SYNLAB Human Genetics

Status: 06/2025

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!

For international orders, please send samples to Leinfelden. National orders can be sent directly to us.
Please contact us before sending prenatal samples to allow better tracking of the samples.

Panel Directory

Disease Types

Whole Exome Sequencing (WES)

Diseases/Diagnostics	TAT	Material
Brain Malformations		
Aicardi-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, 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
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, FKRP, FKTN, FLNA, 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, FKRP, FKTN, 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, FKRP, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Alagille syndrome (ALGS) #,*	3 - 5 w	E
Gene panel: ID112.00, 2 genes (11,1 kb)		
JAG1, NOTCH2		
Arrhythmogenic right ventricular cardiomyopathy (ARVD, ARVC) *	4 - 6 w	E
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		
Atrial and ventricular septal defect (ASD, VSD, AVSD) *	3 - 5 w	E
Gene panel: ID143.00, 12 genes (21,1 kb)		
ACTC1, CITED2, CRELD1, GATA4, GATA5, GATA6, GJA1, MYH6, NKX2-5, NR2F2, TBX20, TLL1		
Bicuspid aortic valve (AOVD) *	3 - 5 w	E
Gene panel: ID301.00, 6 genes (16,7 kb)		
GATA5, NOTCH1, NR2F2, ROBO4, SMAD6, TAB2		
Brain small vessel disease (BSVD) *	3 - 5 w	E
Gen panel: ID325.00, 3 genes (12,0 kb)		
COL4A1, COL4A2, COLGALT1		
Brugada syndrome (BRGDA) *	3 - 5 w	E
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		
Cerebrovascular diseases and stroke *	4 - 6 w	E
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, TGFBR1, TGFBR2, 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, TGFBR1, TGFBR2		
Moyamoya disease (MYMY): 4 genes (22,2 kb)		
ACTA2, GUCY1A1, JAG1, RNF213		
CADASIL, CARASIL: 2 genes (8,4 kb)		
HTRA1, NOTCH3		
CADASIL and CARASIL #,*	3 - 5 w	E
Gene panel: ID167.01, 3 genes (9,4 kb)		
HTRA1, NOTCH3, TREX1		
Cardiac arrhythmia, comprenensive diagnostics *	4 - 6 w	E
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		

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
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
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
CHARGE syndrome * Gene panel: ID307.00, 3 genes (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 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, TGFBR1, TGFBR2, 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, TGFBR1, TGFBR2, TKT, TMEM260, TMEM94, TRAF7, VPS35L, WASHC5, WDPCP, YY1AP1, ZEB2, ZIC3	4 - 6 w	E
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, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Cardiovascular 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, ATP6V0A2, 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, TGFBR1, TGFBR2, 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, TGFBR1, TGFBR2, 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, TGFBR2, TGFBR1 Cutis laxa syndrome (ARCL, ADCL): 11 genes (27,9 kb) ALDH18A1, ATP6V0A2, 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
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
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		
Hereditary hemorrhagic telangiectasia (HHT) * Gene panel: ID155.01, 7 genes (15,6 kb) ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4	3 - 5 w	E
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 (CVDP): 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, TGFBR1, TGFBR2, THSD4 Loeys-Dietz syndrome (LDS): 8 genes (11,1 kb) BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2 Nonsyndromic thoracic aortic aneurysm (AAT): 11 genes (24,0 kb) ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, 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, TGFBR1, TGFBR2	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, TGFBR1, TGFBR2	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, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, UPF3B, ZDHHC9, ZNF469 Marfan syndrome (MFS): 3 genes (11,8 kb) FBN1, TGFBR1, TGFBR2 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, TGFBR1, TGFBR2, 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
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

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
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
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
Sick sinus syndrome (SSS) * Gene panel: ID107.01, 4 genes (16,5 kb) GNB2, HCN4, MYH6, SCN5A	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
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

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
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, TGFBR1, TGFBR2, 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, TGFBR1, TGFBR2 CHARGE syndrome: 2 genes (11,3 kb) CHD7, SEMA3E VCRL syndrome: 3 genes (4,4 kb) HAAO, KYNU, NADSYN1	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, TGFBR1, TGFBR2, THSD4 Thoracic aortic aneurysm, nonsyndromic type (AAT): 10 genes (22,8 kb) ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4 Loeys-Dietz syndrome (LDS): 6 genes (8,4 kb) SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2	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

Diseases/Diagnostics	TAT	Material
Ciliopathies		
Renal ciliopathies, comprehensive diagnostics * Gene panel: ID376.00 Renal ciliopathies, comprehensive diagnostics: 75 genes (181,7 kp) 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
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 Creveld syndrome (EVC): 4 genes (11,5 kb) DYNC2L1, EVC, EVC1, WDR35	3 - 5 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, ATP6V0A2, 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, TGFBR1, TGFBR2, 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, TGFBR1, TGFBR2, 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, TGFBR2, TGFBR1 Cutis laxa syndrome (ARCL, ADCL): 11 genes (27,9 kb) ALDH18A1, ATP6V0A2, 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, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1	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

Diseases/Diagnostics	TAT	Material
Connective Tissue Diseases		
Ehlers-Danlos syndrome (EDS) *	3 - 5 w	E
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		
Loeys-Dietz syndrome (LDS) and similar aortic diseases *	4 - 6 w	E
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, TGFBR1, TGFBR2, THSD4		
Loeys-Dietz syndrome (LDS): 8 genes (11,1 kb)		
BGN, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2		
Nonsyndromic thoracic aortic aneurysm (AAT): 11 genes (24,0 kb)		
ACTA2, FOXE3, LOX, MAT2A, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, 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, TGFBR1, TGFBR2		
Marfan syndrome (MFS) #,*	2 - 4 w	E
Gene panel: ID022.00, 3 genes (11,8 kb)		
FBN1, TGFBR1, TGFBR2		
Marfan syndrome (MFS) and similar syndromes *	4 - 6 w	E
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, NKAP, NPR2, PLOD1, PRDM5, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4, UPF3B, ZDHHC9, ZNF469		
Marfan syndrome (MFS): 3 genes (11,8 kb)		
FBN1, TGFBR1, TGFBR2		
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, TGFBR1, TGFBR2, 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		
Osteogenesis imperfecta (OI) *	3 - 5 w	E
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		
Stickler syndrome (STL) *	3 - 5 w	E
Gene panel: ID062.00, 6 genes (21,2 kb)		
COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3		
Thoracic aortic aneurysm and aortic dissection (AAT, TAAD) *	3 - 5 w	E
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, TGFBR1, TGFBR2, THSD4		
Thoracic aortic aneurysm, nonsyndromic type (AAT): 10 genes (22,8 kb)		
ACTA2, FOXE3, LOX, MFAP5, MYH11, MYLK, PRKG1, TGFBR1, TGFBR2, THSD4		
Loeys-Dietz syndrome (LDS): 6 genes (8,4 kb)		
SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2		

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
Autism spectrum disorders * Gene panel: ID076.04, 168 genes (664,6 kb) AHDC1, ADNP, ADSL, 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, HNRNPU, 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
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, ZFHX4, 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, ZFHX4, 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, TMCO1, 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, TMCO1, 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, 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, EXOC2, EXOC7, EXOC8, FBXW11, FDFT1, 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, LNPK, 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, RALGAPA1, 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, TNR, 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
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, NR0B2, 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, NR0B2, 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
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

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
Rubinstein-Taybi syndrome (RSTS) * Gene panel: ID142.01, 3 genes (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 w	E
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, HMGA2, 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, PAPPA2, 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: ID170.05, 40 genes (78,9 kb) ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, 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) *	3 - 5 w	E
Gene panel: ID111.02, 7 genes (10,1 kb)		
CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR		
Congenital hypothyroidism *	3 - 5 w	E
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 dyshormonogenesis (TDH): 8 genes (23,8 kb)		
DUOX2, DUOXA2, IYD, SLC5A5, SLC26A4, SLC26A7, TG, TPO		
Comined pituitary hormone deficiency (CPHD): 8 genes (11,9 kb)		
HESX1, LHX3, LHX4, OTX2, POU1F1, PROP1, RNPC3, ROBO1		
Diabetes insipidus *	3 - 5 w	E
Gene panel: ID322.00, 5 genes (8,4 kb)		
AQP2, AVP, AVPR2, SLC12A1, WFS1		
Disorder of sex development (DSD) *	4 - 6 w	E
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, NR0B1, NR2F2, NR5A1,, POR, PPP1R12A, PPP2R3C, RSPO1, 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, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2		
46,XX Disorder of sex development (SRXX), nonsyndromic: 6 genes (6,6 kb)		
NR2F2, NR5A1, RSPO1, 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, NR0B1, POR, PPP1R12A, PPP2R3C, RSPO1, 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		
Endocrine hypertension *	3 - 5 w	E
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		
Glucocorticoid deficiency (GCCD) *	3 - 5 w	E
Gene panel: ID222.00, 16 genes (25,1 kb)		
AAAS, ABCD1, CYP11A1, HSD3B2, MC2R, MCM4, MRAP, NNT, NR0B1, NR3C1, PCSK1, POMC, PROP1, STAR, TBX19, TXNRD2		
Growth hormone deficiency (IGHD, CPHD) *	3 - 5 w	E
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		
Hyperaldosteronism (HALD) *	3 - 5 w	E
Gene panel: ID304.00, 6 genes (20,6 kb)		
CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5		

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: ID170.05, 40 genes (78,9 kb) ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, 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
Matury-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
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
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
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, NR0B2, 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, NR0B2, 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: ID293.02 Ovarian dysgenesis (ODG): 18 genes (30,9 kb) BMP15, CLPP, DAP3, ERAL1, ESR2, FSHR, HARS2, HR0B, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 Gonadal dysgenesis, XX type (ODG): 11 genes (19,1 kb) BMP15, ESR2, FSHR,HR0B, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 Gonadal dysgenesis, XX type, with deafness (PRLTS): 7 genes (11,8 kb) CLPP, DAP3, 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: ID387.01, 21 genes (47,3 kb) AIP, CDH23, CDKN1B, DICER1, EPCAM, GNAS, GPR101, MAX, MLH1, MSH2, MSH6, MEN1, PMS2, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, USP8	3 - 5 w	E
Primary and premature ovarian failure (POI, POF) * Gene panel: ID078.06 Primary and premature ovarian failure (POI, POF): 40 genes (88,5 kb) BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, ESR2, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HROB, HSF2BP, INHA, KASH5, LHCGR, MCM8, MCM9, MEIOB, MGA, MRPS22, MSH4, MSH5, NHEJ1, NOBOX, NR5A1, NUP107, POF1B, PSMC3IP, SOHLH1, SPATA22, SPIDR, STAG3, SYCE1, SYCP2L, TP63, XRCC2, ZSWIM7 Premature ovarian failure (POF): 26 genes (65,3 kb) BNC1, C14ORF39, DIAPH2, ERCC6, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, KASH5, MCM8, MEIOB, MGA, MSH4, MSH5, NOBOX, NR5A1, POF1B, SPATA22, STAG3, SYCE1, SYCP2L, TP63, XRCC2 Ovarian dysgenesis (ODG): 11 genes (19,1 kb) BMP15, ESR2, FSHR, HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7	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
Septooptic 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, ATP6V0A1, ATP6V1A, BRAT1, CACNA1A, CACNA1E, CACNB4, CAD, CASR, CDK19, CDKL5, CELF2, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, 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, RHOBTB2, RNF13, ROGDI, RORA, RORB, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SEMA6B, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC38A3, SLC6A1, SLC7A60S, 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, CHRNB2, 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, SLC7A60S, SLC12A5, STX1B, TBC1D24 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	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, ATP6V0A1, ATP6V1A, BRAT1, BSCL2, BTD, 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, RHOBTB2, 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 encepalopathy (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 Metabolic encephalopathy with epilepsy: 29 genes (44,8 kb) ABAT, ADSL, ALDH5A1, ALDH7A1, AMT, BTD, 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, GEFSP) * Gene panel: ID059.03 Febrile seizures with or without epilepsy (FEB, GEFSP): 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 (GEFSP): 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, CHRNB2, 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 lope epilepsy (ENFL): 5 genes (13,5 kb) CHRNA2, CHRNA4, CHRNB2, DEPDC5, KCNT1 Temporal lope 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, SLC7A60S, 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, SLC7A60S, 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, BTD, 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, BTD, 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
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, SLC7A60S, 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, SLC7A60S	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 Aniridie (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, CNNM4, CRX, DRAM2, GNAT2, GUCA1A, GUCY2D, KCNV2, PCARE, PCYT1A, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RP1, RPGR, RPGRIP1, SEMA4A, TLCD3B, TTLL5, 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		
Congentinal 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, TGFBI, 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, TGFBI Stromal corneal dystrophy: 5 genes (11,6 kb) CHST6, DCN, PIKFYVE, TGFBI, UBIAD1 Epithelial corneal dystrophy: 6 genes (12,6 kb) COL17A1, KRT3, KRT12, MCOLN1, TACSTD2, TGFBI 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, TMCO1, 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, TMCO1, 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) AIPL1, 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) # 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: ID383.01, 302 Gene (761,8 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, 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, MKKS, MKS1, MMACHC, MPDZ, MSTO1, MTTP, MVK, MYO7A, NBAS, NDP, NEK2, NEUROD1, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OFD1, OPN1LW, 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, 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, TLCD3B, 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, WDPCP, WDR19, WHRN, ZFYVE26, ZNF408, ZNF423, ZNF513	4 - 6 w	E
Retinitis pigmentosa (RP), comprehensive diagnsotics * Gene panel: ID288.02 Retinintis 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
Septooptic 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, DCDC2, DGUOK, FAH, FOCAD, GALE, GALM, 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 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

Diseases/Diagnostics	TAT	Material
Gastroenterological Diseases		
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
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: ID089.04, 19 genes (59,9 kb) APC, ATM, BARD1, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL, WT1	3 - 5 w	E
Pancreatitis (PCTT) * Gene panel: ID141.02, 18 genes (29,6 kb) APOA5, APOC2, CASR, CEL, CFTR, CPA1, CLDN2, CELA3B, CTRC, CTSB, GPIHBP1, LMF1, LPL, PNLIP, 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

Diseases/Diagnostics	TAT	Material
Hematological Diseases		
Anemias, comprehensive diagnostics * Gene panel: ID392.00 Anemias, comprehensive diagnostics: 187 genes (356,5 kb) ABCB7, ABCG5, ABCG8, ADA, ADA2, ADAMTS13, ADH5, AK1, AK2, ALAD, ALAS2, ALDH2, ALDOA, AMN, ANK1, ATM, ATP11C, ATRX, BLM, BRCA1, BRCA2, BRIP1, C3, C1GALT1C1, CBLIF, CD46, CD59, CDAN1, CDIN1, CFB, CFH, CFHR1, CFHR3, CFI, COL4A1, COX4I2, CPOX, CUBN, CYB5R3, DGKE, DHFR, DKC1, DNASE2, DNAJC21, ELANE, EPAS1, EPB41, EPB42, EPO, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FOXP3, FTCD, G6PC3, G6PD, GATA1, GCLC, GLRX5, GPI, GPX1, GSR, GSS, HAMP, HBA1, HBA2, HBB, HBD, HBG1, HBG2, HEATR3, HFE, HK1, HSPA9, HSCB, IARS2, IREB2, KCNN4, KIF23, KLF1, LARS2, LCAT, LIG4, LPIN2, MAD2L2, MDM4, MMACHC, MMADHC, MPIG6B, MPL, MTHFD1, MTR, MTRR, MYSM1, NBN, NDUFB11, NHEJ1, NHLRC2, NHP2, NT5C3A, PALB2, PFKM, PGK1, PIEZO1, PKLR, PRF1, PUS1, RACGAP1, RAD51, RAD51C, RBSN, REN, RFWD3, RHAG, RMRP, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RUNX1, SBDS, SEC23B, SH2B3, SH2D1A, SLC11A2, SLC19A1, SLC19A2, SLC25A38, SLC30A7, SLC40A1, SLC46A1, SLC2A1, SLC4A1, SLX4, SMAD4, SPTA1, SPTB, SRP72, STAT3, STEAP3, STIM1, TBXAS1, TCN2, TERC, TERT, TF, TFR2, THBD, THPO, TINF2, TMPRSS6, TOMM70, TP53, TPI1, TRNT1, TSR2, UMPS, UBE2T, VPS4A, WRAP53, XK, XRCC2, YARS2 Diamond-Blackfan anemia (DBA): 21 genes (11,1 kb) HEATR3, RPL5, RPL9, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPS7, RPS10, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, TSR2 Sideroblastic anemia (SIDBA): 10 genes (14,9 kb) ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2 Megaloblastic anemia: 13 genes (31,4 kb) AMN, CUBN, DHFR, FTCD, MMADHC, MTHFD1, MTR, MTRR, SLC19A1, SLC19A2, SLC46A1, TCN2, UMPS Congenital nonspherocytic hemolytic anemia (CNSHA): 10 genes (15,3 kb) AK1, G6PD, GATA1, GCLC, GPI, GSR, GSS, HK1, NT5C3A, PKLR Congenital dyserythropoietic anemia (CDAN): 6 genes (12,7 kb) CDAN1, CDIN1, KIF23, KLF1, RACGAP1, SEC23B Spherocytosis (SPH) and Elliptocytose (EL): 6 genes (26,6 kb) ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB Hereditary stomatocytosis: 8 genes (20,8 kb) ABCB6, ABCG5, ABCG8, KCNN4, PIEZO1, RHAG, SLC2A1, SLC4A1	4 - 6 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
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
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
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
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		
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, MPIG6B, 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
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
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

Diseases/Diagnostics	TAT	Material
Hematological Diseases		
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 Thrombotic microangiopathy (TMA): 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 Thrombotic thrombocytopenic purpura (TTP): 1 gene (4,3 kb) ADAMST13 Atypical hemolytic uremic syndrome (AHUS): 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 Autoinflammatoy 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, NLRC4, 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, NLRC4, 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, 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
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
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, CHRNG, 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	4 - 6 w	E
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
Periodic fever syndromes (HPF) * Gene panel: ID088.04,12 genes (26,3 kb) ELANE, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PSTPIP1, RIPK1, TNFRSF1A	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Immunological Diseases		
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, CTNNBL1, 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 immundeficiency (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 Immundeficinecy with hyper-IgM (HIGM): 4 genes (3,2 kb) AICDA, CD40, CD40LG, UNG	3 - 5 w	E
Primary immundeficiency, comprehensive diagnostics * Gene panel: ID380.01, 435 genes (930,7 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, 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, CTNNBL1, 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, 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, MPEG1, MPO, MRTFA, MS4A1, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, 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, RELA, RELB, RFX5, RFXANK, RFXAP, RHBDF2, RHOH, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SASH3, SBDS, SCGN, SEC61A1, SERPING1, SGPL1, SH2D1A, SH3KBP1, SKIC2, SKIC3, SLC19A1, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SLC9A3, SLCO2A1, SMARCAL1, SMARCD2, 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, TGFBR1, TGFBR2, 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, ZNFX1	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
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
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

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
Alport syndrome (ATS) *	3 - 5 w	E
Gene panel: ID099.00, 4 genes (21,0 kb)		
COL4A3, COL4A4, COL4A5, MYH9		
Aminoaciduria *	3 - 5 w	E
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		
Atypical hemolytic uremic syndrome (AHUS) *	3 - 5 w	E
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		
Bardet-Biedl syndrome (BBS) *	3 - 5 w	E
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		
Bartter syndrome (BARTS) *	3 - 5 w	E
Gene panel: ID156.01, 8 genes (17,7 kb)		
BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3		
Branchiootorenal syndrome (BOR) *	3 - 5 w	E
Gene panel: ID315.00, 5 genes (10,2 kb)		
EYA1, SALL1, SIX1, SIX5, TFAP2A		
Congenital anomalies of kidney and urinary tract (CAKUT) *	4 - 6 w	E
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		
Branchiootorenal 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		
Cystic kidney diseases, comprehensive diagnostics *	4 - 6 w	E
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		
Cystinosis (CTNS) and similar metabolic disorders *	3 - 5 w	E
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		
Diabetes insipidus *	3 - 5 w	E
Gene panel: ID322.00, 5 genes (8,4 kb)		
AQP2, AVP, AVPR2, SLC12A1, WFS1		

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: ID335.01, 30 genes (97,7 kb) AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CHEK2, CTR9, DICER1, DIS3L2, FBXW7, FIBP, GPC3, GPC4, KDM3B, NSD1, NYNRIN, MLH1, MSH2, MSH6, PALB2, PMS2, POU6F2, REST, TP53, 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 kp) 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 Thrombotic microangiopathy (TMA): 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 Thrombotic thrombocytopenic purpura (TTP): 1 gene (4,3 kb) ADAMST13 Atypical hemolytic uremic syndrome (AHUS): 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

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
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
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, CIDEC, CREB3L3, CYP27A1, CYP7A1, GCKR, GK, GPD1, GPIHBP1, LDLR, LDLRAP1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPL, NPC1L1, PCSK9, PLIN1, PNPLA5, POLD1, PPARG, SORT1, ZMPSTE24 Hypercholesterinemia: 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, CIDEC, 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, CIDEC, 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, CIDEC, LIPE, LMNA, PLIN1, POLD1, PPARG, ZMPSTE2	3 - 5 w	E
Lipodystrophy (CGL, FPLD)* Gene panel: ID055.04 Lypodystrophy (CGL, FPLD): 15 genes (22,0 kb) ADRA2A, AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEC, 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, CIDEC, 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, CIDEC, 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, MTTP, 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, CIDEC, CREB3L3, CYP27A1, CYP7A1, DHCR7, DHCR24, GCKR, GK, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIMA1, LIPA, LIPC, LIPE, LIPG, LMF1, LMNA, LPA, LPL, MTTP, 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, SLCO1B1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
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 localizes cutaneous amyloidosis (PLCA): 3 genes (7,0 kb) GPNMB, OSMR, IL31RA Cerebral amyloid angiopathy (HCHWA): 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, ETFA, ETFB, PDSS1, PDSS2	3 - 5 w	E
Combined oxidative phosphorylation deficiency (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, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 w	E
Congenital disorder of glycosylation (CDG) * Gene panel: ID035.03 Congenital disorder of glycolysation (CDG): 58 Gene (86,2 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, GET4, MAGT1, MAN1B1, MAN2B2, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, STX5, TMEM165, TUSC3, VMA12, VMA22 Congenital disorder of glycosylation, type I (CDG1): 31 genes (43,6 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAN2B2, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Congenital disorder of glycosylation, type II (CDG2):: 27 genes (42,7 kb) ATP6AP1, ATP6AP2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, GET4, MAN1B1, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, STX5, TMEM165, VMA12, VMA22	3 - 5 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
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

Diseases/Diagnostics	TAT	Material
Metabolic Diseases		
Hyperphosphatasia with impaired intellectual development syndrome (HPMRS) *	2 - 4 w	E
Gene panel: ID292.00, 6 genes (8,2 kb)		
PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY		
Hypoglycemia, hyperinsulinism and ketone metabolism *	3 - 5 w	E
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		
Hypomagnesemia (HOMG)*	3 - 5 w	E
Gene panel: ID054.02, 14 genes (30,9 kb)		
ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B, KCNA1, KCNJ10, RRAGD, SLC12A3, TRPM6		
Hypophosphatasia, hypophosphatemia and rickets *	3 - 5 w	E
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		
Matury-onset diabetes of the young (MODY) *	3 - 5 w	E
Gene panel: ID048.01, 14 genes (22,9 kb)		
ABCC8, APPL1, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1		
Metabolic disorder with epilepsy in childhood and adolescence *	3 - 5 w	E
Gene panel: ID172.00, 15 genes (28,1 kb)		
ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA1, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2		
Metabolic disorder with epilepy in infancy and childhood *	3 - 5 w	E
Gene panel: ID171.00, 18 genes (31,5 kb)		
ALDH5A1, ATP7A, BTD, FOLR1, GAMT, GATM, HEXA, HEXB, HLCS, KCTD7, MTHFR, PHGDH, POLG, PPT1, SLC19A3, SLC2A1, SLC6A8, TPP1		
Metabolic disorder with neonatal epilepsy *	3 - 5 w	E
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		
Metabolic epilepsy *	4 - 6 w	E
Gene panel: ID303.01		
Metabolic epilepsy: 84 genes (122,3 kb)		
ABAT, ACY1, ADSL, ALDH4A1, ALDH5A1, ALDH7A1, AMT, ARG1, ATIC, ATP7A, BCKDHA, BCKDHB, BCKDK, BTD, 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		
Mucopolysaccharidosis (MPS) *	3 - 5 w	E
Gene panel: ID308.00, 12 genes (21,2 kb)		
ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, VPS33A		

Diseases/Diagnostics	TAT	Material
Metabolic Diseases		
Muscular dystrophy-dystroglycanopathy (MDDG) *	3 - 5 w	E
Gene panel: ID179.00		
Muscular dystrophy-dystroglykanopathy (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		
Neonatal diabetes mellitus *	3 - 5 w	E
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		
Neuronal ceroid lipofuscinosis (CLN) *	3 - 5 w	E
Gene panel: ID132.01, 15 genes (20,2 kb)		
ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1		
Peroxisome biogenesis disorder (PBD) *	3 - 5 w	E
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		
Porphyria *	3 - 5 w	E
Gene panel: ID153.01, 10 genes (12,8 kb)		
ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS		
Pulmonary surfactant metabolism dysfunction (SMDP) *	3 - 5 w	E
Gene panel: ID168.01, 6 genes (12,0 kb)		
ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB		
Pyruvate dehydrogenase deficiency (PDHD)	3 - 5 w	E
Gene panel: ID393.00, 7 genes (10,0 kb)		
DLAT, DLD, LIAS, PDHA1, PDHB, PDHX, PDP1		
Renal amyloidosis *	3 - 5 w	E
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		
Storage diseases with cardiac involvement *	3 - 5 w	E
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		
Zellweger syndrome (ZWS) *	3 - 5 w	E
Gene panel: ID084.00, 14 genes (22,3 kb)		
ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26		
3-Methylglutaconaciduria (MGCA) *	3 - 5 w	E
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		

Diseases/Diagnostics	TAT	Material
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
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, TMCO1, 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, sPRED2 Intellectual developmental disordes 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, TMCO1, 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, COPB1, COPB2, 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, COPB2, 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, COPB1, CTNNB1, DYNC1I2, EXOC8, GEMIN4, GPT2, MFSD2A, MTHFS, NARS1, PLAA, PRUNE1, PUS3, SPOP, STAG2, TMX2, TRAPPC6B, SARS1, SMPD4, SVBP, VARS1	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Microcephaly and Macrocephaly		
Hydrocephalus * Gene panel: ID221.04 Hydrocephalus: 20 genes (57,9 kb) AKT3, CCDC88C, CCND2, CFAP43, FANCB, FLVCR2, FOXJ1, GPSM2, HYLS1, 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, HYLS1, KIF7, L1CAM, P4HB, PIK3R2, POMT1, SEC24D, ZIC3	3 - 5 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, TMCO1, 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, TMCO1, 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
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
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
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	4 - 6 w	E
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

Diseases/Diagnostics	TAT	Material
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	3 - 5 w	E
Combined oxidative phosphorylation deficiency (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, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 w	E
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, UQCRFS1, UQCRQ Mitochondrial complex I deficiency, nuclear type (MC1DN): 35 genes (27,4 kb) ACAD9, FOXRED1, 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, TIMMDC1, TMEM126B Mitochondrial complex II deficiency, nuclear type (MC2DN): 4 genes (3,7 kb) SDHA, SDHAF1, SDHB, SDHD Mitochondrial complex III deficiency, nuclear type (MC3DN): 10 genes (7,1 kb) BCS1L, CYC1, LYRM7, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, UQCRFS1 Mitochondrial complex IV deficiency, nuclear type (MC4DN): 21 genes (14,7 kb) COA3, COA5, COA6, COA8, COX4I1, COX6A2, COX6B1, COX8A, COX10, COX14, COX15, COX20, COX5A, COXFA4, LRPPRC, PET100, PET117, SCO1, SCO2, SURF1, TACO1 Mitochondrial complex V deficiency, nuclear type (MC5DN): 6 genes (4,2 kb) ATP5F1A, ATP5F1D, ATP5F1E, ATP5MD, ATPAF2, TMEM70	3 - 5 w	E
Leber hereditary optic neuropathy (LHON) # 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
MELAS syndrome # 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, DNM1L, 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, MMAB, 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, 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, UQCRFS1, 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 # 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

Diseases/Diagnostics	TAT	Material
Mitochondrial Diseases		
Mitochondrial myopathy / encephalopathy # 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 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
Pyruvate dehydrogenase deficiency (PDHD) Gene panel: ID393.00, 7 genes (10,0 kb) DLAT, DLD, LIAS, PDHA1, PDHB, PDHX, PDP1	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, WDPCP	3 - 5 w	E
CHARGE syndrome * Gene panel: ID307.00, 3 genes (12,9 kb) CHD7, SEMA3E, TBX22	3 - 5 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
Combined oxidative phosphorylation deficiency (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, QRSL1, PNPT1, POLRMT, PRORP, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TAMM41, TEFM, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 w	E
Congenital disorder of glycosylation (CDG) * Gene panel: ID035.03 Congenital disorder of glycolysation (CDG): 58 Gene (86,2 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, GET4, MAGT1, MAN1B1, MAN2B2, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, STX5, TMEM165, TUSC3, VMA12, VMA22 Congenital disorder of glycosylation, type I (CDG1): 31 genes (43,6 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DHRSX, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAN2B2, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Congenital disorder of glycosylation, type II (CDG2):: 27 genes (42,7 kb) ATP6AP1, ATP6AP2, B4GALT1, CAMLG, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, GET4, MAN1B1, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, STX5, TMEM165, VMA12, VMA22	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

Diseases/Diagnostics	TAT	Material
Multisystemic Malformation Syndromes		
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, TNNI2, 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
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
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
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

Diseases/Diagnostics	TAT	Material
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, DNM2, 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, DNM2, 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
Congenital myasthenic syndrome (CMS) * Gene panel: ID130.00 Congenital myasthenic syndrome (CMS): 25 genes (55,9 kb) AGRN, ALG2, ALG14, CHAT, CHRNA1, CHRNB1, CHRND, 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, CHRNB1, CHRND, 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
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

Diseases/Diagnostics	TAT	Material
Muscular Diseases		
Charcot-Marie-Tooth disease, axonal type (CMT2, HMSN) *	4 - 6 w	E
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		
Charcot-Marie-Tooth disease, demyelinating type (CMT1, CMT4, HMSN) *	3 - 5 w	E
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 gemes (6,3 kb)		
EGR2, MPZ, CNTNAP1		
Centronuclear myopathy (CNM) *	3 - 5 w	E
Gene panel: ID257.00, 7 genes (21,4 kb)		
CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1, SPEG		
Distal arthrogryposis (DA) *	3 - 5 w	E
Gene panel: ID196.01, 11 genes (40,5 kb)		
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Distal hereditary motor neuronopathy (HMND, HMNR) *	3 - 5 w	E
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		
Distal myopathy (MPD) *	4 - 6 w	E
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		
Limb-girdle muscular dystrophy (LGMD) *	4 - 6 w	E
Gene panel: ID122.03		
Limb-girdle muscular dystrophy (LGMD): 37 genes (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		

Diseases/Diagnostics	TAT	Material
Muscular Diseases		
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
Emery-Dreifuss muscular dystrophy (EDMD) * Gene panel: ID121.00, 6 genes (51,8 kb) EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43	3 - 5 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, ORAI1, 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, SCO2, 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
Muscular dystrophy-dystroglycanopathy (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
Muscular dystrophy, type Duchenne or Becker (DMD, BMD) #,* Gene panel: ID256.00, 1 gene (11,1 kb) DMD	2 - 4 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
Myotonia * Gene panel: ID255.00, 5 genes (25,0 kb) ATP2A1, CLCN1, HINT1, HSPG2, SCN4A	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

Diseases/Diagnostics	TAT	Material
Muscular Diseases		
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, 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	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
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

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
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 gemes (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
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
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
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

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Episodic pain syndrome (FEPS) *	3 - 5 w	E
Gene panel: ID268.00, 4 genes (20,5 kb)		
SCN9A, SCN10A, SCN11A, TRPA1		
Essential tremor (ETM) *	3 - 5 w	E
Gene panel: ID195.01, 4 genes (16,6 kb)		
DRD3, FUS, SCN4A, TENM4		
Frontotemporal dementia (FTD) *	3 - 5 w	E
Gene panel: ID310.01, 19 genes (29,3 kb)		
C9ORF72, CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP		
Hereditary ataxia, comprehensive diagnostics *	4 - 6 w	E
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, MTTP, 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		
Hereditary neuropathy (HMSN, HMN, HSAN), comprehensive diagnostics*	4 - 6 w	E
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, 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, 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		

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Hereditary sensory and autonomic neuropathy (HSAN, HSN) *	3 - 5 w	E
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		
Hyperekplexia (HKPX) *	3 - 5 w	E
Gene panel: ID216.00, 9 genes (16,0 kb)		
ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1		
Hypomyelinating leukodystrophy (HLD) *	3 - 5 w	E
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		
Leukodystrophy and Leukoencephalopathy, comprehensive diagnostics *	4 - 6 w	E
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		
Neurodegeneration with brain iron accumulation (NBIA) *	3 - 5 w	E
Gene panel: ID264.00, 11 genes (20,0 kb)		
ATP13A2, C19ORF12, CP, COASY, CRAT, FA2H, FTL, PANK2, PLA2G6, REPS1, WDR45		
Neurodevelopmental disorders (NED), comprehensive diagnostics *	4 - 6 w	E
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, EXOC2, EXOC7, EXOC8, FBXW11, FDFT1, 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, LNPK, 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, RALGAPA1, 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, TNR, TRAPPC10, TRAPPC4, TRAPPC6B, TRIM8, TRPM3, TTC5, UBE3C, UBE4A, UFC1, VAMP2, VARS1, VPS41, VPS50, WARS1, WARS2, WASF1, WDR45B, ZMIZ1, ZMYM2, ZNF142, ZNF526, ZNF668, ZSWIM6		
Neuronal ceroid lipofuscinosis (CLN) *	3 - 5 w	E
Gene panel: ID132.01, 15 genes (20,2 kb)		
ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1		
Parkinson disease (PARK) *	3 - 5 w	E
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		
Paroxysmal dyskinesia *	3 - 5 w	E
Gene panel: ID286.00, 14 genes (38,9 kb)		
ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24		

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
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
Septooptic dysplasia * Gene panel: ID378.00, 8 genes (10,8 kb) GLI2, HESX1, OTX2, PAX6, PROP1, SOX2, SOX3, TAX1BP3	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, NPTX1, 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) *	3 - 5 w	E
Gene panel: ID185.01, 5 genes (9,7 kb)		
LBX1, MECP2, MYO1H, PHOX2B, RET		
Cystic fibrosis (CF) *	2 - 4 w	E
Gene panel: ID045.00, 1 gene (4,4 kb)		
CFTR		
Interstitial lung disease and pulmonary fibrosis *	3 - 5 w	E
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		
Lung cancer *	4 - 6 w	E
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		
Pulmonary hypertension (PAH) *	3 - 5 w	E
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		
Pulmonary surfactant metabolism dysfunction (SMDP) *	3 - 5 w	E
Gene panel: ID168.01, 6 genes (12,0 kb)		
ABCA3, NKX2-1, SFTPB, SFTPC, CSF2RA, CSF2RB		
Primary ciliar dyskinesia with or without situs inversus (PCD, CILD) *	4 - 6 w	E
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		
Reproductive Disorders		
Adrenogenital syndrome (AGS, CAH) *	3 - 5 w	E
Gene panel: ID111.02, 7 genes (10,1 kb)		
CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR		
Azoospermia and cryptozoospermia *	4 - 6 w	E
Gene panel: ID391.00, 60 genes (141,6 kb)		
ADA2, ADGRG2, AR, C14ORF39, CATIP, CATSPER1, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, FANCM, FKBP6, GCNA, HFM1, KASH5, KCTD19, KLHL10, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PMFBP1, PNLDC1, RAD21L1, RBBP7, RNF212, RPL10L, SHOC1, SOHLH1, SPATA22, SPINK2, SPO11, STAG3, STRA8, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TERB1, TERB2, TEX11, TEX14, TEX15, USP9Y, XRCC2, ZMYND15, ZSWIM7		
Disorder of sex development (DSD) *	4 - 6 w	E
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, NR0B1, NR2F2, NR5A1,, POR, PPP1R12A, PPP2R3C, RSPO1, 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, NR0B1, NR5A1, SOX8, SOX9, SRD5A2, SRY, WT1, ZFPM2		
46,XX Disorder of sex development (SRXX), nonsyndromic: 6 genes (6,6 kb)		
NR2F2, NR5A1, RSPO1, 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, NR0B1, POR, PPP1R12A, PPP2R3C, RSPO1, 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		

Diseases/Diagnostics	TAT	Material
Reproductive Disorders		
Cystic fibrosis (CF) * Gene panel: ID045.00, 1 gene (4,4 kb) CFTR	2 - 4 w	E
Female infertility, omprehensive diagnostics * Gene panel: ID389.00 Female infertility, omprehensive diagnostics: 80 genes (156,6 kb) ASTL, BMP15, BNC1, BTG4, C14ORF39, CDC20, CHEK1, CLPP, CYP11A1, CYP11B1, CYP17A1, CYP21A2, DAP3, DIAPH2, ERAL1, ERCC6, ESR2, FANCM, FBXO43, FIGLA, FMR1, FOXL2, FSHR, GDF9, HARS2, HFM1, HROB, HSD17B4, HSD3B2, HSF2BP, INHA, KASH5, KHDC3L, KPNA7, LARS2, LHCGR, MCM8, MCM9, MEI1, MEIOB, MGA, MOS, MRPS22, MSH4, MSH5, NHEJ1, NLRP2, NLRP5, NLRP7, NOBOX, NR5A1, NUP107, PABPC1L, PADI6, PANX1, PATL2, POF1B, POR, PSMC3IP, REC114, SOHLH1, SPATA22, SPIDR, STAG3, STAR, SYCE1, SYCP2L, TLE6, TOP6BL, TP63, TRIP13, TUBB8, TWNK, WEE2, XRCC2, ZFP36L2, ZP1, ZP2, ZP3, ZSWIM7 Primary and premature ovarian failure (POI, POF): 40 genes (88,5 kb) BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, ESR2, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HROB, HSF2BP, INHA, KASH5, LHCGR, MCM8, MCM9, MEIOB, MGA, MRPS22, MSH4, MSH5, NHEJ1, NOBOX, NR5A1, NUP107, POF1B, PSMC3IP, SOHLH1, SPATA22, SPIDR, STAG3, SYCE1, SYCP2L, TP63, XRCC2, ZSWIM7 Oocyte/zygote/embryo maturation arrest (OZEMA): 26 genes (46,2 kb) ASTL, BTG4, CDC20, CHEK1, FBXO43, KHDC3L, KPNA7, MEI1, MOS, NLRP2, NLRP5, NLRP7, PABPC1L, PADI6, PANX1, PATL2, REC114, TLE6, TOP6BL, TRIP13, TUBB8, WEE2, ZFP36L2, ZP1, ZP2, ZP3 Adrenogenital syndrome (AGS, CAH): 7 genes (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 w	E
Hypogonadotropic hypogonadism with or without anosmia (KAL, HH) * Gene panel: ID170.05, 40 genes (78,9 kb) ANOS1, CHD7, CPE, DMXL2, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, KLB, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3A, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 w	E
Male infertility, comprehensive diagnostics * Gene panel: ID192.06 Male infertility, comprehensive diagnostics: 126 genes (424,9 kb) ACR, ACTL7A, ACTL9, ADA2, ADGRG2, AK7, AK9, AKAP3, AR, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, CFTR, CLDN2, CT55, CYLC1, DDX3Y, DMC1, DMRT1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, HFM1, IFT74, IQCN, KASH5, KCNU1, KCTD19, KLHL10, LRRC23, M1AP, MCM8, MCM9, MCMDC2, MEI1, MEIOB, MLH3, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RAD21L1, RBBP7, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SPO11, SSX1, STAG3, STK33, STRA8, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Spermatogenic failure (SPGF): 107 genes (368,8 kb) ACR, ACTL7A, ACTL9, AK7, AK9, AKAP3, ARMC12, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC146, CCDC34, CCDC62, CCIN, CEP112, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, CT55, CYLC1, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH17, DNALI1, DNHD1, DPY19L2, DRC1, DZIP1, FANCM, FBXO43, FKBP6, FSIP2, GCNA, GGN, IFT74, IQCN, KASH5, KCNU1, KLHL10, LRRC23, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, NUP210L, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RBBP7, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPACA1, SPAG17, SPATA16, SPATA22, SPEF2, SPINK2, SSX1, STAG3, STK33, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TDRD9, TEKT3, TERB1, TERB2, TEX11, TEX14, TEX15, TSGA10, TTC21A, TTC29, USP9Y, USP26, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Obstructive azoospermia (CBAVD, OAZON): 3 genes (8,2 kb) ADGRG2, CFTR, CLDN2	4 - 6 w	E
Multiple morphologic abnormalities of the flagella (MMAF) * Gen-Panel: ID390.00, 37 Gene (213,9 kb) AK7, AKAP3, ARMC2, CCDC34, CCDC146, CFAP43, CFAP44, CFAP47, CFAP54, CFAP57, CFAP58, CFAP61, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, DNAH1, DNAH2, DNAH6, DNAH8, DNAH10, DNAH17, DNALI1, DNHD1, DRC1, DZIP1, FSIP2, IFT74, QRICH2, SPEF2, SSX1, STK33, TTC21A, TTC29, USP26, WDR19	4 - 6 w	E
Oocyte/zygote/embryo maturation arrest (OZEMA) * Gene panel: ID239.03, 26 genes (46,2 kb) ASTL, BTG4, CDC20, CHEK1, FBXO43, KHDC3L, KPNA7, MEI1, MOS, NLRP2, NLRP5, NLRP7, PABPC1L, PADI6, PANX1, PATL2, REC114, TLE6, TOP6BL, TRIP13, TUBB8, WEE2, ZFP36L2, ZP1, ZP2, ZP3	3 - 5 w	E
Ovarian dysgenesis (ODG) * Gene panel: ID293.02 Ovarian dysgenesis (ODG): 18 genes (30,9 kb) BMP15, CLPP, DAP3, ERAL1, ESR2, FSHR, HARS2, HROB, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 Gonadal dysgenesis, XX type (ODG): 11 genes (19,1 kb) BMP15, ESR2, FSHR,HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 Gonadal dysgenesis, XX type, with deafness (PRLTS): 7 genes (11,8 kb) CLPP, DAP3, ERAL1, HARS2, HSD17B4, LARS2, TWNK	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

Diseases/Diagnostics	TAT	Material
Reproductive Disorders		
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
Primary and premature ovarian failure (POI, POF) * Gene panel: ID078.06 Primary and premature ovarian failure (POI, POF): 40 genes (88,5 kb) BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, ESR2, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HROB, HSF2BP, INHA, KASH5, LHCGR, MCM8, MCM9, MEIOB, MGA, MRPS22, MSH4, MSH5, NHEJ1, NOBOX, NR5A1, NUP107, POF1B, PSMC3IP, SOHLH1, SPATA22, SPIDR, STAG3, SYCE1, SYCP2L, TP63, XRCC2, ZSWIM7 Premature ovarian failure (POF): 26 genes (65,3 kb) BNC1, C14ORF39, DIAPH2, ERCC6, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, KASH5, MCM8, MEIOB, MGA, MSH4, MSH5, NOBOX, NR5A1, POF1B, SPATA22, STAG3, SYCE1, SYCP2L, TP63, XRCC2 Ovarian dysgenesis (ODG): 11 genes (19,1 kb) BMP15, ESR2, FSHR, HROB, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7	3 - 5 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, DNM2, 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, DNM2, 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
Arachnodactyly * Gene pnel: ID124.00, 13 genes (35,9 kb) CHST14, CTSC, DSE, EFEMP2, FBN1, FBN2, SCARF2, SKI, TGFB2, TGFB3, TGFBR1, TGFBR2, 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, EFNB1, 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

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Distal arthrogryposis (DA) *	3 - 5 w	E
Gene panel: ID196.01, 11 genes (40,5 kb)		
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Fraser syndrome (FRASRS) *	3 - 5 w	E
Gene panel: ID317.00, 3 genes (24,8 kb)		
FRAS1, FREM2, GRIP1		
Frontonasal dysplasia (FND) *	3 - 5 w	E
Gene panel: ID339.00, 11 genes (22,1 kb)		
ALX1, ALX3, ALX4, ANKH, EFNB1, FGFR1, FGFR2, FGFR3, GLI3, TWIST1, ZSWIM6		
Hand malformations, comprehensive diagnostics *	4 - 6 w	E
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, EFNB1, 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, WDPCP, 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		
Lacrimoauriculodentodigital 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		
Heart-hand syndrome *	3 - 5 w	E
Gene panel: ID165.01, 9 genes (23,8 kb)		
DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5		
Hypophosphatasia, hypophosphatemia and rickets *	3 - 5 w	E
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		
Klippel-Feil syndrome (KFS) *	3 - 5 w	E
Gene panel: ID207.00, 5 genes (12,5 kb)		
GDF3, GDF6, MEOX1, MYO18B, PAX1		
Lethal congenital contracture syndrome (LCCS) *	3 - 5 w	E
Gene panel: ID197.00, 12 genes (34,6 kb)		
ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42		
Mandibulofacial dysostosis (MFD) *	3 - 5 w	E
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		
Multiple epiphyseal dysplasia (EDM) *	3 - 5 w	E
Gene panel: ID202.01, 10 genes (25,9 kb)		
CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, EIF2AK3, KIF7, MATN3, SLC26A2		
Multiple pterygium syndrome *	3 - 5 w	E
Gene panel: ID158.01, 8 genes (16,8 kb)		
CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4		

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Orofacial cleft (OFC)*	4 - 6 w	E
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, SCNM1, 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, SCNM1, 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		
Orofaciodigital syndrome (OFD) *	3 - 5 w	E
Gene panel: ID265.00, 9 genes (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Osteogenesis imperfecta (OI) *	3 - 5 w	E
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		
Osteopetrosis (OPT) and similar bone diseases *	3 - 5 w	E
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, SLCO2A1, 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, SLCO2A1, SOST, TBXAS1, TGFB1, TNFRSF11B, TYROBP		
Osteoporosis *	3 - 5 w	E
Gene panel: ID115.02, 15 Gene (39,0 kb)		
CALCR, COPB2, COL1A1, COL1A2, ESR1, LGR4, LRP5, NOTCH2, PLS3, NHERF1, SGMS2, SLC34A1, VDR, WNT1, WNT11		
Pierre Robin sequence *	3 - 5 w	E
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		
Polydactyly, nonsyndromic type *	3 - 5 w	E
Gene panel: ID166.02, 9 genes (20,9 kb)		
CIBAR1, FBLN1, GLI1, GLI3, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141		
Rubinstein-Taybi syndrome (RSTS) *	3 - 5 w	E
Gene panel: ID142.01, 3 genes (24,3 kb)		
CREBBP, EP300, SRCAP		
Skeletal dysplasia, severe type *	4 - 6 w	E
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 (FBCG): 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		

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
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, ATP6V0A2, 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, KDELR2, 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, SMARCAL1, SMC1A, SMC3, SMOC1, SNRPB, SNX10, SOST, SOX9, SP7, SPARC, STT3A, SUMF1, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCIRG1, TCOF1, TCTN2, TCTN3, TENT5A, TERT, TGFB1, TGFB2, TGFBR2, TMCO1, 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
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
Spondylocostal dysostosis (SCDO) * Gene panel: ID227.00, 7 genes (14,4 kb) DLL3, FLNB, HES7, LFNG, MESP2, RIPPLY2, TBX6	3 - 5 w	E
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

Diseases/Diagnostics	TAT	Material
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, ODAPH, 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, ATP6V0A2, 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
Gorlin syndrome (BCNS) und similar syndromes * Gene panel: ID174.02, 8 genes (25,8 kb) BAP1, CYLD, ELP1, GPR161, NSD1, PTCH1, PTEN, SUFU	3 - 5 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
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, RSPO4, 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

Diseases/Diagnostics	TAT	Material
Skin and Dental Diseases		
Ectodermal dysplasia (ECTD) *	3 - 5 w	E
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 dysplasie (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 dysplasie (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		
Hereditary angioedema (HAE) *	3 - 5 w	E
Gene panel: ID345.00, 7 genes (16,4 kb)		
ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1		
Hermansky-Pudlak syndrome (HPS) *	3 - 5 w	E
Gene panel: ID289.00, 11 genes (22,6 kb)		
AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6		
Hydrops fetalis *	3 - 5 w	E
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, CHRNG, 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		
Hypotrichosis, nonsyndromic type (HYPT) *	3 - 5 w	E
Gene panel: ID146.01, 12 genes (20,1 kb)		
APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE		
Ichthyosis *	3 - 5 w	E
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		
Multiple pterygium syndrome *	3 - 5 w	E
Gene panel: ID158.01, 8 genes (16,8 kb)		
CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4		
Neurofibromatosis (NF) #,*	3 - 5 w	E
Gene panel: ID210.00, 3 genes (11,6 kb)		
NF1, NF2, SPRED1		
Oculocutaneous albinism (OCA) *	3 - 5 w	E
Gene panel: ID082.02, 9 genes (13,3 kb)		
DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1		
Orofaciodigital syndrome (OFD) *	3 - 5 w	E
Gene panel: ID265.00, 9 genes (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Palmoplantar keratoderma (PPK) *	3 - 5 w	E
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		
Erythrokeratodermia 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		

Diseases/Diagnostics	TAT	Material
Skin and Dental Diseases		
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
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, CCBE1, 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, NFLS): 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
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
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, RFWD3, SLX4, TOP3A, UBE2T, WRN, XPA, XPC, XRCC2	4 - 6 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
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

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
Colorectal cancer * 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
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, DNTT, 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, PARG, PARK7, PARP1, PARP2, PARP3, PARPBP, 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, RFWD3, 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
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 syndrome (BCNS) und similar syndromes * Gene panel: ID174.02, 8 genes (25,8 kb) BAP1, 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
Nephroblastoma and Wilms tumor (WT) * Gene panel: ID335.01, 30 genes (97,7 kb) AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CHEK2, CTR9, DICER1, DIS3L2, FBXW7, FIBP, GPC3, GPC4, KDM3B, NSD1, NYNRIN, MLH1, MSH2, MSH6, PALB2, PMS2, POU6F2, REST, TP53, 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

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
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, 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
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: ID089.04, 19 genes (59,9 kb) APC, ATM, BARD1, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL, WT1	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: ID387.01, 21 genes (47,3 kb) AIP, CDH23, CDKN1B, DICER1, EPCAM, GNAS, GPR101, MAX, MLH1, MSH2, MSH6, MEN1, PMS2, PRKAR1A, SDHA, SDHAF2, SDHB, SDHC, SDHD, RET, 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

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
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
Tuberous sclerosis (TSC) * Gene panel: ID332.00, 2 genes (8,9 kb) TSC1, TSC2	2 - 4 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, EHBP1, 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, RTEL1, 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, TGFBR2, 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
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
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 *	4 - 6 w	E
Gene panel: ID850.00, 1223 genes (3235,6 kb)		
AAAS, ABCA12, ABCC6, ABCC9, 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, AHI1, 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, ARFGEF2, ARHGAP29, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, ARMC9, ARSA, ARSB, ARSL, ARX, ASAH1, ASCC1, ASNS, ASPA, ASPM, ASS1, ASXL1, ATAD3A, ATIC, ATP1A2, ATP6V0A2, ATP7A, ATR, ATRX, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT7, B4GAT1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCL11A, BCOR, BCS1L, BFSP2, BGN, BHLHA9, BICD2, BIN1, BLM, BLTP1, BMP1, BMP2, BMP4, BMPER, BMPR1B, BNC2, BPNT2, BRAF, BRAT1, BRCA2, BRIP1, BRPF1, BSND, BTD, BUB1B, C2CD3, CA2, CA8, CACNA1C, CACNA1E, CACNA1G, CANT1, CASK, CASR, CBL, CC2D2A, CCBE1, 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, CRYBA1, 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, DNAL1, DNM1L, DNM2, DNMT3A, DNMT3B, DOCK6, DOK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DSP, DSTYK, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYNC2I1, 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, ERF, ESCO2, ETFA, ETFB, ETFDH, EVC, EVC2, EXOC3L2, EXOSC3, EXT1, EXT2, EXTL3, EYA1, EZH2, FAH, FAM111A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, 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, GALC, GALE, GALK1, GALNS, GALNT2, 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, GMPPB, GNAI3, GNAO1, GNAS, GNB1, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC3, GPC6, GPI, GPSM2, GREB1L, GRHL3, GRIN1, GRIN2B, GRIP1, GSC, GTF2H5, GTPBP3, GUCY2C, GUSB, GZF1, H1-4, HAAO, HADHA, HADHB, HBA1, HBA2, HCCS, HCFC1, HDAC8, HES7, HESX1, HIBCH, HIVEP2, HMGA2, HNF1B, HNF4A, HNRNPK, HOXA1, HOXA13, HOXD13, HPSE2, HR, HRAS, HSD17B3, HSD17B4, HSF4, HSPD1, HSPG2, HUWE1, HYCC1, HYLS1, 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, LIG4, LIPA, LMBR1, LMBRD1, LMNA, LMNB1, LMNB2, LMOD3, LMX1B, LONP1, LRP2, LRP4, LRP5, LRRC56, LTBP3, LTBP4, LYST, LZTFL1, LZTR1, MAB21L2, MACF1, 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, MST01, 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, NACC1, 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, OCRL, ODAD1, ODAD2, ODAD3, OFD1, OPHN1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTX2, P3H1, P4HB, PAFAH1B1, PAK3, PALB2, PAPSS2, PARN, PAX2, PAX3, PAX6, PAX7, PAX8, PBX1, PCGF2, PCNT, PCYT1A, PDCD10, PDE4D, PDGFRB, PDHA1, PEPD, 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, PLAG1, 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, PPIB, 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, RARB, RARS2, RASA1, RAX, RBBP8, RBM10, RBM8A, RBPJ, RECQL4, RELN, REN, RERE, RET, RFT1, RFX6, RIPK4, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNU4ATAC, ROBO1, ROBO3, ROGDI, ROR2, RPGRIP1L, RPL10, RPL11, RPL35A, RPL5, RPS10, RPS17, RPS19, RPS24, RPS26, RPS6KA3, RPS7, RRAS2, RRM2B, RSPH4A, RSPH9, RTEL1, RTTN, RUNX2, RXYLT1, RYR1, SALL1, SALL4, SAMD9, SAMHD1, SATB2, SBDS, SC5D, SCARF2, SCLT1, SCN1A, SCN2A, SCN4A, SCO2, SDCCAG8, SDR9C7, SEC23B, SEC24D, SEPSECS, SERPINF1, SERPINH1, SETBP1, SETD5, SF3B4, SGCG, SGPL1, SH3PXD2B, SHANK3, SHH, SHOC2, SHOX, SIK3, SIL1, SIX3, 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, TAFAZZIN, TALDO1, TAPT1, TBC1D20, TBC1D23, TBC1D24, TBC1D32, TBCD, TBCE, TBCK, TBL1XR1, TBX1, TBX15, TBX18, TBX20, TBX3, TBX4, TBX5, TBX6, TCF12, TCF4, TCIRG1, TCOF1, TCTN1, TCTN2, TCTN3, TELO2, TENM3, TENT5A, TFAP2A, TFAP2B, TGDS, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM1, THOC6, THRA, TINF2, TMCO1, TMEM107, TMEM138, TMEM165, TMEM216, TMEM231, TMEM237, TMEM38B, TMEM67, TMEM94, TMEM98, TMX2, TNNI2, TNNT1, TNNT3, TOE1, TOP3A, TOR1A, TP63, TPM2, TPM3, TRAF3IP1, TRAF7, TRAIP, TRAP1, 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, UMPS, UROS, USP18, USP9X, VAMP1, VEGFC, VIPAS39, VLDLR, VPS13B, VPS33B, VPS53, VRK1, VSX2, WDPCP, 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		

Diseases/Diagnostics	TAT	Material
Preventive Diagnostics: Carrier Screening		
Carrier-Screening *	4 - 6 w	E
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, AGTR1, AGXT, AHI1, AIRE, AKR1D1, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, ALS2, AMACR, AMH, AMHR2, AMT, ANK1, ANO10, ANTXR2, APTX, AQP2, ARG1, ARSA, ARSB, ARSL, ARX, ASL, ASNS, ASPA, ASS1, ATIC, ATM, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AVPR2, B4GALT1, BBS1, BBS10, BBS12, BBS2, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BRIP1, BSND, BTD, BTK, CA2, CAPN3, CASR, CBS, CC2D2A, CCDC88C, CD3D, CD3E, CD40LG, CDH23, CEP290, CERKL, CFP, CFTR, CHM, CHRNA1, CHRND, CHRNE, CHRNG, 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, CTSC, 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, FKRP, FKTN, FLNA, FMO3, FOLR1, FOXN1, FOXP3, FRAS1, FREM2, FUCA1, FXN, G6PC1, G6PC3, G6PD, GAA, GALC, 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, HYLS1, 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, LHCGR, LHX3, LIFR, LIPA, LMBRD1, LMNA, LOXHD1, LPL, LRP2, LRP5, LRPPRC, LYST, MAN2B1, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MED12, MED17, MEFV, MESP2, MFSD8, MGAT2, MID1, MKS1, MLC1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, 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, NR0B1, NR2E3, NTRK1, NUP62, OAT, OCA2, OCRL, 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, PMM2, 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, RAPSN, RARS2, RB1, RDH12, RELN, REN, RLBP1, RNASEH2B, RPE65, RPGR, RPGRIP1L, RS1, RTEL1, SACS, SAMHD1, SBDS, SC5D, SCN2A, SCN8A, SCNN1A, SCNN1B, SCNN1G, SCO2, 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, SMARCAL1, 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, UQCRB, UQCRQ, UROS, USH1C, USH1G, USH2A, VDR, VLDLR, VPS13A, VPS13B, VPS33B, VPS45, VPS53, VRK1, VSX2, WAS, WNT10A, WNT3, WNT7A, WRN, WT1, XPA, XPC, ZIC3, ZMPSTE24, ZNF469		

Whole Exome Sequencing (WES)		
Analyses		
WES solo * (Affected patient)	4 - 12 w	E
WES trio * (Affected patient and parents)	4 - 12 w	E

Notes		
***** = accredited test procedure HG Mannheim and HG München		
# = accredited test procedure HG Freiburg		