

# 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<br>QF-PCR prenatal inkl. MCC | 7                                   | Odlingsmediumrör |
| Riktad analys                         | Amnion            | 13,18,21,X,Y                        | QF-PCR<br>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/<br>Analys                  | Vävnad        | Kromosomområde<br>/ Gen             | Analys/Metod                        | Svarstid<br>(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<br>EDTA |
| Kromosomanalys<br>(Syndromutredning)       | Perifert blod | Screening                           | Kromosomanalys Blod                 | 42                                                                                             | Heparin         |
| Kromosomanalys<br>(Könskromosomutredning)  | Perifert blod | Screening                           | Kromosomanalys Blod                 | 42                                                                                             | Heparin         |
| Kromosomanalys<br>(Infertilitetsutredning) | Perifert blod | Screening                           | Kromosomanalys Blod                 | 42                                                                                             | Heparin         |
| Kromosomanalys (Ägg-<br>el.spermadonation) | Perifert blod | Screening                           | Kromosomanalys Blod                 | 42                                                                                             | Heparin         |
| Kromosomanalys<br>(Upprepade missfall)     | Perifert blod | Screening                           | Kromosomanalys Blod                 | 42                                                                                             | Heparin         |
| FISH-analys<br>(Translokationsutredning)   | Perifert blod | Riktad region                       | FISH Konstitutionellt<br>metafas    | 90#<br>(# vid beställning av<br>unika prober TAT 130<br>dagar)                                 | Heparin         |

| Frågeställning/<br>Analys | Vävnad        | Kromosomområde<br>/ Gen | Analys/Metod                      | Svarstid<br>(dagar) | Rör     |
|---------------------------|---------------|-------------------------|-----------------------------------|---------------------|---------|
| Riktad analys             | Perifert blod | 13,18,21,X,Y            | FISH Konstitutionellt<br>interfas | 7                   | Heparin |

## DNA-baserad diagnostik

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

| Frågeställning/<br>Analys                                                                                                                                                                         | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------|---------------------|------|
| Arytmi och kardiomyopati<br>(inkl. ARVC, Brugada syndrom,<br>LQTS, CPVT, kardiomyopati,<br>HCM, DCM)                                                                                              | Perifert blod | Arytmi och kardiomyopati-panel v3, 187 gener och 1 repeatexpansion<br><br>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*, SLC6, 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<br><br>Screening för patogena repeatexpansioner ingår för följande gen: DMPK | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Ataxi<br>(inkl. CANVAS, episodisk ataxi,<br>FXTAS, spastisk ataxi och<br>spinocerebellär ataxi samt<br>repeatanalys för ataxisjukdomar)<br>Panelen inkluderar även det<br>mitokondriella genomet. | Perifert blod | Ataxi-panel v5, 755 gener och 18 repeatexpansioner<br><br>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, BTBD, 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<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys | Vävnad | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | Analys/Metod | Svarstid<br>(dagar) | Rör |
|---------------------------|--------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------|---------------------|-----|
|                           |        | <p>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, DNMT1, DNMT2, DNMT3, 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, HNRNP2, 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, MTPP, 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,</p> |              |                     |     |

| Frågeställning/<br>Analys    | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Analys/Metod                         | Svarstid<br>(dagar) | Rör  |
|------------------------------|---------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------|---------------------|------|
|                              |               | <p>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</p> <p>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</p> |                                      |                     |      |
| Bartter och Gitelman syndrom | Perifert blod | <p>Gitelman, Bartter och Liddle syndrom panel v1, 13 gener</p> <p>AP2S1, BSND, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, MAGED2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | NGS TruSeq helgenom In silico panel* | 90                  | EDTA |
| Beckwith-Wiedemann syndrom   | Perifert blod | 11p15                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | MLPA metylering                      | 56                  | EDTA |
| Bindvävssjukdomar            | Perifert blod | <p>Bindvävspanel v3, 67 gener</p> <p>ABL1, ACTA2, ADAMTS2, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ASPH, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL12A1, COL1A1, COL1A2,</p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | NGS TruSeq helgenom In silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                                                                                      | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | Analys/Metod                         | Svarstid<br>(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<br><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><br>(Se även cerebrala småkärlssjukdomar)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | NGS TruSeq helgenom In silico panel* | 90                  | EDTA |
| Cerebrala småkärlssjukdomar<br>(bl.a. Moyamoya, Hereditär hemorragisk telangiectasi, Capillary malformation-arteriovenous malformation (CM-AVM) syndrom, Parkes-Weber syndrom) | Perifert blod | Cerebrala småkärlssjukdomspanel v3, 50 gener<br><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><br>(ingår även i Neuropatipanel)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | MLPA enkel                           | 56                  | EDTA |
| Ciliopati<br>(Neurologiska, oftalmologiska, renala och skelettciliopatier, dextrokardi, heterotaxi)                                                                            | Perifert blod | Ciliopatipanel v1, 124 gener<br><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/<br>Analys                | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | Analys/Metod                               | Svarstid<br>(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<br>Konstitutionellt<br>metafas        | 14                  | Heparin |
| Corneal dystrofi                         | Perifert blod | Corneal dystrofi v1, 48 gener<br><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<br>helgenom In<br>silico panel* | 90                  | EDTA    |
| Cystisk Fibros                           | Perifert blod | <i>CFTR</i> (50 mutationer)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Fragmentanalys<br>CFTR                     | 35                  | EDTA    |
| Cystisk Fibros                           | Perifert blod | <i>CFTR</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | NGS TWIST In<br>silico panel*              | 90                  | EDTA    |
| DSD<br>(Disorders of sex<br>development) | Perifert blod | DSD Panel v1, 192 gener och 2 regioner<br><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<br>helgenom In<br>silico panel* | 90                  | EDTA    |

| Frågeställning/<br>Analys     | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Analys/Metod                               | Svarstid<br>(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<br>Region: 11p13 deletionssyndromet (WAGR) och Xp21.2 duplikationssyndromet                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |                                            |                     |      |
| Duchenne, Becker<br>(DMD/BMD) | Perifert blod | DMD                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | MLPA dubbel                                | 56                  | EDTA |
| Duchenne, Becker<br>(DMD/BMD) | Perifert blod | DMD<br>(se även Neuromuskulär panel)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Sangersekvens<br>ering riktad              | 28                  | EDTA |
| Dystoni                       | Perifert blod | Dystonipanel v2, 200 gener och 9 repeatexpansioner<br>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<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                 | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
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|                                                                                                           |               | 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<br>DMPK                     | 35                  | EDTA |
| Ektodermal dysplasi                                                                                       | Perifert blod | Ektodermal dysplasipanel v1, 111 gener<br><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<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Epidermolysis bullosa (EB)                                                                                | Perifert blod | Epidermolysis bullosa panel v2, 31 gener<br><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<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Epilepsi<br>Analysen inkluderar även POLG<br>mutationer associerade med<br>Valproat-inducerad leverskada. | Perifert blod | Epilepsipanel v2, 637 gener, 14 regioner och 3<br>repeatexpansioner<br><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, CHRN2, CIC, CLCN3, CLCN4, CLDN5, CLN3,</i> | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys | Vävnad | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | Analys/Metod | Svarstid<br>(dagar) | Rör |
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|                           |        | <p>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,</p> |              |                     |     |

| Frågeställning/<br>Analys               | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Analys/Metod                         | Svarstid<br>(dagar) | Rör  |
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|                                         |               | <p><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></p> <p>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</p> <p>Screening för patogena repeatexpansioner ingår för följande gener: <i>ARX, ATN1, CSTB</i></p> |                                      |                     |      |
| Erythrocytos                            | Perifert blod | <p>Erythrocytospanel v1, 7 gener</p> <p><i>BPGM, EGLN1, EPAS, EPO, EPOR, JAK2, VHL</i></p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | NGS TruSeq helgenom In silico panel* | 90                  | EDTA |
| Familjär hyperkolesterolemi (FH)        | Perifert blod | <p>Familjär hyperkolesterolemi v2, 7 gener</p> <p><i>ABCG5, ABCG8, APOB, APOE, LDLR, LDLRAP1, PCSK9</i></p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | 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/<br>Analys                                                                                                                                                                         | Vävnad          | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | Analys/Metod                                                                                     | Svarstid<br>(dagar) | Rör  |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------|---------------------|------|
| Helexomsekvensering                                                                                                                                                                               | Perifierat blod | Screening                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | NGS TWIST<br>Exom Trio<br>NGS TWIST<br>Exom Duo<br>NGS TWIST<br>Exom Singleton                   | 90                  | EDTA |
| Helgenomsekvensering<br>Inkl. mitokondriella genomet<br>samt relevanta<br>repeatexpansionssjukdomar                                                                                               | Perifierat blod | Screening                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | NGS TruSeq<br>Helgenom Trio<br>NGS TruSeq<br>Helgenom Duo<br>NGS TruSeq<br>Helgenom<br>Singleton | 90                  | EDTA |
| Hereditär hemorragisk<br>telangiectasi (HHT; Osler-<br>Weber-Rendus syndrom)<br>(bl.a. HHT, Capillary<br>malformation-arteriovenous<br>malformation (CM-AVM)<br>syndrom, Parkes-Weber<br>syndrom) | Periferat blod  | Hereditär hemorragisk telangiectasipanel v1, 6 gener<br><i>ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | NGS TruSeq<br>helgenom In<br>silico panel*                                                       | 90                  | EDTA |
| Hereditär spastisk paraplegi<br>(HSP)                                                                                                                                                             | Periferat blod  | Hereditär spastisk paraplegi panel v2, 115 gener, 1<br>region och 9 repeatexpansioner<br><i>ABCD1, ABHD16A, ACER3, ADAR, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALS2, AMFR,<br/>AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATL1, ATP13A2, B4GALNT1,<br/>BCAS3, BSCL2, C12orf65, C19orf12, CAPN1, CLDN11, COQ4, CPT1C, CTNNB1, CYP27A1,<br/>CYP2U1, CYP7B1, DARS, DDHD1, DDHD2, DDX3X, ELOVL1, ENTPD1, ERLIN1, ERLIN2,<br/>FA2H, FAR1, FARS2, FBXO7, FXN, GALT, GBA2, GBE1, GCH1, GJA1, GLRX5, GPT2,<br/>HACE1, HECTD4, HIKESHI, HPDL, HSPD1, IFIH1, KCNA2, KDM5C, KIDINS220, KIF1A,</i> | NGS TruSeq<br>helgenom In<br>silico panel*                                                       | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                   | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
|-------------------------------------------------------------------------------------------------------------|---------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------|---------------------|------|
|                                                                                                             |               | <p><i>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, SLC25A46, SLC2A1, SPART, SPAST, SPATA5L1, SPG11, SPG21, SPG7, SPTAN1, STN1, TAF8, TECPR2, TFG, TMEM63C, TUBB4A, UBAP1, UCHL1, WASHC5, WDR45B, ZFYVE26</i></p> <p>Region: Xq28 regionen (inklusive MECP2)<br/>Screening för patogena repeatexpansioner ingår för följande gener: <i>ATXN1, ATXN2, ATXN3, ATXN7, ATXN10, CACNA1A, FXN, PPP2R2B, TBP</i></p> |                                            |                     |      |
| HNPP, fam. tryckförämning                                                                                   | Perifert blod | <i>PMP22</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | MLPA enkel                                 | 56                  | EDTA |
| Huntingtons sjukdom                                                                                         | Perifert blod | <i>HTT</i> (Huntingtin)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | Fragmentanalys<br>HTT (HD)                 | 35                  | EDTA |
| Hyperinsulinism och hypoglykemi                                                                             | Perifert blod | <p>Hyperinsulinism och hypoglykemi v1, 47 gener</p> <p><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></p>                                                                                                                                                                                                                                      | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Hyperlipidemi                                                                                               | Perifert blod | <p>Hyperlipidemipanel v1, 18 gener</p> <p><i>ABCA1, ABCG5, ABCG8, APOA1, APOA5, APOB, APOC2, APOE, CREB3L3, CYP27A1, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LMF1, LPL, PCSK9</i></p>                                                                                                                                                                                                                                                                                                                                                                                                                                          | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Hyperparatyreoidism<br>(inkl Familjär hypokalciurisk, hyperkalcemi (FHH), och Multipel endokrin neoplasia)) | Perifert blod | <p>Hyperparatyreoidismpanel v2, 8 gener</p> <p><i>AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET</i></p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Hypofystumör, ärftlig                                                                                       | Perifert blod | <p>Ärftlig hypofystumör v1, 6 gener</p> <p><i>AIP, CDKN1B, DICER1, MEN1, NF1, PRKAR1A</i></p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                                                                                   | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------|---------------------|------|
| Hypogonadotrop<br>hypogonadism<br>(inkl. Kallman syndrom)                                                                                                                   | Perifert blod | Hypogonadotrop hypogonadismpanel v1, 31 gener<br><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<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Hypoparatyreoidism<br>(inkl. Albright hereditary<br>osteodystrophy,<br>pseudohypoparathyroidism,<br>pseudopseudohypoparathyroidism,<br>acrodysostosis and osteoma<br>cutis) | Perifert blod | Hypoparatyreoidismpanel v1, 11 gener<br><i>AIRE, CASR, GATA3, GCM2, GNA11, GNAS, PDE4D, PRKAR1A, PTH, STX16, TBCE</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Hypokondroplasi<br>(Se även <i>FGFR3</i> -relaterad<br>skelettdysplasi)                                                                                                     | Perifert blod | <i>FGFR3</i> (exon 9, 12)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | Sangersekvens<br>ering <i>FGFR3</i>        | 56                  | EDTA |
| Hörselnedsättning                                                                                                                                                           | Perifert blod | Hörselnedsättning panel v1, 289 gener<br><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, 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, GPM2, 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,</i> | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                                                                                         | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              | Analys/Metod                                     | Svarstid<br>(dagar) | Rör  |
|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------|---------------------|------|
|                                                                                                                                                                                   |               | 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 |                                                  |                     |      |
| Hörselnedsättning                                                                                                                                                                 | Perifert blod | GJB2 (exon 2), GJB6<br>(inkl. deletion/duplikationsanalys)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        | Paket:<br>Sangersekvensering GJB2/<br>MLPA enkel | 56                  | EDTA |
| Iktyos, könsbunden                                                                                                                                                                | Perifert blod | STS                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | MLPA enkel                                       | 56                  | EDTA |
| Iktyos, NGS panel<br>(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<br>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                                                                                                                                                                                                                                                                                                                             | NGS TruSeq<br>helgenom In<br>silico panel*       | 90                  | EDTA |
| Immunologisk sjukdom<br>(bl.a immunbrist, inflammatorisk tarmsjukdom, autoinflammatorisk sjukdom, kronisk granulomatös sjukdom, periodisk feber, SCID, kongenital                 | Perifert blod | Immunpanel v1, 355 gener<br>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,                                                                                                                                                                                                                                                                                                                                                                                                                                                      | NGS TruSeq<br>helgenom In<br>silico panel*       | 90                  | EDTA |

| Frågeställning/<br>Analys                  | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | Analys/Metod                      | Svarstid<br>(dagar) | Rör  |
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| neutropeni, och<br>autoimmunitetssyndrom)  |               | CFI, CFP, CFTR, CHD7, CIITA, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTLA4, CTNBNB1, 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, FXP3, 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, MPEP1, MRTFA, MSN, MTHFD1, MVK, MYD88, MYO5B, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NEUROG3, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLR4, 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, SLC20A1, SMARCA11, 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, TMPSR15, 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<br>Fragilt-X       | 35                  | EDTA |
| Infertilitetsutredning,<br>CBAVD           | Perifert blod | CFTR (50 mutationer)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      | Fragmentanalys<br>CFTR            | 35                  | EDTA |
| Infertilitetsutredning,<br>Mikrodeletion Y | Perifert blod | AZF                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       | Fragmentanalys<br>Mikrodeletion Y | 35                  | EDTA |

| Frågeställning/<br>Analys             | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
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| Intellectuell<br>funktionsnedsättning | Perifert blod | <p>Intellectuell funktionsnedsättning panel v1, 1453 gener, 40 regioner och 7 repeatexpansioner</p> <p>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, ATP6VOC, 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, BTB, 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, CCB1, CCDC22, CCDC32, CCDC47, CCDC82, CCDC88C, CCND2, CDC42, CDC6, CDH11, CDH2, CDK10, CDK13, CDK16, CDK19, CDK5RAP2, CDK8, CDKL5, CDON, CECR2, CELF2, CENPF, CENPI, 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, CTDSP1, CTNNA2, CTNNB1, CTNND1, CTNND2, CTR9, CTSB, 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,</p> | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys | Vävnad | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Analys/Metod | Svarstid<br>(dagar) | Rör |
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|                           |        | <p>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, FKR1, 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, GALT, GALE, GALNT2, GALT, GAMT, GATA4, GATAD2B, GATM, GCDH, GCH1, GCSH, GDI1, GEMIN5, GFAP, GFER, GFM1, GIA5, 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, 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, MSMD1,</p> |              |                     |     |

| Frågeställning/<br>Analys | Vävnad | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        | Analys/Metod | Svarstid<br>(dagar) | Rör |
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|                           |        | <p>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, PDS1, PDS2, 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, 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,</p> |              |                     |     |

| Frågeställning/<br>Analys                           | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
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|                                                     |               | <p>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, TMC01, TMEM106B, TMEM127, TMEM147, TMEM165, TMEM216, TMEM222, TMEM237, TMEM240, TMEM63B, TMEM63C, TMEM67, TMEM70, TMEM94, TMTC3, TMX2, TNPO2, TNRC6B, TOE1, TOR1A, TP73, TPP1, TPP2, TRA2B, TRAF7, TRAI, 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, UBT, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC80, UPF3B, UROC1, USP7, USP9X, VAMP2, VARS1, VARS2, VCP, VLDLR, VPS11, VPS13B, VPS41, VPS4A, VPS53, VRK1, WAC, WARAS2, WASF1, WDFY3, WDP, 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, ZFH4, ZFX, ZFYVE26, ZIC1, ZIC2, ZMIZ1, ZMYM2, ZMYM3, ZMYND11, ZMYND8, ZNF142, ZNF292, ZNF335, ZNF462, ZNF526, ZNF699, ZNF711, ZSWIM6</p> <p>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</p> <p>Screening för patogena repeatexpansioner ingår för följande gener: <i>AFF2, CNBP, DMPK, EIF4A3, FMR1, XYLT1, ZIC2</i>.</p> |                                            |                     |      |
| Intestinal pseudo-obstruktion<br>(inkl Hirschprung) | Perifert blod | <p>Intestinal pseudo-obstruktion panel v1, 36 gener och 1 region, 1 repeatexpansion</p> <p><i>ACTA2, ACTG2, BDNF, CELSR3, ECE1, EDN3, EDNRB, ERBB3, FLNA, GDNF, GFRA1, KIFBP, L1CAM, LIG3, LMOD1, MITF, MPV17, MYH11, MYL9, MYLK, NRG1, NRTN, PAX3,</i></p>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                           | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
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|                                                                                                     |               | PHOX2B, POLG, PROK1, PROKR1, PROKR2, RAD21, RET, RMRP, SGO1, SOX10, TTC7A, TYMP, ZEB2<br>Region: 9p21.3<br>Screening för patogena repeatexpansioner ingår för följande gen: PHOX2B                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |                                            |                     |      |
| Kolestas                                                                                            | Perifert blod | Kolestaspanel v1, 58 gener<br>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                                                                                                                                                                                                                                                                                                                                                                    | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Kraniosynostos<br>(bl.a. Apert syndrom, Crouzon syndrom, Pfeiffer syndrom, Saethre-Chotzen syndrom) | Perifert blod | Kraniosynostospanel v4, 107 gener<br>ALPL, ACTB*, ACTG1*, ADAMTSL4, ADNP, AHDC1, ALX3, ALX4, ARID1B, ARSB, ASXL1, B3GAT3*, BCL11B, BRAF*, BMP4, CD96, CDC45, CDT1, CDK13, CHD3, CHD7, COLEC11, CTSK, CYP26B1, EFN1B, ERF, ESCO2, FAM20C, FBN1, FBXO11, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FREM1, GLI3, GPC3, GNB1, GNPTAB, HNRNPK, 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, SMAD6, SMO, SOX6, SPECC1L, TCF12, TCOF1, TFAP2B, TGFB2, TGFB3, TGFB3R1, TGFB3R2, TMCO1, TRAF7, TSHR, TWIST1, WDR19, WDR35, ZEB2*, ZIC1, ZNF462 | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Kortvuxenhet                                                                                        | Perifert blod | Kortvuxenhetspanel v1, 15 gener<br>ACAN, GH1, GHR, GHRHR, GHSR, IGF1, IGF1R, IGFALS, LHX3, LHX4, NPR2, POU1F1, PROP1, SHOX, SOX3                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Liddle syndrom                                                                                      | Perifert blod | Gitelman, Bartter och Liddle syndrompanel v1, 13 gener<br>AP2S1, BSND, CASR, CLCNKA, CLCNKB, GNA1, KCNJ1, MAGED2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A3                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                         | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
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| Makrocefali och överväxt<br>(bl.a. Sotos syndrom) | Perifert blod | Makrocefali och överväxtpanel v2, 55 gener<br><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<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Mikrocefali                                       | Perifert blod | Mikrocefali v1, 208 gener, 6 regioner<br><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, CTNBN1, 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, 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, STAMPB, 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> | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                 | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | Analys/Metod                         | Svarstid<br>(dagar) | Rör  |
|-------------------------------------------|---------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------|---------------------|------|
|                                           |               | Regioner: 2p15p16.1, 5p15, 5q35, 7p22.1, 16p13.3, 17q23.1q23.2                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       |                                      |                     |      |
| Monogen diabetes (MODY)                   | Perifert blod | Monogen diabetes (MODY) v1, 102 gener<br><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<br><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 |
| Multipel endokrin neoplasi 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 |

| Frågeställning/<br>Analys                                                                                                                                                                                        | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------|---------------------|------|
| Neurofibromatos typ1                                                                                                                                                                                             | Perifert blod | <i>NF1, SPRED1</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Neurodegenerativa<br>sjukdomar<br>(bl.a. frontotemporal demens,<br>ALS, Parkinson, prionsjukdom)                                                                                                                 | Perifert blod | Neurodegenerativa sjukdomar v1, 134 gener och 17<br>repeatexpansioner<br><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><br><br>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<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Neuromuskulära<br>sjukdomar<br>(bl.a. myotoni, myasteni, DMD,<br>limb girdle, paramyotonia<br>kongenita, samt generna i<br>ataxipanel)<br><br>Panelen inkluderar även det<br>mitokondriella genomet samt<br>SMA. | Perifert blod | Bred NMD panel v1, 1057 gener och 21<br>repeatexpansioner<br><i>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, BTBD, BVES, C19ORF12, CA8, CACNA1A, CACNA1G, CACNA1S, CACNA2D2, CACNB4, CAMTA1, CAPN1, CAPN3, CARS1, CASK,</i>                                                                                                                                                                                                                                                                                                                                    | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

[illegible]

| Frågeställning/<br>Analys | Vävnad | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | Analys/Metod | Svarstid<br>(dagar) | Rör |
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|                           |        | <p>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, MTTT, 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, NKX6-2, NMNAT1, NOL3, NPC1, NPC2, NPHP1, NPTX1, NR4A2, NTSC2, 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, RTCL1, 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, TAMMM41, TANC2, TANGO2, TARDBP, TBC1D23, TBC1D24, TBCE, TBK1, TCAP, TCF20, TCF4, TCN2, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TELO2,</p> |              |                     |     |

| Frågeställning/<br>Analys                                                                                                                                                 | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   | Analys/Metod                         | Svarstid<br>(dagar) | Rör  |
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|                                                                                                                                                                           |               | <p>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</p> <p>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</p>                                                                                                                                                                                                                                                                                                                                                                                                    |                                      |                     |      |
| <p>Neuropatipanel<br/>(bl.a. Charcot-Marie-Tooth sjukdom, ärftlig tryckkänslig neuropati, hereditär sensorisk och autonom neuropati, transtyretin-medierad amyloidos)</p> | Perifert blod | <p>Neuropatipanel v2, 205 gener</p> <p>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, CTDPI, 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, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MITTP, 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</p> | NGS TruSeq helgenom In silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                               | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
|-------------------------------------------------------------------------|---------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------|---------------------|------|
| Obesitas                                                                | Perifert blod | Obesitaspanel v2, 54 gener, 2 regioner<br><i>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, NROB2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, RAI1, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP</i><br>Regioner: 15q11.13 (PWS/AS), 16p11.2 (BP2-BP3)                                                                                                                                                                                                                             | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Okulär albinism                                                         | Perifert blod | Okulär albinismpanel v1, 32 gener<br><i>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</i>                                                                                                                                                                                                                                                                                                                                                                                                                                  | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Optikusatrofi<br>Panelen inkluderar även det<br>mitokondriella genomet. | Perifert blod | Optikusatrofipanel v1, 86 gener<br><i>ACO2, AFG3L2, ALPK1, ATAD3A, ATG7, AUH, C19orf12, CISD2, DNAJC19, DNAJC30, DNMT1L, 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</i> | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Osteogenesis imperfecta<br>och benskörhet                               | Perifert blod | Osteogenesis imperfecta och benskörhetspanel v1, 67 gener<br><i>ALPL, ANOS, 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</i>                                                                                                                                    | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                                              | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                | Analys/Metod                                | Svarstid<br>(dagar) | Rör  |
|----------------------------------------------------------------------------------------------------------------------------------------|---------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------------------|---------------------|------|
| Pachyonychia congenita                                                                                                                 |               | Pachyonychia congenita v1, 19 gener<br><i>AAGAB, CARD9, COL7A1, CTSC, DSG1, FZD6, GJB6, JUP, KRT16*, KRT17*, KRT5, KRT6A*, KRT6B*, KRT6C*, MBTPS2, PLCD1, RSPO4, TRPV3, USB1</i>                                                                                                                                                                                    | NGS TruSeq<br>helgenom In<br>silico panel*  | 90                  | EDTA |
| Palmopplantar keratodermi<br>och erythrodermi                                                                                          | Perifert blod | Palmopplantar keratodermi och erythrodermi v1, 42 gener<br><i>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</i> | NGS TruSeq<br>helgenom In<br>silico panel*  | 90                  | EDTA |
| Pankreatit                                                                                                                             | Perifert blod | Pankreatitpanel v1, 4 gener<br><i>CELA3B, CFTR, PRSS1, SPINK1</i>                                                                                                                                                                                                                                                                                                   | NGS TruSeq<br>helgenom In<br>silico panel*  | 90                  | EDTA |
| Paragangliom och<br>feokromocytom                                                                                                      | Perifert blod | Paragangliom och feokromocytom panel v2, 17 gener<br><i>DLST, EGLN1, EPAS1, FH, MAX, MDH2, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL</i>                                                                                                                                                                                               | NGS TruSeq<br>helgenom In<br>silico panel** | 90                  | EDTA |
| Periodisk paralyspanel                                                                                                                 | Perifert blod | Periodisk paralyspanel v 1, 4 gener<br><i>CACNA1S, CLCN1, KCNJ2, SCN4A</i>                                                                                                                                                                                                                                                                                          | NGS TruSeq<br>helgenom In<br>silico panel*  | 90                  | EDTA |
| Periodiskt febersyndrom                                                                                                                | Perifert blod | Periodisk febersyndrompanel v2, 22 gener<br><i>ADA2, APOC2, B2M, ELANE, HTR1A, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, OTULIN, POMP, PSMB4, PSMB8, PSTPIP1, TNFAIP3, TNFRSF1A, TRNT1</i>                                                                                                                                                       | NGS TruSeq<br>helgenom In<br>silico panel*  | 90                  | EDTA |
| <i>POLG</i> -relaterad sjukdom<br>Analysen inkluderar även <i>POLG</i><br>mutationer associerade med<br>Valproat-inducerad leverskada. | Perifert blod | <i>POLG</i>                                                                                                                                                                                                                                                                                                                                                         | NGS TruSeq<br>helgenom In<br>silico panel*  | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                              | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                  | Analys/Metod                                                                | Svarstid<br>(dagar) | Rör  |
|--------------------------------------------------------------------------------------------------------|---------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------|---------------------|------|
| Polycystisk njursjukdom                                                                                | Perifert blod | Polycystisk njursjukdomspanel v2, 23 gener<br><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<br>helgenom In<br>silico panel*                                  | 90                  | EDTA |
| Prader-Willi syndrom                                                                                   | Perifert blod | 15q11.2                                                                                                                                                                                                                                                                                                                                                               | MLPA<br>metylering                                                          | 56                  | EDTA |
| Primär ciliär dyskinesi<br>(PCD)<br>(Respiratorisk ciliopati, bl.a. cystisk<br>fibros och bronkiektas) | Perifert blod | PCD panel v2, 44 gener<br><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<br>helgenom In<br>silico panel*                                  | 90                  | EDTA |
| Primär<br>hyperaldosteronism                                                                           | Perifert blod | Primär hyperaldosteronismspanel v1, 5 gener<br><i>CACNA1H, CLCN2, CYP11B1, CYP11B2, KCNJ5</i>                                                                                                                                                                                                                                                                         | NGS TruSeq<br>helgenom In<br>silico panel                                   | 90                  | EDTA |
| Pulmonell arteriell<br>hypertension (PAH)                                                              | Perifert blod | Pulmonell arteriell hypertensionspanel v1, 17 gener<br><i>ABCC8, ACVRL1, AQP1, ATP13A3, BMPR2, CAPNS1, CAV1, EIF2AK4, ENG, GDF2, KCNK3, KDR, NFU1, SRSF2, SMAD9, SOX17, TBX4</i>                                                                                                                                                                                      | NGS TruSeq<br>helgenom In<br>silico panel                                   | 90                  | EDTA |
| RASopatier<br>(bl.a. CFC, Costello syndrom, Noonan<br>syndrom, Legius syndrom, NF1)                    | Perifert blod | RASopatipanel v3, 30 gener<br><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<br>silico panel*<br>NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Retinal degeneration                                                                                   | Perifert blod | Retinal degenerationspanel v1, 365 gener<br><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,</i>                  | NGS TruSeq<br>helgenom In<br>silico panel*                                  | 90                  | EDTA |

| Frågeställning/<br>Analys   | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
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|                             |               | 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, MTTTP, 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, TTL5, 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 meningiompainel v1, 10 gener<br>LZTR1, MEN1, NF2, PRKAR1A, PTCH1, PTEN, SMARCB1, SMARCE1, SUFU, WRN                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys   | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Analys/Metod                         | Svarstid<br>(dagar) | Rör  |
|-----------------------------|---------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------|---------------------|------|
| 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             | Perifert blod | <p>Skelettdysplasipanel v1, 402 gener</p> <p>ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AFF3, AFF4, AGA, AGPS, AIFM1, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANOS, ANTXR2, ARCN1, ARHGAP31, ARSB, ARSL, ASCC1, ASXL1, ASXL2, ATP6VOA2, 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, 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, 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, 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,</p> | NGS TruSeq helgenom In silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                           | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | Analys/Metod                  | Svarstid<br>(dagar) | Rör  |
|---------------------------------------------------------------------------------------------------------------------|---------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------|---------------------|------|
|                                                                                                                     |               | SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMARCA1, 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, TGFB2, 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<br>Utvecklingsförsening/Autism<br>Intellectuell<br>funktionsnedsättning                            | Perifert blod | Screening                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Mikroarray                    | 56                  | EDTA |
| Anlagsbärrutredning för<br>Syndromutredning<br>Utvecklingsförsening/Autism<br>Intellectuell<br>funktionsnedsättning | Perifert blod | Screening                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Mikroarray<br>Riktad          | 56                  | EDTA |
| Transtyretinmedierad<br>amyloidosis                                                                                 | Perifert blod | TTR                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 | NGS TWIST In<br>silico panel* | 90                  | EDTA |
| Tanatofores dysplasi<br>(Se även FGFR3-relaterad<br>skelettdysplasi)                                                | Perifert blod | FGFR3 exon 7, 9, 14, 18                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | Sangersekvensering<br>FGFR3   | 56                  | EDTA |

| Frågeställning/<br>Analys                                                                                                                                                                                                                                                                                    | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------|---------------------|------|
| Trombocytopeni                                                                                                                                                                                                                                                                                               | Perifert blod | Trombocytopenipanel v2, 127 gener<br><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, 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, TBXA51, TERT, THPO, TINF2, TSR2, TUBB1, UBE2T, VWF, WAS, WIPF1, WRAP53, XRCC2, ZCCHC8</i> | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Tuberös skleros                                                                                                                                                                                                                                                                                              | Perifert blod | <i>TSC1, TSC2</i>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                | NGS TWIST In<br>silico panel*              | 90                  | EDTA |
| Tubulopati<br>(inkl. renala tubulära<br>njursjukdomar,<br>elektrolytrubbningar (natrium,<br>kalium, magnesium, fosfat,<br>calcium, acidosis och alkalos),<br>aldosteronavvikelse, nefrogen<br>och neurohypofysär diabetes<br>insipidus, njurstensjukdomar,<br>nefrokalcinosis och metabola<br>njursjukdomar) | Perifert blod | Tubulopatipanel v1, 112 gener<br><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<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Tyroideahormonresistens<br>(inkl. hypotyroidism)                                                                                                                                                                                                                                                             | Perifert blod | Tyroideahormonresistenspanel v2, 35 gener<br><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<br>helgenom In<br>silico panel  | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                                 | Vävnad        | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                          | Analys/Metod                               | Svarstid<br>(dagar) | Rör  |
|---------------------------------------------------------------------------------------------------------------------------|---------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------|---------------------|------|
| Hippel-Lindau (VHL)                                                                                                       | Perifert blod | VHL                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | NGS TruSeq<br>helgenom In<br>silico panel  | 90                  | EDTA |
| Welanders distala myopati                                                                                                 | Perifert blod | TIA1 exon 13                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  | Sangersekvens<br>ering 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<br>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, 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. | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |
| Ärftlig njursjukdom<br>(Inkl. tubulopatipanelen och<br>polycystisk njursjukdomspanelen<br>samt proteinuri, hematuri (inkl | Perifert blod | Ärftlig njursjukdomspanel v1, 290 gener<br>ACE, ACTN4*, ADA2, AGT, AGTR1, AGXT, AHI1, AIRE, ALG5, ALG8, ALG9, ALMS1*, ALPL, AMMECR1, AMN, ANKS6, ANLN, AP2S1, APOA1, APOA4, APOC2, APOE, APRT, AQP2, ARHGDI, ARL13B, ARL6, ATP1A1, ATP6V0A4, ATP6V1B1, ATP7B, AVP, AVPR2,                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     | NGS TruSeq<br>helgenom In<br>silico panel* | 90                  | EDTA |

| Frågeställning/<br>Analys                                                                                                                                                                                                                                                              | Vävnad | Kromosomområde / Gen                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                            | Analys/Metod | Svarstid<br>(dagar) | Rör |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------|---------------------|-----|
| Alports syndrom), FSGS,<br>nefronofitis, autosomalt dominant<br>tubulointerstitiell njursjukdom<br>(exklusive MUC1-ADTKD), aHUS<br>och monogen amyloidos.<br><br>Inkl. ej orsaker till<br>njurmissbildningar (CAKUT) eller<br>monogena<br>autoimmuna/autoinflammatoriska<br>sjukdomar) |        | B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BICC1, BSND, C3, CA2,<br>CACNA1D, CACNA1H, CACNA1S, CASR, CC2D2A, CD151, CD2AP, CD46*, CDC73,<br>CDKN1B, CEP164, CEP290*, CEP41, CEP83, CFB, CFH*, CFHR1, CFHR2, CFHR3, CFHR5,<br>CFI, CHRM3, CLCN2, CLCN5, CLCNKB*, CLDN10, CLDN16, CLDN19, CNNM2, COL4A1,<br>COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CPLANE1, CRB2, CSPP1, CTNS,<br>CUBN*, CUL3, CYP11B1*, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, DAAM2,<br>DCDC2, DGKE, DICER1*, DLC1, DLG5, DMP1, DNAJB11, DYNC2H1, DYNC2I1, DZIP1L,<br>EHHADH, ENPP1, FAH, FAM111A, FAM20A, FAN1, FAT1, FGA, FGF23, FN1, FOXI1,<br>GALNT3, GANAB, GATA3, GATM, GCM2, GLA, GLIS2, GNA11, GNAS, GON7, GRHPR,<br>GSN, HNF1B, HNF4A, HOGA1, HPRT1, HSD11B2, IFT122*, IFT140, IFT172, IFT27, IFT43,<br>IFT74, INF2, INPP5E, INVS, IQCB1, ITGA3, ITS1, ITS2, JAG1, KANK2, KCNA1, KCNJ1,<br>KCNJ10, KCNJ16, KCNJ5, KLHL3, LAGE3, LAMA5, LAMB2, LCAT, LMX1B, LRP2, LYZ,<br>LZTFL1, MAFB, MAGED2, MAGI2, MAPKBP1, MEN1, MKKS, MKS1, MMACHC, MMUT,<br>MOCOS, MT-TF, MYH9, MYO1E, NEK1, NEK8, NLRP3, NOS1AP, NOTCH2*, NPHP1,<br>NPHP3, NPHP4, NPHS1, NPHS2, NPR1, NR3C1, NR3C2, NUP107, NUP133, NUP160,<br>NUP205, NUP85, NUP93, OCRL, OFD1, OSSEP, P3H2, PAX2, PCBD1, PDE3A, PDSS2,<br>PHEX, PKD1*, PKD2, PKHD1, PLCE1, PMM2, PODXL, PRKCSH, PSKH1, PTH, PTH1R,<br>PTPRO, REN, RET, RMND1*, RPGRIP1L, RRAGD, RRM2B, SARS2, SCARB2, SCLT1,<br>SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SGPL1, SLC12A1,<br>SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4,<br>SLC5A2, SLC6A19, SLC7A7, SLC7A9, SMARCA1, STX16, TBC1D8B, TBCE, TCTN1,<br>TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNS2,<br>TP53RK, TPRKB, TRAF3IP1, TRIM8, TRPC6, TRPM6, TRPM7, TSC1, TSC2, TSEN2,<br>TTC21B, TTC8, TTR, TULP3, TXNDC15, UMOD, VDR, VHL, VIPAS39, VPS33B, WDPCP,<br>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.