

Analysutbud Konstitutionella sjukdomar

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 metafase	90# (# vid beställning av unika prober TAT 130 dagar)	Heparin

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
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 <i>FGFR3</i> -relaterad skelettdysplasi)	Perifert blod	<i>FGFR3</i> (exon 10)	Sangersekvens ering <i>FGFR3</i>	56	EDTA
Amyloidos (se även Transtyretinmedierad amyloidos)	Perifert blod	Amyloidospanel v2, 17 gener (<i>APOA1, APOA2, APOA4, APOC2, APOC3, B2M, CST3, EFEMP1, FGA, GSN, LECT2, LYZ, MEFV, MVK, NLRP3, TNFRSF1A, TTR</i>)	NGS TruSeq helgenom In silico panel*	90	EDTA
Angelman syndrom	Perifert blod	15q11.2	MLPA metylering	56	EDTA
Androgenokänslighetssyndrom (AIS)	Perifert blod	<i>AR</i> inklusive repeatanalys	NGS TruSeq helgenom In silico panel*	90	EDTA
Angioödem	Perifert blod	Angioödem v1, 7 gener <i>ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG</i> och <i>SERPING1</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Arytmi och kardiomyopati (inkl. ARVC, Brugada syndrom, LQTS, CPVT, kardiomyopati, HCM, DCM)	Perifert blod	Arytmi och kardiomyopati-panel v3, 187 gener och 1 repeatexpansion ABCC9, ACADVL, ACTA1, ACTN2, AGL, AKAP9, ALG10, ALG10B, ALMS1*, ALPK3, ANK2, ANKRD1, ATAD3A*, BAG3, BRAF*, CACNA1C*, CACNA2D1, CACNB2, CALM1*, CALM2, CALM3, CALR3, CASQ2, CAV3, CAVIN4, CDH2, COA5, CRYAB, CSRP3, CTF1, CTNNA3, DES, DLG1, DMD, DNAJC19, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EMD, EPG5, EYA4, FHL1*, FHL2, FHOD3, FKRP, FKTN, FLII, FLNC*, FOXRED1, FXN*, GAA, GATA6, GATAD1, GLA, GLB1, GPD1L, GUSB*, GYG1, HAMP, HCN4, HFE, HFE2, HJV, HRAS, IDH2, ILK, JPH2, JUP, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLF10, KLHL24, KRAS*, LAMA4, LAMP2, LDB3, LMNA, LZTR1, MAP2K1, MAP2K2, MIB1, MPO, MRAS, MRPL3, MTO1, MT-TI, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYLK3, MYO6, MYOM1, MYOZ2, MYPN, MYZAP, NEBL, NEXN, NKX2-5, NOS1AP, NPPA, NRAP, NRAS, PCCA, PCCB, PDLIM3, PKP2*, PLN, PPA2, PPCS, PPP1CB, PPP1R13L, PRDM16, PRKAG2, PTPN11, RAB3GAP2, RAF1, RANGRF, RASA1, RBM20, RHBDF1, RIT1, RPL3L, RPS6KB1, RRAGC, RRAGD, RYR2, SCN10A, SCN1B, SCN2B, SCN4B, SCN5A, SCO2, SDHA*, SGCB, SGCD, SGCD, SHOC2, SLC25A3, SLC25A4, SLC40A1, SLC6A6, SLMAP, SNTA1, SOS1, SPEG, SYNE1, SYNE2, TAB2, TAFAZZIN, TBX20*, TBX5, TCAP, TECRL, TFR2, TGFB3, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSFM, TTN*, TTR, TULP3, TXNRD2, VCL, XK Screening för patogena repeatexpansioner ingår för följande gen: <i>DMPK</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Ataxi (inkl. CANVAS, episodisk ataxi, FXTAS, spastisk ataxi och spinocerebellär ataxi samt repeatanalys för ataxisjukdomar) Panelen inkluderar även det mitokondriella genomet.	Perifert blod	Ataxi-panel 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,	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		COX6A2, COX6B1, CP, CPLANE1, CPS1, CPT1C, CRAT, CSPP1, CSTB, CTBP1, CTC1, CTDP1, CTNNA2, CTNNB1, CTSB, 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, DNMT2, 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, FKR, 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, LNP, LRP4, LRPPRC, LRSAM1, LYRM7, LYST, MAB21L1, MAG, MAN2B1, MAPK8IP3, MARS1, MARS2, MAST1, MATR3, MBD5, MCOLN1, MECP2, MECP, MED13L, MFN2, MFSB8, 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, MTTT, 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, NDUF51, NDUF52, NDUF53, NDUF54, NDUF56, NDUF57, NDUF58, 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,			

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, UQCRCQ, 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	Perifert blod	Bindvävspanel v3, 67 gener ABL1, ACTA2, ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ASPH, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL12A1, COL1A1, COL1A2,	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
(bl a Ehlers Danlos syndrom, Marfan syndrom, Loeys-Dietz syndrom och TAAD)		<i>COL3A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, DSE, EFEMP1, EFEMP2, ELN, FBN1, FBN2, FKBP14, FBLN5, FLNA, FOXE3, GORAB, IPO8, LOX, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PMEPA1, PRDM5, PRKG1, PYCR1, RIN2, ROBO3, SECISBP2, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFB3R1, TGFB3R2, TNXB, ZNF469</i>			
Bröstcancer - snabbspår	Perifert blod	Bröstcancerpanel v1, 12 gener <i>ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53</i>	NGS TWIST In silico panel*	28	EDTA
CADASIL	Perifert blod	<i>NOTCH3</i> (Se även cerebrala småkärlssjukdomar)	NGS TruSeq helgenom In silico panel*	90	EDTA
Cerebrala småkärlssjukdomar (bl.a. Moyamoya, Hereditär hemorragisk telangiectasi, Capillary malformation-arteriovenous malformation (CM-AVM) syndrom, Parkes-Weber syndrom)	Perifert blod	Cerebrala småkärlssjukdomspanel v3, 50 gener <i>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, SAMHD1, SHOC2, SLC2A10, SMAD4, SOS1, SOS2, SPRED1, TREX1, YY1AP1</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Charcot-Marie-Tooth (CMT1A)	Perifert blod	<i>PMP22</i> (ingår även i Neuropatipanel)	MLPA enkel	56	EDTA
Ciliopati (Neurologiska, oftalmologiska, renala och skelettciliopatier, dextrokardi, heterotaxi)	Perifert blod	Ciliopatipanel v1, 124 gener <i>AHI1, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C2CD3, CBY1, CC2D2A, CCDC39, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP410, CFAP418, CFAP53, CFC1, CIROP, CPLANE1, CRB2, CSPP1, DDX59, DHCR7, DLG5, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EVC, EVC2, EXOC3L2, FAM149B1, GDF1, GLI3, GLIS2, HES7, HNF1B, HYL51, ICK, IFT122, IFT140, IFT172, IFT173, IFT43, IFT52, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, IQCE, KIAA0586, KIAA0753, KIF7, LAMA1, LBR, LZTFL1, MAPKBP1, MEGF8, MKKS, MKS1, MMP21, NEK1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PIBF1, PIK3C2A, PKD1, PKD1L1, PKD2, PKHD1, PMM2, PRKACA, PRKACB, PSKH1, RPGRIP1L, SBDS, SCLT1, SDCCAG8, SMAD2, SUFU, TBC1D32, TCTN1, TCTN2, TCTN3, TMEM107,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		<i>TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TTC21B, TTC8, TXNDC15, VPS13B, WDPCP, WDR19, WDR35, XPNPEP3, ZIC3, ZSWIM6.</i>			
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 <i>AGBL1, ADAMTS18, ALDH18A1, B3GLCT, CHRD1, 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</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Cystisk Fibros	Perifert blod	<i>CFTR</i> (50 mutationer)	Fragmentanalys CFTR	35	EDTA
Cystisk Fibros	Perifert blod	<i>CFTR</i>	NGS TWIST In silico panel*	90	EDTA
DSD (Disorders of sex development)	Perifert blod	DSD Panel v1, 192 gener och 2 regioner <i>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, NDNF, NOBOX, NOG, NROB1, NR2F2, NR3C1, NR5A1, NSMF, NUP107,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		PAX8, PBX1, PCDH17, PCSK1, PGRMC1, PLXNA3, PMM2, POF1B, POLG, POLR2C, POLR3H, POR, POU5F1, PPP1R12A, PRDM13, PROK2, PROKR2, PROP1, PSMC3IP, RCBTB1, RIPK4, RNF216, RPL10, RSP01, 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 Region: 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)	Sangersekvens ering 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, MECP, 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, TSP0AP1, TUBB4A, UBTf, VAC14, VAMP1, VAMP2, VPS13A, VPS13D, VPS16, VPS41, VPS4A, WDR45, WDR73, XPR1, YIF1B, YY1, ZSWIM6	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		Screening för patogena repeatexpansioner ingår för följande gener: <i>ATN1, ATXN1, ATXN2, ATXN3, CACNA1A, CSTB, FXN, PPP2R2B, TBP</i>			
Dystrofia myotonika typ 1	Perifert blod	<i>DMPK</i>	Fragmentanalys DMPK	35	EDTA
Ektodermal dysplasi	Perifert blod	Ektodermal dysplasipanel v1, 111 gener <i>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, SMARCA1, SMARCB1, SMARCE1, SMOC2, SNRPE, SOX9, SOX18, SPINK5, SREBF1, TBC1D24, TFAP2B, TP63, TRAF6, TRPS1, TRPV3, TSPEAR, UBR1, WDR19, WDR35, WNT10A, WNT10B</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Epidermolysis bullosa (EB)	Perifert blod	Epidermolysis bullosa panel v2, 31 gener <i>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</i>	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, 14 regioner och 3 repeatexpansioner <i>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, ARFGF1, ARFGF2, 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, BTBD, C12orf57, C2orf69, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D2, CAD, CAMK2B, CAPRIN1, CARS2, CASK, CC2D2A, CDK19, CDKL5, CELF2, CEP85L, CERS1, CHD2, CHD4, CHD5, CHKA, CHRNA2, CHRNA4, CHRNA7, CHRNA2, CIC, CLCN3, CLCN4, CLDN5, CLN3,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, PIQ, 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, PRR2, PSAP, PTCD3, PTEN, PTPN23, PTS, PUM1, PURA, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB39B, RAB5C, RAC3, RALA, RALGAP1, 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,			

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		<i>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, SMARCC2, 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, UFPS2, UGDH, UGP2, UNC80, USP18, VAMP2, VARS1, VPS11, WARS2, WASF1, WDR26, WDR37, WDR45, WDR45B, WDR73, WNK3, WWOX, YIPF5, YWHAG, ZBTB18, ZBTB47, ZDHHC9, ZEB2, ZNF142, ZNF335</i> Regioner: 1p36, 1q43q44, 4p16.3, 8p23.1, 15q11.13, 15q13.3, 16p12.2, 16p13.11, 17p13.3, 17q12, 22q11.2, Xp11.22p11.23, Xq25, Xq28 Screening för patogena repeatexpansioner ingår för följande gener: <i>ARX, ATN1, CSTB</i>			
Erythrocytos	Perifert blod	Erythrocytospanel v1, 7 gener <i>BPGM, EGLN1, EPAS, EPO, EPOR, JAK2, VHL</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Familjär hyperkolesterolemi (FH)	Perifert blod	Familjär hyperkolesterolemi v2, 7 gener <i>ABCG5, ABCG8, APOB, APOE, LDLR, LDLRAP1, PCSK9</i>	NGS TWIST In silico panel*	90	EDTA
<i>FGFR3</i> -relaterad skelettdysplasi	Perifert blod	<i>FGFR3</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Fragilt-X; FRAXA	Perifert blod	<i>FMR1</i>	Fragmentanalys Fragilt-X	35	EDTA
FXTAS	Perifert blod	<i>FMR1</i>	Fragmentanalys Fragilt-X	35	EDTA
Genotypning	Perifert blod		QF-PCR	35	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Helexomsekvensering	Perifierat blod	Screening	NGS TWIST Exom Trio NGS TWIST Exom Duo NGS TWIST Exom Singleton	90	EDTA
Helgenomsekvensering Inkl. mitokondriella genomet samt relevanta repeatexpansionssjukdomar	Perifierat blod	Screening	NGS TruSeq Helgenom Trio NGS TruSeq Helgenom Duo NGS TruSeq Helgenom Singleton	90	EDTA
Helgenomsekvensering long-read Inkl. mitokondriella genomet samt relevanta repeatexpansionssjukdomar	Perifierat blod	Screening	LRS PacBio Helgenom Trio LRS PacBio Helgenom Duo LRS PacBio Helgenom Singleton, LRS PacBio In silico panel	90	EDTA
Hereditär hemorragisk telangiectasi (HHT; Osler- Weber-Rendus syndrom) (bl.a. HHT, Capillary malformation-arteriovenous malformation (CM-AVM))	Periferat blod	Hereditär hemorragisk telangiectasipanel v1, 6 gener <i>ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
syndrom, Parkes-Weber syndrom)					
Hereditär spastisk paraplegi (HSP)	Perifert blod	Hereditär spastisk paraplegi panel v2, 115 gener, 1 region och 9 repeatexpansioner <i>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, NTSC2, OPA3, PCYT2, PLP1, PNPLA6, POLR3A, PPFIBP1, PRNP, PSEN1, RAB3GAP2, REEP1, REEP2, RETREG1, RNASEH2B, RNF170, RNU7-1, RTN2, SACS, SERAC1, SLC16A2, SLC1A4, SLC25A15, SLC25A46, SLC2A1, SPART, SPAST, SPATA5L1, SPG11, SPG21, SPG7, SPTAN1, STN1, TAF8, TECPR2, TFG, TMEM63C, TUBB4A, UBAP1, UCHL1, WASHC5, WDR45B, ZFYVE26</i> Region: Xq28 regionen (inklusive MECP2) Screening för patogena repeatexpansioner ingår för följande gener: <i>ATXN1, ATXN2, ATXN3, ATXN7, ATXN10, CACNA1A, FXN, PPP2R2B, TBP</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
HNPP, fam. tryckförflamning	Perifert blod	<i>PMP22</i>	MLPA enkel	56	EDTA
Huntingtons sjukdom	Perifert blod	<i>HTT</i> (Huntingtin)	Fragmentanalys HTT (HD)	35	EDTA
Hyperinsulinism och hypoglykemi	Perifert blod	Hyperinsulinism och hypoglykemi v1, 47 gener <i>ABCC8, ACAT1, AGL, AKT2, ALDOA, ALDOB, BTB, CACNA1C, CACNA1D, FBP1, FOXA2, G6PC1, GBE1, GCK, GLUD1, GPC3, GYS2, HADH, HK1, HLCS, HNF1A, HNF4A, INSR, IVD, KCNJ11, KDM6A, KMT2D, LDHA, MAGEL2, MCEE, MMUT, NSD1, OXCT1, PC, PCCA, PCCB, PGM1, PHKA2, PHKB, PHKG2, PMM2, PYGL, SLC16A1, SLC2A2, SLC37A4, TANGO2, UCP2</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Hyperlipidemi	Perifert blod	Hyperlipidemipanel v1, 18 gener <i>ABCA1, ABCG5, ABCG8, APOA1, APOA5, APOB, APOC2, APOE, CREB3L3, CYP27A1, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LMF1, LPL, PCSK9</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Hyperparatyreoidism (inkl Familjär hypokalciurisk, hyperkalcemi (FHH), och Multipel endokrin neoplasii)	Perifert blod	Hyperparatyreoidismpanel v2, 8 gener <i>AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Hypofystumör, ärftlig	Perifert blod	Ärftlig hypofystumör v1, 6 gener <i>AIP, CDKN1B, DICER1, MEN1, NF1, PRKAR1A</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Hypogonadotrop hypogonadism (inkl. Kallman syndrom)	Perifert blod	Hypogonadotrop hypogonadismpanel v1, 31 gener <i>ANOS1, CHD7, CUL4B, DCAF17, FEZF1, FGF8, FGFR1, FSHB, GLI2, GNRH1, GNRHR, HAMP, HFE, IL17RD, KISS1R, KLB, LHB, LHX4, NROB1, NSMF, PROK2, PROKR2, PROP1, SLC29A3, SLC40A1, SOX10, SOX2, TAC3, TACR3, TFR2, WDR11</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Hypoparatyreoidism (inkl. Albright hereditary osteodystrophy, pseudohypoparathyroidism, pseudopseudohypoparathyroidism, acrodysostosis and osteoma cutis)	Perifert blod	Hypoparatyreoidismpanel v1, 11 gener <i>AIRE, CASR, GATA3, GCM2, GNA11, GNAS, PDE4D, PRKAR1A, PTH, STX16, TBCE</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Hypokondroplasi (Se även <i>FGFR3</i> -relaterad skelettdysplasi)	Perifert blod	<i>FGFR3</i> (exon 9, 12)	Sangersekvens ering <i>FGFR3</i>	56	EDTA
Hörselnedsättning	Perifert blod	Hörselnedsättning panel v1, 289 gener <i>ABHD12, ABHD5, ACOX1, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, AMMECR1, ANKH, ARSG, ATP11A, ATP1A3, ATP2B2, ATP6V0A4, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTBD, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CD164, CDC14A,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		<i>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, TSHZ1, TSPEAR, TUBB4B, TWNK, TYR, UBR1, USH1C, USH1G, USH2A, WBP2, WFS1, WHRN, XYLT2, ZNF469</i>			
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

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Iktyos, NGS panel (bl.a. Acral Peeling Skin Syndrome (APSS), Erytrokeratoderma variabilis, (EKV), Harlequin syndrom, könsbunden iktyos, KID syndrom, Sjögren-Larssons syndrom)	Perifert blod	Iktyospanel v3, 71 gener <i>AARS1, ABCA12, ABHD5, ADAMTS17, ALDH3A2, ALOX12B, ALOXE3, AP1B1, AP1S1, ASPRV1, CARD14, 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</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Immunologisk sjukdom (bl.a immunbrist, inflammatorisk tarmsjukdom, autoinflammatorisk sjukdom, kronisk granulomatös sjukdom, periodisk feber, SCID, kongenital neutropeni, och autoimmunitetssyndrom)	Perifert blod	Immunpanel v1, 355 gener <i>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, CTNBL1, 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, MCT5, MEFV, MOGS, MPEP1, MRTFA, MSN, MTHFD1, MVK, MYD88, MYO5B, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NEUROG3, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRP4, 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, RELB, 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, SLC12A1, SMARCA1, SMARCD2, SNORA31, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP54, STAT1, STAT2, STAT3, STAT4,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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
Intellectuell funktionsnedsättning	Perifert blod	Intellectuell funktionsnedsättning panel v1, 1453 gener, 40 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, 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, B3GALCT, 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, BTBD, 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,	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, CTDPI, CTNNA2, CTNNB1, CTNND1, CTNND2, CTR9, CTS, 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, DEPD5, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHTKD1, DHX30, DHX37, DHX9, DIAPH1, DIS3L2, DKC1, DLD, DLG1, DLG3, DLG4, DLL1, DMD, DMPK, DMXL2, DNAJC12, DNAJC19, DNMT1, DNMT1L, 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, GLYCK, 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,			

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, MOC51, MOC52, 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, QRIC1, RAB11A, RAB11B,			

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, RHOTB2, 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, TEO2, 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, TRAP1, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREX1, TRIM8, TRIO, TRIP12, TRIT1, TRMT1, TRMT10A, TRNT1, TRPM3, TRRAP, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TSHB, TSPAN7, TSPAP1, 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, WDRCP, WDR26, WDR37, WDR4, WDR45, WDR45B, WDR62, WDR73, WDR81, WIPI2, WNK3, WNT1, WT1, WWOX, XRCC4, XYL1, YARS1, YIF1B, YIPF5, YWHAE, YWHAG, YY1, ZBTB18, ZBTB20, ZBTB24, ZBTB47, ZBTB7A, ZC4H2, ZDHHC9,			

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		<i>ZEB2, ZFX, ZFYVE26, ZIC1, ZIC2, ZMIZ1, ZMYM2, ZMYM3, ZMYND11, ZMYND8, ZNF142, ZNF292, ZNF335, ZNF462, ZNF526, ZNF699, ZNF711, ZSWIM6</i> Regioner: 1p36, 1q21.1, 1q43q44, 2p21, 2p15p16.1, 2q11.2, 2q13, 2q27.3, 3q24, 3q29, 4p16.3, 5p15, 5q35, 7p22.1, 7q11.23, 8p23.1, 10q22.3q23.2, 11p13, 11p11.2, 14q32.2, 15q11.2, 15q11.13, 15q13.3, 15q24, 15q25.2, 16p13.3, 16p13.11, 16p12.2, 16p11.2, 17p13.3, 17q11.2, 17q12, 17q21.3, 17q23.1q23.2, 22q11.2, Xp11.23, Xp11.22p11.23, Xp11.22, Xq25, Xq28 Screening för patogena repeatexpansioner ingår för följande gener: <i>AFF2, CNBP, DMPK, EIF4A3, FMR1, XYLT1, ZIC2</i>.			
Intestinal pseudo-obstruktion (inkl Hirschsprung)	Perifert blod	Intestinal pseudo-obstruktion panel v1, 36 gener och 1 region, 1 repeatexpansion <i>ACTA2, ACTG2, BDNF, CELSR3, ECE1, EDN3, EDNRB, ERBB3, FLNA, GDNF, GFRA1, KIFBP, L1CAM, LIG3, LMOD1, MIF, MPV17, MYH11, MYL9, MYLK, NRG1, NRTN, PAX3, PHOX2B, POLG, PROK1, PROKR1, PROKR2, RAD21, RET, RMRP, SGO1, SOX10, TTC7A, TYMP, ZEB2</i> Region: 9p21.3 Screening för patogena repeatexpansioner ingår för följande gen: <i>PHOX2B</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Kolestas	Perifert blod	Kolestaspanel v1, 58 gener <i>ABCB11, ABCB4, ABCC2, ADK, AKR1D1, ALDOB, AMACR, ATP7B, ATP8B1, BAAT, BCS1L, CFTR, CLDN1, COG7, CYP27A1, CYP7B1, DCDC2, DGUOK, FAH, GALE, GALM, GALT, GBA1, HADHA, HNF1B, HSD3B7, JAG1, KIF12, LIPA, MPI, MPV17, MVK, MYO5B, NBAS, NOTCH2, NPC1, NPC2, NR1H4, PEX1, PEX12, PEX26, PEX6, PKHD1, POLG, RINT1, SERPINA1, SLC25A13, SMPD1, TALDO1, TJP2, TRMU, UGT1A1, UNC45A, USP53, VIPAS39, VPS33B, YARS1, ZFYVE19</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Kraniosynostos (bl.a. Apert syndrom, Crouzon syndrom, Pfeiffer syndrom, Saethre-Chotzen syndrom)	Perifert blod	Kraniosynostospanel v4, 107 gener <i>ALPL, ACTB*, ACTG1*, ADAMTSL4, ADNP, AHDC1, ALX3, ALX4, ARID1B, ARSB, ASXL1, B3GAT3*, BCL11B, BRAF*, BMP4, CD96, CDC45, CDT1, CDK13, CHD3, CHD7, COLEC11, CTSK, CYP26B1, EFN1, ERF, ESCO2, FAM20C, FBN1, FBXO11, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FREM1, GLI3, GPC3, GNB1, GNPTAB, HNRNP, HUWE1, IDS*, IDUA, IFT122*, IFT140, IFT43, IGF1R, IHH, IL11RA, IRX5, JAG1, KAT6A, KAT6B, KMT5B, KRAS*, LTBP1, MASP1, MEGF8, MSX2*, MAN2B1, MAP3K20, NFIA, NFIX, ORC1, ORC4, ORC6, P4HB, PAX3, PHEX, POR, PPP3CA, PRRX1, PTPN11, POR, RAB23, RECQL4, RNU12, RSPRY1, RUNX2, SCARF2, SEC24D, SIX1, SKI, SLC25A24, SMAD2, SMAD3,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		<i>SMAD6, SMO, SOX6, SPECC1L, TCF12, TCOF1, TFAP2B, TGFB2, TGFB3, TGFB1, TGFB2, TMCO1, TRAF7, TSHR, TWIST1, WDR19, WDR35, ZEB2*, ZIC1, ZNF462</i>			
Kortvuxenhet	Perifert blod	Kortvuxenhetspanel v1, 15 gener <i>ACAN, GH1, GHR, GHRHR, GHSR, IGF1, IGF1R, IGFALS, LHX3, LHX4, NPR2, POU1F1, PROP1, SHOX, SOX3</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Liddle syndrom	Perifert blod	Gitelman, Bartter och Liddle syndrompanel v1, 13 gener <i>AP2S1, BSND, CASR, CLCNKA, CLCNKB, GNA1, KCNJ1, MAGED2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Makrocefali och överväxt (bl.a. Sotos syndrom)	Perifert blod	Makrocefali och överväxtpanel v2, 55 gener <i>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</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Mikrocefali	Perifert blod	Mikrocefali v1, 208 gener, 6 regioner <i>AARS1, ACBD6, ADARB1, AFG2B, ANKLE2, AP4E1, ARF3, ARPC4, ASPM, ATP1A2, ATP6V0A1, ATP6V0C, ATR, ATRX, BLM, BPTF, BRCA2, BRIP1, BUB1, BUB1B, CAMK2B, CAMSAP1, CASK, CCDC88A, CCND2, CDK5RAP2, CDT1, CENPF, CEP135, CEP152, CEP55, CEP57, CHAMP1, CHKA, CIT, CKAP2L, COASY, CPAP, CREBBP, CRNKL1, CSNK2A1, CTCF, CTNNA1, CTU2, DDX11, DHCR7, DIAPH1, DNA2, DNMT3A, DOHH, DONSON, DPM1, DROSHA, DYNC1I2, DYRK1A, EEF1D, EFTUD2, EIF2S3, EIF5A, ERCC4, ERCC6, ERCC8, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FBRSL1, FLVCR1, FOXG1, FRA10AC1, GINS3, GMNN, GPT2, GRM7, GTF2E2, GTF3C3, H4C3, HDAC8, HHAT, HMGB1, HPDL, IARS1, IER3IP1, IGF1, IGF1R, INPP4A, INTS11, KIF11, KIF14, KIFBP, KMT2B, KNL1, LAGE3, LARP7, LIG4, LMNB1, LMNB2, MCPH1, MECP2, METTL5, MFSD2A, MINPP1, MORC2, MRPL49, MSMO1, MYCN, NAPB, NARS1, NBN, NCAPD2, NCAPD3, NDE1,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		<i>NHEJ1, NIPBL, NSD2, NSRP1, NUP107, NUP188, NUP214, ORC1, ORC4, ORC6, OSGEP, PCDH12, PCNT, PDHA1, PLK4, PNKP, PNPLA8, POC1A, POGZ, PPFIBP1, PPIL1, PQBP1, PRIM1, PRUNE1, PTPN23, PUF60, PUS7, RAD21, RAD50, RAD51, RBBP8, RNU4-2, RNU4ATAC, RPL10, RTTN, SARS1, SASS6, SLC1A4, SLC25A19, SLC38A3, SLC4A10, SLC9A6, SLF2, SLX4, SMARCA5, SMC1A, SMC3, SMC5, SMG8, SPOUT1, STAMBP, STIL, SVBP, TMEM167A, TMX2, TNPO2, TOP3A, TP53RK, TRAIP, TRAPPC10, TRAPPC12, TRAPPC6B, TRAPPC9, TRIO, TRMT10A, TSEN15, TSEN54, TTC5, TTI1, TUBG1, TUBGCP2, TUBGCP4, TUBGCP6, UBA5, UFC1, UFM1, UGP2, UNC80, VRK1, WARS1, WDR11, WDR37, WDR4, WDR62, WDR73, WLS, XRCC4, YIPF5, ZEB2, ZNF335, ZNF526</i> Regioner: 2p15p16.1, 5p15, 5q35, 7p22.1, 16p13.3, 17q23.1q23.2			
Monogen diabetes (MODY)	Perifert blod	Monogen diabetes (MODY) v1, 102 gener <i>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</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Meckel syndrom och Joubert syndrom	Perifert blod	Meckel syndrom och Joubert syndrom v1, 38 gener <i>AHI1, ARL13B, ARMC9, B9D1, B9D2, C5ORF42, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, INPP5E, KATNIP, KIAA0586, KIF14, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, TXNDC15, ZNF423</i>	NGS TWIST In silico panel*	90	EDTA
Miller-Dieker syndrom	Perifert blod	17p13.3	MLPA enkel	56	EDTA
Multipel endokrin neoplasi typ 1 (MEN1)	Perifert blod	<i>MEN1</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Multipel endokrin neoplasia typ 2 (MEN2)	Perifert blod	<i>RET</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Myotonia och paramyotonia kongenita panel	Perifert blod	<i>CLCN1, SCN4A</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Neurofibromatos typ1	Perifert blod	<i>NF1, SPRED1</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Neurodegenerativa sjukdomar (bl.a. frontotemporal demens, ALS, Parkinson, prionsjukdom)	Perifert blod	Neurodegenerativa sjukdomar v1, 134 gener och 17 repeatexpansioner <i>ABCD1, AFG3L2, ALS2, ANG, ANXA11, APP, ARSA, ATP13A2, ATP1A3, ATP7B, AUH, C19orf12, CACNA1A, CACNA1G, CCNF, CHCHD10, CHCHD2, CHMP2B, CLCN2, CLN6, COASY, COL4A1, COL4A2, CP, CSF1R, CTSA, CTSF, CYLD, CYP27A1, CYP7B1, DARS2, DCTN1, DNAJB2, DNAJC5, DNAJC6, DNAJC7, DNMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, EPM2A, ERBB4, FBXO7, FIG4, FTL, FUS, FXN, GBA1, GBE1, GCH1, GFAP, GLA, GRN, GSN, HEXA, HEXB, HNRNPA1, HTRA1, ITM2B, JAM2, KCNC3, KCND3, KIF5A, LAMB1, LRRK2, LYST, MAPT, MATR3, MYORG, NAA60, NEK1, NHLRC1, NOTCH3, NPC1, NPC2, OPA3, OPTN, PANK2, PARK7, PDGFB, PDGFRB, PFN1, PINK1, PLA2G6, PLD3, PRKN, PRKRA, PRNP, PRPH, PSAP, PSEN1, PSEN2, RAB32, RAB39B, RFC1, RNF216, SETX, SIGMAR1, SLC20A2, SLC30A10, SNCA, SOD1, SORL1, SPAST, SPG11, SPG21, SPG7, SPR, SQSTM1, SS18L1, STUB1, SYNJ1, TARDBP, TBK1, TMEM240, TREM2, TREX1, TTC19, TTR, TUBA4A, TUBB4A, TYROBP, UBQLN2, VAPB, VCP, VPS13A, VPS35*, VRK1, WDR45, XK, XPR1</i> Screening för patogena repeatexpansioner ingår för följande gener: <i>AR, ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, FXN, HTT, JPH3, NOP56, PPP2R2B, RFC1, TBP</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Neuromuskulära sjukdomar (bl.a. myotoni, myasteni, DMD, limb girdle, paramyotonia)	Perifert blod	Bred NMD panel v1, 1057 gener och 21 repeatexpansioner <i>AAAS, AARS1, AARS2, ABCA1, ABCA2, ABCB7, ABCD1, ABHD12, ABHD5, ACAD9, ACADM, ACADVL, ACO2, ACOX1, ACTA1, ACTL6B, ACTN2, ADA2, ADAR, ADGRG1,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

[illegible]

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, LNP, LPIN1, LRIF1, LRP4, LRPPRC, LRSAM1, LYRM7, LYST, MAB21L1, MAG, MAN2B1, MAP3K20, MAPK8IP3, MARS1, MARS2, MAST1, MATR3, MBD5, MCM3AP, MCOLN1, MECP2, MECP, MED13L, MEGF10, MFN2, MFSB8, 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, MTPP, 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, NDUF51, NDUF52, NDUF53, NDUF54, NDUF56, NDUF57, NDUF58, NDUFV1, NDUFV2, NEB, NEFH, NEFL, NEU1, NEXMIF, NF2, NFASC, NGF, NHLRC1, NIPA1, NKX2-1, NKX2-2, NMNAT1, NOL3, NPC1, NPC2, NPHP1, NPTX1, NR4A2, NTSC2, NTNG2, NTRK1, NUBPL, NUP62, NUS1, OBSCN, OFD1, OGDH, OGDHL, OPA1, OPA3, OPHN1, OPTN, ORA1, 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, SH3TC2, SHMT2, SIGMAR1, SIL1, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC18A3, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2,			

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, UQCQR, 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, årtlig tryckkänslig neuropati, hereditär sensorisk och autonom neuropati, transtyretin-medierad amyloidosis)	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, GALT, 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,	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, 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, 2 regioner 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 Regioner: 15q11.13 (PWS/AS), 16p11.2 (BP2-BP3)	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 genom.	Perifert blod	Optikusatrofipanel v1, 86 gener ACO2, AFG3L2, ALPK1, ATAD3A, ATG7, AUH, C19orf12, CISD2, DNAJC19, DNAJC30, DNMI1L, EPRS1, FDXR, ISCA2, MECP, 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 benskörheth	Perifert blod	Osteogenesis imperfecta och benskörhetspanel v1, 67 gener	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, 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, RSP04, TRPV3, USB1	NGS TruSeq helgenom In silico panel*	90	EDTA
Palmopantar keratodermi och erythrodermi	Perifert blod	Palmopantar keratodermi och erythrodermi 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, RSP01, SASH1, SERPINA12, SERPINB7, SLURP1, SMARCAD1, SNAP29, TAT, TRPV3, WNT10A	NGS TruSeq helgenom In silico panel*	90	EDTA
Pankreatit	Perifert blod	Pankreatitpanel v1, 4 gener CELA3B, CFTR, PRSS1, SPINK1	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, NLR4, NLRP12, NLRP3, NOD2, OTULIN, POMP, PSMB4, PSMB8, PSTPIP1, TNFAIP3, TNFRSF1A, TRNT1	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
<i>POLG</i> -relaterad sjukdom Analysen inkluderar även <i>POLG</i> mutationer associerade med Valproat-inducerad leverskada.	Perifert blod	<i>POLG</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Polycystisk njursjukdom	Perifert blod	Polycystisk njursjukdomspanel v2, 23 gener <i>ALG5, ALG8, ALG9, COL4A1, DNAJB11, DZIP1L, GANAB, HNF1B, IFT140, JAG1, NEK8, NOTCH2*, PKD1*, PKD2, PKHD1, PMM2, PRKCSH, SEC61A1, SEC63, TSC1, TSC2, UMOD, VHL</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Prader-Willi syndrom	Perifert blod	15q11.2	MLPA metylering	56	EDTA
Primär ciliär dyskinesi (PCD) (Respiratorisk ciliopati, bl.a. cystisk fibros och bronkiektas)	Perifert blod	PCD panel v2, 44 gener <i>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</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Primär hyperaldosteronism	Perifert blod	Primär hyperaldosteronismspanel v1, 5 gener <i>CACNA1H, CLCN2, CYP11B1, CYP11B2, KCNJ5</i>	NGS TruSeq helgenom In silico panel	90	EDTA
Pulmonell arteriell hypertension (PAH)	Perifert blod	Pulmonell arteriell hypertensionspanel v1, 17 gener <i>ABCC8, ACVRL1, AQP1, ATP13A3, BMPR2, CAPNS1, CAV1, EIF2AK4, ENG, GDF2, KCNK3, KDR, NFU1, SARS2, SMAD9, SOX17, TBX4</i>	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 <i>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</i>	NGS TWIST In silico panel* NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Retinal degeneration	Perifert blod	Retinal degenerationpanel v1, 365 gener <i>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-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, MTPP, 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</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Schwannomatos och meningiom	Perifert blod	Schwannomatos och meningiompanel v1, 10 gener <i>LZTR1, MEN1, NF2, PRKAR1A, PTCH1, PTEN, SMARCB1, SMARCE1, SUFU, WRN</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
<i>SHOX</i> -relaterad kortvuxenhet	Perifert blod	<i>SHOX</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Silver-Russel syndrom	Perifert blod	11p15	MLPA metylering	56	EDTA
Skelettdysplasi	Perifert blod	Skelettdysplasipanel v1, 402 gener <i>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, CEP290, CFAP410, CHST14, CHST3, CHSY1, CLIK1, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRIPT, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTNS, 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, FAH, 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,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		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, RPRIP1L, 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, SLC02A1, SMAD3, SMAD4, SMARCA11, 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, TGFBI, TGFBI2, 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 Intellectuell funktionsnedsättning	Perifert blod	Screening	Mikroarray	56	EDTA
Anlagsbärande utredning för Syndromutredning Utvecklingsförsening/Autism Intellectuell funktionsnedsättning	Perifert blod	Screening	Mikroarray Riktad	56	EDTA
Transtyretinmedierad amyloidosis	Perifert blod	TTR	NGS TWIST In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Tanatofor dysplasi (Se även <i>FGFR3</i> -relaterad skelettdysplasi)	Perifert blod	<i>FGFR3</i> exon 7, 9, 14, 18	Sangersekvensering <i>FGFR3</i>	56	EDTA
Trombocytopeni	Perifert blod	Trombocytopenipanel v2, 127 gener <i>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, PLA1, 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, TBXA51, TERT, THPO, TINF2, TSR2, TUBB1, UBE2T, VWF, WAS, WIPF1, WRAP53, XRCC2, ZCCHC8</i>	NGS TruSeq helgenom In silico panel*	90	EDTA
Tuberös skleros	Perifert blod	<i>TSC1, TSC2</i>	NGS TWIST In silico panel*	90	EDTA
Tubulopati (inkl. renala tubulära njursjukdomar, elektrolytrubbningar (natrium, kalium, magnesium, fosfat, calcium, acidos och alkalos), aldosteronavvikelser, nefrogen och neurohypofysär diabetes insipidus, njurstensjukdomar, nefrokalcinos och metabola njursjukdomar)	Perifert blod	Tubulopatipanel v1, 112 gener <i>AGXT, AIRE, ALPL, AMMECR1, AP2S1, APRT, AQP2, ATP1A1, ATP6V0A4, ATP6V1B1, ATP7B, AVP, AVPR2, BSND, CA2, CACNA1D, CACNA1H, CACNA1S, CASR, CDC73, CDKN1B, CLCN2, CLCN5, 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.</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
Tyroideahormonresistens (inkl. hypotyroidism)	Perifert blod	Tyroideahormonresistenspanel v2, 35 gener <i>CASR, CDCA8, DUOX2, DUOX2A, 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</i>	NGS TruSeq helgenom In silico panel	90	EDTA
Hippel-Lindau (VHL)	Perifert blod	<i>VHL</i>	NGS TruSeq helgenom In silico panel	90	EDTA
Welanders distala myopati	Perifert blod	<i>TIA1</i> exon 13	Sangersekvens ering <i>TIA1</i>	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 <i>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, 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, RFWO3, RIT1, RPA1, RPL11, RPL15, RPL18, RPL23, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL36, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTE1, RUNX1, SAMD9, SAMD9L, SBDS, SDHB, SDHC, SDHD, SEC23B, SH2D1A, SHOC2, SLFN14, SLX4, SOS1, SRC, SRP54, SRP72, STIM1, STN1, TBXA2R, TBXA51, TCIRG1, TERC, TERT, THBD, THPO, TINF2,</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

Frågeställning/ Analys	Vävnad	Kromosomområde / Gen	Analys/Metod	Svarstid (dagar)	Rör
		<i>TNFRSF13B, TP53, TSR2, TUBB1, TYK2, UBE2T, UROS, USB1, VHL, VPS13B, VPS45, VWF, WAS, WIPF1, WRAP53, XRCC2, ZCCHC8.</i>			
Ärftlig njursjukdom (Inkl. tubulopatipanelen och polycystisk njursjukdomspanelen samt proteinuri, hematuri (inkl Alports syndrom), FSGS, nefronoftis, autosomalt dominant tubulointerstitiell njursjukdom (exklusive MUC1-ADTKD), aHUS och monogen amyloidosis. Inkl. ej orsaker till njurmissbildningar (CAKUT) eller monogena autoimmuna/autoinflammatoriska sjukdomar)	Perifert blod	Ärftlig njursjukdomspanel v1, 290 gener <i>ACE, ACTN4*, ADA2, AGT, AGTR1, AGXT, AHI1, AIRE, ALG5, ALG8, ALG9, ALMS1*, ALPL, AMMECR1, AMN, ANKS6, ANLN, AP2S1, APOA1, APOA4, APOC2, APOE, APRT, AQP2, ARHGDI1, 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, DDCDC2, 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, RRGD, RRM2B, SARS2, SCARB2, SCLT1, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SGPL1, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC5A2, SLC6A19, SLC7A7, SLC7A9, SMARCA1, STX16, TBC1D8B, TBCE, TCTN1, TCTN2, TCTN3, TMEM138, TMEM139, TMEM231, TMEM237, TMEM67, TNS2, TP53RK, TPRKB, TRAF3IP1, TRIM8, TRPC6, TRPM6, TRPM7, TSC1, TSC2, TSEN2, TTC21B, TTC8, TTR, TULP3, TXNDC15, UMOD, VDR, VHL, VIPAS39, VPS33B, WPCP, WDR19, WDR35, WDR72, WDR73, WNK1, WNK4, WT1, XDH, XPNPEP3, XPO5, YRDC.</i>	NGS TruSeq helgenom In silico panel*	90	EDTA

* 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.