NEXT GENERATION SANGER SEQUENCING PLATFORMS (NGS)

Category	Disease Category	Disease	Number of Genes	Genes	Comment	Test Specification	Price in €
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	COMPREHENSIVE HEREDITARY CANCER	116	AIP, ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CYLD, DDB2, DICER1, DIS3L2, DKC1, EGFR, ELANE, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, EXO1, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GATA2, GPC3, GREM1, HNF1A, HOXB13, HRAS, KIT, KRAS, MAX, MEN1, MET, MITF, MLH1, MLH3, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NSD1, PALB2, PHOX2B, PMS1, PMS2, POLD1, POLE, PPM1D, PRF1, PRKAR1A, PTCH1, PTEN, PTPN11, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RUNX1, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCB1, STK11, SUFU, TERC, TERT, TINF2, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XPA, XPC, XRCC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	BREAST, OVARIAN, COLORECTAL CANCER	30	BRCA1, BRCA2, APC, ATM, BAP1, BARD1, BMPR1A, BRIP1, CDH1, CDKN2A CDK4, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MITF, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53		Sequencing and Deletion-Duplication Testing of all Genes	359
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	BREAST AND OVARIAN CANCER	12	BRCA1, BRCA2, CHEK2, BRIP1, BARD1, CDH1, TP53, PTEN, RAD51C, MRE11A, NBN, ATM		Sequencing	680
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	COLON CANCER	6	APC, MUTYH, MLH1, MSH2, MSH6, PMS1, PMS2, EPCAM, BMPR1A, SMAD4, STK11, PTEN		Sequencing and Deletion-Duplication Testing of all Genes	680
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	GASTROINTESTINAL CANCER	38	APC, ATM, AXIN2, BLM, BMPR1A, BRCA1, BRCA2, BUB1B, CDH1, CDKN2A, EPCAM, FANCC, GREM1, HNF1A, KIT, MEN1, MLH1, MSH2, MSH6, MUTYH, NF1, PALB2, PMS2, POLD1, POLE, PTEN, RHBDF2, SDHB, SDHC, SDHD, SMAD4, SMARCB1, STK11, TMEM127, TP53, TSC1, TSC2, VHL		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	LEUKEMIA	25	ATM, BLM, BRCA2, CDKN2A, CEBPA, DKC1, ELANE, FANCA, GATA2, HRAS, KRAS, MLH1, MSH2, MSH6, NBN, NF1, NRAS, PMS2, PTPN11, RUNX1, SBDS, TERC, TERT, TINF2, TP53		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	LUNG CANCER	4	BRCA2, CDKN2A, EGFR, TP53		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	MELANOMA AND SKIN CANCER	19	BAP1, BRCA1, BRCA2, CDK4, CDKN2A, DDB2, ERCC2, ERCC3, ERCC4, ERCC5, MITF, PTCH1, PTEN, RB1, SUFU, TP53, WRN, XPA, XPC		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	PANCREATIC CANCER	22	APC, ATM, BMPR1A, BRCA1, BRCA2, BUB1B, CDKN2A, EPCAM, FANCC, MEN1, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, SMAD4, STK11, TP53, TSC1, TSC2, VHL		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	PARAGANGLIOMA-PHEOCHROMOCYTOMA	11	FH, MAX, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	PEDIATRIC CANCER	52	ALK, APC, AXIN2, BAP1, BLM, BMPR1A, CDC73, CDKN1C, CEBPA, DICER1, DIS3L2, EPCAM, EZH2, FH, GATA2, GPC3, HRAS, MAX, MEN1, MLH1, MSH2, MSH6, NBN, NF1, NF2, NSD1, PHOX2B, PMS2, PRF1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, RET, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCB1, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1		Sequencing and Deletion-Duplication Testing of all Genes	
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	RENAL CANCER (PANEL 1)	25	BAP1, CDC73, CDKN1C, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, HNF1A, MET, MLH1, MSH2, MSH6, PMS2, PTEN, SDHB, SDHC, SDHD, SMARCB1, TP53, TSC1, TSC2, VHL, WT1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	RENAL CANCER (PANEL 2)	19	BAP1, FH, FLCN, MET, MITF, MLH1, MSH2, MSH6, PMS2, PTEN, SDHA, SDHB, SDHC, SDHD, TP53, TSC1, TSC2, VHL, EPCAM (Deletion/Duplication Testing only)		Sequencing of 18 Genes and Deletion-Duplication Testing of 19 Genes	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	THYROID CANCER	11	APC, CDC73, DICER1, MEN1, PRKAR1A, PTEN, RET, SDHB, SDHD, TP53, WRN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	NEUROFIBROMATOSIS	7	KIT, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	TUBEROUS SCLEROSIS	2	TSC1, TSC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	XERODERMA PIGMENTOSUM (PANEL 1)	7	DDB2, ERCC2, ERCC3, ERCC4, ERCC5, XPA, XPC		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cancer	XERODERMA PIGMENTOSUM (PANEL 2)	9	DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC		Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	COMPREHENSIVE CARDIOLOGY	165	A2ML1, AARS2, ABCB4, ABCC9, ACAD9, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, AGK, AGL, AKAP9, ALPK3, ANK2, ANKRD1, ANO5, APOA1, APOB, BAG3, BRAF, CACNA1C, CACNA2D4, CACNB2, CALM1, CALM2, CALM3, CAPN3, CASQ2, CAV3, CBL, CHKB, COX15, CPT1A, CPT2, CRYAB, CSRP3, CTNNA3, DAG1, DBH, DES, DMD, DNAJC19, DSC2, DSG2, DSP, DYSF, EMD, ENPP1, ETFA, ETFB, ETFDH, EYA4, FBXO32, FHL1, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATA5, GATAD1, GBE1, GFM1, GJA5, GLA, GLB1, GMPPB, GNE, GUSB, HCN4, HFE, HRAS, ISPD, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, KRAS, LAMA2, LAMP2, LARGE, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOT, MYPN, NEXN, NF1, NKX2-5, NOS1AP, NRAS, NSUN2, PKP2, PLEC, PLEKHM2, PLN, PNPLA2, POMGNT1, POMT1, POMT2, PRDM16, PRKAG2, PTPN11, RAF1, RASA2, RBM20, RIT1, RRAS, RYR2, SCN10A, SCN1B, SCN3B, SCN5A, SCNN1B, SCNN1G, SCO2, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A4, SMCHD1, SOS1, SPRED1, TAZ, TBX5, TCAP, TGFB3, TMEM43, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRIM32, TRPM4, TSFM, TTN,		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	AORTA ANOMALIES	37	ABCC6, ACTA2, ADAMTS2, CBS, COL11A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, EFEMP2, ELN, ENPP1, FBLN5, FBN1, FBN2, FKBP14, FLNA, GATA5, MFAP5, MYH11, NOTCH1, PLOD1, SKI, SLC2A10, SLC39A13, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, TNXB, ZNF469		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	ARRHYTHMIA	50	ABCB4, AKAP9, ANK2, CACNA1C, CACNA2D4, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CTNNA3, DBH, DES, DSC2, DSG2, DSP, GATA6, GJA5, HCN4, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDB3, LMNA, MYH6, NKX2-5, NOS1AP, PKP2, PLN, RYR2, SCN10A, SCN1B, SCN3B, SCN5A, TBX5, TGFB3, TMEM43, TNNI3, TNNT2, TPM1, TRDN, TRPM4, TTN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY (ARVC)	14	CTNNA3, DES, DSC2, DSG2, DSP, JUP, LDB3, LMNA, PKP2, PLN, RYR2, TGFB3, TMEM43, TTN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	ATRIAL FIBRILLATION	22	ABCB4, CACNA2D4, CACNB2, GATA6, GJA5, HCN4, KCNA5, KCND3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDB3, LMNA, RYR2, SCN10A, SCN1B, SCN3B, SCN5A, TBX5		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	BRUGADA SYNDROME	11	CACNA1C, CACNB2, CAV3, HCN4, KCND3, KCNE3, KCNH2, SCN1B, SCN3B, SCN5A, TRPM4		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	CARDIOMYOPATHY	134	A2ML1, AARS2, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALPK3, ANKRD1, ANO5, APOA1, BAG3, BRAF, CAPN3, CASQ2, CAV3, CBL, CHKB, COX15, CPT1A, CPT2, CRYAB, CSRP3, CTNNA3, DAG1, DBH, DES, DMD, DNAJC19, DSC2, DSG2, DSP, DYSF, EMD, ETFA, ETFB, ETFDH, EYA4, FBXO32, FHL1, FKRP, FKTN, FLNC, FOXRED1, FXN, GAA, GATAD1, GBE1, GFM1, GLA, GLB1, GMPPB, GNE, GUSB, HCN4, HFE, HRAS, ISPD, JPH2, JUP, KRAS, LAMA2, LAMP2, LARGE, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MTO1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYOT, MYPN, NEXN, NF1, NRAS, NSUN2, PKP2, PLEC, PLEKHM2, PLN, PNPLA2, POMGNT1, POMT1, POMT2, PRDM16, PRKAG2, PTPN11, RAF1, RASA2, RBM20, RIT1, RRAS, RYR2, SCN5A, SCNN1B, SCNN1G, SCO2, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A4, SMCHD1, SOS1, SPRED1, TAZ, TCAP, TGFB3, TMEM43, TMEM70, TNNC1, TNNI3, TNNT2, TPM1, TRIM32, TSFM, TTN, TTR, VCL, VCP, XK		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CPVT)	7	CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TRDN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	CONGENITAL HEART MALFORMATION	28	ACTA2, ACTC1, BMPR2, CHD7, CTC1, ELN, FLNA, FOXC1, FOXH1, G6PC3, GATA4, GATA5, GATA6, GJA1, GJA5, HRAS, JAG1, NF1, NKX2-5, NODAL, NOTCH1, NOTCH2, TBX1, TBX5, TEAP2B, TLL1, ZFPM2, ZIC3		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	CONGENITAL HEART MALFORMATION (PANEL 2)	18	ACTC1, BMPR2, CFC1, ELN, G6PC3, GATA4, GATA6, GDF1, GJA1, JAG1, MYH6, NKX2-3, NKX2-5, NKX2-6, RBM10, TBX1, TBX20, TLL1		Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	CONNECTIVE TISSUE DISORDERS	22	ACTA2, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL11A1, COL11A2, FBN1, FBN2, FLNA, MYH11, MYLK, NOTCH1, NTM, PRKG1, SMAD3, TGFB2, TGFBR1, TGFBR2, TGFBR3		Sequencing	1700
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	DILATED CARDIOMYOPATHY (DCM)	27	ABCC9, BAG3, DES, DMD, DSC2, DSG2, DSP, EMD, FBXO32, HCN4, JUP, LAMP2, LMNA, MYBPC3, MYH6, MYH7, PKP2, PLEKHM2, PLN, RAF1, RBM20, SCN5A, TCAP, TNNT2, TPM1, TTN, VCL		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	EHLERS-DANLOS SYNDROME	33	ABCC6, ADAMTS2, ALDH18A1, ATP6V0A2, ATP7A, CBS, CHST14, COL11A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, PLOD1, PYCR1, SLC39A13, SMAD3, TGFB2, TGFBR1, TGFBR2, TNXB, ZNF469		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	EHLERS-DANLOS SYNDROME (PANEL 2)	11	B3GALT6, XYLT1, B3GAT3, B4GALT7, PLOD1, FKBP14, SLC39A13, CHST14, DSE, ADAMTS2, RIN2		Sequencing	1100
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	HYPERLIPIDEMIA	11	ABCA1, ABCG5, ABCG8, APOA1, APOB, APOC3, APOE, LDLR, LDLRAP1, LPL, PCSK9		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	HYPERTROPHIC CARDIOMYOPATHY (HCM)	19	ACTC1, ACTN2, ALPK3, CSRP3, GAA, GLA, JPH2, LAMP2, MYBPC3, MYH7, MYL2, MYL3, PRKAG2, RAF1, SOS1, TNNI3, TNNT2, TPM1, TTR			
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	LEFT VENTRICULAR NON-COMPACTION CARDIOMYOPATHY (LVNC)	29	ABCC9, BAG3, DES, DMD, DSC2, DSG2, DSP, EMD, FBXO32, HCN4, JPH2, JUP, LAMP2, LMNA, MYBPC3, MYH6, MYH7, PKP2, PLEKHM2, PLN, RAF1, RBM20, RYR2, SCN5A, TCAP, TNNT2, TPM1, TTN, VCL		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	LIDDLE SYNDROME	2	SCNN1B, SCNN1G		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	LONG QT SYNDROME (LQTS)	15	AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NOS1AP, SCN5A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	MARFAN SYNDROME	25	ADAMTSL4, CBS, COL11A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, FBN1, FBN2, MED12, PLOD1, SKI, SLC2A10, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, UBE3B, VCAN			
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	NOONAN SYNDROME	19	A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, NSUN2, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SPRED1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	PULMONARY ARTERY HYPERTENSION (PAH)	11	ACVRL1, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, KCNA5, KCNK3, RASA1, SMAD4, TBX4		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	SHORT QT SYNDROME (SQTS)	5	CACNA1C, CACNB2, KCNH2, KCNJ2, KCNQ1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Cardiology	SUDDEN DEATH	154	ABCA1, ABCC6, ABCC9, ACTA2, ACTC1, ACTN2, ADRB1, ADRB2, ADRB3, AGL, AKAP9, ANK2, ANKRD1, APOA1, APOB, APOE, BAG3, BRAF, CACNA1B, CACNA1C, CACNA1D, CACNA2D1, CACNB2, CALR3, CASQ2, CAV3, CBL, COL3A1, CRYAB, CSRP3, CTF1, DES, DMD DPP6, DSC2, DSG2, DSP, DTNA, EFEMP2, EMD, ENPP1, EYA4, FBLN5, FBN1, FBN2, FHL1, FHL2, FKTN, FLNC, FXN, GAA, GJA1, GJA5, GLA, GPD1L, HCN1, HCN4, HRAS, ILK, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE1L, KCNE2, KCNE3, KCNE4, KCNH2, KCNJ11, KCNJ12, KCNJ2, KCNJ3, KCNJ5, KCNJ8, KCNQ1, KCNQ2, KRAS, LAMA4, LAMP2,LDB3, LDLR, LMNA, LPL, LRP6, MAP2K1, MAP2K2, MEF2A, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOT, MYOZ2, MYPN, NEBL, NEXN, NOTCH1, NPPA, NRAS, PCSK9, PDLIM3, PKP2, PKP4, PLEC, PLN, PNN,PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RANGRF, RBM20, RPSA, RYR2, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN10A, SCNN1B, SCNN1G, SDHA, SGCD, SHOC2, SLC25A4,SLC2A10, SMAD3, SNTA1, SOS1, SPRED1, SYNE1,SYNE2, TAZ, TCAP, TGFB3, TGFBR1, TGFBR2, TGFBR3, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TTN, VCL		Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	ADAMS-OLIVER SYNDROME	6	ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	ALBINISM	19	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITF, MYO5A, OCA2, RAB27A, SLC45A2, TYR, TYRP1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	CUTIS LAXA	8	ALDH18A1, ATP4A, ATP6V0A2, EFEMP2, ELN, FBLN5, PYCR1, SLC2A10		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	CUTIS LAXA (PANEL 2)	13	ELN, FBLN4 (EFEMP2), FBLN5, LTBP4, ATP6V0A2, PYCR1, ALDH18A1, ATP7A, COG7, TALDO1, SCYL1BP1 (GORAB), NAA10, RIN2		Sequencing	1600
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	DYSKERATOSIS CONGENITA	11	AK2, CTC1, DKC1, NHP2, NOP10, RTEL1, TERC, TERT, TINF2, USB1, WRAP53		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	ECTODERMAL DYSPLASIA	19	ABCC9, BCS1L, DSP, EDA, EDAR, ERCC2, EVC, EVC2, GJB2, GJB6, IFT122, IKBKG, JUP, PORCN, RMRP, SHOC2, TP63, WDR35, WNT10A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	EHLERS-DANLOS SYNDROME	33	ABCC6, ADAMTS2, ALDH18A1, ATP6V0A2, ATP7A, CBS, CHST14, COL11A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, PLOD1, PYCR1, SLC39A13, SMAD3, TGFB2, TGFBR1, TGFBR2, TNXB, ZNF469		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	EPIDERMOLYSIS BULLOSA	25	ATP2C1, CDSN, COL17A1, COL7A1, DSG1, DSG2, DSG4, DSP, DST, EXPH5, FERMT1, GRIP1, ITGA3, ITGA6, ITGB4, KRT1, KRT14, KRT5, LAMA3, LAMB3, LAMC2, MMP1, PKP1, PLEC, TGM5		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	EPIDERMOLYSIS BULLOSA (PANEL 2)	21	CD151, CDSN, CHST8, COL7A1, COL17A1, DSP, DST, EXPH5 , FERMT1, ITGA3, ITGA6, ITGB4, JUP, KRT5, KRT14, LAMA3, LAMB3, LAMC2, PKP1, PLEC1, TGM5		Sequencing	1300
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	HERMANSKY-PUDLAK SYNDROME	22	ABCA3, AP3B1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	ICHTHYOSIS	19	ABCA12, ALG8, ALOX12B, ALOXE3, CYP4F22, EBP, ERCC2, FLG, GJA1, GJB2, GJB3, KRT1, KRT2, KRT9, PEX7, PHYH, STS, SUMF1, TGM1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	NEUROFIBROMATOSIS	7	KIT, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	PACHYONYCHIA CONGENITA	8	AAGAB, GJB6, KRT16, KRT17, KRT6A, KRT6B, KRT6C, TRPV3		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	PROGERIA AND PROGEROID SYNDROMES	16	AGPAT2, ALDH18A1, B4GALT7, BLM, BSCL2, COL3A1, ERCC2, ERCC4, ERCC5, ERCC6, ERCC8, LMNA, PYCR1, RECQL4, WRN, ZMPSTE24		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Dermatology	WAARDENBURG SYNDROME	6	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	COMPREHENSIVE HEARING LOSS	158	ABHD12, ACTG1, ADCY1, ADGRV1, ALMS1, ANKH, ATP6V1B1, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CDH23, CDKN1C, CEACAM16, CHD7, CHSY1, CIB2, CLDN14, CLIC5, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCDC2, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DLX5, DSPP, EDN3, EDNRB, ELMOD3, EPS8, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FGFR3, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, HARS, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, LHFPL5, LOXHD1, LRP2, LRTOMT, MANBA, MARVELD2, MET, MIR96, MITF, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RMND1, RPS6KA3, SALL4, SEMA3E, SERPINB6, SIX1, SIX5, SLC17A8, SLC19A2, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, STRC, SUCLA2, SUCLG1, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMC2, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TSPEAR, TYR, USH1C, USH1G, USH2A, VCAN, WFS1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	ALPORT SYNDROME	6	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	BRANCHIO-OTO-RENAL (BOR) SYNDROME	4	EYA1, SIX1, SIX5, TFAP2A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT)	4	ACVRL1, ENG, RASA1, SMAD4		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	NON-SYNDROMIC HEARING LOSS	93	ACTG1, ADCY1, BDP1, BSND, CABP2, CCDC50, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, COCH, COL11A2, COL4A6, CRYM, DCDC2, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DSPP, ELMOD3, EPS8, ESPN, ESRRB, EYA4, FAM65B, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, HGF, HOMER2, ILDR1, KARS, KCNQ4, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MIR96, MSRB3, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, NARS2, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PNPT1, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SERPINB6, SIX1, SLC17A8, SLC26A4, SLC26A5, SLITRK6, SMPX, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMC2, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, WFS1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	PENDRED SYNDROME	3	FOXI1, KCNJ10, SLC26A4		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	SYNDROMIC HEARING LOSS	70	ABHD12, ACTG1, ADGRV1, ALMS1, ANKH, ATP6V1B1, BCS1L, BSND, BTD, CACNA1D, CD151, CDH23, CDKN1C, CHD7, CHSY1, CIB2, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, DFNB31, DLX5, EDN3, EDNRB, EYA1, FGF3, FOXI1, GATA3, HARS, HOXB1, KCNE1, KCNJ10, KCNQ1, LRP2, MANBA, MITF, MYH9, MYO7A, NDP, NLRP3, PAX3, PCDH15, PDZD7, POLR1C, POLR1D, SEMA3E, SIX1, SIX5, SLC19A2, SLC26A4, SLITRK6, SMAD4, SNAI2, SOX10, TCOF1, TFAP2A, TIMM8A, TYR, USH1C, USH1G, USH2A, VCAN, WFS1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	USHER SYNDROME	13	ABHD12, ADGRV1, CDH23, CIB2, CLRN1, DFNB31, HARS, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ear-Nose-Throat	WAARDENBURG SYNDROME	6	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	ABNORMAL GENITALIA/ DISORDERS OF SEX DEVELOPMENT	39	AMH, AMHR2, ANOS1, AR, ARX, ATRX, BCOR, CDKN1C, CEP41, CHD7, CREBBP, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DYNC2H1, FIG4, FRAS1, GATA4, GNRHR, HSD17B3, HSD3B2, IL17RD, IRF6, LHCGR, MKS1, NR0B1, NR5A1, POR, PROKR2, RSPO1, SOX9, SRD5A3, SRY, STAR, TACR3, WT1, ZFPM2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	CONGENITAL ADRENAL HYPERPLASIA	7	CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, PRKAR1A, STAR		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	GLUCOCORTICOID DEFICIENCY	6	MC2R, MRAP, NNT, NR3C1, POMC, STAR		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	HYPERPARATHYROIDISM	6	AIRE, CASR, CDC73, CDKN1B, MEN1, RET		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	HYPOGLYCEMIA, HYPERINSULINISM AND KETONE METABOLISM	18	ABCC8, ACAT1, FBP1, GCK, GLUD1, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, OXCT1, PCK1, PCK2, PDX1, SLC16A1, UCP2		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	HYPOMAGNESEMIA	17	CLDN16, CLDN19, CNNM1, CNNM2, CNNM4, EGF, FXYD2, HNF1B, KCNA1, MAGT1, MMGT1, NIPA2, SLC12A3, SLC41A2, SLC41A3, TRPM6, TRPM7		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	HYPOTHYROIDISM AND RESISTANCE TO THYROID HORMONE	17	DUOX2, GNAS, HESX1, NKX2-1, NKX2-5, PAX8, POU1F1, PROP1, SLC16A2, SLC26A4, SLC5A5, TG, THRA, THRB, TPO, TSHB, TSHR		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	KALLMANN SYNDROME	8	ANOS1, CHD7, FGF8, FGFR1, GNRHR, PROK2, PROKR2, TACR3		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	MODY	12	ABCC8, BLK, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	MONOGENIC DIABETES MELLITUS	25	ABCC8, BLK, EIF2AK3, FOXP3, G6PC2, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, INS, INSR, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PPARG, SLC16A1, SLC2A2, UCP2, WFS1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	MONOGENIC OBESITY	36	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CRTC1, CUL4B, DYRK1B, GNAS, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, PYY, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDPCP		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	PREMATURE OVARIAN FAILURE	13	CYP17A1, CYP19A1, FOXL2, FSHR, GALT, GNAS, LHCGR, LMNA, NR5A1, POLG, POR, STAR, WT1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Endocrinology	PREMATURE OVARIAN FAILURE (PANEL 2)	17	NOBOX, LHR, STAR, BMP15, PSMC3IP, WT1, DIAPH2, FSHB, GDF9, FOXL2, WNT4, FSHR, FIGLA, CYP17A1, CYP19A1, POF1B		Sequencing of 16 genes and repeat FMR1	2800
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	CHOLESTASIS	31	ABCB11, ABCB4, ABCC2, ATP8B1, CFTR, CYP7B1, DGUOK, EPCAM, FAH, JAG1, LCT, MKS1, MYO5B, NEUROG3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, SERPINA1, SLC25A13, SLC26A3, SMPD1, SPINT2, TJP2, TMEM216, TRMU, TTC37, UGT1A1, VPS33B		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	CHOLESTASIS (PANEL 2)	15	ABCB11, ABCB4, AKR1D1, ATP8B1, BAAT, CLDN1, HSD3B7, JAG1, NOTCH2, NR1H4, SERPINA1, SLC25A13, TJP2, VIPAS39, VPS33B		Sequencing	1700
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	CONGENITAL DIARRHEA	9	DGAT1, EPCAM, GUCY2C, LCT, MYO5B, NEUROG3, SLC26A3, SPINT2, TTC37		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	CONGENITAL HEPATIC FIBROSIS	29	AHI1, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP290, INVS, IQCB1, MKKS, MKS1, NPHP1, NPHP3, NPHP4, OFD1, PKD2, PKHD1, RPGRIP1L, TMEM67, TRIM32, TTC21B, TTC8, WDR35		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	GASTROINTESTINAL ATRESIA	7	CHD7, FANCB, FANCC, GLI3, MID1, SOX2, TTC7A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	HIRSCHSPRUNG DISEASE	9	EDN3, EDNRB, KIF1BP, MITF, NRG1, PAX3, RET, SOX10, ZEB2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	PANCREATITIS	5	CFTR, CPA1, CTRC, PRSS1, SPINK1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Gastroenterology	POLYCYSTIC LIVER DISEASE	5	LRP5, PKD1, PKD2, PRKCSH, SEC63		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	COMPREHENSIVE HEMATOLOGY	175	ABCA3, ABCB7, ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, AK2, ALAS2, AMN, ANK1, ANKRD26, AP3B1, ATM, ATR, ATRX, BLM, BLOC1S3, BLOC1S6, BRCA2, BRIP1, C15ORF41, CDAN1, CDKN2A, CEBPA, CSF2RA, CTC1, CTSC, CUBN, CXCR4, CYCS, DKC1, DTNBP1, ELANE, EPB42, ERCC4, F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FGA, FGB, FGG, FLNA, G6PC3, G6PD, GATA1, GATA2, GGCX, GP1BA, GP1BB, GP9, GPI, GPR143, GSS, HAX1, HBA1, HBA2, HBB, HFE, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, KLF1, KRAS, LMAN1, LPIN2, LYST, MAGT1, MASTL, MLH1, MPL, MSH2, MSH6, MTR, MYH9, MYO5A, NBEAL2, NBN, NF1, NHP2, NOP10, NRAS, OCA2, P2RY12, PALB2, PC, PDHA1, PDHX, PKLR, PMS2, PRF1, PROC, PROS1, PTPN11, PUS1, RAB27A, RAD51C, RBM8A, RECQL4, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SBDS, SEC23B, SERPINC1, SFTPB, SFTPC, SH2D1A, SLC19A2, SLC45A2, SLC4A1, SLFN14, SLX4, SPTA1, SPTB, STX11, STXBP2, TBXA2R, TCIRG1, TERC, TERT, THBD, TINF2, TMPRSS6, TP53, TPI1, TUBB1, TYR, TYRP1, UNC13D, USB1, VKORC1, VWF, WAS, WRAP53, XIAP, XRCC2, YARS2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	ANEMIA	68	ABCB7, ADAMTS13, ALAS2, AMN, ANK1, ATM, ATR, ATRX, BLM, BRCA2, BRIP1, C15ORF41, CDAN1, CUBN, EPB42, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PD, GATA1, GPI, GSS, HBA1, HBA2, HBB, HFE, KLF1, LPIN2, MTR, NBN, PALB2, PC, PDHA1, PDHX, PKLR, PUS1, RAD51C, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, SBDS, SEC23B, SLC19A2, SLC4A1, SLX4, SPTA1, SPTB, THBD, TMPRSS6, TPI1, XRCC2, YARS2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	BLEEDING DISORDER/COAGULOPATHY	54	ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, AP3B1, BLOC1S3, BLOC1S6, CYCS, DTNBP1, F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FGA, FGB, FGG, FLNA, GATA1, GGCX, GP1BA, GP1BB, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, LMAN1, MASTL, MPL, MYH9, NBEAL2, P2RY12, PROC, PROS1, RBM8A, RUNX1, SERPINC1, SLFN14, TBXA2R, THBD, TUBB1, VKORC1, VWF, WAS		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	BLOOM SYNDROME	1	BLM		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	BONE MARROW FAILURE SYNDROME	93	ACTB, AK2, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRCA2, BRIP1, CDKN2A, CEBPA, CSF2RA, CTC1, CTSC, CXCR4, DKC1, DTNBP1, ELANE, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, G6PC3, GATA1, GATA2, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, ITK, JAGN1, KRAS, LYST, MAGT1, MLH1, MPL, MSH2, MSH6, MYO5A, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PMS2, PRF1, PTPN11, RAB27A, RAD51C, RECQL4, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SBDS, SH2D1A, SLX4, STX11, STXBP2, TCIRG1, TERC, TERT, TINF2, TP53, UNC13D, USB1, WAS, WRAP53, XIAP, XRCC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	COAGULATION FACTOR DEFICIENCY	16	F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FGA, FGB, FGG, GGCX, LMAN1, VKORC1, VWF		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	CONGENITAL NEUTROPENIA	13	ACTB, CSF2RA, CTSC, ELANE, G6PC3, GATA2, HAX1, IFNGR2, JAGN1, LYST, SBDS, TCIRG1, WAS		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	DIAMOND-BLACKFAN ANEMIA	12	GATA1, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	DYSKERATOSIS CONGENITA	11	AK2, CTC1, DKC1, NHP2, NOP10, RTEL1, TERC, TERT, TINF2, USB1, WRAP53		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	FANCONI ANEMIA	22	ATM, ATR, BLM, BRCA2, BRIP1, CXCR4, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, NBN, PALB2, RAD51C, SLX4, XRCC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS	13	FAS, ITK, LYST, MAGT1, MYO5A, PRF1, RAB27A, RECQL4, SH2D1A, STX11, STXBP2, UNC13D, XIAP		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	HEREDITARY LEUKEMIA	25	ATM, BLM, BRCA2, CDKN2A, CEBPA, DKC1, ELANE, FANCA, GATA2, HRAS, KRAS, MLH1, MSH2, MSH6, NBN, NF1, NRAS, PMS2, PTPN11, RUNX1, SBDS, TERC, TERT, TINF2, TP53		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	HERMANSKY-PUDLAK SYNDROME	22	ABCA3, AP3B1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	NEUTROPENIA	8	CSF3R, ELANE, G6PC3, GATA2, GFI1, HAX1, VPS45, WAS		Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	PLATELET FUNCTION DISORDER	18	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GP1BA, GP1BB, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, NBEAL2, P2RY12, RUNX1, TBXA2R		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	RED BLOOD CELL MEMBRANE DISORDER	5	ANK1, EPB42, SLC4A1, SPTA1, SPTB		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Hematology	THROMBOCYTOPENIA	25	ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, CYCS, FLNA, GATA1, GP1BA, GP1BB, GP9, HOXA11, ITGA2, ITGA2B, ITGB3, MASTL, MPL, MYH9, NBEAL2, RBM8A, RUNX1, SLFN14, THBD, TUBB1, WAS		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	COMPREHENSIVE PRIMARY IMMUNODEFICIENCY	232	ACP5, ACTB, ADA, ADAR, ADIPOQ, ADIPOR1, ADIPOR2, AICDA, AIRE, AK2, AP3B1, ARMC4, ATM, BLM, BTK, C1QA, C1QB, C1QBP, C1QC, C1R, C1S, C2, C3, C3AR1, C4A, C4B, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CASP10, CASP8, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD8A, CD93, CECR1, CFB, CFD, CFH, CFHR1, CFHR3, CFI, CFP, CIITA, CLU, COLEC11, CORO1A, CR1, CR2, CRP, CSF2RA, CTC1, CTLA4, CTSC, CYBA, CYBB, DCLRE1C, DDX58, DGKE, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DNMT3B, DOCK2, DOCK8, DRC1, DYX1C1, ELANE, FAS, FCN1, FCN2, FCN3, FERMT3, FOXP3, G6PC3, G6PD, GATA2, HAX1, HYDIN, IFIH1, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKG, IL10RA, IL10RB, IL12RB1, IL1RN, IL2RA, IL2RG, IL36RN, IL7, IL7R, ISG15, ITGB2, ITK, JAGN1, JAK3, LCK, LIG4, LPIN2, LRBA, LRRC6, LYST, MAGT1, MALT1, MASP1, MASP2, MAT2A, MEFV, MRE11A, MVK, NBN, NCF1, NCF2, NCF4, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, OFD1, ORAI1, PIGA, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, PRF1, PRKDC, PSMB8, PSTPIP1, PTPRC, PTX3, RAB27A, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RPGR, RSPH1, RSPH4A, RSPH9, RTEL1, SAMHD1, SBDS, SERPING1, SH2D1A, SLC37A4, SMARCAL1, SP110, SPAG1, SPINK5, STAT1, STAT2, STAT3, STAT4, STAT5B, STIM1, STK4, STXBP2, TAP1, TAP2, TAPBP, TBX1, TCIRG1, TERC, TERT, THBD, TINF2, TMEM173, TNFRSF13B, TNFRSF1A, TNFRSF4, TRAC, TREX1, TYK2, UNC119, USB1, VSIG4, VTN, WAS, WRAP53, XIAP, ZAP70, ZMYND10		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	AUTOINFLAMMATORY SYNDROME	25	ACP5, ADAR, CARD14, DDX58, ELANE, IFIH1, IL1RN, IL36RN, ISG15, LPIN2, MEFV, MVK, NLRP12, NLRP3, NOD2, PLCG2, PSMB8, PSTPIP1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TMEM173, TNFRSF1A, TREX1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	BONE MARROW FAILURE SYNDROME	93	ACTB, AK2, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRCA2, BRIP1, CDKN2A, CEBPA, CSF2RA, CTC1, CTSC, CXCR4, DKC1, DTNBP1, ELANE, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, G6PC3, GATA1, GATA2, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, ITK, JAGN1, KRAS, LYST, MAGT1, MLH1, MPL, MSH2, MSH6, MYO5A, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PMS2, PRF1, PTPN11, RAB27A, RAD51C, RECQL4, RPL11, RPL15, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS29, RPS7, RTEL1, RUNX1, SBDS, SH2D1A, SLX4, STX11, STXBP2, TCIRG1, TERC, TERT, TINF2, TP53, UNC13D, USB1, WAS, WRAP53, XIAP, XRCC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	CHRONIC GRANULOMATOUS DISEASE	7	CYBA, CYBB, G6PD, NCF1, NCF2, NCF4, NOD2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	COMPLEMENT SYSTEM DISORDER	82	ADIPOQ, ADIPOR1, ADIPOR2, ARMC4, C1QA, C1QB, C1QBP, C1QC, C1R, C1S, C2, C3, C3AR1, C4A, C4B, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD46, CD55, CD59, CD93, CFB, CFD, CFH, CFHR1, CFHR3, CFI, CFP, CLU, COLEC11, CR1, CR2, CRP, DGKE, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, FCN1, FCN2, FCN3, HYDIN, LRRC6, MASP1, MASP2, MAT2A, NME8, OFD1, PIGA, PTX3, RPGR, RSPH1, RSPH4A, RSPH9, SERPING1, SPAG1, THBD, VSIG4, VTN, ZMYND10		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	CONGENITAL NEUTROPENIA	13	ACTB, CSF2RA, CTSC, ELANE, G6PC3, GATA2, HAX1, IFNGR2, JAGN1, LYST, SBDS, TCIRG1, WAS		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	DYSKERATOSIS CONGENITA	11	AK2, CTC1, DKC1, NHP2, NOP10, RTEL1, TERC, TERT, TINF2, USB1, WRAP53		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Immunology	SEVERE COMBINED IMMUNODEFICIENCY	68	ADA, AK2, ATM, BLM, CARD11, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CIITA, CORO1A, DCLRE1C, DNMT3B, DOCK8, IFNGR1, IKBKG, IL12RB1, IL2RA, IL2RG, IL7R, ITGB2, ITK, JAK3, LCK, LIG4, LRBA, MAGT1, MALT1, NHEJ1, ORAI1, PIK3CD, PMS2, PNP, PRKDC, PTPRC, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, RTEL1, SH2D1A, SMARCAL1, SP110, SPINK5, STAT1, STAT2, STAT3, STAT4, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TBX1, TNFRSF4, TRAC, TYK2, UNC119, WAS, ZAP70		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	COMPREHENSIVE CONGENITAL MALFORMATION AND SKELETAL DYSPLASIA	429	ABCC6, ACP5, ACTA1, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS2, ADAMTSL2, ADGRG1, AGPS, AGRN, AKT1, AKT3, ALPL, ALX3, ALX4, AMER1, ANKH, ANO5, ARFGEF2, ARHGAP31, ARID1A, ARID1B, ARSE, ARX, ASPA, ASPM, ASXL1, ATP6V0A2, ATR, B3GALNT2, B3GALT6, B4GALT7, BHLHA9, BIN1, BMP1, BMP2, BMP4, BMPR1A, BMPR1B, BRWD3, CA2, CANT1, CAPN3, CASK, CASR, CBS, CCM2, CCND2, CDC6, CDK5RAP2, CDKN1C, CDON, CDT1, CENPF, CENPJ, CEP152, CEP164, CEP63, CFL2, CHAT, CHD7, CHD8, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CLCN5, CLCN7, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL4A4, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COL9A3, COLQ, COMP, CREBBP, CRTAP, CSPP1, CTSK, CUL4B, CUL7, CYP27B1, DCX, DHCR24, DHCR7, DIS3L2, DLL3, DLL4, DLX3, DNM2, DNMT3A, DOCK6, DOK7, DPAGT1, DVL1, DYM, DYNC1H1, DYNC2H1, DYRK1A, EBP, ECEL1, EDN3, EDNRB, EFEMP2, EFNB1, EFTUD2, EGR2, EIF2AK3, EIF2B5, ELN, EMD, EMX2, ENAM, ENPP1, EOGT, EP300, ERCC5, ERCC6, ERCC8, ESCO2, EVC, EVC2, EXOSC3, EXT1, EXT2, EYA1, EZH2, FAM20A, FAM20C, FAM83H, FANCB, FANCC, FBLN5, FBN1, FBN2, FGD1, FGF23, FGF3, FGF8, FGFR1, FGFR2, FGFR3, FH, FHL1, FKBP10, FKBP14 FKRP, FKTN, FLNA, FLNB, FOXH1, FOXL2, FREM1, GAA, GBA, GBE1, GDF5, GFAP, GFPT1, GH1, GHR, GHRHR, GJA1, GLE1, GLI2, GLI3, GMPPB, GNAS, GNPAT, GPC3, GPSM2, GRIA3, HDAC8, HEPACAM, HESX1, HOXA13, HOXD13, HSPG2, HUWE1, IFT122, IFT140, IFT172, IFT80, IGF1, IGF1R, IGFALS, IHH, IKBKG, INSR, IRF6, IRS1, ISPD, KAT6B, KBTBD13, KDM6A, KIAA0196, KIF11, KIF1BP, KIF7, KIT, KLHL40, KMT2D, KRAS, KRIT1, L1CAM, LAMA2, LAMP2, LARGE, LBR, LEMD3, LHX3, LHX4, LIFR, LIG4, LMNA, LMX1B, LRP4, LRP5, LTBP2, MASP1, MATN3, MBD5, MCPH1, MED12, MEF2C, MID1, MITF, MLC1, MMP9, MPZ, MRE11A, MSX2, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH7, NALCN, NDE1, NEB, NEK1, NF1, NF2, NFIX, NHEJ1, NIPBL, NODAL, NOG, NOTCH1, NOTCH2, NPR2, NR5A1, NRG1, NSD1, NSDHL, OBSL1, OCRL, OFD1, OPHN1, ORC1, ORC4, ORC6, OTX2, P3H1, PAFAH1B1, PAPSS2, PAX3, PAX6, PCNT, PDCD10, PEX7, PHEX, PHF6, PIEZO2, PIGA, PIK3CA, PITX2, PLOD1, PLOD2, PMM2, PNKP, POLR1C, POMGNT1, POMT1, POMT2, POR, POU1F1, PPIB, PQBP1, PRKAR1A, PROP1, PTCH1, PTEN, PTH1R, PTPN11, PYCR1, RAB39B, RAB3GAP1, RAB3GAP2, RAF1, RAPSN, RARS2, RASA1, RBBP8, RBPJ, RECQL4, RELN, RET, RMRP, RNU4ATAC, ROR2, RUNX2, RYR1, SBDS, SCO2, SELENON, SERPINF1, SERPINH1, SF3B4,		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	ACHONDROGENESIS, HYPOCHONDROGENESIS, FIBROCHONDROGENESIS	5	COL11A1, COL11A2, COL2A1, SLC26A2, TRIP11		Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	ADAMS-OLIVER SYNDROME	6	ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	ARTHROGRYPOSES	58	ACTA1, AGRN, BIN1, CASK, CFL2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, COL6A2, COLQ, DHCR24, DOK7, DPAGT1, ECEL1, EGR2, ERCC5, ERCC6, EXOSC3, FBN2, FHL1, FKTN, GBA, GBE1, GFPT1, GLE1, KAT6B, KLHL40, MPZ, MTM1, MUSK, MYBPC1, MYH2, MYH3, NALCN, NEB, PIEZO2, PLOD2, PMM2, RAPSN, RARS2, SCO2, SELENON, TGFB3, TK2, TNNI2, TNNT1, TPM2, TPM3, TRPV4, TSEN2, TSEN54, VPS33B,		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	BRACHYDACTYLY / SYNDACTYLY	12	BMPR1B, ESCO2, GDF5, GNAS, HOXA13, HOXD13, IHH, NOG, RECQL4, ROR2, SOX9, TP63		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	BRUCK SYNDROME	2	PLOD2, FKBP10		Sequencing	1000
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	CEREBRAL CAVERNOUS MALFORMATION	4	CCM2, KRIT1, PDCD10, RASA1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	CHONDRODYSPLASIA PUNCTATA	7	AGPS, ARSE, EBP, GNPAT, LBR, NSDHL, PEX7		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	COCKAYNE SYNDROME	2	ERCC6, ERCC8		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	CRANIOSYNOSTOSIS	34	ALPL, ALX3, ALX4, BMP4, EDN3, EDNRB, EFNB1, ESCO2, FGFR1, FGFR2, FGFR3, FLNB, FREM1, GDF5, GLI3, IFT122, IFT140, MASP1, MITF, MSX2, NOG, PAX3, POR, RECQL4, RET, SKI, SOX10, TCF12, TGFBR1, TGFBR2, TTR, TWIST1, WDR19, WDR35		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	DENTINOGENESIS IMPERFECTA, DENTIN DYSPLASIA, TOOTH AGENESIS	4	DSPP, MSX1, PTH1R, WNT10A		Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	DWARFISM	15	ATR, CDC6, CDT1, CENPJ, CEP152, CEP63, CUL7, NOTCH2, OBSL1, ORC1, ORC4, ORC6, PCNT, RBBP8, RNU4ATAC		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	ECTRODACTYLY	4	LMBR1, TP63, WNT3, WNT10B		Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	FACIAL DYSOSTOSIS	29	ALPL, ALX3, ALX4, CREBBP, DLL3, EFNB1, EFTUD2, EP300, EVC, EVC2, FGFR1, FGFR2, FGFR3, GHR, HDAC8, HSPG2, LIFR, NIPBL, POLR1C, RMRP, SF3B4, SMC1A, SMC3, SOX9, SRCAP, TCF12, TCOF1, TWIST1, UBE2A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	FETAL AKINESIA	10	CHRNA1, CHRND, CHRNE, CHRNG, COLQ, DOK7, GLE1, KLHL40, MUSK, RAPSN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	GASTROINTESTINAL ATRESIA	7	CHD7, FANCB, FANCC, GLI3, MID1, SOX2, TTC7A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	HETEROTAXY / SITUS INVERSUS	25	ACVR2B, ANKS6, CCDC103, CCDC11, CCDC114, CCDC39, CCDC40, DNAAF1, DNAAF2, DNAAF3, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, FOXH1, GDF1, HEATR2, INVS, LEFTY2, LRRC6, NKX2		Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	HIRSCHSPRUNG DISEASE	9	EDN3, EDNRB, KIF1BP, MITF, NRG1, PAX3, RET, SOX10, ZEB2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	HOLOPROSENCEPHALY	12	CDON, FGF8, FGFR1, FOXH1, GLI2, GLI3, NODAL, PTCH1, SHH, SIX3, TGIF1, ZIC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	KABUKI SYNDROME	7	CHD7, EYA1, FLNB, IRF6, KDM6A, KMT2D, SIX5		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	LISSENCEPHALY	15	ACTB, ACTG1, ARX, DCX, FKTN, LARGE, PAFAH1B1, POMGNT1, POMT1, POMT2, RELN, TUBA1A, TUBB2B, VLDLR, YWHAE		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MACROCEPHALY / OVERGROWTH SYNDROME	38	ABCC6, AKT1, AKT3, ASPA, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EIF2B5, EZH2, GFAP, GLI3, GPC3, GPSM2, GRIA3, HEPACAM, HUWE1, KIAA0196, KIF7, L1CAM, MED12, MLC1, NFIX, NSD1, OFD1, PIGA, PIK3CA, PTCH1, PTEN, RAB39B, SYNJ, TSC1, TSC2, UBE3B		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MEIER-GORLIN SYNDROME	5	CDC6, CDT1, ORC1, ORC4, ORC6		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	METAPHYSEAL DYSPLASIA	10	ANKH, CDKN1C, COL10A1, FGFR3, FLNA, MMP9, PTH1R, RMRP, RUNX2, SBDS		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MICROCEPHALY AND PONTOCEREBELLAR HYPOPLASIA	34	AKT3, ASPM, ATR, CASK, CDK5RAP2, CENPF, CENPJ, CEP152, CEP164, CEP63, DYNC1H1, DYRK1A, EFTUD2, EXOSC3, KIF11, LIG4, MBD5, MCPH1, MRE11A, NDE1, NHEJ1, OPHN1, PAFAH1B1, PCNT, PNKP, POMT1, PQBP1, RARS2, STIL, TSEN2, TSEN54, TUBB2B, VRK1, WDR62		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MICROMELIC DYSPLASIA	24	ADAMTS10, ADAMTSL2, BMPR1B, DVL1, EXT1, FBN1, FGFR3, GDF5, GNAS, IFT122, IFT140, IHH, LIFR, LTBP2, NPR2, PRKAR1A, ROR2, SHOX, SMAD4, SOX9, TRPS1, WDR19, WDR35, WNT5A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	MULTIPLE SYNOSTOSES AND SYMPHALANGISM	6	FGF9, FLNB, GDF5, HOXA11, NOG, TTR		Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	NEUROFIBROMATOSIS	7	KIT, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	NEURONAL MIGRATION DISORDER	47	ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARX, B3GALNT2, CHD7, COL4A1, COL4A2, COL4A4, DCX, DYNC1H1, EMX2, FH, FKTN, FLNA, GMPPB, GPSM2, ISPD, KIF1BP, KIF7, L1CAM, LAMA2, LARGE, MED12, MEF2C, NDE1, NSDHL, PAFAH1B1, PIK3CA, POMGNT1, POMT1, POMT2, RAB3GAP1, RAB3GAP2, RELN, SLC12A6, SRPX2, TMEM5, TUBA1A, TUBA8, TUBB2B, TUBB3, VLDLR, WDR62, WWOX		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOGENESIS IMPERFECTA	61	ACTA1, ALPL, ANO5, ATP6V0A2, B3GALNT2, B4GALT7, BMP1, CAPN3, CFL2, CHKB, CLCN5, COL1A1, COL1A2, COL6A1, COL6A2, COL6A3, CRTAP, DNM2, EMD, ENPP1, FGF23, FHL1, FKBP10, FKRP, FKTN, FLNA, FLNB, GAA, GMPPB, ISPD, KBTBD13, KLHL40, LAMA2, LAMP2, LARGE, LMNA, LRP5, MYH7, NEB, OCRL, P3H1, PHEX, PIEZO2, PLOD2, POMGNT1, POMT1, POMT2, PPIB, PYCR1, RAPSN, RYR1, SELENON, SERPINF1, SERPINH1, SIL1, SLC34A3, TMEM43, TMEM5, TNNT1, TPM2, TPM3		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOLYSIS	7	BANF1, MMP2, SQSTM1, TNFRSF11A, TREM2, TYROBP, WNK1		Sequencing	1750
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOPETROSIS	21	AMER1, ANKH, CA2, CLCN7, COL1A1, CTSK, DLX3, FAM20C, GJA1, IKBKG, LEMD3, LRP4, LRP5, PTH1R, SLC29A3, SLCO2A1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TYROBP		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOPETROSIS (PANEL 2)	6	CA2, CLCN7, OSTM1, PLEKHM1, TCIRG1, TNFRSF11A		Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOPETROSIS (PANEL 3)	6	ANKH, DLX3, HPGD, LRP4, SOST, TGFB1, TNFRSF11B		Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	OSTEOPOROSIS	3	LRP5, WNT1, PLS3		Sequencing	1650
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	PATELLAR DYSOSTOSES	3	LMX1B, PITX1, TBX4		Sequencing	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	POLYDACTYLY - SYNDACTYLY - TRIPHALANGISM	6	GLI3, HOXD10, HOXD13, LMBR1, LRP4, PITX1		Sequencing	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	POLYMICROGYRIA	14	ADGRG1, AKT3, CHD7, FH, GPSM2, KIF1BP, NDE1, NSDHL, SRPX2, TUBA1A, TUBA8, TUBB2B, TUBB3, WDR62		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	PSEUDOXANTHOMA ELASTICUM	4	ABCC6, ENPP1, GGCX, VEGFA (Hotspots)		Sequencing	1000
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	RHEUMATOID-LIKE OSTEOARTHROPATHIES	7	ACAN, COL2A1, HPGD, IL1RN, LPIN2, TRPV4, WISP3		Sequencing	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SECKEL SYNDROME	6	ATR, CENPJ, CEP152, CEP63, PCNT, RBBP8		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SEPTO-OPTIC DYSPLASIA	4	HESX1, OTX2, PAX6, SOX2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SEX DEVELOPMENT DISORDERS (DSD), HERMAPHRODITISM	29	AMH, AMHR2, AR, ARX, CBX2, CYP11A1, CYP11B1, CYP17A1, CYP19A1, DHCR7, DHH, DMRT1, HSD11B1, HSD17B3, HSD3B2, INSL3, LHCGR, MAMLD1, NR3C1, NR5A1, POR, RSPO1, RXFP2, SOX9, SRD5A2, STAR, TSPYL1, WT1, WNT4		Sequencing	1200
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SHORT RIB DYSPLASIA / ASPHYXIATING THORACIC DYSPLASIA	15	CSPP1, DYNC2H1, EVC, EVC2, GLI2, IFT122, IFT140, IFT172, IFT80, NEK1, TCTN3, TTC21B, WDR19, WDR34, WDR35		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SHORT STATURE	61	AKT1, ATR, BMP2, BMP4, BMPR1A, CDC6, CDT1, CENPJ, CEP152, CEP63, CREBBP, CUL7, DHCR7, EP300, EYA1, FGD1, FGF3, FGFR3, FOXL2, GH1, GHR, GHRHR, GLI2, HESX1, IGF1, IGF1R, IGFALS, INSR, IRS1, KRAS, LHX3, LHX4, NIPBL, NOTCH2, NR5A1, OBSL1, ORC1, ORC4, ORC6, OTX2, PCNT, PITX2, POU1F1, PROP1, PTCH1, PTPN11, RAF1, RBBP8, RNU4ATAC, SHH, SHOX, SIX3, SMC1A, SOS1, SOX2, SOX3, STAT5B, TBX19, TBX3, TGIF1,		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SKELETAL DYSPLASIA WITH ABNORMAL MINERALIZATION	27	ALPL, ANKH, B4GALT7, CASR, CLCN5, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, CRTAP, CYP27B1, ENPP1, FBN1, FGF23, FKBP10, P3H1, PHEX, PLOD2, PPIB, SERPINF1, SLC34A3, SLC39A13, SOX9, TNFRSF11A, TNFRSF11B, VDR		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	SPONDYLOMETAPHYSEAL / SPONDYLOEPI-(META)-PHYSEAL DYSPLASIA	18	ACP5, B3GALT6, CANT1, CHST3, COL11A1, COL11A2, COL2A1, DYM, EIF2AK3, HSPG2, MATN3, PAPSS2, RMRP, SLC39A13, SMARCAL1, TRAPPC2, TRPV4, WISP3		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	STICKLER SYNDROME	8	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LRP2, VCAN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Malformations	WARBURG MICRO SYNDROME	3	RAB3GAP1, RAB3GAP2, RAB18		Sequencing	3100

Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	COMPREHENSIVE METABOLISM	354	ABCC8, ABCD1, ABCD4, ACAA1, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADAMTSL2, ADAR, ADCK3, ADSL, AGA, AGK, AGL, AGPAT2, AKT2, ALAD, ALAS2, ALDH5A1, ALDH7A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMT, ANO10, ANTXR2, APTX, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATP13A2, ATP6V0A2, AUH, B3GLCT, B4GALT1, BCKDHA, BCKDHB, BSCL2, BTD, C10ORF2, C12ORF65, CACNA1S, CAV1, CAV3, CBS, CD320, CKMT1A, CKMT1B, CKMT2, CLCN1, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CNNM1, CNNM2, CNNM4, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COQ2, COQ6, COQ9, CPOX, CPS1, CPT1A, CPT1B, CPT2, CTNS, CTSA, CTSC, CTSD, CTSK, DBT, DDOST, DGUOK, DHCR7, DHDDS, DLD, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DYM, ECHS1, EGF, ENO3, EPM2A, ETFA, ETFB, ETFDH, FBP1, FBXL4, FECH, FH, FLNA, FLNB, FOLR1, FUCA1, FXYD2, G6PC, GAA, GALC, GALNS, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GCK, GIF, GLA, GLB1, GLDC, GLUD1, GMPPA, GNE, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GPHN, GUSB, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HAMP, HCFC1, HEXA, HEXB, HFE, HFE2, HGSNAT, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HRAS, HSD17B10, HSD17B4, HYAL1, IDS, IDUA, IFIH1, INSR, ISCU, IVD, KCNA1, KCNJ11, KCNJ2, L2HGDH, LAMA2, LAMP2, LDB3, LDHA, LIPA, LMBRD1, LMNA, LPIN1, MAGT1, MAN1B1, MANBA, MCCC1, MCCC2, MCEE, MCOLN1, MFN2, MFSD8, MGAT2, MMAA, MMAB, MMACHC, MMADHC, MMGT1, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MTHFR, MTR, MTRR, MUT, MYOT, NAGLU, NAGS, NDUFS1, NEU1, NHLRC1, NIPA2, NPC1, NPC2, OAT, OPA1, OPA3, OTC, OXCT1, PAH, PC, PCBD1, PCCA, PCCB, PCK1, PCK2, PDHA1, PDHB, PDHX, PDSS1, PDSS2, PDX1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG1, PHKG2, PHYH, PLIN1, PMM2, POLG, POLG2, PPARG, PPOX, PPT1, PRKAG2, PRKAG3, PRODH, PSAP, PTRF, PTS, PYGL, PYGM, QDPR, RAI1, RBCK1, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, RPN2, RRM2B, RYR1, SAMHD1, SCN4A, SEC23B, SGSH, SLC12A3, SLC16A1, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A3, SLC25A4, SLC2A2, SLC30A10, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A4, SLC3A1, SLC40A1, SLC41A2, SLC41A3, SLC46A1, SLC6A19, SLC6A8, SLC7A7, SLC7A9, SMPD1, SPG7, SRD5A3, SSR4, STT3A, STT3B, SUCLA2, SUCLG1, SUMF1, SUOX, TAZ, TBC1D4, TCF4, TCN2, TFR2,		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	AICARDI-GOUTIÈRES SYNDROME	7	ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	COENZYME Q10 DEFICIENCY	11	ADCK3, ANO10, APTX, COQ2, COQ6, COQ9, ETFA, ETFB, ETFDH, PDSS1, PDSS2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)		CONGENITAL DISORDERS OF GLYCOSYLATION	47	ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B3GLCT, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, GMPPA, GNE, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, PGM1, PMM2, RFT1, RPN2, SEC23B, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, STT3A, STT3B,		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	CREATINE METABOLISM DEFICIENCY	6	CKMT1A, CKMT1B, CKMT2, GAMT, GATM, SLC6A8		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	CYSTINURIA	2	SLC3A1, SLC7A9		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	FATTY ACID OXIDATION SYNDROME	25	ACAA1, ACAD9, ACADL, ACADM, ACADS, ACADVL, ALDH5A1, CPT1A, CPT2, ECHS1, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, PPARG, SLC22A5, SLC25A20, TAZ		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	GLYCOGEN STORAGE DISORDER	29	AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	HEMOCHROMATOSIS	5	HAMP, HFE, HFE2, SLC40A1, TFR2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	HYPERLIPIDEMIA	11	ABCA1, ABCG5, ABCG8, APOA1, APOB, APOC3, APOE, LDLR, LDLRAP1, LPL, PCSK9		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	HYPERLIPIDEMIA (PANEL 2)	11	ABCG5, ABCG8, APOB, APOC2, APOE, GPIHBP1, LDLR, LDLRAP1, LMF1, LPL, PCSK9		Sequencing	1550
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	HYPERPHENYLALANINEMIA	5	GCH1, PAH, PCBD1, PTS, QDPR		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	HYPOGLYCEMIA, HYPERINSULINISM AND KETONE METABOLISM	18	ABCC8, ACAT1, FBP1, GCK, GLUD1, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, OXCT1, PCK1, PCK2, PDX1, SLC16A1, UCP2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	HYPOMAGNESEMIA	17	CLDN16, CLDN19, CNNM1, CNNM2, CNNM4, EGF, FXYD2, HNF1B, KCNA1, MAGT1, MMGT1, NIPA2, SLC12A3, SLC41A2, SLC41A3, TRPM6, TRPM7		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	LIPODYSTROPHY	10	AGPAT2, AKT2, BSCL2, CAV1, LMNA, PLIN1, PPARG, PTRF, TBC1D4, ZMPSTE24		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	LYSOSOMAL DISORDERS AND MUCOPOLYSACCHARIDOSIS	99	ABCC8, ACY1, ADAMTSL2, ADSL, AGA, ALDH5A1, ALDH7A1, AMT, ANTXR2, ARG1, ARSA, ARSB, ASAH1, ASPA, ATP13A2, BTD, CLN3, CLN5, CLN6, CLN8, COL11A2, COL2A1, CTNS, CTSA, CTSC, CTSD, CTSK, DHCR7, DPYD, DYM, ETFA, ETFB, ETFDH, FH, FOLR1, FUCA1, GAA, GALC, GALNS, GAMT, GBA, GCDH, GLA, GLB1, GLDC, GNE, GNPTAB, GNPTG, GNS, GPC3, GUSB, HEXA, HEXB, HGSNAT, HPD, HRAS, HYAL1, IDS, IDUA, L2HGDH, LAMA2, LDB3, LIPA, MAN1B1, MANBA, MCOLN1, MFSD8, MOCS1, MOCS2, MYOT, NAGLU, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX16, PEX26, PEX3, PEX5, PEX6, PGK1, PHYH, PPT1, PRODH, PSAP, QDPR, RAI1, SGSH, SLC17A5, SLC25A15, SLC46A1, SMPD1, SUMF1, SUOX, TCF4, TPP1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	METABOLIC MYOPATHY AND RHABDOMYOLYSIS	40	ACAD9, ACADL, ACADM, ACADVL, AGL, ALDOA, C10ORF2, CAV3, CPT1B, CPT2, ENO3, ETFA, ETFB, ETFDH, GAA, GBE1, GYG1, GYS1, HADHA, HADHB, ISCU, LDHA, LPIN1, OPA1, OPA3, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKG1, POLG, PYGM, RBCK1, RRM2B, RYR1, SLC22A5, SUCLA2, TK2, TYMP		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	MITOCHONDRIAL DISEASE	37	MTRNR1, MTRNR2, MTND1, MTND2, MTND3, MTND4L, MTND4, MTND5, MTND6, MTCO1, MTCO2, MTCO3, MTATP8, MTATP6, MTCYB, MTTF, MTTV, MTTL1, MTTI, MTTQ, MTTM, MTTW, MTTA, MTTN, MTTC, MTTY, MTTS1, MTTD, MTTK, MTTG, MTTR, MTTH, MTTS2, MTTL2, MTTE, MTTT, MTTP		Sequencing of the Entire mitochondrial genome (16569 bp) including all the 37 genes (2 rRNA genes, 22 tRNA genes, and 13 protein-coding genes) and non-coding regions	750

Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	MITOCHONDRIAL DNA DEPLETION SYNDROME	25	AGK, APTX, AUH, C10ORF2, C12ORF65, DGUOK, FBXL4, MFN2, MPV17, NDUFS1, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A3, SLC25A4, SPG7, SUCLA2, SUCLG1, TIMM8A, TK2, TMEM126A, TYMP, WFS1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	MITONUCLEAR GENES	132	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, 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, SLC37A4, SOD1, SPG7, SUCLA2, SUCLG1, SURF1, TACO1, TAZ, TIMM8A, TK2, TMEM126A, TMEM70, TRMU, TSFM, TTC19, TUFM, TYMP, UQCRB, UQCRQ,		Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	OBESITY	36	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CRTC1, CUL4B, DYRK1B, GNAS, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, PYY, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDPCP		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	ORGANIC ACIDEMIA/ACIDURIA & COBALAMIN DEFICIENCY	32	ABCD4, ACAT1, ACSF3, BCKDHA, BCKDHB, CBS, CD320, DBT, DLD, ETFA, ETFB, ETFDH, GCDH, GIF, HCFC1, HMGCL, IVD, LMBRD1, MCCC1, MCCC2, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, PCCA, PCCB, TCN2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	PERIODIC PARALYSIS	4	CACNA1S, CLCN1, KCNJ2, SCN4A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	PEROXISOMAL DISORDERS	18	ABCD1, ACOX1, AMACR, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Metabolic Disorders	PORPHYRIA	9	ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	ALPORT SYNDROME	6	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	BARDET-BIEDL SYNDROME	18	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, IFT172, MKKS, MKS1, SDCCAG8, TMEM67, TRIM32, TTC8		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	BARTTER SYNDROME	8	BSND, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, SLC12A1, SLC12A3		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	BRANCHIO-OTO-RENAL (BOR) SYNDROME	4	EYA1, SIX1, SIX5, TFAP2A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	CILIOPATHY	83	AHI1, ALMS1, ANKS6, ARL13B, ARL6, ARMC4, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21ORF59, C5ORF42, CC2D2A, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CEP164, CEP290, CEP41, CEP83, CFTR, CSPP1, DCDC2, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, GLIS2, HYDIN, IFT172, INPP5E, INVS, IQCB1, KIAA0586, KIF7, LRRC6, MKKS, MKS1, NEK8, NME8, NPHP1, NPHP3, NPHP4, OFD1, RPGR, RPGRIP1L, RSPH1, RSPH4A, RSPH9, SDCCAG8, SPAG1, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRIM32, TTC21B, TTC8, WDR19, ZMYND10, ZNF423		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	CYSTIC KIDNEY DISEASE	9	BICC1, EYA1, HNF1B, PAX2, PKD1, PKD2, PKHD1, SIX5, UMOD		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	DIABETES INSIPIDUS	3	AQP2, AVP, AVPR2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	HEMOLYTIC UREMIC SYNDROME	13	ADAMTS13, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, THBD		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	HYPOMAGNESEMIA	17	CLDN16, CLDN19, CNNM1, CNNM2, CNNM4, EGF, FXYD2, HNF1B, KCNA1, MAGT1, MMGT1, NIPA2, SLC12A3, SLC41A2, SLC41A3, TRPM6, TRPM7		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	HYPOPHOSPHATEMIC RICKETS	10	CLCN5, DMP1, ENPP1, FAH, FGF23, KL, PHEX, SLC34A1, SLC34A3, VDR		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	JOUBERT SYNDROME	29	AHI1, ARL13B, B9D1, B9D2, C5ORF42, CC2D2A, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, NPHP1, NPHP3, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	LIDDLE SYNDROME	2	SCNN1B, SCNN1G		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	MECKEL SYNDROME	11	B9D1, B9D2, CC2D2A, CEP290, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM216, TMEM231, TMEM67		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	MONOGENIC OBESITY	36	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CRTC1, CUL4B, DYRK1B, GNAS, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, POMC, PPARG, PYY, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDPCP		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	NEPHRONOPHTHISIS	19	ANKS6, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, ZNF423		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	NEPHROTIC SYNDROME	29	ACTN4, ADCK4, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, CRB2, DGKE, EMP2, INF2, ITGA3, LAMB2, LMX1B, MYH9, MYO1E, NPHS1, NPHS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, TTC21B, WT1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	POLYCYSTIC KIDNEY DISEASE	8	BICC1, LRP5, NOTCH2, PKD1, PKD2, PKHD1, PRKCSH, SEC63		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	PRIMARY CILIARY DYSKINESIA	32	ARMC4, C21ORF59, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, HYDIN, INVS, LRRC6, NME8, OFD1, RPGR, RSPH1, RSPH4A, RSPH9, SPAG1, ZMYND10		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	PRIMARY HYPEROXALURIA	3	AGXT, GRHPR, HOGA1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	PSEUDOHYPOALDOSTERONISM	9	CUL3, HSD11B2, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	RENAL MALFORMATION	15	ACE, BMP4, DSTYK, EYA1, FANCB, FOXC2, FREM1, GATA3, HNF1B, PAX2, REN, RET, SIX1, SIX5, WT1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	RENAL TUBULAR ACIDOSIS	5	ATP6V0A4, ATP6V1B1, CA2, SLC4A1, SLC4A4		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Nephrology	SENIOR-LOKEN SYNDROME	7	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	AMYOTROPHIC LATERAL SCLEROSIS (ALS)	31	ALS2, ANG, ATL1, BSCL2, CHCHD10, CHMP2B, DCTN1, FIG4, FUS, GBE1, GRN, HEXA, HNRNPA1, HSPD1, KIAA0196, KIF5A, OPTN, PRF1, REEP1, SETX, SLC52A2, SLC52A3, SOD1, SPAST, SPG11, SPG20, SQSTM1, TARDBP, UBQLN2, VAPB, VCP		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	ATAXIA	141	ABCB7, ABHD12, ACO2, ADCK3, AFG3L2, AHI1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATN1, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C10ORF2, C5ORF42, CA8, CACNA1A, CACNB4, CAMTA1, CASK, CC2D2A, CCDC28B, CCDC88C, CEP290, CEP41, CLCN2, CLN5, CLPP, COX20, CSTB, CWF19L1, CYP27A1, DNAJC19, DNMT1, EEF2, ELOVL4, ELOVL5, FBXL4, FGF14, FLVCR1, FMR1, FXN, GBA2, GFAP, GOSR2, GRID2, GRM1, GSS, HARS2, HTT, INPP5E, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, KIF7, LAMA1, LARS2, MARS2, MKKS, MKS1, MRE11A, MTPAP, MTTP, NEDD4, NOL3, NOP56, NPHP1, OFD1, OPA1, OPHN1, PAX6, PDYN, PEX7, PHYH, PNKD, PNKP, PNPLA6, POLG, PPP2R2B, PRKCG, PRRT2, RPGRIP1L, RUBCN, SACS, SETX, SIL1, SLC1A3, SLC2A1, SLC52A2, SLC9A6, SNX14, SPG7, SPTBN2, STUB1, SYT14, TBP, TCTN1, TCTN2, TCTN3, TDP1, TGM6, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRIM32, TTBK2, TTC8, TTPA, TUBB4A, VAMP1, VLDLR, WDPCP, WDR81, WFS1, WWOX, ZNF423, ZNF592		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	AUTISM SPECTRUM DISORDERS	13	CACNA1C, DHCR7, EN2, GAMT, MECP2, NLGN3, NLGN4X, NSD1, PDE8B, PTEN, RPL10, TSC1, TSC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	CEROID LIPOFUSCINOSIS AND PROGRESSIVE MYOCLONIC EPILEPSY	28	AFG3L2, ASAH1, ATP13A2, CERS1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, CTSF, DNAJC5, EPM2A, FOLR1, GOSR2, GRN, KCNC1, KCTD7, MFSD8, NEU1, NHLRC1, PPT1, PRICKLE1, PRICKLE2, SCARB2, SERPINI1, TBC1D24, TPP1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	CHARCOT-MARIE-TOOTH NEUROPATHY	86	AARS, AIFM1, AMACR, ARHGEF10, ATL1, ATL3, ATP7A, BAG3, BSCL2, C12ORF65, CCT5, COX10, COX6A1, CTDP1, DCAF8, DCTN1, DHTKD1, DNM2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, FXN, GAN, GARS, GDAP1, GJB1, GNB4, GNE, HADHB, HARS, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS, KIF1A, KIF1B, KIF5A, LDB3, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MPZ, MTMR2, MYOT, NDRG1, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, POLG, PRPS1, PRX, RAB7A, REEP1, SACS, SBF1, SBF2, SCN9A, SETX, SH3TC2, SLC12A6, SMAD3, SPG11, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TYMP, VCP, WNK1, YARS		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	COENZYME Q10 DEFICIENCY	11	ADCK3, ANO10, APTX, COQ2, COQ6, COQ9, ETFA, ETFB, ETFDH, PDSS1, PDSS2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	CONGENITAL HYPOVENTILATION	7	ASCL1, BDNF, BMP2, EDN3, PHOX2A, PHOX2B, RET		Sequencing	1650
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYASTHENIA	17	AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, DPAGT1, GFPT1, LAMB2, MUSK, PLEC, RAPSN, SCN4A, STIM1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	DEMENTIA	17	APOE, APP, CHMP2B, CSF1R, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, SIGMAR1, SORL1, TARDBP, TREM2, UBE3A, UBQLN2, VCP		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	DYSTONIA	6	GCH1 (DYT5), TH, SGCE (DYT11), SPR, ATP1A3 (DYT12), PRKRA (DYT16)		Sequencing	2900
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	EPILEPSY	194	ABCD1, ADAR, ADSL, AFG3L2, AGA, AIMP1, ALDH5A1, ALDH7A1, ALG13, AMACR, AMT, ARG1, ARHGEF9, ARSA, ARX, ASAH1, ASPA, ATP13A2, ATRX, BTD, CACNA1A, CACNA1H, CACNB4, CASK, CASR, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL4A1, COX15, CPT2, CSF1R, CSTB, CTSD, CTSF, CUL4B, DARS2, DCX, DEPDC5, DNAJC5, DNM1, DOCK7, DPYD, EARS2, EEF1A2, EFHC1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPM2A, ETFA, ETFB, ETFDH, FAM126A, FH, FLNA, FOLR1, FOXG1, FOXRED1, GABRA1, GABRB3, GABRG2, GALC, GAMT, GCDH, GCH1, GFAP, GJC2, GLDC, GNAO1, GNE, GOSR2, GPHN, GRIA3, GRIN2A, GRIN2B, GRN, HCN1, HEPACAM, HNRNPU, HSD17B10, HSPD1, IQSEC2, KCNA1, KCNA2, KCNB1, KCNC1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIF1A, L2HGDH, LGI1, MARS2, MBD5, MECP2, MED12, MEF2C, MFSD8, MLC1, MOCS1, MTHFR, MTOR, NDUFAF5, NECAP1, NEU1, NHLRC1, NOTCH3, NRXN1, OFD1, OPHN1, PCDH19, PGK1, PHF6, PIGA, PLCB1, PLP1, PNKP, PNPO, POLR3A, POLR3B, PPT1, PRICKLE1, PRICKLE2, PRODH, PRRT2, PSAP, PTS, PURA, QDPR, RAB39B, RELN, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SERPINI1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A15, SLC25A22, SLC2A1, SLC35A2, SLC46A1, SLC6A1, SLC6A8, SLC9A6, SMS, SNAP25, SOX10, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SUMF1, SUOX, SYN1, SYNGAP1, SZT2, TBC1D24, TCF4, TPP1, TREX1, TSC1, TSC2, TUBB4A, UBE2A, UBE3A, WDR45, WWOX, ZEB2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	EPILEPSY AND X-LINKED DEVELOPMENTAL DELAY	22	ARHGEF9, ARX, ATRX, CASK, CDKL5, CUL4B, GRIA3, HSD17B10, IQSEC2, KDM5C, MECP2, MED12, OFD1, OPHN1, PCDH19, PGK1, PHF6, RAB39B, SLC9A6, SMS, SYN1, UBE2A			
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	EPILEPTIC ENCEPHALOPATHY	83	ADAR, ADSL, ALDH7A1, ALG13, AMT, ARHGEF9, ARX, CACNA1A, CASK, CDKL5, CHD2, CNTNAP2, CPT2, DCX, DNM1, DOCK7, EEF1A2, FLNA, FOXG1, GABRA1, GABRB3, GABRG2, GAMT, GLDC, GNAO1, GPHN, GRIN2A, GRIN2B, HCN1, HEPACAM, HNRNPU, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, KIF1A, MBD5, MECP2, MEF2C, MOCS1, MTHFR, NECAP1, NRXN1, PCDH19, PIGA, PLCB1, PNKP, PNPO, PURA, RNASEH2A, RNASEH2B, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SNAP25, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SZT2, TBC1D24, TCF4, TREX1, TSC1, TSC2, UBE3A, WDR45, WWOX, ZEB2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	LEUKODYSTROPHY AND LEUKOENCEPHALOPATHY	42	ABCD1, ADAR, AIMP1, ARSA, ASPA, CLCN2, COL4A1, COX15, CSF1R, DARS2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, FAM126A, FOLR1, FOXRED1, GALC, GFAP, GJC2, HEPACAM, HSPD1, L2HGDH, MARS2, MLC1, NDUFAF5, NOTCH3, PLP1, POLR3A, POLR3B, PSAP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SOX10, SUMF1, TREX1, TUBB4A		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MENTAL RETARDATION (AUTOSOMAL RECESSIVE)	20	AP4B1, AP4E1, AP4M1, AP4S1, CA8, CC2D1A, CNTNAP2, CRBN, ERLIN2, GRIK2, MAN1B1, NRXN1, PRSS12, ST3GAL3, STXBP1, TRAPPC9, TUSC3, VLDLR, ZC3H14, ZNF526		Sequencing	3950
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MENTAL RETARDATION (X-LINKED)	94	ABCD1, ACSL4, AFF2, AGTR2, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CDKL5, CUL4B, DCX, DKC1, DLG3, ELK1, FANCB, FGD1, FLNA, FMR1, FTSJ1, GDI1, GK, GPC3, GRIA3, HCCS, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIAA2022, KLF8, L1CAM, LAMP2, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MTM1, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NSDHL, NXF5, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRPS1, RAB39B, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TIMM8A, TSPAN7, UBE2A, UPF3B, ZCCHC12, ZDHHC15, ZDHHC9, ZNF41, ZNF674, ZNF711, ZNF81		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MICROCEPHALY	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		Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MIGRAINE	10	ATP1A2, ATP1A3, CACNA1A, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY (COLLAGEN TYPE VI-RELATED)	6	COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY (CONGENITAL)	25	B3GALNT2, B3GNT1, CHKB, COL12A1, COL6A1, COL6A3, DAG1, DPM1, DPM3, FKRP, FKTN, GMPPB, GOSR2, ISPD, ITGA7, LAMA2, LARGE, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, ST3GAL4, TMEM5		Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY (EMERY-DREIFUSS)	6	DMD, EMD, FHL1, LMNA, TMEM43, TTN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY (LIMB GIRDLE AND CONGENITAL)	33	ANO5, CAPN3, CAV3, COL4A1, COL4A2, DES, DMD, DNAJB6, DYSF, FKRP, FKTN, GMPPB, ISPD, LAMA2, LARGE, LIMS2, LMNA, MYOT, PNPLA2, POMGNT1, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MUSCULAR DYSTROPHY / MYOPATHY	51	ACTA1, ANO5, CAPN3, CAV3, CFL2, COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, DES, DMD, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, GMPPB, ISPD, KBTBD13, KLHL40, KLHL41, LAMA2, LARGE, LIMS2, LMNA, LMOD3, MTM1, MYOT, NEB, PNPLA2, POMGNT1, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, SMCHD1, TCAP, TMEM43, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TTN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYASTHENIA	17	AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, DPAGT1, GFPT1, LAMB2, MUSK, PLEC, RAPSN, SCN4A, STIM1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYASTHENIA (PANEL 2)	40	AGRN, BLK, C5, CCL21, CCR6, CD2, CD28, CD40, CD58, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, GFPT1, IL2, IL21, IL2RA, IL2RB,IL6ST, IRF5, MUSK, PRDM1, PRKCQ, PTPRC, RAG1, RAPSN, RBPJ, SCN4A, STAT4, TAGAP, TNFAIP3, TNFRSF14, TRAF1, TRAF3IP2, TRAF6, VAMP1		Sequencing	2050
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYOPATHY	34	ACTA1, ATP2A1, BAG3, BIN1, CAV3, CFL2, CLCN1, CNTN1, COL6A1, COL6A2, COL6A3, CRYAB, DES, DNM2, DYSF, FHL1, FLNC, GNE, ISCU, KBTBD13, LDB3, MATR3, MTM1, MYH2, MYH7, MYOT, NEB, RYR1, SEPN1, TNNT1, TPM2, TPM3, TTN, VCP		Sequencing	2050
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYOPATHY (CENTRONUCLEAR)	6	BIN1, CCDC78, DNM2, MTM1, MYF6, RYR1		Sequencing	1700
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYOPATHY (DISTAL)	18	ANO5, BAG3, CAV3, CRYAB, DES, DNAJB6, DYSF, FHL1, FLNC, GNE, LDB3, MATR3, MYH7, MYOT, TCAP, TIA1, TTN, VCP		Sequencing	1800
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	MYOPATHY (NEMALINE)	11	ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MTM1, NEB, TNNT1, TPM2, TPM3			
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION (NBIA)	7	PANK2, PLA2G6, C19ORF12, FTL, FA2H, ATP13A2, CP		Sequencing	2950
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	PARKINSON DISEASE	12	ATP13A2, DNAJC6, FBXO7, LRRK2, MAPT, PARK2, PARK7, PINK1, PLA2G6, SLC6A3, SNCA, VPS35		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	PARKINSON DISEASE (PANEL 2)	23	ADH1C, ATP13A2, ATP1A3, DCTN1, EIF4G1, FBXO7, GBA, GCH1, GIGYF2, HTRA2, LRRK2, MAPT, PARK2, PARK7, PDXK, PINK1, PLA2G6, POLG1, SNCA, SNCAIP, SNCB, UCHL1, VPS35		Sequencing	2050
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	PARKINSON DISEASE WITH DYSTONIA	5	TAF1, SLC6A3, ATP1A3, PRKRA, PLA2G6		Sequencing	2750
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	PERIODIC PARALYSIS	4	CACNA1S, CLCN1, KCNJ2, SCN4A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	SPASTIC PARAPLEGIA	35	AFG3L2, ALS2, ATL1, B4GALNT1, BSCL2, C12ORF65, C19ORF12, CYP7B1, DDHD1, DDHD2, FA2H, FXN, GALC, GBA2, GJC2, HSPD1, KDM5C, KIAA0196, KIF1A, KIF5A, L1CAM, MARS2, NIPA1, PLP1, PNPLA6, REEP1, SACS, SETX, SLC16A2, SLC33A1, SPAST, SPG11, SPG20, SPG7, SPR		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	SPASTIC PARAPLEGIA (PANEL 2)	27	ALS2, AP5Z1, ATL1, BSCL2, CCT5, CYP7B1, FA2H, GJC2, HSPD1, KDM5C, KIAA0196, KIF1A, KIF5A, L1CAM, NIPA1, PLP1, PNPLA6, REEP1, SLC16A2, SLC33A1, SPAST, SPG7, SPG11, SPG20, SPG21, ZFYVE26, ZFYVE27		Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	SPINAL MUSCULAR ATROPHY	29	AARS, ASAH1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, FBXO38, GARS, HEXA, HSPB1, HSPB3, HSPB8, IGHMBP2, LAS1L, PLEKHG5, REEP1, SCO2, SLC5A7, SMN1, SMN2, TRPV4, UBA1, VAPB, YRK1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Neurology	ZELLWEGER SYNDROME	14	EX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH		Sequencing	1500
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	ACHROMATOPSIA	6	ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	ALBINISM	19	AP3B1, BLOC1S3, BLOC1S6, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITF, MYO5A, OCA2, RAB27A, SLC45A2, TYR, TYRP1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	BARDET-BIEDL SYNDROME	18	ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, IFT172, MKKS, MKS1, SDCCAG8, TMEM67, TRIM32, TTC8		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	BRITTLE CORNEA	2			Sequencing	600

Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CATARACT	54	ABCB6, ADAMTSL4, AGK, ALDH18A1, BCOR, BFSP2, COL11A1, COL18A1, COL2A1, COL4A1, CRYAA, CRYAB, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CYP27A1, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXE3, FTL, FYCO1, FZD4, GALK1, GALT, GCNT2, GJA1, GJA3, GJA8, HSF4, LIM2, MAF, MYH9, NDP, NF2, NHS, OCRL, OPA3, PAX6, PITX3, RAB3GAP1, RECQL4, SIL1, SLC33A1, TDRD7, TFAP2A, TMEM70, WFS1, WRN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CONE ROD DYSTROPHY	32	ABCA4, ADAM9, AIPL1, BEST1, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CERKL, CLN3, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, FBLN5, GNAT2, GUCA1A, GUCY2D, KCNV2, MERTK, PDE6C, PDE6H, PROM1, PRPH2, RAX2, RDH5, RPGR, RPGRIP1, SEMA4A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CONGENITAL STATIONARY NIGHT BLINDNESS	17	CABP4, CACNA1F, CACNA2D4, CYP4V2, GNAT1, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RDH5, RHO, RLBP1, RPE65, SAG, TRPM1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CORNEAL DYSTROPHY	15	CHST6, COL5A1, CYP4V2, FOXE3, GJA8, KRT12, LCAT, LOXHD1, MAF, PITX2, SLC4A11, TCF4, TGFBI, ZEB1, ZNF469		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	CORNEAL DYSTROPHY (PANEL 2)	20	CHST6, COL5A1, COL8A2, CYP4V2, DCN, GSN, KRT3, KRT12, LOXHD1, PIKFYVE, PRDM5, SLC4A11, SOD1, ZEB1, ZNF469, TACSTD2, TCF4, TGFBI, UBIAD1, VSX1		Sequencing	1550
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	ECTOPIA LENTIS	3	LTBP2, ADAMTSL4, FBN1		Sequencing	1600
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	FLECKED RETINA DISORDERS	11	ABCA4, CHM, CYP4V2, ELOVL4, PROM1, PRPH2, RDH5, RHO, RLBP1, RS1, VPS13E		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	GLAUCOMA	17	CNTNAP2, COL4A1, CYP1B1, FOXC1, FOXE3, LMX1B, LTBP2, MAF, MYOC, OPA1, OPA3, OPTN, PAX6, PITX2, TBK1, TMEM126A, WDR36		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	JOUBERT SYNDROME	29	AHI1, ARL13B, B9D1, B9D2, C5ORF42, CC2D2A, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, NPHP1, NPHP3, OFD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	LEBER CONGENITAL AMAUROSIS	25	AIPL1, ALMS1, BBS4, CABP4, CEP290, CNGA3, CRB1, CRX, DTHD1, GUCY2D, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MYO7A, NMNAT1, RD3, RDH12, RDH5, RPE65, RPGRIP1, SPATA7, TULP1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	MACULAR DYSTROPHY	17	ABCA4, BEST1, CERKL, CNGB3, CRB1, ELOVL4, FBLN5, IMPG1, PROM1, PRPH2, RAX2, RDH12, RDH5, RLBP1, RP1L1, RPGR, RS1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	MACULAR DYSTROPHY (PANEL 2)	21	ABCA4, ARMS2, C2, C3, C9, CCR3, CFB, CFH, CFI, CST3, CXCL8, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, IL6, IL1A, NLRP3, RAX2, TLR4		Sequencing	1850
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	MICROPHTHALMIA, ANOPHTHALMIA AND ANTERIOR SEGMENT DYSGENESIS	32	ABCB6, BCOR, BMP4, CHD7, COL4A1, CYP1B1, ERCC2, ERCC5, ERCC6, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, GJA1, HCCS, HESX1, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, POBP1, RAB3GAP1, SHH, SIX3, SOX2, STRA6, TFAP2A, VPS13B, ZIC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	NEURO-OPHTHALMOLOGY	26	APTX, C10ORF2, C12ORF65, FRMD7, GPR143, HESX1, MFN2, NDUFS1, OPA1, OPA3, OTX2, PAX6, POLG, ROBO3, RRM2B, SALL4, SETX, SLC25A4, SOX2, SPG7, TIMM8A, TK2, TMEM126A, TUBB3, TYMP, WFS1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	OPTIC ATROPHY	10	C12ORF65, MFN2, NDUFS1, OPA1, OPA3, POLG, SPG7, TIMM8A, TMEM126A, WFS1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINAL DYSTROPHY	181	ABCA4, ABHD12, ADAM9, ADGRV1, AHI1, AIPL1, ALMS1, ARL13B, ARL6, ATF6, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C2ORF71, C5ORF42, C8ORF37, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDHR1, CEP164, CEP290, CEP41, CERKL, CHM, CIB2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CRB1, CRX, CSPP1, CYP4V2, DFNB31, DHDDS, DTHD1, EFEMP1, ELOVL4, EYS, FAM161A, FBLN5, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNPTG, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HARS, HK1, HMX1, IDH3B, IFT140, IFT172, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, KCNJ13, KCNV2, KIAA0586, KIF11, KIF7, KLHL7, LCA5, LRAT, LRIT3, LRP2, LRP5, MAK, MERTK, MKKS, MKS1, MVK, MYO7A, NDP, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPA1, OPA3, OTX2, PANK2, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PDZD7, PEX1, PEX2, PEX7, PHYH, PRCD, PROM1, PRPF3, PRPF31, PRPF8, PRPH2, RAX2, RBP3, RD3, RDH12, RDH5, RGR, RHO, RLBP1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SNRNP200, SPATA7, TCTN1, TCTN2, TCTN3, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTPA, TULP1, USH1C, USH1G, USH2A, VCAN, VPS13B, WDR19, ZNF423, ZNF513		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINITIS PIGMENTOSA	80	ABCA4, ABHD12, AIPL1, ARL6, BBS1, BBS2, BEST1, C2ORF71, C8ORF37, CDHR1, CEP290, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FLVCR1, GNPTG, GUCY2D, HK1, IDH3B, IMPDH1, IMPG2, KLHL7, LCA5, LRAT, MAK, MERTK, MVK, NMNAT1, NR2E3, NRL, OAT, OFD1, PANK2, PDE6A, PDE6B, PDE6G, PEX1, PEX2, PEX7, PHYH, PRCD, PROM1, PRPF3, PRPF31, PRPF8, PRPH2, RBP3, RDH12, RDH5, RGR, RHO, RLBP1, RP1, RP2, RPE65, RPGR, RPGRIP1, RS1, SAG, SEMA4A, SNRNP200, SPATA7, TOPORS, TTC8, TTPA, TULP1, USH1C, USH2A, VPS13B, WDR19, ZNF513		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINITIS PIGMENTOSA (AUTOSOMAL DOMINANT) (PANEL 3)	21	ASCC3L1, CM, CRX, FSCN2, GUCA1B, IMPDH1, KU-1L7, NR2E3, NRL, PRPF3, PRPF8, PRPF31, PRPH2-RDS, RDH12, RHO, ROM1, RP1, RP9, SEMA4A, TOPORS, VMD2-BEST1		Sequencing	2600
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINITIS PIGMENTOSA (AUTOSOMAL RECESSIVE) (PANEL 2)	56	BCA4, ASCC3L1, BEST1, C2ORF71, C8ORF37, CA4, CERKL, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, EYS, FAM161A, FSCN2, GUCA1B, IDH3B, IMPDH1, IMPG2, KLHL7, LRAT, MAK, MERTK, NR2E3, NRL, OFD, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF6, PRPF8, PRPF31, PRPH2, RBP3, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SPATA7, TTC8, TOPORS, TULP1, USH2A, ZNF513		Sequencing	1600
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	RETINOBLASTOMA	1	RB1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	SENIOR-LOKEN SYNDROME	7	CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8		Sequencing and Deletion-Duplication Testing of all Genes	1290

Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	STARGARDT DISEASE AND MACULAR DYSTROPHIES	13	ABCA4, BEST1, C1QTNF5, CDH3, CNGB3, ELOVL4, FSCN2, PROM1, PRPH2, RDH12, RP1L1, RPGR, TIMP3		Sequencing	2950
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	STICKLER SYNDROME	8	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LRP2, VCAN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	USHER SYNDROME	13	ABHD12, ADGRV1, CDH23, CIB2, CLRN1, DFNB31, HARS, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	USHER SYNDROME (PANEL 2)	12	ABHD12, CDH23, CLRN1, DFNB31, GPR98, HARS/USH3B, MYO7A, PCDH15, USH1C, USH1G, USH1J, USH2A		Sequencing	2100
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	USHER SYNDROME (PANEL 3)	20	ABHD12, CDH23, CIB2, CLRN1, COL4A6, DFNB31, DSPP (Excluding Exon 5), GIPC3, GPR98, HARS, KARS, LHFPL5, LOXHD1, MYO7A, PCDH15, PDZD7, TNC, USH2A, USH1C, USH1G		Sequencing	1490
Next Generation and Sanger Sequencing Platforms (NGS)	Ophthalmology	VITREORETINOPATHY	17	BEST1, CAPN5, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, FZD4, KCNJ13, LRP5, NDP, NR2F3, RS1, TSPAN12, VCAN		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	COMPREHENSIVE PULMONOLOGY	61	ABCA3, CCDC39, CCDC40, CFTR, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, CSF2RA, DKC1, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, EDN3, EFEMP2, ELMOD2, ELN, FBLN5, FLCN, FOXF1, GLRA1, HPS1, HPS4, ITGA3, LTBP4, MECP2, NF1, NKX2-1, NME8, PARN, PHOX2B, RAPSN, RET, RSPH4A, RSPH9, RTEL1, SCN4A, SCNN1A, SCNN1B, SERPINA1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC6A5, SLC7A7, SMPD1, STAT3, TERC, TERT, TINF2, TSC1, TSC2, ZEB2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	BRONCHIECTASIS	15	CCDC39, CCDC40, CFTR, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, NME8, RSPH4A, RSPH9, SCNN1A, SCNN1B		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	CENTRAL HYPOVENTILATION AND APNEA	15	CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, EDN3, GLRA1, MECP2, PHOX2B, RAPSN, RET, SCN4A, SLC6A5, ZEB2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	CYSTIC LUNG DISEASE	8	EFEMP2, ELN, FBLN5, FLCN, LTBP4, SERPINA1, TSC1, TSC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	HERMANSKY-PUDLAK SYNDROME	22	ABCA3, AP3B1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	INTERSTITIAL LUNG DISEASE	24	ABCA3, CSF2RA, DKC1, ELMOD2, HPS1, HPS4, ITGA3, NF1, NKX2-1, PARN, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, STAT3, TERC, TERT, TINF2, TSC1, TSC2		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	NEONATAL RESPIRATORY DISTRESS - SURFACTANT DYSFUNCTION	5	ABCA3, FOXF1, NKX2-1, SFTPB, SFTPC		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	PRIMARY CILIARY DYSKINESIA	32	ARMC4, C21ORF59, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DRC1, DYX1C1, HYDIN, INVS, LRRC6, NME8, OFD1, RPGR, RSPH1, RSPH4A, RSPH9, SPAG1, ZMYND10		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Pulmonology	PULMONARY ARTERY HYPERTENSION (PAH)	11	ACVRL1, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, KCNA5, KCNK3, RASA1, SMAD4, TBX4		Sequencing and Deletion-Duplication Testing of all Genes	1290
Next Generation and Sanger Sequencing Platforms (NGS)	Whole Exome (WES)	WHOLE EXOME (PANEL 1: PATIENT ONLY)	+/- 23.000	WHOLE EXOME		WES of patient only	1500
Next Generation and Sanger Sequencing Platforms (NGS)	Whole Exome (WES)	WHOLE EXOME (PANEL 2: TRIO OF PATIENT and 2 PARENTS)	+/- 23.000	WHOLE EXOME		WES TRIO analysis of patient and 2 parents to detect de novo variants	2500
Next Generation and Sanger Sequencing Platforms (NGS)	Whole Exome (WES)	WHOLE EXOME (PANEL 3: 2 SIBS and 2 PARENTS)	+/- 23.000	WHOLE EXOME		WES of 2 sibs and 2 parents to detect autosomal recessive variants	2700
Next Generation and Sanger Sequencing Platforms (NGS)	Whole Exome (WES)	WHOLE EXOME (PANEL 4: 3 AFFECTED FAMILY MEMBERS)	+/- 23.000	WHOLE EXOME		WES of 3 affected family members to detect autosomal dominant variants	2700