Fosterdiagnostik

Frågeställning/Analys	Vävnad	Kromosomområde Gen	Analys/Metod	Svarstid (dagar)	Rör
Kromosomanalys	CVS	Screening	Kromosomanalys CVS	21	Odlingsmediumrör
Kromosomanalys	Amnion	Screening	Kromosomanalys Amnion	21	Sterilt rör
Mikroarrayanalys vid fosterdiagnostik	CVS	Screening	Mikroarray prenatal	14	Odlingsmediumrör
Mikroarrayanalys vid fosterdiagnostik	Amnion	Screening	Mikroarray prenatal	14	Sterilt rör
Riktad analys	CVS	13,18,21,X,Y	QF-PCR QF-PCR prenatal inkl. MCC	7	Odlingsmediumrör
Riktad analys	Amnion	13,18,21,X,Y	QF-PCR QF-PCR prenatal inkl. MCC	7	Sterilt rör
Riktat test	Blod (maternellt)	13,18,21,X,Y	NIPT	21	BCT Streck rör
Riktat test	Blod (maternellt)	13,18,21,X,Y	NIPT Akut	14	BCT Streck rör
Riktat test	Blod (maternellt)	13,18,21,X,Y	NIPT skickeprov	21	BCT Streck rör
Misstänkt ärftlig sjukdom	CVS	Varierar beroende på frågeställning	Varierar beroende på frågeställning	Varierar beroende på frågeställning	Odlingsmediumrör
Misstänkt ärftlig sjukdom	Amnion	Varierar beroende på frågeställning	Varierar beroende på frågeställning	Varierar beroende på frågeställning	Sterilt rör

Missfall/intrauterin fosterdöd/abortmaterial	Vävnadsbiopsi	Varierar beroende på frågeställning	Varierar beroende på frågeställning	Varierar beroende på frågeställning	Sterilt rör

Kromosomanalys/FISH

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Akuta frågeställningar	Perifert blod	Varierar beroende på frågeställning	Varierar beroende på frågeställning	Varierar beroende på frågeställning. Vid frågor kontakta läkare på klinisk genetik 018-6122018	Heparin EDTA
Kromosomanalys (Syndromutredning)	Perifert blod	Screening	Kromosomanalys Blod	42	Heparin
Kromosomanalys (Könskromosomutredning)	Perifert blod	Screening	Kromosomanalys Blod	42	Heparin
Kromosomanalys (Infertilitetsutredning)	Perifert blod	Screening	Kromosomanalys Blod	42	Heparin
Kromosomanalys (Ägg-el.spermadonation)	Perifert blod	Screening	Kromosomanalys Blod	42	Heparin
Kromosomanalys (Upprepade missfall)	Perifert blod	Screening	Kromosomanalys Blod	42	Heparin
FISH-analys (Translokationsutredning)	Perifert blod	Riktad region	FISH Konstitutionellt metafas	90[#] ([#] vid beställning av unika prober TAT 130 dagar)	Heparin
Riktad analys	Perifert blod	13,18,21,X,Y	FISH Konstitutionellt interfas	7	Heparin

DNA-baserad diagnostik

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Riktad analys	Perifert blod	13,18,21,X,Y	QF-PCR	7	EDTA
22q11 del/dup syndrom	Perifert blod	22q11	MLPA enkel	56	EDTA
Akondroplasi (Se även *FGFR3*-relaterad skelettdysplasi)	Perifert blod	*FGFR3* (exon 10)	Sangersekvensering *FGFR3*	56	EDTA
Amyloidos	Perifert blod	Amyloidospanel v2, 17 gener *(APOA1, APOA2, APOA4, APOC2, APOC3, B2M, CST3, EFEMP1, FGA, GSN, LECT2, LYZ, MEFV, MVK, NLRP3, TNFRSF1A, TTR)*	NGS TruSeq helgenom In silico panel*	90	EDTA
Angelman syndrom	Perifert blod	15q11.2	MLPA metylering	56	EDTA
Androgenokänslighetssyndrom (AIS)	Perifert blod	*AR* inklusive repeatanalys	NGS TruSeq helgenom In silico panel*	90	EDTA
Arytmi och kardiomyopati (inkl. ARVC, Brugada syndrom, LQTS, CPVT, kardiomyopati, HCM, DCM)	Perifert blod	Arytmi och kardiomyopatipanel v2, 100 gener *ABCC9, ACADVL, ACTC1, ACTN2, AGL, ALMS1, ALPK3, BAG3, BRAF, CACNA1C, CACNA1D, CALM1, CALM2, CALM3, CASQ2, CBL, CDH2, CPT2, CRYAB, CSRP3, DES, DMD, DNAJC19, DOLK, DSC2, DSG2, DSP, ELAC2, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GATA4, GATA5, GJA5, GLA, HCN4, HFE, HRAS, JUP, KCNE1, KCNH2, KCNJ2, KCNQ1, KRAS, LAMP2, LMNA, LZTR1, MAP2K1, MAP2K2, MRAS, MTO1, MYBPC3, MYH7, MYL2, MYL3, MYL4, MYLK3, NF1, NKX2-5, NRAS, PCCA, PCCB, PKP2, PLN, PPA2, PPCS, PPP1CB, PRKAG2, PTPN11, RAF1, RASA1, RBM20, RIT1, RYR2, SCN5A, SDHA, SGCD, SHOC2, SLC22A5, SOS1, SOS2, SPRED1, TAZ, TBX20, TCAP, TMEM43,*	NGS TWIST In silico panel*	90	EDTA

		TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, VCL,			
Ataxi (inkl. CANVAS, episodisk ataxi, FXTAS, spastisk ataxi och spinocerebellär ataxi samt repeatanalys för ataxisjukdomar) Panelen inkluderar även det mitokondriella genomet.	Perifert blod	Ataxipanel v5, 755 gener och 18 repeatexpansioner *AAAS, AARS1, AARS2, ABCA2, ABCB7, ABCD1, ABHD12, ACO2, ACOX1, ACTL6B, ADA2, ADAR, ADGRG1, ADPRS, ADSL, AFG3L2, AGTPBP1, AHI1, AIFM1, AIMP1, ALDH18A1, ALDH5A1, ALG6, ALS2, AMACR, ANG, ANO10, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOB, APTX, AR, ARG1, ARL13B, ARL3, ARL6IP1, ARMC9, ARSA, ARV1, ARX, ASL, ASS1, ATAD3A, ATCAY, ATG5, ATG7, ATL1, ATM, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B3, ATP2B4, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRX, AUH, B4GALNT1, B9D1, B9D2, BBS1, BCKDHA, BCKDHB, BCS1L, BEAN1, BICD2, BOLA3, BRAT1, BSCL2, BTD, C19orf12, CA8, CACNA1A, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CAPN1, CARS1, CASK, CAV1, CC2D2A, CCDC88C, CCT5, CDK16, CDKL5, CEP104, CEP120, CEP290, CEP41, CHAMP1, CHCHD10, CHMP1A, CHMP2B, CHP1, CLCN2, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, COA5, COA7, COA8, COASY, COG1, COG4, COG5, COG7, COG8, COL18A1, COQ2, COQ4, COQ6, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6A2, COX6B1, CP, CPLANE1, CPS1, CPT1C, CRAT, CSPP1, CSTB, CTBP1, CTC1, CTDP1, CTNNA2, CTNNB1, CTSA, CTSD, CTSF, CUL4B, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DAB1, DARS1, DARS2, DBT, DCX, DDHD1, DDHD2, DGAT2, DHDDS, DHFR, DHX30, DKC1, DLAT, DLD, DMPK, DNAJC19, DNAJC3, DNAJC5, DNM1L, DNM2, DNMT1, DOCK3, DPM1, DPM2, DSTYK, DYNC1H1, DYRK1A, EBF3, EEF2, EGR2, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, ENTPD1, EPM2A, EPRS1, ERBB4, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ETHE1, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM149B1, FARS2, FASTKD2, FAT1, FAT2, FBXL4, FDXR, FGF12, FGF14, FIG4, FITM2, FKRP, FKTN, FLVCR1, FMR1, FOLR1, FOXG1, FOXRED1, FRMD4A, FTL, FUS, FXN, FZR1, GABRB1, GABRB2, GABRB3, GAD1, GALC, GALNT2, GAMT, GAN, GBA1, GBA2, GBE1, GCDH, GCH1, GCLC, GDAP2, GEMIN4, GEMIN5, GFAP, GJA1, GJB1, GJC2, GLB1, GLS, GMPPB, GOSR2, GPAA1, GPI, GRIA2, GRIA4, GRID2, GRIK2, GRM1, GRN, GSS, GTPBP2, HACE1, HARS1, HARS2, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HIP1R, HK1, HLCS, HNRNPA1, HNRNPH2, HPDL, HSD17B4, HSPD1, HTRA1, IBA57, IFIH1, IFT140, INPP5E, IQSEC1, IRF2BPL, ITM2B, ITPR1, JAM2, KATNIP, KCNA1, KCNA2, KCNC1, KCNC3, KCND3, KCNJ10, KCNMA1, KCNN2, KCNQ2, KCTD7, KIAA0586, KIDINS220, KIF1A, KIF1B, KIF1C, KIF5A, KIF7, KLC2, KY, L1CAM, L2HGDH, LAMA1, LARGE1, LARS2, LETM1, LIG4, LMNB1, LMNB2, LNPK, LRP4, LRPPRC, LRSAM1, LYRM7, LYST, MAB21L1, MAG, MAN2B1, MAPK8IP3,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		MARS1, MARS2, MAST1, MATR3, MBD5, MCOLN1, MECP2, MECR, MED13L, MFN2, MFSD8, MGAT2, MGME1, MICU1, MKS1, MLC1, MMACHC, MMADHC, MME, MORC2, MPDU1, MPV17, MPZ, MRE11, MSTO1, MT-ATP6, MT-ATP8, MTCL1, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTPAP, MTRFR, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MVK, NADK2, NALCN, NANS, NARS1, NAT8L, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEFH, NEFL, NEU1, NEXMIF , NF2, NFASC, NHLRC1, NIPA1, NKX2-1, NKX6-2, NMNAT1, NOL3, NPC1, NPC2, NPHP1, NPTX1, NR4A2, NT5C2, NTNG2, NUBPL, NUP62, NUS1, OFD1, OGDH, OGDHL, OPA1, OPA3, OPHN1, OPTN, OTC, OTUD4, PANK2, PARS2, PAX6, PAX9, PC, PCDH12, PCDH19, PCLO, PCNA, PCYT2, PDE6D, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFN1, PGK1, PGM3, PHYH, PIBF1, PIEZO2, PIGG, PIGS, PIGV, PIK3R5, PITRM1, PLA2G6, PLD3, PLP1, PMM2, PMP22, PMPCA, PMPCB, PNKD, PNKP, PNP, PNPLA6, PNPT1, POLG, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMT1, POU4F1, PPT1, PRDM8, PRDX3, PRF1, PRICKLE1, PRICKLE2, PRKCG, PRNP, PRPS1, PRRT2, PRX, PSAP, PSEN1, PTRH2, PTS, PUM1, PURA, PYCR2, QARS1, RAB11B, RAD50, RARS1, RARS2, REEP1, REEP2, RELN, REPS1, RFC4, RFT1, RNASEH1, RNASEH2B, RNASET2, RNF168, RNF170, RNF216, RNF220, ROGDI, RORA, RPGRIP1L, RPIA, RRM2B, RTEL1, RTN2, RTN4IP1, RUBCN, SACS, SAMD9L, SARS1, SARS2, SCARB2, SCN1A, SCN2A, SCN8A, SCO1, SCYL1, SDHA, SDHAF1, SDHB, SDHD, SELENOI, SEPSECS, SERAC1, SETX, SGCE, SH3TC2, SHMT2, SIGMAR1, SIL1, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC25A15, SLC25A46, SLC2A1, SLC30A9, SLC33A1, SLC39A4, SLC44A1, SLC46A1, SLC52A2, SLC52A3, SLC5A6, SLC6A1, SLC6A19, SLC9A1, SLC9A6, SNAP25, SNX14, SOD1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, STN1, STUB1, STXBP1, STXBP2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SVBP, SYNE1, SYNGAP1, SYT14, TACO1, TANC2, TANGO2, TARDBP, TBC1D23, TBC1D24, TBCE, TBK1, TCF20, TCF4, TCN2, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TELO2, TFG, TGM6, TH, THG1L, TINF2, TMEM106B, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM63A, TMEM67, TMEM70, TOE1, TOP3A, TPK1, TPP1, TPRKB, TRAPPC11, TRAPPC6B, TRIM32, TRMT5, TRNT1, TRPC3, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTC19, TTC21B, TTC8, TTPA, TTR, TUBA1A, TUBA4A,			

		TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TWNK, TXN2, TYMP, TYROBP, UBA5, UBAP1, UBE3A, UBQLN2, UBR4, UBTF, UCHL1, UNC80, UQCRB, UQCRQ, UROC1, VAMP1, VAPB, VARS2, VCP, VLDLR, VPS11, VPS13D, VPS37A, VPS41, VPS53, VRK1, VWA3B, WARS2, WASHC5, WDR26, WDR45B, WDR62, WDR73, WDR81, WFS1, WWOX, XPA, XRCC1, XRCC4, YME1L1, ZBTB18, ZFYVE26, ZIC1, ZIC4, ZNF423, ZSWIM6 Screening för patogena repeatexpansioner ingår för följande gener: *ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, BEAN1, CACNA1A, DAB1, FGF14, FMR1, FXN, NOP56, PPP2R2B, RFC1, TBP, ZFHX3*			
Bartter och Gitelman syndrom	Perifert blod	Gitelman, Bartter och Liddle syndrom panel v1, 13 gener *AP2S1, BSND, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, MAGED2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3*	NGS TruSeq helgenom In silico panel*	90	EDTA
Beckwith-Wiedemann syndrom	Perifert blod	11p15	MLPA metylering	56	EDTA
Bindvävssjukdomar (bl a Ehlers Danlos syndrom, Marfan syndrom, Loeys-Dietz syndrom och TAAD)	Perifert blod	Bindvävspanel v2, 44 gener *ACTA2, ADAMTS2, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469*	NGS TWIST In silico panel*	90	EDTA
Bröstcancer - snabbspår	Perifert blod	Bröstcancerpanel v1, 12 gener *ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53*	NGS TWIST In silico panel*	28	EDTA
CADASIL	Perifert blod	*NOTCH3* (Se även cerebrala småkärlssjukdomar)	NGS TruSeq helgenom In silico panel*	90	EDTA
Cerebrala småkärlssjukdomar (bl.a. Moyamoya, Hereditär hemorragisk telangiektasi, Capillary malformation-arteriovenous malformation (CM-AVM) syndrom, Parkes-Weber syndrom)	Perifert blod	Cerebrala småkärlssjukdomspanel v3, 50 gener *A2ML1, ABCC6, ACTA2, ACVRL1, ANGPTL6, APP, ATP1A2, BRAF, CACNA1A, CBL, CCM2, COL3A1, COL4A1, COL4A2, COL5A1, COLGALT1, CST3, ENG, EPHB4, FOXC1, GDF2, GLA, GUCY1A1, HRAS, HTRA1, KRAS, KRIT1, LZTR1, MAP2K1, MAP2K2, NF1, NOTCH3, NRAS, PDCD10, PTPN11, RAF1, RASA1, RASA2, RIT1, RNF213, RRAS,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		SAMHD1, SHOC2, SLC2A10, SMAD4, SOS1, SOS2, SPRED1, TREX1, YY1AP1			
Charcot-Marie-Tooth (CMT1A)	Perifert blod	*PMP22* (ingår även i Neuropatipanel)	MLPA enkel	56	EDTA
Cri du Chat syndrom	Perifert blod	5p15	MLPA enkel	56	EDTA
Cri du Chat syndrom	Perifert blod	5p	FISH Konstitutionellt metafas	14	Heparin
Corneal dystrofi	Perifert blod	Corneal dystrofi v1, 48 gener AGBL1, ADAMTS18, ALDH18A1, B3GLCT, CHRDL1, CHST6, COL5A1, COL17A1, COL8A2, DCN, FOXE3, GJA1, GJA8, GLA, GRHL2, GSN, HMX1, KERA, KRT12, KRT3, LCAT, LOXHD1, LTBP2, MAF, MCOLN1, MIR184, NLRP3, OVOL2, PAX6, PIK3R1, PIKFYVE, PITX2, PRDM5, PRDX3, RAB18, RAB3GAP1, RAB3GAP2, SLC16A12, SLC4A11, STS, TACSTD2, TCF4, TGFBI, TUBA3D, UBIAD1, VSX1, ZEB1, ZNF469	NGS TruSeq helgenom In silico panel*	90	EDTA
Cystisk Fibros	Perifert blod	*CFTR* (50 mutationer)	Fragmentanalys CFTR	35	EDTA
Cystisk Fibros	Perifert blod	*CFTR*	NGS TWIST In silico panel*	90	EDTA
DSD (Disorders of sex development)	Perifert blod	DSD Panel v1, 192 gener och 2 regioner *AARS2, AKR1C2, AKR1C3, AKR1C4, ALDOA, AMH, AMHR2, ANOS1, AR, ARHGAP35, ARX, ATF3, ATRX, BMP15, BMP4, BMP7, BMPR1B, BNC1, BUB1B, C14orf39, CBX2, CCDC141, CDKN1C, CHD7, CLPP, CPE, CTU2, CUL4B, CYB5A, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP21A2, DACH2, DCAF17, DHCR7, DHH, DHX37, DIAPH2, DMRT1, DMRT2, DUSP6, EIF2B4, EIF2B5, EIF4ENIF1, EMX2, ERAL1, ERCC6, ESR1, ESR2, FANCM, FEZF1, FGD1, FGF17, FGF8, FGFR1, FGFR2, FIGLA, FKBP4, FLRT3, FMR1, FOXL2, FRAS1, FREM2, FSHB, FSHR, GALT, GATA4, GDF9, GGPS1, GLI2, GNRH1, GNRHR, GRIP1, HAMP, HARS2, HFE, HFM1, HHAT, HNF1B, HOXA13, HOXA4, HOXB6, HS6ST1, HSD17B3, HSD17B4, HSD3B2, HSF2BP, IGSF10, IL17RD, INSL3, KASH5, KHDRBS1, KISS1, KISS1R, KLB, LARS2, LEP, LEPR, LHB, LHCGR, LHX1, LHX3, LHX4, LHX9, LMNA, MAMLD1, MAP3K1, MCM8, MCM9, MEIOB, MID1, MRPS22, MSH4, MSH5, MYRF, NANOS3,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		NDNF, NOBOX, NOG, NR0B1, NR2F2, NR3C1, NR5A1, NSMF, NUP107, PAX8, PBX1, PCDH17, PCSK1, PGRMC1, PLXNA3, PMM2, POF1B, POLG, POLR2C, POLR3H, POR, POU5F1, PPP1R12A, PRDM13, PROK2, PROKR2, PROP1, PSMC3IP, RCBTB1, RIPK4, RNF216, RPL10, RSPO1, SAMD9, SEMA3A, SEMA3F, SGO2, SGPL1, SLC29A3, SLC40A1, SOHLH1, SOHLH2, SOX10, SOX11, SOX2, SOX3, SOX8, SOX9, SPIDR, SPRY4, SRD5A2, SRY, STAG3, STAR, STS, SYCE1, SYCP2L, TAC3, TACR3, TCF12, TFR2, TOE1, TP63, TSPYL1, TWNK, VAMP7, WDR11, WNT4, WT1, WWOX, XRCC2, ZFPM2, ZSWIM7 samt 11p13 deletionssyndromet (WAGR) och Xp21.2 duplikationssyndromet			
Duchenne, Becker (DMD/BMD)	Perifert blod	*DMD*	MLPA dubbel	56	EDTA
Duchenne, Becker (DMD/BMD)	Perifert blod	*DMD* (se även Neuromuskulär panel)	Sangersekvensering riktad	28	EDTA
Dystoni	Perifert blod	Dystonipanel v2, 200 gener och 9 repeatexpansioner *ABAT, ACER3, ACOX1, ACTB, ADAR, ADCY5, AFG3L2, ALDH18A1, ANO3, AP1S2, APTX, ARFGEF3, ARSA, ARX, ASL, ATM, ATP13A2, ATP1A2, ATP1A3, ATP5MC3, ATP7B, BCAP31, BCS1L, C19orf12, CACNA1A, CACNA1G, CACNB4, CAMK4, CHMP2B, CLN3, CLN5, CLPB, COASY, COX10, COX15, COX20, CP, CSF1R, CSTB, CYP27A1, DCAF17, DCTN1, DDC, DHDDS, DLAT, DLD, DNAJC12, ECHS1, EIF2AK2, FA2H, FBXO7, FITM2, FOLR1, FOXG1, FOXRED1, FTL, FUCA1, FXN, GCDH, GCH1, GJC2, GLB1, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GRIN1, GTPBP2, HCFC1, HECW2, HEXA, HIBCH, HNRNPH1, HPCA, HPRT1, HSPD1, HTRA2, IFIH1, IMPDH2, IRF2BPL, KCNA1, KCNMA1, KCNQ2, KCTD17, KIF1C, KMT2B, L2HGDH, LRPPRC, LYST, MAPT, MARS2, MECR, MED27, MRE11, MTFMT, MYORG, NDUFA1, NDUFA10, NDUFA12, NDUFA2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NGLY1, NKX2-1, NKX6-2, NPC1, NPC2, NUP54, OPA3, PANK2, PCCA, PCCB, PDE10A, PDE2A, PDGFB, PDGFRB, PDHA1, PDHX, PET100, PINK1, PLA2G6, PNKD, PNKP, PNPT1, POLR3A, PPP2R5D, PRKN, PRKRA, PRNP, PRRT2, PTS, QDPR, RAB39B, RNASEH2B, RNASEH2C, RNASET2, RNF216, RNU7-1, SAMHD1, SCN1A, SCN8A, SERAC1, SETX, SGCE, SHQ1, SLC16A2, SLC18A2, SLC19A3, SLC20A2, SLC2A1, SLC30A10, SLC30A9, SLC39A14, SLC6A3, SLC6A8, SNORD118, SPATA5L1, SPG11, SPR, SQSTM1, SUCLA2, SUOX, SURF1, SYNJ1, SYT1, TAF1, TARS2, TBC1D24, TH, THAP1, TIMM8A, TMEM151A, TOR1A, TPK1, TREX1, TSPOAP1,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		TUBB4A, UBTF, VAC14, VAMP1, VAMP2, VPS13A, VPS13D, VPS16, VPS41, VPS4A, WDR45, WDR73, XPR1, YIF1B, YY1, ZSWIM6 Screening för patogena repeatexpansioner ingår för följande gener: *ATN1, ATXN1, ATXN2, ATXN3, CACNA1A, CSTB, FXN, PPP2R2B, TBP*			
Dystrofia myotonika typ 1	Perifert blod	*DMPK*	Fragmentanalys DMPK	35	EDTA
Ektodermal dysplasi	Perifert blod	Ektodermal dysplasipanel v1, 111 gener *ANTXR1, APCDD1, ARID1A, ARID1B, ATP7A, ATP6V1B2, AXIN2, BCS1L, BMP4, CDH3, CDSN, CLDN1, CSTB, CTNND1, CTSC, CTSK, DSG4, DSP, EDA, EDA2R, EDAR, EDARADD, EGFR, ERCC2, ERCC3, ERCC8, EVC, EVC2, FGF10, FGFR2, FGFR3, GJA1, GJB2, GJB6, GRHL2, GTF2E2, GTF2H5, HEPHL1, HOXC13, HR, IFT122, IFT140, IFT43, IFT52, IKBKG, INSR, IRF6, JUP, KANK2, KCTD1, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT25, KRT6A, KRT6B, KRT6C, KRT71, KRT74, KRT83, KRT85, LIPH, LPAR6, LRP6, LSS, LTBP3, MBTPS2, MPLKIP, MSX1, NECTIN1, NECTIN4, NFKB1, PAX9, PEX1, PEX6, PIGL, PKP1, POC1A, PORCN, PTH1R, RHOA, RIN2, RNF113A, ROGDI, RPL21, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SMOC2, SNRPE, SOX9, SOX18, SPINK5, SREBF1, TBC1D24, TFAP2B, TP63, TRAF6, TRPS1, TRPV3, TSPEAR, UBR1, WDR19, WDR35, WNT10A, WNT10B*	NGS TruSeq helgenom In silico panel*	90	EDTA
Epidermolysis bullosa (EB)	Perifert blod	Epidermolysis bullosa panel v2, 31 gener *ATP2C1, CAST, CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSG1, DSP, DST, EXPH5, FERMT1, FLG2, ITGA3, ITGA6, ITGB4, KLHL24, KRT1, KRT10, KRT14, KRT2, KRT5, KRT6C, LAMA3, LAMB3, LAMC2, PKP1, PLEC, SERPINB8, TGM5*	NGS TruSeq helgenom In silico panel*	90	EDTA
Epilepsi Analysen inkluderar även POLG mutationer associerade med Valproat-inducerad leverskada.	Perifert blod	Epilepsipanel v2, 637 gener, 13 regioner och 3 repeatexpansioner *AARS1, ABAT, ABCA2, ACOX1, ACTL6B, ADAM22, ADAR, ADARB1, ADGRG1, ADPRS, ADSL, AFF3, AFG3L2, AGO1, AIMP1, AKT3, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG13, ALG14, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMACR, AMPD2, AMT, ANKRD11, AP1G1, AP2M1, AP3B2, AP4B1, AP4S1, APC2, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGEF9, ARID1B, ARSA, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASXL3, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP5PO, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATRX, BAP1, BCKDHA, BCKDHB, BCS1L, BLTP1, BOLA3, BRAF, BRAT1, BSCL2, BTD, C12orf57, C2orf69, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D2, CAD, CAMK2B, CAPRIN1, CARS2, CASK,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		CC2D2A, CDK19, CDKL5, CELF2, CEP85L, CERS1, CHD2, CHD4, CHD5, CHKA, CHRNA2, CHRNA4, CHRNA7, CHRNB2, ClC, CLCN3, CLCN4, CLDN5, CLN3, CLN5, CLN6, CLN8, CLPB, CLTC, CNKSR2, CNNM2, CNOT9, CNPY3, CNTNAP2, COG7, COL18A1, COL4A1, COL4A2, COQ2, COQ4, COQ9, CPA6, CPLX1, CREBBP, CRELD1, CSNK2B, CSTB, CTNNA2, CTSD, CTSF, CUL3, CUL4B, CUX2, CYFIP2, CYP27A1, D2HGDH, DBT, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DEPDC5, DHDDS, DHFR, DHPS, DHX30, DIAPH1, DLL1, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK7, DOLK, DPAGT1, DPH5, DPM1, DPYD, DROSHA, DTYMK, DYNC1H1, DYRK1A, EARS2, ECHS1, EEF1A2, EFHC1, EFTUD2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EMC10, EML1, ENTPD1, EPG5, EPM2A, ESAM, ETHE1, EXOSC3, EXT2, FAR1, FARS2, FASTKD2, FBXL4, FBXO11, FBXO28, FGF12, FGF13, FGFR3, FKTN, FLNA, FOLR1, FOXG1, FOXRED1, FRMD5, FRRS1L, FUCA1, FUT8, GABBR2, GABRA1, GABRA2, GABRA3, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNT2, GAMT, GBA1, GCH1, GCSH, GFAP, GLB1, GLDC, GLRA2, GLUD1, GLUL, GM2A, GNAO1, GNAQ, GNB1, GNB5, GOSR2, GOT2, GPAA1, GPHN, GRIA2, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GTPBP2, GTPBP3, GUF1, H3-3A, H3-3B, HACE1, HAX1, HCFC1, HCN1, HCN2, HECTD4, HECW2, HEPACAM, HERC2, HEXA, HEXB, HID1, HMGCL, HNRNPH2, HNRNPR, HNRNPU, HPDL, HRAS, HSD17B10, HSD17B4, HTRA2, IER3IP1, IFIH1, IKBKG, IQSEC2, IRF2BPL, ITPA, KANSL1, KAT5, KAT8, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND2, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, KDM6B, KIF1A, KIF2A, KIF5A, KIF5C, KLHL20, KMT2E, KPTN, KRAS, LARS1, LETM1, LGI1, LIAS, LMBRD2, LMNB2, MACF1, MADD, MAF, MAP2K1, MAP2K2, MAST4, MBD5, MBOAT7, MDH2, MECP2, MED11, MED12, MED17, MED27, MEF2C, MFF, MFSD8, MINPP1, MLC1, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MTHFR, MTHFS, MTOR, NACC1, NAGA, NAGLU, NAPB, NARS1, NARS2, NBEA, NDE1, NDUFA1, NDUFA10, NDUFAF2, NDUFAF5, NDUFS4, NDUFS8, NDUFV1, NECAP1, NEDD4L, NEUROD2, NEXMIF, NGLY1, NHLRC1, NPC1, NPC2, NPRL2, NPRL3, NR4A2, NRROS, NRXN1, NSD1, NSDHL, NSRP1, NTRK2, NUP214, NUS1, OCLN, OGDHL, OPHN1, OTUD6B, OTUD7A, OXR1, P4HTM, PABPC1, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PARS2, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCYT2, PDHA1, PDHX, PET100, PGM2L1, PHACTR1, PHGDH, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3R2, PIP5K1C, PLA2G6, PLAA, PLCB1, PLK1, PLP1, PLPBP, PLXNA1, PMM2, PMPCB, PNKP, PNPO, PNPT1, POLG, POMGNT1, POMT1, PPFIBP1, PPIL1, PPP1R3F, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRDM8, PRICKLE1, PRICKLE2, PRIMA1, PRMT7, PRODH, PRPF8, PRRT2, PSAP, PTCD3, PTEN, PTPN23, PTS, PUM1, PURA, QARS1, QDPR, RAB11A,			

		RAB11B, RAB18, RAB39B, RAB5C, RAC3, RALA, RALGAPA1, RARS2, RELN, RFT1, RHEB, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, ROGDI, RORA, RORB, RTN4IP1, RTTN, RUSC2, SAMHD1, SARS1, SATB1, SATB2, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SEPSECS, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGSH, SHQ1, SIK1, SLC12A5, SLC13A5, SLC16A2, SLC19A3, SLC1A2, SLC1A4, SLC25A1, SLC25A12, SLC25A22, SLC2A1, SLC32A1, SLC35A2, SLC35A3, SLC38A3, SLC39A8, SLC45A1, SLC6A1, SLC6A8, SLC6A9, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMS, SNAP25, SNIP1, SNORD118, SPATA5, SPATA5L1, SPTAN1, SPTBN1, ST3GAL3, ST3GAL5, STAG1, STAMBP, STRADA, STX1B, STXBP1, SUCLA2, SUMF1, SUOX, SURF1, SYN1, SYNGAP1, SYNJ1, SZT2, TAF8, TANGO2, TBC1D24, TBC1D2B, TBCD, TBCK, TBL1XR1, TCF4, TDP2, TFE3, TIAM1, TIMM50, TMEM222, TMEM63B, TMX2, TNPO2, TPK1, TPP1, TRA2B, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TREX1, TRIM8, TRIT1, TRPM3, TRPM6, TSC1, TSC2, TSEN54, TUBA1A, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, U2AF2, UBA5, UBAP2L, UBE2A, UBE3A, UBR7, UFM1, UFSP2, UGDH, UGP2, UNC80, USP18, VAMP2, VARS1, VPS11, WARS2, WASF1, WDR26, WDR37, WDR45, WDR45B, WDR73, WNK3, WWOX, YIPF5, YWHAG, ZBTB18, ZBTB47, ZDHHC9, ZEB2, ZNF142, ZNF335 Regioner: 1p36, 1q43q44, 4p16.3, 8p23.1, 15q11,13, 15q13.3, 16p12.2, 16p13.11, 17p13.3, 17q12, 22q11.2, Xp11.22p11.23, Xq28 Screening för patogena repeatexpansioner ingår för följande gener: *ARX, ATN1, CSTB*			
Erytrocytos	Perifert blod	Erytrocytospanel v1, 7 gener *BPGM, EGLN1, EPAS, EPO, EPOR, JAK2, VHL*	NGS TruSeq helgenom In silico panel*	90	EDTA
Familjär hyperkolesterolemi (FH)	Perifert blod	Familjär hyperkolesterolemi v2, 7 gener *ABCG5, ABCG8, APOB, APOE, LDLR, LDLRAP1, PCSK9*	NGS TWIST In silico panel*	90	EDTA
FGFR3-relaterad skelettdysplasi	Perifert blod	*FGFR3*	NGS TruSeq helgenom In silico panel*	90	EDTA
Fragilt-X; FRAXA	Perifert blod	*FMR1*	Fragmentanalys Fragilt-X	35	EDTA
FXTAS	Perifert blod	*FMR1*	Fragmentanalys Fragilt-X	35	EDTA

Genotypning	Perifert blod		QF-PCR	35	EDTA
Helexomsekvensering	Perifiert blod	Screening	NGS TWIST Exom Trio NGS TWIST Exom Duo NGS TWIST Exom Singleton	90	EDTA
Helgenomsekvensering Inkl. mitokondriella genomet samt relevanta repeatexpansionssjukdomar	Perifiert blod	Screening	NGS TruSeq Helgenom Trio NGS TruSeq Helgenom Duo NGS TruSeq Helgenom Singleton	90	EDTA
Hereditär hemorragisk telangiektasi (HHT; Osler-Weber-Rendus syndrom) (bl.a. HHT, Capillary malformation-arteriovenous malformation (CM-AVM) syndrom, Parkes-Weber syndrom)	Perifert blod	Hereditär hemorragisk telangiektasipanel v1, 6 gener *ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4*	NGS TruSeq helgenom In silico panel*	90	EDTA
Hereditär spastisk paraplegi (HSP)	Perifert blod	Hereditär spastisk paraplegi panel v2, 115 gener, 1 region och 9 repeatexpansioner *ABCD1, ABHD16A, ACER3, ADAR, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMFR, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BCAS3, BSCL2, C12orf65, C19orf12, CAPN1, CLDN11, COQ4, CPT1C, CTNNB1, CYP27A1, CYP2U1, CYP7B1, DARS, DDHD1, DDHD2, DDX3X, ELOVL1, ENTPD1, ERLIN1, ERLIN2, FA2H, FAR1, FARS2, FBXO7, FXN, GALC, GBA2, GBE1, GCH1, GJA1, GLRX5, GPT2, HACE1, HECTD4, HIKESHI, HPDL, HSPD1, IFIH1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KPNA3, L1CAM, LETM1, MAG, MAPK8IP3, NDUFA12, NIPA1, NKX6-2, NSRP1, NT5C2, OPA3, PCYT2, PLP1, PNPLA6, POLR3A, PPFIBP1, PRNP, PSEN1, RAB3GAP2, REEP1, REEP2, RETREG1, RNASEH2B, RNF170, RNU7-1, RTN2, SACS, SERAC1, SLC16A2, SLC1A4, SLC25A15,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		SLC25A46, SLC2A1, SPART, SPAST, SPATA5L1, SPG11, SPG21, SPG7, SPTAN1, STN1, TAF8, TECPR2, TFG, TMEM63C, TUBB4A, UBAP1, UCHL1, WASHC5, WDR45B, ZFYVE26 Region: Xq28 Screening för patogena repeatexpansioner ingår för följande gener: *ATXN1, ATXN2, ATXN3, ATXN7, ATXN10, CACNA1A, FXN, PPP2R2B, TBP*			
HNPP, fam. tryckförlamning	Perifert blod	*PMP22*	MLPA enkel	56	EDTA
Huntingtons sjukdom	Perifert blod	*HTT* (Huntingtin)	Fragmentanalys HTT (HD)	35	EDTA
Hyperlipidemi	Perifert blod	*Hyperlipidemipanel v1, 18 gener* *ABCA1, ABCG5, ABCG8, APOA1, APOA5, APOB, APOC2, APOE, CREB3L3, CYP27A1, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LMF1, LPL, PCSK9*	NGS TruSeq helgenom In silico panel*	90	EDTA
Hyperparatyreoidism (inkl. Familjär hypokalciurisk hyperkalcemi (FHH),Albright hereditary osteodystrophy, pseudohypoparathyroidism, pseudopseudohypoparathyroidism, acrodysostosis and osteoma cutis)	Perifert blod	Hyperparatyreoidismpanel v2, 8 gener *AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET*	NGS TruSeq helgenom In silico panel*	90	EDTA
Hypofystumör, ärftlig	Perifert blod	Ärftlig hypofystumör v1, 6 gener *AIP, CDKN1B, DICER1, MEN1, NF1, PRKAR1A*	NGS TruSeq helgenom In silico panel*	90	EDTA
Hypogonadotrop hypogonadism (inkl. Kallman syndrom)	Perifert blod	Hypogonadotrop hypogonadismpanel v1, 31 gener *ANOS1, CHD7, CUL4B, DCAF17, FEZF1, FGF8, FGFR1, FSHB, GLI2, GNRH1, GNRHR, HAMP, HFE, IL17RD, KISS1R, KLB, LHB, LHX4, NR0B1, NSMF, PROK2, PROKR2, PROP1, SLC29A3, SLC40A1, SOX10, SOX2, TAC3, TACR3, TFR2, WDR11*	NGS TruSeq helgenom In silico panel*	90	EDTA
Hypoparatyreoidism (inkl. Albright hereditary osteodystrophy, pseudohypoparathyroidism,	Perifert blod	Hypoparatyreoidismpanel v1, 11 gener *AIRE, CASR, GATA3, GCM2, GNA11, GNAS, PDE4D, PRKAR1A, PTH, STX16, TBCE*	NGS TruSeq helgenom In silico panel*	90	EDTA

pseudopseudohypoparathyroidism, acrodysostosis and osteoma cutis)					
Hypokondroplasi (Se även *FGFR3*-relaterad skelettdysplasi)	Perifert blod	*FGFR3* (exon 9, 12)	Sangersekvensering *FGFR3*	56	EDTA
Hörselnedsättning	Perifert blod	Hörselnedsättning panel v1, 289 gener *BHD12, ABHD5, ACOX1, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, AMMECR1, ANKH, ARSG, ATP11A, ATP1A3, ATP2B2, ATP6V0A4, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTD, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CD164, CDC14A, CDC42, CDH23, CDK9, CEACAM16, CEP250, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, EIF3F, ELMOD3, EPS8, EPS8L2, ERAL1, ESPN, ESRRB, EYA1, EYA4, FAM136A, FDXR, FGF3, FGFR2, FGFR3, FITM2, FOXC1, FOXI1, GATA3, GDF6, GIPC3, GJA1, GJB2, GJB3, GJB6, GPSM2, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, HARS1, HARS2, HGF, HOMER2, HOXA2, HOXB1, HSD17B4, ILDR1, JAG1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, KMT2D, LARS2, LHFPL5, LHX3, LMX1A, LOXHD1, LOXL3, LRP2, LRRC51/LRTOMT, MAN2B1, MANBA, MARVELD2, MASP1, MCM2, MEOX1, MET, MGP, MITF, MPZL2, MSRB3, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH14, MYH9, MYO15A, MYO18B, MYO3A, MYO6, MYO7A, NAGLU, NARS2, NDP, NDRG1, NEFL, NF2, NLRP3, NOG, NR2F1, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX1, PAX3, PCDH15, PCGF2, PDE1C, PDZD7, PEX1, PEX26, PEX6, PHYH, PISD, PJVK, PNPT1, POLR1B, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RAI1, RDX, REST, RIPOR2, RMND1, ROR1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERAC1, SERPINB6, SH3TC2, SIX1, SIX2, SIX5, SLC12A2, SLC17A8, SLC19A2, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SPATA5, SPNS2, STAG2, STRC, SUCLA2, SUCLG1, SYNE4, SYT2, TBC1D24, TBL1X, TBX1, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMEM126A, TMEM132E, TMEM43, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TRRAP,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		TSHZ1, TSPEAR, TUBB4B, TWNK, TYR, UBR1, USH1C, USH1G, USH2A, WBP2, WFS1, WHRN, XYLT2, ZNF469			
Hörselnedsättning	Perifert blod	*GJB2* (exon 2), GJB6 (inkl. deletion/duplikationsanalys)	Paket: Sangersekvensering GJB2/ MLPA enkel	56	EDTA
Iktyos, könsbunden	Perifert blod	*STS*	MLPA enkel	56	EDTA
Iktyos, NGS panel (bl.a. Acral Peeling Skin Syndrome (APSS), Erytrokeratodermia variablis, (EKV), Harlequin syndrom, könsbunden iktyos, KID syndrom, Sjögren-Larssons syndrom)	Perifert blod	Iktyospanel v2, 70 gener *AARS1, ABCA12, ABHD5, ADAMTS17, ALDH3A2, ALOX12B, ALOXE3, AP1B1, AP1S1, ASPRV1, CASP14, CAST, CDSN, CERS3, CHST8, CLDN1, CLDN10, CSTA, CYP4F22, DOLK, DSG1, DSP, EBP, ELOVL1, ELOVL4, ERCC2, ERCC3, FLG, FLG2, GJA1, GJB2, GJB3, GJB4, GJB6, GTF2E2, GTF2H5, KDSR, KRT1, KRT10, KRT2, LIPN, LORICRIN, MARS1, MBTPS2, MPDU1, MPLKIP, NIPAL4, NSDHL, PEX7, PHGDH, PHYH, PIGL, PNPLA1, POMP, PSAT1, RNF113A, SDR9C7, SERPINB8, SLC27A4, SNAP29, SPINK5, SRD5A3, SREBF1, ST14, STS, SULT2B1, SUMF1, TARS1, TGM1, TGM5*	NGS TruSeq helgenom In silico panel*	90	EDTA
Immunologisk sjukdom (bl.a immunbrist, inflammatorisk tarmsjukdom, autoinflammatorisk sjukdom, kronisk granulomatös sjukdom, periodisk feber, SCID, kongential neutronpeni, och autoimmunitetssyndrom)	Perifert blod	Immunpanel v1, 355 gener *ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, AP3B1, AP3D1, ARPC1B, ARPC5, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLNK, BTK, C1QA, C1QC, C1S, C2, C2orf69, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CIITA, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTLA4, CTNNBL1, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1B, DCLRE1C, DEF6, DGAT1, DNAAF11, DNAH1, DNAJB13, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, ELANE, ELF4, EPG5, ERCC6L2, EXTL3, FADD, FAS, FASLG, FCHO1, FERMT3, FNIP1, FOXJ1, FOXN1, FOXP3, G6PC3, G6PD, GAS8, GATA2, GFI1, GIMAP5, GINS1, GUCY2C, HAX1, HELLS, HMOX1, HYDIN, HYOU1, ICOS, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7, IL7R, INO80, IRAK4, IRF2BP2, IRF4, IRF7, IRF8, IRF9, ISG15, ITGB2, ITK, ITPKB, JAGN1, JAK1, JAK3, LACC1, LAMTOR2, LAT, LCK, LCP2, LCT, LIG1, LIG4, LIPA, LPIN2, LRBA, LYN, LYST, MAGT1, MALT1, MAP3K14, MCM10, MCM4, MCTS1, MEFV, MOGS, MPEG1, MRTFA, MSN,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		MTHFD1, MVK, MYD88, MYO5B, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NEUROG3, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NSMCE3, OAS1, ODAD1, ODAD2, ORAI1, OTULIN, PARN, PAX1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PNP, POLA1, POMP, PRIM1, PRKCD, PRKDC, PSMB10, PSMB8, PSMB9, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RC3H1, REL, RELA, RFX5, RFXANK, RFXAP, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RPGR, RPSA, RSPH3, RTEL1, SAMHD1, SASH3, SEC61A1, SERPING1, SH2D1A, SKIC2, SKIC3, SLC26A3, SLC29A3, SLC35C1, SLC37A4, SLC39A4, SLC39A7, SLC46A1, SLC51B, SLC7A7, SLC9A3, SLCO2A1, SMARCAL1, SMARCD2, SNORA31, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP54, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK4, STX11, STXBP2, STXBP3, SYK, TAFAZZIN, TAP1, TAP2, TBX1, TCF3, TCN2, TET2, TFRC, TGFB1, TICAM1, TLR2, TLR3, TLR7, TLR8, TMC6, TMC8, TMPRSS15, TNFAIP3, TNFRSF1A, TNFRSF9, TOP2B, TPP2, TRAC, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC12, TTC7A, TYK2, UBA1, UBE2T, UNC13D, UNC93B1, UNG, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WNT2B, XIAP, ZAP70, ZBTB24, ZMYND10, ZNF341, ZNFX1			
Infertilitetsutredning, POI	Perifert blod	*FMR1*	Fragmentanalys Fragilt-X	35	EDTA
Infertilitetsutredning, CBAVD	Perifert blod	*CFTR* (50 mutationer)	Fragmentanalys CFTR	35	EDTA
Infertilitetsutredning, Mikrodeletion Y	Perifert blod	AZF	Fragmentanalys Mikrodeletion Y	35	EDTA
Intellektuell funktionsnedsättning	Perifert blod	Intellektuell funktionsnedsättning panel v1, 1453 gener, 48 regioner och 7 repeatexpansioner *AAAS, AARS1, AASS, ABAT, ABCA2, ABCC9, ABCD1, ABCD4, ABHD16A, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACER3, ACO2, ACOX1, ACSL4, ACTB, ACTG1, ACTL6A, ACTL6B, ACY1, ADAM22, ADAR, ADARB1, ADAT3, ADD1, ADD3, ADGRG1, ADK, ADNP, ADSL, AFF2, AFF3, AFF4, AGA, AGO1, AGO2, AGTPBP1, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AKT3, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG12, ALG13, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALMS1, ALX4, AMER1, AMPD2, AMT, ANK2, ANK3, ANKRD11, ANKRD17, AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, COA8, ARCN1, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGEF9, ARID1A, ARID1B, ARID2,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		ARID5A, ARL13B, ARL6, ARMC9, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD1, ATAD3A, ATG7, ATIC, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP6V1B2, ATP7A, ATP8A2, ATP9A, ATR, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, AUTS2, B3GALNT2, B3GLCT, B4GALNT1, B4GALT7, B9D2, BAP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCL11A, BCL11B, BCOR, BCS1L, BICRA, BLM, BLOC1S1, BLTP1, BMP4, BMPR1A, BNC1, BOLA3, BPTF, BRAF, BRAT1, BRD4, BRF1, BRPF1, BRSK2, BRWD3, BSCL2, BTD, BUB1, BUB1B, C12orf4, C12orf57, MTRFR, C2CD3, C2orf69, CPLANE1, CA2, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1I, CACNA2D1, CAD, CAMK2A, CAMK2B, CAMK4, CAMTA1, CAPN15, CAPRIN1, CARS1, CASK, CASP2, CBL, CBS, CC2D1A, CC2D2A, CCBE1, CCDC22, CCDC32, CCDC47, CCDC82, CCDC88C, CCND2, CDC42, CDC6, CDH11, CDH2, CDK10, CDK13, CDK16, CDK19, CDK5RAP2, CDK8, CDKL5, CDON, CECR2, CELF2, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP290, CEP41, CEP55, CEP57, CEP83, CEP85L, CERT1, CHAMP1, CHD2, CHD3, CHD4, CHD5, CHD7, CHD8, CHKA, CHKB, CHMP1A, CHRNA7, CIC, CIT, CKAP2L, CLCN3, CLCN4, CLCN6, CLDN11, CLDN5, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLTC, CNBP, CNKSR2, CNNM2, CNOT1, CNOT2, CNOT3, CNOT9, CNTNAP1, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL4A1, COL4A2, COLEC11, COPB2, COQ4, COQ8A, COX10, COX11, COX15, CPE, CPLX1, CPS1, CRADD, CRB2, CREBBP, CRPPA, CSDE1, CSNK1G1, CSNK2A1, CSNK2B, CSPP1, CSTB, CTBP1, CTCF, CTDP1, CTNNA2, CTNNB1, CTNND1, CTNND2, CTR9, CTSA, CTSD, CTU2, CUL3, CUL4B, CUX1, CUX2, CWC27, CWF19L1, STEEP1, CYB5R3, CYC1, CYFIP2, D2HGDH, DAG1, DAGLA, DARS1, DARS2, DBT, DCAF17, DCHS1, DCPS, DCX, DDB1, DDC, DDHD2, DDX11, DDX23, DDX3X, DDX59, DDX6, DEAF1, DEGS1, DEPDC5, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHTKD1, DHX30, DHX37, DHX9, DIAPH1, DIS3L2, DKC1, DLD, DLG1, DLG3, DLG4, DLL1, DMD, DMPK, DMXL2, DNAJC12, DNAJC19, DNM1, DNM1L, DNMT3A, DNMT3B, DOCK3, DOCK6, DOCK7, DOHH, DOLK, DPAGT1, DPF2, DPH1, DPH5, DPM1, DPM2, DPP6, DPYD, DPYS, DPYSL5, DTYMK, DYM, DYNC1H1, DYRK1A, EARS2, EBF3, EBP, EDEM3, EED, EEF1A2, EFTUD2, EHMT1, EIF2AK2, EIF2AK3, EIF2S3, EIF3F, EIF4A2, EIF4A3, EIF5A, ELAC2, ELN, ELOVL4, ELP2, EMC1, EMC10, EML1, EMX2, ENTPD1, EP300, EPG5, ERBB4, ERCC1, ERCC2, ERCC3, ERCC5, ERCC6, ERCC6L2, ERCC8, ERI1, ERLIN2, ESAM, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXT2, EXTL3, EZH2, FAM111A, HYCC1, FAM20C, FAM50A, FAR1, FARS2, FARSA, FAT4, FBRSL1, FBXL3, FBXL4, FBXO11, FBXO28, FBXO31, FBXW11, FBXW7, FGD1, FGF12, FH, FIG4, FILIP1, FKRP, FKTN, FLNA, FLVCR2, FMN2, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FOXRED1, FRMD5, FRMPD4, FTCD,		

		FTSJ1, FUCA1, FUT8, FXN, GABBR2, GABRA1, GABRA2, GABRA5, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAK, GALC, GALE, GALNT2, GALT, GAMT, GATA4, GATAD2B, GATM, GCDH, GCH1, GCSH, GDI1, GEMIN5, GFAP, GFER, GFM1, GJA5, GJC2, GK, GLB1, GLDC, GLI2, GLI3, GLIS3, GLRA2, GLS, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAI1, GNAO1, GNAS, GNB1, GNB2, GNB5, GNPAT, GNPTAB, GNPTG, GNS, GPAA1, GPC3, GPC4, GPT2, GRIA1, GRIA2, GRIA3, GRIA4, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM1, GRM7, GTF2E2, GTF2H5, GTPBP2, GTPBP3, GUSB, H1-4, H3-3A, H3-3B, H4C3, H4C5, HACE1, HADHA, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECTD4, HECW2, HEPACAM, HERC1, HERC2, HESX1, HEXA, HEXB, HGSNAT, HIBCH, HID1, HIVEP2, HK1, HLCS, HMGB1, HMGCL, HNF1B, HNMT, HNRNPH1, HNRNPH2, HNRNPK, HNRNPR, HNRNPU, HOXA1, HPD, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA2, HUWE1, IARS1, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT172, IGF1, IGF1R, IKBKG, IL1RAPL1, IMPDH2, INPP5E, INPP5K, INTS1, INTS11, IQSEC2, IRF2BPL, IRX5, ITPA, ITPR1, IVD, JAM3, JARID2, JPH3, KANSL1, KARS1, KAT5, KAT6A, KAT6B, KAT8, KCNA2, KCNB1, KCNC1, KCND2, KCNH1, KCNH5, KCNJ10, KCNJ11, KCNJ6, KCNK3, KCNK9, KCNMA1, KCNN2, KCNN3, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM1A, KDM2B, KDM3B, KDM4B, KDM5A, KDM5B, KDM5C, KDM6A, KDM6B, KIAA0586, KIDINS220, KIF11, KIF14, KIF1A, KIFBP, KIF21B, KIF2A, KIF4A, KIF5A, KIF5C, KIF7, KLF7, KLHL20, KLHL7, KMT2A, KMT2B, KMT2C, KMT2D, KMT2E, KMT5B, KNL1, KPTN, KRAS, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMC3, LAMP2, LARGE1, LARP7, LARS1, LETM1, LHX2, LIAS, LIG4, LINGO4, LINS1, LIPT1, LMBRD2, LMNB1, LONP1, LRP2, LRPPRC, LSS, LYRM7, LZTR1, MAB21L1, MAB21L2, MACF1, MADD, MAF, MAGEL2, MAN1B1, MAN2B1, MAN2C1, MANBA, MAOA, MAOB, MAP1B, MAP2K1, MAP2K2, MAPK1, MAPK8IP3, MAPRE2, MASP1, MAST1, MAST4, MAT1A, MBD5, MBOAT7, MBTPS2, MCCC1, MCCC2, MCM3AP, MCOLN1, MCPH1, MDH2, MECP2, MED11, MED12, MED13, MED13L, MED17, MED23, MED25, MED27, MEF2C, MEIS2, METTL23, METTL5, MFF, MFSD2A, MFSD8, MGAT2, MICU1, MID1, MINPP1, MKKS, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MN1, MOCS1, MOCS2, MOGS, MORC2, MPDU1, MPLKIP, PALS1, MRPS22, MRPS34, MSL3, MSMO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MTSS2, MVK, MYCN, MYH10, MYH11, MYO5A, MYT1L, NAA10, NAA15, NACC1, NAGA, NAGLU, NALCN, NANS, NAPB, NARS1, NBEA, NCDN, NCKAP1, NDE1, NDP, NDST1, NDUFA1, NDUFA2, NDUFS1, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEDD4L, NEMF, NEU1, NEUROD2, NEUROG1, NEXMIF, NF1, NFASC, NFIA, NFIX, NFU1, NGLY1, NHS, NIPBL, NKAP, NKX2-1, NLGN3, NONO, NOTCH2NLC, NOVA2, NPC1, NPC2, NPHP1, NR2F1, NR2F2, NR4A2, NRAS, NRCAM, NRROS, NRXN1, NSD1, NSD2, NSDHL, NSRP1, NSUN2, NT5C2, NTNG2, NTRK1, NTRK2, NUBPL, NUDT2,			

		NUP214, NUS1, OCLN, OCRL, ODC1, OFD1, OGDHL, OGT, OPA3, OPHN1, OSGEP, OTC, OTOA, OTUD5, OTUD6B, OTUD7A, OTX2, OXR1, P4HTM, PABPC1, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PAK3, PAN2, PARN, PAX6, PAX8, PBX1, PC, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCGF2, PCNT, PCYT2, PDE4D, PDGFRB, PDHA1, PDHB, PDHX, PDSS1, PDSS2, PDZD8, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGAP2, PGAP3, PGK1, PGM2L1, PGM3, PHACTR1, PHF21A, PHF6, PHF8, PHGDH, PHIP, PI4KA, PIBF1, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3CA, PIK3R2, PIP5K1C, PITRM1, PLA2G6, PLAA, PLCB1, PLK1, PLK4, PLP1, PLPBP, PLXNA1, PMM2, PMPCB, PNKP, PNPLA6, PNPT1, POGZ, POLA1, POLG, POLR1C, POLR2A, POLR3A, POLR3B, POLRMT, POMGNT1, POMGNT2, POMT1, POMT2, PORCN, POU3F2, POU3F3, PPFIBP1, PPIL1, PPM1D, PPP1CB, PPP1R12A, PPP1R15B, PPP1R21, PPP1R3F, PPP2CA, PPP2R1A, PPP2R2B, PPP2R5D, PPP3CA, PPT1, PQBP1, PRDM13, PREPL, PRICKLE2, PRKAR1B, PRMT7, PRPF8, PRPS1, PRR12, PRSS12, PRUNE1, PSAP, PSMC3, PSMD12, PSPH, PTCH1, PTCHD1, PTDSS1, PTEN, PTF1A, PTPN11, PTPN23, PTPN4, PTRHD1, PTS, PUF60, PUM1, PURA, PUS1, PUS3, PUS7, PYCR1, PYCR2, QARS1, QDPR, QRICH1, RAB11A, RAB11B, RAB18, RAB23, RAB39B, RAB3GAP1, RAB3GAP2, RAB5C, RAC1, RAC3, RAD21, RAF1, RAI1, RALA, RALGAPA1, RAP1B, RARB, RARS1, RARS2, RBBP8, RBL2, RBM10, RBSN, RELN, RERE, RFT1, RFX3, RFX4, RFX7, RHOBTB2, RIT1, RLIM, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF125, RNF13, RNU4-2, RNU7-1, ROBO1, ROGDI, ROR2, RORA, RPGRIP1L, RPIA, RPL10, RPS17, RPS6KA3, RRM2B, RSRC1, RTEL1, RTN4IP1, RTTN, RXYLT1, SAMD9, SAMHD1, SARS1, SARS2, SATB1, SATB2, SBF1, SC5D, SCAF4, SCAMP5, SCAPER, SCN1A, SCN2A, SCN3A, SCN8A, SCO2, SCYL1, SDCCAG8, SDHA, SDHAF1, SEMA6B, SEPSECS, SERAC1, SET, SETBP1, SETD1A, SETD1B, SETD2, SETD5, SFXN4, SGPL1, SGSH, SH2B1, SHANK1, SHANK2, SHANK3, SHH, SHMT2, SHOC2, SHQ1, SHROOM4, SIAH1, SIK1, SIL1, SIN3A, SIN3B, SIX3, SKI, SKIC3, SLC12A2, SLC12A5, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A1, SLC1A2, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A22, SLC2A1, SLC30A9, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC38A3, SLC39A14, SLC39A8, SLC3A1, SLC46A1, SLC4A4, SLC5A6, SLC6A1, SLC6A17, SLC6A19, SLC6A3, SLC6A8, SLC6A9, SLC9A6, SLX4, SMAD4, SMARCA2, SMARCA4, SMARCA5, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SMC1A, SMC3, SMG8, SMOC1, SMPD1, SMPD4, SMS, SNAP25, SNAP29, SNIP1, SNORD118, SNRPB, SNRPN, SNX14, SNX27, SON, SOS1, SOS2, SOX10, SOX11, SOX2, SOX3, SOX4, SOX5, SOX6, SOX9, SPART, SPATA5, SPATA5L1, SPECC1L, SPEN, SPG11, SPOP, SPR, SPRED1, SPRED2, SPTAN1, SPTBN1, SPTBN2, SPTBN4, SRCAP, SRD5A3, SRRM2, SRSF1, SSR4, ST3GAL3, ST3GAL5, STAG1, STAG2,			

		STAMBP, STIL, STRA6, STRADA, STT3A, STX1B, STXBP1, SUCLG1, SUFU, SUMF1, SUOX, SUPT16H, SURF1, SUZ12, SVBP, SYN1, SYNCRIP, SYNGAP1, SYNJ1, SYP, SYT1, SZT2, TAF1, TAF2, TAF4, TAF6, TAF8, TAFAZZIN, TANC2, TANGO2, TAOK1, TASP1, TAT, WWTR1, TBC1D20, TBC1D23, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBP, TBR1, TBX1, TBX2, TBX4, TBX6, TCEAL1, TCF20, TCF4, TCF7L2, TCN2, TCTN2, TCTN3, TDP2, TECPR2, TEFM, TELO2, TENM3, TET3, TFE3, TGIF1, TH, THOC2, THOC6, THRA, THUMPD1, TIAM1, TIMM50, TLK2, TMCO1, TMEM106B, TMEM127, TMEM147, TMEM165, TMEM216, TMEM222, TMEM237, TMEM240, TMEM63B, TMEM63C, TMEM67, TMEM70, TMEM94, TMTC3, TMX2, TNPO2, TNRC6B, TOE1, TOR1A, TP73, TPP1, TPP2, TRA2B, TRAF7, TRAIP, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM8, TRIO, TRIP12, TRIT1, TRMT1, TRMT10A, TRNT1, TRPM3, TRRAP, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TSHB, TSPAN7, TSPOAP1, TTC19, TTC5, TTC8, TTI1, TTI2, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP6, TUSC3, TWIST1, U2AF2, UBA5, UBAP2L, UBE2A, UBE2QL1, UBE3A, UBE3B, UBE4A, UBR1, UBR7, UBTF, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC80, UPF3B, UROC1, USP7, USP9X, VAMP2, VARS1, VARS2, VCP, VLDLR, VPS11, VPS13B, VPS41, VPS4A, VPS53, VRK1, WAC, WARS2, WASF1, WDFY3, WDPCP, WDR26, WDR37, WDR4, WDR45, WDR45B, WDR62, WDR73, WDR81, WIPI2, WNK3, WNT1, WT1, WWOX, XRCC4, XYLT1, YARS1, YIF1B, YIPF5, YWHAE, YWHAG, YY1, ZBTB18, ZBTB20, ZBTB24, ZBTB47, ZBTB7A, ZC4H2, ZDHHC9, ZEB2, ZFHX4, ZFX, ZFYVE26, ZIC1, ZIC2, ZMIZ1, ZMYM2, ZMYM3, ZMYND11, ZMYND8, ZNF142, ZNF292, ZNF335, ZNF462, ZNF526, ZNF699, ZNF711, ZSWIM6 Screening för patogena repeatexpansioner ingår för följande gener: *AFF2, CNBP, DMPK, EIF4A3, FMR1, XYLT1, ZIC2.*			
Intestinal pseudo-obstruktion (inkl Hirschprung)	Perifert blod	Intestinal pseudo-obstruktion panel v1, 36 gener och 1 region *ACTA2, ACTG2, BDNF, CELSR3, ECE1, EDN3, EDNRB, ERBB3, FLNA, GDNF, GFRA1, KIFBP, L1CAM, LIG3, LMOD1, MITF, MPV17, MYH11, MYL9, MYLK, NRG1, NRTN, PAX3, PHOX2B, POLG, PROK1, PROKR1, PROKR2, RAD21, RET, RMRP, SGO1, SOX10, TTC7A, TYMP, ZEB2* Region: 9p21.3	NGS TruSeq helgenom In silico panel*	90	EDTA
Kraniosynostos panel (bl.a. Apert syndrom, Crouzon syndrom, Pfeiffer syndrom, Saethre-Chotzen syndrom)	Perifert blod	Kraniosynostospanel v3, 65 gener *ALPL, ASXL1, B3GAT3, CD96, CDC45, CDT1, COLEC11, CYP26B1, EFNA4, EFNB1, ERF, ESCO2, FBN1, FGF9, FGFR1, FGFR2, FGFR3, FREM1, GLI3, GPC3, IFT122, IFT140, IFT43, IGF1R, IL11RA, KAT6A, KAT6B, MASP1, MEGF8, MSX2, NFIA, ORC1, ORC4, ORC6, P4HB, PHEX, POR, PPP3CA, RAB23, RECQL4, RSPRY1, RUNX2, SCARF2,*	NGS TWIST In silico panel*	90	EDTA

		SEC24D, SIX2, SKI, SLC25A24, SMAD2, SMAD3, SMAD6, SOX6, SPECC1L, STAT3, TCF12, TCOF1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMCO1, TWIST1, WDR19, WDR35, ZEB2, ZIC1			
Kortvuxenhetspanel	Perifert blod	Kortvuxenhetspanel v1, 15 gener *ACAN, GH1, GHR, GHRHR, GHSR, IGF1, IGF1R, IGFALS, LHX3, LHX4, NPR2, POU1F1, PROP1, SHOX, SOX3*	NGS TruSeq helgenom In silico panel*	90	EDTA
Liddle syndrom	Perifert blod	Gitelman, Bartter och Liddle syndrompanel v1, 13 gener *AP2S1, BSND, CASR, CLCNKA, CLCNKB, GNA1, KCNJ1, MAGED2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3*	NGS TruSeq helgenom In silico panel*	90	EDTA
Makrocefali och överväxt (bl.a. Sotos syndrom)	Perifert blod	Makrocefali och överväxtpanel v2, 55 gener *AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EED, EIF2B5, EZH2, GFAP, GJA4 , GLI3, GPC3, GPSM2, GRIA3, HEPACAM, HUWE1, KDM1A, KIF7, KPTN, L1CAM, MAX, MED12, MLC1, MPDZ, NFIB, NFIX, NSD1, OFD1, PIGA, PIK3CA, PIK3R1, PIK3R2, PTCH1, PTEN, RAB39B, RASA1, RNF135, SETD2, SYN1, TMEM94, UPF3B, ZBTB20, MTOR, PDGFRB, RNF125 , SUZ12, ZBTB7A*	NGS TruSeq helgenom In silico panel*	90	EDTA
Monogen diabetes (MODY)	Perifert blod	Monogen diabetes (MODY) v1, 102 gener *ABCC8, AGPAT2, AKT2, APPL1, BSCL2, CAV1, CAVIN1, CEL, CIDEC, CISD2, DCAF17, DNAJC3, DUT, DYRK1B, EIF2AK3, FBN1, FOXP3, GATA4, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, IER3IP1, INS, INSR, KCNJ11, KCNJ6, LIPE, LMNA, MAFA, MANF, MFN2, MIA3, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTX2, NEUROD1, NEUROG3, OTULIN, PAX6, PCBD1, PCNT, PDX1, PIK3R1, PLAGL1, PLIN1, POLD1, PPARG, PPP1R15B, PRKCE, PSMB8, PTF1A, RFX6, SLC16A1, SLC19A2, SLC29A3, TRMT10A, UCP2, WFS1, WRN, ZBTB20, ZFP57, ZMPSTE24*	NGS TruSeq helgenom In silico panel*	90	EDTA
Meckel syndrom och Joubert syndrom	Perifert blod	Meckel syndrom och Joubert syndrom v1, 38 gener *AHI1, ARL13B, ARMC9, B9D1, B9D2, C5ORF42, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KATNIP, KIAA0586, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1,*	NGS TWIST In silico panel*	90	EDTA

		RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15, ZNF423			
Miller-Dieker syndrom	Perifert blod	17p13.3	MLPA enkel	56	EDTA
Multipel endokrin neoplasi typ 1 (MEN1)	Perifert blod	*MEN1*	NGS TruSeq helgenom In silico panel*	90	EDTA
Multipel endokrin neoplasi typ 2 (MEN2)	Perifert blod	*RET*	NGS TruSeq helgenom In silico panel*	90	EDTA
Myotonia och paramyotonia kongenita panel	Perifert blod	*CLCN1, SCN4A*	NGS TruSeq helgenom In silico panel*	90	EDTA
Neurofibromatos typ1	Perifert blod	*NF1, SPRED1*	NGS TWIST In silico panel*	90	EDTA
Neuromuskulär panel (bl.a. myotoni, myasteni, DMD, limb girdle, paramyotonia kongenita) Panelen inkluderar även det mitokondriella genomet samt SMA.	Perifert blod	Neuromuskulär panel v2, 347 gener och 3 repeatexpansioner *AARS1, ABHD5, ACAD9, ACADM, ACADVL, ACTA1, ACTN2, ADSS1, AGL, AGRN, ALDOA, ALG14, ALG2, ALS2, AMPD1, ANO5, AR, ASAH1, ASCC1, ASCC3, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BET1, BICD2, BIN1, BSCL2, BVES, CACNA1A, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CIAO1, CLCN1, CNBP, CNTN1, COL12A1, COL13A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ4, COQ8A, COX6A2, CPT1B, CPT2, CRPPA, CRYAB, CYP2C8, DAG1, DCTN1, DES, DGUOK, DHX16, DMD, DMPK, DNAJB2, DNAJB4, DNAJB6, DNM2, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DTNA, DUX4, DYNC1H1, DYSF, ECEL1, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FAM111B, FBP2, FBXO38, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GARS1, GBE1, GFER, GFPT1, GGPS1, GIPC1, GMPPB, GNE, GOLGA2, GOSR2, GYG1, GYS1, HACD1, HADHA, HADHB, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HRAS, HSPB1, HSPB3, HSPB8, HTRA2, IGHMBP2, INPP5K, ISCU, ITGA7, JAG2, KBTBD13, KCNJ2, KLHL40, KLHL41, KLHL9, KY, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LDB3, LDHA, LETM1, LGI4, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP4,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		MAP3K20, MATR3, MEG3, MEGF10, MICU1, MLIP, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO1, MT-CO2, MT-CO2, MT-CO3, MT-CYB, MTM1, MTMR14, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MUSK, MYBPC1, MYBPC3, MYF5, MYF6, MYH1, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NEB, NEFL, OBSCN, ORAI1, PABPN1, PAX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PHKG1, PIEZO2, PLEC, PLEKHG5, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PPA2, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, REEP1, RFC4, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SELENON, SETX, SGCA, SGCB, SGCD, SGCG, SIGMAR1, SIL1, SLC18A3, SLC22A12, SLC22A5, SLC25A1, SLC25A4, SLC25A42, SLC2A9, SLC52A1, SLC52A2, SLC52A3, SLC5A7, SMCHD1, SMN1, SMN2, SMPX, SNAP25, SNRPN, SNUPN, SPEG, SPG11, SPTBN4, SQSTM1, SRPK3, STAC3, STIM1, STIM2, SUCLA2, SVIL, SYNE1, SYNE2, SYT15, SYT2, TAFAZZIN , TAMM41, TANGO2, TCAP, TIA1, TK2, TMEM43, TNNC2, TNNI1, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRIP4, TRPV4, TSEN54, TSFM, TTN, TYMP, UBA1, UBQLN1, UNC13A, UNC45B, VAMP1, VAPB, VCP, VMA21, VPS33B, VRK1, VWA1, YARS2, ZC4H2 Screening för patogena repeatexpansioner ingår för följande gener: *AR*, *CNBP* och *DMPK*.			
Neuromuskulära sjukdomar, bred panel (inkluderar ataxipanel v5, neuropatipanel v2 och neuromuskulär panel v2)	Perifert blod	Bred NMD panel v1, 1059 gener och 21 repeatexpansioner *AAAS, AARS1, AARS2, ABCA1, ABCA2, ABCB7, ABCD1, ABHD12, ABHD5, ACAD9, ACADM, ACADVL, ACO2, ACOX1, ACTA1, ACTL6B, ACTN2, ADA2, ADAR, ADGRG1, ADPRS, ADSL, ADSS1, AFG3L2, AGL, AGRN, AGTPBP1, AHI1, AIFM1, AIMP1, ALDH18A1, ALDH5A1, ALDOA, ALG14, ALG2, ALG6, ALS2, AMACR, AMPD1, ANG, ANO10, ANO5, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOB, APTX, AR, ARG1, ARL13B, ARL3, ARL6IP1, ARMC9, ARSA, ARV1, ARX, ASAH1, ASCC1, ASCC3, ASL, ASS1, ATAD3A, ATCAY, ATG5, ATG7, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2B3, ATP2B4, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRX, AUH, B3GALNT2, B4GALNT1, B4GAT1, B9D1, B9D2, BAG3, BBS1, BCKDHA, BCKDHB, BCS1L, BEAN1, BET1, BICD2, BIN1, BOLA3, BRAT1, BSCL2, BTD, BVES, C19ORF12, CA8, CACNA1A, CACNA1G, CACNA1S, CACNA2D2, CACNB4, CAMTA1, CAPN1, CAPN3, CARS1, CASK, CASQ1, CAV1, CAV3, CAVIN1, CC2D2A, CCDC78, CCDC88C, CCT5, CD59, CDK16, CDKL5, CEP104, CEP120, CEP290, CEP41, CFAP276, CFL2, CHAMP1,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		CHAT, CHCHD10, CHKB, CHMP1A, CHMP2B, CHP1, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CIAO1, CLCN1, CLCN2, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNBP, CNTN1, CNTNAP1, COA5, COA7, COA8, COASY, COG1, COG4, COG5, COG7, COG8, COL12A1, COL13A1, COL18A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ2, COQ4, COQ6, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6A1, COX6A2, COX6B1, CP, CPLANE1, CPOX, CPS1, CPT1B, CPT1C, CPT2, CRAT, CRPPA, CRYAB, CSPP1, CSTB, CTBP1, CTC1, CTDP1, CTNNA2, CTNNB1, CTSA, CTSD, CTSF, CUL4B, CWF19L1, CYP27A1, CYP2C8, CYP2U1, CYP7B1, DAB1, DAG1, DARS1, DARS2, DBT, DCTN1, DCX, DDHD1, DDHD2, DEGS1, DES, DGAT2, DGUOK, DHDDS, DHFR, DHX16, DHX30, DKC1, DLAT, DLD, DMD, DMPK, DNAJB2, DNAJB4, DNAJB6, DNAJC19, DNAJC3, DNAJC5, DNM1L, DNM2, DNMT1, DOCK3, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DRP2, DST, DSTYK, DTNA, DUX4, DYNC1H1, DYRK1A, DYSF, EBF3, ECEL1, EEF2, EGR2, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, ELP1, EMD, ENO3, ENTPD1, EPG5, EPM2A, EPRS1, ERBB4, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXOSC8, EXOSC9, FA2H, FAH, FAM111B, FAM149B1, FARS2, FASTKD2, FAT1, FAT2, FBLN5, FBP2, FBXL4, FBXO38, FDX2, FDXR, FGD4, FGF12, FGF14, FHL1, FIG4, FITM2, FKBP14, FKRP, FKTN, FLAD1, FLNC, FLVCR1, FMR1, FOLR1, FOXG1, FOXRED1, FRMD4A, FTL, FUS, FXN, FXR1, FZR1, GAA, GABRB1, GABRB2, GABRB3, GAD1, GALC, GALNT2, GAMT, GAN, GARS1, GBA1, GBA2, GBE1, GCDH, GCH1, GCLC, GDAP1, GDAP2, GEMIN4, GEMIN5, GFAP, GFER, GFPT1, GGPS1, GIPC1, GJA1, GJB1, GJC2, GLA, GLB1, GLS, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GPAA1, GPI, GRIA2, GRIA4, GRID2, GRIK2, GRM1, GRN, GSN, GSS, GTPBP2, GYG1, GYS1, HACD1, HACE1, HADHA, HADHB, HARS1, HARS2, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HINT1, HIP1R, HK1, HLCS, HMBS, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HNRNPH2, HPDL, HRAS, HSD17B4, HSPB1, HSPB3, HSPB8, HSPD1, HTRA1, HTRA2, HYCC1, IARS2, IBA57, IFIH1, IFT140, IGHMBP2, INF2, INPP5E, INPP5K, IQSEC1, IRF2BPL, ISCU, ITGA7, ITM2B, ITPR1, JAG2, JAM2, KATNIP, KBTBD13, KCNA1, KCNA2, KCNC1, KCNC3, KCND3, KCNJ10, KCNJ2, KCNMA1, KCNN2, KCNQ2, KCTD7, KIAA0586, KIDINS220, KIF1A, KIF1B, KIF1C, KIF5A, KIF7, KLC2, KLHL40, KLHL41, KLHL9, KY, L1CAM, L2HGDH, LAMA1, LAMA2, LAMA5, LAMB2, LAMP2, LARGE1, LARS2, LDB3, LDHA, LETM1, LGI4, LIG4, LIMS2, LITAF, LMNA, LMNB1, LMNB2, LMOD3, LNPK, LPIN1, LRIF1, LRP4, LRPPRC, LRSAM1, LYRM7, LYST, MAB21L1, MAG, MAN2B1, MAP3K20, MAPK8IP3, MARS1, MARS2, MAST1, MATR3, MBD5, MCM3AP, MCOLN1, MECP2, MECR, MED13L, MEG3, MEGF10, MFN2, MFSD8, MGAT2, MGME1, MICAL1, MICU1, MKS1, MLC1, MLIP, MMACHC, MMADHC, MME, MORC2, MPDU1, MPV17, MPZ, MRE11, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-			

		CO3, MT-CYB, MTFMT, MTM1, MTMR14, MTMR2, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTPAP, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTCL1, MTRFR, MUSK, MVK, MYBPC1, MYBPC3, MYF5, MYF6, MYH1, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NADK2, NAGA, NALCN, NANS, NARS1, NAT8L, NAXE, NDC1, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEB, NEFH, NEFL, NEU1, NEXMIF, NF2, NFASC, NGF, NHLRC1, NIPA1, NKX2-1, NKX6-2, NMNAT1, NOL3, NPC1, NPC2, NPHP1, NPTX1, NR4A2, NT5C2, NTNG2, NTRK1, NUBPL, NUP62, NUS1, OBSCN, OFD1, OGDH, OGDHL, OPA1, OPA3, OPHN1, OPTN, ORAI1, OTC, OTUD4, PABPN1, PANK2, PARS2, PAX6, PAX7, PAX9, PC, PCDH12, PCDH19, PCLO, PCNA, PCYT2, PDE6D, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGK1, PGM1, PGM3, PHKA1, PHKB, PHKG1, PHYH, PIBF1, PIEZO2, PIGG, PIGS, PIGV, PIK3R5, PITRM1, PLA2G6, PLD3, PLEC, PLEKHG5, PLP1, PMM2, PMP2, PMP22, PMPCA, PMPCB, PNKD, PNKP, PNP, PNPLA2, PNPLA6, PNPT1, POGLUT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, POU4F1, PPA2, PPOX, PPT1, PRDM12, PRDM8, PRDX3, PREPL, PRF1, PRICKLE1, PRICKLE2, PRKAG2, PRKCG, PRNP, PRPS1, PRRT2, PRX, PSAP, PSEN1, PTPN11, PTRH2, PTS, PUM1, PURA, PUS1, PYCR2, PYGM, PYROXD1, QARS1, RAB11B, RAB7A, RAD50, RAPSN, RARS1, RARS2, RBCK1, REEP1, REEP2, RELN, REPS1, RETREG1, RFC1, RFC4, RFT1, RNASEH1, RNASEH2B, RNASET2, RNF168, RNF170, RNF216, RNF220, ROGDI, RORA, RPGRIP1L, RPIA, RRM2B, RTEL1, RTN2, RTN4IP1, RUBCN, RXYLT1, RYR1, RYR3, SACS, SAMD9L, SARS1, SARS2, SBF1, SBF2, SCARB2, SCN10A, SCN11A, SCN1A, SCN2A, SCN4A, SCN8A, SCN9A, SCO1, SCYL1, SDHA, SDHAF1, SDHB, SDHD, SELENOI, SELENON, SEPSECS, SEPTIN9, SERAC1, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SHMT2, SIGMAR1, SIL1, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC18A3, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC22A12, SLC22A5, SLC25A1, SLC25A15, SLC25A19, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC2A9, SLC30A9, SLC33A1, SLC39A4, SLC44A1, SLC46A1, SLC52A1, SLC52A2, SLC52A3, SLC5A6, SLC5A7, SLC6A1, SLC6A19, SLC9A1, SLC9A6, SMCHD1, SMN1, SMN2, SMPX, SNAP25, SNRPN, SNUPN, SNX14, SOD1, SORD, SOX10, SPART, SPAST, SPEG, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, SRPK3, STAC3, STIM1, STIM2, STN1,			

		STUB1, STXBP1, STXBP2, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SVBP, SVIL, SYNE1, SYNE2, SYNGAP1, SYT14, SYT15, SYT2, TACO1, TAFAZZIN , TAMM41, TANC2, TANGO2, TARDBP, TBC1D23, TBC1D24, TBCE, TBK1, TCAP, TCF20, TCF4, TCN2, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TELO2, TFG, TGM6, TH, THG1L, TIA1, TINF2, TK2, TMEM106B, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM43, TMEM63A, TMEM67, TMEM70, TNNC2, TNNI1, TNNI2, TNNT1, TNNT3, TNPO3, TOE1, TOP3A, TOR1AIP1, TPK1, TPM2, TPM3, TPP1, TPRKB, TRAPPC11, TRAPPC6B, TRDN, TRIM2, TRIM32, TRIP4, TRMT5, TRNT1, TRPA1, TRPC3, TRPV4, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTC19, TTC21B, TTC8, TTN, TTPA, TTR, TUBA1A, TUBA4A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TWNK, TXN2, TYMP, TYROBP, UBA1, UBA5, UBAP1, UBE3A, UBQLN1, UBQLN2, UBR4, UBTF, UCHL1, UNC13A, UNC45B, UNC80, UQCRB, UQCRQ, UROC1, VAMP1, VAPB, VARS2, VCP, VLDLR, VMA21, VPS11, VPS13A, VPS13D, VPS33B, VPS37A, VPS41, VPS53, VRK1, VWA1, VWA3B, WARS1, WARS2, WASHC5, WDR26, WDR45B, WDR62, WDR73, WDR81, WFS1, WNK1, WWOX, XK, XPA, XRCC1, XRCC4, YARS1, YARS2, YME1L1, ZBTB18, ZC4H2, ZFYVE26, ZIC1, ZIC4, ZNF423, ZSWIM6 Screening för patogena repeatexpansioner ingår för följande gener: *AR, ATN1, ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, BEAN1, CACNA1A, CNBP, DAB1, DMPK, FGF14, FMR1, FXN, NOP56, PPP2R2B, RFC1, TBP, ZFHX3*			
Neuropatipanel (bl.a. Charcot-Marie-Tooth sjukdom, ärftlig tryckkänslig neuropati, hereditär sensorisk och autonom neuropati, transtyretin-medierad amyloidos)	Perifert blod	Neuropatipanel v2, 205 gener *AARS1, ABCA1, ABHD12, AGTPBP1, AIFM1, APOA1, APTX, ARSA, ASAH1, ATL1, ATL3, ATM, ATP1A1, ATP7A, B4GALNT1, BAG3, BCKDHB, BICD2, BSCL2, CD59, CFAP276, CHCHD10, CNTNAP1, COA7, COX6A1, CPOX, CTDP1, CYP27A1, CYP7B1, DARS2, DCTN1, DEGS1, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, ERCC6, ERCC8, EXOSC9, FAH, FBLN5, FBXO38, FGD4, FIG4, FLVCR1, FXN, GALC, GAN, GARS1, GBA2, GDAP1, GJB1, GJC2, GLA, GNB4, GSN, HADHA, HADHB, HARS1, HEXA, HINT1, HK1, HMBS, HSPB1, HSPB8, HYCC1, IARS2, IGHMBP2, INF2, KCNA2, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, LYST, MARS1, MCM3AP, MFN2, MICAL1, MMACHC, MME, MORC2, MPV17, MPZ, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTMR2, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTRFR, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, NAGA, NDC1, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, OPA3, PDHA1, PDK3, PEX10, PEX7, PHYH, PLEKHG5, PMM2, PMP2, PMP22, PNKP, POLG, POLG2, POLR3A, PPOX, PRDM12, PRNP, PRPS1, PRX, PTPN11, RAB7A, REEP1,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		RETREG1, SACS, SBF1, SBF2, SCN10A, SCN11A, SCN9A, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SORD, SOX10, SPAST, SPG11, SPTBN4, SPTLC1, SPTLC2, SURF1, SYT2, TFG, TRIM2, TRPA1, TRPV4, TTPA, TTR, TUBB3, TYMP, UBA1, VAPB, VPS13A, VRK1, VWA1, WARS1, WNK1, XK, XPA, YARS1, ZFYVE26			
Obesitas	Perifert blod	Obesitaspanel v2, 54 gener *ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, GNAS, IFT172, IFT27, IFT74, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MKKS, MKS1, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, RAI1, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP*	NGS TruSeq helgenom In silico panel*	90	EDTA
Okulär albinism	Perifert blod	Okulär albinismpanel v1, 32 gener *AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CACNA1F, DCT, DTNBP1, EDN3, EDNRB, FRMD7, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, PAX6, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1*	NGS TruSeq helgenom In silico panel*	90	EDTA
Optikusatrofi Panelen inkluderar även det mitokondriella genomet.	Perifert blod	Optikusatrofipanel v1, 86 gener *ACO2, AFG3L2, ALPK1, ATAD3A, ATG7, AUH, C19orf12, CISD2, DNAJC19, DNAJC30, DNM1L, EPRS1, FDXR, ISCA2, MECR, MFF, MFN2, MGME1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTPAP, MTRFR, SSBP1, NARS2, NBAS, NDUFA12, NDUFAF3, NDUFS1, NR2F1, OPA1, OPA3, PDSS1, POLG, PRPS1, RTN4IP1, SLC19A2, SLC19A3, SLC25A46, SLC44A1, SLC52A2, SNX10, SPG7, SUCLA2, TFG, TIMM8A, TMEM126A, TSFM, UCHL1, WFS1, YME1L1, ZNHIT3*	NGS TruSeq helgenom In silico panel*	90	EDTA
Osteogenesis imperfecta och benskörhet	Perifert blod	Osteogenesis imperfecta och benskörhetspanel v1, 67 gener *ALPL, ANO5, ASCC1, B3GAT3, B4GALT7, BMP1, CA2, CLCN5, CLCN7, COL1A1, COL1A2, CREB3L1, CRTAP, CTNS, CTSK, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, FGFR1, FKBP10, GNAS, GORAB, IFITM5, LRP5, LRRK1, MBTPS2, MESD, NBAS, NOTCH2, NTRK1, OCRL, OSTM1, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SFRP4, SGMS2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SNX10, SP7, SPARC, SUCO, TAPT1, TCIRG1, TENT5A, TMEM38B,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		TNFRSF11A, TNFRSF11B, TNFSF11, TRIP4, VDR, WNT1, WNT3A, XYLT2			
Pachyonychia congenita		Pachyonychia congenita v1, 19 gener *AAGAB, CARD9, COL7A1, CTSC, DSG1, FZD6, GJB6, JUP, KRT16*, KRT17*, KRT5, KRT6A*, KRT6B*, KRT6C*, MBTPS2, PLCD1, RSPO4, TRPV3, USB1*	NGS TruSeq helgenom In silico panel*	90	EDTA
Palmoplantar keratodermi och erytrodermi		Palmoplantar keratodermi och erytrodermi v1, 42 gener *AAGAB, AQP5, CFTR, COG6, COL14A1, CTSC, DSG1, DSP, ENPP1, GJA1, GJB2, GJB4, GJB6, GRHL2, JUP, KANK2, KRT1, KRT14, KRT16, KRT1, KRT6A, KRT6B, KRT6C, KRT9, LORICRIN, LSS, MBTPS2, MPZ, MT-TS1, PERP, PKP1, RHBDF2, RSPO1, SASH1, SERPINA12, SERPINB7, SLURP1, SMARCAD1, SNAP29, TAT, TRPV3, WNT10A*	NGS TruSeq helgenom In silico panel*	90	EDTA
Paragangliom och feokromocytom	Perifert blod	Paragangliom och feokromocytom panel v2, 17 gener *DLST, EGLN1, EPAS1, FH, MAX, MDH2, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL*	NGS TruSeq helgenom In silico panel**	90	EDTA
Periodisk paralyspanel	Perifert blod	Periodisk paralyspanel v 1, 4 gener CACNA1S, CLCN1, KCNJ2, SCN4A	NGS TruSeq helgenom In silico panel*	90	EDTA
Periodiskt febersyndrom	Perifert blod	*Periodisk febersyndrompanel v2, 22 gener* *ADA2, APOC2, B2M, ELANE, HTR1A, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, OTULIN, POMP, PSMB4, PSMB8, PSTPIP1, TNFAIP3, TNFRSF1A, TRNT1*	NGS TruSeq helgenom In silico panel*	90	EDTA
POLG-relaterad sjukdom Analysen inkluderar även POLG mutationer associerade med Valproat-inducerad leverskada.	Perifert blod	*POLG*	NGS TruSeq helgenom In silico panel*	90	EDTA
Polycystisk njursjukdom	Perifert blod	Polycystisk njursjukdomspanel v2, 23 gener *ALG5, ALG8, ALG9, COL4A1, DNAJB11, DZIP1L, GANAB, HNF1B, IFT140, JAG1, NEK8, NOTCH2*, PKD1*, PKD2, PKHD1, PMM2, PRKCSH, SEC61A1, SEC63, TSC1, TSC2, UMOD, VHL*	NGS TruSeq helgenom In silico panel*	90	EDTA
Prader-Willi syndrom	Perifert blod	15q11.2	MLPA metylering	56	EDTA

Primär ciliär dyskinesi PCD)	Perifert blod	PCD panel v2, 44 gener CCDC39, CCDC40, CCNO, CFAP298, CFAP300, CFTR, DNAAF1, DNAAF11, DNAAF19, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, DRC2, DRC4, FOXJ1, GAS2L2, HYDIN, LRRC56, MCIDAS, NEK10, ODAD1, ODAD2, ODAD3, ODAD4, OFD1, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, SPEF2, ZMYND10	NGS TruSeq helgenom In silico panel*	90	EDTA
Primär hyperaldosteronism	Perifert blod	Primär hyperaldosteronismpanel v1, 5 gener *CACNA1H, CLCN2, CYP11B1, CYP11B2. KCNJ5*	NGS TruSeq helgenom In silico panel	90	EDTA
RASopatier (bl.a. CFC, Costello syndrom, Noonan syndrom, Legius syndrom, NF1)	Perifert blod	RASopatipanel v3, 30 gener *ACTB, ACTG1, BRAF, CBL, CDC42, FBXW11, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2, SYNGAP1*	NGS TWIST In silico panel* NGS TruSeq helgenom In silico panel*	90	EDTA
Retinal degeneration	Perifert blod	Retinal degenerationpanel v1, 365 gener *ABCA4, ABCC6, ABCD1, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AGBL5, AHI1, AIPL1, AIRE, ALMS1, ALPK1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARR3, ARSG, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CFAP418, CHM, CIB2, CISD2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, COQ2, CPE, CPLANE1, CRB1, CRPPA, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTSD, CWC27, CYP4V2, DHDDS, DHX38, DNAJC5, DRAM2, DTHD1, DYNC2H1, EFEMP1, ELOVL4, EMC1, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT74, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KATNIP, KCNJ13, KCNV2, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MED12, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MMACHC, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-*	NGS TruSeq helgenom In silico panel*	90	EDTA

		RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MTTP, MVK, MYO7A, NAGLU, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTX2, P3H2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDSS1, PDSS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PISD, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RIMS2, RLBP1, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SCLT1, SDCCAG8, SEMA4A, SGSH, SLC24A1, SLC25A46, SLC38A8, SLC45A2, SLC6A6, SLC7A14, SNRNP200, SPATA7, SPP2, SRD5A3, SSBP1, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TLCD3B, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, VPS13B, WDPCP, WDR19, WFS1, WHRN, ZNF408, ZNF423, ZNF513			
Schwannomatos och meningiom	Perifert blod	Schwannomatos och meningiompanel v1, 10 gener *LZTR1, MEN1, NF2, PRKAR1A, PTCH1, PTEN, SMARCB1, SMARCE1, SUFU, WRN*	NGS TruSeq helgenom In silico panel*	90	EDTA
SHOX-relaterad kortvuxenhet	Perifert blod	*SHOX*	NGS TruSeq helgenom In silico panel*	90	EDTA
Silver-Russel syndrom	Perifert blod	11p15	MLPA metylering	56	EDTA
Skelettdysplasi (bred)	Perifert blod	Skelettdysplasipanel v1, 400 gener *ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AFF3, AFF4, AGA, AGPS, AIFM1, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARSB, ARSL, ASCC1, ASXL1, ASXL2, ATP6V0A2, ATR, B3GALT6, B3GAT3, B3GLCT, B4GALT7, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CC2D2A, CCDC8, CCN6, CDC45, CDC6, CDH3, CDKN1C, CDT1, CEP120, CEP152,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		CEP290, CFAP410, CHST14, CHST3, CHSY1, CILK1, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRIPT, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DDRGK1, DHCR24, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DOCK6, DONSON, DVL1, DVL2, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EFTUD2, EIF2AK3, ENPP1, EOGT, ERF, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FBN1, FBN2, FERMT3, FGF10, FGF16, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FTO, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GH1, GHR, GHRHR, GHSR, GJA1, GLB1, GLI3, GMNN, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GSC, GUSB, GZF1, HDAC8, HES7, HGSNAT, HPGD, HSPG2, IARS2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT80, IFT81, IGF1, IGF1R, IGF2, IGFALS, IHH, IL1RN, INPPL1, INTU, KAT6B, KIAA0586, KIAA0753, KIF22, KIF7, KL, KMT2A, KMT2D, LBR, LEMD3, LFNG, LHX3, LHX4, LIFR, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP2, LTBP3, MAFB, MAN2B1, MAP3K7, MATN3, MBTPS1, MBTPS2, MEGF8, MEOX1, MESD, MESP2, MGP, MKS1, MMP13, MMP2, MMP9, MNX1, MSX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIX, NIPBL, NKX3-2, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NSD1, NSDHL, NTRK1, NXN, OBSL1, OCRL, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PGM3, PHEX, PIGV, PIK3C2A, PISD, PITX1, PKDCC, PLOD2, PLS3, POC1A, POLR1A, POLR1C, POLR1D, POP1, POR, POU1F1, PPIB, PRKAR1A, PROP1, PTDSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB23, RAB33B, RBBP8, RECQL4, RIPPLY2, ROR2, RPGRIP1L, RSPRY1, RUNX2, SALL1, SALL4, SBDS, SC5D, SEC24D, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMARCAL1, SMC1A, SMC3, SNRPB, SNX10, SOST, SOX3, SOX9, SP7, SPARC, SQSTM1, SUCO, SUMF1, TAB2, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN3, TENT5A, TGFB1, TGFB2, TGFBR2, TMEM165, TMEM216, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAF3IP1, TRAPPC2, TREM2, TRIP11, TRIP4, TRPS1, TRPV4, TRPV6, TTC21B, TWIST1, TYROBP, UFSP2, VDR, WDR19, WNT1, WNT10B, WNT3A, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, ZMPSTE24, ZNF687, ZSWIM6			
Smith-Magenis syndrom	Perifert blod	17p11.2	MLPA enkel	56	EDTA

Spinal Muskelatrofi (typ I-III)	Perifert blod	*SMN1/2*	MLPA enkel	56	EDTA
Syndromutredning Utvecklingsförsening/Autism Intellektuell funktionsnedsättning	Perifert blod	Screening	Mikroarray	56	EDTA
Anlagsbärarutredning för Syndromutredning Utvecklingsförsening/Autism Intellektuell funktionsnedsättning	Perifert blod	Screening	Mikroarray Riktad	56	EDTA
Tanatofor dysplasi (Se även *FGFR3*-relaterad skelettdysplasi)	Perifert blod	*FGFR3* exon 7, 9, 14, 18	Sangersekvensering *FGFR3*	56	EDTA
Trombocytopeni	Perifert blod	Trombocytopenipanel v2, 127 gener *ABCG5, ABCG8, ACD , ACTN1, ADA, ADAMTS13, ANKRD26, ANO6, AP3B1, BLOC1S3, BLOC1S6, BRCA2, CTC1, CTLA4, CYCS, DDX41, DIAPH1, DKC1, DNAJC21, DTNBP1, EFL1, ERCC4, ERCC6L2, ETV6, F10, F11, F13A1, F13B, F2, F5, F7, F8, F9, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FGA, FGB, FGG, FERMT3, FLI1, FYB1, GATA1, GATA2, GFI1B, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, LIG4, LYST, MAD2L2, MECOM , MYH9, MYSM1, NBEAL2, NHP2, NOP10, P2RY12, PALB2, PARN, PLAU, POT1, PRKACG, RASGRP2, RAD51, RAD51C, RBM8A, RFWD3, RPL11, RPL15, RPL18, RPL23, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL36, RPL5, RPS7, RPS10, RPS15A, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS29, RTEL1, RUNX1, SBDS, SLFN14, SLX4, SRC, STIM1, STN1, TBXA2R, TBXAS1, TERT, THPO, TINF2, TSR2, TUBB1, UBE2T, VWF, WAS , WIPF1, WRAP53, XRCC2, ZCCHC8*	NGS TruSeq helgenom In silico panel*	90	EDTA
Tuberös skleros	Perifert blod	*TSC1, TSC2*	NGS TWIST In silico panel*	90	EDTA
Tubulopati	Perifert blod	Tubulopatipanel v1, 112 gener *AGXT, AIRE, ALPL, AMMECR1, AP2S1, APRT, AQP2, ATP1A1, ATP6V0A4, ATP6V1B1, ATP7B, AVP, AVPR2, BSND, CA2, CACNA1D, CACNA1H, CACNA1S, CASR, CDC73, CDKN1B, CLCN2, CLCN5,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, CTNS, CUL3, CYP11B1*, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, DMP1, EHHADH, ENPP1, FAH, FAM111A, FAM20A, FGF23, FOXI1, GALNT3, GATA3, GATM, GCM2, GLA, GNA11, GNAS, GRHPR, HNF1B, HNF4A, HOGA1, HPRT1, HSD11B2, KCNA1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KLHL3, LCAT, MAGED2, MEN1, MOCOS, MT-TF, MMUT, NR3C1, NR3C2, OCRL, PCBD1, PDE3A, PHEX, PTH, PTH1R, RET, RMND1*, RRAGD, RRM2B, SARS2, SCN4A, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC5A2, SLC6A19, SLC7A7, SLC7A9, STX16, TBCE, TRPM6, TRPM7, UMOD, VDR, VIPAS39, VPS33B, WDR72, WNK1, WNK4, XDH.*			
Tyroideahormonresistens (inkl. hypotyroidism)	Perifert blod	Tyroideahormonresistenspanel v2, 35 gener *CASR, CDCA8, DUOX2, DUOXA2, FOXE1, GCM2, GLIS3, GNAS, HESX1, IGSF1, IRS4, IYD, LHX3, LHX4, NKX2-1, NKX2-5, OTX2, PAX8, POU1F1, PROP1, SECISBP2, SLC16A2, SLC26A4, SLC26A7, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TRH, TRHR, TSHB, TSHR, TUBB1*	NGS TruSeq helgenom In silico panel	90	EDTA
Hippel-Lindau (VHL)	Perifert blod	*VHL*	NGS TruSeq helgenom In silico panel	90	EDTA
Welanders distala myopati	Perifert blod	*TIA1* exon 13	Sangersekvensering TIA1	56	EDTA
Williams syndrom	Perifert blod	7q11.23	MLPA enkel	56	EDTA
Wolf-Hirschhorn syndrom	Perifert blod	4p telomer	MLPA enkel	56	EDTA
Ärftlig hematologi	Perifert blod	Ärftlig hematologipanel v3, 226 gener *ABCG5, ABCG8, ACD, ACTN1, ADA, ADAMTS13, AIRE, AK2, ALAS2, ANKRD26, ANO6, AP3B1, ATG2B, ATM, BAP1, BLM, BLOC1S3, BLOC1S6, BPGM, BRAF, BRCA1, BRCA2, BRIP1, CASP10, CBL, CD27, CD40LG, CDAN1, CDIN1, CDKN2A, CEBPA, CHEK2, CLPB, COPZ1, CSF3R, CTC1, CTLA4, CXCR4, CYCS, DCLRE1B, DDX41, DIAPH1, DKC1, DNAJC21, DOCK8, DTNBP1, EFL1, EGLN1, ELANE, EPAS1, EPCAM, EPO, EPOR, ERCC4, ERCC6L2, ETV6, F10, F11, F13A1, F13B, F2, F5, F7, F8, F9, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FERMT3, FGA, FGB, FGG, FLI1, FYB1, G6PC3, GATA1, GATA2, GFI1, GFI1B, GP1BA, GP1BB, GP6, GP9, GRHL2, GSKIP, HAX1, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF1,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		IL17RA, IL2RG, IRF8, ITGA2B, ITGB3, ITK, JAGN1, JAK2, KLF1, KRAS, LIG4, LYST, LZTR1, MAD2L2, MAGT1, MDM4, MECOM, MLH1, MPL, MSH2, MSH6, MYH9, MYSM1, NAF1, NBN, NF1, NF2, NHP2, NOP10, NPM1, NRAS, P2RY12, PALB2, PARN, PAX5, PIK3CD, PLAU, PML, PMS2, POLA2, POLD1, POLE, POT1, PRF1, PRKACG, PTEN, PTPN11, RAD51, RAD51C, RAD51D, RAF1, RASGRP2, RBBP6, RBM8A, RFWD3, RIT1, RPA1, RPL11, RPL15, RPL18, RPL23, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL36, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHB, SDHC, SDHD, SEC23B, SH2D1A, SHOC2, SLFN14, SLX4, SOS1, SRC, SRP54, SRP72, STIM1, STN1, TBXA2R, TBXAS1, TCIRG1, TERC, TERT, THBD, THPO, TINF2, TNFRSF13B, TP53, TSR2, TUBB1, TYK2, UBE2T, UROS, USB1, VHL, VPS13B, VPS45, VWF, WAS, WIPF1, WRAP53, XRCC2, ZCCHC8.			
Ärftlig njursjukdom (bl.a. Alport syndrom, nefronoftis, nefrostiskt syndrom)	Perifert blod	Ärftlig njursjukdomspanel v1, 290 gener *ACE, ACTN4*, ADA2, AGT, AGTR1, AGXT, AHI1, AIRE, ALG5, ALG8, ALG9, ALMS1*, ALPL, AMMECR1, AMN, ANKS6, ANLN, AP2S1, APOA1, APOA4, APOC2, APOE, APRT, AQP2, ARHGDIA, ARL13B, ARL6, ATP1A1, ATP6V0A4, ATP6V1B1, ATP7B, AVP, AVPR2, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BSND, C3, CA2, CACNA1D, CACNA1H, CACNA1S, CASR, CC2D2A, CD151, CD2AP, CD46*, CDC73, CDKN1B, CEP164, CEP290*, CEP41, CEP83, CFB, CFH*, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CHRM3, CLCN2, CLCN5, CLCNKB*, CLDN10, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CSPP1, CTNS, CUBN*, CUL3, CYP11B1*, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, DAAM2, DCDC2, DGKE, DICER1*, DLC1, DLG5, DMP1, DNAJB11, DYNC2H1, DYNC2I1, DZIP1L, EHHADH, ENPP1, FAH, FAM111A, FAM20A, FAN1, FAT1, FGA, FGF23, FN1, FOXI1, GALNT3, GANAB, GATA3, GATM, GCM2, GLA, GLIS2, GNA11, GNAS, GON7, GRHPR, GSN, HNF1B, HNF4A, HOGA1, HPRT1, HSD11B2, IFT122*, IFT140, IFT172, IFT27, IFT43, IFT74, INF2, INPP5E, INVS, IQCB1, ITGA3, ITSN1, ITSN2, JAG1, KANK2, KCNA1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KLHL3, LAGE3, LAMA5, LAMB2, LCAT, LMX1B, LRP2, LYZ, LZTFL1, MAFB, MAGED2, MAGI2, MAPKBP1, MEN1, MKKS, MKS1, MMACHC, MMUT, MOCOS, MT-TF, MYH9, MYO1E, NEK1, NEK8, NLRP3, NOS1AP, NOTCH2*, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NPR1, NR3C1, NR3C2, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, OCRL, OFD1, OSGEP, P3H2, PAX2, PCBD1, PDE3A, PDSS2, PHEX, PKD1*, PKD2, PKHD1, PLCE1, PMM2, PODXL, PRKCSH, PSKH1, PTH, PTH1R, PTPRO, REN, RET, RMND1*, RPGRIP1L, RRAGD, RRM2B, SARS2, SCARB2, SCLT1, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SGPL1, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC5A2,*	NGS TruSeq helgenom In silico panel*	90	EDTA

		SLC6A19, SLC7A7, SLC7A9, SMARCAL1, STX16, TBC1D8B, TBCE, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNS2, TP53RK, TPRKB, TRAF3IP1, TRIM8, TRPC6, TRPM6, TRPM7, TSC1, TSC2, TSEN2, TTC21B, TTC8, TTR, TULP3, TXNDC15, UMOD, VDR, VHL, VIPAS39, VPS33B, WDPCP, WDR19, WDR35, WDR72, WDR73, WNK1, WNK4, WT1, XDH, XPNPEP3, XPO5, YRDC.			

* Sekvensering samt deletions/duplikationsanalys ingår i alla genpaneler NGS TWIST In silico panel samt NGS TruSeq helgenom In silico panel).

Alla genpaneler baserade på NGS TWIST In silico panel alternativt NGS TruSeq helgenom In silico panel kan expanderas till helexom- respektive helgenomsekvensering av alla sjukdomsassocierade gener. Vänligen skicka ny remiss om så önskas.