

SYNLAB MVZ Humangenetik Mannheim

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Next Generation Sequencing (NGS)

Next-generation sequencing allows the analysis of up to several million single DNA molecules in one sequencing run by "massive multiple parallel sequencing". Sequencing of single DNA molecules takes place either on PCR products (so-called amplicon libraries) or on specifically prepared DNA fragments (so-called enrichment libraries).

On average, approx. 4 - 6 sequence variants (Small Nucleotide Variants (SNVs)) are detected per analyzed gene, the relevance of which must be assessed by the reporting scientist for the interpretation of findings. Access to public, commercial and inhouse databases and knowledgeable alert variant analysis classification and filtration are therefore crucial for high quality reporting.

We offer NGS sequencing solutions from **whole exome analysis** through large and small **panel analyses** down to **single gene** and single nucleotide analysis. If analyses can be targeted or are technically not accessible by NGS technology we do as well offer genetic analyses including Array CGH, Sanger sequencing, CNV analysis based on MLPA assays and Repeat analyses.

For NGS sequencing the bioinformatic analysis is based on illumina Dragen pipeline technology. For clinical annotation filtration and reporting Varvis (company Limbus Medtech) is used. Analyses are evaluated by scientists, medical reports are validated by certified human geneticists. Reporting can be offered in German or English language.

If no clinical report is needed we can also provide sequence analysis only with raw data transfer (FastQ files).

For preconfigured gene panels, please see the downloadable PDF file.

On request we can include additional genes in the panels and we also provide custom panel analyses.

Finally we offer whole exome analysis (WES) based on TWIST whole exome supplemented with a customized spike-in that encompasses intronic and regulatory pathogenic variants (class 4 and class 5) listed in ClinVar and HGMD professional from 2022. Thus, also known pathogenic intronic variants are included in the exome analysis. WES can be offered for single analysis (only the affected patient) or preferentially for trio-constellation, that includes the affected patient and the parents. The trio-constellation analysis gives the best results as it improves classification and interpretation of variants, and allows to determine phasing of variants within one gene without time consuming segregation analysis.

Sample requirements

Material

- [E] EDTA Blood > 1 ml (preferably 3 ml)
- [S] Saliva (Oragene DNA OG-500/OGD-500 , OG-575 & OGD-575 or OG-510/OGD-510 kit)
- [D] Extracted DNA > 2 µg (concentration 20 – 150 ng/µl)
- [F] DNA extracted from formalin-fixed paraffin-embedded (FFPE) tissue

Sending Samples

Human genetics patient samples have a minimal likelihood of containing pathogens and can therefore be sent as "Exempted Medical Samples" if certain basic requirements for the packaging are met.

Patient samples with a minimal probability of containing pathogens can be sent as exempt medical samples without specifying a UN number ("P 650 light") provided the following packaging conditions are observed:

Packaging: "EXEMPTED MEDICAL PROBE" and "EXEMPT HUMAN SPECIMEN"

Triple packaging consisting of

- waterproof primary vessel (Monovette)
- waterproof secondary packaging (receptacle with absorbent material)
- sufficient solid outer packaging (ZHMA shipping bag).

When sending by post, remember the following points:

A box-shaped packaging made of cardboard or a shipping envelope made of tear-resistant paper or plastic film is permitted as transport packaging.

It can be sent as a parcel (weight: up to 2,000 g), maxi (weight: up to 1,000 g) or large letters (weight: up to 500 g).

The measures mentioned are intended to prevent any release of the sample!

International orders as well as national orders can be sent directly to us.

For prenatal samples please contact us before sending to allow better tracking of the samples.

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Whole Exome Sequencing (WES)

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Diseases/Diagnostics	TAT	Material
Brain Malformations		
Alcardi-Goutières syndrome (AGS) * Gene panel: ID058.01, 9 genes (13,0 kb) ADAR, IFIH1, LSM11, RNASEH2A, RNASEH2B, RNASEH2C, RNU7-1, SAMHD1, TREX1	3 - 5 w	E
Cerebellar dysgenesis, X-linked * Gene panel: ID219.00, 11 genes (24,5 kb) ABCB7, CASK, DKC1, FMR1, L1CAM, MECP2, MID1, OFD1, OPHN1, SLC9A6, ZIC3	3 - 5 w	E
Complex cortical dysplasia with other brain malformations (CDCBM) * Gene panel: ID271.02, 15 genes (52,0 kb) ADGRG1, APC2, CAMSAP1, CTNNA2, DYNC1H1, KIF2A, KIF5C, KIF26A, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2	3 - 5 w	E
Holoprosencephaly (HPE) * Gene panel: ID169.02, 17 genes (48,8 kb) CDON, CNOT1, DLL1, DISP1, FGF8, FGFR1, GAS1, GLI2, PRRX1, PTCH1, SHH, SIX3, STAG2, STIL, TGIF1, WDR62, ZIC2	3 - 5 w	E
Joubert syndrome (JBTS) * Gene panel: ID028.03, 40 genes (104,1 kb) AHI1, ARL13B, ARL3, ARMCF9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 w	E
Lissencephaly (LIS) * Gene panel: ID133.00, 12 genes (46,4 kb) ARX, CDK5, CEP85L, DCX, KATNB1, LAMB1, MACF1, NDE1, PAFAH1B1, RELN, TMTC3, TUBA1A	3 - 5 w	E
Meckel syndrome (MKS) * Gene panel: ID032.02, 13 genes (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 w	E
Neuronal migration disorders, comprehensive diagnostics * Gene panel: ID180.01 Neuronal migration disorders, comprehensive diagnostics: 82 genes (264,4 kb) ACTB, ACTG1, ADGRG1, AKT3, APC2, ARF1, ARFGEF2, ARX, B3GALNT2, B4GAT1, CAMSAP1, CCND2, CDK5, CEP85L, COL3A1, COL4A1, COL4A2, COLGALT1, CRADD, CRPPA, CTNNA2, DAG1, DCHS1, DCX, DYNC1H1, EML1, EMX2, ERMARD, FAT4, FH, FIG4, FKR, FKTN, FLNA, GMP, GMPPB, KATNB1, KIF1BP, KIF2A, KIF5C, KIF26A, LAMB1, LAMC3, LARGE1, MACF1, MAP1B, MTOR, NDE1, NEDD4L, OCLN, PAFAH1B1, PHGDH, PI4KA, PIK3CA, PIK3R2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSAT1, RAB18, RAB3GAP1, RAB3GAP2, RAC3, RELN, RTTN, RXYLT1, SHH, SIX3, TBC1D20, TMTC3, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUBGCP2, WDR62 Periventricular nodular heterotopia (PVNH): 6 genes (26,1 kb) ARF1, ARFGEF2, ERMARD, FLNA, MAP1B, NEDD4L Complex cortical dysplasia (CDCBM): 12 genes (45,7 kb) APC2, CTNNA2, KIF2A, KIF5C, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1 Walker-Warburg syndrome (MDDGA): 14 genes (23,7 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKR, FKTN, GMP, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Lissencephaly (LIS): 12 genes (46,4 kb) ARX, CDK5, CEP85L, DCX, KATNB1, LAMB1, MACF1, NDE1, PAFAH1B1, RELN, TMTC3, TUBA1A Polymicrogyria: 15 genes (42,0 kb) ADGRG1, AKT3, CCND2, COL3A1, FIG4, KIFBP, OCLN, PI4KA, PIK3CA, RTTN, PIK3R2, TUBA1A, TUBA8, TUBB2B, WDR62 Schizencephaly: 7 genes (19,7 kb) COL4A1, COL4A2, COLGALT1, EMX2, SHH, SIX3, WDR62	4 - 6 w	E
Periventricular nodular heterotopia (PVNH) * Gene panel: ID306.00, 6 genes (26,1 kb) ARF1, ARFGEF2, ERMARD, FLNA, MAP1A, NEDD4L	3 - 5 w	E
Polymicrogyria * Gene panel: ID176.01, 15 genes (42,0 kb) ADGRG1, AKT3, CCND2, COL3A1, FIG4, KIFBP, OCLN, RTTN, PI4KA, PIK3CA, PIK3R2, TUBA1A, TUBA8, TUBB2B, WDR62	3 - 5 w	E
Pontocerebellar hypoplasia (PCH) * Gene panel: ID071.02, 26 genes (60,5 kb) AMPD2, CASK, CDC40, CHMP1A, CLP1, COASY, EXOSC1, EXOSC3, EXOSC8, EXOSC9, MINPP1, PCLO, PPIL1, RARS2, RELN, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN2, TSEN15, TSEN34, TSEN54, VPS51, VPS53, VRK1	3 - 5 w	E
Schizencephaly * Gene panel: ID173.00, 7 genes (19,7 kb) COL4A1, COL4A2, COLGALT1, EMX2, SHH, SIX3, WDR62	3 - 5 w	E
Septooptic dysplasia * Gene panel: ID378.00, 8 genes (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	3 - 5 w	E
Walker-Warburg syndrome (WWS, MDDGA) * Gene panel: ID178.00, 14 genes (23,7 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKR, FKTN, GMP, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Arrhythmogenic right ventricular cardiomyopathy (ARVD, ARVC) * Gene panel: ID010.03, 27 genes (185,3 kb) ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN	4 - 6 w	E
Atrial and ventricular septal defect (ASD, VSD, AVSD) * Gene panel: ID143.00, 12 genes (21,1 kb) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NKX2-5, NR2F2, TBX20, TLL1	3 - 5 w	E
Bicuspid aortic valve (AOVD) * Gene panel: ID301.00, 6 genes (16,7 kb) GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2	3 - 5 w	E
Brugada syndrome (BRGDA) * Gene panel: ID014.02, 23 genes (56,4 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4	3 - 5 w	E
Cardiac arrhythmia, comprenensive diagnostics * Gene panel: ID026.03 Cardiac arrhythmia, comprenensive diagnostics: 71 genes (274,2 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, EMD, FGF12, GJA5, GNB2, GNB5, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LEMD2, LMNA, MYH6, MYL4, NKX2-5, NPPA, NUP155, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TANGO2, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TTN Long QT syndrome (LQT): 18 genes (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN Brugada syndrome (BRGDA): 23 genes (56,4 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, FGF12, GPD1L, HCN4, KCND2, KCND3, KCNE3, KCNE5, KCNH2V, KCNJ8, PKP2, RANGRF, SCN1B, SCN2B, SCN3B, SCN5A, SCN10A, SEMA3A, SLMAP, TRPM4 Early repolarization syndrome (ERS): 12 genes (38,9 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A Familial atrial fibrillation (ATFB): 17 genes (29,5 kb) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A Short QT syndrome (SQT): 7 genes (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3 Sick sinus syndrome (SSS): 4 genes (16,5 kb) GNB2, HCN4, MYH6, SCN5A Catecholaminergic polymorphic ventricular tachycardia (CPVT): 9 genes (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN Arrhythmogenic right ventricular dysplasia (ARVD): 15 genes (150,6 kb) CDH2, CTNNA3, DES, DSC2, DSG2, DSP, JUP, PLN, LMNA, PKP2, PRKAG2, RYR2, TGFB3, TMEM43, TTN	4 - 6 w	E
Cardiomyopathy, comprehensive diagnostics * Gene panel: ID027.05 Cardiomyopathy, comprehensive diagnostics: 154 genes (487,0 kb) ABCC9, ACTA1, ACTC1, ACTN2, ALPK3, ANKRD1, APOA1, B2M, BAG3, BAG5, BRAF, CACNA1C, CACNB2, CALR3, CAP2, CASQ2, CAV3, CDH2, COA5, COA6, CORIN, COX15, CRYAB, CSRP3, CTF1, CTNNA3, DES, DMD, DMPK, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FGA, FHL1, FHL2, FHOD3, FKRP, FKTN, FLII, FLNC, FTH1, FXN, GAA, GATA4, GATAD1, GET3, GLA, GSN, HADHA, HAMP, HCN4, HFE, HJV, HRAS, ILK, JPH2, JUP, KCNQ1, KIF20A, KLF10, KLHL24, KRAS, KY, LAMA4, LAMP2, LDB3, LIMS2, LMNA, LMOD2, LRRC10, LYZ, LZTR1, MAP2K1, MAP2K2, MAPK1, MCM10, MIB1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOT, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NNT, NONO, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, PYROXD1, RAF1, RBM20, RIT1, RPL3L, RRAS2, RYR2, SCN5A, SCO2, SDHA, SGCD, SHOC2, SLC40A1, SLC25A4, SOS1, SOS2, SPRED2, SVIL, SYNE1, SYNE2, TAFAZZIN, TBX5, TBX20, TCAP, TFR2, TGFB3, TJP1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TRPM4, TTN, TTR, UNC45B, VCL, VEZF1 Dilated cardiomyopathy (DCM, CMD): 68 genes (301,9 kb) ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CAP2, CRYAB, CSRP3, DES, DMD, DSG2, DSP, DTNA, EMD, EYA4, FKTN, FLII, FLNC, GATAD1, GET3, HFE, ILK, JPH2, LAMA4, LAMP2, LDB3, LMNA, LMOD2, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1 Hypertrophic cardiomyopathy (HCM, CMH): 56 genes (253,8 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, CACNA1C, CALR3, CAV3, CORIN, CRYAB, CSRP3, DES, DSP, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, KLF10, KLHL24, KRAS, LAMP2, LDB3, MAP2K1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SLC25A4, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL Restrictive cardiomyopathy (RCM): 15 genes (135,7 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR Arrhythmogenic right ventricular cardiomyopathy (ARVD, ARVC): 27 genes (185,3 kb) ACTC1, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, RYR2, SCN5A, TGFB3, TJP1, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN Nondilated left ventricular cardiomyopathy (NDLVC, LVNC): 35 genes (226,1 kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
CADASIL and CARASIL * Gene panel: ID167.01, 3 genes (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 w	E
Catecholaminergic polymorphic ventricular tachycardia (CPVT) * Gene panel: ID012.03, 9 genes (33,9 kb) ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN	3 - 5 w	E
Cerebrovascular diseases and stroke * Gene panel: ID234.02 Cerebrovascular diseases and stroke: 44 genes (130,0 kb) ACE, ADA2, ACTA2, ALOX5AP, APOE, APP, CBS, CCM2, COL3A1, COL4A1, COL4A2, COLGALT1, CST3, F2, F5, FBN1, FLNA, GAA, GLA, GSN, GUCY1A1, HTRA1, ITM2B, JAG1, KRIT1, MTHFR, MYH11, MYLK, NOS3, NOTCH3, OTC, PDCD10, POLG, PRKCH, PRNP, RNF213, SLC2A10, SMAD3, TGFB2, TGFB3, TGFBFR1, TGFBFR2, TREX1, TTR Cerebral amyloid angiopathy: 6 genes (7,1 kb) APP, CST3, GSN, ITM2B, PRNP, TTR Brain small vessel disease (BSVD): 3 genes (12,0 kb) COL4A1, COL4A2, COLGALT1 Cerebral cavernous malformation (CCM): 3 genes (4,2 kb) CCM2, KRIT1, PDCD10 Cerebral aneurysm and dissection: 10 genes (32,8 kb) ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB3, TGFBFR1, TGFBFR2 Moyamoya disease (MYMY): 4 genes (22,2 kb) ACTA2, GUCY1A1, JAG1, RNF213 CADASIL, CARASIL: 2 genes (8,4 kb) HTRA1, NOTCH3	4 - 6 w	E
Connective tissue disorders with aortic involvement * Gene panel: ID137.05 Connective tissue disorders with aortic involvement: 67 genes (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFBFR1, TGFBFR2, THSD4, TNXB, ZNF469 Thoracic aortic aneurysm and aortic dissection (TAA/D): 17 genes (42,6 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBFR1, TGFBFR2, THSD4 Ehlers-Danlos syndrome (EDS): 20 genes (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan syndrome (MFS): 5 genes (22,2 kb) CBS, FBN1, FBN2, TGFBFR2, TGFBFR1 Cutis laxa syndrome (ARCL, ADCL): 11 genes (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1 Stickler syndrome (STL): 4 genes (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani syndrome (WMS): 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 w	E
Congenital heart defects, comprehensive diagnostics * Gene panel: ID019.02 Congenital heart defects, comprehensive diagnostics: 149 genes (472,9 kb) ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR2B, ADAMTS10, ADAMTS17, ADAMTS19, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CFAP45, CFAP52, CFAP53, CFC1, CHD4, CHD7, CIROP, CITED2, CREBBP, CRELD1, DHCR7, DLL4, DNAAF1, DNAH5, DNAH9, DNAH11, DOCK6, DPYSL5, DTNA, EHMT1, ELN, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FLT4, FOXC1, FOXF1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GPC3, HAAO, HAND1, HOXA1, HRAS, ISL1, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MMP21, MNS1, MRAS, MYH11, MYH6, MYRF, NADSYN1, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NONO, NOTCH1, NOTCH2, NR2F2, NRAS, NSD1, ODAD2, PIGL, PITX2, PKD1L1, PLD1, PPP1CB, PRDM6, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, ROBO4, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMAD6, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, SPRED2, STAG2, STRA6, TAB2, TBX1, TBX20, TBX3, TBX5, TFAP2B, TGDS, TGFBFR1, TGFBFR2, TKT, TLL1, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZFPM2, ZIC3 Isolated congenital heart defects: 48 genes (149,9 kb) ACTC1, ACVR2B, ADAMTS19, DNAAF1, DNAH5, DNAH9, DNAH11, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CITED2, CRELD1, ELN, FLNA, FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, HAND1, ISL1, JAG1, MMP21, MED13L, MNS1, MYH6, NKX2-5, NKX2-6, NR2F2, NODAL, NOTCH1, ODAD2, PKD1L1, PLD1, PRDM6, ROBO4, SMAD6, TAB2, TBX1, TBX20, TFAP2B, TLL1, ZFPM2, ZIC3 Syndromic congenital heart defects: 109 genes (354,9 kb) ABL1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFBFR1, TGFBFR2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Congenital multiple heart defects (CHTD) * Gene panel: ID302.01, 9 genes (21,5 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, PLXND1, SMAD2, TAB2, ZIC3	3 - 5 w	E
Conotruncal heart malformations (CTHM) * Gene panel: ID160.01, 12 genes (24,8 kb) CFC1, FLT4, FOXH1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3	3 - 5 w	E
Cutis laxa (ARCL, ADCL) * Gene panel: ID109.02, 11 genes (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 w	E
CHARGE syndrome * Gene panel: ID307.00, 3 genes (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 w	E
Dilated cardiomyopathy (CMD, DCM) * Gene panel: ID008.05, 68 genes (301,9 kb) ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CAP2, CRYAB, CSRP3, DES, DMD, DSG2, DSP, DTNA, EMD, EYA4, FKTN, FLII, FLNC, GATAD1, GET3, HFE, ILK, JPH2, LAMA4, LAMP2, LDB3, LMNA, LMOD2, LRRC10, MIB1, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYPN, NEBL, NEXN, NKX2-5, OBSCN, PDLIM3, PKP2, PLEKHM2, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, TBX20, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1	4 - 6 w	E
Early repolarization syndrome (ERS) * Gene panel: ID330.00, 12 genes (38,9 kb) ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A	3 - 5 w	E
Ehlers-Danlos syndrome (EDS) * Gene panel: ID039.05 Ehlers-Danlos syndrome (EDS): 20 genes (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Ehlers-Danlos syndrome (EDS), autosomal dominant: 8 genes (36,1 kb) C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1 Ehlers-Danlos syndrome (EDS), autosomal recessive: 13 genes (47,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469	3 - 5 w	E
Endocrine hypertension * Gene panel: ID270.03 Endocrine hypertension: 33 genes (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronism (HALD): 5 genes (19,1 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronism type II (PHA2): 4 genes (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Pheochromocytoma/paraganglioma syndrome (PPGL): 11 genes (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenal cushing syndrome (PPNAD): 4 genes (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle syndrome (LIDLS): 3 genes (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertension with low renin level: 20 genes (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 w	E
Familial atrial fibrillation (ATFB) * Gene panel: ID016.02, 17 genes (29,5 kb) ABCC9, GJA5, KCNA5, KCNE1, KCNE2, KCNE5, KCNH2, KCNJ2, KCNQ1, MYL4, NPPA, NUP155, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A	3 - 5 w	E
Heart-hand syndrome * Gene panel: ID165.01, 9 genes (23,8 kb) DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5	3 - 5 w	E
Hereditary hemorrhagic telangiectasia (HHT) * Gene panel: ID155.01, 7 genes (15,6 kb) ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4	3 - 5 w	E
Hypertrophic cardiomyopathy (CMH, HCM) * Gene panel: ID007.05, 56 genes (253,8 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, CACNA1C, CALR3, CAV3, CORIN, CRYAB, CSRP3, DES, DSP, FHL1, FHOD3, FLNC, GAA, GLA, JPH2, KLF10, KLHL24, LAMP2, KRAS, LDB3, MAP2K1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, OBSCN, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RYR2, SLC25A4, TCAP, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Isolated congenital heart defects * Gene panel: ID017.04 Isolated congenital heart defects: 43 genes (103,4 kb) ACTC1, ACVR2B, ADAMTS19, DNAAF1, DNAH5, DNAH9, DNAH11, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CITED2, CRELD1, ELN, FLNA, FLT4, FOXH1, GATA4, GATA5, GATA6, GDF1, GJA1, HAND1, ISL1, JAG1, MMP21, MED13L, MNS1, MYH6, NKX2-5, NKX2-6, NR2F2, NODAL, NOTCH1, ODAD2, PKD1L1, PLD1, PRDM6, ROBO4, SMAD6, TAB2, TBX1, TBX20, TFAP2B, TLL1, ZFPM2, ZIC3 Atrial and/or ventricular septal defect (ASD, VSD): 12 genes (21,1 kb) ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NR2F2, NKX2-5, TBX20, TLL1 Tetralogy of Fallot (TOF): 12 genes (23,5 kb) FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2 Visceral heterotaxy (HTX): 13 genes (26,2 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, MMP21, MNS1, NODAL, PKD1L1, ZIC3 Conotruncal heart malformations (CTHM): 12 genes 24,8 (kb) FLT4, FOXH1, CFC1, GATA5, GATA6, GDF1, MED13L, NKX2-5, NKX2-6, TBX1, ZFPM2, ZIC3 Aortic valve disease (AOVD): 7 genes (19,1 kb) ELN, GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB Hypoplastic left heart syndrome (HLHS): 5 genes (5,2 kb) GDF1, GJA1, HAND1, NKX2-5, NR2F2 Congenital heart defect, multiple type (CHTD): 7 genes (14,4 kb) FLT4, GATA5, GDF1, NR2F2, PLD1, TAB2, ZIC3 Cardiac valvular dysplasia (CVD): 3 genes (14,8 kb) ADAMTS19, FLNA, PLD1	4 - 6 w	E
Loeys-Dietz syndrome (LDS) and similar aortic diseases * Gene panel: ID009.07 Loeys-Dietz syndrome (LDS) and similar aortic diseases: 38 genes (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Loeys-Dietz syndrome (LDS): 8 genes (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2 Nonsyndromic thoracic aortic aneurysm (AAT): 11 genes (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Syndromes with aortic aneurysm: 29 genes (91,7 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2	4 - 6 w	E
Long QT syndrome (LQT) * Gene panel: ID013.01, 18 genes (52,5 kb) AKAP9, ALG10B, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, SCN4B, SCN5A, SNTA1, TRDN	3 - 5 w	E
Marfan syndrome (MFS) * Gene panel: ID022.00, 3 genes (11,8 kb) FBN1, TGFB1, TGFB2	2 - 4 w	E
Marfan syndrome (MFS) and similar syndromes * Gene panel: ID194.05 Marfan syndrome (MFS) and similar syndromes: 48 genes (153,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTS14, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan syndrome (MFS): 3 genes (11,8 kb) FBN1, TGFB1, TGFB2 Loeys-Dietz syndrome (LDS) and Thoracic aortic aneurysm (AAT): 17 genes (42,6 kb) ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Stickler syndrome (STL): 6 genes (21,1 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Weill-Marchesani syndrome (WMS): 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Ehlers-Danlos syndrome (EDS): 10 genes (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469	4 - 6 w	E
Muscular diseases with cardiac involvement * Gene panel: ID123.03 Muscular diseases with cardiac involvement: 19 genes (193,6 kb) BAG3, CRYAB, DES, DMD, EMD, FHL1, FLNC, KY, LDB3, LMNA, MYL2, MYOT, PYROXD1, SVIL, SYNE1, SYNE2, TMEM43, TTN, UNC45B Myofibrillar myopathy (MFM): 12 genes (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Emery-Dreifuss muscular dystrophy (EMDM): 6 genes (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43 Duchenne/Becker muscular dystrophy (DMD, BMD): 1 gene (11,1 kb) DMD	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Nondilated left ventricular cardiomyopathy (NDLVC, LVNC) * Gene panel: ID011.04, 35 genes (226,1 kb) ACTC1, ACTN2, DES, DMD, DMPK, DSP, DTNA, FLNC, GATA4, HCN4, ILK, LDB3, LMNA, MIB1, MYBPC3, MYH7, MYL2, MYL3, NKX2-5, NNT, NONO, OBSCN, PLN, PRDM16, RBM20, RYR2, SCN5A, TAFAZZIN, TBX5, TBX20, TMEM43, TMEM70, TNNT2, TPM1, TTN	4 - 6 w	E
Noonan syndrome (NS) * Gene panel: ID023.06, 16 genes (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2	3 - 5 w	E
Pulmonary hypertension (PAH, PPH) * Gene panel: ID281.01, 23 genes (49,9 kb) ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, G6PC3, GDF2, KCNA5, KCNK3, KDR, NFU1, NOTCH3, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4	3 - 5 w	E
RAS-related disorders * Gene panel: ID015.05 RAS-related disorders: 21 genes (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Noonan syndrome (NS): 15 genes (24,7 kb) BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Cardiofaciocutaneous syndrome (CFC): 4 genes (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 LEOPARD syndrome (LPRD): 3 genes (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 w	E
Restrictive cardiomyopathy (RCM) * Gene panel: ID105.03, 15 genes (135,7 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MYBPC3, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTN, TTR	4 - 6 w	E
Short QT syndrome (SQT) * Gene panel: ID233.01, 7 genes (22,1 kb) CACNA1C, CACNA2D1, CACNB2, KCNH2, KCNJ2, KCNQ1, SLC4A3	3 - 5 w	E
Storage diseases with cardiac involvement * Gene panel: ID149.03 Storage diseases with cardiac involvement: 16 genes (20,7 kb) APOA1, B2M, FGA, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, LYZ, PRKAG2, SLC40A1, TFR2, TTR Cardiac glycogen storage disease (GSD): 3 genes (5,8 kb) GAA, LAMP2, PRKAG2 Hemochromatosis (HFE): 6 genes (7,3 kb) FTH1, HAMP, HFE, HJV, SLC40A1, TFR2 Amyloidosis (AMYLD): 6 genes (6,4 kb) APOA1, B2M, FGA, GSN, LYZ, TTR	3 - 5 w	E
Syndromic congenital heart defects * Gene panel: ID252.02 Syndromic congenital heart defects: 109 genes (354,9 kb) ABL1, ACTA2, ACTB, ACTG1, ADAMTS10, ADAMTS17, AFF4, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BRAF, CBL, CCDC22, CDK13, CHD4, CHD7, CREBBP, DHCR7, DLL4, DOCK6, DPYSL5, DTNA, EHMT1, EOGT, EP300, EVC, EVC2, FBN1, FBN2, FLNA, FOXC1, FOXF1, FOXP1, GATA6, GPC3, HAAO, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYNU, LTBP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MED12, MED13L, MEGF8, MEIS2, MGP, MRAS, MYH11, MYRF, NADSYN1, NF1, NIPBL, NONO, NOTCH1, NOTCH2, NRAS, NSD1, PIGL, PITX2, PPP1CB, PRKAR1A, PRKD1, PTPN11, RAB23, RAF1, RBM10, RBPJ, RERE, RIT1, RRAS2, SALL1, SALL4, SEMA3E, SHOC2, SMARCA4, SMARCB1, SMARCE1, SMC3, SOS1, SOS2, STAG2, SPRED2, STRA6, TBX1, TBX3, TBX5, TFAP2B, TGDS, TGFB1, TGFB2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3 Noonan syndrome (NS): 16 genes (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Cardiofaciocutaneous syndrome (CFC): 4 genes (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 Ritscher-Schinzel syndrome (RTSC): 4 genes (10,0 kb) DPYSL5, CCDC22, VPS35L, WASHC5 Adams-Oliver syndrome (AOS): 6 genes (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ Kabuki syndrome (KABUK): 2 genes (20,8 kb) KDM6A, KMT2D Alagille-Syndrom (ALGS): 2 Gene (10,7 kb) JAG1, NOTCH2 Marfan syndrome (MFS): 3 genes (12,0 kb) FBN1, TGFB1, TGFB2 CHARGE syndrome: 2 genes (11,3 kb) CHD7, SEMA3E VCRL syndrome: 3 genes (4,4 kb) HAAO, KYNU, NADSYN1	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Sick sinus syndrome (SSS) * Gene panel: ID107.01, 4 genes (16,5 kb) GNB2, HCN4, MYH6, SCN5A	3 - 5 w	E
Sudden cardiac death * Gene panel: ID349.01 Sudden cardiac death: 127 genes (393,9 kb) ABCC9, ACTC1, ACTN2, AKAP9, ALG10B, ALPK3, ANK2, ANKRD1, BAG3, BAG5, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CAP2, CASQ2, CAV3, CDH2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FGF12, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATAD1, GET3, GJA5, GLA, GNAI2, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KLHL24, LAMA4, LAMP2, LDB3, LEMD2, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK2, MYOZ2, MYPN, MYZAP, NEXN, NKX2-5, NPPA, NUP155, PKP2, PLN, PPA2, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RANGRF, RBM20, RPL3L, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SEMA3A, SGCD, SLC4A3, SLMAP, SNTA1, TAFAZZIN, TBX5, TCAP, TECRL, TGFB3, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TSPYL1, TTN, TTR, VCL, VEZF1 Arrhythmia (BRGDA, LQT) and sudden cardiac death: 74 genes (281,6 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, DTNA, EMD, FGF12, GJA5, GNAI2, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LEMD2, LMNA, MYBPC3, MYH6, MYL4, NKX2-5, NPPA, NUP155, PPA2, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TSPYL1, TTN Cardiomyopathy (HCM, DCM) and sudden cardiac death: 71 genes (261,3 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, BAG5, CAP2, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSG2, DSP, EMD, EYA4, FHL1, FHOD3, FKRP, FKTN, FLII, FLNC, GATAD1, GET3, GLA, JPH2, JUP, KLHL24, LAMA4, LAMP2, LDB3, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, MYZAP, NEXN, NKX2-5, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, TAFAZZIN, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TTN, TTR, VCL, VEZF1	4 - 6 w	E
Tetralogy of Fallot (TOF) * Gene panel: ID144.01, 12 genes (29,5 kb) FLT4, GATA4, GATA5, GATA6, GDF1, ISL1, JAG1, NKX2-5, NR2F2, TAB2, TBX1, ZFPM2	3 - 5 w	E
Thoracic aortic aneurysm and aortic dissection (AAT, TAAD) * Gene panel: ID020.01 Thoracic aortic aneurysm and aortic dissection (AAT, TAAD): 16 genes (41,0 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Thoracic aortic aneurysm, nonsyndromic type (AAT): 10 genes (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Loeys-Dietz syndrome (LDS): 6 genes (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2	3 - 5 w	E
Visceral heterotaxy (HTX) * Gene panel: ID145.01, 18 genes (72,4 kb) ACVR2B, CFAP45, CFAP52, CFAP53, CFC1, CIROP, CRELD1, GDF1, DNAAF1, DNAH5, DNAH9, DNAH11, MMP21, MNS1, NODAL, ODAD2, PKD1L1, ZIC3	3 - 5 w	E
Ciliopathies		
Bardet-Biedl syndrome (BBS) * Gene panel: ID093.02, 21 genes (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	3 - 5 w	E
Joubert syndrome (JBTS) * Gene panel: ID028.03, 40 genes (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 w	E
Meckel syndrome (MKS) * Gene panel: ID032.02, 13 genes (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 w	E
Nephronophthisis (NPHP) * Gene panel: ID030.02, 22 genes (70,9 kb) ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	3 - 5 w	E
Primary ciliar dyskinesia with or without situs inversus (PCD, CILD) * Gene panel: ID085.03, 50 genes (164,8 kb) BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10	4 - 6 w	E
Senior-Loken syndrome (SLSN) * Gene panel: ID029.01, 8 genes (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Ciliopathies		
Short-rib thoracic dysplasia with or without polydactyly (SRTD) * Gene panel: ID067.00 Short-rib thoracic dysplasia with or without polydactyly (SRTD): 20 genes (68,2 kb) CEP120, DYNC2H1, DYNC2L1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Short-rib thoracic dysplasia (Jeune-ATD, SRPS, SRTD): 18 genes (61,3 kb) CEP120, DYNC2H1, DYNC2L1, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Ellis-van Crefeld syndrome (EVC): 4 genes (11,5 kb) DYNC2L1, EVC, EVC1, WDR35	3 - 5 w	E
Renal ciliopathies, comprehensive diagnostics * Gene panel: ID376.00 Renal ciliopathies, comprehensive diagnostics: 75 genes (181,7 kb) AHI1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP418, CPLANE1, CSPP1, DCDC2, FAM149B1, GLIS2, IFT172, IFT27, IFT74, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, PDE6D, PIBF1, RPGRIP1L, SDCCAG8, SLC41A1, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TRIM32, TTC21B, TTC8, TXNDC15, WPCP, WDR19, XPNPEP3, ZNF423 Nephronophthisis (NPHP): 17 genes (50,3 kb) ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423 Bardet-Biedl syndrome (BBS): 22 genes (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WPCP Senior-Loken syndrome (SLSN): 8 genes (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19 Joubert syndrome (JBTS): 40 genes (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423 Meckel syndrome (MKS): 14 genes (36,2 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231, TXNDC15	4 - 6 w	E
Connective Tissue Diseases		
Connective tissue disorders with aortic involvement * Gene panel: ID137.05 Connective tissue disorders with aortic involvement: 67 genes (225,7 kb) ABCC6, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, AEBP1, ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, COL11A1, COL12A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP1, LTBP2, LTBP4, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRDM5, PRKG1, PYCR1, ROBO4, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2, THSD4, TNXB, ZNF469 Thoracic aortic aneurysm and aortic dissection (TAA/D): 17 genes (42,6 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Ehlers-Danlos syndrome (EDS): 20 genes (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Marfan syndrome (MFS): 5 genes (22,2 kb) CBS, FBN1, FBN2, TGFB2, TGFB1 Cutis laxa syndrome (ARCL, ADCL): 11 genes (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1 Stickler syndrome (STL): 4 genes (14,7 kb) COL2A1, COL9A1, COL9A2, COL11A1 Weill-Marchesani syndrome (WMS): 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 w	E
Cutis laxa (ARCL, ADCL) * Gene panel: ID109.02, 11 genes (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 w	E
Ehlers-Danlos syndrome (EDS) * Gene panel: ID039.05 Ehlers-Danlos syndrome (EDS): 20 genes (79,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469 Ehlers-Danlos syndrome (EDS), autosomal dominant: 8 genes (36,1 kb) C1R, C1S, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL12A1 Ehlers-Danlos syndrome (EDS), autosomal recessive: 13 genes (47,6 kb) ADAMTS2, AEBP1, B3GALT6, B4GALT7, CHST14, COL1A2, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Connective Tissue Diseases		
Loeys-Dietz syndrome (LDS) and similar aortic diseases * Gene panel: ID009.07 Loeys-Dietz syndrome (LDS) and similar aortic diseases: 38 genes (112,5 kb) ACTA2, AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP4, MAT2A, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Loeys-Dietz syndrome (LDS): 8 genes (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2 Nonsyndromic thoracic aortic aneurysm (AAT): 11 genes (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Syndromes with aortic aneurysm: 29 genes (91,7 kb) AEBP1, ALDH18A1, BGN, C1S, C1R, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, LTBP4, NOTCH1, PLOD1, ROBO4, SKI, SLC2A10, SMAD2, SMAD3, SMAD6, TGFB2, TGFB3, TGFB1, TGFB2	4 - 6 w	E
Marfan syndrome (MFS) * Gene panel: ID022.00, 3 genes (11,8 kb) FBN1, TGFB1, TGFB2	2 - 4 w	E
Marfan syndrome (MFS) and similar syndromes * Gene panel: ID194.05 Marfan syndrome (MFS) and similar syndromes: 48 genes (153,2 kb) ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, DLG4, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MFAP5, MYH11, MYLK, NKP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan syndrome (MFS): 3 genes (11,8 kb) FBN1, TGFB1, TGFB2 Loeys-Dietz syndrome (LDS) and Thoracic aortic aneurysm (AAT): 17 genes (42,6 kb) ACTA2, COL3A1, BGN, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Stickler syndrome (STL): 6 genes (21,1 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Weill-Marchesani syndrome (WMS): 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 Ehlers-Danlos syndrome (EDS): 10 genes (39,1 kb) CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469	4 - 6 w	E
Osteogenesis imperfecta (OI) * Gene panel: ID066.02, 21 genes (39,2 kb) ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TENT5A, WNT1	3 - 5 w	E
Stickler syndrome (STL) * Gene panel: ID062.00, 6 genes (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 w	E
Thoracic aortic aneurysm and aortic dissection (AAT, TAAD) * Gene panel: ID020.01 Thoracic aortic aneurysm and aortic dissection (AAT, TAAD): 16 genes (41,0 kb) ACTA2, COL3A1, FBN1, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2, THSD4 Thoracic aortic aneurysm, nonsyndromic type (AAT): 10 genes (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFB1, TGFB2, THSD4 Loeys-Dietz syndrome (LDS): 6 genes (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFB1, TGFB2	3 - 5 w	E
Developmental and Growth Disorders		
Autism spectrum disorders * Gene panel: ID076.04, 168 genes (664,6 kb) AHD1, ADNP, ADL, AFF2, AGO1, ALDH5A1, ANK2, ANKRD11, ARHGEF9, ARID1B, ARX, ASH1L, ASTN2, ASXL3, ATP1A1, AUTS2, BAZ2B, BCKDK, BCL11A, BRSK2, CACNA1C, CAPRIN1, CASK, CASZ1, CC2D1A, CDKL5, CELF4, CHD2, CHD7, CHD8, CIC, CNOT3, CNTN4, CNTNAP2, CREBBP, CSDE1, CSNK2A1, CTNNB1, CUL3, DDX3X, DEAF1, DHCR7, DIP2A, DLGAP2, DNMT3A, DPP6, DSCAM, DYNC1H1, DYRK1A, EBF3, EHMT1, EIF4E, EP300, FMR1, FOXG1, FOXP1, FOXP2, FRMPD4, GIGYF1, GLRA2, GRIA2, GRIN2A, GRIN2B, HERC2, HNRNP, IL1RAPL1, IQSEC2, IRF2BPL, KATNAL2, KCNQ3, KDM5A, KDM5C, KDM6B, KMT2A, KMT2C, KMT2E, KMT5B, MAGEL2, MAOA, MBD5, MBOAT7, MECP2, MED13, MED13L, MEF2C, MEIS2, MYT1L, NAA15, NEXMIF, NF1, NHS, NIPBL, NLGN1, NLGN3, NLGN4X, NOVA2, NR1I3, NR4A2, NRXN1, NRXN2, NRXN3, NSD1, OPHN1, PAH, PAX5, PCDH19, PDZD8, PHF12, PHF2, PHF21A, PHF3, POGZ, PPP2R5D, PSMD12, PTCHD1, PTEN, RAB39B, RAI1, RELN, RERE, RFX3, RIMS1, RORB, RPL10, SATB2, SCN1A, SCN2A, SEMA5A, SETD2, SETD5, SGSH, SHANK2, SHANK3, SLC6A1, SLC6A8, SLC9A6, SLC9A9, SMARCB1, SMARCC2, SON, SOX5, SPAST, STXBP1, SYN1, SYNGAP1, TANC2, TBL1XR1, TBR1, TCF20, TCF4, TLK2, TMLHE, TRIP12, TRPC6, TRRAP, TSC1, TSC2, UBE3A, UPF3B, VAMP2, VPS13B, WAC, WDFY3, ZMYM3, ZMYND8, ZNF292, ZNF462, ZSWIM6	4 - 6 w	E
Coffin-Siris syndrome (CSS) * Gene panel: ID118.02, 14 genes (46,5 kb) ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
Congenital disorder of glycosylation (CDG) * Gene panel ID035.02 Congenital disorder of glycosylation (CDG): 51 genes (74,7 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, MAGT1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3 Congenital disorder of glycosylation, type I (CDG1): 29 genes (39,6 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Congenital disorder of glycosylation, type II (CDG2): 22 genes (35,1 kb) ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, TMEM165, TMEM199	3 - 5 w	E
Cornelia de Lange syndrome (CDLS) * Gene panel: ID033.02, 8 genes (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 w	E
Developmental language disorder (DLD, CAS) Gene panel: ID368.00 Developmental language disorder (DLD, CAS): 34 genes (119,7 kb) ATP2C2, BCL11A, BUD13, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, NFXL1, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TM4SF20, TNRC6B, UPF2, WDR5, ZFXH4, ZNF142, ZNF277 Childhood apraxia of speech (CAS): 29 genes (110,2 kb) BCL11A, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TNRC6B, UPF2, WDR5, ZFXH4, ZNF142 Specific language impairment (SLI): 5 genes (9,5 kb) TP2C2, BUD13, NFXL1, TM4SF20, ZNF277	4 - 6 w	E
FG syndrome (FGS) * Gene panel: ID215.00, 3 genes (17,2 kb) CASK, FLNA, MED12	3 - 5 w	E
Growth abnormalities and macrocephaly * Gene panel: ID072.03 Growth abnormalities and macrocephaly: 30 genes (80,9 kb) AKT1, BRAF, CUL4B, DNMT3A, EED, EZH2, GPC3, GPC4, H1-4, HRAS, HUWE1, KRAS, NF1, NFIX, NRAS, NSD1, OFD1, PIK3CA, PPP1CB, PTEN, PTPN11, RAF1, RIT1, RNF125, RRAS2, SETD2, SHOC2, SOS1, SPRED1, SUZ12 Overgrowth syndrome with macrocephaly: 11 genes (33,0 kb) DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, RNF125, SETD2, SUZ12 Noonan syndrome with macrocephaly: 10 genes (15,2 kb) BRAF, KRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1	3 - 5 w	E
Growth hormone deficiency (IGHD, CPHD) * Gene panel: ID211.01 Growth hormone deficiency (IGHD, CPHD): 15 genes (28,2 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A, SOX3 Isolated growth hormone deficiency (IGHD): 5 genes (6,4 kb) BTK, GH1, GHRHR, GHSR, SOX3 Combined pituitary hormone deficiency (CPHD): 10 genes (21,8 kb) GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A	3 - 5 w	E
Hyperphosphatasia with impaired intellectual development syndrome (HPMRS) * Gene panel: ID292.00, 6 genes (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	2 - 4 w	E
Intellectual developmental disorder, autosomal recessive (MRT, IDD) * Gene panel: ID037.03 Intellectual developmental disorder, autosomal-recessive (MRT, IDD): 77 Gene (201,7 kb) ABCA2, ACTL6B, ADAT3, ALG14, ALKBH8, ANK3, APC2, ASCC3, CAMK2A, CASP2, CC2D1A, CEP104, CPE, CRADD, CRBN, DEAF1, EDC3, EIF3F, ELP2, FBXL3, FBXO31, FERRY3, FMN2, GNB5, GRIA1, GRIK2, HERC2, HNMT, IMPA1, IQSEC1, KDM5B, KPTN, LGI3, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NEMF, NSUN2, NSUN6, NUDT2, OTUD6B, PDE2A, PDZD8, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, PUS7, RSRC1, RUSC2, SCAPER, SLC45A1, SLC6A17, ST3GAL3, TAF13, TAF2, TECR, TMEM94, TNIK, TPR, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, WIPI2, ZBTB11, ZC3H14 Intellectual developmental disorder, autosomal-recessive, nonsyndromic (MRT): 58 Gene (157,9 kb) ADAT3, ALKBH8, ANK3, APC2, ASCC3, CAMK2A, CASP2, CC2D1A, CEP104, CRADD, CRBN, EDC3, EIF3F, ELP2, FBXO31, FERRY3, FMN2, GRIA1, GRIK2, HERC2, HNMT, IMPA1, KDM5B, KPTN, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NSUN2, NSUN6, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, RSRC1, RUSC2, SLC6A17, ST3GAL3, TAF2, TAF13, TECR, TNIK, TPR, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, ZBTB11, ZC3H14 Intellectual developmental disorder, autosomal-recessive, syndromic (IDD): 20 Gene (48,5 kb) ABCA2, ALG14, CPE, DEAF1, FBXL3, GNB5, IQSEC1, LGI3, NEMF, NUDT2, OTUD6B, PDE2A, PDZD8, PUS7, SCAPER, SLC45A1, TAF2, TMEM94, WIPI2, ZBTB11	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
Intellectual developmental disorder, autosomal dominant (MRD, IDD) * Gene panel: ID036.06 Intellectual developmental disorder, autosomal dominant (MRD, IDD): 105 genes (354,6 kb) ACTL6B, ADNP, AHDC1, AP2M1, ARID1A, ARID1B, ARID2, ASH1L, ATP2B1, AUTS2, BCL11A, BCL11B, BICRA, BRPF1, CACNG2, CAMK2A, CAMK2B, CAMK2G, CCNK, CDH15, CDK8, CERT1, CHAMP1, CHD3, CHD8, CIC, CLTC, CNOT2, CNOT3, CTCF, CTNNB1*, DDX6, DEAF1, DHX9, DLG4, DPF2, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, FBXO11, FOXP1, GATAD2B, GNB1, GRIA1, GRIN1, GRIN2B, HIVEP2, HNRNPC, KAT6A, KCNQ5, KDM3B, KDM4B, KIF1A, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MEF2C, MTSS2, MYT1L, NAA15, NR4A2, NUS1, PACS1, PAK1, PHF21A, POGZ, PPP2R1A, PPP2R5D, PURA*, RAC1, RFX7, RORA, SET, SETBP1, SETD1B, SETD2, SETD5, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX11, SOX4, SOX6, SRRM2, STAG1, SYNGAP1, TAF4, TANC2, TBL1XR1, TBR1, TCP1, TLK2, TNPO2, TRIO, TRIP12, USP7, ZBTB18, ZMYND11, ZNF292 Intellectual developmental disorder, autosomal dominant, nonsyndromic (MRD): 62 genes (224,4 kb) AP2M1, ARID1A, ARID1B, ASH1L, ATP2B1, AUTS2, CACNG2, CAMK2A, CAMK2B, CAMK2G, CDH15, CERT1, CIC, CLTC, CTCF, DEAF1, DHX9, DLG4, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, GATAD2B, GNB1, GRIA1, GRIN2B, HNRNPC, HIVEP2, KCNQ5, KDM4B, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MYT1L, NAA15, NUS1, PACS1, POGZ, PPP2R1A, PPP2R5D, RAC1, RFX7, SET, SETBP1, SETD2, SETD5, SMARCA4, SMARCB1, SOX11, SRRM2, STAG1, SYNGAP1, TAF4, TBL1XR1, TLK2, TRIO, ZBTB18, ZMYND11, ZNF292 Intellectual developmental disorder, autosomal dominant, syndromic (IDD): 55 genes (168,0 kb) ACTL6B, ADNP, AHDC1, ARID1A, ARID1B, ARID2, BCL11A, BCL11B, BICRA, BRPF1, CCNK, CDK8, CERT1, CHAMP1, CHD3, CHD8, CNOT2, CNOT3, CTNNB1, DDX6, DEAF1, DPF2, FBXO11, FOXP1, GATAD2B, GRIN1, KAT6A, KDM3B, KIF1A, MEF2C, MTSS2, NR4A2, PACS1, PAK1, PHF21A, POGZ, PPP2R1A, PPP2R5D, PURA, RORA, SETD1B, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX11, SOX4, SOX6, TANC2, TBR1, TCP1, TNPO2, TRIP12, USP7	4 - 6 w	E
Intellectual developmental disorder, X-linked (XLID, MRX, MRXS) * Gene panel: ID038.06 Intellectual developmental disorder, X-linked (XLID, MRX, MRXS): 77 genes (194,6 kb) ACSL4, AFF2, AP1S2, ARX, ATP6AP2, ATRX, BRWD3, CASK, CLCN4, CLIC2, CNKSR2, CSTF2, CUL4B, DDX3X, DLG3, EIF2S3, FAM50A, FGD1, FGF13, FMR1, FRMPD4, FTSJ1, GDI1, GLRA2, GRIA3, HCFC1, HNRNPH2, HS6ST2, HUWE1, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIF4A, KLHL15, LAS1L, MECP2, MED12, MID2, MSL3, NEXMIF, NKAP, NONO, OGT, OPHN1, PAK3, PHF6, PHF8, POLA1, PQBP1, PRPS1, RAB39B, RBMX, RLIM, RPL10, RPS6KA3, SLC16A2, SLC9A6, SLC9A7, SLITRK2, SMS, STEEP1, SYN1, SYP, TAF1, THOC2, TSPAN7, UBE2A, UPF3B, USP9X, USP27X, WNK3, ZMYM3, ZC4H2, ZDHHC9, ZFX, ZNF711 Intellectual developmental disorder, X-linked, nonsyndromic (XLID, MRX): 29 genes (77,8 kb) ACSL4, AFF2, ARX, BRWD3, DLG3, FGF13, FRMPD4, FTSJ1, GDI1, HCFC1, IL1RAPL1, IQSEC2, KIF4A, KLHL15, MID2, NEXMIF, OGT, PAK3, RAB39B, RPS6KA3, SLC9A7, STEEP1, SYN1, SYP, THOC2, TSPAN7, USP9X, USP27X, ZNF711 Intellectual developmental disorder, X-linked, syndromic (MRXS): 52 genes (130,8 kb) AFF2, AP1S2, ARX, ATP6AP2, ATRX, CASK, CLCN4, CLIC2, CNKSR2, CSTF2, CUL4B, DDX3X, EIF2S3, FAM50A, FGD1, FMR1, GLRA2, GRIA3, HNRNPH2, HS6ST2, HUWE1, IGBP1, KDM5C, LAS1L, MECP2, MED12, MSL3, NKAP, NONO, OPHN1, PHF6, PHF8, POLA1, PQBP1, PRPS1, RAB39B, RBMX, RLIM, RPL10, SLC16A2, SLC9A6, SLITRK2, SMS, TAF1, UBE2A, UPF3B, USP9X, ZC4H2, WNK3, ZMYM3, ZDHHC9, ZFX	4 - 6 w	E
Intellectual developmental disorder and macrocephaly * Gene panel: ID131.03 Intellectual developmental disorder and macrocephaly: 48 genes (147,3 kb) ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMC01, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9 Intellectual developmental disorder, autosomal dominant (MRD) and macrocephaly: 21 genes (70,8 kb) CAMK2G, CHD3, CHD8, DEAF1, GATAD2B, HEPACAM, MTOR, NFIB, PAK1, PHF21A, PPP2R5D, PTEN, RAC1, RNF125, SETD2, SHANK3, SPOP, TRIO, TRIP12, ZBTB7A, ZBTB20 Intellectual developmental disorder, autosomal recessive (MRT) and macrocephaly: 10 genes (20,2 kb) ADK, ALKBH8, APC2, CRADD, KIF7, KPTN, MLC1, TBC1D7, TMC01, ZBTB7A Intellectual developmental disorder, X-linked (MRX) and macrocephaly: 18 genes (57,9 kb) BRWD3, CUL4B, DDX3X, FMR1, GRIA3, HUWE1, IGBP1, KDM5C, L1CAM, MECP2, MED12, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B, ZDHHC9	4 - 6 w	E
Intellectual developmental disorder and microcephaly * Gene panel: ID129.02 Intellectual developmental disorder and microcephaly: 80 genes (229,8 kb) ACSL4, ADAT3, ATRX, AUTS2, CAMK2B, CAMK2G, CASK, CERT1, CHAMP1, CKAP2L, CTCF, CTNNB1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCFC1, HIVEP2, HNMT, HNRNPH2, HUWE1, IQSEC1, KDM5C, KIF11, KIF1A, L1CAM, LINGO1, LINS1, MBD5, MBOAT7, MCPH1, MECP2, METTL5, MYCN, NEXMIF, NSUN2, OGT, PAK3, PGAP1, PHF6, POGZ, POLA1, PPP2R1A, PQBP1, PUS3, RAC1, RBBP8, RLIM, RPL10, SET, SETD2, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TAF1, TAF13, TAF2, THOC2, TLK2, TRAPPC9, TRIO, TRMT1, TTI2, WDR11, WDR73, ZBTB18, ZC4H2, ZEB2 Intellectual developmental disorder, autosomal dominant (MRD) and microcephaly: 32 genes (100,5 kb) AUTS2, CAMK2B, CAMK2G, CERT1, CHAMP1, CTCF, CTNNB1, DPP6, DYRK1A, EFTUD2, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5, MYCN, POGZ, PPP2R1A, RAC1, SET, SETD2, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TLK2, TRIO, ZBTB18, ZEB2 Intellectual developmental disorder, autosomal recessive (MRT) and microcephaly: 21 genes (41,0 kb) ADAT3, CKAP2L, EDC3, GPT2, HNMT, LINGO1, LINS1, MBOAT7, MCPH1, METTL5, NSUN2, PGAP1, PUS3, RBBP8, TAF13, TAF2, TRAPPC9, TRMT1, TTI2, WDR11, WDR73 Intellectual developmental disorder, X-linked (MRX) and microcephaly: 27 genes (88,3 kb) ACSL4, ATRX, CASK, DDX3X, EIF2S3, HCFC1, HNRNPH2, HUWE1, IQSEC1, KDM5C, L1CAM, MECP2, NEXMIF, OGT, PAK3, PHF6, POLA1, PQBP1, RLIM, RLIM, SHROOM4, SLC16A2, SLC6A8, SLC9A6, TAF1, THOC2, ZC4H2	4 - 6 w	E
Kabuki syndrome (KABUK) * Gene panel: ID127.00, 2 genes (20,8 kb) KDM6A, KMT2D	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
Neurodevelopmental disorders (NED), comprehensive diagnostics * Gene panel: ID358.00, 169 genes (431,8 kb) ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6VOA1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC2, EXOC7, EXOC8, FBXW11, FDF1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNP1, HNRNP, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, LNP, MADD, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAP1, RBL2, RERE, SARS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMP1, TIAM1, TMEM147, TMEM222, TMX2, TNF, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WFS1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	4 - 6 w	E
Noonan syndrome (NS) * Gene panel: ID023.06, 16 genes (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2	3 - 5 w	E
Obesity * Gene panel: ID183.03 Obesity: 57 genes (139,3 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FTO, GNAS, IFT27, IFT74, IFT172, INPP5E, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NROB2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WPCP Obesity, nonsyndromic: 18 genes (26,0 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NROB2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Obesity, syndromic: 40 genes (113,8 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, GNAS, IFT172, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WPCP Bardet-Biedl syndrome (BBS): 22 genes (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WPCP	4 - 6 w	E
Overgrowth, comprehensive diagnostics * Gene panel: ID299.01, 64 genes (211,3 kb) ABCC9, AKT1, AKT2, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DICER1, DIS3L2, DNMT3A, EED, EZH2, FBN1, FIBP, GFAP, GLI3, GPC3, GPC4, GRIA3, H1-4, HEPACAM, HERC1, HUWE1, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, MTOR, NFIX, NONO, NPR2, NSD1, OFD1, PDGFRB, PHF21A, PIGA, PIK3CA, PIK3R2, PPP2R5C, PPP2R5D, PTCH1, PTEN, RAB39B, RNF125, RNF135, SETD2, SHANK3, STRADA, SUFU, SUZ12, SYN1, TBC1D7, TCF20, TMEM94, TRIP12, UPF3B, ZBTB20, ZDHHC9	4 - 6 w	E
Overgrowth syndromes (SOTOS, BWS) * Gene panel: ID073.05 Overgrowth syndromes (SOTOS, BWS): 15 genes (41,2 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, PDGFRB, PTEN, SETD2, SUZ12, RNF125 Sotos-like overgrowth syndromes: 6 genes (23,1 kb) EED, EZH2, NFIX, NSD1, SETD2, SUZ12 Beckwith-Wiedemann-like overgrowth syndromes: 5 genes (10,1 kb) CDKN1C, DIS3L2, GPC3, GPC4, OFD1	3 - 5 w	E
Pitt-Hopkins syndrome (PTHS) * Gene panel: ID106.00, 3 genes (10,6 kb) CNTNAP2, NRXN1, TCF4	3 - 5 w	E
Progeria and progeroid syndromes * Gene panel: ID147.01, 25 genes (61,9 kb) ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24	3 - 5 w	E
RAS-related disorders * Gene panel: ID015.05 RAS-related disorders: 21 genes (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Noonan syndrome (NS): 15 genes (24,7 kb) BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Cardiofaciocutaneous syndrome (CFC): 4 genes (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 LEOPARD syndrome (LPRD): 3 genes (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 w	E
Rett syndrome (RTT) and similar syndromes * Gene panel: ID125.01, 16 genes (45,3 kb) CDKL5, FOXG1, GABBR2, GABRA2, GABRB2, GRIN2B, HTT, IQSEC2, MECP2, MEF2C, NTNG2, SHANK3, STXBP1, TCF4, UBE3A, WDR45	3 - 5 w	E
Rubinstein-Taybi syndrome (RSTS) * Gene panel: ID142.01, 3 genes (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
Seckel syndrome (SCKL) * Gene panel: ID113.00, 9 genes (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP	3 - 5 w	E
Short stature, comprehensive diagnostics * Gene panel: ID340.02 Short stature, comprehensive diagnostics: 207 genes (511,0 kb) ACAN, ACP5, ACTB, ACTG1, AGPS, ALPL, AMMECR1, ANKRD11, ARCN1, ARSB, ATR, B3GALT6, B3GAT3, B4GALT7, BCS1L, BGN, BLM, BMP2, BMPR1B, BRAF, BRCA1, BRCA2, BRIP1, BTK, CBL, CCDC8, CDC45, CDC6, CDKN1C, CDT1, CENPJ, CEP152, CEP63, CFAP410, COL10A1, COL11A2, COL27A1, COL2A1, COMP, CREBBP, CRIPT, CSGALNACT1, CUL7, DDR2, DDRGK1, DHCR7, DNA2, DONSON, DPH1, EP300, ERCC4, ERCC6, ERCC8, EXOC6B, EXOSC2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGD1, FGFR1, FGFR3, FLNB, FN1, GALNS, GDF5, GH1, GHR, GHRHR, GHSR, GLB1, GLI2, GMNN, GNPAT, GPX4, GRHL2, GSC, GUSB, GZF1, HDAC8, HESX1, HMG2, HRAS, HYAL1, IDUA, IGF1, IGF1R, IGF2, IGFALS, INSR, IRS1, IRS4, KIF22, KMT2A, KRAS, LARP7, LFNG, LHX3, LHX4, LTBP3, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MCM5, MRAS, NBAS, NBN, NEPRO, NIN, NIPBL, NKX2-5, NKX3-2, NOTCH2, NPR2, NRAS, NSMCE2, OBSL1, ORC1, ORC4, ORC6, OTX2, PALB2, PAM16, PAPP2, PAX8, PCNT, PEX5, PEX7, PHEX, PIK3R1, PISD, PLAG1, PLCB3, POC1A, POLR3GL, POP1, POU1F1, PPP1CB, PPP1R15B, PPP3CA, PRKG2, PRMT7, PROP1, PTH1R, PTPN11, PUS7, RAD21, RAD51, RAD51C, RAF1, RBBP8, RFWD3, RIT1, RMRP, RNPC3, RNU4ATAC, ROBO1, RPL13, RRAS2, RSPRY1, RTTN, SCUBE3, SGMS2, SHOC2, SHOX, SLC10A7, SLC26A2, SLX4, SMARCA2, SMC1A, SMC3, SOS1, SOS2, SOX2, SOX3, SPRED2, SRCAP, STAT5B, TALDO1, TBCE, TBL1X, TBX15, TBX19, TBX2, THRA, TKT, TONSL, TOP3A, TRAIP, TRAPPC2, TRHR, TRIM37, TRIP11, TRMT10A, TRPV4, TSHB, TSHR, UBE2T, XRCC2, XRCC4 Growth hormone deficiency (IGHD, CPHD): 14 genes (23,0 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1, SOX3 Noonan syndrome (NS): 16 genes (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Meier-Gorlin syndrome (MGORS): 8 genes (12,6 kb) CDC45, CDC6, CDT1, GMNN, MCM5, ORC1, ORC4, ORC6 Seckel syndrome (SCKL): 9 genes (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP Congenital hypothyroidism (CHNG): 8 genes (13,0 kb) IRS4, NKX2-5, PAX8, TBL1X, THRA, TRHR, TSHB, TSHR Skeletal dysplasia (SED, SMD, AMD): 35 genes (83,9 kb) ACAN, B3GALT6, BGN, BMPR1B, CFAP410, COL11A2, COL2A1, COMP, DDRGK1, DDR2, EXOC6B, FGFR3, FN1, GDF5, GPX4, KIF22, NEPRO, NKX3-2, NPR2, PAM16, PAPSS2, PISD, PLCB3, RMRP, RNU4ATAC, POP1, PRKG2, RPL13, RSPRY1, SIK3, SLC26A2, TONSL, TRAPPC2, TRIP11, TRPV4	4 - 6 w	E
Three M syndrome (3M) * Gene panel: ID214.00, 3 genes (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 w	E
Ear, Nose and Throat Diseases		
Branchiootorenal syndrome (BOR) * Gene panel: ID315.00, 5 genes (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A	3 - 5 w	E
Nonsyndromic deafness, comprehensive diagnostics * Gene panel: ID237.03 Nonsyndromic deafness, comprehensive diagnostics: 133 genes (374,8 kb) ABCC1, ACTG1, ADCY1, AFG2B, AIFM1, ATOH1, ATP11A, ATP2B2, BDP1, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A1, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA4, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GPRASP2, GRAP, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, HOMER2, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MAP1B, MARVELD2, MCM2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PDZD7, PI4KB, PJVK, PKHD1L1, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SLC12A2, SLC17A8, SLC26A4, SLC26A5, SLC44A4, SMPX, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, THOC1, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, USP48, WBP2, WFS1, WHRN Nonsyndromic deafness, autosomal dominant (DFNA): 60 genes (180,1 kb) ABCC1, ACTG1, ATOH1, ATP11A, ATP2B2, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MAP1B, MCM2, MT-RNR1, MYH14, MYH9, MYO3A, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PI4KB, PLS1, POU4F3, PTPRQ, REST, RIPOR2, SCD5, SLC12A2, SIX1, SLC17A8, SLC44A4, TBC1D24*, TECTA, THOC1, TMC1, TNC, TRRAP, USP48, WFS1 Nonsyndromic deafness, autosomal recessive (DFNB): 83 genes (229,9 kb) ADCY1, AFG2B, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJVK, PKHD1L1, PPIP5K2, PNPT1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SPNS2, STRC, STX4, SYNE4, TBC1D24*, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN Nonsyndromic deafness, X-linked (DFNX): 6 genes (11,8 kb) AIFM1, COL4A6, GRASP2, POU3F4, PRPS1, SMPX	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Ear, Nose and Throat Diseases		
Nonsyndromic deafness, autosomal dominant (DFNA) * Gene panel: ID091.04, 60 genes (180,1 kb) ABCC1, ACTG1, ATOH1, ATP11A, ATP2B2, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPHA10, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MAP1B, MCM2, MT-RNR1, MYH14, MYH9, MYO3A, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PI4KB, PLS1, POU4F3, PTPRQ, REST, RIPOR2, SCD5, SLC12A2, SIX1, SLC17A8, SLC44A4, TBC1D24, TECTA, THOC1, TMC1, TNC, TRRAP, USP48, WFS1	4 - 6 w	E
Nonsyndromic deafness, autosomal recessive (DFNB) * Gene panel: ID092.04, 83 genes (229,9 kb) ADCY1, AFG2B, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GAS2, GIPC3, GJB2, GJB3, GJB6, GPR156, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MINAR2, MPZL2, MSRB3, MT-RNR1, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PJVK, PKHD1L1, PPIP5K2, PNPT1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SPNS2, STRC, STX4, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TMTC4, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN	4 - 6 w	E
Nonsyndromic deafness, X-linked (DFNX) * Gene panel: ID290.01, 8 genes (17,1 kb) AIFM1, COL4A5, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX, TIMM8A	3 - 5 w	E
Hypogonadotropic hypogonadism with or without anosmia (KAL, HH) * Gene panel: ID170.04, 37 genes (62,5 kb) ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 w	E
Perrault syndrome (PRLTS) * Gene panel: ID388.00, 9 genes (14,1 kb) CLPP, DAP3, ERAL1, HARS2, HSD17B4, LARS2, MRPL49, PRORP, TWNK	3 - 5 w	E
Syndromic deafness, comprehensive diagnostics * Gene panel: ID190.01 Syndromic deafness, comprehensive diagnostics: 109 genes (317,3 kb) ABHD12, ADGRV1, AFG2A, AIFM1, ALMS1, ANKH, ARSG, ATP1A3, ATP6V0A4, ATP6V1B1, BCAP31, BCS1L, BRAF, BSND, CACNA1D, CATSPER2, CD151, CDH23, CHD7, CHSY1, CIB2, CISD2, CLPP, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL9A1, COL9A2, COL9A3, DCAF17, DIAPH3, DLX5, DNMT1, DSPP, EDN3, EDNRB, ERAL1, ESPN, EXOSC2, EYA1, FGF3, FGFR3, FOXC1, FOXI1, GATA3, GJA1, GJB2, GPSM2, HARS1, HARS2, HSD17B4, KCNE1, KCNJ10, KCNQ1, KITLG, LARS2, LHX3, LRP2, MAF, MANBA, MITF, MPZ, MYH14, MYH9, MYO7A, NLRP3, PAX3, PCDH15, PDZD7, PEX1, PEX6, PLOD3, PMP22, POLD1, POLR1C, POLR1D, PRDM5, PRPS1, PTPN11, RAF1, RPGR, RPS6KA3, SALL1, SALL4, SEMA3E, SIX1, SIX5, SLC19A2, SLC26A4, SLC4A11, SLC52A2, SLC52A3, SNAI2, SOX10, TCOF1, TFAP2A, TIMM8A, TWNK, TYR, USH1C, USH1G, USH2A, WFS1, WHRN, ZNF469 Usher syndrome (USH): 13 genes (70,3 kb) ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USHC, USH1G, USH2A, WHRN Stickler syndrome (STL): 6 genes (21,2 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2 Alport syndrome (ATS): 4 genes (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9 Waardenburg syndrome (WS): 8 genes (9,4 kb) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Perrault syndrome (PRLTS): 6 genes (9,3 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK LEOPARD syndrome (LPRD): 3 genes (6,0 kb) BRAF, PTPN11, RAF1 CHARGE syndrome: 2 genes (11,3 kb) CHD7, SEMA3E	4 - 6 w	E
Usher syndrome (USH) * Gene panel: ID034.01 Usher syndrome (USH): 13 genes (70,3 kb) ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN Usher syndrome, type I (USH1): 6 genes (26,2 kb) CDH23, CIB2, MYO7A, PCDH15, USH1C, USH1G Usher syndrome, type II (USH2): 4 genes (40,4 kb) ADGRV1, PDZD7, USH2A, WHRN Usher syndrome, type III and type IV (USH3, USH4): 3 genes (3,8 kb) ARSG, CLRN1, HARS1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Endocrine Disorders		
Adrenogenital syndrome (AGS, CAH) * Gene panel: ID111.02, 7 genes (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 w	E
Congenital hypothyroidism * Gene panel: ID369.00 Congenital hypothyroidism: 37 genes (69,0 kb) CDCA8, DIO1, DUOX2, DUOXA2, FOXE1, GLIS3, GNAS, HESX1, IGSF1, IRS4, IYD, LHX3, LHX4, NKX2-1, NKX2-5, OTX2, PAX8, POU1F1, PRKAR1A, PROP1, RNPC3, ROBO1, SECISBP2, SLC16A2, SLC26A4, SLC26A7, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TRH, TRHR, TSHB, TSHR, TUBB1 Congenital nongoitrous hypothyroidism (CHNG): 8 genes (13,0 kb) IRS4, NKX2-5, PAX8, TBL1X, THRA, TRHR, TSHB, TSHR Thyroid dysmorphogenesis (TDH): 8 genes (23,8 kb) DUOX2, DUOXA2, IYD, SLC5A5, SLC26A4, SLC26A7, TG, TPO Combined pituitary hormone deficiency (CPHD): 8 genes (11,9 kb) HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1	3 - 5 w	E
Diabetes insipidus * Gene panel: ID322.00, 5 genes (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 w	E
Disorder of sex development (DSD) * Gene panel: ID117.03 Disorder of sex development (DSD): 49 genes (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NROB1, NR2F2, NR5A1,, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Disorder of sex development (SRXY), nonsyndromic: 21 genes (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NROB1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Disorder of sex development (SRXX), nonsyndromic: 6 genes (6,6 kb) NR2F2, NR5A1, RSP01, SOX9, SRY, WNT4 Disorder of sex development (DSD), syndromic: 37 genes (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NROB1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX9, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenital syndrome (AGS, CAH): 7 genes (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 w	E
Endocrine hypertension * Gene panel: ID270.03 Endocrine hypertension: 33 genes (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronism (HALD): 5 genes (19,1 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronism type II (PHA2): 4 genes (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Pheochromocytoma/paraganglioma syndrome (PPGL): 11 genes (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenal cushing syndrome (PPNAD): 4 genes (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle syndrome (LIDLS): 3 genes (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertension with low renin level: 20 genes (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 w	E
Glucocorticoid deficiency (GCCD) * Gene panel: ID222.00, 16 genes (25,1 kb) AAAS, ABCD1, CYP11A1, HSD3B2, MC2R, MCM4, MRAP, NNT, NROB1, NR3C1, PCSK1, POMC, PROP1, STAR, TBX19, TXNRD2	3 - 5 w	E
Growth hormone deficiency (IGHD, CPHD) * Gene panel: ID211.01 Growth hormone deficiency (IGHD, CPHD): 15 genes (28,2 kb) BTK, GH1, GHRHR, GHSR, GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A, SOX3 Isolated growth hormone deficiency (IGHD): 5 genes (6,4 kb) BTK, GH1, GHRHR, GHSR, SOX3 Combined pituitary hormone deficiency (CPHD): 10 genes (21,8 kb) GLI2, HESX1, IGSF10, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, SEMA3A	3 - 5 w	E
Hyperaldosteronism (HALD) * Gene panel: ID304.00, 6 genes (20,6 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Endocrine Disorders		
Hyperinsulinemic hypoglycemia (HHF) * Gene panel: ID126.00, 8 genes (16,9 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A	3 - 5 w	E
Hyperparathyroidism (HRPT) * Gene panel: ID338.00, 10 genes (19,2 kb) AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET, SLC12A1, TRPV6	3 - 5 w	E
Hypoglycemia, hyperinsulinism and ketone metabolism * Gene panel: ID280.00 Hypoglycemia, hyperinsulinism and ketone metabolism: 44 genes (85,9 kb) ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4 Glycogen storage disease (GSD): 24 genes (48,4 kb) AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4 Hyperinsulinemic hypoglycemia (HHF): 9 genes (18,8 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A	3 - 5 w	E
Hypogonadotropic hypogonadism with or without anosmia (KAL, HH) * Gene panel: ID170.04, 37 genes (62,5 kb) ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 w	E
Hypoparathyroidism * Gene panel: ID353.00, 16 genes (24,8 kb) AIRE, CASR, CYP24A1, FAM111A, GATA3, GCM2, GNA11, GNAS, HADHA, HADHB, PTH, SLC34A1, SOX3, STX16, TBCE, TBX1	3 - 5 w	E
Mature-onset diabetes of the young (MODY) * Gene panel: ID048.01, 14 genes (22,9 kb) ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	3 - 5 w	E
Neuroendocrine neoplasia * Gene panel: ID386.00, 19 genes (29,2 kb) AIP, CDC73, CDKN1B, DLST, FH, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL	3 - 5 w	E
Neonatal diabetes mellitus * Gene panel: ID162.01 Neonatal diabetes mellitus: 29 genes (53,6 kb) ABCC8, BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57 Permanent neonatal diabetes mellitus (PNDM): 10 genes (15,9 kb) ABCC8, FOXP3, GCK, INS, KCNJ11, MNX1, NEUROD1, SLC19A2, SLC2A2, ZFP57 Syndromal neonatal diabetes mellitus: 21 genes (40,5 kb) BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5	3 - 5 w	E
Obesity * Gene panel: ID183.03 Obesity: 57 genes (139,3 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FTO, GNAS, IFT27, IFT74, IFT172, INPP5E, KIDINS220, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NROB2, NTRK2, PCSK1, PGM2L1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Obesity, nonsyndromic: 18 genes (26,0 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NROB2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Obesity, syndromic: 40 genes (113,8 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, GNAS, IFT172, IFT27, IFT74, INPP5E, KIDINS220, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PGM2L1, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl syndrome (BBS): 22 genes (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 w	E
Ovarian dysgenesis (ODG) * Gene panel: ID293.01 Ovarian dysgenesis (ODG): 16 Gene (27,8 kb) BMP15, CLPP, ERAL1, ESR2, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 XX gonadal dysgenesis (ODG): 10 Gene (17,1 kb) BMP15, ESR2, FSHR, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX gonadal dysgenesis with deafness (PRLTS): 6 genes (10,7 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK	3 - 5 w	E
Pheochromocytoma/paraganglioma syndrome (PPGL) * Gene panel: ID042.03, 14 genes (22,8 kb) DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Endocrine Disorders		
Pituitary adenoma (PITA) * Gene panel: ID387.00, 8 genes (20,7 kb) AIP, CDH23, CDKN1B, GNAS, GPR101, MEN1, PRKAR1A, USP8	3 - 5 w	E
Premature ovarian failure (POF) * Gene panel: ID078.05 Premature ovarian failure (POF): 23 genes (51,1 kb) BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, INHA, LHCGR, MCM8, MSH5, NOBOX, NR5A1, POF1B, STAG3, SYCE1, XRCC2 Premature ovarian failure (POF), X-linked: 4 genes (8,2 kb) BMP15, DIAPH2, FMR1, POF1B Premature ovarian failure (POF), autosomal recessive: 10 genes (25,0 kb) C14ORF39, GDF9, FANCM, HFM1, HSF2BP, MCM8, MSH5, STAG3, SYCE1, XRCC2 Premature ovarian failure (POF), autosomal dominant: 6 genes (12,7 kb) BNC1, ERCC6, FIGLA, FOXL2, NOBOX, NR5A1	3 - 5 w	E
Pseudoaldosteronism (LIDLS) and Pseudohypoaldosteronism (PHA) * Gene panel: ID250.00 Pseudoaldosteronism (LIDLS) and Pseudohypoaldosteronism (PHA): 8 genes (23,8 kb) CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4 Pseudohypoaldosteronism, type I (PHA1): 4 genes (8,8 kb) NR3C2, SCNN1A, SCNN1B, SCNN1G Pseudohypoaldosteronism, type II (PHA2): 4 genes (15,7 kb) CUL3, KLHL3, WNK1, WNK4 Liddle syndrome (LIDLS): 3 genes (5,9 kb) SCNN1A, SCNN1B, SCNN1G	3 - 5 w	E
Septo optic dysplasia * Gene panel: ID378.00, 8 genes (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	3 - 5 w	E
Thyroid cancer * Gene panel: ID220.02, 26 genes (60,5 kb) ACD, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, POT1, PRKAR1A, PTEN, RET, SDHB, SDHD, SEC23B, SRGAP1, SRRM2, TINF2, TP53	3 - 5 w	E
Epilepsy and Migraine		
Absence epilepsy (JAE, CAE) * Gene panel: ID057.02, 10 genes (21,8 kb) CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, KCNMA1, RORB, SLC2A1, SLC12A5	3 - 5 w	E
Benign neonatal and infantile seizures (BFNS, BFIS) * Gene panel: ID134.01, 6 genes (19,8 kb) CHRNA2, KCNQ2, KCNQ3, PRRT2, SCN2A, SCN8A	3 - 5 w	E
Developmental and epileptic encephalopathy (DEE, EIEE) * Gene panel: ID080.03 Developmental and epileptic encephalopathy (DEE, EIEE): 105 genes (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Developmental and epileptic encephalopathy (DEE, EIEE), autosomal dominant: 51 genes (143,1 kb) ATP1A2, ATP1A3, ATP6V0A1, ATP6V1A, CACNA1A, CACNA1E, CDK19, CELF2, CHD2, CUX2, CYFIP2, DNM1, EEF1A2, FBXO28, FGF12, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GNAO1, GRIN2B, GRIN2D, HCN1, HNRNPU, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, NEUROD2, NSF, NTRK2, PACS2, PHACTR1, PPP3CA, RHOBTB2, RNF13, SCN1A, SCN2A, SCN3A, SCN8A, SIK1, SLC1A2, SPTAN1, STXBP1, YWHAG Developmental and epileptic encephalopathy (DEE, EIEE), autosomal recessive: 45 genes (105,0 kb) AARS1, ACTL6B, ADAM22, AP3B2, ARV1, CAD, CNPY3, CPLX1, DALRD3, DENND5A, DMXL2, DOCK7, FRRS1L, GAD1, GLS, GOT2, GRIN1, GUF1, HID1, ITPA, MDH1, MDH2, NECAP1, PARS2, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, SCN1B, SLC12A5, SLC13A5, SLC25A12, SLC25A22, SLC38A3, ST3GAL3, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX Developmental and epileptic encephalopathy (DEE, EIEE), X-linked: 9 genes (20,3 kb) ALG13, ARHGEF9, ARX, CDKL5, FGF13, PCDH19, PIGA, SLC35A2, SMC1A	4 - 6 w	E
Epilepsy with severe developmental disorder * Gene panel: ID060.00, 20 genes (68,4 kb) ARHGEF9, ARX, CACNA1A, CDKL5, FOXG1, KCNQ2, MECP2, MEF2C, MTHFR, PCDH19, SCN1A, SCN2A, SCN8A, SCN9A, SLC25A22, SLC2A1, SPTAN1, STXBP1, SYNGAP1, UBE3A	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Epilepsy and Migraine		
Epilepsy, comprehensive diagnostics * Gene panel: ID061.05 Epilepsy, comprehensive diagnostics: 163 genes (388,9 kb) AARS1, ACTL6B, ADAM22, ALDH7A1, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ASAH1, ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, BRAT1, CACNA1A, CACNA1E, CACNB4, CAD, CASR, CDK19, CDKL5, CELF2, CERS1, CHD2, CHRNA2, CHRNA4, CHRN2, CILK1, CLCN2, CLN8, CNPY3, CNTN2, CNTNAP2, CPA6, CPLX1, CSTB, CUX2, CYFIP2, DALRD3, DENND5A, DEPDC5, DMXL2, DNM1, DOCK7, EEF1A2, EFHC1, EPM2A, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GAL, GLS, GNAO1, GOT2, GOSR2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GUF1, HCN1, HCN2, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC1, KCNC2, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNT2, KCTD7, LGI1, LMNB2, MDH1, MDH2, MECP2, MEF2C, MTOR, NECAP1, NEUROD2, NHLRC1, NPRL2, NPRL3, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PNPO, POLG, PLPBP, PPP3CA, PRDM8, PRICKLE1, PRRT2, RELN, RHOTB2, RNF13, ROGDI, RORA, RORB, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC38A3, SLC6A1, SLC7A6OS, SMC1A, SNIP1, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Focal epilepsy: 17 genes (50,9 kb) CHRNA2, CHRNA4, CHRN2, CNTNAP2, CPA6, DEPDC5, GAL, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PCDH19, RELN, SCN3A, SRPX2, TBC1D24 Generalized epilepsy: 39 genes (72,6 kb) ALDH7A1, ASAH1, CACNB4, CASR, CERS1, CILK1, CLCN2, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, HCN1, HCN2, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP, POLG, PRDM8, PRICKLE1, RORB, SCARB2, SCN1A, SCN1B, SEMA6B, SLC2A1, SLC6A1, SLC7A6OS, SLC12A5, STX1B, TBC1D24 Epileptic encephalopathy (DEE, EIEE): 105 genes (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, MDH1, MDH2, NECAP1, NEUROD2, NSF, NTRK2, PACS2, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOTB2, RNF13, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG	4 - 6 w	E
Epileptic encephalopathy, comprehensive diagnostics * Gene panel: ID047.04 Epileptic encephalopathy, comprehensive diagnostics: 163 genes (371,9 kb) AARS1, ABAT, ACTL6B, ADAM22, ADAR, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, AP3B2, ARHGEF9, ARV1, ARX, ASNS, ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, BRAT1, BSCL2, BTB, CACNA1A, CACNA1E, CAD, CCDC88A, CDK19, CDKL5, CELF2, CHD2, CLCN4, CNPY3, CPLX1, CNTNAP2, CUX2, CYFIP2, D2HGDH, DALRD3, DENND5A, DNM1, DNM1L, DMXL2, DOCK7, EEF1A2, ETHE1, FGF12, FGF13, FOLR1, FOXG1, FBXO28, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GAMT, GCSH, GLDC, GLS, GNAO1, GOT2, GPHN, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, IDH2, IFIH1, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, LIAS, MECP2, MEF2C, MDH1, MDH2, MFF, MOCS1, MOCS2, MTHFR, NAXD, NAXE, NECAP1, NEUROD2, NRXN1, NSF, NTRK2, PACS2, PARS2, PC, PCDH19, PHACTR1, PHGDH, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PNPO, POLG, PPP3CA, PURA, RHOTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SERPINI1, SIK1, SLC1A2, SLC2A1, SLC12A5, SLC13A5, SLC6A8, SLC6A9, SLC9A6, SLC19A3, SLC25A1, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TCF4, TPK1, TRAK1, TREX1, UBA5, UGDH, UGP2, WDR45, WWOX, YWHAG Developmental and epileptic encephalopathy (DEE, EIEE): 105 genes (268,3 kb) AARS1, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX, ATP1A2, ATP1A3, ATP6VOA1, ATP6V1A, CACNA1A, CACNA1E, CAD, CDK19, CDKL5, CELF2, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DALRD3, DENND5A, DMXL2, DNM1, DOCK7, EEF1A2, FBXO28, FGF12, FGF13, FRRS1L, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GLS, GNAO1, GOT2, GRIN1, GRIN2B, GRIN2D, GUF1, HCN1, HID1, HNRNPU, ITPA, KCNA2, KCNB1, KCNC2, KCNQ2, KCNT1, KCNT2, LIAS, MECP2, MEF2C, MDH1, MDH2, MTHFR, NAXD, NAXE, NECAP1, NEUROD2, NRXN1, NSF, NTRK2, PACS2, PARS2, PC, PCDH19, PHACTR1, PHGDH, PIGA, PIGB, PIGP, PIGQ, PIGS, PLCB1, PNKP, PPP3CA, RHOTB2, RNASEH2A, RNASEH2B, RNASEH2C, RNF13, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SERPINI1, SIK1, SLC1A2, SLC2A1, SLC12A5, SLC13A5, SLC6A8, SLC6A9, SLC9A6, SLC19A3, SLC25A1, SLC25A12, SLC25A22, SLC35A2, SLC38A3, SMC1A, SPTAN1, ST3GAL3, STXBP1, SYNJ1, SZT2, TBC1D24, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Metabolic encephalopathy with epilepsy: 29 genes (44,8 kb) ABAT, ADSL, ALDH5A1, ALDH7A1, AMT, BTB, D2HGDH, FOLR1, GAMT, GCSH, GLDC, GPHN, IDH2, LIAS, MDH2, MOCS1, MOCS2, MTHFR, PC, PHGDH, PNPO, POLG, SLC1A2, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, TPK1	4 - 6 w	E
Febrile seizures with or without epilepsy (FEB, GEFS) * Gene panel: ID059.03 Febrile seizures with or without epilepsy (FEB, GEFS): 9 genes (35,9 kb) ADGRV1, CPA6, GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B Familial febrile seizures (FEB): 5 genes (30,3 kb) ADGRV1, CPA6, GABRG2, HCN2, SCN1A Generalized epilepsy with febrile seizures (GEFS): 7 genes (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 w	E
Focal epilepsy * Gene panel: ID208.01 Focal epilepsy: 17 genes (50,9 kb) CHRNA2, CHRNA4, CHRN2, CNTNAP2, CPA6, DEPDC5, GAL, GRIN2A, KCNT1, LGI1, NPRL2, NPRL3, PCDH19, RELN, SCN3A, SRPX2, TBC1D24 Focal epilepsy with variable foci (FFEVF): 4 genes (13,7 kb) DEPDC5, NPRL2, NPRL3, SCN3A Nocturnal frontal lobe epilepsy (ENFL): 5 genes (13,5 kb) CHRNA2, CHRNA4, CHRN2, DEPDC5, KCNT1 Temporal lobe epilepsy (ETL): 4 genes (13,4 kb) CPA6, GAL, LGI1, RELN	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Epilepsy and Migraine		
Generalized epilepsy * Gene panel: ID040.05 Generalized epilepsy: 39 genes (72,6 kb) ALDH7A1, ASAH1, CACNB4, CASR, CERS1, CILK1, CLCN2, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, HCN1, HCN2, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP, POLG, PRDM8, PRICKLE1, RORB, SCARB2, SCN1A, SCN1B, SEMA6B, SLC2A1, SLC6A1, SLC7A6OS, SLC12A5, STX1B, TBC1D24 Myoclonic epilepsy (EJM, EPM): 24 genes (45,3 kb) ASAH1, CERS1, CILK1, CLCN2, CNTN2, CSTB, CACNB4, EFHC1, EPM2A, GABRA1, GABRD, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SCN1A, SEMA6A, SLC7A6OS, TBC1D24 Absence epilepsy (EJA, ECA): 9 genes (18,3 kb) CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, RORB, SLC2A1, SLC12A5 Generalized epilepsy with febrile seizures (GEFSP): 7 genes (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 w	E
Generalized epilepsy with febrile seizures plus (GEFSP, GEFS+) * Gene panel: ID235.03, 8 genes (17,2 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, SLC32A1, STX1B	3 - 5 w	E
Hemiplegic migraine (FHM) * Gene panel: ID064.02, 7 genes (23,1 kb) ATP1A2, ATP1A3, CACNA1A, PRRT2, SCN1A, SLC1A3, SLC2A1	3 - 5 w	E
Metabolic disorder with epilepsy in childhood and adolescence * Gene panel: ID172.00, 15 genes (28,1 kb) ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA1, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2	3 - 5 w	E
Metabolic disorder with epilepsy in infancy and childhood * Gene panel: ID171.00, 18 genes (31,5 kb) ALDH5A1, ATP7A, BTBD, FOLR1, GAMT, GATM, HEXA, HEXB, HLCS, KCTD7, MTHFR, PHGDH, POLG, PPT1, SLC19A3, SLC2A1, SLC6A8, TPP1	3 - 5 w	E
Metabolic disorder with neonatal epilepsy * Gene panel: ID135.00, 25 genes (39,4 kb) ABAT, ADSL, ALDH7A1, AMT, BCKDHA, BCKDHB, CPS1, CTSD, DBT, DDC, DLD, DPYD, ETHE1, FH, GCSH, GLDC, GPHN, IVD, L2HGDH, MOCS1, MOCS2, OTC, PCCA, PCCB, PNPO	3 - 5 w	E
Metabolic epilepsy * Gene panel: ID303.01 Metabolic epilepsy: 84 genes (122,3 kb) ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTBD, CLN3, CLN5, CLN6, CLN8, CNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GM2A, GPHN, GRN, HEXA, HEXB, HIBCH, HLCS, IDH2, IVD, KCTD7, L2HGDH, LIAS, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, NEU1, OTC, PAH, PC, PCBD1, PCCA, PCCB, PGK1, PHGDH, PLPBP, PNPO, POLG, PPM1K, PPT1, PRODH, PTS, QDPR, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, SLC46A1, SUOX, TPK1, TPP1 Glycine encephalopathy (GCE): 5 genes (8,0 kb) AMT, GCSH, GLDC, LIAS, SLC6A9 Molybdenum cofactor deficiency (MOCOD): 3 genes (3,7 kb) GPHN, MOCS1, MOCS2 Cerebral creatine deficiency syndrome (CCDS): 3 genes (3,9 kb) GAMT, GATM, SLC6A8 Maple syrup urine disease (MSUD): 5 genes (6,6 kb) BCKDHA, BCKDHB, DBT, DLD, PPM1K 2-Hydroxyglutaric aciduria: 3 genes (5,3 kb) L2HGDH, D2HGDH, IDH2, SLC25A1 GM2-gangliosidosis: 3 genes (3,8 kb) HEXA, HEXB, GM2A Neuronal ceroid lipofuscinosis (CLN): 12 genes (14,3 kb) CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	4 - 6 w	E
Migraine (MGR) * Gene panel: ID065.01 Migraine (MGR): 16 genes (44,8 kb) ALPK1, ATP1A2, ATP1A3, CACNA1A, CSNK1D, ESR1, HTRA1, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1, TNF, TREX1 Familial hemiplegic migraine (FHM): 3 genes (16,6 kb) ATP1A2, CACNA1A, SCN1A	3 - 5 w	E
Myoclonic epilepsy (EPM, EJM) * Gene panel: ID116.03 Myoclonic epilepsy (EPM, EJM): 24 genes (45,3 kb) ASAH1, CERS1, CILK1, CLCN2, CNTN2, CSTB, CACNB4, EFHC1, EPM2A, GABRA1, GABRD, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SCN1A, SEMA6A, SLC7A6OS, TBC1D24 Juvenile myoclonic epilepsy (EJM): 12 genes (28,0 kb) CACNB4, CASR, CILK1, CLCN2, EFHC1, GABRA1, GABRD, RORB, SCN1A, SLC2A1, SLC12A5, TBC1D24 Progressive myoclonic epilepsy (EPM): 16 genes (24,0 kb) ASAH1, CERS1, CLN8, CSTB, EPM2A, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SEMA6B, SLC7A6OS	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Eye Diseases		
Achromatopsia (ACHM) * Gene panel: ID164.02, 6 genes (10,4 kb) ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H	3 - 5 w	E
Age-related macular degeneration (ARMD) * Gene panel: ID186.00, 16 genes (51,0 kb) ABCA4, APOE, ARMS2, C2, C3, C9, CFB, CFH, CFI, CST3, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1, RAX2	3 - 5 w	E
Albinism, comprehensive diagnostics * Gene panel: ID175.05 Albinism, comprehensive diagnostics: 33 genes (73,8 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DCT, DTNBP1, EDN3, EDNRB, EPG5, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1 Oculocutaneous albinism (OCA, OA): 9 genes (13,2 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Hermansky-Pudlak syndrome (HPS): 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6 Waardenburg syndrome (WS): 8 genes (9,3 kb) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Griscelli syndrome (GS): 3 genes (8,0 kb) MLPH, MYO5A, RAB27A	3 - 5 w	E
Anophthalmia and Microphthalmia (MCOP) * Gene panel: ID263.02 Anophthalmia and Microphthalmia (MCOP): 46 genes (107,6 kb) ABCB6, ALDH1A3, BCOR, BMP4, CHD7, CRYAA, FRAS1, FREM1, FREM2, FOXE3*, FZD5, GDF3, GDF6, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MFRP, MITF, NAA10, NHEJ1, OTX2, PAX2, PAX6, PITX3, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SHH, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, TMEM98, VAX1, VSX2, YAP1 Microphthalmia, isolated (MCOP): 6 genes (8,6 kb) ALDH1A3, GDF3, GDF6, MFRP, PRSS56, VSX2 Microphthalmia, syndromic (MCOPS): 13 genes (24,9 kb) BCOR, BMP4, HCCS, HMGB3, MAB21L2, NAA10, OTX2, SOX2, RARB, RAX, STRA6, TENM3, VAX1 Microphthalmia, with coloboma (MCOPCB): 11 genes (22,1 kb) ABCB6, GDF3, GDF6, FZD5, NHEJ1, PAX6, RBP4, SHH, STRA6, TENM3, VSX2 Syndromes with mikrophthalmia: 34 genes (90,4 kb) BCOR, BMP4, CHD7, CRYAA, FRAS1, FREM1, FREM2, FOXE3, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MITF, NAA10, OTX2, PAX2, PITX3, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TFAP2A, TMEM98, TENM3, VAX1, YAP1	4 - 6 w	E
Anterior segment dysgenesis (ASGD) * Gene panel: ID182.02 Anterior segment dysgenesis (ASGD): 11 genes (21,5 kb) B3GLCT, CPAMD8, CYP1B1, ELP4, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN, TRIM44 Peters anomaly: 7 genes (8,8 kb) B3GLCT, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3 Axenfeld-Rieger anomaly: 3 genes (3,6 kb) FOXC1, PAX6, PITX2 Aniridia (AN): 3 genes (3,6 kb) ELP4, PAX6, TRIM44	3 - 5 w	E
Cataract (CTRCT) * Gene panel: ID206.01 Cataract (CTRCT): 37 genes (59,7 kb) AGK, BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, DNMBP, EPHA2, FOXE3, FYCO1, GCNT2, GJA3, GJA8, HSF4, LEMD2, LIM2, LSS, MAF, MIP, NHS, PITX3, SIPA1L3, SLC16A12, TDRD7, UNC45B, VIM, WFS1 Cataract (CTRCT), autosomal dominant: 26 genes (29,2 kb) BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, EPHA2, GJA3, GJA8, HSF4, MAF, MIP, PITX3, SLC16A12, UNC45B, VIM, WFS1 Cataract (CTRCT), autosomal recessive: 15 genes (30,0 kb) AGK, BFSP1, CRYAA, CRYAB, CRYBB1, CRYBB3, DNMBP, FOXE3, FYCO1, GCNT2, LEMD2, LIM2, LSS, SIPA1L3, TDRD7 Cataract (CTRCT), X-linked: 1 gene (4,9 kb) NHS	3 - 5 w	E
Cone dystrophy (COD) and Cone-rod dystrophy (CORD) * Gen-Panel: ID101.03, 38 Gene (94,5 kb) ABCA4, ADAM9, AIPL1, ATF6, CACNA1F, CACNA2D4, CABP4, CDH3, CDHR1, CEP78, CEP250, CFAP418, CNGA3, CNGB3, CNM4, CRX, DRAM2, GNAT2, GUCA1A, GUCY2D, KCNV2, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RP1, RPGR, RPGRIP1, SEMA4A, TLCD3B, TLL5, UNC119	3 - 5 w	E
Congenital fibrosis of the extraocular muscles (CFEOM) * Gene panel: ID063.00, 5 genes (10,5 kb) COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3	3 - 5 w	E
Congenital stationary night blindness (CSNB) * Gene panel: ID267.00, 14 genes (39,2 kb) CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, SAG, RHO, SLC24A1, TRPM1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Eye Diseases		
Congenital nystagmus (NYS) * Gene panel: ID331.00 Congenital nystagmus (NYS): 31 genes (71,0 kb) AHR, ATF6, CACNA1F, CEP290, CNGA3, CNGB3, CRB1, CRX, DAGLA, DCT, FRMD7, GPR143, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, LRMDA, NMNAT1, NYX, OCA2, PAX6, ROBO1, RPE65, RPGRIP1, SLC24A5, SLC38A8, SLC45A2, TULP1, TYR, TYRP1 Nystagmus, nonsyndromic (NYS): 3 genes (8,3 kb) FRMD7, GPR143, ROBO1 Foveal hypoplasia, nonsyndromic (FVH): 3 genes (5,1 kb) AHR, PAX6, SLC38A8 Oculocutaneous albinism (OCA): 7 genes (11,1 kb) DCT, LRMDA, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Leber congenital amaurosis (LCA): 12 genes (29,5 kb) CEP290*, CRB1, CRX, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, RPE65, RPGRIP1, TULP1	4 - 6 w	E
Corneal dystrophy (CD) * Gene panel: ID329.01 Corneal dystrophy (CD): 27 genes (68,9 kb) AGBL1, CHST6, COL8A2, COL17A1, CYP4V2, DCN, GRHL2, GSN, KERA, KRT3, KRT12, LCAT, LOXHD1, MCOLN1, OVOL2, PAX6, PIKFYVE, PRDM5, SLC4A11, TACSTD2, TCF4, TGFB1, TUBA3D, UBIAD1, VSX1, ZEB1, ZNF469 Fuchs endothelial corneal dystrophy (FECD): 6 genes (20,2 kb) AGBL1, COL8A2, LOXHD1, SLC4A11, TCF4, ZEB1 Posterior polymorphous corneal dystrophy (PPCD): 4 genes (8,2 kb) COL8A2, GRHL2, OVOL2, ZEB1 Lattice corneal dystrophy (CDL): 3 genes (5,4 kb) GSN, TACSTD2, TGFB1 Stromal corneal dystrophy: 5 genes (11,6 kb) CHST6, DCN, PIKFYVE, TGFB1, UBIAD1 Epithelial corneal dystrophy: 6 genes (12,6 kb) COL17A1, KRT3, KRT12, MCOLN1, TACSTD2, TGFB1 Brittle cornea syndrome (BCN): 2 genes (13,8 kb) PRDM5, ZNF468 Keratoconus (KTCN): 2 genes (2,5 kb) TUBA3D, VSX1	3 - 5 w	E
Fraser syndrome (FRASRS) * Gene panel: ID317.00, 3 genes (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 w	E
Fuchs endothelial corneal dystrophy (FECD) * Gene panel: ID261.00, 5 genes (13,5 kb) AGBL1, COL8A2, SLC4A11, TCF4, ZEB1	3 - 5 w	E
Glaucoma (GLC) * Gene panel: ID275.01 Glaucoma (GLC): 27 genes (53,5 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CPAMD8, CYP1B1, FOXC1, FOXE3, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, PAX6, PITX2, PITX3, PXDN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMC01, WDR36 Primary glaucoma (GLC1, GLC3): 20 genes (37,8 kb) ASB10, ATOH7, CDKN2B, COL8A2, COL18A1, CYP1B1, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1, OPTN, RAMP2, SIX6, SLC4A11, TBK1, TEK, TMC01, WDR36 Anterior segment dysgenesis (ASGD): 8 genes (17,5 kb) CPAMD8, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN	3 - 5 w	E
Hermansky-Pudlak syndrome (HPS) * Gene panel: ID289.00, 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 w	E
High myopia (MYP) * Gene panel: ID079.03 High myopia (MYP): 25 genes (81,1 kb) ARR3, CACNA1F, CNGB3, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, CPSF1, GPR179, GRM6, GZF1, IRX5, LOXL3, LRPAP1, LRP2, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLITRK6, ZNF644 High myopia (MYP), nonsyndromic (MYP): 12 genes (24,5 kb) ARR3, CNGB3, CPSF1, LOXL3, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, ZNF644 Syndromes with high myopia: 14 genes (58,7 kb) CACNA1F, COL11A1, COL2A1, COL9A1, COL9A2, COL9A3, COL18A1, GPR179, GRM6, GZF1, LRP2, IRX5, P3H2, SLITRK6	3 - 5 w	E
Leber congenital amaurosis (LCA) * Gene panel: ID187.01, 19 genes (38,8 kb) AIP1, CEP290, CRB1, CRX, GDF6, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RD3, RDH12, RPE65, RPGRIP1, SPATA7, TULP1, USP45	3 - 5 w	E
Leber hereditary optic neuropathy (LHON) ^{CO} Gene panel: ID701.00, 17 genes (10,5 kb) MT-ATP6, MT-CO1, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-TE, MT-TL1, MT-TM, MT-TQ, MT-TT	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Eye Diseases		
Macular dystrophy (MD) * Gene panel: ID139.02, 18 genes (44,6 kb) ABCA4, BEST1, CDH3, CHST6, CNGB3, CRB1, CTNNA1, ELOVL4, IMPG1, IMPG2, MAPKAPK3, MFSD8, PROM1, PRPH2, RDH12, RP1L1, SIX6, TIMP3	3 - 5 w	E
Oculocutaneous albinism (OCA) * Gene panel: ID082.02, 9 genes (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 w	E
Optic atrophy (OPA) * Gene panel ID081.05 Optic atrophy (OPA): 38 genes (60,6 kb) ACO2, AFG3L2, ATP1A3, C19ORF12, CISD2, DNAJC30, DNM1L, EPRS1, FDXR, ISCA2, KLC2, MCAT, MECR, MFF, MFN2, MIEF1, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, RTN4IP1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, SSBP1, TBCE, TIMM8A, TMEM126A, UCHL1, WFS1, YME1L1, ZNHIT3 Optic atrophy (OPA), nonsyndromic: 12 genes (18,5 kb) ACO2, AFG3L2, DNM1L, MCAT, MECR, MIEF1, OPA1, OPA3, RTN4IP1, SSBP1, TMEM126A, YME1L1 Optic atrophy (OPA), syndromic: 29 genes (46,6 kb) ATP1A3, C19ORF12, CISD2, DNAJC30, EPRS1, FDXR, ISCA2, KLC2, MECR, MFF, MFN2, MTRFR, NBAS, NDUFA12, NR2F1, OPA1, OPA3, PDXK, PRPS1, SDHA, SLC25A46, SLC44A1, SLC52A2, SPG7, TBCE, TIMM8A, UCHL1, WFS1, ZNHIT3	3 - 5 w	E
Progressive external ophthalmoplegia with mtDNA deletions (PEOA, PEOB) * Gene panel: ID300.00 Progressive external ophthalmoplegia with mtDNA deletions (PEOA, PEOB): 10 genes (17,9 kb) DGUOK, DNA2, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, TK2, TOP3A, TWNK Progressive external ophthalmoplegia, autosomal dominant (PEOA): 6 genes (12,4 kb) DNA2, POLG, POLG2, RRM2B, SLC25A4, TWNK Progressive external ophthalmoplegia, autosomal recessive (PEOB): 5 genes (9,2 kb) DGUOK, POLG, RNASEH1, TK2, TOP3A	3 - 5 w	E
Retinal diseases, comprehensive diagnostics * Gene panel ID383.00, 306 genes (764,2 kb) ABCA4, ABCC6, ABHD12, ACBD5, ACO2, ADAM9, ADAMTS18, ADGRV1, AFG3L2, AGBL5, AHI1, AHR, AIPL1, AIRE, ALDH3A2, ALMS1, ALPK1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARSG, ATF6, ATOH7, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP164, CEP250, CEP290, CEP78, CERKL, CFAP20, CFAP410, CFAP418, CFH, CHM, CIB2, CLCC1, CLEC3B, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL18A1, COL2A1, COL4A1, COL9A1, COL9A2, COL9A3, COQ2, COQ8B, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTNND1, CTSD, CWC27, CYP4V2, DCT, DHDDS, DHX38, DNAJC5, DRAM2, DYNC2H1, EFEMP1, ELOVL4, ERCC6, ERCC8, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FSCN2, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS1, HCCS, HGSNAT, HK1, HKDC1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT43, IFT74, IKBKG, IMPDH1, IMPG1, IMPG2, INPP5E, IQCB1, KCNJ13, KCNV2, KIAA1549, KIF3B, KIF11, KIZ, KLHL7, LAMA1, LAMP2, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MAN2B1, MAPKAPK3, MCOLN1, MED12, MERTK, MFRP, MFSD8, MIR204, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MTPP, MVK, MYO7A, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPN1LW, OPN1MW, OTX2, P3H2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6G, PDZD7, PDE6H, PDSS1, PEX1, PEX2, PEX6, PEX7, PHYH, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, POMGNT2, POMT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PYGM, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS2, RLBP1, RNU4ATAC, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SAMD7, SCAPER, SDCCAG8, SEMA4A, SGSH, SLC24A1, SLC38A8, SLC6A6, SLC7A14, SNRNP200, SPATA7, SRD5A3, SSBP1, STN1, STX3, SUMF1, TIMM8A, TIMP3, TINF2, TLCDB3, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRAF3IP1, TREX1, TRIM32, TRNT1, TRPM1, TSPAN12, TTC8, TTLL5, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, UNC119, USH1C, USH1G, USH2A, USP45, VCAN, VPS13B, VWA8, WDCP, WDR19, WHRN, ZFYVE26, ZNF408, ZNF423, ZNF513	4 - 6 w	E
Retinitis pigmentosa (RP), comprehensive diagnostics * Gene panel: ID288.02 Retinitis pigmentosa (RP), comprehensive diagnostics: 87 genes (216,9 kb) ABCA4, AGBL5, AHR, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS2, BEST1, CC2D2A, CDHR1, CERKL, CFAP418, CHM, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, CRX, DHDDS, DHX38, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, HGSNAT, HK1, IDH3A, IDH3B, IFT140, IFT172, IFT43, HKDC1, IMPDH1, IMPG1, IMPG2, KIAA1549, KIF3B, KIZ, KLHL7, LRAT, MAK, MERTK, NEK2, NR2E3, NRL, OFD1, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, ROM1, RP1, RP1L1, RP2, RP9, RPE65, RPGR, SAG, SEMA4A, SLC7A14, SNRNP200, SPATA7, TOPORS, TTC8, TULP1, USH2A, ZNF408, ZNF513 Retinitis pigmentosa (RP), autosomal dominant: 28 genes (59,2 kb) AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPDH1, IMPG1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, RP1, RP9, RPE65, SAG, SEMA4A, SNRNP200, TOPORS Retinitis pigmentosa (RP), autosomal recessive: 61 genes (162,4 kb) ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, CC2D2A, CDHR1, CERKL, CFAP418, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, HKDC1, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, PRPH2, RAX2, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513 Retinitis pigmentosa (RP), X-linked: 4 genes (8,5 kb) CHM, RP2, RPGR, OFD1	4 - 6 w	E
Retinitis pigmentosa (RP), autosomal dominant * Gene panel: ID053.03, 29 genes (64,9 kb) AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPG1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RDH12, RGR, RHO, RP1, RP9, RPE65, SAG, SEMA4A, SNRNP200, TOPORS, VWA8	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Eye Diseases		
Retinitis pigmentosa (RP), autosomal recessive * Gene panel: ID050.04, 60 genes (161,3 kb) ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, CC2D2A, CDHR1, CERKL, CFAP418, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, HKDC1, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, RAX2, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, ZNF513	4 - 6 w	E
Senior-Loken syndrome (SLSN) * Gene panel: ID029.01, 8 genes (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 w	E
Septo optic dysplasia * Gene panel: ID378.00, 8 genes (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	3 - 5 w	E
Stargardt disease (STGD) * Gene panel: ID102.00, 4 genes (11,4 kb) ABCA4, ELOVL4, PROM1, PRPH2	3 - 5 w	E
Stickler syndrome (STL) * Gene panel: ID062.00, 6 genes (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 w	E
Usher syndrome (USH) * Gene panel: ID034.01 Usher syndrome (USH): 13 genes (70,3 kb) ADGRV1, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7, USH1C, USH1G, USH2A, WHRN Usher syndrome, type I (USH1): 6 genes (26,2 kb) CDH23, CIB2, MYO7A, PCDH15, USH1C, USH1G Usher syndrome, type II (USH2): 4 genes (40,4 kb) ADGRV1, PDZD7, USH2A, WHRN Usher syndrome, type III and type IV (USH3, USH4): 3 genes (3,8 kb) ARSG, CLRN1, HARS1	3 - 5 w	E
Vitreoretinopathy * Gene panel ID352.00 Vitreoretinopathy: 23 genes (58,9 kb) ATOH7, BEST1, CAPN5, COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, PAK2, RCBTB1, RS1, TSPAN12, VCAN, ZNF408 Exudative Vitreoretinopathy (EVR): 15 genes (34,9 kb) ATOH7, CAPN5, CTNNB1, BEST1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RS1, TSPAN12, VCAN, ZNF408 Syndromes with Vitreoretinopathy (STL, KNO): 10 genes (27,6 kb) COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL18A1, KIF11, NDP, PAK2, RCBTB1	3 - 5 w	E
Walker-Warburg syndrome (WWS, MDDGA) * Gene panel: ID178.00, 14 genes (23,7 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 w	E
Weill-Marchesani syndrome (WMS) * Gene panel: ID230.00, 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 w	E
Gastroenterological Diseases		
Alagille syndrome (ALGS) * Gene panel: ID112.00, 2 genes (11,1 kb) JAG1, NOTCH2	3 - 5 w	E
Cholestasis, comprehensive diagnostics * Gene panel: ID367.00, 81 genes (179,0 kb) ABCB11, ABCB4, ABCC2, ABCD3, ABCG5, ABCG8, ACOX2, ADK, AKR1D1, ALDOB, AMACR, ATP7B, ATP8B1, BAAT, BCS1L, CCDC115, CFTR, CLDN1, COG7, CYP27A1, CYP7B1, DCDL2, DGUOK, FAH, FOCAD, GALE, GALT, GBA1, HADHA, HNF1B, HSD3B7, IFT56, JAG1, KIF12, LARS1, LIPA, LSR, MPI, MPV17, MVK, MYO5B, NBAS, NOTCH2, NPC1, NPC2, NR1H4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PKHD1, POLG, RINT1, SCYL1, SEMA7A, SERPINA1, SLC25A13, SLC51A, SLC51B, SMPD1, TALDO1, TJP2, TRMU, TULP3, UGT1A1, UNC45A, USP53, VIPAS39, VPS33B, VPS50, YARS1, ZFYVE19	4 - 6 w	E
Colorectal cancer and polyposis * Gene panel: ID006.09, 23 genes (64,1 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53	3 - 5 w	E
Gastric cancer * Gene panel: ID090.04, 25 genes (73,6 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MBD4, MLH1, MSH2, MSH6, MUTYH, PMS2, PDGFRA, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53	3 - 5 w	E
Gastrointestinal stromal tumor (GIST) * Gene panel: ID226.00, 8 genes (19,0 kb) KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Gastroenterological Diseases		
Hirschsprung disease (HSCR) * Gene panel: ID177.01, 20 genes (37,1 kb) ECE1, EDN3, EDNRB, DNMT3B, GDNF, GFRA1, KIFBP, L1CAM, NRG1, NRTN, NTF3, NTRK3, PHOX2B, PSPN, RET, SEMA3A, SEMA3C, SEMA3D, SOX10, ZEB2	3 - 5 w	E
Intrahepatic cholestasis * Gene panel: ID159.04 Intrahepatic cholestasis: 25 genes (64,9 kb) ABCB4, ABCB11, ABCC2, ABCD3, ABCG5, ABCG8, ACOX2, AKR1D1, AMACR, ATP8B1, BAAT, CYP7B1, HSD3B7, JAG1, KIF12, MYO5B, NOTCH2, NR1H4, SEMA7A, SLC25A13, SLC51A, TJP2, USP53, VPS33B, ZFYVE19 Progressive intrahepatic cholestasis (PFIC): 13 genes (35,2 kb) ABCB4, ABCB11, ABCG8, ATP8B1, KIF12, MYO5B, NR1H4, SEMA7A, SLC51A, TJP2, USP53, VPS33B, ZFYVE19 Congenital bile acid synthesis defect (CBAS): 7 genes (10,0 kb) ABCD3, ACOX2, AKR1D1, AMACR, BAAT, CYP7B1, HSD3B7 Alagille syndrome (ALGS): 2 genes (11,1 kb) JAG1, NOTCH2	3 - 5 w	E
Lynch syndrome (LYNCH, HNPCC) * Gene panel: ID002.02, 5 genes (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	3 - 5 w	E
Pancreatic cancer * Gene panel: ID089.03, 17 genes (55,2 kb) APC, ATM, BRCA1, BRCA2, CDKN2A, CTSC, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL	3 - 5 w	E
Pancreatitis (PCTT) * Gene panel: ID141.02, 18 genes (29,6 kb) APOA5, APOC2, CASR, CEL, CFTR, CPA1, CLDN2, CELA3B, CTSC, CTSB, GPIHBP1, LMF1, LPL, PNLP, PRSS1, SPINK1, TRPV6, UBR1	3 - 5 w	E
Polyposis syndrome (PS, FAP) * Gene panel: ID005.06, 15 genes (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 w	E
Polycystic liver disease (PCLD) * Gene panel: ID305.02 Polyzystic liver disease (PCLD): 16 genes (55,7 kb) ALG5, ALG6, ALG8, ALG9, DNAJB11, DZIP1L, GANAB, IFT140, LRP5, NEK8, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63 Polycystic liver disease with or without kidney cysts (PCLD): 7 genes (14,0 kb) ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B Polycystic kidney disease with polycystic liver disease (PKD): 9 genes (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1	3 - 5 w	E
Visceral neuropathy and myopathy, comprehensive diagnostics * Gene panel: ID238.02 Visceral neuropathy and myopathy, comprehensive diagnostics: 36 Gene (83,7 kb) ACTA2, ACTG2, CHRM3, DNMT3B, ECE1, EDN3, EDNRB, ERBB2, ERBB3, FLNA, GDNF, GFRA1, KIFBP, L1CAM, LIG3, LMOD1, MYH11, MYL9, MYLK, NRG1, NRTN, NTF3, NTRK3, PHOX2B, POLG, PSPN, RAD21, RET, RRM2B, SEMA3A, SEMA3C, SEMA3D, SGO1, SOX10, TYMP, ZEB2 Hirschsprung disease (HSCR): 20 genes (37,1 kb) ECE1, EDN3, EDNRB, DNMT3B, GDNF, GFRA1, KIFBP, L1CAM, NRG1, NRTN, NTF3, NTRK3, PHOX2B, PSPN, RET, SEMA3A, SEMA3C, SEMA3D, SOX10, ZEB2 Intestinal pseudoobstruction, neuropathic type (VSCN): 9 genes (28,6 kb) ERBB2, ERBB3, FLNA, LIG3, POLG, RAD21, RRM2B, SGO1, TYMP Intestinal pseudoobstruction, myopathic type (VSCM): 8 genes (19,9 kb) ACTA2, ACTG2, CHRM3, LMOD1, MYL9, MYLK, MYH11, RAD21	3 - 5 w	E
Hematological Diseases		
Atypical hemolytic uremic syndrome (AHUS) * Gene panel: ID163.04, 20 genes (42,1 kb) ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 w	E
Bone marrow failure (BMF) * Gene panel ID357.00 Bone marrow failure (BMF): 15 genes (29,9 kb) ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTEL1, SRP72, TERC, TERT, TP53, ZCCHC8 Bone marrow failure syndrome (BMFS): 8 genes (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Telomere-related bone marrow failure and pulmonary fibroses (PFBMFT): 6 genes (13,0 kb) PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8	3 - 5 w	E
Erythrocytosis (ECYT) * Gene panel: ID138.02, 11 genes (13,9 kb) BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Hematological Diseases		
Fanconi anemia (FANC) * Gene panel: ID043.02, 21 genes (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2	3 - 5 w	E
Hemophilia * Gene panel: ID154.01, 5 genes (18,9 kb) F8, F9, LMAN1, MCFD2, VWF	3 - 5 w	E
Hermansky-Pudlak syndrome (HPS) * Gene panel: ID289.00, 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 w	E
Myelodysplastic syndrome (MDS) and Acute myeloid leukemia (AML) * Gene panel: ID321.01 Myelodysplastic syndrome (MDS) and Acute myeloid leukemia (AML): 121 genes (244,1 kb) ACD, ADA2, ADH5, ALDH2, ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CHEK2, CLPB, CSF3R, CTC1, DCLRE1B, DDX41, DKC1, DNAJC21, DNMT3A, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GFI1, HAX1, HEATR3, HRAS, IKZF1, JAGN1, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MBD4, MDM4, MECOM, MLH1, MRAS, MSH2, MSH6, MYSM1, NAF1, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PARN, PAX5, PMS2, PTPN11, RAD51, RAD51C, RAF1, RBBP6, RFWD3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SOS1, SOS2, SRP54, SRP72, STAT3, STN1, TERC, TERT, TINF2, TP53, TSR2, TYMS, UBE2T, UNC13D, VPS45, WAS, WRAP53, XRCC2, ZCCHC8 Acute myeloid leukemia (AML): 12 genes (28,4 kb) ANKRD26, CEBPA, DDX41, ETV6, GATA2, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53 Diamond-Blackfan anemia (DBA): 20 genes (11,3 kb) GATA1, HEATR3, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2 Shwachman-Diamond syndrome (SDS): 4 genes (7,2 kb) DNAJC21, EFL1, SBDS, SRP54 Bone marrow failure syndrome (BMFS): 8 genes (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Pulmonary fibrosis and bone marrow failure (PFBMFT): 6 genes (13,0 kb) PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8 Dyskeratosis congenita (DKC): 13 genes (21,8 kb) ACD, CTC1, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53 Congenital neutropenia (SCN): 10 genes (13,9 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS Fanconi anemia (FANC): 20 genes (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2	4 - 6 w	E
Neutropenia * Gene panel: ID189.02 Neutropenia: 30 genes (51,0 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAFAZZIN, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1 Severe congenital neutropenia (SCN): 11 genes (15,4 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS Syndromes with neutropenia: 21 genes (37,5 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAFAZZIN, USB1, VPS13B, WDR1	3 - 5 w	E
Platelet disorders, comprehensive diagnostics * Gene panel: ID274.02 Platelet disorders, comprehensive diagnostics: 72 genes (155,0 kb) ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S5, BLOC1S6, CD36, CDC42, CYCS, DIAPH1, DTNBP1, EPHB2, ETV6, FERMT3, FLI1, FYB1, GALE, GATA1, GFI1B, GNE, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF1, IKZF5, ITGA2, ITGA2B, ITGB3, JAK2, LYST, MASTL, MECOM, MP1G6B, MPL, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, PTPRJ, RAP1B, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, TBXA2R, TBXAS1, THPO, TPM4, TUBA8, TUBB1, WAS, WDR1, WIPF1 Thrombocytopenia, nonsyndromic (THC): 16 genes (29,1 kb) ACTB, ANKRD26, CYCS, ETV6, FYB1, GALE, GNE, IKZF5, MASTL, PTPRJ, RAP1B, SRC, THPO, TUBB1, TUBA8, WAS Thrombocythemia (THCYT): 3 genes (6,4 kb) JAK2, MPL, THPO Bleeding disorder, platelet-type (BDPLT): 24 genes (53,6 kb) ACTN1, ANO6, CD36, EPHB2, FLI1, GFI1B, GP1BA*, GP1BB, GP6, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1, TPM4 Hermansky-Pudlak syndrome (HPS): 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	4 - 6 w	E
Porphyria * Gene panel: ID153.01, 10 genes (12,8 kb) ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Hematological Diseases		
Sideroblastic anemia (SIDBA) * Gene panel: ID355.00, 10 genes (14,9 kb) ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2	3 - 5 w	E
Spherocytosis (SPH) and Elliptocytosis (EL) * Gene panel: ID203.01 Spherocytosis (SPH) and Elliptocytosis (EL) : 6 genes (26,8 kb) ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB Spherocytosis (SPH): 5 genes (24,2 kb) ANK1, EPB42, SLC4A1, SPTA1, SPTB Elliptocytosis (EL): 4 genes (18,4 kb) EPB41, SLC4A1, SPTA1, SPTB	3 - 5 w	E
Thrombocytopathy (BDPLT, HPS) * Gene panel: ID119.01 Thrombocytopathy (BDPLT, HPS): 37 genes (91,0 kb) ACTN1, ANO6, AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CD36, DTNBP1, EPHB2, FERMT3, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGA2B, ITGB3, LYST, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1 Bleeding disorder, platelet-type (BDPLT), with thrombocytopenia: 15 genes (37,9 kb) ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14 Bleeding disorder, platelet-type (BDPLT), without thrombocytopenia: 10 genes (20,3 kb) ANO6, EPHB2, GP6, ITGA2B, ITGB3, P2RY12, PTGS1, RASGRP2, TBXA2R, TBXAS1 Hermansky-Pudlak syndrome (HPS): 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 w	E
Thrombocytopenia * Gene panel: ID104.03 Thrombocytopenia (THC): 49 genes (98,5 kb) ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CD36, CDC42, CYCS, DIAPH1, ETV6, FLI1, FYB1, GALE, GATA1, GFI1B, GNE, GP1BA, GP1BB, GP9, HOXA11, IKZF1, IKZF5, ITGA2, ITGA2B, ITGB3, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, PLAU, PRKACG, PTPRJ, RAP1B, RBM8A, RUNX1, SLFN14, SRC, STIM1, THPO, TPM4, TUBA8, TUBB1, WAS, WDR1, WIPF1 Thrombocytopenia, nonsyndromic (THC): 13 genes (26,3 kb) ANKRD26, CYCS, ETV6, FYB1, GNE, IKZF5, MASTL, PTPRJ, SRC, THPO, TUBA8, TUBB1, WAS Bleeding disorder, platelet-type (BDPLT): 16 genes (38,8 kb) ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14, TPM4	3 - 5 w	E
Thrombophilia (THPH) * Gene panel: ID150.01, 12 genes (25,2 kb) F2, F5, F9, F13B, HABP2, HRG, MTHFR, PROC, PROS1, SERPINC1, SERPIND1, THBD	3 - 5 w	E
Thrombotic microangiopathy (TMA) * Gene panel: ID707.00, 23 genes (44,7 kb) ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD	3 - 5 w	E
Immunological Diseases		
Atypical hemolytic uremic syndrome (AHUS) * Gene panel: ID163.04, 20 genes (42,1 kb) ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 w	E
Autoinflammatory syndromes, comprehensive diagnostics * Gene panel: ID087.05 Autoinflammatory syndromes, comprehensive diagnostics: 51 genes (99,3 kb) ADA2, ALPK1, ARPC1B, ARPC5, CARD14, DNASE2, DOCK11, DPP9, ELANE, ELF4, HCK, IKBKG, IL1RN, IL36RN, JAK1, LPIN2, LYN, MEFV, MVK, NCKAP1L, NFKB1, NLR4, NLRP1, NLRP12, NLRP3, NOD2, NTRK1, OTULIN, PLCG2, POMP, PSMB4, PSMB8, PSMB9, PSMB10, PSMG2, PSTPIP1, RBCK1, RELA, RIPK1, RNF31, SHARPIN, SLC29A3, SOCS1, STING1, SYK, TBK1, TNFAIP3, TNFRSF1A, TNFRSF11A, TRNT1, WDR1 Recurrent fever syndromes: 14 genes (29,9 kb) ELANE, LPIN2, MEFV, MVK, NLR4, NLRP12, NLRP3, NOD2, OTULIN, PSMB8, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 w	E
Bone marrow failure (BMF) * Gene panel ID357.00 Bone marrow failure (BMF): 15 genes (29,9 kb) ADH5, ALDH2, DNAJC21, DUT, ERCC6L2, MDM4, MYSM1, PARN, RPA1, RTE1, SRP72, TERC, TERT, TP53, ZCCHC8 Bone marrow failure syndrome (BMFS): 8 genes (16,1 kb) ADH5, ALDH2, DNAJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Telomere-related bone marrow failure and pulmonary fibroses (PFBMFT): 6 genes (13,0 kb) PARN, RPA1, RTE1, TERC, TERT, ZCCHC8	3 - 5 w	E
Chronic granulomatous disease (CGD) * Gene panel: ID379.00, 8 genes (8,8 kb) CYBB, CYBA, CYBC1, G6PD, NCF1, NCF2, NCF4, RAC2	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Immunological Diseases		
Hyper-IgE syndrome with recurrent infections (HIES) * Gene panel: ID240.01, 10 genes (29,5 kb) DOCK8, DSG1, IL6R, IL6ST, PGM3, SPINK5, STAT3, STAT6, TYK2, ZNF341	3 - 5 w	E
Hydrops fetalis * Gene panel: ID370.00, 148 genes (404,7 kb) ACAD9, AHCY, ALG1, ALG12, ALG8, ALG9, ALPK3, ANGPT2, ARSB, ASAH1, ATP1A2, BRAF, CALCRL, CBL, CCBE1, CDAN1, CEP55, CFH, CHD7, CHRNA1, CHRND, CHRNA2, COL2A1, CTSA, DHCR7, DHCR24, DMPK, DNAH9, DOK7, DYNC1H1, EBP, EHBP1L1, EP300, EPHB4, ERCC5, FAT4, FBXW11, FGFR3, FH, FLT4, FOXC2, FOXP3, GAA, GALNS, GATA1, GATB, GBA1, GBE1, GLA, GLB1, GLDN, GLE1, GLUL, GNPTAB, GUSB, HADHA, HADHB, HBA1, HBA2, HNF1B, HRAS, IDUA, KIDINS220, KLF1, KLHL40, KMT2D, KRAS, LARS2, LBR, LIPA, LRP6, LZTR1, MAP2K1, MAP2K2, MAPK1, MDFIC, MKKS, MRAS, MUSK, MVK, MYH3, MYRF, NDUFB10, NEB, NEU1, NEXN, NF1, NPC1, NPC2, NRAS, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIEZO1, PKLR, PMM2, POU3F3, PPP1CB, PTH1R, PTPN11, RAF1, RAPSN, RASA1, RASA2, RHD, RIT1, RPL11, RPL15, RRAS, RRAS2, RYR1, SCN4A, SF3B4, SGPL1, SHOC2, SLC17A5, SLC22A5, SLC30A5, SMPD1, SOS1, SOS2, SOX18, SPRED1, SPRED2, SPTB, STAT3, SUMF1, SUZ12, TALDO1, TAFAZZIN, THSD1, UROS, WAC, WDFY3, ZEB2, ZNF148	4 - 6 w	E
Neutropenia * Gene panel: ID189.02 Neutropenia: 30 genes (51,0 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA1, GATA2, GFI1, GINS1, HAX1, JAGN1, PGM3, RAC2, SBDS, SMARCD2, SRP54, TAFAZZIN, TCIRG1, USB1, VPS13B, VPS45, WAS, WDR1 Severe congenital neutropenia (SCN): 11 genes (15,4 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, TCIRG1, VPS45, WAS Syndromes with neutropenia: 21 genes (37,5 kb) ADA2, CD40, CD40LG, CEBPE, CLPB, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, GATA1, GATA2, GINS1, PGM3, RAC2, SBDS, SMARCD2, TAFAZZIN, USB1, VPS13B, WDR1	3 - 5 w	E
Periodic fever syndromes (HPF) * Gene panel: ID088.04,12 genes (26,3 kb) ELANE, MEFV, MVK, NLRCA, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 w	E
Primary antibody deficiency (CVID, AGM)* Gen-Panel: ID382.00 Primary antibody deficiency (CVID, AGM): 38 genes (70,7 kb) AICDA, BLNK, BTK, CARD11, CD19, CD40, CD40LG, CD79A, CD79B, CD81, CR2, CTNNB1, CXCR4, FNIP1, ICOS, IGLL1, IKZF1, IL21, IRF2BP2, LRBA, LRRC8A, MS4A1, NFKB1, NFKB2, OAS1, PIK3CD, PIK3R1, PLCG2, RAC2, SEC61A1, SH3KBP1, SLC39A7, SPI1, TCF3, TNFRSF13B, TNFRSF13C, TOP2B, UNG Common variable immunodeficiency (CVID): 14 genes (28,0 kb) CD19, CD81, CR2, ICOS, IKZF1, IL21, IRF2BP2, LRBA, MS4A1, NFKB1, NFKB2, SEC61A1, TNFRSF13B, TNFRSF13C Agammaglobulinaemia (AGM): 11 genes (16,2 kb) BLNK, BTK, CD79A, CD79B, IGLL1, LRRC8A, PIK3R1*, SH3KBP1, SLC39A7, SPI1, TCF3 Immunodeficiency with hyper-IgM (HIGM): 4 genes (3,2 kb) AICDA, CD40, CD40LG, UNG	3 - 5 w	E
Primary immunodeficiency, comprehensive diagnostics * Gene panel: ID380.00, 441 genes (942,4 kb) ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AGR2, AICDA, AIRE, AK2, ALPI, ALPK1, ANGPT1, ANKZF1, AP1S3, AP3B1, AP3D1, ARHGEF1, ARPC1B, ARPC5, ATAD3A, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C2ORF69, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C9, CARD10, CARD11, CARD14, CARD8, CARD9, CARMIL2, CASP10, CASP8, CBLB, CCBE1, CD19, CD247, CD27, CD28, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFHR1, CFI, CFP, CFTR, CHD7, CIB1, CIITA, CLPB, COL7A1, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTNNB1, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1B, DCLRE1C, DEF6, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK11, DOCK2, DOCK8, DPP9, DUT, EFL1, ELANE, ELF4, EPG5, ERBIN, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FGL2, FLT3LG, FMNL2, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC3, G6PD, GATA1, GATA2, GFI1, GIMAP5, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HMOX1, HPS1, HPS4, HPS6, HSPA1L, HTRA2, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNG, IFNGR1, IFNGR2, IGLL1, IKBKB, IKBKG, IKZF1, IKZF2, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1R1, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IPO8, IRAK4, IRF1, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, ITPKB, IVNS1ABP, JAGN1, JAK1, JAK3, KCNA5, KDM6A, KMT2A, KMT2D, KRAS, LACC1, LAMTOR2, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LYN, LYST, MAGT1, MALT1, MAP3K14, MBL2, MCM10, MCM4, MCTS1, MECOM, MEFV, MOGS, MPEP1, MPO, MRTFA, MS4A1, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRCA, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NPC1, NRAS, NSMCE3, NUDCD3, OAS1, ORAI1, OTULIN, PARN, PAX1, PEPD, PGM3, PI4KA, PIK3CD, PIK3CG, PIK3R1, PLCG2, PLG, PNP, POLA1, POLD1, POLD3, POLE, POLR3A, POLR3C, POLR3F, POMP, PRF1, PRIM1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSTPIP1, PTCRA, PTEN, PTPN2, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RC3H1, RECQL4, REL, RELB, RFX5, RFXANK, RFXAP, RHBDF2, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU7-1, RORC, RPSA, RTE1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCGN, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A1, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SLC9A3, SLC02A1, SMARCA1, SMARCD2, SNORA31, SOCS1, SP110, SPI1, SPINK5, SPPL2A, SRP54, STAT1, STAT2, STAT3, STAT4, STAT5B, STAT6, STIM1, STING1, STK4, STX11, STXBP2, STXBP3, SYK, TAP1, TAP2, TAPBP, TAFAZZIN, TBK1, TBX1, TBX21, TCF3, TCN2, TERC, TERT, TET2, TFRC, TGFB1, TGFB1R1, TGFB2, TICAM1, TINF2, TLR3, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TOM1, TOP2B, TPP2, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC7A, TYK2, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, XIAP, ZAP70, ZBTB24, ZNF341, ZNFXX1	4 - 6 w	E
Severe combined immunodeficiency (SCID) * Gene panel: ID381.00, 21 genes (47,6 kb) ADA, AK2, BCL11B, CD3E, CD3D, CIITA, CORO1A, DCLRE1C, FOXN1, IL2RG, IL7R, LAT, LIG4, JAK3, NHEJ1, POLD3, PRKDC, PTPRC, RAC2, RAG1, RAG2	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
Alport syndrome (ATS) * Gene panel: ID099.00, 4 genes (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9	3 - 5 w	E
Aminoaciduria * Gene panel: ID318.00 Aminoaciduria: 13 genes (21,1 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC1A1, SLC2A2, SLC3A1, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC34A1, SLC36A2 Cystinuria: 2 genes (3,5 kb) SLC3A1, SLC7A9 Hyperglycinuria: 3 genes (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Fanconi renotubular syndrome (FRTS): 5 genes (7,7 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC34A1	3 - 5 w	E
Atypical hemolytic uremic syndrome (AHUS) * Gene panel: ID163.04, 20 genes (42,1 kb) ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN	3 - 5 w	E
Bardet-Biedl syndrome (BBS) * Gene panel: ID093.02, 21 genes (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	3 - 5 w	E
Bartter syndrome (BARTS) * Gene panel: ID156.01, 8 genes (17,7 kb) BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3	3 - 5 w	E
Branchiotoorenal syndrome (BOR) * Gene panel: ID315.00, 5 genes (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A	3 - 5 w	E
Congenital anomalies of kidney and urinary tract (CAKUT) * Gene panel: ID229.03 Congenital anomalies of kidney and urinary tract (CAKUT): 62 genes (198,3 kb) ACE, ACTG2, AGT, AGTR1, ANOS1, BICC1, BMP4, BNC2, CDC5L, CEP55, CHD1L, CHRM3, CRKL, DSTYK, EYA1, FAT4, FGF20, FRAS1, FREM1, FREM2, GATA3, GLI3, GREB1L, GRIP1, HNF1B, HPSE2, ITGA8, KIF14, LIFR, LRIG2, LRP4, MUC1, MYH11, NEK8, NPHP3, NRIP1, PAX2, PBX1, REN, RET, ROBO1, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX11, SOX17, TBC1D1, TBX18, TFAP2A, TNXB, TRAP1, UMOD, UPK3A, WBP11, WNT4 Renal hypodysplasia/aplasia and renal agenesis: 25 genes (75,9 kb) ANOS1, BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GFRA1, GREB1L, HNF1B, ITGA8, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4 Vesicoureteral reflux (VUR): 10 genes (33,7 kb) DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB Branchiotoorenal syndrome (BOR): 5 genes (10,2 kb) EYA1, SALL1, SIX1, SIX5, TFAP2A Renal tubular dysgenesis (RTD): 4 genes (7,7 kb) ACE, AGT, AGTR1, REN Fraser syndrome (FRASRS): 3 genes (24,8 kb) FRAS1, FREM2, GRIP1 MMIH syndrome (MMIHS): 5 Gene (15,1 kb) ACTG2, LMOD1, MYH11, MYL9, MYLK	4 - 6 w	E
Cystic kidney diseases, comprehensive diagnostics * Gene panel: ID100.09 Cystic kidney diseases, comprehensive diagnostics: 53 genes (165,3 kb) ALG5, ALG6, ALG8, ALG9, ANKS6, BICC1, CEP83, CEP164, CEP290, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, FLCN, GANAB, GLIS2, HNF1B, IFT140, IFT172, INVS, IQCB1, LRP5, MAPKBP1, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, PRKCSH, REN, RPGRIP1L, SDCCAG8, SEC61A1, SEC61B, SEC63, SLC41A1, TSC1, TSC2, TTC21B, TMEM67, UMOD, VHL, WDR19, XPNPEP3, ZNF423 Polycystic kidney disease (PKD): 9 genes (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1 Polycystic liver disease with kidney cysts (PCLD): 7 genes (14,0 kb) ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B Medullary cystic kidney disease (MCKD, ADTKD): 5 genes (7,7 kb) HNF1B, MUC1, REN, SEC61A1, UMOD Nephronophthisis (NPHP): 17 genes (50,3 kb) ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423 Tuberous sclerosis (TSC): 2 genes (8,9 kb) TSC1, TSC2	4 - 6 w	E
Cystinosis (CTNS) and similar metabolic disorders * Gene panel: ID706.01, 16 genes (30,1 kb) ATP7B, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, EHHADH, FAH, GALT, GATM, HNF4A, KCNJ1, NDUFAF6, OCRL, SLC12A1, SLC34A1	3 - 5 w	E
Diabetes insipidus * Gene panel: ID322.00, 5 genes (8,4 kb) AQP2, AVP, AVPR2, SLC12A1, WFS1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
Endocrine hypertension * Gene panel: ID270.03 Endocrine hypertension: 33 genes (81,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, DLST, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL, WNK1, WNK4, YY1AP1 Hyperaldosteronism (HALD): 5 genes (19,1 kb) CACNA1H, CACNA1D, CLCN2, CYP11B1, KCNJ5 Pseudohypoaldosteronism type II (PHA2): 4 genes (15,8 kb) CUL3, KLHL3, WNK1, WNK4 Pheochromocytoma/paraganglioma syndrome (PPGL): 11 genes (11,8 kb) DLST, MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL Adrenal cushing syndrome (PPNAD): 4 genes (8,9 kb) NR3C1, PDE11A, PDE8B, PRKAR1A Liddle syndrome (LIDLS): 3 genes (5,9 kb) SCNN1A, SCNN1B, SCNN1G Hypertension with low renin level: 20 genes (58,7 kb) CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP17A1, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4	3 - 5 w	E
Fanconi renotubular syndrome (FRTS) * Gene panel: ID359.00, 7 genes (10,4 kb) CTNS, EHHADH, GATM, HNF4A, NDUFAF6, SLC2A2, SLC34A1	3 - 5 w	E
Fraser syndrome (FRASRS) * Gene panel: ID317.00, 3 genes (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 w	E
Galloway-Mowat syndrome (GAMOS) * Gene panel: ID251.01, 10 genes (12,5 kb) GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC	3 - 5 w	E
Glomerulonephritis * Gene panel: ID103.01 Glomerulonephritis: 18 genes (36,9 kb) C1QA, C1QB, C1QC, C2, C3, CD46, CFB, CFI, CFH, CFHR1, CFHR2, CFHR3, CFHR5, DGKE, FN1, PRKCD, SOX18, THBD C3 glomerulopathy (C3G): 3 genes (7,2 kb) CFI, CFH, CFHR5 C1q deficiency (C1QD): 3 genes (2,3 kb) C1QA, C1QB, C1QC	3 - 5 w	E
Hyperoxaluria * Gene panel: ID363.00 Hyperoxaluria: 6 Gene (8,6 kb) AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6 Primary hyperoxaluria (PH): 3 genes (3,2 kb) AGXT, GRHPR, HOGA1 Calcium oxalate nephrolithiasis (CAON): 3 genes (5,4 kb) OXGR1, SLC26A1, SLC26A6	3 - 5 w	E
Hypomagnesemia (HOMG) * Gene panel: ID054.02, 14 genes (30,9 kb) ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6	3 - 5 w	E
Hypophosphatasia, hypophosphatemia and rickets * Gene panel: ID269.03 Hypophosphatasia, hypophosphatemia and rickets: 16 genes (27,8 kb) ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR Hypophosphatemic rickets (HR): 9 genes (16,1 kb) CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3 Vitamin D-dependent hypophosphatemic rickets (VDDR): 4 genes (5,8 kb) CYP2R1, CYP3A4, CYP27B1, VDR Hypophosphatasia (HPP): 1 gene (1,6 kb) ALPL	3 - 5 w	E
Joubert syndrome (JBTS) * Gene panel: ID028.03, 40 genes (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423	4 - 6 w	E
Metabolic kidney diseases * Gene panel: ID705.00, 29 genes (53,4 kb) AGXT, APOA1, ATP7B, B2M, BSND, CLCN5, CLCNKA, CLCNKB, CTNS, FAH, FGA, GALT, GLA, GRHPR, GSN, HOGA1, KCNJ1, LYZ, MEFV, MVK, NLRC4, NLRP12, NLRP3, OCRL, PLCG2, SLC12A1, SLC26A1, TNFRSF1A, TTR	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
Meckel syndrome (MKS) * Gene panel: ID032.02, 13 genes (35,1 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231	3 - 5 w	E
Microhaematuria * Gene panel: ID385.00, 11 genes (44,0 kb) CFHR5, COL4A1, COL4A3, COL4A4, COL4A5, FN1, INF2, MYH9, PIGA, PIGT, UMOD	3 - 5 w	E
Nephroblastoma and Wilms tumor (WT) * Gene panel: ID335.00, 21 genes (70,4 kb) AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CEP57, CTR9, DICER1, DIS3L2, GPC3, GPC4, NSD1, PALB2, POU6F2, REST, TRIM28, TRIM37, TRIP13, WT1	3 - 5 w	E
Nephrocalcinosis * Gene panel: ID361.00, 29 genes (54,1 kb) ADCY10, AGXT, ALPL, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, FAH, FAM20A, GRHPR, HOGA1, KCNJ1, MAGED2, OCRL, OXGR1, RRAGD, SLC12A1, SLC4A1, SLC34A1, SLC34A3, VIPAS39, VPS33B	3 - 5 w	E
Nephronophthisis (NPHP) * Gene panel: ID030.02, 22 genes (70,9 kb) ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423	3 - 5 w	E
Nephrotic syndrome (SRNS, NPHS) and Focal segmental glomerulosclerosis (FSGS) * Gene panel: ID098.06 Nephrotic syndrome (SRNS, NPHS) and Focal segmental glomerulosclerosis (FSGS): 43 genes (129,7 kb) ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, DAAM2, DGKE, EMP2, INF2, KANK2, KIRREL1, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PAX2, PDSS2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, TRPC6, WT1 Nephrotic syndrome (SRNS, NPHS): 30 genes (89,3 kb) ARHGAP24, ARHGDIA, AVIL, COQ2, COQ6, COQ8B, DAAM2, DGKE, EMP2, KANK2, KIRREL1, LAMA5, LAMB2, MAGI2, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PDSS2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, WT1 Focal-segmental glomerulosclerosis (FSGS): 18 genes (49,5 kb) ACTN4, ANLN, APOL1, ARHGAP24, COQ2, COQ6, COL4A3, COL4A4, COL4A5, CRB2, CD2AP, INF2, LMX1B, MYO1E, PAX2, PDSS2, SMARCAL1, TRPC6	4 - 6 w	E
Pseudoaldosteronism (LIDLS) and Pseudohypoaldosteronism (PHA) * Gene panel: ID250.00 Pseudoaldosteronism (LIDLS) and Pseudohypoaldosteronism (PHA): 8 genes (23,8 kb) CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4 Pseudohypoaldosteronism, type I (PHA1): 4 genes (8,8 kb) NR3C2, SCNN1A, SCNN1B, SCNN1G Pseudohypoaldosteronism, type II (PHA2): 4 genes (15,7 kb) CUL3, KLHL3, WNK1, WNK4 Liddle syndrome (LIDLS): 3 genes (5,9 kb) SCNN1A, SCNN1B, SCNN1G	3 - 5 w	E
Polycystic kidney disease (PKD) * Gene panel: ID295.03 Polycystic kidney disease (PKD): 9 genes (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1 Polycystic kidney disease, autosomal dominant (PKD, ADPKD): 7 genes (27,4 kb) ALG5, DNAJB11, GANAB, IFT140, NEK8, PKD1, PKD2 Polycystic kidney disease, autosomal recessive (PKD, ARPKD): 2 genes (14,5 kb) DZIP1L, PKHD1	3 - 5 w	E
Polycystic liver disease (PCLD) * Gene panel: ID305.02 Polyzystic liver disease (PCLD): 16 genes (55,7 kb) ALG5, ALG6, ALG8, ALG9, DNAJB11, DZIP1L, GANAB, IFT140, LRP5, NEK8, PKD1, PKD2, PKHD1, PRKCSH, SEC61B, SEC63 Polycystic liver disease with or without kidney cysts (PCLD): 7 genes (14,0 kb) ALG6, ALG8, ALG9, LRP5, PRKCSH, SEC63, SEC61B Polycystic kidney disease with polycystic liver disease (PKD): 9 genes (41,8 kb) ALG5, DNAJB11, DZIP1L, GANAB, IFT140, NEK8, PKD1, PKD2, PKHD1	3 - 5 w	E
Renal amyloidosis * Gene panel: ID320.01 Renal amyloidosis: 9 genes (13,2 kb) APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TNFRSF1A, TTR Hereditary systemic amyloidosis (AMYLD): 6 genes (6,3 kb) APOA1, B2M, FGA, GSN, LYZ, TTR Periodic fever syndromes with amyloidosis: 3 genes (6,8 kb) MEFV, NLRP3, TNFRSF1A	3 - 5 w	E
Renal cancer * Gene panel: ID041.04, 34 genes (76,8 kb) BAP1, CDC73, CDKN1C, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MITF, MLH1, MSH2, MSH6, PBRM1, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
Renal hypodysplasia, aplasia and agenesis * Gene panel: ID319.00, 23 genes (72,5 kb) BICC1, BMP4, CEP55, DSTYK, FAT4, FGF20, FREM1, GATA3, GREB1L, HNF1B, ITGA8, NEK8, NPHP3, NRIP1, PAX2, PBX1, ROBO1, RET, SALL1, TBX18, UPK3A, WBP11, WNT4	3 - 5 w	E
Renal tubular acidosis (RTA) * Gene panel: ID297.00, 9 genes (18,5 kb) ATP6V0A4, ATP6V1B1, CA2, FOXI1, SLC4A1, SLC4A4, VIPAS39, VPS33B, WDR72	3 - 5 w	E
Renal tubular dysgenesis (RTD) * Gene panel: ID316.00, 4 genes (7,7 kb) ACE, AGT, AGTR1, REN	2 - 4 w	E
Renal ciliopathies, comprehensive diagnostics * Gene panel: ID376.00 Renal ciliopathies, omprehensive diagnostics: 75 genes (181,7 kb) AHI1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP418, CPLANE1, CSPP1, DCDC2, FAM149B1, GLIS2, IFT172, IFT27, IFT74, INPP5E, INVS, IQCB1, KATNIP, KIAA0586, KIAA0753, KIF14, KIF7, LZTFL1, MAPKBP1, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, PDE6D, PIBF1, RPGRIP1L, SDCCAG8, SLC41A1, SUFU, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TMEM67, TOGARAM1, TRAF3IP1, TRIM32, TTC21B, TTC8, TXNDC15, WDPCP, WDR19, XPNPEP3, ZNF423 Nephronophthisis (NPHP): 17 genes (50,3 kb) ANKS6, CEP83, CEP164, DCDC2, GLIS2, INVS, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, SLC41A1, TMEM67, TTC21B, WDR19, XPNPEP3, ZNF423 Bardet-Biedl syndrome (BBS): 22 genes (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP Senior-Loken syndrome (SLSN): 8 genes (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19 Joubert syndrome (JBTS): 40 genes (104,1 kb) AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2, CC2D2A, CEP104, CEP120, CEP290, CEP41, CPLANE1, CSPP1, FAM149B1, IFT74, INPP5E, KATNIP, KIAA0586, KIAA0753, KIF7, NPHP1, MKS1, OFD1, PDE6D, PIBF1, RPGRIP1L, SUFU, TCTN1, TCTN2, TCTN3, TMEM67, TMEM107, TMEM138, TMEM216, TMEM218, TMEM231, TMEM237, TOGARAM1, TTC21B, ZNF423 Meckel syndrome (MKS): 14 genes (36,2 kb) B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM67, TMEM107, TMEM216, TMEM231, TXNDC15	4 - 6 w	E
Senior-Loken syndrome (SLSN) * Gene panel: ID029.01, 8 genes (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 w	E
Thrombotic microangiopathy (TMA) * Gene panel: ID707.00, 23 genes (44,7 kb) ADAMTS13, C2, C3, C4BPA, C4BPB, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, CLU, DGKE, MMACHC, MMADHC, MTHFD1, MMUT, PIGA, PLG, THBD	3 - 5 w	E
Tubulointerstitial kidney disease, autosomal dominant (ADTKD) * Gene panel: ID296.00, 6 genes (8,8 kb) DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD	3 - 5 w	E
Urolithiasis, nephrolithiasis and nephrocalcinosis * Gene panel: ID231.04 Urolithiasis, nephrolithiasis and nephrocalcinosis: 37 genes (68,8 kb) ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, BSND, CASR, CLCN5, CLDN16, CLDN19, CLCNKB, CYP24A1, G6PC1, GRHPR, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, OCRL, OXGR1, RRAGD, SLC2A9, SLC3A1, SLC4A1, SLC4A4, SLC6A19, SLC6A20, SLC7A9, SLC12A1, SLC22A12, SLC26A1, SLC34A1, SLC34A3, SLC36A2, XDH Hypercalciuria: 15 genes (30,2 kb) ADCY10, BSND, CASR, CLCN5, CLCNKB, CLDN16, CLDN19, CYP24A1, KCNJ1, MAGED2, OCRL, RRAGD, SLC12A1, SLC34A1, SLC34A3 Hyperoxaluria: 4 genes (6,3 kb) AGXT, GRHPR, HOGA1, SLC26A1, OXGR1 Hyperglycinuria: 3 genes (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Hypocitraturia: 4 genes (11,1 kb) ATP6V0A4, ATP6V1B1, SLC4A1, SLC4A4 Xanthinuria: 2 genes (6,7 kb) MOCOS, XDH Cystinuria: 2 genes (3,5 kb) SLC3A1, SLC7A9	3 - 5 w	E
Urothelial cancer * Gene panel: ID337.00, 34 genes (105,0 kb) APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC	4 - 6 w	E
Vesicoureteral reflux (VUR) * Gene panel: ID314.00, 10 genes (33,7 kb) DSTYK, HPSE2, LRIG2, NRIP1, PAX2, PBX1, ROBO2, SOX17, TBX18, TNXB	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Lipometabolic Disorders		
Combined hyperlipidemia * Gene panel: ID025.06 Combined hyperlipidemia: 40 genes (72,8 kb) ABCG5, ABCG8, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEA, CREB3L3, CYP27A1, CYP7A1, GCKR, GK, GPD1, GPIHBP1, LDLR, LDLRAP1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, NPC1L1, PCSK9, PLIN1, PNPLA5, POLD1, PPARG, SORT1, ZMPSTE24 Hypercholesterolemia: 13 genes (36,4 kb) ABCG5, ABCG8, APOB, APOE, CYP7A1, CYP27A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1 Hypertriglyceridemia: 29 genes (51,1 kb) AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEA, CREB3L3, GCKR, GK, GPD1, GPIHBP1, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, PLIN1, POLD1, PPARG, ZMPSTE24	3 - 5 w	E
HDL-associated lipometabolic disorder * Gene panel: ID096.01, 15 genes (25,5 kb) ABCA1, ANGPTL3, ANGPTL4, APOA1, APOA2, APOA4, APOC3, CETP, LCAT, LIPC, LIPG, NPC1, NPC2, SCARB1, SMPD1	3 - 5 w	E
Hypercholesterolemia * Gene panel: ID024.02, 13 genes (36,4 kb) ABCG5, ABCG8, APOB, APOE, CYP27A1, CYP7A1, LDLR, LDLRAP1, LIPA, NPC1L1, PCSK9, PNPLA5, SORT1	3 - 5 w	E
Hypertriglyceridemia * Gene panel: ID095.03 Hypertriglyceridemia: 32 genes (54,1 kb) ADRA2A, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CIDEA, CREB3L3, GCKR, GK, GPD1, GPIHBP1, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24 Hyperchylomicronemia: 6 genes (6,1 kb) APOC2, APOE, APOA5, GPIHBP1, LMF1, LPL Lipodystrophy: 15 genes (22,0 kb) AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEA, LIPE, LMNA, PLIN1, POLD1, PPARG, ZMPSTE24	3 - 5 w	E
Lipodystrophy (CGL, FPLD)* Gene panel: ID055.04 Lipodystrophy (CGL, FPLD): 15 genes (22,0 kb) ADRA2A, AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEA, LIPE, LMNA, PCYT1A, PLAAT3, PLIN1, POLD1, PPARG, ZMPSTE24 Congenital generalized lipodystrophy (CGL): 5 genes (4,9 kb) AGPAT2, BSCL2, CAV1, CAVIN1, PCYT1A Familial partial lipodystrophy (FPLD): 8 genes (11,5 kb) ADRA2A, CAV1, CIDEA, LIPE, LMNA, PLAAT3, PLIN1, PPARG	3 - 5 w	E
Lipodystrophy, comprehensive diagnostics * Gene panel: ID343.00, 45 genes (80,0 kb) ADRA2A, AGPAT2, AKT2, ALDH18A1, BANF1, BSCL2, CAV1, CAVIN1, CIDEA, EPHX1, ERCC6, ERCC8, FBN1, HRAS, KCNJ6, LEP, LEPR, LIPE, LMNA, MCM3, MCM7, MFN2, MTX2, NSMCE2, OPA3, OTULIN, PCYT1A, PDGFRB, PIK3R1, PLIN1, POLD1, POLR3A, POMP, PPARG, PSMA3, PSMB4, PSMB8, PSMB9, PSMG2, PTPN11, SLC25A24, SLC29A3, SPRTN, WRN, ZMPSTE24	3 - 5 w	E
Lipometabolic disorder due to LDL deficiency * Gene panel: ID094.01, 10 genes (29,6 kb) ANGPTL3, ANGPTL4, ANGPTL8, APOB, APOE, LIMA1, MTPP, NPC1L1, PCSK9, SAR1B	3 - 5 w	E
Lipometabolic disorders, comprehensive diagnostics * Gene panel: ID044.03, 62 genes (114,6 kb) ABCA1, ABCG5, ABCG8, ACADM, ACADS, ACADVL, ADRA2A, AGPAT2, AKT2, ANGPTL3, ANGPTL4, ANGPTL8, APOA1, APOA2, APOA4, APOA5, APOB, APOC2, APOC3, APOE, BSCL2, CAV1, CAVIN1, CETP, CIDEA, CREB3L3, CYP27A1, CYP7A1, DHCR7, DHCR24, GCKR, GK, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIMA1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPA, LPL, MTPP, NPC1, NPC1L1, NPC2, PCSK9, PCYT1A, PLAAT3, PLIN1, PNPLA5, POLD1, PPARG, SAR1B, SCARB1, SMPD1, SORT1, ZMPSTE24	4 - 6 w	E
Statin-associated myopathy * Gene panel: ID097.00, 11 genes (37,2 kb) ACADM, ACADS, ACADVL, AMPD1, CACNA1S, CAV3, CPT2, LPIN1, PYGM, RYR1, SLC01B1	3 - 5 w	E
Metabolic Diseases		
Amyloidosis * Gene panel: ID375.00 Amyloidosis: 18 genes (25,0 kb) APP, APOA1, APOA2, APOC2, B2M, CST3, FGA, GPNMB, GSN, IL31RA, ITM2B, LYZ, MEFV, NLRP3, OSMR, PRNP, TNFRSF1A, TTR Hereditary systemic amyloidosis (AMYLD): 8 genes (7,0 kb) APOA1, APOA2, APOC2, B2M, FGA, GSN, LYZ, TTR Primary localized cutaneous amyloidosis (PLCA): 3 genes (7,0 kb) GPNMB, OSMR, IL31RA Cerebral amyloid angiopathy (CHWA): 4 genes (4,3 kb) APP, CST3, ITM2B, PRNP	3 - 5 w	E
Coenzyme Q10 deficiency (COQ10D) * Gene panel: ID225.01, 15 genes (18,7 kb) ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFB, PDSS1, PDSS2	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Metabolic Diseases		
Combined defect of oxidative phosphorylation (COXPD) * Gene panel: ID287.01, 60 genes (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C2ORF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRS1, PNPT1, POLRMT, PRORP, PTC3, RMND1, SFXN4, SLC25A26, TARS2, TMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 w	E
Congenital disorder of glycosylation (CDG) * Gene panel ID035.02 Congenital disorder of glycosylation (CDG): 51 genes (74,7 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, MAGT1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3 Congenital disorder of glycosylation, type I (CDG1): 29 genes (39,6 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Congenital disorder of glycosylation, type II (CDG2): 22 genes (35,1 kb) ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, TMEM165, TMEM199	3 - 5 w	E
Folate metabolism disorder* Gene panel: ID334.00, 10 genes (18,7 kb) CBS, FOLR1, FOLR2, FTCD, MTHFD1, MTHFR, MTR, MTRR, SLC19A1, SLC46A1	3 - 5 w	E
Glycogen storage disease (GSD) * Gene panel: ID108.00, 23 genes (46,9 kb) AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, SLC2A2, SLC37A4	3 - 5 w	E
Glycosylphosphatidylinositol biosynthesis defect (GPIBD) * Gene panel: ID291.00 Glycosylphosphatidylinositol biosynthesis defect (GPIBD): 22 genes (33,3 kb) GPAA1, PGAP1, PGAP2, PGAP3, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGL, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIGY Multiple congenital anomalies-hypotonia-seizures syndrome (MCAHS): 4 genes (7,7 kb) PIGA, PIGN, PIGQ, PIGT Hyperphosphatasia with mental retardation syndrome (HPMRS): 6 genes (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	3 - 5 w	E
Hemochromatosis (HFE) and Hemosiderosis * Gene panel: ID114.04, 10 genes (14,6 kb) BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TF, TFR2	3 - 5 w	E
Homocystinuria * Gene panel: ID191.01, 9 genes (15,3 kb) ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR, PRDX1	3 - 5 w	E
Hypercalcemia * Gene panel: ID262.00, 8 genes (14,6 kb) AP2S1, CASR, CDC73, CYP24A1, GCM2, GNA11, SLC34A1, SLC12A1	3 - 5 w	E
Hyperinsulinemic hypoglycemia (HHF) * Gene panel: ID126.00, 8 genes (16,9 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A	3 - 5 w	E
Hyperoxaluria * Gene panel: ID363.00, 6 Gene (8,6 kb) AGXT, GRHPR, HOGA1, OXGR1, SLC26A1, SLC26A6	3 - 5 w	E
Hyperphosphatasia with impaired intellectual development syndrome (HPMRS) * Gene panel: ID292.00, 6 genes (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	2 - 4 w	E
Hypoglycemia, hyperinsulinism and ketone metabolism * Gene panel: ID280.00 Hypoglycemia, hyperinsulinism and ketone metabolism: 44 genes (85,9 kb) ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4 Glycogen storage disease (GSD): 24 genes (48,4 kb) AGL, ALDOA, ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4 Hyperinsulinemic hypoglycemia (HHF): 9 genes (18,8 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A	3 - 5 w	E
Hypomagnesemia (HOMG)* Gene panel: ID054.02, 14 genes (30,9 kb) ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXD2, HNF1B, KCNA1, KCNJ10, RRGD, SLC12A3, TRPM6	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Metabolic Diseases		
Hypophosphatasia, hypophosphatemia and rickets * Gene panel: ID269.03 Hypophosphatasia, hypophosphatemia and rickets: 16 genes (27,8 kb) ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR Hypophosphatemic rickets (HR): 9 genes (16,1 kb) CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3 Vitamin D-dependent hypophosphatemic rickets (VDDR): 4 genes (5,8 kb) CYP2R1, CYP3A4, CYP27B1, VDR Hypophosphatasia (HPP): 1 gene (1,6 kb) ALPL	3 - 5 w	E
Maturity-onset diabetes of the young (MODY) * Gene panel: ID048.01, 14 genes (22,9 kb) ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1	3 - 5 w	E
Metabolic disorder with epilepsy in childhood and adolescence * Gene panel: ID172.00, 15 genes (28,1 kb) ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA1, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2	3 - 5 w	E
Metabolic disorder with epilepsy in infancy and childhood * Gene panel: ID171.00, 18 genes (31,5 kb) ALDH5A1, ATP7A, BTBD, FOLR1, GAMT, GATM, HEXA, HEXB, HLCS, KCTD7, MTHFR, PHGDH, POLG, PPT1, SLC19A3, SLC2A1, SLC6A8, TPP1	3 - 5 w	E
Metabolic disorder with neonatal epilepsy * Gene panel: ID135.00, 25 genes (39,4 kb) ABAT, ADSL, ALDH7A1, AMT, BCKDHA, BCKDHB, CPS1, CTSD, DBT, DDC, DLD, DPYD, ETHE1, FH, GCSH, GLDC, GPHN, IVD, L2HGDH, MOCS1, MOCS2, OTC, PCCA, PCCB, PNPO	3 - 5 w	E
Metabolic epilepsy * Gene panel: ID303.01 Metabolic epilepsy: 84 genes (122,3 kb) ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTBD, CLN3, CLN5, CLN6, CLN8, CNNM2, CPS1, CTSD, CTSF, D2HGDH, DBT, DHFR, DLD, DNAJC5, DPYD, ETFA, ETFB, ETFDH, ETHE1, FH, FOLR1, GAMT, GATM, GCDH, GCH1, GCSH, GLDC, GLUL, GM2A, GPHN, GRN, HEXA, HEXB, HIBCH, HLCS, IDH2, IVD, KCTD7, L2HGDH, LIAS, MDH2, MFSD8, MOCS1, MOCS2, MTHFR, NEU1, OTC, PAH, PC, PCBD1, PCCA, PCCB, PGK1, PHGDH, PLPBP, PNPO, POLG, PPM1K, PPT1, PRODH, PTS, QDPR, SLC2A1, SLC6A8, SLC6A9, SLC19A3, SLC25A1, SLC46A1, SUOX, TPK1, TPP1 Glycine encephalopathy (GCE): 5 genes (8,0 kb) AMT, GCSH, GLDC, LIAS, SLC6A9 Molybdenum cofactor deficiency (MOCOD): 3 genes (3,7 kb) GPHN, MOCS1, MOCS2 Cerebral creatine deficiency syndrome (CCDS): 3 genes (3,9 kb) GAMT, GATM, SLC6A8 Maple syrup urine disease (MSUD): 5 genes (6,6 kb) BCKDHA, BCKDHB, DBT, DLD, PPM1K 2-Hydroxyglutaric aciduria: 3 genes (5,3 kb) L2HGDH, D2HGDH, IDH2, SLC25A1 GM2-gangliosidosis: 3 genes (3,8 kb) HEXA, HEXB, GM2A Neuronal ceroid lipofuscinosis (CLN): 12 genes (14,3 kb) CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	4 - 6 w	E
Mucopolysaccharidosis (MPS) * Gene panel: ID308.00, 12 genes (21,2 kb) ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, VPS33A	3 - 5 w	E
Muscular dystrophy-dystroglycanopathy (MDDG) * Gene panel: ID179.00 Muscular dystrophy-dystroglycanopathy (MDDG): 15 genes (24,0 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muscular dystrophy-dystroglycanopathy (MDDGA) with brain and eye anomalies: 14 genes (22,8 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muscular dystrophy-dystroglycanopathy (MDDGB) with or without impaired intellectual development: 8 genes (13,1 kb) DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMT1, POMT2 Muscular dystrophy-dystroglycanopathy (MDDGC), limb-girdle: 11 genes (18,5 kb) CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, POMGNT1, POMGNT2, POMK, POMT1, POMT2	3 - 5 w	E
Neonatal diabetes mellitus * Gene panel: ID162.01 Neonatal diabetes mellitus: 29 genes (53,6 kb) ABCC8, BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GCK, GLIS3, HNF1B, IER3IP1, IL2RA, INS, INSR, KCNJ11, LRBA, MNX1, NEUROD1, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, SLC2A2, STAT3, WFS1, YIPF5, ZFP57 Permanent neonatal diabetes mellitus (PNDM): 10 genes (15,9 kb) ABCC8, FOXP3, GCK, INS, KCNJ11, MNX1, NEUROD1, SLC19A2, SLC2A2, ZFP57 Syndromal neonatal diabetes mellitus: 21 genes (40,5 kb) BSCL2, CISD2, EIF2AK3, FOXP3, GATA4, GATA6, GLIS3, HNF1B, IER3IP1, IL2RA, INSR, LRBA, NEUROG3, NKX2-2, PDX1, PTF1A, RFX6, SLC19A2, STAT3, WFS1, YIPF5	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Metabolic Diseases		
Neuronal ceroid lipofuscinosis (CLN) * Gene panel: ID132.01, 15 genes (20,2 kb) ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1	3 - 5 w	E
Peroxisome biogenesis disorder (PBD) * Gene panel: ID083.01 Peroxisome biogenesis disorder (PBD): 14 genes (19,9 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Zellweger syndrome (PBD, type A): 12 genes (18,3 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Neonatal adrenoleukodystrophy/ Infantile refsum syndrome (PBD, type B): 11 genes (17,0 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX11B, PEX13, PEX16, PEX26 Heimler syndrome (PBD, type C): 2 genes (6,8 kb) PEX1, PEX6	3 - 5 w	E
Porphyria * Gene panel: ID153.01, 10 genes (12,8 kb) ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	3 - 5 w	E
Pulmonary surfactant metabolism dysfunction (SMDP) * Gene panel: ID168.01, 6 genes (12,0 kb) ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB	3 - 5 w	E
Renal amyloidosis * Gene panel: ID320.01 Renal amyloidosis: 9 genes (13,2 kb) APOA1, B2M, FGA, GSN, LYZ, MEV, NLRP3, TNFRSF1A, TTR Hereditary systemic amyloidosis (AMYLD): 6 genes (6,3 kb) APOA1, B2M, FGA, GSN, LYZ, TTR Periodic fever syndromes with amyloidosis: 3 genes (6,8 kb) MEV, NLRP3, TNFRSF1A	3 - 5 w	E
Storage diseases with cardiac involvement * Gene panel: ID149.03 Storage diseases with cardiac involvement: 16 genes (20,7 kb) APOA1, B2M, FGA, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, LYZ, PRKAG2, SLC40A1, TFR2, TTR Cardiac glycogen storage disease (GSD): 3 genes (5,8 kb) GAA, LAMP2, PRKAG2 Hemochromatosis (HFE): 6 genes (7,3 kb) FTH1, HAMP, HFE, HJV, SLC40A1, TFR2 Amyloidosis (AMYLD): 6 genes (6,4 kb) APOA1, B2M, FGA, GSN, LYZ, TTR	3 - 5 w	E
Zellweger syndrome (ZWS) * Gene panel: ID084.00, 14 genes (22,3 kb) ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	3 - 5 w	E
3-Methylglutaconaciduria (MGCA) * Gene panel: ID249.01, 16 genes (18,9 kb) AGK, ATPAF2, ATP5F1D, ATP5F1E, AUH, CLPB, DNAJC19, ECHS1, HTRA2, OPA3, POLG, SERAC1, SUCLA2, TAFAZZIN, TIMM50, TMEM70	3 - 5 w	E
Microcephaly and Macrocephaly		
Galloway-Mowat syndrome (GAMOS) * Gene panel: ID251.01, 10 genes (12,5 kb) GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC	3 - 5 w	E
Growth abnormalities and macrocephaly * Gene panel: ID072.03 Growth abnormalities and macrocephaly: 30 genes (80,9 kb) AKT1, BRAF, CUL4B, DNMT3A, EED, EZH2, GPC3, GPC4, H1-4, HRAS, HUWE1, KRAS, NF1, NFIX, NRAS, NSD1, OFD1, PIK3CA, PPP1CB, PTEN, PTPN11, RAF1, RIT1, RNF125, RRAS2, SETD2, SHOC2, SOS1, SPRED1, SUZ12 Overgrowth syndrome with macrocephaly: 11 genes (33,0 kb) DNMT3A, EED, EZH2, GPC3, GPC4, NFIX, NSD1, OFD1, RNF125, SETD2, SUZ12 Noonan syndrome with macrocephaly: 10 genes (15,2 kb) BRAF, KRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1	3 - 5 w	E
Hydrocephalus * Gene panel: ID221.04 Hydrocephalus: 20 genes (57,9 kb) AKT3, CCDC88C, CCND2, CFAP43, FANCB, FLVCR2, FOXJ1, GPSM2, HYL1, KIF7, L1CAM, MPDZ, P4HB, PIK3R2, POMT1, SEC24D, SMARCC1, TRIM71, WDR81, ZIC3 Congenital hydrocephalus (HYC): 6 genes (27,7 kb) CCDC88C, L1CAM, MPDZ, SMARCC1, TRIM71, WDR81 Syndromes with hydrocephalus: 14 genes (29,0 kb) AKT3, CCND2, FANCB, FLVCR2, FOXJ1, GPSM2, HYL1, KIF7, L1CAM, P4HB, PIK3R2, POMT1, SEC24D, ZIC3	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Microcephaly and Macrocephaly		
Macrocephaly, comprehensive diagnostics * Gene panel: ID070.03 Macrocephaly, comprehensive diagnostics: 135 genes (364,6 kb) ABCC9, ADK, AKT1, AKT3, ALKBH8, AMER1, ANKH, APC2, ASPA, ASXL2, BRAF, BRWD3, CAMK2G, CCDC22, CCDC88C, CCND2, CDH2, CDKN1C, CHD1, CHD3, CHD8, CRADD, CSF1R, CUL4B, DIS3L2, DDX3X, DEAF1, DNMT3A, DVL1, DVL3, EED, EML1, EXT2, EZH2, FGFR3, FIBP, FMR1, FOXP1, GATAD2B, GCDH, GFAP, GLI3, GPC3, GPC4, GRIA3, GUSB, H1-4, HEPACAM, HERC1, HRAS, HSD17B4, HUWE1, IGBP1, INPPL1, KDM5C, KIF7, KPTN, KRAS, L1CAM, LAMB1, LBR, LZTR1, MAN2B1, MAP2K1, MAP2K2, MAPK1, MECP2, MED12, MITF, MLC1, MPDZ, MRAS, MSL3, MTOR, NF1, NFIB, NFIX, NONO, NRAS, NSD1, NXN, OFD1, OPHN1, PAK1, PHF21A, PIGA, PIGM, PIGN, PIGT, PIK3CA, PIK3R2, PKDCC, PPP1CB, PPP2R5D, PTCH1, PTCH2, PTEN, PTPN11, RAB39B, RAC1, RAF1, RIN2, RIT1, RNF125, RNF135, ROR2, RRAS2, SEC23B, SETD2, SHANK3, SHOC2, SHROOM4, SNX10, SOS1, SOS2, SPOP, SPRED1, SPRED2, STRADA, SUFU, SUZ12, TBC1D7, TCIRG1, TMC01, TNFRSF11A, TRIO, TRIP12, UPF3B, WASHC5, WDR81, WNT5A, ZBTB7A, ZBTB20, ZBTB42, ZDHHC9 Cowden syndrome (CWS): 4 genes (8,2 kb) AKT1, PIK3CA, PTEN, SEC23B Robinow syndrome (RRS, DRS): 5 genes (9,5 kb) DVL1, DVL3, NXN, ROR2, WNT5A Overgrowth syndromes: 15 genes (38,9 kb) CDKN1C, DIS3L2, DNMT3A, EED, EZH2, FIBP, GPC3, GPC4, NFIX, NSD1, OFD1, PTEN, RNF125, SETD2, SUZ12 RAS-related syndromes: 21 genes (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED1, sSPRED2 Intellectual developmental disorders with macrocephaly: 49 genes (147,9 kb) ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, H1-4, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMC01, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9	4 - 6 w	E
Microcephaly, comprehensive diagnostics * Gene panel: ID069.02 Microcephaly, comprehensive diagnostics: 129 genes (371,7 kb) ADARB1, ANKLE2, ARCN1, ARFGEF2, ASPM, ATR, BLM, CARS1, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CENPT, CEP135, CEP152, CEP63, CIT, CKAP2L, COBP1, COBP2, CRIPT, CTNNB1, CTU2, DIAPH1, DNA2, DNMT3A, DONSON, DPP6, DYNC1I2, DYRK1A, EFTUD2, EIF2S3, ERCC1, ERCC2, ERCC5, ERCC6, EXOC8, EXT2, FOXG1, GEMIN4, GPT2, IER3IP1, KAT6A, KATNB1, KCNA4, KIF11, KIF14, KNL1, LAGE3, LMNB1, LMNB2, MCPH1, MED17, MFSD2A, MSMO1, MTHFS, MYCN, NARS1, NBN, NCAPD2, NCAPD3, NCAPH, NDE1, NHEJ1, NIN, NSMCE2, NUP107, NUP133, NUP37, OCLN, OSGEP, PCDH12, PCNT, PHC1, PHGDH, PLAA, PLEKHG2, PLK4, PNKP, PPP1R15B, PQBP1, PRUNE1, PSAT1, PUS3, PUS7, QARS1, RAB18, RAB3GAP1, RAB3GAP2, RAD50, RBBP8, RTTN, SARS1, SASS6, SLC1A4, SLC25A19, SLC9A6, SMPD4, SPOP, STAG2, STAMBP, STIL, SVBP, TBC1D20, THOC6, TMX2, TOP3A, TP53RK, TPRKB, TRAIP, TRAPPC6B, TRAPPC14, TRIO, TRMT10A, TUBGCP2, TUBGCP4, TUBGCP6, VARS1, VPS13B, WDFY3, WDR4, WDR62, WDR73, XRCC4, YIPF5, ZEB2, ZNF335 Primary microcephaly (MCPH): 27 genes (110,8 kb) ANKLE2, ASPM, CDK6, CIT, CDK5RAP2, CENPE, CENPJ, CEP135, CEP152, COBP2, KIF14, KNL1, MCPH1, MFSD2A, NCAPD2, NCAPD3, NCAPH, NUP37, PHC1, SASS6, STIL, TRAPPC14, WDFY3, WDR62, ZNF335 Seckel syndrome (SCKL): 9 genes (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP Warburg Micro syndrome (WARBM): 4 genes (8,9 kb) RAB18, RAB3GAP1, RAB3GAP2, TBC1D20 Galloway-Mowat syndrome (GAMOS): 8 genes (11,4 kb) LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73 Cerebrooculofacioskeletal syndrome (COFS): 4 genes (11,3 kb) ERCC1, ERCC2, ERCC5, ERCC6 Microcephaly and chorioretinopathy (MCCRP): 4 genes (13,5 kb) PLK4, TUBGCP4, TUBGCP6, KIF11 Neurodevelopmental disorder with microcephaly (NEDM): 21 genes (39,7 kb) ADARB1, COBP1, CTNNB1, DYNC1I2, EXOC8, GEMIN4, GPT2, MFSD2A, MTHFS, NARS1, PLAA, PRUNE1, PUS3, SPOP, STAG2, TMX2, TRAPPC6B, SARS1, SMPD4, SVBP, VARS1	4 - 6 w	E
Intellectual developmental disorder and macrocephaly * Gene panel: ID131.03 Intellectual developmental disorder and macrocephaly: 48 genes (147,3 kb) ADK, ALKBH8, APC2, BRWD3, CAMK2G, CHD3, CHD8, CRADD, CUL4B, DDX3X, DEAF1, FMR1, GATAD2B, GRIA3, HEPACAM, HUWE1, IGBP1, KDM5C, KIF7, KPTN, L1CAM, MECP2, MED12, MLC1, MSL3, MTOR, NFIB, NONO, OPHN1, PAK1, PHF21A, PPP2R5D, PTEN, RAB39B, RAC1, RNF125, SETD2, SHANK3, SHROOM4, SPOP, TBC1D7, TMC01, TRIO, TRIP12, UPF3B, ZBTB7A, ZBTB20, ZDHHC9 Intellectual developmental disorder, autosomal dominant (MRD) and macrocephaly: 21 genes (70,8 kb) CAMK2G, CHD3, CHD8, DEAF1, GATAD2B, HEPACAM, MTOR, NFIB, PAK1, PHF21A, PPP2R5D, PTEN, RAC1, RNF125, SETD2, SHANK3, SPOP, TRIO, TRIP12, ZBTB7A, ZBTB20 Intellectual developmental disorder, autosomal recessive (MRT) and macrocephaly: 10 genes (20,2 kb) ADK, ALKBH8, APC2, CRADD, KIF7, KPTN, MLC1, TBC1D7, TMC01, ZBTB7A Intellectual developmental disorder, X-linked (MRX) and macrocephaly: 18 genes (57,9 kb) BRWD3, CUL4B, DDX3X, FMR1, GRIA3, HUWE1, IGBP1, KDM5C, L1CAM, MECP2, MED12, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B, ZDHHC9	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Microcephaly and Macrocephaly Intellectual developmental disorder and microcephaly * Gene panel: ID129.02 Intellectual developmental disorder and microcephaly: 80 genes (229,8 kb) ACSL4, ADAT3, ATRX, AUTS2, CAMK2B, CAMK2G, CASK, CERT1, CHAMP1, CKAP2L, CTCF, CTNNB1, DDX3X, DPP6, DYRK1A, EDC3, EFTUD2, EHMT1, EIF2S3, GPT2, GRIN2B, HCFC1, HIVEP2, HNMT, HNRNP2, HUWE1, IQSEC1, KDM5C, KIF11, KIF1A, L1CAM, LINGO1, LINS1, MBD5, MBOAT7, MCPH1, MECP2, METTL5, MYCN, NEXMIF, NSUN2, OGT, PAK3, PGAP1, PHF6, POGZ, POLA1, PPP2R1A, PQBP1, PUS3, RAC1, RBBP8, RLIM, RPL10, SET, SETD2, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TAF1, TAF13, TAF2, THOC2, TLK2, TRAPPC9, TRIO, TRMT1, TTI2, WDR11, WDR73, ZBTB18, ZC4H2, ZEB2 Intellectual developmental disorder, autosomal dominant (MRD) and microcephaly: 32 genes (100,5 kb) AUTS2, CAMK2B, CAMK2G, CERT1, CHAMP1, CTCF, CTNNB1, DPP6, DYRK1A, EFTUD2, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5, MYCN, POGZ, PPP2R1A, RAC1, SET, SETD2, SMARCA4, SMARCB1, SMARCE1, SOX11, SOX4, SYNGAP1, TLK2, TRIO, ZBTB18, ZEB2 Intellectual developmental disorder, autosomal recessive (MRT) and microcephaly: 21 genes (41,0 kb) ADAT3, CKAP2L, EDC3, GPT2, HNMT, LINGO1, LINS1, MBOAT7, MCPH1, METTL5, NSUN2, PGAP1, PUS3, RBBP8, TAF13, TAF2, TRAPPC9, TRMT1, TTI2, WDR11, WDR73 Intellectual developmental disorder, X-linked (MRX) and microcephaly: 27 genes (88,3 kb) ACSL4, ATRX, CASK, DDX3X, EIF2S3, HCFC1, HNRNP2, HUWE1, IQSEC1, KDM5C, L1CAM, MECP2, NEXMIF, OGT, PAK3, PHF6, POLA1, PQBP1, RLIM, RLIM, SHROOM4, SLC16A2, SLC6A8, SLC9A6, TAF1, THOC2, ZC4H2 Pontocerebellar hypoplasia (PCH) * Gene panel: ID071.02, 26 genes (60,5 kb) AMPD2, CASK, CDC40, CHMP1A, CLP1, COASY, EXOSC1, EXOSC3, EXOSC8, EXOSC9, MINPP1, PCLO, PPIL1, RARS2, RELN, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN2, TSEN15, TSEN34 TSEN54, VPS51, VPS53, VRK1 Primary microcephaly, autosomal recessive (MCPH) * Gene panel: ID031.02, 25 genes (107,1 kb) ANKLE2, ASPM, CDK6, CIT, CDK5RAP2, CENPE, CENPJ, CEP135, CEP152, COPB2, KIF14, KNL1, MAP11, MCPH1, MFSD2A, NCAPD2, NCAPD3, NCAPH, NUP37, PHC1, SASS6, STIL, WDFY3, WDR62, ZNF335 Seckel syndrome (SCKL) * Gene panel: ID113.00, 9 genes (33,5 kb) ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAP	4 - 6 w 3 - 5 w 4 - 6 w 3 - 5 w	E E E E
Mitochondrial Diseases Coenzyme Q10 deficiency (COQ10D) * Gene panel: ID225.01, 15 genes (18,7 kb) ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2 Combined defect of oxidative phosphorylation (COXPD) * Gene panel: ID287.01, 60 genes (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C20RF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCO3, RMND1, SFXN4, SLC25A26, TARS2, TMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2 Deficiency of mitochondrial complexes I to V (MCDN) * Gene panel: ID074.02 Deficiency of mitochondrial complexes I to V (MCDN): 76 genes (57,0 kb) ACAD9, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MD, ATPAF2, BCS1L, COA3, COA5, COA6, COA8, COX4I1, COX5A, COX6A2, COX6B1, COX8A, COX10, COX14, COX15, COX20, COXFA4, CYC1, FOXRED1, LRPPRC, LYRM7, MTFMT, NDUFA1, NDUFA2, NDUFA6, NDUFA9, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB3, NDUFB8, NDUFB9, NDUFB10, NDUFB11, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, PET100, PET117, SCO1, SCO2, SDHA, SDHAF1, SDHB*, SDHD, SURF1, TACO1, TIMMDC1, TMEM70, TMEM126B, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRC3, UQCRC4, UQCRC5, UQCRC6, UQCRC7, UQCRC8, UQCRC9, UQCRC10, UQCRC11, UQCRC12, UQCRC13, UQCRC14, UQCRC15, UQCRC16, UQCRC17, UQCRC18, UQCRC19, UQCRC20, UQCRC21, UQCRC22, UQCRC23, UQCRC24, UQCRC25, UQCRC26, UQCRC27, UQCRC28, UQCRC29, UQCRC30, UQCRC31, UQCRC32, UQCRC33, UQCRC34, UQCRC35, UQCRC36, UQCRC37, UQCRC38, UQCRC39, UQCRC40, UQCRC41, UQCRC42, UQCRC43, UQCRC44, UQCRC45, UQCRC46, UQCRC47, UQCRC48, UQCRC49, UQCRC50, UQCRC51, UQCRC52, UQCRC53, UQCRC54, UQCRC55, UQCRC56, UQCRC57, UQCRC58, UQCRC59, UQCRC60, UQCRC61, UQCRC62, UQCRC63, UQCRC64, UQCRC65, UQCRC66, UQCRC67, UQCRC68, UQCRC69, UQCRC70, UQCRC71, UQCRC72, UQCRC73, UQCRC74, UQCRC75, UQCRC76, UQCRC77, UQCRC78, UQCRC79, UQCRC80, UQCRC81, UQCRC82, UQCRC83, UQCRC84, UQCRC85, UQCRC86, UQCRC87, UQCRC88, UQCRC89, UQCRC90, UQCRC91, UQCRC92, UQCRC93, UQCRC94, UQCRC95, UQCRC96, UQCRC97, UQCRC98, UQCRC99, UQCRC100, UQCRC101, UQCRC102, UQCRC103, UQCRC104, UQCRC105, UQCRC106, UQCRC107, UQCRC108, UQCRC109, UQCRC110, UQCRC111, UQCRC112, UQCRC113, UQCRC114, UQCRC115, UQCRC116, UQCRC117, UQCRC118, UQCRC119, UQCRC120, UQCRC121, UQCRC122, UQCRC123, UQCRC124, UQCRC125, UQCRC126, UQCRC127, UQCRC128, UQCRC129, UQCRC130, UQCRC131, UQCRC132, UQCRC133, UQCRC134, UQCRC135, UQCRC136, UQCRC137, UQCRC138, UQCRC139, UQCRC140, UQCRC141, UQCRC142, UQCRC143, UQCRC144, UQCRC145, UQCRC146, UQCRC147, UQCRC148, UQCRC149, UQCRC150, UQCRC151, UQCRC152, UQCRC153, UQCRC154, UQCRC155, UQCRC156, UQCRC157, UQCRC158, UQCRC159, UQCRC160, UQCRC161, UQCRC162, UQCRC163, UQCRC164, UQCRC165, UQCRC166, UQCRC167, 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Diseases/Diagnostics	TAT	Material
Mitochondrial Diseases		
MELAS syndrome ^{co} Gene panel: ID700.01, 22 genes (9,2 kb) MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND3, MT-ND4, MT-ND5, MT-ND6, MT-RNR2, MT-TC, MT-TE, MT-TF, MT-TH, MT-TK, MT-TL1, MT-TL2, MT-TQ, MT-TS1, MT-TS2, MT-TV, MT-TW	3 - 5 w	E
Mitochondrial diseases, nuclear type * Gene panel: ID704.01, 312 genes (406,1 kb) AARS2, ABCB7, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH4A1, ALDH6A1, AMACR, AMT, ANO10, APTX, ATL1, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MK, ATPAF2, AUH, BCAT2, BCKDHA, BCKDHB, BCS1L, BOLA3, BTD, C1QBP, CARS2, CISD2, COA3, COA5, COA6, COA8, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX5A, COX6A2, COX6B1, CPS1, CPT1A, CPT2, CYB5R3, CYC1, CYCS, CYP27A1, D2HGDH, DARS2, DBT, DECR1, DGUOK, DHODH, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNMT1L, EARS2, ELAC2, ERCC6, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBP1, FBXL4, FH, FOXRED1, FXN, GAMT, GARS1, GATB, GATC, GATM, GCDH, GCK, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GTPBP3, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HK1, HLCS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSPD1, HTRA2, IDH1, IDH2, IDH3B, ISCU, IVD, KARS1, KIF1B, KIF5A, L2HGDH, LARS2, LIAS, LRPPRC, LYRM4, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MFN2, MGME1, MICOS13, MIEF2, MIPEP, MLYCD, MMAA, MMBAB, MMADHC, MMUT, MPV17,MRM2, MRPL12, MRPL3, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS25, MRPS28, MRPS34, MRPS7, MTFMT, MTO1, MTPAP, MTRFR, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF8, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NSUN3, NUBPL, OAT, OPA1, OPA3, OTC, OXCT1, PANK2, PARK7, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PET117, PINK1, PNK1, PNKD, PNPLA2, PNPT1, POLG, POLG2, PPOX, PRKN, PTCD3, PUS1, QRSL1, RARS2, REEP1, RMND1, RMRP, RNASEH1, RRM2B, SACS, SAMHD1, SARS2, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A10, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC33A1, SLC6A8, SPART, SPAST, SPG7, STAR, SUCLA2, SUCLG1, SURF1, TACO1, TAFAZZIN, TARS2, TFAM, TIMM22, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TOP3A, TPK1, TRIT1, TRMT10C, TRMT5, TRMU, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UNG, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRRS1, UQCRQ, VARS2, WFS1, WWOX, XPNPEP3, YARS2, YWHAE	4 - 6 w	E
Mitochondrial DNA depletion syndrome (MTDPS) * Gene panel: ID324.00, 19 genes (25,6 kb) AGK, DGUOK, FBXL4, MGME1, MPV17, MRM2, OPA1, POLG, POLG2, RRM2B, SLC25A4, SLC25A10, SLC25A21, SUCLA2, SUCLG1, TFAM, TK2, TWNK, TYMP	3 - 5 w	E
Mitochondrial genome ^{co} Gene panel: ID703.00, 37 genes (11,4 kb) 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	3 - 5 w	E
Mitochondrial myopathy / encephalopathy ^{co} Gene panel: ID702.00, 27 genes (7,0 kb) MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND2, MT-ND5, 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-TW	3 - 5 w	E
Progressive external opthalmoplegia with mtDNA deletions (PEOA, PEOB) * Gene panel: ID300.00 Progressive external opthalmoplegia with mtDNA deletions (PEOA, PEOB): 10 genes (17,9 kb) DGUOK, DNA2, POLG, POLG2, RNASEH1, RRM2B, SLC25A4, TK2, TOP3A, TWNK Progressive external opthalmoplegia, autosomal dominant (PEOA): 6 genes (12,4 kb) DNA2, POLG, POLG2, RRM2B, SLC25A4, TWNK Progressive external opthalmoplegia, autosomal recessive (PEOB): 5 genes (9,2 kb) DGUOK, POLG, RNASEH1, TK2, TOP3A	3 - 5 w	E
Multisystemic Malformation Syndromes		
Alagille syndrome (ALGS) * Gene panel: ID112.00, 2 genes (11,1 kb) JAG1, NOTCH2	3 - 5 w	E
Alport syndrome (ATS) * Gene panel: ID099.00, 4 genes (21,0 kb) COL4A3, COL4A4, COL4A5, MYH9	3 - 5 w	E
Bardet-Biedl syndrome (BBS) * Gene panel: ID093.02, 21 genes (39,0 kb) ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDCPC	3 - 5 w	E
CHARGE syndrome * Gene panel: ID307.00, 3 genes (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 w	E
Combined defect of oxidative phosphorylation (COXPD) * Gene panel: ID287.01, 60 genes (80,3 kb) AARS2, AIFM1, ATP5F1A, C1QBP, C20RF69, CARS2, CRLS1, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL39, MRPL44, MRPL49, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NFS1, NSUN3, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Multisystemic Malformation Syndromes		
Coffin-Siris syndrome (CSS) * Gene panel: ID118.02, 14 genes (46,5 kb) ARID1A, ARID1B, ARID2, BICRA, DPF2, PHF6, SMARCA2, SMARCA4, SMARCB1, SMARCC2, SMARCD1, SMARCE1, SOX4, SOX11	3 - 5 w	E
Congenital disorder of glycosylation (CDG) * Gene panel ID035.02 Congenital disorder of glycosylation (CDG): 51 genes (74,7 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, MAGT1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3 Congenital disorder of glycosylation, type I (CDG1): 29 genes (39,6 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Congenital disorder of glycosylation, type II (CDG2): 22 genes (35,1 kb) ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, TMEM165, TMEM199	3 - 5 w	E
Cornelia de Lange syndrome (CDLS) * Gene panel: ID033.02, 8 genes (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 w	E
FG syndrome (FGS) * Gene panel: ID215.00, 3 genes (17,2 kb) CASK, FLNA, MED12	3 - 5 w	E
Kabuki syndrome (KABUK) * Gene panel: ID127.00, 2 genes (20,8 kb) KDM6A, KMT2D	3 - 5 w	E
Marfan syndrome (MFS) * Gene panel: ID022.00, 3 genes (11,8 kb) FBN1, TGFBR1, TGFBR2	2 - 4 w	E
Malformation syndromes with predominantly facial involvement * Gene panel: ID279.00 Malformation syndromes with predominantly facial involvement: 25 genes (90,9 kb) C2CD3, COL11A2, CPLANE1, DDX59, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GRIP1, IFT57, INTU, KIAA0753, MEGF8, MYH3, MYMK, MYT1, OFD1, RAB23, RBM10, TCTN3, TGDS, TMEM107, TNNT2, TWIST1 Acrocephalosyndactyly syndrome (ACS): 6 genes (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1 Orofaciodigital syndrome (OFD): 9 genes (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107 Fraser syndrome (FRASRS): 3 genes (24,8 kb) FRAS1, FREM2, GRIP1 Pierre Robin syndrome: 4 genes (9,7 kb) COL11A2, MYMK, RBM10, TGDS	3 - 5 w	E
Noonan syndrome (NS) * Gene panel: ID023.06, 16 genes (27,4 kb) BRAF, CBL, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2	3 - 5 w	E
Orofaciodigital syndrome (OFD) * Gene panel: ID265.00, 9 genes (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 w	E
RAS-related disorders * Gene panel: ID015.05 RAS-related disorders: 21 genes (40,1 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, PPP1CB, SHOC2, SOS1, SOS2, SPRED1, SPRED2 Noonan syndrome (NS): 15 genes (24,7 kb) BRAF, KRAS, LZTR1, MAPK1, MRAS, NRAS, PPP1CB, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED2 Cardiofaciocutaneous syndrome (CFC): 4 genes (5,3 kb) BRAF, KRAS, MAP2K1, MAP2K2 LEOPARD syndrome (LPRD): 3 genes (6,0 kb) BRAF, PTPN11, RAF1	3 - 5 w	E
Rubinstein-Taybi syndrome (RSTS) * Gene panel: ID142.01, 3 genes (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 w	E
Stickler syndrome (STL) * Gene panel: ID062.00, 6 genes (21,2 kb) COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3	3 - 5 w	E
Tuberous sclerosis (TSC) * Gene panel: ID332.00, 2 genes (8,9 kb) TSC1, TSC2	2 - 4 w	E

Diseases/Diagnostics	TAT	Material
Multisystemic Malformation Syndromes		
VACTERL association * Gene panel: ID258.02, 27 genes (72,8 kb) BRCA2, CHD7, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FGF8, FOXF1, GLI3, HAAO, HOXD13, HSPA6, MNX1, RECQL4, SALL1, KYNU, MYCN, NADSYN1, RAD51C, TRAP1, WBP11, ZIC3	3 - 5 w	E
Weill-Marchesani syndrome (WMS) * Gene panel: ID230.00, 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 w	E
Zellweger syndrome (ZWS) * Gene panel: ID084.00, 14 genes (22,3 kb) ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	3 - 5 w	E
Muscular Diseases		
Arthrogryposis * Gene panel: ID200.01 Arthrogryposis: 61 genes (209,7 kb) ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, CNTNAP1, DHCR24, DNMT2, DOK7, DSE, ECEL1, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERGIC1, FBN2, FKBP10, FLVCR2, GBE1, GLDN, GLE1, KLHL40, KLHL41, LGI4, LMOD3, NALCN, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, MYLPF, NEB, NEK9, NUP88, PIEZO2, PIP5K1C, PLOD2, RAPSN, RYR1, SCYL2, SYNE1, TNNI2, TNNT3, TOR1A, TPM2, TPM3, TRIP4, TRPV4, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2 Arthrogryposis multiplex congenita (AMC): 6 genes (58,2 kb) ERGIC1, TOR1A, LGI4, NEB, SCYL2, SYNE1 Distal arthrogryposis (DA): 11 genes (40,3 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2*, TNNT3, TPM2, UBA1 Lethal congenital contracture syndrome (LCCS): 11 genes (31,5 kb) ADCY6, ADGRG6, CNTNAP1, DNMT2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42 Fetal akinesia deformation sequence (FADS): 4 genes (7,6 kb) DOK7, MUSK, NUP88, RAPSN	4 - 6 w	E
Centronuclear myopathy (CNM) * Gene panel: ID257.00, 7 genes (21,4 kb) CCDC78, DNMT2, BIN1, MAP3K20, MTMR14, MTM1, SPEG	3 - 5 w	E
Charcot-Marie-Tooth disease, axonal type (CMT2, HMSN) * Gene panel: ID052.03 Charcot-Marie-Tooth disease, axonal type (CMT2, HMSN): 45 genes (111,8 kb) AARS1, AIFM1, ATP1A1, CADM3, COX6A1, DHTKD1, DNMT2, DYNC1H1, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, JAG1, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PNKP, PRPS1, RAB7A, SLC12A6, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth disease, axonal, autosomal dominant (CMT2): 26 genes (74,2 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNMT2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, JAG1, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, SLC12A6, TRPV4, VCP Charcot-Marie-Tooth disease, axonal, autosomal recessive (CMT2): 10 genes (24,4 kb) GDAP1, IGHMBP2, LMNA, LRSAM1, MFN2, MME, MPV17, PNKP, SPG11, TRIM2 Charcot-Marie-Tooth disease, intermediate (CMTDI, CMTRI): 11 genes (18,5 kb) COX6A1, DNMT2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth disease, X-linked (CMTX): 4 genes (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Charcot-Marie-Tooth disease with optic atrophy (CMT6): 3 genes (4,5 kb) MFN2, PDXK, SLC25A46	4 - 6 w	E
Charcot-Marie-Tooth disease, demyelinating type (CMT1, CMT4, HMSN) * Gene panel: ID051.03 Charcot-Marie-Tooth disease, demyelinating type (CMT1, CMT4, HMSN): 29 genes (69,5 kb) CNTNAP1, COX6A1, DNMT2, EGR2, FBLN5, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, ITPR3, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1 Charcot-Marie-Tooth disease, demyelinating, autosomal dominant (CMT1): 11 genes (19,0 kb) EGR2, FBLN5, GDAP1, ITPR3, LITAF, MPZ, NEFL, POLR3B, PMP2, PMP22 Charcot-Marie-Tooth disease, demyelinating, autosomal recessive (CMT4): 12 genes (33,8 kb) EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth disease, intermediate (CMTDI, CMTRI): 11 genes (18,5 kb) COX6A1, DNMT2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Neuropathy of Déjerine-Sottas (CMT3, DSS): 4 genes (7,1 kb) EGR2, MPZ, PMP22, PRX Congenital hypomyelinating neuropathy (CHN): 3 genes (6,3 kb) EGR2, MPZ, CNTNAP1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Muscular Diseases		
Charcot-Marie-Tooth disease (CMT, HMSN), comprehensive diagnostics * Gene panel: ID312.02 Charcot-Marie-Tooth disease (CMT, HMSN), comprehensive diagnostics: 87 genes (217,3 kb) AARS1, AIFM1, ARHGEF10, ATP1A1, ATP7A, BSCL2, CADM3, CNTNAP1, COQ7, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A7, SORD, SPG11, SPTAN1, SPTLC1, SURF1, SYT2, TRIM2, TRPV4, VCP, VRK1, VWA1, WARS1, YARS1 Charcot-Marie-Tooth disease, demyelating (CMT1, CMT4, HMSN1): 20 genes (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth disease, axonal (CMT2): 35 genes (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth disease, intermediate (CMTDI, CMTRI): 11 genes (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth disease, X-linked (CMTX): 4 genes (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophic neuropathy of Déjerine-Sottas (CMT3, DSS): 4 genes (7,1 kb) EGR2, MPZ, PMP22, PRX Peripheral neuropathy with optic atrophy (HMSN6): 3 genes (4,5 kb) MFN2, PDXK, SLC25A46 Congenital hypomyelating neuropathy (CHN): 3 genes (6,3 kb) EGR2, MPZ, CNTNAP1 Distal hereditary motor neuropathy (HMND, HMNR): 24 genes (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	4 - 6 w	E
Congenital myasthenic syndrome (CMS) * Gene panel: ID130.00 Congenital myasthenic syndrome (CMS): 25 genes (55,9 kb) AGRN, ALG2, ALG14, CHAT, CHRNA1, CHRN1, CHRN2, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, LRP4, MUSK, MYO9A, PREPL, RAPSN, SCN4A, SLC5A7, SLC18A3, SLC25A1, SNAP25, SYT2, VAMP1 Congenital myasthenic syndrome (CMS), presynaptic: 8 genes (21,9 kb) AGRN, CHAT, MYO9A, SLC5A7, SLC18A3, SLC25A1, SYT2, VAMP1 Congenital myasthenic syndrome (CMS), synaptic or postsynaptic: 18 genes (40,1 kb) AGRN, ALG2, ALG14, CHRNA1, CHRN1, CHRN2, CHRNE, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, LRP4, MUSK, PREPL, RAPSN, SCN4A, SNAP25	3 - 5 w	E
Congenital myopathy (CMYP) * Gene panel: ID212.02 Congenital myopathy (CMYP): 50 genes (269,7 kb) ACTA1, ACTN2, BAG3, BIN1, CACNA1S, CCDC78, CFL2, CNTN1, CRYAB, DES, DNAJB4, DNM2, FLNC, FXR1, HACD1, KBTBD13, KLHL40, KLHL41, KY, LDB3, LMOD2, MAP3K20, MEGF10, MTM1, MTMR14, MYBPC1, MYH2, MYH7, MYL1, MYL2, MYOD1, MYOT, MYPN, NEB, PAX7, PYROXD1, RYR1, RYR3, SCN4A, SVIL, TPM2, SELENON, SPEG, STAC3, TNNC2, TNNT1, TPM2, TPM3, TTN, UNC45B Central core myopathy (CCD): 1 gene (15,1 kb) RYR1 Nemaline myopathy (NEM): 11 genes (40,6 kb) ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, TNNT1, TPM2, TPM3, MYPN, NEB Centronuclear myopathy (CNM): 7 genes (20,3 kb) BIN1, CCDC78, DNM2, MAP3K20, MTM1, MTMR14, SPEG Myofibrillar myopathy (MFM): 12 genes (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYOT, PYROXD1, TTN	3 - 5 w	E
Distal arthrogryposis (DA) * Gene panel: ID196.01, 11 genes (40,5 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNT2, TNNT3, TPM2, UBA1	3 - 5 w	E
Distal hereditary motor neuronopathy (HMND, HMNR) * Gene panel: ID254.01 Distal hereditary motor neuronopathy (HMND, HMNR): 24 genes (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Distal hereditary motor neuronopathy, autosomal dominant type (HMND): 15 genes (38,4 kb) BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1 Distal hereditary motor neuronopathy, autosomal recessive type (HMNR): 10 genes (14,4 kb) ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SORD, VRK1, VWA1	3 - 5 w	E
Distal myopathy (MPD) * Gene panel: ID328.01, 30 genes (186,0 kb) ACTA1, ACTN2, ADSS1, ANO5, BAG3, CAV3, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FLNC, GNE, HNRNPA1, HNRPA2B1, HSPB8, KY, LDB3, MATR3, MYH7, MYOT, NEB, PYROXD1, SMPX, SQSTM1, TIA1, TCAP, TTN, VCP	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Muscular Diseases		
Emery-Dreifuss muscular dystrophy (EDMD) * Gene panel: ID121.00, 6 genes (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43	3 - 5 w	E
Nemaline myopathy (NEM) * Gene panel: ID199.00, 11 genes (40,6 kb) ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3	3 - 5 w	E
Lethal congenital contracture syndrome (LCCS) * Gene panel: ID197.00, 12 genes (34,6 kb) ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42	3 - 5 w	E
Limb-girdle muscular dystrophy (LGMD) * Gene panel: ID122.03 Limb-girdle muscular dystrophy (LGMD): 37 Gene (196,8 kb) ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DNAJB6, DPM3, DYSF, FKRP, FKTN, GMPPB, HNRNPDL, JAG2, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN Limb-girdle muscular dystrophy, autosomal recessive (LGMDR): 34 Gene (191,8 kb) ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DPM3, DYSF, FKRP, FKTN, GMPPB, JAG2, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TRAPPC11, TRIM32, TT Limb-girdle muscular dystrophy, autosomal dominant (LGMDD): 7 genes (23,2 kb) CAPN3, COL6A1, COL6A2, COL6A3, DNAJB6, HNRNPDL, TNPO3 Limb-girdle muscular dystrophy-dystroglycanopathy (MDDGC): 11 genes (17,6 kb) CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, POMGNT1, POMGNT2, POMK, POMT1, POMT2 Ullrich muscular dystrophy (UCMD): 3 genes (15,7 kb) COL6A1, COL6A2, COL6A3 Bethlem myopathy (BTHLM): 3 genes (15,7 kb) COL6A1, COL6A2, COL6A3	4 - 6 w	E
Muscular diseases, comprehensive diagnostics * Gene panel: ID336.00 Muscular diseases, comprehensive diagnostics: 246 genes (749,8 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, AGRN, ALDOA, ALG14, ALG2, AMPD1, ANO5, ASAH1, ASCC1, ATP2A1, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, BVES, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC78, CFL2, CHAT, CHCHD10, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CLCN1, CNBP, CNTN1, COL12A1, COL13A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRPPA, CRYAB, DAG1, DES, DGUOK, DMD, DNA2, DNAJB6, DNM2, DNMT3B, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, ECEL1, EGR2, EMD, ENO3, EPG5, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, FDX2, FHL1, FKBP14, FKRP, FKTN, FLAD1, FLNC, FXR1, GAA, GARS1, GBE1, GDAP1, GFPT1, GMPPB, GNE, GYG1, GYS1, HADH, HADHA, HADHB, HINT1, HNRNPA1, HNRNPA2B1, HNRNPDL, HSPB8, IGHMBP2, INPP5K, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LAMA2, LAMA5, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LIG3, LIMS2, LMNA, LMOD3, LPIN1, LRIF1, LRP4, MAP3K20, MATR3, MEGF10, MFN2, MGME1, MICU1, MPV17, MPZ, MTM1, MTMR14, MTRFR, MUSK, MYBPC1, MYH14, MYH2, MYH3, MYH7, MYH8, MYL1, MYL2, MYMK, MYO18B, MYO9A, MYOT, MYPN, NEB, NEFL, OPA1, OPA1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PLEC, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, PREPL, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RNASEH1, RRM2B, RXYLT1, RYR1, RYR3, SCN4A, SC02, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SLC18A3, SLC22A5, SLC25A1, SLC25A20, SLC25A26, SLC25A3, SLC25A32, SLC25A4, SLC52A3, SLC5A7, SMCHD1, SMN1, SNAP25, SPEG, SPG7, SPG11, SQSTM1, STAC3, STIM1, SUCLA2, SUCLG1, SVIL, SYNE1, SYNE2, SYT2, TAFAZZIN, TCAP, TFAM, TIA1, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRIP4, TRMT5, TRPV4, TSFM, TTN, TUBB3, TWNK, TYMP, UBA1, UNC45B, VAMP1, VCP, VMA21, VRK1, YARS2 Congenital, distal and metabolic myopathies: 110 genes (382,6 kb) ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTN2, ADSS1, AGK, AGL, ALDOA, AMPD1, ANO5, BAG3, BIN1, CAV3, CCDC78, CFL2, CNTN1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, ECEL1, SELENON, ENO3, ETFA, ETFB, ETFDH, FLAD1, FLNC, FXR1, GAA, GBE1, GNE, GYG1, GYS1, HADH, HADHA, HADHB, ISCU, ITGA7, KBTBD13, KLHL40, KLHL41, KY, LDB3, LAMP2, LDHA, LIG3, LMOD3, LPIN1, MAP3K20, MATR3, MEGF10, MGME1, MTM1, MTMR14, MYBPC1, MYH2, MYH3, MYH7, MYH8, MYMK, MYOT, MYPN, NEB, TNNI2, OPA1, PAX7, PFKM, PGK1, PGM1, PHKA1, PHKB, PIEZO2, PNPLA2, POLG, POLG2, PRKAG2, PUS1, PYGM, PYROXD1, RBCK1, RRM2B, RYR1, SLC22A5, SLC25A20, SLC25A4, SPEG, STAC3, SUCLA2, SUCLG1, TAFAZZIN, TCAP, TIA1, TK2, TNNT1, TNNT3, TPM2, TPM3, TTN, TYMP, UBA1, YARS2 Limb-girdle muscular dystrophy and other muscular dystrophies: 47 genes (270,0 kb) ANO5, B3GALNT2, B4GAT1, BVES, CAPN3, COL12A1, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DMD, DNAJB6, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GMPPB, HNRNPDL, LAMA2, LARGE1, LIMS2, LMNA, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, RXYLT1, SGCA, SGCB, SGCD, SGCG, SYNE1, SYNE2, TCAP, TMEM43, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN	4 - 6 w	E
Muscular diseases with cardiac involvement * Gene panel: ID123.03 Muscular diseases with cardiac involvement: 19 genes (193,6 kb) BAG3, CRYAB, DES, DMD, EMD, FHL1, FLNC, KY, LDB3, LMNA, MYL2, MYOT, PYROXD1, SVIL, SYNE1, SYNE2, TMEM43, TTN, UNC45B Myofibrillar myopathy (MFM): 12 genes (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Emery-Dreifuss muscular dystrophy (EMDM): 6 genes (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43 Duchenne/Becker muscular dystrophy (DMD, BMD): 1 gene (11,1 kb) DMD	4 - 6 w	E
Myofibrillar myopathy (MFM) * Gene panel: ID161.01, 12 genes (130,7 kb) BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Muscular Diseases		
Myotonia * Gene panel: ID255.00, 5 genes (25,0 kb) ATP2A1, CLCN1, HINT1, HSPG2, SCN4A	3 - 5 w	E
Muscular dystrophy-dstroglycanopathy (MDDG) * Gene panel: ID179.00 Muscular dystrophy-dstroglycanopathy (MDDG): 15 genes (24,0 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muscular dystrophy-dstroglycanopathy with brain und eye anomalies (MDDGA): 14 genes (22,8 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1 Muscular dystrophy-dstroglycanopathy with or without impaired intellectual development (MDDGB): 8 genes (13,1 kb) DPM3, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMT1, POMT2 Muscular dystrophy-dstroglycanopathy, limb-girdle (MDDGC): 11 genes (18,5 kb) CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, POMGNT1, POMGNT2, POMK, POMT1, POMT2	3 - 5 w	E
Periodic paralysis * Gene panel: ID253.00, 7 genes (16,6 kb) CACNA1S, KCNE3, KCNJ2, KCNJ5, KCNJ12, KCNJ18, SCN4A	3 - 5 w	E
Spinal muscular atrophy (SMA) * Gene panel: ID152.02 Spinal muscular atrophy (SMA): 39 genes (87,9 kb) AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1 Proximal spinal muscular atrophy (SMA): 13 genes (34,2 kb) AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB Distal spinal muscular atrophy (DSMA, HMN): 24 genes (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	3 - 5 w	E
Walker-Warburg syndrome (WWS, MDDGA) * Gene panel: ID178.00, 14 genes (23,7 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 w	E
Neurodegenerative Diseases		
Alzheimer disease (AD) * Gene panel: ID157.01, 7 genes (16,8 kb) ABCA7, ADAM10, APOE, APP, PSEN1, PSEN2, TF	3 - 5 w	E
Amyotrophic lateral sclerosis (ALS) * Gene panel: ID209.03 Amyotrophic lateral sclerosis (ALS): 35 genes (80,2 kb) ALS2, ANG, ANXA11, ATXN2, C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, LRP12, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1*, SPG11, SPTLC1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP Amyotrophic lateral sclerosis, juvenile type: 5 genes (22,4 kb) ALS2, SETX, SIGMAR1, SPG11, SPTLC1 Amyotrophic lateral sclerosis and frontotemporal dementia (FTDALS): 14 genes (22,6 kb) C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, FUS, OPTN, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP	3 - 5 w	E
Basal ganglia calcification (IBGC) * Gene panel: ID327.00, 6 genes (11,2 kb) JAM2, MYORG, PDGFB, PDGFRB, SLC20A2, XPR1	3 - 5 w	E
Brain atrophy and demyelinating brain diseases * Gene panel: ID278.00 Brain atrophy and demyelinating brain diseases: 55 genes (96,0 kb) AIMP1, AIMP2, ASPA, B3GALNT2, B4GAT1, CNP, COL4A1, CRPPA, DAG1, DARS1, DEGS1, EPRS1, FAM126A, FARSA, FARSB, FKRP, FKTN, GFAP, GJC2, GMPPB, GRM7, HIKESHI, HSPD1, LARGE1, MAPT, MAT1A, MED17, MTHFS, PLP1, POLR1C, POLR3A, , POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSEN1, PYCR2, RARS1, RXYLT1, SLC25A12, SOX10, TBCD, TMEM106B, TMEM63A, TRAPPC12, TRAPPC4, TRAPPC6B, UBTF, UFM1, VPS11 Brain atrophy: 13 genes (24,2 kb) EXOC7, EXOC8, FARSA, FARSB, GRM7, MAPT, MED17, PSEN1, TBCD, TRAPPC4, TRAPPC6B, TRAPPC12, UBTF Demyelinating brain diseases: 27 genes (43,0 kb) AIMP1, AIMP2, ASPA, CNP, DARS1, DEGS1, EPRS1, FAM126A, GFAP, GJC2, HIKESHI, HSPD1, MAT1A, MTHFS, PLP1, POLR1C, POLR3A, POLR3B, PYCR2, RARS1, SLC25A12, SOX10, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Walker-Warburg syndrome (MDDGA): 14 genes (23,6 kb) B3GALNT2, B4GAT1, CRPPA, DAG1, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 w	E
CADASIL and CARASIL * Gene panel: ID167.01, 3 genes (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 w	E
Choreatiform dyskinesia * Gene panel: ID272.01, 17 genes (40,6 kb) ADCY5, ATP1A2, ATP1A3, FRRS1L, GNAO1, HPRT1, KCNMA1, NKX2-1, PDE2A, PDE10A, PNKD, PRNP, PRRT2, RNF216, SLC2A1, VPS13A, XK	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Charcot-Marie-Tooth disease, axonal type (CMT2, HMSN) * Gene panel: ID052.03 Charcot-Marie-Tooth disease, axonal type (CMT2, HMSN): 45 genes (111,8 kb) AARS1, AIFM1, ATP1A1, CADM3, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, JAG1, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PNKP, PRPS1, RAB7A, SLC12A6, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth disease, axonal, autosomal dominant (CMT2): 26 genes (74,2 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, JAG1, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, SLC12A6, TRPV4, VCP Charcot-Marie-Tooth disease, axonal, autosomal recessive (CMT2): 10 genes (24,4 kb) GDAP1, IGHMBP2, LMNA, LRSAM1, MFN2, MME, MPV17, PNKP, SPG11, TRIM2 Charcot-Marie-Tooth disease, intermediate (CMTDI, CMTRI): 11 genes (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth disease, X-linked (CMTX): 4 genes (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Charcot-Marie-Tooth disease with optic atrophy (CMT6): 3 genes (4,5 kb) MFN2, PDXK, SLC25A46	4 - 6 w	E
Charcot-Marie-Tooth disease, demyelinating type (CMT1, CMT4, HMSN) * Gene panel: ID051.03 Charcot-Marie-Tooth disease, demyelinating type (CMT1, CMT4, HMSN): 29 genes (69,5 kb) CNTNAP1, COX6A1, DNM2, EGR2, FBLN5, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, ITPR3, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1 Charcot-Marie-Tooth disease, demyelinating, autosomal dominant (CMT1): 11 genes (19,0 kb) EGR2, FBLN5, GDAP1, ITPR3, LITAF, MPZ, NEFL, POLR3B, PMP2, PMP22 Charcot-Marie-Tooth disease, demyelinating, autosomal recessive (CMT4): 12 genes (33,8 kb) EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth disease, intermediate (CMTDI, CMTRI): 11 genes (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Neuropathy of Déjerine-Sottas (CMT3, DSS): 4 genes (7,1 kb) EGR2, MPZ, PMP22, PRX Congenital hypomyelinating neuropathy (CHN): 3 genes (6,3 kb) EGR2, MPZ, CNTNAP1	3 - 5 w	E
Charcot-Marie-Tooth disease (CMT, HMSN), comprehensive diagnostics * Gene panel: ID312.02 Charcot-Marie-Tooth disease (CMT, HMSN), comprehensive diagnostics: 87 genes (217,3 kb) AARS1, AIFM1, ARHGEF10, ATP1A1, ATP7A, BSCL2, CADM3, CNTNAP1, COQ7, COX6A1, DCTN1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A7, SORD, SPG11, SPTAN1, SPTLC1, SURF1, SYT2, TRIM2, TRPV4, VCP, VRK1, VWA1, WARS1, YARS1 Charcot-Marie-Tooth disease, demyelating (CMT1, CMT4, HMSN1): 20 genes (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth disease, axonal (CMT2): 35 genes (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth disease, intermediate (CMTDI, CMTRI): 11 genes (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth disease, X-linked (CMTX): 4 genes (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophic neuropathy of Déjerine-Sottas (CMT3, DSS): 4 genes (7,1 kb) EGR2, MPZ, PMP22, PRX Peripheral neuropathy with optic atrophy (HMSN6): 3 genes (4,5 kb) MFN2, PDXK, SLC25A46 Congenital hypomyelating neuropathy (CHN): 3 genes (6,3 kb) EGR2, MPZ, CNTNAP1 Distal hereditary motor neuropathy (HMND, HMNR): 24 genes (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	4 - 6 w	E
Distal hereditary motor neuronopathy (HMND, HMNR) * Gene panel: ID254.01 Distal hereditary motor neuronopathy (HMND, HMNR): 24 genes (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Distal hereditary motor neuronopathy, autosomal dominant type (HMND): 15 genes (38,4 kb) BSCL2, DCTN1, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SPTAN1, SYT2, TRPV4, WARS1 Distal hereditary motor neuronopathy, autosomal recessive type (HMNR): 10 genes (14,4 kb) ATP7A, DNAJB2, COQ7, IGHMBP2, PLEKHG5, REEP1, SIGMAR1, SORD, VRK1, VWA1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Dystonia (DYT) * Gene panel: ID128.04 Dystonia (DYT): 25 genes (55,9 kb) ACTB, ADCY5, ANO3, AOPEP, ATP1A3, COL6A3, ECHS1, GCH1, GNAL, HPCA, KCTD17, KMT2B, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A, VPS16 Dystonia (DYT), isolated type: 15 genes (39,4 kb) ADCY5, ANO3, COL6A3, ECHS1, GNAL, HPCA, KCNN2, KCTD17, KMT2B, SHQ1, SLC2A1, THAP1, TOR1A, TUBB4A, VPS11 Dystonia (DYT), combined type: 29 genes (47,2 kb) ACTB, ADCY5, AOPEP, ATP1A3, ATP5MC3, BCAP31, ECHS1, EIF2AK2, GCH1, MECR, NUP54, PLA2G6, PNKD, PRKRA, PRRT2, SGCE, SHQ1, SLC2A1, SLC6A3, SLC18A2, SLC30A10, SLC39A14, SPR, TAF1, TH, TIMM8A, TMEM151A, VPS16, WARS2	3 - 5 w	E
Episodic ataxia (EA) * Gene panel: ID184.02, 6 genes (20,6 kb) ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3	3 - 5 w	E
Episodic pain syndrome (FEPS) * Gene panel: ID268.00, 4 genes (20,5 kb) SCN9A, SCN10A, SCN11A, TRPA1	3 - 5 w	E
Essential tremor (ETM) * Gene panel: ID195.01, 4 genes (16,6 kb) DRD3, FUS, SCN4A, TENM4	3 - 5 w	E
Frontotemporal dementia (FTD) * Gene panel: ID310.01 Frontotemporal dementia (FTD): 19 genes (29,3 kb) C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Frontotemporal dementia and Amyotrophic Lateralsklerosis (FTDALS): 17 genes (27,1 kb) C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Frontotemporal dementia, inclusion body myopathy and Paget disease (IBMPFD): 3 genes (4,6 kb) HNRNPA1, HNRNPA2B1, VCP	3 - 5 w	E
Hereditary ataxia, comprehensive diagnostics * Gene panel: ID276.02 Hereditary ataxia, comprehensive diagnostics: 128 genes (373,1 kb) ABCB7, ABHD12, ACO2, AFG3L2, ANGPTL1, AIFM1, ANO10, APOB, APTX, ATCAY, ATG5, ATG7, ATM, ATP1A3, ATP2B3, ATP8A2, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CASK, CCDC88C, CHP1, CLCN2, CLN5, COA7, COQ2, COQ4, COQ8A, CWF19L1, CYP27A1, DAB1, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF12, FGF14, FLVCR1, GDAP2, GOSR2, GRID2, GRM1, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, LAMA1, MARS2, MME, MRE11, MTCL1, MTPAP, MTPP, NBN, NKX6-2, NPTX1, OPHN1, PCDH12, PCNA, PDYN, PEX7, PHYH, PIK3R5, PITRM1, PLD3, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRDX3, PRKCG, PRPS1, PTF1A, PUM1, RFC1, RNF216, RNU12, RUBCN, SACS, SAMD9L, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC2A1, SLC25A46, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG7, SPTBN2, SQSTM1, STUB1, SYNE1, SYT14, TDP1, TDP2, TGM6, THG1L, TMEM240, TPP1, TRPC3, TSFM, TTBK2, TTPA, TUBB4A, TWNK, TXN2, UBA5, VAMP1, VLDLR, VPS13D, VPS41, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1 Episodic ataxia (EA): 6 genes (20,5 kb) ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3 Spastic ataxia (SPAX): 7 genes (25,6 kb) AFG3L2, CHP1, COQ4, KIF1C, MARS2, MTPAP, NKX6-2, SACS, VAMP1 Spinocerebellar ataxia, autosomal dominant (SCA): 27 genes (91,6 kb) AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, NPTX1, PDYN, PLD3, PRKCG, PUM1, SAMD9L, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2 Spinocerebellar ataxia, autosomal recessive (SCAR): 32 genes (106,7 kb) ANO10, ATG5, ATG7, COA7, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PITRM1, PMPCA, PRDX3, RNU12, RUBCN, SCYL1, SETX, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, UBA5, VPS13D, VPS41, VWA3B, WWOX, XRCC1 Cerebellar ataxia with mental retardation (CAMRQ): 4 genes (12,9 kb) ATP8A2, CA8, VLDLR, WDR81 Cerebellar ataxia, X-linked: 7 genes (15,9 kb) ABCB7, AIFM1, ATP2B3, CASK, OPHN1, PRPS1, SLC9A6 Ataxia-oculomotor apraxia (AOA): 4 genes (13,3 kb) APTX, PIK3R5, PNKP, SETX Ataxia teleangiectatica (AT): 5 genes (15,4 kb) APTX, ATM, MRE11, NBN, PCNA	4 - 6 w	E
Hereditary sensory and autonomic neuropathy (HSAN, HSN) * Gene panel: ID086.02, 16 genes (52,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	3 - 5 w	E
Hyperekplexia (HKPX) * Gene panel: ID216.00, 9 genes (16,0 kb) ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1	3 - 5 w	E
Hypomyelinating leukodystrophy (HLD) * Gene panel: ID277.00, 22 genes (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Hereditary neuropathy (HMSN, HMN, HSAN), comprehensive diagnostics* Gene panel: ID374.00 Hereditary neuropathy (HMSN, HMN, HSAN), comprehensive diagnostics: 134 genes (343,6 kb) AARS1, ABCA1, ACOX1, AIFM1, AR, ARHGEF10, ATL1, ATL3, ATP1A1, ATP7A, BICD2, BSCL2, CADM3, CFAP276, CHCHD10, CNTNAP1, COQ7, COX20, COX6A1, CPOX, CYP27A1, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DRP2, DST, DYNC1H1, EGR2, ELP1, EMILIN1, FBLN5, FBXO38, FGD4, FIG4, GAN, GARS1, GBF1, GDAP1, GJB1, GNB4, GSN, HARS1, HEXA, HEXB, HINT1, HK1, HMBS, HSPB1, HSPB3, HSPB8, IGHMBP2, INF2, ITPR3, JAG1, JPH1, KARS1, KIF1A, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MAG, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYH14, NAGLU, NDRG1, NEFH, NEFL, NGF, NTRK1, OPA1, PDK3, PDXK, PIGB, PLEKHG5, PMP2, PMP22, PNKP, POLR3B, PPOX, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SBF1, SBF2, SCN9A, SCN10A, SCN11A, SCO2, SEPTIN9, SETX, SH3TC2, SIGMAR1, SLC12A6, SLC25A46, SLC5A6, SLC5A7, SLC52A2, SLC52A3, SMN1, SORD, SPG11, SPTAN1, SPTLC1, SPTLC2, SURF1, SYT2, TECPR2, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, VCP, VRK1, VWA1, WARS1, WNK1, YARS1 Charcot-Marie-Tooth disease, demyelinating (CMT1, CMT4, HMSN1): 20 genes (50,3 kb) EGR2, FGD4, FBLN5, FIG4, GDAP1, HK1, ITPR3, LITAF, MPZ, MTMR2, NDRG1, NEFL, PMP2, PMP22, POLR3B, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth disease, axonal (CMT2, HMSN2): 35 genes (97,3 kb) AARS1, ATP1A1, CADM3, DHTKD1, DNM2, DYNC1H1, GARS1, GBF1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, JAG1, JPH1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PNKP, RAB7A, SLC12A6, SPG11, TRIM2, TRPV4, VCP Charcot-Marie-Tooth disease, intermediate (CMTDI, CMTRI): 11 genes (18,5 kb) COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1 Charcot-Marie-Tooth disease, X-linked (CMTX, HMSN): 4 genes (4,9 kb) AIFM1, GJB1, PDK3, PRPS1 Hypertrophic Déjerine-Sottas neuropathy (DSS, HMSN3): 4 genes (7,1 kb) EGR2, MPZ, PMP22, PRX Congenital hypomyelating neuropathy (CHN): 3 genes (6,3 kb) EGR2, MPZ, CNTNAP1 Distal hereditary motor neuropathy (HMND, HMNR): 24 genes (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1 Hereditary sensorische und autonomic neuropathy (HSAN, HSN): 16 genes (57,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, TECPR2, WNK1	4 - 6 w	E
Leukodystrophy and Leukoencephalopathy, comprehensive diagnostics * Gene panel: ID204.04 Leukodystrophy and Leukoencephalopathy, comprehensive diagnostics : 122 genes (208,5 kb) AARS2, ABCD1, ACBD5, ACER3, ACOX1, ADAR, AIFM1, AIMP1, AIMP2, ALDH3A2, ARSA, ASPA, AUH, BOLA3, CLCN2, CLDN11, CNP, COA7, COA8, COL4A1, COL4A2, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS1, DARS2, DEGS1, EARS2, EIF2AK1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, FA2H, FAM126A, FDX2, FOLR1, FOXRED1, FUCA1, GALC, GBE1, GCDH, GFAP, GFM1, GJC2, GLB1, HEPACAM, HIKESHI, HMGCL, HSD17B4, HSPD1, HTRA1, IBA57, IFIH1, ISCA1, ISCA2, KARS1, KCNT1, L2HGDH, LMNB1, LYRM7, MARS2, MCOLN1, MLC1, MTFMT, NAXD, NAXE, NFU1, NKX6-2, NOTCH3, NUBPL, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PLAA, PLEKHG2, PLP1, PMPCB, POLR1C, POLR3A, POLR3B, POLR3K, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SDHAF1, SLC13A3, SLC16A2, SLC17A5, SOX10, SPTAN1, STN1, SUMF1, TMEM106B, TMEM63A, TREM2, TREX1, TUBB4A, TYMP, TYROBP, UFM1, VPS11, ZFYVE26 Leukodystrophy with hypomyelination (HLD): 22 genes (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Leukodystrophy with peroxisome biogenesis disorder (PBD): 15 genes (20,8 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH Orthochromatic leukodystrophy: 10 genes (14,6 kb) ASPA, CSF1R, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GFAP, HEPACAM, MLC1 Metachromatic leukodystrophy: 3 genes (4,2 kb) ARSA, PSAP, SUMF1 Aicardi-Goutières syndrome (AGS): 7 genes (11,9 kb) ADAR, IFIH1, RNASE2A, RNASE2B, RNASE2C, SAMHD1, TREX1 CADASIL, CARASIL: 2 genes (8,4 kb) HTRA1, NOTCH3	4 - 6 w	E
Neurodegeneration with brain iron accumulation (NBIA) * Gene panel: ID264.00, 11 genes (20,0 kb) ATP13A2, C19ORF12, CP, COASY, CRAT, FA2H, FTL, PANK2, PLA2G6, REPS1, WDR45	3 - 5 w	E
Neurodevelopmental disorders (NED), comprehensive diagnostics * Gene panel: ID358.00, 169 genes (431,8 kb) ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, EXOC7, EXOC7, EXOC8, FBXW11, FDF1, FRA10AC1, FRMD5, GABBR2, GEMIN4, GEMIN5, GNAI1, GNAO1, GNB2, GPT2, GRIA2, GRIA4, GRIK2, GRIN1, GRM7, H3-3A, H3-3B, H4C11, H4C3, H4C5, H4C9, HECTD4, HECW2, HNRNPH1, HNRNPR, HPDL, HS2ST1, INTS1, INTS8, IRF2BPL, KAT5, KCNN2, KDM6B, LNPB, MADD, MAPK8IP3, MED27, MEF2C, MFSD2A, MTHFS, MTOR, NAE1, NARS1, NBEA, NCDN, NFASC, NOVA2, NRCAM, NSRP1, NTNG2, ODC1, OGDHL, OTUD5, PCDHGC4, PGAP1, PGM2L1, PI4KA, PIGA, PIGG, PIGK, PIGU, PLAA, PLXNA1, POLR2A, PPFIBP1, PPP1R21, PPP2CA, PRKAR1B, PRUNE1, PSMB1, PSMC1, PTPN23, PURA, PUS3, RAB11B, RAC3, RALA, RALGAP1, RBL2, RERE, SARS1, SEC31A, SETD1A, SHMT2, SHQ1, SMG8, SMG9, SMPD4, SNIP1, SPOP, SPTBN4, STAG2, SUPT16H, SVBP, SYT1, TAF2, TAF8, TBC1D2B, TCEAL1, THUMPD1, TIAM1, TMEM147, TMEM222, TMX2, TNF, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Parkinson disease (PARK) * Gene panel: ID077.02 Parkinson disease (PARK): 37 genes (93,8 kb) ADH1C, ATP13A2, ATP1A3, ATP6AP2, CHCHD2, DCTN1, DNAJC6, EIF4G1, FBXO7, GBA1, GCH1, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, PARK7, PINK1, PLA2G6, POLG, PRKRA, PRKN, PSAP, RAB39B, SLC18A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SYNJ1, TAF1, TH, TRPM7, UCHL1, UQCRC1, VPS13C, VPS35 Parkinson disease (PARK), early-onset: 9 genes (30,3 kb) ATP13A2, DNAJC6, FBXO7, PARK7, PINK1, PLA2G6, PRKN, SYNJ1, VPS13C Parkinson disease (PARK), late-onset: 12 genes (27,3 kb) ADH1C, CHCHD2, EIF4G1, GBA1, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, SNCA, UCHL1, VPS35 Dystonia-parkinsonism (DYT): 9 genes (18,3 kb) ATP1A3, GCH1, PRKRA, SLC6A3, SLC18A2, SLC30A10, SLC39A14, TAF1, TH	3 - 5 w	E
Paroxysmal dyskinesia * Gene panel: ID286.00, 14 genes (38,9 kb) ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24	3 - 5 w	E
Peroxisome biogenesis disorder (PBD) * Gene panel: ID083.01 Peroxisome biogenesis disorder (PBD): 14 genes (19,9 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Zellweger syndrome (PBD, type A): 12 genes (18,3 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Neonatal adrenoleukodystrophy/ Infantile refsum syndrome (PBD, type B): 11 genes (17,0 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX11B, PEX13, PEX16, PEX26 Heimler syndrome (PBD, type C): 2 genes (6,8 kb) PEX1, PEX6	3 - 5 w	E
Sleep disorders * Gene panel: ID371.00, 13 genes (25,3 kb) ADRB1, BHLHE41, CRY1, CRY2, CSNK1D, GRM1, HCRT, MOG, NPSR1, PER2, PER3, PRNP, TIMELESS	3 - 5 w	E
Spastic ataxia (SPAX) * Gene panel: ID228.00, 12 genes (34,8 kb) AFG3L2, CAPN1, CHP1, GJC2, KIF1C, MARS2, MTPAP, NKX6-2, POLR3A, SACS, SPG7, VAMP1	3 - 5 w	E
Spastic paraplegia (HSP, SPG) * Gene panel: ID148.04 Spastic paraplegia (HSP, SPG): 67 genes (138,1 kb) ABCD1, ABHD16A, ALDH18A1, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, KPNA3, L1CAM, MAG, MTRFR, NFU1, NIPA1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP1, REEP2, RNF170, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, SPTAN1, SPTSSA, TFG, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27 Spastic paraplegia (SPG), autosomal dominant: 20 genes (43,1 kb) ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, KPNA3, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, SPTAN1, SPTSSA, UBAP1, WASHC5, ZFYVE27 Spastic paraplegia (SPG), autosomal recessive: 50 genes (101,2 kb) ABHD16A, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, FICD, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NFU1, NT5C2, PCYT2, PI4KA, PLP1, PNPLA6, REEP2, RNF170, SELENOI, SPART, SPG7, SPG11, SPG21, SPTSSA, TFG, UCHL1, VPS37A, ZFYVE26 Spastic paraplegia (SPG), X-linked: 3 genes (6,9 kb) ABCD1, L1CAM, PLP1	4 - 6 w	E
Spinal muscular atrophy (SMA) * Gene panel: ID152.02 Spinal muscular atrophy (SMA): 39 genes (87,9 kb) AR, ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, COQ7, DCTN1, DNAJB2, DYNC1H1, EMILIN1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SLC25A46, SMN1, SMN2, SORD, SPTAN1, SYT2, TRIP4, TRPV4, UBA1, VAPB, VRK1, VWA1, WARS1 Proximal spinal muscular atrophy (SMA): 13 genes (34,2 kb) AR, ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, GARS1, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB Distal spinal muscular atrophy (DSMA, HMN): 24 genes (54,7 kb) ATP7A, BSCL2, COQ7, DCTN1, DNAJB2, EMILIN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SORD, SPTAN1, SYT2, TRPV4, VRK1, VWA1, WARS1	3 - 5 w	E
Spinocerebellar ataxia, autosomal dominant (SCA, ADCA) * Gene panel: ID236.03, 27 genes (91,6 kb) AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, NPXT1, PDYN, PLD3, PRKCG, PUM1, SAMD9L, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2	3 - 5 w	E
Spinocerebellar ataxia, autosomal recessive (SCAR, SCAN) * Gene panel: ID213.04, 36 genes (117,6 kb) ANO10, ATG5, ATG7, COA7, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PITRM1, PMPCA, PNPLA6, POLG, PRDX3, RNU12, RUBCN, SCYL1, SETX, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, TWNK, UBA5, VPS13D, VPS41, VWA3B, WDR73, WWOX, XRCC1	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Pulmonary and Respiratory Diseases		
Congenital central hypoventilation syndrome (CCHS) * Gene panel: ID185.01, 5 genes (9,7 kb) LBX1, MECP2, MYO1H, PHOX2B, RET	3 - 5 w	E
Interstitial lung disease and pulmonary fibrosis * Gene panel: ID341.01 Interstitial lung disease und pulmonary fibrosis: 40 genes (80,9 kb) ABCA3, ACD, AP3B1, AP3D1, COPA, CSF2RA, CSF2RB, DCLRE1B, DKC1, FAM111B, FARSA, FARSB, FLNA, FOXF1, GBA1, HPS1, HPS4, ITGA3, MARS1, NAF1, NHP2, NKX2-1, NOP10, OAS1, PARN, RPA1, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, STING1, TERC, TERT, TINF2, WRAP53, ZCCHC8 Pulmonary alveolar proteinosis (SMDP, ILD): 10 genes (17,7 kb) ABCA3, CSF2RA, CSF2RB, MARS1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC7A7, OAS1 Telomere-related pulmonary fibrosis (PFBMFT, DKC): 14 genes (22,7 kb) ACD, DCLRE1B, DKC1, NAF1, NHP2, NOP10, PARN, RPA1, RTEL1, TERC, TERT, TINF2, WRAP53, ZCCHC8 Syndromes with pulmonary fibrosis: 16 genes (32,6 kb) ACD, AP3B1, AP3D1, DKC1, FAM111B, HPS1, HPS4, NAF1, NHP2, NOP10, PARN, RTEL1, SLC34A2, TERT, TINF2, WRAP53 Syndromes with diffuse parenchymal lung disease: 10 genes (25,0 kb) COPA, FARSA, FARSB, FLNA, FOXF1, GBA1, ITGA, NKX2-1, SMPD1, STING1	3 - 5 w	E
Lung cancer * Gene panel: ID260.02, 33 genes (122,5 kb) ATM, ATR, BAP1, BRCA1, BRCA2, BLM, CDH1, CDKN2A, CHEK2, DICER1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCG, FANCD2, FGFR3, FLCN, JAK2, MET, MSH6, MUTYH, NBN, NF1, NKX2-1, PALB2, PRKN, RAD50, RECQL4, SDHA, TSC2, TP53	4 - 6 w	E
Pulmonary hypertension (PAH) * Gene panel: ID281.01, 23 genes (49,9 kb) ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, G6PC3, GDF2, KCNA5, KCNK3, KDR, NFU1, NOTCH3, SARS2, SMAD1, SMAD4, SMAD9, SOX17, TBX4	3 - 5 w	E
Pulmonary surfactant metabolism dysfunction (SMDP) * Gene panel: ID168.01, 6 genes (12,0 kb) ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB	3 - 5 w	E
Primary ciliar dyskinesia with or without situs inversus (PCD, CILD) * Gene panel: ID085.03, 50 genes (164,8 kb) BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10	4 - 6 w	E
Reproductive Disorders		
Adrenogenital syndrome (AGS, CAH) * Gene panel: ID111.02, 7 genes (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 w	E
Disorder of sex development (DSD) * Gene panel: ID117.03 Disorder of sex development (DSD): 49 genes (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, NROB1, NR2F2, NR5A1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX8, SOX9, SRD5A2, SRY, STAR, TOE1, TSPYL1, WNT4, WT1, ZFPM2 46,XY Disorder of sex development (SRXY), nonsyndromic: 21 genes (37,5 kb) AKR1C2, AR, CBX2, CYP11A1, DHH, DHX37, DMRT1, GATA4, HHAT, HSD17B3, LHCGR, MAMLD1, MAP3K1, NROB1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2 46,XX Disorder of sex development (SRXX), nonsyndromic: 6 genes (6,6 kb) NR2F2, NR5A1, RSP01, SOX9, SRY, WNT4 Disorder of sex development (DSD), syndromic: 37 genes (73,1 kb) AMH, AMHR2, ANOS1, AR, ARX, ATRX, CDKN1C, CHD7, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DHCR7, DMRT1, CTU2, GATA4, HHAT, HOXA13, HSD3B2, HSD17B3, LHCGR, MYRF, NROB1, POR, PPP1R12A, PPP2R3C, RSP01, SAMD9, SGPL1, SOX9, SRD5A2, STAR, TOE1, TSPYL1, WNT4, WT1 Adrenogenital syndrome (AGS, CAH): 7 genes (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	4 - 6 w	E
Hypogonadotropic hypogonadism with or without anosmia (KAL, HH) * Gene panel: ID170.04, 37 genes (62,5 kb) ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 w	E
Ovarian dysgenesis (ODG) * Gene panel: ID293.01 Ovarian dysgenesis (ODG): 16 Gene (27,8 kb) BMP15, CLPP, ERAL1, ESR2, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 XX gonadal dysgenesis (ODG): 10 Gene (17,1 kb) BMP15, ESR2, FSHR, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX gonadal dysgenesis with deafness (PRLTS): 6 genes (10,7 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Reproductive Disorders		
Oocyte/zygote/embryo maturation arrest (OZEMA) * Gene panel: ID239.02, 22 genes (36,7 kb) ASTL, BTG4, CDC20, CHEK1, FBXO43, KPNA7, MOS, NLRP2, NLRP5, PABPC1L, PADI6, PANX1, PATL2, REC114, TLE6, TRIP13, TUBB8, WEE2, ZFP36L2, ZP1, ZP2, ZP3	3 - 5 w	E
Male infertility (SPGF, CBAVD) * Gene panel: ID192.05 Male infertility (SPGF, CBAVD): 108 genes (368,8 kb) ACR, ACTL7A, ACTL9, ADGRG2, AK7, AK9, AKAP3, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP251, CFAP43, CFAP44, CFAP47, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFTR, CT55, CYLC1, DMRT1, DNAH1, DNAH10, DNAH17, DNAH2, DNAH8, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, HFM1, IFT74, IQCN, KASH5, KCNU1, KCTD19, KLHL10, LRRC23, M1AP, MEI1, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PLCZ1, PMFBP1, PNLD1, PPP2R3C, QRIC2, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPEF2, SPINK2, SSX1, STAG3, STK33, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEK3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Spermatogenic failure (SPGF): 102 genes (349,2 kb) ACR, ACTL7A, ACTL9, AK7, AK9, AKAP3, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP251, CFAP43, CFAP44, CFAP47, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CT55, CYLC1, DNAH1, DNAH10, DNAH17, DNAH2, DNAH8, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, IFT74, IQCN, KASH5, KCNU1, KLHL10, LRRC23, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PLCZ1, PMFBP1, PNLD1, PPP2R3C, QRIC2, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPEF2, SPINK2, SSX1, STAG3, STK33, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEK3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Congenital bilateral aplasia of vas deferens (CBAVD): 2 genes (7,5 kb) ADGRG2, CFTR	4 - 6 w	E
Perrault syndrome (PRLTS) * Gene panel: ID388.00, 9 genes (14,1 kb) CLPP, DAP3, ERAL1, HARS2, HSD17B4, LARS2, MRPL49, PRORP, TWNK	3 - 5 w	E
Premature ovarian failure (POF) * Gene panel: ID078.05 Premature ovarian failure (POF): 23 Gene (51,1 kb) BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, INHA, LHCGR, MCM8, MSH5, NOBOX, NR5A1, POF1B, STAG3, SYCE1, XRCC2 Premature ovarian failure (POF), X-linked: 4 genes (8,2 kb) BMP15, DIAPH2, FMR1, POF1B Premature ovarian failure (POF), autosomal recessive: 10 genes (25,0 kb) C14ORF39, GDF9, FANCM, HFM1, HSF2BP, MCM8, MSH5, STAG3, SYCE1, XRCC2 Premature ovarian failure (POF), autosomal dominant: 6 genes (12,7 kb) BNC1, ERCC6, FIGLA, FOXL2, NOBOX, NR5A1	3 - 5 w	E
Primary ciliary dyskinesia with or without situs inversus (PCD, CILD) * Gene panel: ID085.03, 50 genes (164,8 kb) BRWD1, CCDC103, CCDC39, CCDC40, CCDC65, CCNO, CFAP74, CFAP298, CFAP300, CLXN, DAW1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAAF11, DNAH1, DNAH5, DNAH7, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC56, MCIDAS, NEK10, NME5, NME8, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TP73, TTC12, ZMYND10	4 - 6 w	E
Skeletal and Bone Diseases		
Acrocephalosyndactyly (ACS) * Gene panel: ID311.00, 6 genes (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1	3 - 5 w	E
Adams-Oliver syndrome (AOS) * Gene panel: ID259.00, 6 genes (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ	3 - 5 w	E
Arthrogryposis * Gene panel: ID200.01 Arthrogryposis: 61 genes (209,7 kb) ACTA1, ADCY6, ADGRG6, ASCC1, BICD2, CHRNA1, CHRND, CHRNG, CHST14, CNTN1, CNTNAP1, DHCR24, DNMT2, DOK7, DSE, ECEL1, ERBB3, ERCC1, ERCC2, ERCC5, ERCC6, ERGIC1, FBN2, FKBP10, FLVCR2, GBE1, GLDN, GLE1, KLHL40, KLHL41, LGI4, LMOD3, NALCN, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, MYLPF, NEB, NEK9, NUP88, PIEZO2, PIP5K1C, PLOD2, RAPSN, RYR1, SCYL2, SYNE1, TNNI2, TNNT3, TOR1A, TPM2, TPM3, TRIP4, TRPV4, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2 Arthrogryposis multiplex congenita (AMC): 6 genes (58,2 kb) ERGIC1, TOR1A, LGI4, NEB, SCYL2, SYNE1 Distal arthrogryposis (DA): 11 genes (40,3 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1 Lethal congenital contracture syndrome (LCCS): 11 genes (31,5 kb) ADCY6, ADGRG6, CNTNAP1, DNMT2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42 Fetal akinesia deformation sequence (FADS): 4 genes (7,6 kb) DOK7, MUSK, NUP88, RAPSN	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Arachnodactyly * Gene panel: ID124.00, 13 genes (35,9 kb) CHST14, CTSC, DSE, EFEMP2, FBN1, FBN2, SCARF2, SKI, TGFB2, TGFB3, TGFB1, TGFB2, SMAD3	3 - 5 w	E
Brachydactyly (BD) * Gene panel: ID218.02 Brachydactyly (BD): 21 genes (60,1 kb) ADAMTS10, ADAMTS17, BMP2, BMPR1B, CHST11, CHSY1, FBN1, GDF5, HOXD13, HUWE1, IHH, LTBP2, NOG, PDE3A, PITX1, PRMT7, PTHLH, ROR2, RUNX2, TBC1D24, TRPV4 Brachydactyly (BD), nonsyndromic: 8 genes (10,5 kb) BMP2, BMPR1B, GDF5, HOXD13, IHH, NOG, PTHLH, ROR2 Brachydactyly (BD), syndromic: 16 genes (52,5 kb) ADAMTS10, ADAMTS17, CHST11, CHSY1, FBN1, GDF5, HOXD13, HUWE1, LTBP2, NOG, PDE3A, PITX1, PRMT7, RUNX2, TBC1D24, TRPV4	3 - 5 w	E
Craniosynostosis (CRS) * Gene panel: ID224.02 Craniosynostosis (CRS): 36 genes (84,9 kb) ALPL, ALX4, ASXL1, CD96, CDC45, COLEC10, COLEC11, CYP26B1, EFN1, ERF, ESCO2, FGFR1, FGFR2, FGFR3, FREM1, GLI3, IFT43, IFT122, IL11RA, MASP1, MEGF8, MSX2, P4HB, POR, PPP3CA, RAB23, RECQL4, SCARF2, SEC24D, SKI, SMAD6, TCF12, TWIST1, WDR19, WDR35, ZIC1 Craniosynostosis (CRS), nonsyndromic: 8 genes (10,5 kb) ALX4, ERF*, IL11RA, MSX2, SMAD6, TCF12, TWIST1, ZIC1 Acrocephalosyndactyly (ACS): 6 genes (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1 Cranioectodermal dysplasia (CED): 4 genes (12,1 kb) IFT43, IFT122, WDR19, WDR35 Trigonocephaly (TRIGNO): 5 genes (17,0 kb) ASXL1, CD96, FGFR1, FREM1, PPP3CA	3 - 5 w	E
Distal arthrogryposis (DA) * Gene panel: ID196.01, 11 genes (40,5 kb) ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNT2, TNNT3, TPM2, UBA1	3 - 5 w	E
Fraser syndrome (FRASRS) * Gene panel: ID317.00, 3 genes (24,8 kb) FRAS1, FREM2, GRIP1	3 - 5 w	E
Frontonasal dysplasia (FND) * Gene panel: ID339.00, 11 genes (22,1 kb) ALX1, ALX3, ALX4, ANKH, EFN1, FGFR1, FGFR2, FGFR3, GLI3, TWIST1, ZSWIM6	3 - 5 w	E
Hand malformations, comprehensive diagnostics * Gene panel: ID298.00 Hand malformation, comprehensive diagnostics: 110 genes (295,9 kb) ADAMTS10, ADAMTS17, AKT3, BHLHA9, BMP2, BMPR1B, C2CD3, CACNA1C, CCND2, CCNQ, CDH3, CHST11, CHSY1, CIBAR1, CKAP2L, CPLANE1, CREBBP, DACT1, DDX59, DHCR7, DHODH, DLL4, DLX5, DOCK6, EFN1, EFTUD2, EOGT, EP300, ESCO2, EVC2, FBLN1, FBN1, FGF10, FGF16, FGF9, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GATA6, GDF5, GDF6, GJA1, GLI1, GLI2, GLI3, GRIP1, HOXA13, HOXD13, HUWE1, IFT57, IGF2, IHH, INTU, IQCE, IRF6, KIAA0753, KIAA0825, KIF7, LMBR1, LMNA, LRP4, LTBP2, MAP3K20, MECOM, MEGF8, MYCN, NAA10, NECTIN1, NECTIN4, NOG, NOTCH1, OFD1, PAX3, PDE3A, PDE4D, PIK3CA, PIK3R2, PITX1, PRKAR1A, PRMT7, PTHLH, RAB23, RBM8A, RBPJ, RECQL4, RIPK4, ROR2, RUNX2, SALL1, SALL4, SF3B4, SMO, SMOC1, SOST, TBC1D24, TBX15, TBX3, TBX5, TCTN3, TMEM107, TP63, TRPV4, TWIST1, WPCP, WNT10B, WNT7A, YY1AP1, ZNF141 Brachydactyly (BD), nonsyndromic: 9 genes (14,0 kb) BMP2, BMPR1B, GDF5, HOXD13, IHH, NOG, PDE3A, PTHLH, ROR2 Polydactyly (PAPA, PPD), nonsyndromic: 9 genes (20,9 kb) CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141 Syndactyly (SDTY), nonsyndromic: 8 genes (18,5 kb) BHLHA9, FBLN1, GJA1, GLI3, HOXD13, LMBR1, LRP4, NECTIN4 Ectrodactyly (SHFM): 7 genes (10,6 kb) CDH3, DLX5, FGFR1, IGF2, TP63, WNT7A, WNT10B Acrocephalosyndactyly (ACS): 6 genes (17,0 kb) FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1 Orofaciodigital syndrome (OFD): 9 genes (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107 Lacrimalauriculodentodigital syndrome (LADD): 3 genes (5,5 kb) FGF10, FGFR2, FGFR3 Multiple synostoses syndrome (SYNS): 6 genes (8,3 kb) FGF9, GDF5, GDF6, HOXA11, MECOM, NOG Acrodysostosis syndrome (ACRDYS): 3 genes (4,9 kb) PDE4D, PRKAR1A, SF3B4	4 - 6 w	E
Heart-hand syndrome * Gene panel: ID165.01, 9 genes (23,8 kb) DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Hypophosphatasia, hypophosphatemia and rickets * Gene panel: ID269.03 Hypophosphatasia, hypophosphatemia and rickets: 16 genes (27,8 kb) ALPL, CLCN5, CYP2R1, CYP27B1, CYP3A4, DMP1, ENPP1, FAH, FAM20C, FGF23, KL, NHERF1, PHEX, SLC34A1, SLC34A3, VDR Hypophosphatemic rickets (HR): 9 genes (16,1 kb) CLCN5, DMP1, ENPP1, FAM20C, FGF23, NHERF1, PHEX, SLC34A1, SLC34A3 Vitamin D-dependent hypophosphatemic rickets (VDDR): 4 genes (5,8 kb) CYP2R1, CYP3A4, CYP27B1, VDR Hypophosphatasia (HPP): 1 gene (1,6 kb) ALPL	3 - 5 w	E
Klippel-Feil syndrome (KFS) * Gene panel: ID207.00, 5 genes (12,5 kb) GDF3, GDF6, MEOX1, MYO18B, PAX1	3 - 5 w	E
Lethal congenital contracture syndrome (LCCS) * Gene panel: ID197.00, 12 genes (34,6 kb) ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42	3 - 5 w	E
Mandibulofacial dysostosis (MFD) * Gene panel: ID188.01 Mandibulofacial dysostosis (MFD): 11 genes (22,0 kb) DHODH, EDNRA, EFTUD2, POLR1A, POLR1B, POLR1C, POLR1D, RPS28, SF3B4, TCOF1, TSR2 Treacher-Collins syndrome (TCS): 4 genes (9,4 kb) POLR1B, POLR1C, POLR1D, TCOF1	3 - 5 w	E
Multiple epiphyseal dysplasia (EDM) * Gene panel: ID202.01, 10 genes (25,9 kb) CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, EIF2AK3, KIF7, MATN3, SLC26A2	3 - 5 w	E
Multiple pterygium syndrome * Gene panel: ID158.01, 8 genes (16,8 kb) CHRNA1, CHRN1, CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1	3 - 5 w	E
Orofacial cleft (OFC) * Gene panel: ID266.01 Orofacial cleft (OFC): 50 genes (151,0 kb) ARHGAP29, BMP4, BRD4, C2CD3, CDH1, CHD7, CDH11, CPLANE1, CYFIP1, DDX59, DHCR7, DLX4, ESCO2, FGFR1, FGFR2, FLNB, GRHL3, HDAC8, IFT57, INTU, IRF6, KDM1A, KDM6A, KIAA0753, KMT2D, LRRC32, MEIS2, MID1, MSX1, NECTIN1, NEK1, NIPA1, NIPA2, NIPBL, OFD1, RAD21, RAB34, RIPK4, SCN11, SLC26A2, SMC1A, SMC3, SPECC1L, TBX22, TCTN3, TGDS, TMEM107, TP63, TUBGCP5, ZRSR2 Orofacial cleft (OFC), nonsyndromic: 9 genes (15,1 kb) ARHGAP29, BMP4, DLX4, GRHL3, IRF6, MSX1, NECTIN1, TBX22, TP63 Orofaciodigital syndrome (OFD): 13 genes (36,5 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, NEK1, OFD1, RAB34, SCN11, TCTN3, TMEM107, ZRSR2 Cornelia de Lange syndrome (CDLS): 6 genes (22,9 kb) BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3 Kabuki syndrome (KABUK): 3 genes (22,9 kb) KDM1A, KDM6A, KMT2D Van-der-Woude syndrome (VWS): 2 genes (3,3 kb) GRHL3, IRF6	4 - 6 w	E
Orofaciodigital syndrome (OFD) * Gene panel: ID265.00, 9 genes (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 w	E
Osteogenesis imperfecta (OI) * Gene panel: ID066.02, 21 genes (39,2 kb) ANO5, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LRP5, MBTPS2, MESD, P3H1, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TMEM38B, TENT5A, WNT1	3 - 5 w	E
Osteopetrosis (OPT) and similar bone diseases * Gene panel: ID346.01 Osteopetrosis (OPT) and similar bone diseases: 32 genes (68,1 kb) AMER1, ANKH, CA2, CLCN7, CSF1R, CTSK, DLX3, FAM20C, FERMT3, GJA1, HPGD, LEMD3, LRP4, LRP5, LRP6, LRRK1, OSTM1, PLEKHM1, PTDSS1, PTH1R, SLC4A2, SLC29A3, SLC02A1, SNX10, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP Osteopetrosis (OPTA, OPTB): 13 genes (30,2 kb) CA2, CLCN7, FERMT3, LRP5, LRP6, OSTM1, PLEKHM1, SLC4A2, SLC29A3, SNX10, TCIRG1, TNFRSF11A, TNFSF11 Syndroms with dense bone dysplasia: 19 genes (38,0 kb) AMER1, ANKH, CSF1R, CTSK, DLX3, FAM20C, GJA1, HPGD, LEMD3, LRP4, LRRK1, PTDSS1, PTH1R, SLC02A1, SOST, TBXAS1, TGFB1, TNFRSF11B, TYROBP	3 - 5 w	E
Osteoporosis * Gene panel: ID115.01, 13 genes (29,4 kb) CALCR, COL1A1, COL1A2, ESR1, LRG4, LRP5, PLS3, SGMS2, SLC34A1, SLC9A3R1, UGT2B17, VDR, WNT1	3 - 5 w	E
Polydactyly, nonsyndromic type * Gene panel: ID166.02, 9 genes (20,9 kb) CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Pierre Robin sequence * Gene panel: ID294.00, 33 genes (72,6 kb) AMER1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, EIF4E3, GNAI3, MYMK, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 w	E
Rubinstein-Taybi syndrome (RSTS) * Gene panel: ID142.01, 3 genes (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 w	E
Short-rib thoracic dysplasia with or without polydactyly (SRTD) * Gene panel: ID067.00 Short-rib thoracic dysplasia with or without polydactyly (SRTD): 20 genes (68,2 kb) CEP120, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Short-rib thoracic dysplasia (Jeune-ATD, SRPS, SRTD): 18 genes (61,3 kb) CEP120, DYNC2H1, DYNC2LI1, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60 Ellis-van Crefeld syndrome (EVC): 4 genes (11,5 kb) DYNC2L1, EVC, EVC1, WDR35	3 - 5 w	E
Skeletal dysplasia, severe type * Gene panel: ID056.01 Skeletal dysplasia, severe type: 46 genes (144,2 kb) AGPS, ALPL, BMPER, CANT1, CEP120, CILK1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, CRTAP, DLL3, DYNC2H1, EBP, FAM20C, FAM111A, FGFR2, FGFR3, FLNA, FLNB, GDF5, GNPAT, GPX4, HSPG2, IFT52, IFT80, IFT81, INPPL1, KIAA0586, LBR, LIFR, NEK1, NSDHL, P3H1, PEX5, PEX7, PPIB, PTH1R, SLC26A2, SLC35D1, SOX9, TRIP11, TRPV4, WDR34, WDR35 Achondrogenesis (ACG): 4 genes (14,1 kb) COL2A1, GDF5, SLC26A2, TRIP11 Fibrochondrogenesis (FCG): 2 genes (10,6 kb) COL11A1, COL11A2 Thanatophoric dysplasia (TD): 2 genes (6,9 kb) COL1A2, FGFR3 Chondrodysplasia, lethal: 12 genes (24,5 kb) AGPS, EBP, FLNB, GDF5, GNPAT, GPX4, LBR, PEX5, PEX7, PTH1R, SLC26A2, SLC35D1 Osteogenesis imperfecta (OI), lethal: 5 genes (12,6 kb) CRTAP, COL1A1, COL1A2, P3H1, PPIB Short-rib thoracic dysplasia (SRTD), lethal: 9 genes (35,0 kb) CEP120, DYNC2H1, IFT52, IFT80, IFT81, KIAA0586, NEK1, WDR34, WDR35	4 - 6 w	E
Skeletal dysplasia, comprehensive diagnostis * Gene panel: ID356.00, 407 genes (969,6 kb) ABCC9, ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, AFF3, AGA, AGPS, ALG12, ALG3, ALG9, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARL6, ARSB, ARSL, ASXL1, ASXL2, ATP6VOA2, ATP7A, B3GAT3, B3GLCT, B4GALT7, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, BPNT2, C2CD3, CA2, CANT1, CASR, CC2D2A, CCDC8, CCN6, CCNQ, CDC45, CDH3, CDKN1C, CDT1, CEP120, CEP290, CFAP410, CHST14, CHST3, CHSY1, CILK1, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL9A1, COL9A2, COL9A3, COLEC11, COMP, COPB2, CREB3L1, CREBBP, CRTAP, CSGALNACT1, CSPP1, CTSA, CTSC, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DHCR24, DHCR7, DHODH, DIS3L2, DLL3, DLL4, DLX3, DLX5, DMP1, DNMT3A, DOCK6, DPAGT1, DPM1, DVL1, DVL2, DVL3, DYM, DYNC2H1, DYNC2I1, DYNC2I2, DYNC2LI1, DYNLT2B, EBP, EED, EFTUD2, EIF2AK3, ENPP1, EOGT, ERF, ESCO2, EVC, EVC2, EXT1, EXT2, EXTL3, EZH2, FAM111A, FAM20C, FBN1, FBN2, FERMT3, FGF10, FGF16, FGF23, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FLNA, FLNB, FN1, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GHR, GJA1, GLB1, GLI3, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GSC, GUSB, GZF1, HDAC8, HES7, HGSNAT, HHAT, HOXD13, HPGD, HS2ST1, HSPG2, IDH1, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IHH, IL11RA, IL1RN, INPPL1, KAT6B, KDELRL2, KIAA0753, KIF22, KIF7, KMT2D, LBR, LEMD3, LIFR, LMBR1, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LRRK1, LTBP1, LTBP3, MAFB, MAN2B1, MAP3K7, MASP1, MATN3, MBTPS1, MEGF8, MEOX1, MESD, MESP2, MGP, MKKS, MKS1, MMP13, MMP2, MPDU1, MSX2, MTX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIX, NIPBL, NKX3-2, NLRP3, NOG, NOTCH1, NOTCH2, NPR2, NPR3, NSD1, NSDHL, NXN, OBSL1, OFD1, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAPSS2, PAX3, PCNT, PCYT1A, PDE3A, PDE4D, PEX5, PEX7, PGM3, PHEX, PHGDH, PIGT, PIGV, PIK3C2A, PIK3R1, PISD, PITX1, PKDCC, PLOD2, PLS3, POC1A, POLR1A, POLR1B, POLR1C, POLR1D, POP1, POR, PPIB, PRKAR1A, PRKG2, PRMT7, PSAT1, PSPH, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23, RAB33B, RASGRP2, RBM8A, RBPJ, RECQL4, RFT1, RINT1, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RPL13, RUNX2, SALL1, SALL4, SBDS, SCARF2, SCUBE3, SEC24D, SERPINF1, SERPINH1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHOX, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A1, SLC34A3, SLC35C1, SLC35D1, SLC39A13, SLCO2A1, SMAD3, SMAD4, SMAD6, SMARCA1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, STT3A, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFB2, TMC01, TMEM165, TMEM216, TMEM231, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TP63, TRAPPC2, TREM2, TRIP11, TRPS1, TRPV4, TRPV6, TTC21B, TTC8, TWIST1, TYROBP, UFSP2, UNC45A, VDR, WBP11, WDPCP, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC4, XYLT1, XYLT2, YY1, ZMPSTE24, ZNF687, ZSWIM6	4 - 6 w	E
Spondylocostal dysostosis (SCDO) * Gene panel: ID227.00, 7 genes (14,4 kb) DLL3, FLNB, HES7, LFNG, MESP2, RIPPLY2, TBX6	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Spondyloepiphyseal and spondylometaphyseal dysplasia (SED, SMD, SEMD) * Gene panel: ID110.01 Spondyloepiphyseal and spondylometaphyseal dysplasia (SED, SMD, SEMD): 39 genes (90,7 kb) ACAN, ACP5, AIFM1, B3GALT6, BGN, CFAP410, CHST3, COL2A1, COL10A1, COL11A2, COMP, DDR2, DDRGK1, EXOC6B, FN1, GPX4, KIF22, MATN3, MBTPS1, MMP13, NANS, NEPRO, NKX3-2, PAM16, PAPSS2, PCYT1A, PISD, PLCB3, POP1, RPL13, RSPRY1, SIK3, SMARCAL1, TONSL, TRAPPC2, TRIP11, TRPV4, UFSP2, WISP3 Spondyloepiphyseal dysplasia (SED): 9 genes (25,9 kb) ACAN, CHST3, COL2A1, COMP, MBTPS1, SMARCAL1, TRAPPC2, TRPV4, WISP3 Spondylometaphyseal dysplasia (SMD): 11 genes (30,2 kb) ACP5, CFAP410, COL2A1, COL10A1, FN1, GPX4, PAM16, PCYT1A, PLCB3, TRIP11, TRPV4 Spondyloepimetaphyseal dysplasia (SEMD): 21 genes (47,6 kb) ACAN, AIFM1, B3GALT6, BGN, COL2A1, DDR2, DDRGK1, EXOC6B, KIF22, MATN3, MMP13, NANS, NEPRO, PAPSS2, PISD, POP1, RPL13, RSPRY1, SIK3, TONSL, UFSP2	3 - 5 w	E
Three M syndrome (3M) * Gene panel: ID214.00, 3 genes (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 w	E
Weill-Marchesani syndrome (WMS) * Gene panel: ID230.00, 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	3 - 5 w	E
Skin and Dental Diseases		
Adams-Oliver syndrome (AOS) * Gene panel: ID259.00, 6 genes (23,2 kb) ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ	3 - 5 w	E
Albinism, comprehensive diagnostics * Gene panel: ID175.05 Albinism, comprehensive diagnostics: 33 genes (73,8 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DCT, DTNBP1, EDN3, EDNRB, EPG5, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, KITLG, LRMDA, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1 Oculocutaneous albinism (OCA, OA): 9 genes (13,2 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1 Hermansky-Pudlak syndrome (HPS): 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6 Waardenburg syndrome (WS): 8 genes (9,3 kb) EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR Griscelli syndrome (GS): 3 genes (8,0 kb) MLPH, MYO5A, RAB27A	3 - 5 w	E
Amelogenesis imperfecta (AI) * Gene panel: ID232.01, 17 genes (29,4 kb) ACP4, AMBN, AMELX, AMTN, DLX3, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, MMP20, ODAF, RELT, SLC24A4, WDR72	3 - 5 w	E
Cowden syndrome (CWS) * Gene panel: ID075.01, 8 genes (12,8 kb) AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1	3 - 5 w	E
Cutaneous malignant melanoma (CMM) * Gene panel: ID193.01, 12 genes (26,4 kb) BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3	3 - 5 w	E
Cutis laxa (ARCL, ADCL) * Gene panel: ID109.02, 11 genes (27,9 kb) ALDH18A1, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 w	E
Dyskeratosis congenita (DKC) * Gene panel: ID347.01, 15 genes (24,1 kb) ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53	3 - 5 w	E
Ectodermal dysplasia (ECTD) * Gene panel: ID136.04 Ectodermal dysplasia (ECTD): 44 genes (74,6 kb) AP1B1, CDH3, CHUK, CST6, DLX3, DSG4, EDA, EDAR, EDARADD, GJA1, GJB2, GJB6, GRHL2, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT74, KRT81, KRT83, KRT85, KRT86, LEF1, LRP6, MSX1, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, WNT10A Ectodermal dysplasia (hair/tooth/nail/glandular type), nonsyndromic: 20 Gene (27,5 kb) CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT74, KRT85, LEF1, LRP6, MSX1, NFKBIA, TRAF6, TSPEAR, WNT10A Ectodermal dysplasia (hair/tooth/nail/glandular type), syndromic: 28 Gene (51,0 kb) AP1B1, CDH3, CHUK, DLX3, DSG4, GJA1, GJB2, GJB6, GRHL2, IKBKG, KRT16, KRT17, KRT81, KRT83, KRT86, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRPS1, TWIST2	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Skin and Dental Diseases		
Ectodermal dysplasia (ECTD), comprehensive diagnostics Gene panel: ID366.00 Ectodermal dysplasia (ECTD), comprehensive diagnostics: 92 genes (206,6 kb) ANAPC1, ANTXR1, AP1B1, APCDD1, ARID1A, ARID1B ATP6V1B2, AXIN2, C3ORF52, CDH1, CDH3, CDSN, CHUK, CST6, CTNND1, CTSK, DLX3, DSG4, DSP, EDA, EDAR, EDARADD, EVC, EVC2, FGF10, FGFR2, FGFR3, GJA1, GJB2, GJB6, GRHL2, HEPHL1, HOXC13, HR, IFT122, IFT140, IFT43, IFT52, IKBKG, INSR, KCTD1, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT25, KRT74, KRT81, KRT83, KRT85, KRT86, LEF1, LIPH, LPAR6, LRP6, MBTPS2, MSX1, NECTIN1, NECTIN4, NFKB2, NFKBIA, NLRP1, PAX9, PEX1, PEX6, PKP1, PORCN, PRKD1, RIPK4, ROGDI, RSP04, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SNRPE, SREBF1, ST14, TBC1D24, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, UBR1, WDR19, WDR35, WNT10A Ectodermal dysplasie (hair/tooth/nail/glandular type), nonsyndromic: 20 genes (27,5 kb) CST6, DLX3, EDA, EDAR, EDARADD, GJB6, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT74, KRT85, LEF1, LRP6, MSX1, NFKBIA, TRAF6, TSPEAR, WNT10A Ectodermal dysplasie (hair/tooth/nail/glandular type), syndromic: 28 genes (51,0 kb) AP1B1, CDH3, CHUK, DLX3, DSG4, GJA1, GJB2, GJB6, GRHL2, IKBKG, KRT16, KRT17, KRT81, KRT83, KRT86, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRPS1, TWIST2 Complex syndromes with ectodermal dysplasia: 42 genes (120,5 kb) ANTXR1, ARID1A, ARID1B, ATP6V1B2, CDH1, CTNND1, CTSK, DSG4, DSP, EVC, EVC2, FGF10, FGFR2, FGFR3, HEPHL1, IFT122, IFT140, IFT43, IFT52, INSR, KCTD1, KRT14, KRT16, KRT17, KRT81, KRT83, KRT86, NLRP1, PEX1, PEX6, ROGDI, SETBP1, SLC25A24, SMARCA4, SMARCAD1, SMARCB1, SMARCE1, SREBF1, TBC1D24, UBR1, WDR19, WDR35	4 - 6 w	E
Epidermolysis bullosa (EB) * Gene panel: ID198.02 Epidermolysis bullosa (EB): 34 genes (120,6 kb) ATP2A2, ATP2C1, CAST, CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSG1, DSP, DST, EXPH5, FERMT1, FLG2, IKBKG, ITGA3, ITGA6, ITGB4, JUP, KLHL24, KRT1, KRT10, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, SERPINB8, SLC39A4, SPINK5, TGM5 Epidermolysis bullosa simplex (EBS): 7 genes (33,4 kb) CD151, DST, EXPH5, KLHL24, KRT14, KRT5, PLEC Epidermolysis bullosa junctionalis (JEB): 7 genes (28,4 kb) COL17A1, ITGA3, ITGA6, ITGB4, LAMA3, LAMB3, LAMC2 Epidermolysis bullosa dystrophica (DEB): 1 gene (8,8 kb) COL7A1 Kindler Epidermolysis bullosa (KNDLRS): 1 gene (2,0 kb) FERMT1 Syndromes with epidermolysis bullosa: 20 genes (63,9 kb) ATP2A2, ATP2C1, CAST, CD151, CDSN, CHST8, CSTA, DSG1, DSP, FERMT1, FLG2, IKBKG, ITGA3, JUP, PKP1, PLEC, SERPINB8, SLC39A4, SPINK5, TGM5	4 - 6 w	E
Gorlin-Goltz syndrome (BCNS) und similar syndromes * Gene panel: ID174.01, 7 genes (23,6 kb) CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 w	E
Hereditary angioedema (HAE) * Gene panel: ID345.00, 7 genes (16,4 kb) ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1	3 - 5 w	E
Hermansky-Pudlak syndrome (HPS) * Gene panel: ID289.00, 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 w	E
Hydrops fetalis * Gene panel: ID370.00, 148 genes (404,7 kb) ACAD9, AHCY, ALG1, ALG12, ALG8, ALG9, ALPK3, ANGPT2, ARSB, ASAH1, ATP1A2, BRAF, CALCRL, CBL, CCBE1, CDAN1, CEP55, CFH, CHD7, CHRNA1, CHRND, CHRNA1, COL2A1, CTSA, DHCR24, DHCR7, DMPK, DNAH9, DOK7, DYNC1H1, EBP, EHBP1L1, EP300, EPHB4, ERCC5, FAT4, FBXW11, FGFR3, FH, FLT4, FOXC2, FOXP3, GAA, GALNS, GATA1, GATB, GBA1, GBE1, GLA, GLB1, GLDN, GLE1, GLUL, GNPTAB, GUSB, HADHA, HADHB, HBA1, HBA2, HNF1B, HRAS, IDUA, KIDINS220, KLF1, KLHL40, KMT2D, KRAS, LARS2, LBR, LIPA, LRP6, LZTR1, MAP2K1, MAP2K2, MAPK1, MDFIC, MKKS, MRAS, MUSK, MVK, MYH3, MYRF, NDUFB10, NEB, NEU1, NEXN, NF1, NPC1, NPC2, NRAS, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHGDH, PIEZO1, PKLR, PMM2, POU3F3, PPP1CB, PTH1R, PTPN11, RAF1, RAPSN, RASA1, RASA2, RHD, RIT1, RPL11, RPL15, RRAS, RRAS2, RYR1, SCN4A, SF3B4, SGPL1, SHOC2, SLC17A5, SLC22A5, SLC30A5, SMPD1, SOS1, SOS2, SOX18, SPRED1, SPRED2, SPTB, STAT3, SUMF1, SUZ12, TALDO1, TAFAZZIN, THSD1, UROS, WAC, WDFY3, ZEB2, ZNF148	3 - 5 w	E
Hypotrichosis, nonsyndromic type (HYPT) * Gene panel: ID146.01, 12 genes (20,1 kb) APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE	3 - 5 w	E
Ichthyosis * Gene panel: ID217.01 Ichthyosis: 38 genes (73,0 kb) ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1B1, AP1S1, ASPRV1, CASP14, CERS3, CLDN1, CLDN10, CSTA, CYP4F22, ELOVL4, ERCC2, FLG, GJB2, KRT1, KRT10, KRT2, LIPN, LORICRIN, MBTPS2, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SLC27A4, SNAP29, SPINK5, SREBF1, ST14, STS, SULT2B1, TGM1 Congenital ichthyosis, autosomal recessive (ARCI): 14 genes (28,7 kb) ABCA12, ALOX12B, ALOXE3, CASP14, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, SDR9C7, SLC27A4, ST14, SULT2B1, TGM1 Ichthyosis, autosomal dominant or X-linked: 7 genes (21,3 kb) ASPRV1, FLG, GJB2, KRT1, KRT2, KRT10, STS	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Skin and Dental Diseases		
Multiple pterygium syndrome * Gene panel: ID158.01, 8 genes (16,8 kb) CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1, CHRNA1	3 - 5 w	E
Neurofibromatosis (NF) * Gene panel: ID210.00, 3 genes (11,6 kb) NF1, NF2, SPRED1	3 - 5 w	E
Oculocutaneous albinism (OCA) * Gene panel: ID082.02, 9 genes (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 w	E
Orofaciodigital syndrome (OFD) * Gene panel: ID265.00, 9 genes (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 w	E
Pachyonychia congenita (PC) * Gene panel: ID120.01, 13 genes (30,1 kb) AAGAB, ATP2A2, DSG1, DSP, GJB6, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, MBTPS2, TRPV3	3 - 5 w	E
Palmoplantar keratoderma (PPK) * Gene panel: ID323.00 Palmoplantar keratoderma (PPK): 33 genes (58,2 kb) AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJA1, GJB2, GJB3, GJB4, JUP, KDSR, KRT1, KRT6A, KRT6B, KRT6C, KRT9, KRT10, KRT14, KRT16, KRT17, KRT83, LORICRIN, MBTPS2, PERP, RHBDF2, SERPINB7, SLURP1, SMARCAD1, TAT, TRPM4, TRPV3, WNT10A Palmoplantar keratoderma (PPK), nonsyndromic: 11 Gene (25,7 kb) AAGAB, AQP5, DSG1, DSP, KRT1, KRT6C, KRT9, KRT10, KRT16, SERPINB7, TRPV3 Palmoplantar keratoderma (PPK), syndromic: 15 Gene (22,4 kb) CTSC, ENPP1, GJA1, GJB2, KRT14, MBTPS2, JUP, LORICRIN, PERP, RHBDF2, SLURP1, SMARCAD1, TAT, TRPV3, WNT10A Erythrokeratoderma variabilis progressiva (EKVP): 7 Gene (9,5 kb) GJA1, GJB3, GJB4, KDSR, KRT83, PERP, TRPM4 Pachyonychia congenita (PC): 4 Gene (6,1 kb) KRT6A, KRT6B, KRT16, KRT17	3 - 5 w	E
Peeling skin syndrome (PSS) * Gene panel: ID309.00, 6 genes (13,6 kb) CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5	3 - 5 w	E
Pierre Robin sequence * Gene panel: ID294.00, 33 genes (72,6 kb) AMER1, AP3D1, BMP2, COG1, COL2A1, COL11A1, COL11A2, DHODH, EDN1, EFTUD2, EIF4E3, GNAI3, MYMK, PDHA1, PGAP3, PGM1, PIGA, PLCB4, POLR1B, POLR1C, POLR1D, RBM10, SATB2, SCUBE3, SF3B4, SLC10A7, SLC26A2, SNRPB, SOX9, TBX1, TCOF1, TGDS, WASHC5	3 - 5 w	E
Primary lymphedema (LMPHM) * Gene panel: ID372.00, 21 genes (69,4 kb) ADAMTS3, ANGPT2, CALCRL, CCB1, CELSR1, EPHB4, ERG, FAT4, FLT4, FOXC2, GATA2, GJA1, GJC2, KIF11, MDFIC, PIEZO1, PTPN14, SOX18, THSD1, TIE1, VEGFC	3 - 5 w	E
Progeria and progeroid syndromes * Gene panel: ID147.01, 25 genes (61,9 kb) ALDH18A1, B3GALT6, B4GALT7, BANF1, BLM, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LEMD2, LMNA, MTX2, PDGFRB, POLD1, POLR3A, PYCR1, RECQL, RECQL4, SLC25A24, TOMM7, WRN, ZMPSTE24	3 - 5 w	E
Selective tooth agenesis (STHAG) * Gene panel: ID151.02, 16 genes (26,9 kb) EDA, EDAR, EDARADD, GREM2, IRF6, KDF1, KREMEN1, LRP6, LTBP3, MSX1, PAX9, PTH1R, TP63, TSPEAR, WNT10A, WNT10B	3 - 5 w	E
Systemic diseases with multiple café-au-lait spots * Gene panel: ID351.00 Systemic diseases with multiple café-au-lait spots: 9 genes (27,6 kb) BRAF, MLH1, MSH2, MSH6, NF1, PMS2, PTPN11, RAF1, SPRED1 Mismatch repair cancer syndrome (MMRCS): 4 genes (11,8 kb) MLH1, MSH2, MSH6, PMS2 LEOPARD syndrome (LPRD): 3 genes (6,0 kb) BRAF, PTPN11, RAF1 Neurofibromatosis (NF1, NF1S): 2 genes (9,8 kb) NF1, SPRED1	3 - 5 w	E
Tuberous sclerosis (TSC) * Gene panel: ID332.00, 2 genes (8,9 kb) TSC1, TSC2	2 - 4 w	E
Xeroderma pigmentosum (XP) * Gene panel: ID282.00, 10 genes (23,5 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
BRCA1 and BRCA2-associated cancer * Gene panel: ID001.00, 2 genes (15,8 kb) BRCA1, BRCA2	2 - 4 w	E
Breast cancer * Gene panel: ID021.02, 12 genes (41,0 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53	3 - 5 w	E
Breast and ovarian cancer (HBOC) * Gene panel: ID003.04 Breast and ovarian cancer (HBOC): 19 genes (62,5 kb) ATM, BARD1, BRIP1, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53 Breast cancer: 10 genes (38,9 kb) ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, STK11, TP53 Ovarian cancer: 14 genes (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 w	E
Breast and ovarian cancer, comprehensive diagnostics * Gene panel: ID068.01, 50 genes (151,5 kb) ABRAXAS1, ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, POLD1, POLE, PTEN, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RECQL, RECQL4, SDHB, SDHC, SDHD, SLX4, SMARCA4, STK11, TP53, XRCC2	4 - 6 w	E
Chromosomal instability syndromes * Gene panel: ID326.01, 40 genes (121,0 kb) ANAPC1, ATM, BLM, BRCA1, BRCA2, BRIP1, DDB2, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, MRE11, NBN, PALB2, PCNA, POLH, RAD50, RAD51, RAD51C, RECQL4, RFWF3, SLX4, TOP3A, UBE2T, WRN, XPA, XPC, XRCC2	4 - 6 w	E
Colorectal cancer * Gene panel: ID049.03, 18 genes (55,7 kb) APC, ATM, BMPR1A, CHEK2, EPCAM, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11	3 - 5 w	E
Colorectal cancer and polyposis * Gene panel: ID006.09 Colorectal cancer and polyposis: 23 genes (64,1 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53 Colorectal cancer: 18 genes (55,7 kb) APC, ATM, CHEK2, BMPR1A, EPCAM, MBD4, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11 Colorectal polyposis: 15 genes (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 w	E
Colorectal cancer with microsatellite instability (MSI) * Gene panel: ID283.00, 9 genes (25,4 kb) EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE	3 - 5 w	E
Colorectal cancer, comprehensive diagnostics * Gene panel: ID285.02, 34 genes (89,7 kb) APC, ATM, AXIN2, BLM, BMPR1A, CDH1, CHEK2, EPCAM, EXO1, FLCN, GALNT12, GREM1, MBD4, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN*, NTHL1, PLA2G2A, PMS2, POLD1, POLE, PTEN, RFC1, RPA1, RNF43, RPS20, SMAD4, STK11, TGFBR2, TP53	3 - 5 w	E
Constitutional mismatch repair deficiency syndrome (CMMRDS, MMRCS) * Gene panel: ID362.00, 5 genes (12,7 kb) EPCAM, MLH1, MSH2, MSH6, PMS2	3 - 5 w	E
Cowden syndrome (CWS) * Gene panel: ID075.01, 8 genes (12,8 kb) AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1	3 - 5 w	E
Cutaneous malignant melanoma (CMM) * Gene panel: ID193.01, 12 genes (26,4 kb) BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3	3 - 5 w	E
DNA repair deficiency syndromes, comprehensive diagnostics * Gene panel: ID348.00, 221 genes (507,3 kb) ABRAXAS1, ALKBH2, ALKBH3, ANAPC1, APEX1, APEX2, APLF, APTX, ATM, ATR, ATRIP, ATRX, BARD1, BLM, BRCA1, BRCA2, BRIP1, CCNH, CDK7, CETN2, CHAF1A, CHEK1, CHEK2, CLK2, DCLRE1A, DCLRE1B, DCLRE1C, DDB1, DDB2, DMC1, DNA2, DNPH1, DNNT, DUT, EME1, EME2, ENDOV, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, EXO1, EXO5, FAAP100, FAAP20, FAAP24, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FEN1, GEN1, GTF2E2, GTF2H1, GTF2H2, GTF2H3, GTF2H4, GTF2H5, H2AX, HELQ, HERC2, HFM1, HLTf, HMCES, HUS1, LIG1, LIG3, LIG4, MAD2L2, MBD4, MDC1, MGMT, MLH1, MLH3, MMS19, MNAT1, MPG, MPLKIP, MRE11, MSH2, MSH3, MSH4, MSH5, MSH6, MUS81, MUTYH, NABP2, NBN, NEIL1, NEIL2, NEIL3, NHEJ1, NTHL1, NUDT1, NUDT15, NUDT18, OGG1, PALB2, PARC, PARK7, PARP1, PARP2, PARP3, PARBP, PAXIP1, PCNA, PDS5B, PER1, PMS1, PMS2, PNKP, POLA1, POLB, POLD1, POLD2, POLD3, POLD4, POLE, POLE2, POLE3, POLE4, POLG, POLH, POLI, POLK, POLL, POLM, POLN, POLQ, PRIMPOL, PRKDC, PRPF19, RAD1, RAD17, RAD18, RAD23A, RAD23B, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54B, RAD54L, RAD9A, RBBP8, RDM1, RECQL, RECQL4, RECQL5, REV1, REV3L, RIF1, RFWF3, RMI1, RNF168, RNF4, RNF8, RPA1, RPA2, RPA3, RPA4, RRM2B, SEM1, SETMAR, SHLD1, SHLD2, SHLD3, SHPRH, SLX1A, SLX1B, SLX4, SMC5, SMC6, SMUG1, SPIDR, SPO11, SPRTN, SWI5, SWSAP1, TDG, TDP1, TDP2, TOP3A, TOPBP1, TP53, TP53BP1, TREX1, TREX2, UBE2A, UBE2B, UBE2N, UBE2T, UBE2V2, UNG, USP1, UVSSA, WDR48, WRN, XAB2, XPA, XPC, XRCC1, XRCC2, XRCC3, XRCC4, XRCC5, XRCC6, ZSWIM7	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
Dyskeratosis congenita (DKC) * Gene panel: ID347.01, 15 genes (24,1 kb) ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53	3 - 5 w	E
Endometrial cancer* Gene panel: ID364.00, 12 genes (29,1 kb) EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, STK11, TP53	3 - 5 w	E
Endometrial cancer, comprehensive diagnostics * Gene panel: ID365.00, 26 genes (92,9 kb) ATM, APC, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, STK11, TP53	3 - 5 w	E
Fanconi anemia (FANC) * Gene panel: ID043.02, 21 genes (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2	3 - 5 w	E
Gastric cancer * Gene panel: ID090.04, 25 genes (73,6 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MBD4, MLH1, MSH2, MSH6, MUTYH, PMS2, PDGFRA, PTEN, SDHA, SDHB, SDHC, SDHD, SMAD4, STK11, TP53	3 - 5 w	E
Gastrointestinal stromal tumor (GIST) * Gene panel: ID226.00, 8 genes (19,0 kb) KIT, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD	3 - 5 w	E
Glioblastoma (GLM) * Gene panel: ID313.00, 16 genes (61,0 kb) APC, BRCA1, BRCA2, CDKN2A, EPCAM, MLH1, MSH2, MSH6, NF1, NF2, PMS2, POT1, PTEN, TP53, TSC1, TSC2	3 - 5 w	E
Gorlin-Goltz syndrome (BCNS) and similar syndromes * Gene panel: ID174.01, 7 genes (23,6 kb) CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 w	E
Lung cancer * Gene panel: ID260.02, 33 genes (122,5 kb) ATM, ATR, BAP1, BRCA1, BRCA2, BLM, CDH1, CDKN2A, CHEK2, DICER1, EGFR, ERBB2, ERCC2, FANCA, FANCC, FANCG, FANCD2, FGFR3, FLCN, JAK2, MET, MSH6, MUTYH, NBN, NF1, NKX2-1, PALB2, PRKN, RAD50, RECQL4, SDHA, TSC2, TP53	4 - 6 w	E
Lynch syndrome (LYNCH, HNPCC) * Gene panel: ID002.02, 5 genes (12,7 kb) MLH1, MSH2, MSH6, PMS2, EPCAM	3 - 5 w	E
Medulloblastoma * Gene panel: ID205.02, 22 genes (75,7 kb) APC, BRCA2, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTCH1, PTCH2, PTEN, SMARCB1, SMARCA4, SUFU, TP53, VHL	3 - 5 w	E
Myelodysplastic syndrome (MDS) and Acute myeloid leukemia (AML) * Gene panel: ID321.01 Myelodysplastic syndrome (MDS) and Acute myeloid leukemia (AML): 121 genes (244,1 kb) ACD, ADA2, ADH5, ALDH2, ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CHEK2, CLPB, CSF3R, CTC1, DCLRE1B, DDX41, DKC1, DNJC21, DNMT3A, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, G6PC3, GATA1, GATA2, GFI1, HAX1, HEATR3, HRAS, IKZF1, JAGN1, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MBD4, MDM4, MECOM, MLH1, MRAS, MSH2, MSH6, MYSM1, NAF1, NBN, NF1, NHP2, NOP10, NRAS, PALB2, PARN, PAX5, PMS2, PTPN11, RAD51, RAD51C, RAF1, RBBP6, RFWD3, RIT1, RPA1, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SOS1, SOS2, SRP54, SRP72, STAT3, STN1, TERC, TERT, TINF2, TP53, TSR2, TYMS, UBE2T, UNC13D, VPS45, WAS, WRAP53, XRCC2, ZCCHC8 Acute myeloid leukemia (AML): 12 genes (28,4 kb) ANKRD26, CEBPA, DDX41, ETV6, GATA2, RUNX1, SAMD9, SAMD9L, SRP72, TERC, TERT, TP53 Diamond-Blackfan anemia (DBA): 20 genes (11,3 kb) GATA1, HEATR3, RPL5, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2 Shwachman-Diamond syndrome (SDS): 4 genes (7,2 kb) DNJC21, EFL1, SBDS, SRP54 Bone marrow failure syndrome (BMFS): 8 genes (16,1 kb) ADH5, ALDH2, DNJC21, ERCC6L2, MDM4, MYSM1, SRP72, TP53 Pulmonary fibrosis and bone marrow failure (PFBMFT): 6 genes (13,0 kb) PARN, RPA1, RTEL1, TERC, TERT, ZCCHC8 Dyskeratosis congenita (DKC): 13 genes (21,8 kb) ACD, CTC1, DCLRE1B, DKC1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, WRAP53 Congenital neutropenia (SCN): 10 genes (13,9 kb) CLPB, CSF3R, ELANE, G6PC3, GFI1, HAX1, JAGN1, SRP54, VPS45, WAS Fanconi anemia (FANC): 20 genes (60,7 kb) BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, UBE2T, XRCC2	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
Nephroblastoma and Wilms tumor (WT) * Gene panel: ID335.00, 21 genes (70,4 kb) AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CEP57, CTR9, DICER1, DIS3L2, GPC3, GPC4, NSD1, PALB2, POU6F2, REST, TRIM28, TRIM37, TRIP13, WT1	3 - 5 w	E
Neuroendocrine neoplasia * Gene panel: ID386.00, 19 genes (29,2 kb) AIP, CDC73, CDKN1B, DLST, FH, MAX, MEN1, NF1, PRKAR1A, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL	3 - 5 w	E
Neurofibromatosis (NF) * Gene panel: ID210.00, 3 genes (11,6 kb) NF1, NF2, SPRED1	3 - 5 w	E
Ovarian cancer * Gene panel: ID004.04, 14 genes (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 w	E
Pancreatic cancer * Gene panel: ID089.03, 17 genes (56,2 kb) APC, ATM, BRCA1, BRCA2, CDKN2A, CTRC, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL	3 - 5 w	E
Pheochromocytoma/paraganglioma syndrome (PPGL) * Gene panel: ID042.03, 14 genes (22,8 kb) DLST, FH, MAX, MDH2, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 w	E
Pituitary adenoma (PITA) * Gene panel: ID387.00, 8 genes (20,7 kb) AIP, CDH23, CDKN1B, GNAS, GPR101, MEN1, PRKAR1A, USP8	3 - 5 w	E
Plasmocytoma * Gene panel: ID354.01, 40 genes (89,5 kb) ARID1A, ATM, BLM, BTK, CASP8, CASP10, CDKN2A, CBL, CTLA4, DDX41, EFL1, ETV6, FANCA, FAS, FASLG, HCLS1, KDM1A, KLHDC8B, KRAS, LAPTM5, MLH1, MSH2, MSH6, MYD88, NBN, NF1, NRAS, PAX5, PRF1, PMS2, POT1, PRKCD, PTPN11, RBM8A, SBDS, SH2B3, SH2D1A, TP53, USP45, WAS	3 - 5 w	E
Polyposis syndrome (PS, FAP) * Gene panel: ID005.06, 15 genes (39,4 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MBD4, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 w	E
Prostate cancer * Gene panel: ID140.03, 30 genes (101,5 kb) AR, ATM, ATR, BAP1, BARD1, BRCA1, BRCA2, BRIP1, CYP3A43, CDH1, CHEK2, ELAC2, EPCAM, FANCM, HOXB13, MLH1, MRE11, MSR1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, RECQL, RNASEL, TP53, TRRAP	3 - 5 w	E
Renal cancer * Gene panel: ID041.04, 34 genes (76,8 kb) BAP1, CDC73, CDKN1C, CDKN2B, CHEK2, CTR9, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MITF, MLH1, MSH2, MSH6, PBRM1, PMS2, PTEN, REST, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TRIM28, TSC1, TSC2, VHL, WT1	3 - 5 w	E
Soft tissue sarcoma and Osteosarcoma * Gene panel: ID223.02, 55 genes (155,1 kb) APC, ATM, ATR, ATRX, BLM, BRCA2, BUB1B, CDKN1C, CDKN2A, CEP57, CHEK2, DICER1, DKC1, EPCAM, ERCC2, EXT1, EXT2, FAH, FANCC, FH, HRAS, KIT, MEN1, MLH1, MRE11, MSH2, MSH6, MTAP, NBN, NF1, PALB2, PDGFRA, PMS2, POT1, PRKAR1A, PTCH1, PTEN, RB1, RECQL4, RPS19, SDHA, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, SQSTM1, SUFU, TBXT, TNFRSF11A, TP53, VHL, WAS, WRN, ZNF687	4 - 6 w	E
Thyroid cancer * Gene panel: ID220.02, 26 genes (60,5 kb) ACD, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, POT1, PRKAR1A, PTEN, RET, SDHB, SDHD, SEC23B, SRGAP1, SRRM2, TINF2, TP53	3 - 5 w	E
Tumor syndromes, comprehensive diagnostics * Gene panel: ID018.05, 357 genes (796,5 kb) ABRAXAS1, ACD, ADA2, ADH5, AIP, AKT1, ALDH2, ALK, AMER1, ANAPC1, ANKRD26, APC, APTX, AR, ARID1A, ASXL1, ATM, ATR, ATRX, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BTK, BUB1B, CASP10, CASP8, CASR, CBL, CCND1, CDC73, CDCA7, CDH1, CDH23, CDK4, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CEBPA, CEP57, CHEK2, CLPB, CSF3R, CTC1, CTLA4, CTNNA1, CTR9, CTRC, CYLD, CYP3A43, DCLRE1B, DCLRE1C, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, DNA2, DNAJC21, DNMT3A, DNMT3B, DUT, EFL1, EGFR, EHPB1, ELAC2, ELANE, ELP1, ENOSF1, EPCAM, ERBB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ETV6, EXO1, EXT1, EXT2, EZH2, FAH, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGFR3, FH, FLCN, FOXE1, G6PC3, GALNT12, GATA1, GATA2, GDNF, GEN1, GFI1, GNAS, GPC3, GPC4, GPR101, GPR161, GREM1, GTF2E2, GTF2H5, HABP2, HAVCR2, HAX1, HCLS1, HEATR3, HELLS, HERC2, HFM1, HNF1A, HNF1B, HOXB13, HRAS, IKZF1, JAGN1, JAK2, KDM1A, KIF1B, KIT, KLHDC8B, KRAS, LAPTM5, LIG1, LIG3, LIG4, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAP2K5, MAPK1, MAX, MBD4, MC1R, MDH2, MDM4, MECOM, MEN1, MET, MGMT, MINPP1, MITF, MLH1, MLH3, MPLKIP, MRAS, MRE11, MSH2, MSH3, MSH4, MSH5, MSH6, MSR1, MTAP, MUTYH, MYD88, MYSM1, NAF1, NBN, NDUFA13, NF1, NF2, NHEJ1, NHP2, NKX2-1, NOP10, NRAS, NSD1, NTHL1, NTRK1, OGG1, PALB2, PALLD, PARK7, PARN, PAX5, PBRM1, PCNA, PDGFRA, PHOX2B, PIK3CA, PLA2G2A, PMS1, PMS2, PNKP, POLA1, POLD1, POLE, POLG, POLH, POT1, POU6F2, PRF1, PRIMPOL, PRKAR1A, PRKCD, PRKDC, PRKN, PRSS1, PTCH1, PTCH2, PTEN, PTPN11, RABL3, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD54B, RAD54L, RAF1, RASA2, RB1, RBBP6, RBBP8, RBM8A, RECQL, RECQL4, REST, RET, RFC1, RFWD3, RHBDF2, RINT1, RIT1, RNASEL, RNF139, RNF168, RNF43, RPA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS19, RPS20, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RRAS2, RRM2B, RTE1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SH2B3, SH2D1A, SHOC2, SLC25A11, SLX4, SMAD4, SMAD7, SMARCA4, SMARCB1, SMARCE1, SMC5, SOS1, SOS2, SPIDR, SPINK1, SPRED1, SPRTN, SQSTM1, SRGAP1, SRP54, SRP72, STAT3, STK11, STN1, SUFU, TBXT, TDP1, TDP2, TERC, TERF2IP, TERT, TGFB2, TINF2, TMEM127, TNFRSF11A, TOP3A, TP53, TREX1, TRIM28, TRIM37, TRIP13, TRRAP, TSC1, TSC2, TSR2, TYMS, TYR, UBE2A, UBE2T, UNC13D, UNG, USB1, USP8, USP45, UVSSA, VHL, VPS45, WAS, WRAP53, WRN, WT1, WWP1, XPA, XPC, XRCC1, XRCC2, XRCC3, XRCC4, ZBTB24, ZCCHC8, ZNF687, ZSWIM7	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
Tumor syndromes in childhood * Gene panel: ID333.00 Tumor syndromes in childhood: 139 genes (341,1kb) ACD, ALK, ANKRD26, APC, ATM, BAP1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, DNAJC21, EFL1, ELANE, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, GATA2, GPC3, HRAS, IKZF1, KIF1B, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MRAS, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NKX2-1, NOP10, NHP2, NRAS, NSD1, PALB2, PARN, PAX5, PHOX2B, PMS2, POLE, POLH, POU6F2, PRKAR1A, PTCH1, PTCH2, PTEN, PTPN11, RAD51, RAD51C, RAF1, RB1, RECQL4, REST, RET, RFWD3, RIT1, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, SOS2, SRP72, STK11, SUFU, TERT, TMEM127, TINF2, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2 Hematological malignancies: 49 genes (126,4 kb) ACD, ANKRD26, ATM, BLM, BRCA1, BRCA2, BRIP1, CEBPA, DDX41, DKC1, DNAJC21, EFL1, ELANE, ERCC4, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GATA2, IKZF1, MAD2L2, NBN, NHP2, NOP10, PALB2, PARN, PAX5, RAD51, RAD51C, RFWD3, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SLX4, SRP72, TERT, TINF2, TP53, UBE2T, WRAP53, XRCC2 Neural tumors: 32 genes (111,9 kb) ALK, APC, BRCA2, CDKN2A, CHEK2, DICER1, ELP1, EPCAM, ERCC2, FANCM, KIF1B, LZTR1, MLH1, MSH2, MSH6, NBN, NF1, NF2, PALB2, PHOX2B, PMS2, PTCH1, PTCH2, PTEN, RB1, SMARCA4, SMARCB1, SUFU, TP53, TSC1, TSC2, VHL Endocrine tumors: 15 genes (21,2 kb) CDC73, CDKN1B, DLST, KIF1B, MAX, MEN1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL RAS-related disorders: 18 genes (36,6 kb) BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PTPN11, RAF1, RIT1, RRAS2, SHOC2, SOS1, SOS2 Wilms tumor (WT): 10 genes (29,5 kb) BRCA2, CDKN1C, GPC3, DIS3L2, POU6F2, TRIM28, WT1, TRIM37, CDC73, REST Xeroderma pigmentosum (XP): 9 genes (19,0 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC MMR deficiency syndrome (MMRCS): 5 genes (12,7 kb) MLH1, MSH2, PMS2, MSH6, EPCAM	4 - 6 w	E
Tuberous sclerosis (TSC) * Gene panel: ID332.00, 2 genes (8,9 kb) TSC1, TSC2	2 - 4 w	E
Urothelial cancer * Gene panel: ID337.00, 34 genes (105,0 kb) APC, ATM, BAP1, BARD1, BLM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, ERCC2, ERCC3, ERCC5, FANCC, FH, GEN1, MITF, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, RAD50, RAD51B, RAD51C, RB1, RECQL4, SDHA, TP53, XPC	4 - 6 w	E
Xeroderma pigmentosum (XP) * Gene panel: ID282.00, 10 genes (23,5 kb) DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Prenatal Diagnostics: Fetal Anomalies		
Fetal anomalies * Gene panel: ID850.00, 1223 genes (3235,6 kb) AAAS, ABCA12, ABCO6, ABCO9, ABHD5, ABL1, ACAD9, ACADVL, ACAN, ACE, ACOX1, ACP5, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTG2, ACVR2B, ACY1, ADAMTS10, ADAMTS17, ADAMTS3, ADAMTSL2, ADAR, ADGRG1, ADGRG6, ADNP, ADSL, AFF4, AGK, AGL, AGPS, AHCY, AHDC1, AH11, AKT1, AKT2, AKT3, ALDH18A1, ALDH1A3, ALDH3A2, ALDH7A1, ALDOA, ALG1, ALG12, ALG2, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPL, ALX1, ALX3, ALX4, AMACR, AMER1, AMMECR1, AMPD2, AMT, ANAPC1, ANKH, ANKRD11, ANKS6, ANOS1, ANTXR1, ANTXR2, AP1S2, AP4E1, AR, ARCN1, ARFGF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARMCO9, ARSA, ARSB, ARSL, ARX, ASAH1, ASCC1, ASNS, ASPA, ASPM, ASS1, ASXL1, ATAD3A, ATIC, ATP1A2, ATP6VOA2, ATP7A, ATR, ATRX, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BACP31, BCL11A, BCOR, BCS1L, BFPSP2, BGN, BHLHA9, BICD2, BIN1, BLM, BLTP1, BMP1, BMP2, BMP4, BMP6, BMPER, BMPR1B, BNC2, BPNT2, BRAF, BRAT1, BRCA2, BRIP1, BRPF1, BSND, BTD, BUB1B, C2CD3, CA2, CA8, CACNA1C, CACNA1E, CACNA1G, CANT1, CASK, CASR, CBL, CC2D2A, CCBF1, CCDC103, CCDC39, CCDC40, CCDC8, CCDC88C, CCND2, CCNQ, CDAN1, CDC45, CDC6, CDH1, CDH3, CDK13, CDK5RAP2, CDK8, CDKL5, CDKN1C, CDON, CDT1, CELSR1, CENPF, CENPJ, CEP104, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP55, CEP57, CEP63, CEP83, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFAP418, CFAP53, CFC1, CFL2, CFTR, CHAMP1, CHAT, CHD4, CHD7, CHKB, CHMP1A, CHRNA1, CHRNA3, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHSY1, CHUK, CILK1, CIT, CKAP2L, CLCN7, CLP1, CLPB, CNOT1, CNOT3, CNTNAP1, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL12A1, COL13A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A1, COL9A2, COLEC10, COLEC11, COLQ, COQ4, COQ9, COX7B, CPLANE1, CPT2, CRADD, CRB2, CREB3L1, CREBBP, CRIPT, CRLF1, CRPPA, CRTAP, CRYAA, CRYA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CSF1R, CSNK2A1, CSPP1, CTC1, CTCF, CTNNB1, CTNND1, CTSA, CTSD, CTSK, CTU2, CUL4B, CUL7, CWC27, CYP11A1, CYP11B1, CYP17A1, CYP1B1, CYP21A2, CYP26B1, CYP2U1, CYP4F22, DAG1, DARS1, DCHS1, DCX, DDR2, DDX11, DDX3X, DDX59, DENND5A, DHCR24, DHCR7, DHFR, DHODH, DIAPH1, DIS3L2, DISP1, DKC1, DLL3, DLL4, DLX5, DMPK, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJB11, DNAI1, DNML1, DNM2, DNMT3A, DNMT3B, DOCK6, DOF3, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DSP, DSTYK, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC211, DYNC2I2, DYNC2LI1, DYNLT2B, DYRK1A, DZIP1L, EBF3, EBP, ECEL1, EDA, EDNRA, EDNRB, EED, EFNB1, EFTUD2, EHMT1, EIF2AK3, EIF2B2, EIF2B3, EIF2S3, EIF4A3, EIF5A, ELAC2, ELN, ELOVL4, EMD, EML1, EMX2, ENPP1, EOGT, EP300, EPG5, EPHB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERCC9, ESCO2, ETFA, ETFB, ETFDH, EVC, EVC2, EXOC3L2, EXOSC3, EXT1, EXT2, EXTL3, EYA1, EYH2, EZH2, FAH, FAM111A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANGC, FANCI, FANCL, FAR1, FAT4, FBLN5, FBN1, FBN2, FBXL4, FGD1, FGF10, FGF3, FGF8, FGFR1, FGFR2, FGFR3, FH, FIG4, FKBP10, FKBP14, FKRP, FKTN, FLNA, FLNB, FLNC, FLT4, FLVCR2, FOLR1, FOXC1, FOXC2, FOXE1, FOXE3, FOXF1, FOXG1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMD4A, FTL, FUT8, FYCO1, FZD2, G6PC3, GAA, GALT, GALE, GALK1, GALS, GALT2, GANAB, GATA2, GATA3, GATA4, GATA6, GBA1, GBA2, GBE1, GCDH, GDF1, GDF5, GDF6, GFAP, GFM1, GFPT1, GJA1, GJA3, GJA8, GJC2, GLA, GLB1, GLDC, GLDN, GLE1, GLI1, GLI2, GLI3, GLIS3, GLUL, GMNN, GMPBP, GNAI3, GNAO1, GNAS, GNB1, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC6, GPI, GPM2, GREB1L, GRHL3, GRIN1, GRIN2B, GRIP1, GSC, GTF2H5, GTPBP3, GUCY2C, GUSB, GZF1, H1-4, HAAO, HADHA, HADHB, HBA1, HBA2, HCCS, HCF1, HDAC8, HES7, HESX1, HIBCH, HIVEP2, HMG2, HNF1B, HNF4A, HNRNP, HOXA1, HOXA13, HOXD13, HPSE2, HR, HRAS, HSD17B3, HSD17B4, HSFA, HSPD1, HSPG2, HUWE1, HYCC1, HYL51, IARS1, IDH1, IDS, IDUA, IER3IP1, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGF2, IGHMBP2, IHH, IKBKG, IL11RA, IL1RAPL1, INPP5E, INPPL1, INSR, INTU, INVS, IQCB1, IRF6, ITGA3, ITGA6, ITGA8, ITGB4, JAG1, KANSL1, KAT6A, KAT6B, KATNB1, KCNJ1, KCNJ2, KCTD1, KDM5C, KDM6A, KIAA0586, KIAA0753, KIF11, KIF14, KIF1A, KIF1B, KIF22, KIF2A, KIF5C, KIF7, KLF1, KLHL40, KLHL41, KLHL7, KMT2A, KMT2C, KMT2D, KNL1, KRAS, KRIT1, KYNU, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMC3, LARGE1, LARP7, LBR, LFNG, LGI4, LHX3, LHX4, LIFR, LGI4, LIPA, LMBR1, LMBRD1, LMNA, LMNB1, LMNB2, LMOD3, LMX1B, LONP1, LR2, LRP4, LRP5, LRRCS6, LTBP3, LTBPA, LYST, LZTF1, LZTR1, MAB21L2, MACEF1, MAF, MAFB, MAGEL2, MAP2K1, MAP2K2, MAP3K1, MAP3K20, MAP3K7, MAPRE2, MASP1, MATN3, MBTPS2, MCOLN1, MCPH1, MED12, MEF2C, MEGF10, MEGF8, MEIS2, MEOX1, MESD, MESP2, MFRP, MFSD2A, MGP, MID1, MKKS, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MMP13, MMP21, MN1, MNX1, MOCS1, MOCS2, MOGS, MPDU1, MPLKIP, MRAS, MRPS22, MSL3, MSMO1, MSTO1, MSX1, MSX2, MTM1, MTO1, MTOR, MTRFR, MUSK, MYBPC1, MYCN, MYH10, MYH11, MYH2, MYH3, MYH6, MYH7, MYH8, MYH9, MYL1, MYMK, MYO18B, MYO9A, MYOCD, MYPN, MYRF, MYT1, NAA10, NACCC1, NADSYN1, NAGA, NALCN, NANS, NBAS, NBN, NDE1, NDP, NDUFAF5, NEB, NECTIN1, NECTIN4, NEDD4L, NEK1, NEK8, NEK9, NEU1, NF1, NFIX, NHEJ1, NHS, NIPAL4, NIPBL, NKX2-5, NKX3-2, NODAL, NOG, NOTCH1, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPR2, NR0B1, NR2F2, NR5A1, NRAS, NSD1, NSDHL, NUBPL, NUP107, NXN, OBSL1, OCLN, OCLR, ODAD1, ODAD2, ODAD3, OFD1, OPHN1, ORC1, ORC4, ORC6, OSGPE, OSTM1, OTX2, P3H1, P4HB, PAFAH1B1, PAK3, PALB2, PAPSS2, PARN, PAX2, PAX3, PAX6, PAX7, PAX8, PBX1, PCGF2, PCNT, PCYT1A, PDCD10, PDE4D, PDGFRB, PDHA1, PEDP, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAP2, PGAP3, PGM1, PGM3, PHF6, PHF8, PHGDH, PHIP, PHOX2B, PIBF1, PIEZO1, PIEZO2, PIGA, PIGL, PIGN, PIGO, PIGT, PIGV, PIK3C2A, PIK3CA, PIK3R1, PIK3R2, PITX1, PITX2, PITX3, PKD1, PKD1L1, PKD2, PKHD1, PKLR, PLG1, PLG, PLK4, PLOD1, PLOD2, PMM2, PNKP, PNPLA1, POC1A, POGZ, POLE, POLG2, POLR1A, POLR1B, POLR1C, POLR1D, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POP1, POR, PORCN, POU1F1, PP1B, PPP1CB, PPP2R1A, PPP2R5D, PQBP1, PRG4, PRIM1, PRKAG2, PRKAR1A, PRKD1, PRMT7, PRRX1, PRSS56, PRUNE1, PSAP, PSAT1, PSPH, PTCH1, PTDSS1, PTF1A, PTH1R, PTHLH, PTPN11, PTPN14, PTS, PUF60, PYCR1, PYGM, QRICH1, RAB18, RAB23, RAB33B, RAB3GAP1, RAB3GAP2, RAC1, RAD21, RAF1, RAI1, RAPSN, RAR, RARS2, RASA1, RAX, RBBP8, RBM10, RBM8A, RBPJ, RECQL4, RELN, REN, RERE, RET, RFT1, RFX6, RPK4, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU4ATAC, ROBO1, ROBO3, ROGDI, ROR2, RPGRIP1L, RPL10, RPL11, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS6KA3, RPS7, RRA52, RRM2B, RSPH4A, RSPH9, RTKL1, RTTN, RUNX2, RXYLT1, RYR1, SALL1, SALL4, SAMD9, SAMHD1, SATB2, SBDS, SC5D, SCARF2, SCLT1, SCN1A, SCN2A, SCN4A, SCO2, SDCCAG8, SDR9C7, SEC23B, SEC24D, SEPECS, SERPINF1, SERPINA1, SETBP1, SETD5, SF3B4, SGCG, SGLP1, SH3PX2D2B, SHANK3, SHH, SHOC2, SHOX, SIK3, SIL1, SIK3, SIX5, SIX6, SKI, SKIC3, SLC10A7, SLC12A1, SLC12A6, SLC13A5, SLC16A2, SLC17A5, SLC18A3, SLC25A19, SLC25A20, SLC25A24, SLC25A38, SLC26A2, SLC26A3, SLC27A4, SLC29A3, SLC2A10, SLC33A1, SLC35A2, SLC35C1, SLC35D1, SLC39A8, SLC5A7, SLC6A9, SLX4, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCC1, SMARCE1, SMC1A, SMC3, SMCHD1, SMG9, SMN1, SMO, SMOC1, SMPD1, SMPD4, SMS, SNORD118, SNRPB, SNX10, SNX14, SON, SOS1, SOS2, SOST, SOX10, SOX17, SOX18, SOX2, SOX3, SOX6, SOX9, SP7, SPAG1, SPARC, SPATA5, SPECC1L, SPEG, SPG11, SPRED1, SRCAP, SRD5A2, SRD5A3, SRY, ST14, STAC3, STAG2, STAMBP, STAR, STIL, STRA6, STRADA, SUCLG1, SUFU, SULT2B1, SUMF1, SUZ12, TAB2, TAF1, TAFAZ1N, TALDO1, TAP1, TBC1D20, TBC1D23, TBC1D24, TBC1D32, TBDC, TBCE, TBCK, TBL1XR1, TBX1, TBX15, TBX18, TBX20, TBX3, TBX4, TBX5, TBX6, TCF12, TCF4, TCIRG1, TCOF1, TCTN1, TCTN2, TCTN3, TEO2, TENM3, TENT5A, TFAP2A, TFAP2B, TGDS, TGF2, TGF3, TGFBR1, TGFBR2, TGIF1, TGM1, THOC6, THRA, TINF2, TMC01, TMEM107, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM38B, TMEM67, TMEM98, TMX2, TNNT2, TNNT3, TOE1, TOP3A, TOR1A, TP63, TPM2, TPM3, TRAF3IP1, TRAF7, TRAP1, TRAP12, TRAPPC12, TRAPPC9, TREX1, TRIM37, TRIP11, TRIP12, TRIP4, TRMT10A, TRPS1, TRPV4, TRPV6, TSC1, TSC2, TSEN2, TSEN34, TSEN54, TSFM, TTC21B, TTC7A, TTC8, TTN, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TWIST1, TWIST2, TXNDC15, TXNL4A, UBA1, UBE2T, UBE3B, UBR1, UMP, UROS, USP18, USP9X, VAMP1, VEGFC, VIPAS39, VLDLR, VPS13B, VPS33B, VPS53, VRK1, VSX2, WDRCP, WDR19, WDR26, WDR35, WDR62, WDR73, WDR81, WNT1, WNT10B, WNT5A, WNT7A, WRAP53, WT1, XRCC4, XYLT1, XYLT2, YY1, ZBTB18, ZBTB20, ZC4H2, ZEB2, ZFP57, ZIC1, ZIC2, ZIC3, ZMPSTE24, ZMYND10, ZSWIM6	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Preventive Diagnostics: Carrier Screening		
Carrier-Screening * Gene panel: ID350.00, 625 genes (1591,4 kb) AAAS, ABCA12, ABCA3, ABCA4, ABCB11, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACE, ACOX1, ACSF3, ADA, ADAMTS13, ADAMTS2, ADGRG1, ADGRV1, AFF2, AGA, AGL, AGPS, AGT, AGR1, AGXT, AHI1, AIRE, AKR1D1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, ALS2, AMACR, AMH, AMHR2, AMT, ANK1, ANO10, ANTXR2, ARB1, AQP2, ARG1, ARSA, ARSB, ARSL, ARX, ASL, ASNS, ASPA, ASS1, ATIC, ATM, ATP6VOA2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AVPR2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BRIP1, BSND, BTBD, BTK, CA2, CAPN3, CASR, CBS, CC2D2A, CCDC88C, CD3D, CD3E, CD40LG, CDH23, CEP290, CERKL, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNE, CHRNA, CIITA, CLCN1, CLDN1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL11A2, COL17A1, COL1A2, COL27A1, COL4A3, COL4A4, COL4A5, COL7A1, COQ2, COQ8A, CPS1, CPT1A, CPT2, CRB1, CRLF1, CRTAP, CSTB, CTNS, CTSD, CTSK, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP27A1, CYP27B1, D2HGDH, DBT, DCLRE1C, DCX, DDB2, DDC, DGUOK, DHCR24, DHCR7, DHDDS, DKC1, DLD, DLL3, DMD, DMP1, DNAH5, DNAI1, DNAI2, DNAJC19, DNMT3B, DOK7, DOLK, DPAGT1, DPM1, DPYD, DSP, DUOX2, DUOXA2, DYNC2H1, DYSF, EDA, EDN3, EDNRB, EFEMP2, EGR2, EIF2AK3, EIF2B5, ELP1, EMD, ENPP1, EPB42, EPM2A, ERBB3, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYS, F11, F8, F9, FAH, FAM126A, FAM161A, FAM20C, FANCA, FANCB, FANCC, FANCG, FBLN5, FBP1, FGA, FGB, FGD4, FGG, FH, FKBP, FKTN, FLNA, FMO3, FOLR1, FOXN1, FOXP3, FRAS1, FREM2, FUCA1, FXN, G6PC1, G6PC3, G6PD, GAA, GALT, GALE, GALK1, GALNS, GALT, GAMT, GATA1, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GDF5, GFM1, GJA1, GJB1, GJB2, GJC2, GLA, GLB1, GLDC, GLE1, GLI3, GNE, GNPTAB, GNPTG, GNS, GP1BA, GP9, GRHPR, GRIP1, GSS, GTF2H5, GUCY2D, GUSB, GYS2, HADH, HADHA, HADHB, HAMP, HAX1, HBB, HESX1, HEXA, HEXB, HGD, HGSNAT, HIBCH, HJV, HLCS, HMGCL, HMGCS2, HOGA1, HPD, HPRT1, HPS1, HPS3, HPS4, HSD17B10, HSD17B3, HSD17B4, HSD3B2, HSD3B7, HSPG2, HYAL1, HYL1, IDS, IDUA, IGBP1, IGF1, IGHMBP2, IGSF1, IL2RG, IL7R, INPP5E, INS, INSR, INVS, IQCB1, ITGA6, ITGB4, IVD, IYD, JAG1, JAK3, KCNJ1, KCNJ11, KCNQ1, KCNQ2, KCTD7, L1CAM, LAMA2, LAMA3, LAMB2, LAMB3, LAMC2, LARGE1, LBR, LCA5, LDLR, LDLRAP1, LHCRG, LHX3, LIFR, LIPA, LMBRD1, LMNA, LOXHD1, LPL, LRP2, LRP5, LRPPRC, LYST, MAN2B1, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MED12, MED17, MEV1, MESP2, MFSD8, MGAT2, MID1, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMTU, MOCS1, MOCS2, MOGS, MPI, MPL, MPV17, MPZ, MRPS16, MRPS22, MTHFR, MTM1, MTR, MTRR, MTTP, MUTYH, MVK, MYO15A, MYO5A, MYO7A, NAGA, NAGLU, NAGS, NBN, NDP, NDRG1, NDUFAF5, NDUFS6, NEU1, NEUROG3, NHLRC1, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NROB1, NR2E3, NTRK1, NUP62, OAT, OCA2, OCRM, OFD1, OPA3, OSTM1, OTC, P3H1, PAH, PAX6, PAX8, PC, PCBD1, PCCA, PCCB, PCDH15, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PEX1, PEX10, PEX2, PEX6, PEX7, PFKM, PHGDH, PHKB, PKHD1, PKLR, PLA2G6, PLCE1, PLEC, PLEKHG5, PLG, PLOD1, PLP1, PNM2, PMP22, PNPO, POLG, POMGNT1, POMT1, POMT2, POR, POU1F1, PPT1, PQBP1, PREPL, PRF1, PROC, PROP1, PRPS1, PRRT2, PRX, PSAP, PSAT1, PTH1R, PTPRC, PTS, PUS1, PYGL, PYGM, QDPR, RAB23, RAB27A, RAB3GAP1, RAB3GAP2, RAG1, RAG2, RAPSIN, RARS2, RB1, RDH12, RELN, REN, RLBP1, RNASEH2B, RPE65, RPRG, RRGRI1L, RS1, RTEL1, SACS, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCN11A, SCN11B, SCN11G, SCOD, SEPSECS, SERPINA1, SFTPB, SFTPC, SGCA, SGCB, SGCD, SGCG, SGSH, SH2D1A, SIL1, SLC12A1, SLC12A3, SLC12A6, SLC16A1, SLC16A2, SLC17A5, SLC19A3, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A22, SLC26A2, SLC26A3, SLC26A4, SLC2A1, SLC34A2, SLC35A1, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A4, SLC3A1, SLC45A2, SLC4A1, SLC4A11, SLC5A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMARCA1, SMPD1, SNAP29, SP110, SPR, SRD5A2, ST3GAL5, STAR, STRA6, SUCLG1, SUMF1, SUOX, TAFAZZIN, TAT, TBCE, TCIRG1, TECPR2, TF, TFR2, TG, TGM1, TH, TIMM8A, TK2, TMEM216, TMEM67, TNFRSF11B, TNNT1, TPO, TPP1, TREX1, TRHR, TRIM32, TRIM37, TRMU, TSEN54, TSHB, TSHR, TSPYL1, TTC37, TTN, TTPA, TWNK, TYMP, TYR, TYRP1, UBA1, UBR1, UGT1A1, UQCRCB, UQCRCQ, UROS, USH1C, USH1G, USH2A, VDR, VLDLR, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, WAS, WNT10A, WNT3, WNT7A, WRN, WT1, XPA, XPC, ZIC3, ZMPSTE24, ZNF469	4 - 6 w	E