS with CNV analysis
Stargardt Disease	4	ABCA4, CNGB3, ELOVL4, PROM1	NGS with CNV analysis
	1	ABCA4	NGS MLPA
Usher Syndrome	20	ABHD12, ADGRV1 (GPR98), CDH23, CIB2, CLRN1, COL4A6, DSPP (excluding exon 5), GIPC3, HARS1, KARS1, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G, WHRN (DFNB31)	NGS with CNV analysis
	1	USH2A	MLPA
	1	PCDH15	MLPA
Vitelliform Macular Dystrophy	3	BEST1, IMPG1, PRPH2	NGS with CNV analysis
	2	BEST1, PRPH2	MLPA
	1	BEST1	NGS
Vitreoretinopathy	20	ATOH7, BEST1, CAPN5, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, COL18A1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, RS1, TSPAN12, VCAN, ZNF408	NGS with CNV analysis

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Vitreoretinopathy	20	ATOH7, BEST1, CAPN5, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, COL18A1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, RS1, TSPAN12, VCAN, ZNF408	NGS with CNV analysis
X-Linked Retinitis Pigmentosa	3	OFD1, RP2, RPGR (ORF15 included)	NGS with CNV analysis
	2	RP2, RPGR	MLPA
	2	RP2, RPGR (ORF15 included)	Targeted regions sequencing by NGS, Sanger Sequencing
	1	RPGR (ORF15 region only)	Sanger Sequencing
X-Linked Retinoschisis	1	RS1	Sanger Sequencing
Eye Diseases	294	294 genes associated with different eye diseases	NGS with CNV analysis
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS with CNV analysis
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

Version: 2021-11-24

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

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ASPER
OTOGENETICS

Asper Otogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Method
Alport Syndrome	3	COL4A3, COL4A4, COL4A5	NGS with CNV analysis
	2	COL4A5, COL4A6	MLPA
Aminoglycoside-Induced Deafness	1/1	MT-RNR1	Sanger sequencing
Branchiootorenal Syndrome	3	EYA1, SIX1, SIX5	NGS with CNV analysis
	1	EYA1	MLPA
Jervell and Lange-Nielson Syndrome	2	KCNE1, KCNQ1	Sanger Sequencing
Palmoplantar Keratoderma with Deafness	1	GJB2	Sanger Sequencing
Pendred Syndrome	1	SLC26A4	NGS MLPA
Sensorineural Hearing Loss	78	ACTG1, ADGRV1, ATP2B2, ATP6V1B1, BSND, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLRN1, COCH, COL11A2, CRYM, DFNA5, DFNB59, DIABLO, DIAPH1, DIAPH3, ESPN, ESRRB, EYA4, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, HARS1, HGF, ILDR1, KCNJ10, KCNQ4, LHFPL5, LOXHD1, LRTOMT, MAN2B1, 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, WHRN, WFS1	NGS with CNV analysis
	1	GJB2	Sanger Sequencing
	11	GJB2, GJB3, GJB6, KCNQ4, MYO7A, MYO15A, MT-RNR1, MT-TS1, SLC26A4, SLC26A5, TMC1	Targeted regions sequencing by NGS
Stickler Syndrome	6	COL2A1, COL11A1, COL11A2, COL9A1, COL9A2, COL9A3	NGS with CNV analysis
Treacher-Collins Syndrome	3	POLR1C, POLR1D, TCOF1	NGS with CNV analysis
	1	TCOF1	MLPA

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Usher Syndrome	20	ABHD12, ADGRV1 (GPR98), CDH23, CIB2, CLRN1, COL4A6, DSPP (excluding exon 5), GIPC3, HARS1, KARS1, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G, WHRN (DFNB31)	NGS with CNV analysis
	1	USH2A	MLPA
	1	PCDH15	MLPA
	9	ADGRV1 (GPR98), CDH23, CLRN1, MYO7A, PCDH15, USH2A, USH1C, USH1G, WHRN (DFNB31)	Targeted regions sequencing by NGS
Waardenburg Syndrome	7	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR	NGS with CNV analysis
	3	MITF, PAX3, SOX10	MLPA
Zellweger Spectrum Disorders	14	PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH	NGS with CNV analysis
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

Version: 2021-06-03

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
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- TAT 2-9 weeks, excl. WES 10 weeks and WGS 15 weeks

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ASPER
PHARMACOGENETICS

Asper Pharmacogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Drugs	Method
Aminoglycoside-Induced Deafness	1/1	MT-RNR1	Amikacin, gentamycin, kanamycin, streptomycin, tobramycin	Sanger sequencing
Antidepressants PGx	2	CYP2C19, CYP2D6	SSRIs (citalopram, escitalopram, fluvoxamine, paroxetine, sertraline) TCAs (amitriptyline, clomipramine, desipramine, doxepin, imipramine, nortriptyline, trimipramine)	NGS, Long PCR
Statin-Induced Myopathy	1/1	SLCO1B1	Simvastatin, atorvastatin	Sanger sequencing

Version: 2021-03-23

Service includes

- DNA extraction
- Genotyping
- Bioinformatic analysis of detected variants
- Interpretation
- Results delivery by registered mail (on request)
- TAT 10-15 working days

Online ordering, specimen requirements and shipping instructions are available at www.asperbio.com.

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ASPER
REPROGENETICS

Asper Reprogenetics

Disease/Condition name	No of detectable genes/markers	Genes	Method
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 with CNV analysis
Carriership	550	550 genes associated with inherited diseases	NGS with CNV analysis
Cystic Fibrosis	1	CFTR	NGS MLPA
Female Infertility	128	ANOS1, AR, AXL, BMP15, BMP4, CASR, CCDC141, CFTR, CLPP, CPEB1, DUOX1, DUOX2, DUOXA2, DUSP6, EIF2B1, EIF2B2, EIF2B4, EIF2B5, EIF4ENIF1, ERCC6, ESR1, ESR2, F2, F5, FEZF1, FGF17, FGF8, FGFR1, FIGLA, FLRT3, FMR1 (incl CGG trinucleotide repeat expansion), FOXE1, FOXL2, FSHB, FSHR, GCM2, GDF9, GHR, GLIS3, GNAS, GNRH1, GNRHR, HARS2, HESX1, HFM1, HSD17B4, HS6ST1, IGSF1, IL17RD, INHA, IRS4, IYD, KISS1, KISS1R, LARS2, LHCGR, LHB, LHX3, LHX4, LHX8, MCM8, MCM9, MRPS22, MSH5, MTHFR, NANOS3, NKX2-1, NKX2-5, NLRP2, NLRP5, NOBOX, NR0B1, NR5A1, NSMF, NUP107, OTX2, PADI6, PATL2, PAX8, PDE3A, PLCZ1, POLR3B, POU1F1, PROC, PROK2, PROKR2, PROP1, PROS1, PSMC3IP, SECISBP2, SEMA3A, SERPINC1, SERPINE1, SLC26A4, SLC5A5, SMC1B, SOHLH1, SOX10, SOX2, SOX3, SPIDR, SPRY4, SRA1, STAG3, SYCE1, SYCE3, SYCP3, TAC3, TACR3, TBL1X, TG, THBD, THRA, THRB, TPO, TRH, TRHR, TSHB, TSHR, TTF1, TUBB8, WDR11, WEE2, WNT4, WT1, ZP1, ZP2, ZP3	NGS with CNV analysis
Folate-Dependent Neural Tube Defects	1	MTHFR	Targeted mutation analysis
Fragile X Syndrome	1	FMR1	Repeat Expansion/ Fragment Length Analysis
Male Factor Infertility	138	AK7, ADGRG2, AMH, AMHR2, ANOS1, AR, ARMC2, AURKC, AXL, BMP4, BNC2, BRDT, CASR, CATSPER1, CCDC39, CFAP43, CFAP44, CFAP69, CFTR, CYP17A1, CYP11B1, DDX25, DMC1, DMRT1, DNAH1, DNAH11, DNAH5, DNAH6, DNAI1, DPY19L2, DUOX1, DUOX2, DUOXA2, FANCM, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FOXE1, FSHB, FSHR, FSIP2, GCM2, GHR, GLIS3, GNAS, GNRH1, GNRHR, HESX1, HSD3B2, HS6ST1, IGSF1, IL17RD, INSL3, IRS4, IYD, KISS1R, HS6ST1, IGSF1, IL17RD, INSL3, IRS4, IYD, KISS1R,	NGS with CNV analysis

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Disease/Condition name	No of detectable genes/markers	Genes	Method
Male Factor Infertility	138	KLHL10, LHB, LHX3, LHX4, LRRC6, MAGEB4, MAMLD1, M1AP, MEI1, MEIOB, NANOS1, NKX2-1, NKX2-5, NLRP14, NR5A1, NR0B1, NSMF, OTX2, PANK2, PAX8, PDE3A, PLCZ1, PMFBP1, POU1F1, PPP2R3C, PROK2, PROKR2, PROP1, QRICH2, RNF212, RSPO1, SECISBP2, SEMA3A , SEPTIN12 (SEPT12), SLC26A4, SLC26A8, SLC5A5, SLC9A3, SOHLH1, SOX10, SOX2, SOX3, SOX8, SOX9, SPATA16, SPINK2, SRA1, SRD5A2, STAG3, STX2, SUN5, SYCE1, SYCP3, TAC3, TACR3, TAF4B, TBL1X, TDRD9, TEX11, TEX14, TEX15, TG, THRA, THRB, TPO, TRH, TRHR, TRIM37, TSGA10, TSHB, TSHR, TTC21A, TTF1, USP26, USP9Y, UTP14C, WDR11, WDR66, XRCC2, ZMYND15	NGS with CNV analysis
	N/A	Klinefelter Syndrome (47XXY)	PCR
	3/8 (+9 for extended analysis)	AZF deletions	PCR
Maternal Cell Contamination	NA/6		Fragment Analysis
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 with CNV analysis
Whole Exome Sequencing (WES)		Solo sample	NGS with CNV analysis
		Trio samples	NGS with CNV analysis
Whole Genome Sequencing (WGS)		Solo sample Trio samples	NGS
Familial mutation			Targeted Sequencing
Deletion/duplication analysis of selected regions			Chromosomal Microarray Analysis

Version: 2021-07-09

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