

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, CYP11B1, 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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Bardet-Biedl Syndrome, McKusick-Kaufman Syndrome, Borjeson-Forssman-Lehmann Syndrome, Alstrom Syndrome, Albright Hereditary Osteodystrophy	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
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
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, TGFB1, UBIAD1, VSX1	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	MT-ND1, MT-ND4, MT-ND6	176
Norrie Disease	Sanger Sequencing	1	NDP	262

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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	1567
			Trio samples	3100
Whole Genome Sequencing (WGS)	NGS		Solo sample	4500
			Trio samples	8800

* Clinical interpretation is not available

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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	552	552 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

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	1567
			Trio samples	3100
Whole Genome Sequencing (WGS)	NGS		Solo sample	4500
			Trio samples	8800

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Breast and Ovarian/ Endometrial Cancer	Targeted mutation analysis by APEX	5/87	BRCA1, BRCA2, CHEK2, RAD51, NBN	316
	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, BMPRIA, 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	MLH1	515
		1	MSH2	515
		1	MSH6	515
	NGS	3	MLH1, MSH2, MSH6	1030
MLPA	2/NA	MLH1, MSH2	257	
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	NBN	788
		1/1	NBN	90

Disease/Condition name	Method	No of detectable genes/markers	Genes	Price (EUR)
Polyposis Syndromes	NGS	6	APC, BMPRIA, 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	1567
			Trio samples	3100
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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, TGFB1, TGFB2, TGFB3	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, SPRED1	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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Alzheimer Disease	Sanger Sequencing	1/2	APOE	87
Charcot-Marie-Tooth Disease	NGS	56	AARS, AIFM1, ARHGEF10, BSCL2, C12orf65, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HARS, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS, KIF5A, KIF1B, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MORC2, MPZ, MTMR2, NAGLU, NDRG1, PDK3, PLEKHG5, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SETX, SH3TC2, SURF1, TRIM2, TRPV4, VCP, YARS	1314
	MLPA	1/NA	PMP22	316
Cornelia de Lange Syndrome	NGS	5	HDAC8, NIPBL, RAD21, SMC3, SMC1A	1030
Craniosynostosis	NGS, Sanger Sequencing	7	FGFR1, FGFR2, FGFR3, IL11RA, MSX2, RECQL4, TWIST1	1051
Dystonia	NGS	39	ACTB, ADCY5, ANO3, ARSA, ATM, ATP1A3, ATP7B, CACNA1B, CIZ1, COL6A3, DRD2, GCDH, GCH1, GNAL, GNAO1, HPCA, KCNMA1, KCTD17, PANK2, PLA2G6, PNKD, PRKN, PRKRA, PRRT2, RELN, SGCE, SLC2A1, SLC6A3, SLC25A1, SLC30A10, SLC39A14, SPR, TAF1, TBCE, TH, THAP1, TIMM8A, TOR1A, TUBB4A	1314
Epilepsy	NGS	126	AARS, ADAR, ADSL, ALDH7A1, ALG3, ALG13, ARHGEF9, ARX, ATP1A2, ATP1A3, ATP6AP2, ATRX, BRAT1, CACNA1A, CACNA1D, CACNA1H, CACNB4, CASK, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRN2, CLCN2, CPA6, CSTB, DEPDC5, DNM1, DOCK7, EEF1A2, EFHC1, EPM2A, FLNA, FOXG1, GABRA1, GABRB3, GABRD, GABRG2, GAMT, GATM, GNAO1, GOSR2, GPHN, GRIN1, GRIN2A, GRIN2B, HCN1, HUWE1, ITPA, IQSEC2, KCNA1, KCNA2, KCNB1, KCNC1, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KIAA2022, KIF1A, KIF5C, LGI1, MBD5, MCCC1, MECP2, MEF2C, MOCS1, MOCS2, MTOR, NECAP1, NHLRC1, NRXN1, PCDH19, PIK3R2, PIGA, PIGO, PIGT, PLCB1, PNKP, PNPO, POLG, PRICKLE1, PRRT2, PURA, RELN, ROGDI, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SERPINI1, SIK1, SLC12A5, SLC13A5, SLC25A22, SLC2A1, SLC35A2, SLC35A3, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SNIP1, SPATA5, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBPI, SYN1, SYNGAP1, SYP, SZT2, TBC1D24, TCF4, TSC1, TSC2, TUBB3, UBE3A, WDR45, WWOX, ZDHHC9	1546

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Fragile X Syndrome	Repeat Expansion/ Fragment Length Analysis	1	FMR1	262
Frontotemporal Dementia	NGS	5	CHMP2B, GRN, MAPT, TARDBP, PSEN1	1051
Hereditary Spastic Paraplegia	NGS	34	ATL1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, B4GALNT1, BSCL2, CYP7B1, CYP2U1, DDHD2, ERLIN2, FA2H, GBA2, GJC2, HSPD1, KIAA0196, KIF1A, KIF5A, LICAM, NIPA1, PLP1, PNPLA6, REEP1, RTN2, SLC16A2, SPAST, SPG7, SPG11, SPG20, SPG21, TECPR2, VPS37A, ZFYVE26	1314
	Sanger Sequencing	1/1	MT-ATP6	87
Joubert Syndrome	NGS	29	AH11, 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	1314
Menkes Disease	Sanger Sequencing	1	ATP7A	1030
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
Mitochondrial Diseases	Sanger Sequencing/ NGS	37	Mitochondrial Genome	1030
	NGS	133	AARS2, ABCB7, ACAD9, ACADL, ACADM, ACADS, ACADVL, ADCK3, AFG3L2, AIFM1, ALAS2, APTX, ATP5E, ATPAF2, AUH, BCS1L, BOLA3, C10orf2, C12orf65, CISD2, COA5, COQ2, COQ6, COQ9, COX10, COX15, COX6B1, CPT1A, CPT2, DARS2, DGUOK, DLAT, DLD, DNAJC19, DNM1L, ETFA, ETFB, ETFDH, ETHE1, FASTKD2, FBP1, FH, FOXRED1, G6PC, GAMT, GATM, GFER, GFM1, GYS2, HARS2, HLCS, HADH, HADHA, HSPD1, ISCU, LRPPRC, MFN2, MPV17, MRPS16, MRPS22, MTFMT, MTPAP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, OPA1, OPA3, PC, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, POLG, POLG2, PUS1, RARS2, REEP1, RRM2B, SARS2, SCO1, SCO2, SDHA, SDHAF1, SETX, SLC19A3, SLC25A20, SLC25A3, SLC25A4, SLC6A8	1314

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Mitochondrial Diseases	NGS	133	SLC37A4, SOD1, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TAZ, TIMM8A, TK2, TMEM126A, TMEM70, TRMU, TSFM, TTC19, TUFM, TYMP, UQCRB, UQCRQ, WFS1, YARS2	1314
Mitochondrial Encephalomyopathy Lactic Acidosis and Stroke-Like Episodes (MELAS Syndrome)	Sanger Sequencing	1/1	MT-TL1	87
Neurodegeneration with Brain Iron Accumulation	NGS	10	ATP13A2, COASY, C19orf12, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45	1051
Parkinson Disease	NGS	20	ADH1C, ATP13A2, ATXN2, DNAJC6, EIF4G1, FBXO7, GBA, GIGYF2, HTRA2, LRRK2, MAPT, PARK2, PARK7, PINK1, PLA2G6, SLC6A3, SNCA, TBP (excluding exon 3), UCHL1, VPS35	1314
Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency	Sanger Sequencing	1	ACADS	515
Smith-Lemli-Opitz Syndrome	Sanger Sequencing	1	DHCR7	525
Spinocerebellar Ataxias	NGS	64	ABCB7, ABHD12, ACO2, ADCK3, AFG3L2, ANO10, APTX, ATCAY, ATM, ATP8A2, CASK, CCDC88C, CLCN2, CLN5, C10orf2, CWF19L1, CYP27A1, DARS2, DNMT1, EEF2, ELOVL4, ELOVL5, FGF14, FLVCR1, FXN, GOSR2, GRM1, ITPR1, KCNC3, KCND3, KCNJ10, LAMA1, NOP56, OPHN1, PDYN, PHYH, PNKP, PNPLA6, POLG, PRKCG, PTF1A, RNF216, RUBCN, SACS, SETX, SIL1, SLC9A1, SLC9A6, SLC52A2, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TTBK2, TDP1, TGM6, TMEM240, TPP1, TTPA, TUBB4A, WFS1, WWOX, ZNF592	1314
	Repeat Expansion Analysis	12	ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, ATN1, BEAN1, CACNA1A, FXN, NOP56, PPP2R2B, TBP	2677
Very Long Chain Acyl-Coenzyme A Dehydrogenase (VLCAD) Deficiency	Sanger Sequencing	1	ACADVL	515
Wilson Disease	Targeted mutation analysis by APEX	1/117	ATP7B	316
	Sanger Sequencing	1	ATP7B	773

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

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

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	1567
			Trio samples	3100
Whole Genome Sequencing (WGS)	NGS		Solo sample	4500
			Trio samples	8800

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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
Whole Exome Sequencing (WES)	NGS		Solo sample	1567
			Trio samples	3100
Whole Genome Sequencing (WGS)	NGS		Solo sample	4500
			Trio samples	8800

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Brain malformations	NGS	147	ACTB, ACTG1, ADGRG1, AHI1, AKT3, AMPD2, AMT, AP4M1, ARFGEF2, ARL13B, ARX, ASPM, ATP6VOA2, 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
Jeune Syndrome	NGS	15	CEP120, CSPP1, DYNC2H1, DYNC2LI1, IFT52, IFT80, IFT140, IFT172, KIAA0586, NEK1, TTC21B, WDR19, WDR34, WDR35, WDR60	1546
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 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

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

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