SYNLAB Human Genetics

Status: 12/2023

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 based on a spike-in Twist whole exome capture library analysed on illumina sequencers.

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)
[no] 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.
For prenatal samples please contact us before sending to allow better tracking of the samples.
Samples send in germany can be sent directly to us.

Index

Disease Types

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: ID271.01, 13 genes (47,1 kb) APC2, CAMSAP1, CTNNA2, DYNC1H1, KIF2A, KIF5C, KIF26A, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1	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, TOGAGRAM1, 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
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		
Arrhythmogenic right ventricular cardiomyopathy (ARVD, ARVC) *	4 - 6 w	E
Gene panel: ID010.02, 15 genes (143,9 kb)		
DH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, PLN, LMNA, PKP2, PRKAG2, TGFB3, TMEM43, 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
Gene 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		
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		
Cardiomyopathy, comprehensive diagnostics *	4 - 6 w	E
Gene panel: ID027.02		
Cardiomyopathy, comprehensive diagnostics: 132 genes (456,7 kb)		
ABCC9, ACTA1, ACTC1, ACTN2, ALMS1, ALPK3, ANKRD1, BAG3, BAG5, BRAF, CALR3, CASQ2, CAV3, CAVIN4, CBL, CDH2, COA5, COA6, COX15, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, EMD, EYA4, FHL1, FHL2, FHOD3, FKRP, FKTN, FLNC, FXN, GAA, GATAD1, GET3, GLA, HADHA, HCN4, HFE, HRAS, ILK, JPH2, JUP, KIF20A, KLF10, KLHL24, KRAS, LAMA2, LAMA4, LAMP2, LDB3, LIMS2, LMNA, LMOD2, LZTR1, MAP2K1, MAP2K2, MAPK1, MCM10, MIB1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYO6, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NPPA, NRAS, OBSCN, PDLIM3, PKP2, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, PTPN11, RAF1, RBM20, RIT1, RPL3L, RRAS2, RYR2, SCN2B, SCN5A, SCO2, SDHA, SGCB, SGCD, SGCG, SHOC2, SLC25A4, SOS1, SOS2, SPRED2, SYNE1, SYNE2, TAFAZZIN, TBX20, TCAP, TGFB3, TMEM43, TMPO, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TRPM4, TTN, TTR, TXNRD2, VCL, VEZF1		
Dilatated cardiomyopathy (DCM, CMD): 63 genes (287,5 kb)		
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CRYAB, CSRP3, DES, DMD, DNAJC19, DSG2, DSP, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GATAD1, GET3, HFE, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEBL, NEXN, NKX2-5, PDLIM3, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, SYNE2, TAFAZZIN, TBX20, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL, VEZF1		
Hypertrophic cardiomyopathy (HCM, CMH): 56 genes (209,1 kb)		
ACTC1, ACTN2, ALPK3, ANKRD1, BRAF, CALR3, CAV3, CSRP3, DES, FHL1, FHOD3, FLNC, GAA, GLA, HRAS, JPH2, KLF10, KLHL24, KRAS, LAMP2, LDB3, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NRAS, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RIT1, RRAS2, SOS1, SOS2, SPRED2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL		
Restrictive cardiomyopathy (RCM): 14 genes (31,4 kb)		
ACTC1, BAG3, DES, FLNC, KIF20A, MCM10, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTR		
Arrhythmogenic right ventricular cardiomyopathy (ARVD, ARVC): 15 genes (150,6 kb)		
CDH2, CTNNA3, DES, DSC2, DSG2, DSP, JUP, LMNA, PKP2, PLN, PRKAG2, RYR2, TGFB3, TMEM43, TTN		
Left ventricular noncompaction cardiomyopathy (LVNC, NCCM): 18 genes (169,0 kb)		
ACTC1, ACTN2, DMD, DTNA, HCN4, LDB3, LMNA, MIB1, MYBPC3, MYH7, NKX2-5, PRDM16, RYR2, SCN5A, TAFAZZIN, TNNT2, TPM1, TTN		

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Catecholaminergic polymorphic ventricular tachycardia (CPVT) *	3 - 5 w	E
Gene panel: ID012.03, 9 genes (33,9 kb)		
ANK2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN		
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		
Congenital heart defects, comprehensive diagnostics *	4 - 6 w	E
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		
Congenital isolated heart defects *	4 - 6 w	E
Gene panel: ID017.04		
Congenital isolated 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		

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Congenital syndromic heart defects * Gene panel: ID252.02, 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 Weill-Marchesani syndrome (WMS): 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2 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 syndrome (ALGS): 2 genes (11,1 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
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
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
CADASIL and CARASIL #,* Gene panel: ID167.01, 3 genes (9,4 kb) HTRA1, NOTCH3, TREX1	3 - 5 w	E
CHARGE syndrome * Gene panel: ID307.00, 3 genes (12,9 kb) CHD7, SEMA3E, TBX22	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		
Dilated cardiomyopathy (CMD, DCM) *	4 - 6 w	E
Gene panel: ID008.03		
Dilated cardiomyopathy (CMD, DCM): 63 genes (287,5 kb)		
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BAG5, CRYAB, CSRP3, DES, DMD, DNAJC19, DSG2, DSP, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GATAD1, GET3, HFE, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, LMOD2, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEBL, NEXN, NKX2-5, PDLIM3, PLN, PPCS, PRDM16, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, SYNE1, SYNE2, TAFAZZIN, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TBX20, TTN, VCL, VEZF1		
Dilated cardiomyopathy, nonsyndromic, dominant (CMD1): 42 genes (201,2 kb)		
ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, CRYAB, CSRP3, DES, DSG2, EYA4, FKTN, FLNC, LAMA4, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYPN, NEBL, NEXN, NKX2-5, PDLIM3, PLN, PRDM16, PSEN1, PSEN2, RAF1, RBM20, SCN5A, SDHA, SGCD, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TBX20, TTN, VCL, VEZF1		
Dilated cardiomyopathy, nonsyndromic, recessive (CMD2): 8 genes (9,7 kb)		
BAG5, GATAD1, GET3, JPH2, LMOD2, PPCS, RPL3L, TNNI3		
Dilated cardiomyopathy, nonsyndromic, X-linked (CMD3): 2 genes (11,9 kb)		
DMD, TAFAZZIN		
Dilated cardiomyopathy, syndromic: 18 genes (86,9 kb)		
DES, DMD, DNAJC19, DSP, EMD, FHL1, FKRP, HFE, JUP, LAMP2, LMNA, MYH7, MYL2, SYNE1, SYNE2, TAFAZZIN, TCAP, TMEM43		
Early repolarization syndrome (ERS) *	3 - 5 w	E
Gene panel: ID330.00, 12 genes (38,9 kb)		
ABCC9, CACNA1C, CACNA2D1, CACNB2, DPP6, GPD1L, KCND3, KCNE1, KCNH2, KCNJ8, SCN5A, SCN10A		
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		
Endocrine hypertension *	3 - 5 w	E
Gene panel: ID270.02		
Endocrine hypertension: 32 genes (80,9 kb)		
CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP11B2, CYP17A1, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL, WNK1, WNK4, YY1AP1		
Hyperaldosteronism (HALD): 6 genes (20,6 kb)		
CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5		
Pseudohypoaldosteronism type II (PHA2): 4 genes (15,8 kb)		
CUL3, KLHL3, WNK1, WNK4		
Pheochromocytoma/paraganglioma syndrome (PPGL): 9 genes (9,5 kb)		
MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, 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		
Familial atrial fibrillation (ATFB) *	3 - 5 w	E
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		
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		
Hereditary hemorrhagic telangiectasia (HHT) *	3 - 5 w	E
Gene panel: ID155.01, 7 genes (15,6 kb)		
ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4		
Hypertrophic cardiomyopathy (CMH, HCM) *	4 - 6 w	E
Gene panel: ID007.03		
Hypertrophic cardiomyopathy (HCM, CMH): 56 genes (209,1 kb)		
ACTC1, ACTN2, ALPK3, ANKRD1, BRAF, CALR3, CAV3, CSRP3, DES, FHL1, FHOD3, FLNC, GAA, GLA, HRAS, JPH2, KLF10, KLHL24, KRAS, LAMP2, LDB3, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, NRAS, PDLIM3, PLN, PRKAG2, PTPN11, RAF1, RRAS2, RIT1, SOS1, SOS2, SPRED2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, TTR, VCL		
Hypertrophic cardiomyopathy, nonsyndromic (CMH): 35 genes (176,6 kb)		
ACTC1, ACTN2, ALPK3, ANKRD1, CAV3, CSRP3, DES, FHL1, FHOD3, FLNC, JPH2, KLF10, KLHL24, LDB3, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, PDLIM3, PLN, PRKAG2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTN, VCL		
Hypertrophic cardiomyopathy, syndromic: 22 genes (33,6 kb)		
BRAF, CALR3, GAA, GLA, HRAS, KRAS, LAMP2, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NRAS, PRKAG2, PTPN11, RAF1, RRAS2, RIT1, SOS1, SOS2 , SPRED2, TTR		

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
Left ventricular noncompaction cardiomyopathy (LVNC, NCCM) *	4 - 6 w	E
Gene panel: ID011.02, 18 genes (169,0 kb)		
ACTC1, ACTN2, DMD, DTNA, HCN4, LDB3, LMNA, MIB1, MYBPC3, MYH7, NKX2-5, PRDM16, RYR2, SCN5A, TAFAZZIN, TNNT2, TPM1, TTN		
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		
Long QT syndrome (LQT) *	3 - 5 w	E
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		
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: ID194.03		
Marfan syndrome (MFS) and similar syndromes: 39 genes (134,2 kb)		
ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL11A1, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MTHFR, MYH11, MYLK, PLOD1, PRDM5, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469		
Marfan syndrome (MFS): 3 genes (11,8 kb)		
FBN1, TGFBR1, TGFBR2		
Thoracic aortic aneurysm (TAA/D): 13 genes (36,4 kb)		
ACTA2, COL3A1, FBN1, FOXE3, LOX, MYH11, MYLK, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2		
Ehlers-Danlos syndrome (EDS): 10 genes (39,1 kb)		
CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469		
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		
Multiple congenital heart defects (CHTD) *	3 - 5 w	E
Gene panel: ID302.00, 7 genes (14,4 kb)		
FLT4, GATA5, GDF1, NR2F2, PLD1, TAB2, ZIC3		
Muscular diseases with cardiac involvement *	4 - 6 w	E
Gene panel: ID123.02		
Muscular diseases with cardiac involvement: 34 genes (220,8 kb)		
BAG3, BVES, CAV3, CRYAB, DES, DMD, DPM3, EMD, FHL1, FKRP, FKTN, FLNC, JAG2, KY, LDB3, LAMA2, LIMS2, LMNA, MYL2, MYOT, POMT1, PYROXD1, SGCA, SGCB, SGCD, SGCG, SVIL, SYNE1, SYNE2, TCAP, TMEM43, TOR1AIP1, TTN, UNC45B		
Myofibrillar myopathy (MFM): 12 genes (130,7 kb)		
BAG3, CRYAB, DES, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B		
Limb-girdle muscular dystrophy (LGMD): 17 genes (131,7 kb)		
BVES, CAV3, DES, DPM3, FKRP, FKTN, JAG2, LAMA2, LIMS2, POMT1, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TTN		
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		
Noonan syndrome (NS) *	3 - 5 w	E
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		
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		

Diseases/Diagnostics	TAT	Material
Cardiovascular Diseases		
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: ID105.01, 14 genes (31,4 kb) ACTC1, BAG3, DES, FLNC, KIF20A, MCM10, MYH7, MYL2, MYL3, MYPN, TNNI3, TNNT2, TPM1, TTR	3 - 5 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: ID149.01, 13 genes (21,5 kb) ATP7B, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, PRKAG2, SLC40A1, TFR2, TTR	3 - 5 w	E
Sudden cardiac death * Gene panel: ID349.00 Sudden cardiac death: 110 genes (366,9 kb) ABCC9, ACTC1, ACTN2, AKAP9, ALG10B, ALPK3, ANK2, ANKRD1, BAG3, BAG5, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DPP6, DSC2, DSG2, DSP, EMD, EYA4, FGF12, FHL1, FHOD3, FKRP, FKTN, FLNC, GATAD1, GLA, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, NKX2-5, PKP2, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RANGRF, RBM20, RPL3L, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SDHA, SEMA3A, SGCD, SLC4A3, SLMAP, SNTA1, TBX5, TCAP, TECRL, TGFB3, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TTN, TTR, VCL Arrhythmia (BRGDA, LQT) and sudden cardiac death: 65 genes (268,2 kb) ABCC9, AKAP9, ALG10B, ANK2, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DES, DPP6, DSC2, DSG2, DSP, EMD, FGF12, GNB2, GPD1L, HCN4, JPH2, JUP, KCNA5, KCND2, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, LMNA, MYBPC3, MYH6, NKX2-5, PKP2, PLN, PRKAG2, RANGRF, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SEMA3A, SLC4A3, SLMAP, SNTA1, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TRDN, TRPM4, TTN Cardiomyopathy (HCM, DCM) and sudden cardiac death: 63 genes (247,7 kb) ABCC9, ACTC1, ACTN2, ALPK3, ANKRD1, BAG3, BAG5, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSG2, DSP, EMD, EYA4, FHL1, FHOD3, FKRP, FKTN, FLNC, GATAD1, GLA, JPH2, JUP, LAMA4, LAMP2, LDB3, LMNA, MYBPC3, MYH6, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEXN, NKX2-5, PLN, PPCS, PRDM16, PRKAG2, PSEN1, PSEN2, RAF1, RBM20, RPL3L, SCN5A, SDHA, SGCD, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRIM63, TTN, TTR, VCL	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

Diseases/Diagnostics	TAT	Material
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, TOGAGRAM1, 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: ID085.02, 42 genes (132,9 kb) ARMC4, CCDC103, CCDC39, CCDC40, CCDC65, CCDC114, CCDC151, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH5, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC6, LRRC56, MCIDAS, NEK10, NME8, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, TTC12, TTC25, ZMYND10	4 - 6 w	E
Senior-Loken syndrome (SLSN) * Gene panel: ID029.01, 8 genes (27,9 kb) CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19	3 - 5 w	E
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
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

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		
Hereditary hemorrhagic telangiectasia (HHT) *	3 - 5 w	E
Gene panel: ID155.01, 7 genes (15,6 kb)		
ACVRL1, BMPR2, ENG, EPHB4, GDF2, RASA1, SMAD4		
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: ID194.03		
Marfan syndrome (MFS) and similar syndromes: 39 genes (134,2 kb)		
ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS, CHST14, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL11A1, DSE, EFEMP2, FBN1, FBN2, FKBP14, FLNA, FOXE3, LOX, LTBP2, MED12, MTHFR, MYH11, MYLK, PLOD1, PRDM5, SKI, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, ZNF469		
Marfan syndrome (MFS): 3 genes (11,8 kb)		
FBN1, TGFBR1, TGFBR2		
Thoracic aortic aneurysm (TAA/D): 13 genes (36,4 kb)		
ACTA2, COL3A1, FBN1, FOXE3, LOX, MYH11, MYLK, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2		
Ehlers-Danlos syndrome (EDS): 10 genes (39,1 kb)		
CHST14, DSE, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PLOD1, PRDM5, ZNF469		
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		
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: ID076.03 Autism spectrum disorders: 106 genes (377,1 kb) ADNP, ADSL, AFF2, ALDH5A1, ARX, ARID1B, ASH1L, ASTN2, ATP1A1, AUTS2, BCL11A, CACNA1C, CASK, CC2D1A, CDKL5, CHD2, CHD7, CHD8, CNOT3, CNTN4, CNTNAP2, CTNNB1, CUL3, DEAF1, DHCR7, DLGAP2, DPP6, DYRK1A, EHMT1, EIF4E, FMR1, FOXG1, FOXP1, FOXP2, FRMPD4, GRIA3, GRIN2A, GRIN2B, HERC2, IL1RAPL1, IQSEC2, KDM5C, KMT2C, KTM5B, MAOA, MBD5, MECP2, MED12, MED13L, MEF2C, MYT1L, NAA15, NAGLU, NEXMIF, NHS, NLGN1, NLGN3, NLGN4X, NR1I3, NRXN1, NRXN2, NRXN3, OPHN1, PAH, PCDH19, PHF6, PHF8, PHF21A, POGZ, PQBP1, PTCHD1, PTEN, RAB39B, RAI1, RPL10, SCN1A, SCN2A, SEMA5A, SETD2, SGSH, SHANK2, SHANK3, SLC6A1, SLC6A8, SLC9A6, SLC9A9, SLC35A3, SMARCB1, SYN1, SYNGAP1, TBL1XR1, TBR1, TCF4, TLK2, TMLHE, TRRAP, TSC1, TSC2, UBE2A, UBE3A, UPF3B, VPS13B, ZEB2, ZIC1, ZMYND11, ZNF292 Susceptibility to autism (AUTS): 14 genes (36,5 kb) CHD8, CNTNAP2, EIF4E, MECP2, NLGN1, NLGN3, NLGN4X, PTCHD1, PTEN, RPL10, SHANK2, SLC9A9, TBR1, TMLHE	4 - 6 w	E
Coffin-Siris syndrome (CSS) * Gene panel: ID118.01, 10 genes (34,2 kb) ARID1A, ARID1B, ARID2, DPF2, SMARCC2, SMARCA4, SMARCB1, SMARCE1, SOX4, SOX11	3 - 5 w	E
Congenital disorder of glycosylation (CDG) * Gene panel ID035.02 Congenital disorder of glycolysation (CDG): 51 genes (74,7 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, MAGT1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3 Congenital disorder of glycolysation, type I (CDG1): 29 genes (39,6 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Congenital disorder of glycolysation, type II (CDG2): 22 genes (35,1 kb) ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, TMEM165, TMEM199	3 - 5 w	E
Cornelia de Lange syndrome (CDLS) * Gene panel: ID033.02, 8 genes (32,0 kb) ANKRD11, BRD4, HDAC8, NIPBL, RAD21, SMC1A, SMC3, SMS	3 - 5 w	E
Developmental language disorder (DLD, CAS) Gene panel: ID368.00 Developmental language disorder (DLD, CAS): 34 genes (119,7 kb) ATP2C2, BCL11A, BUD13, CDK13, CHD3, CNTNAP2, DDX3X, EBF3, ERC1, FOXP1, FOXP2, GALT, GNAO1, GNB1, GRIN2A, KAT6A, KANSL1, MEIS2, NFXL1, POGZ, PURA, SETBP1, SETD1A, SETD1B, SHANK3, SRCAP, SRPX2, TM4SF20, TNRC6B, UPF2, WDR5, 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

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
Intellectual developmental disorder, autosomal dominant (MRD) *	4 - 6 w	E
Gene panel: ID036.05		
Intellectual developmental disorder, autosomal dominant (MRD): 74 genes (258,2 kb)		
ADNP, AHDC1, AP2M1, ARID1A, ARID1B, ARID2, ASH1L, ATP2B1, AUTS2, BICRA, CACNG2, CAMK2A, CAMK2B, CAMK2G, CDH15, CERT1, CHAMP1, CIC, CLTC, CTCF, CTNNB1, DEAF1, DLG4, DPF2, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, GATAD2B, GNB1, GRIA1, GRIN1, GRIN2B, HIVEP2, KAT6A, KCNQ5, KDM4B, KIF1A, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MEF2C, MYT1L, NAA15, NUS1, PACS1, POGZ, PPP2R1A, PPP2R5D, PURA, RAC1, SET, SETBP1, SETD2, SETD5, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMARCD1, SOX4, SOX11, STAG1, SYNGAP1, TBL1XR1, TLK2, TRIO, TRIP12, ZBTB18, ZMYND11, ZNF292		
Intellectual developmental disorder, autosomal dominant, nonsyndromic (MRD): 57 genes (203,8 kb)		
AP2M1, ARID1A, ARID1B, ASH1L, ATP2B1, AUTS2, CACNG2, CAMK2A, CAMK2B, CAMK2G, CDH15, CERT1, CIC, CLTC, CTCF, DEAF1, DLG4, DPP6, DYNC1H1, DYRK1A, EEF1A2, EPB41L1, GATAD2B, GNB1, GRIA1, GRIN2B, HIVEP2, KCNQ5, KDM4B, KMT2B, KMT5B, LMAN2L, MBD5, MED13, MYT1L, NAA15, NUS1, PACS1, POGZ, PPP2R1A, PPP2R5D, RAC1, SET, SETBP1, SETD2, SETD5, SMARCA4, SMARCB1, SOX11, STAG1, SYNGAP1, TBL1XR1, TLK2, TRIO, ZBTB18, ZMYND11, ZNF292		
Intellectual developmental disorder, autosomal dominant, syndromic: 26 genes (86,4 kb)		
ADNP, AHDC1, ARID1A, ARID1B, ARID2, BICRA, CHAMP1, CTNNB1, DEAF1, DPF2, GATAD2B, GRIN1, KAT6A, KIF1A, MEF2C, PACS1, POGZ, PURA, SMARCA4, SMARCB1, SMARCC2, SMARCE1, SMARCD1, SOX4, SOX11, TRIP12		
Intellectual developmental disorder, autosomal recessive (MRT) *	4 - 6 w	E
Gene panel: ID037.02, 54 genes (141,2 kb)		
ADAT3, ALKBH8, ANK3, APC2, C12ORF4, CAMK2A, CC2D1A, CEP104, CRADD, CRBN, EDC3, EIF3F, ELP2, FBXO31, FMN2, GRIA1, GRIK2, HERC2, HNMT, IMPA1, KDM5B, KPTN, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NAA20, NDST1, NSUN2, PGAP1, PGAP2, PIDD1, PIGC, PRSS12, RSRC1, RUSC2, SLC6A17, ST3GAL3, TAF2, TAF13, TECR, TNIK, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, WDR11, ZBTB11, ZC3H14		
Intellectual developmental disorder, X-linked (XLID, MRX, MRXS) *	4 - 6 w	E
Gene panel: ID038.05		
Intellectual developmental disorder, X-linked (XLID, MRX, MRXS): 72 genes (178,2 kb)		
ACSL4, AFF2, AP1S2, ARX, ATP6AP2, ATRX, BRWD3, CASK, CLCN4, CLIC2, CNKSR2, 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, SMS, STEEP1, SYN1, SYP, TAF1, THOC2, TSPAN7, UBE2A, UPF3B, USP9X, USP27X, ZC4H2, ZDHHC9, 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): 47 genes (114,4 kb)		
AFF2, AP1S2, ARX, ATP6AP2, ATRX, CASK, CLCN4, CLIC2, CNKSR2, 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		
Intellectual developmental disorder and macrocephaly *	4 - 6 w	E
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		
Intellectual developmental disorder and microcephaly *	4 - 6 w	E
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		

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
Kabuki syndrome (KABUK) #,* Gene panel: ID127.00, 2 genes (20,8 kb) KDM6A, KMT2D	3 - 5 w	E
Neurodevelopmental disorders (NED), comprehensive diagnostics * Gene panel: ID358.00, 169 genes (431,8 kb) ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, 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
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: ID147.00, 27 genes (67,0 kb) ALDH18A1, ANAPC1, B3GALT6, B4GALT7, BANF1, BLM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LMNA, MDM2, MTX2, NAA10, POLD1, POLR3A, POLR3GL, PYCR1, RECQL4, SLC25A24, 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
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
Sotos syndrome (SOTOS) and similar malformation syndromes Gene panel: ID181.01, 4 genes (18,8 kb) APC2, EZH2, NFIX, NSD1	3 - 5 w	E
Three M syndrome (3M) * Gene panel: ID214.00, 3 genes (12,4 kb) CCDC8, CUL7, OBSL1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Developmental and Growth Disorders		
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
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
Hypogonadotropic hypogonadism with or without anosmia (KAL, HH) * Gene panel: ID170.04, 37 genes (62,5 kb) ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 w	E
Nonsyndromic deafness, comprehensive diagnostics * Gene panel: ID237.02 Nonsyndromic deafness, comprehensive diagnostics: 117 genes (326,4 kb) ABCC1, ACTG1, ADCY1, AIFM1, BDP1, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL11A1, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA4, GAB1, GIPC3, GJB2, GJB3, GJB6, GREB1L, GPRASP2, GRAP, GRHL2, GRXCR1, GRXCR2, GSDME, HGF, HOMER2, ILDR1, KARS1, KCNQ4, KITLG, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MCM2, MET, MPZL2, MSRB3, MYH14, MYH9*, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NLRP3, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PDZD7, PJVK, PLS1, PNPT1, POU3F4, POU4F3, PPIP5K2, PRPS1, PTPRQ, RDX, REST, RIPOR2, ROR1, S1PR2, SCD5, SERPINB6, SIX1, SLC12A2, SLC17A8, SLC26A4, SLC26A5, SLC44A4, SMPX, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C, WBP2, FS1, WHRN Nonsyndromic deafness, autosomal dominant (DFNA): 48 genes (144,6 kb) ABCC1, ACTG1, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MCM2, MYH14, MYH9, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PLS1, POU4F3, PTPRQ, REST, SCD5, SIX1, SCL12A2, SLC17A8, SLC44A4, TBC1D24, TECTA, TMC1, TNC, TRRAP, WFS1 Nonsyndromic deafness, autosomal recessive (DFNB): 75 genes (207,8 kb) ADCY1, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL4A6, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MPZL2, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PPIP5K2, PJVK, PNPT1, POU3F4, PRPS1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SMPX, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, 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
Nonsyndromic deafness, autosomal dominant (DFNA) * Gene panel: ID091.03, 48 genes (144,6 kb) ABCC1, ACTG1, CCDC50, CD164, CEACAM16, COCH, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, EYA4, GJB2, GJB3, GJB6, GREB1L, GRHL2, GSDME, HOMER2, KCNQ4, KITLG, LMX1A, MCM2, MYH14, MYH9, MYO6, MYO7A, NLRP3, OSBPL2, P2RX2, PDE1C, PLS1, POU4F3, PTPRQ, REST, SCD5, SIX1, SCL12A2, SLC17A8, SLC44A4, TBC1D24, TECTA, TMC1, TNC, TRRAP, WFS1	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Ear, Nose and Throat Diseases		
Nonsyndromic deafness, autosomal recessive (DFNB) *	4 - 6 w	E
Gene panel: ID092.03, 75 genes (207,8 kb)		
ADCY1, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN9, CLDN14, CLIC5, CLRN2, COCH, COL4A6, COL11A2, DCDC2, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, GAB1, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GRAP, GRXCR1, GRXCR2, HGF, ILDR1, KARS1, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MET, MPZL2, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OTOA, OTOF, OTOG, OTOGL, PCDH15, PDZD7, PPIP5K2, PJVK, PNPT1, POU3F4, PRPS1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4, SLC26A5, SMPX, SPNS2, STRC, SYNE4, TBC1D24, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TRIOBP, TPRN, TSPEAR, USH1C, WBP2, WHRN		
Nonsyndromic deafness, X-linked (DFNX) *	3 - 5 w	E
Gene panel: ID290.00, 7 genes (16,8 kb)		
AIFM1, COL4A5, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX		
Syndromic deafness, comprehensive diagnostics *	4 - 6 w	E
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		
Usher syndrome (USH) *	3 - 5 w	E
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		
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		

Diseases/Diagnostics	TAT	Material
Endocrine Disorders		
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: ID270.02		
Endocrine hypertension: 32 genes (80,9 kb)		
CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1, CYP11B2, CYP17A1, HSD11B2, KCNJ5, KLHL3, NF1, MAX, NR3C1, NR3C2, PDE3A, PDE8B, PDE11A, PRKAR1A, RET, SCNN1A, SCNN1B, SCNN1G, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL, WNK1, WNK4, YY1AP1		
Hyperaldosteronism (HALD): 6 genes (20,6 kb)		
CACNA1H, CACNA1D, CLCN2, CYP11B1, CYP11B2, KCNJ5		
Pseudohypoaldosteronism type II (PHA2): 4 genes (15,8 kb)		
CUL3, KLHL3, WNK1, WNK4		
Pheochromocytoma/paraganglioma syndrome (PPGL): 9 genes (9,5 kb)		
MAX, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, 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		
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		
Hyperinsulinemic hypoglycemia (HHF) *	3 - 5 w	E
Gene panel: ID126.00, 8 genes (16,9 kb)		
ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF4A		
Hyperparathyroidism (HRPT) *	3 - 5 w	E
Gene panel: ID338.00, 10 genes (19,2 kb)		
AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET, SLC12A1, TRPV6		
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		
Hypogonadotropic hypogonadism with or without anosmia (KAL, HH) *	3 - 5 w	E
Gene panel: ID170.04, 37 genes (62,5 kb)		
ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11		

Diseases/Diagnostics	TAT	Material
Endocrine Disorders		
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
Obesity * Gene panel: ID183.02 Obesity: 54 genes (130,1 kb) ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FFAR4, FTO, IFT27, IFT74, IFT172, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP Obesity, nonsyndromic: 19 genes (27,1 kb) ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FFAR4, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3 Obesity, syndromic: 36 genes (103,6 kb) ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, IFT172, IFT27, IFT74, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, PHF6, PHIP, RAB23, SDCCAG8, TRIM32, TTC8, TUB, VPS13B, WDPCP Bardet-Biedl syndrome (BBS): 22 genes (44,3 kb) ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CEP290, CFAP418, IFT27, IFT74, IFT172, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP	4 - 6 w	E
Ovarian dysgenesis (ODG) * Gene panel: ID293.01 Ovarian dysgenesis (ODG): 16 Gene (27,8 kb) BMP15, CLPP, ERAL1, ESR2, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 XX gonadal dysgenesis (ODG): 10 Gene (17,1 kb) BMP15, ESR2, FSHR, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX gonadal dysgenesis with deafness (PRLTS): 6 genes (10,7 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK	3 - 5 w	E
Pheochromocytoma/paraganglioma syndrome (PPGL) * Gene panel: ID042.02, 16 genes (29,4 kb) DLST, FH, KIF1B, GDNF, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	3 - 5 w	E
Premature ovarian failure (POF) * Gene panel: ID078.05 Premature ovarian failure (POF): 23 genes (51,1 kb) BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, INHA, LHCGR, MCM8, MSH5, NOBOX, NR5A1, POF1B, STAG3, SYCE1, XRCC2 Premature ovarian failure (POF), X-linked: 4 genes (8,2 kb) BMP15, DIAPH2, FMR1, POF1B Premature ovarian failure (POF), autosomal recessive: 10 genes (25,0 kb) C14ORF39, GDF9, FANCM, HFM1, HSF2BP, MCM8, MSH5, STAG3, SYCE1, XRCC2 Premature ovarian failure (POF), autosomal dominant: 6 genes (12,7 kb) BNC1, ERCC6, FIGLA, FOXL2, NOBOX, NR5A1	3 - 5 w	E
Pseudoaldosteronism (LIDLS) and Pseudohypoaldosteronism (PHA) * Gene panel: ID250.00 Pseudoaldosteronism (LIDLS) and Pseudohypoaldosteronism (PHA): 8 genes (23,8 kb) CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4 Pseudohypoaldosteronism, type I (PHA1): 4 genes (8,8 kb) NR3C2, SCNN1A, SCNN1B, SCNN1G Pseudohypoaldosteronism, type II (PHA2): 4 genes (15,7 kb) CUL3, KLHL3, WNK1, WNK4 Liddle syndrome (LIDLS): 3 genes (5,9 kb) SCNN1A, SCNN1B, SCNN1G	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
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
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
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, SLC7A6OS, SMC1A, SNIP1, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TCF4, TRAK1, UBA5, UGDH, UGP2, WWOX, YWHAG Focal epilepsy: 17 genes (50,9 kb) CHRNA2, CHRNA4, 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, SLC7A6OS, 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
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		
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
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
Generalized epilepsy * Gene panel: ID040.05 Generalized epilepsy: 39 genes (72,6 kb) ALDH7A1, ASAH1, CACNB4, CASR, CERS1, CILK1, CLCN2, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, HCN1, HCN2, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP, POLG, PRDM8, PRICKLE1, RORB, SCARB2, SCN1A, SCN1B, SEMA6B, SLC2A1, SLC6A1, SLC7A6OS, SLC12A5, STX1B, TBC1D24 Myoclonic epilepsy (EJM, EPM): 24 genes (45,3 kb) ASAH1, CERS1, CILK1, CLCN2, CNTN2, CSTB, CACNB4, EFHC1, EPM2A, GABRA1, GABRD, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG, PRDM8, PRICKLE1, SCARB2, SCN1A, SEMA6A, SLC7A6OS, TBC1D24 Absence epilepsy (EJA, ECA): 9 genes (18,3 kb) CASR, CLCN2, EFHC1, GABRA1, GABRB3, GABRG2, RORB, SLC2A1, SLC12A5 Generalized epilepsy with febrile seizures (GEFSP): 7 genes (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 w	E
Generalized epilepsy with febrile seizures plus (GEFSP) * Gene panel: ID235.02, 7 genes (15,7 kb) GABRD, GABRG2, HCN1, HCN2, SCN1A, SCN1B, STX1B	3 - 5 w	E
Hemiplegic migraine (FHM) * Gene panel: ID064.01, 4 genes (18,9 kb) ATP1A2, ATP1A3, CACNA1A, SCN1A	3 - 5 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
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 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

Diseases/Diagnostics	TAT	Material
Epilepsy and Migraine		
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 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
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
Eye Diseases		
Achromatopsia (ACHM) * Gene panel: ID164.01, 7 genes (11,5 kb) ATF6, CNGA3, CNGB3, GNAT2, OPN1LW, 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
Anterior segment dysgenesis (ASGD) * Gene panel: ID182.02 Anterior segment dysgenesis (ASGD): 9 genes (18,8 kb) B3GLCT, CPAMD8, CYP1B1, FOXC1, FOXE3, PAX6, PITX2, PITX3, PXDN Peters anomaly: 6 genes (7,8 kb) B3GLCT, CYP1B1, FOXC1, PAX6, PITX2, PITX3 Axenfeld-Rieger anomaly: 2 genes (3,3 kb) FOXC1, PITX2	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Eye Diseases		
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-rod or cone dystrophy (CORD, COD) * Gene panel: ID101.02 Cone-rod or cone dystrophy (CORD, COD): 29 genes (72,4 kb) ABCA4, ADAM9, AIPL1, C8ORF37, CACNA1F, CACNA2D4, CDHR1, CEP78, CEP250, CNNM4, CRX, DRAM2, GUCA1A, GUCY2D, KCNV2, OPN1LW, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5, UNC119 Cone-rod dystrophy (CORD): 24 genes (63,4 kb) ABCA4, ADAM9, AIPL1, C8ORF37, CACNA1F, CDHR1, CEP78, CEP250, CNNM4, CRX, DRAM2, GUCA1A, GUCY2D, PITPNM3, POC1B, PROM1, RAB28, RAX2, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5, UNC119 Cone dystrophy (COD): 8 genes (15,3 kb) CACNA2D4, GUCA1A, GUCY2D, KCNV2, OPN1LW, PDE6C, PDE6H, RPGR	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
Corneal dystrophy (CD) * Gene panel: ID329.00 Corneal dystrophy (CD): 26 genes (67,0 kb) AGBL1, CHST6, COL8A2, COL17A1, CYP4V2, DCN, FOXE3, GRHL2, GSN, KRT3, KRT12, LCAT, LOXHD1, 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: 5 genes (10,9 kb) COL17A1, KRT3, KRT12, 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

Diseases/Diagnostics	TAT	Material
Eye Diseases		
Leber congenital amaurosis (LCA) *	3 - 5 w	E
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		
Leber hereditary optic neuropathy (LHON) #	3 - 5 w	E
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		
Macula dystrophy (MD) *	3 - 5 w	E
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		
Microphthalmia, anophthalmia and coloboma *	4 - 6 w	E
Gene panel: ID263.01		
Microphthalmia, anophthalmia and coloboma: 50 genes (120,8 kb)		
ABCB6, ALDH1A3, BCOR, BMP4, CC2D2A, CHD7, C12ORF57, CRYAA, FRAS1, FREM1, FREM2, FOXE3, GDF3, GDF6, GLI2, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MAF, MFRP, MITF, NAA10, OTX2, PAX2, PAX6, PDE6D, PIGL, PITX3, PTCH1, PRSS56, RARB, RAX, RBP4, RPGRIP1L, SALL2, SEMA3A, SHH, SIX3, SMOC1, SOX2, SRD5A3, STRA6, TENM3, TFAP2A, TMEM67, VAX1, VSX2, YAP1		
Microphthalmia, syndromic (MCOPS): 12 genes (23,9 kb)		
BCOR, BMP4, HCCS, HMGB3, MAB21L2, NAA10, OTX2, SOX2, RARB, STRA6, TENM3, VAX1		
Microphthalmia, isolated (MCOP): 7 genes (9,7 kb)		
ALDH1A3, GDF3, GDF6, MFRP, PRSS56, RAX, VSX2		
Microphthalmia, isolated, with coloboma (MCOPCB): 8 genes (18,2 kb)		
ABCB6, GDF3, GDF6, RBP4, SHH, STRA6, TENM3, VSX2		
Ocular coloboma, isolated: 2 genes (4,3 kb)		
PAX6, SALL2		
Myopia (MYP) *	3 - 5 w	E
Gene panel: ID079.02		
Myopia (MYP): 20 genes (61,1 kb)		
ARR3, CNGB3, CPSF1, COL11A1, COL2A1, COL9A1, COL9A2, COL18A1, GZF1, IRX5, LRPAP1, LRP2, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLITRK6, ZNF644		
Myopia (MYP), nonsyndromic: 11 genes (22,3 kb)		
ARR3, CNGB3, CPSF1, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, ZNF644		
Myopia (MYP), syndromic: 10 genes (41,0 kb)		
COL11A1, COL2A1, COL9A1, COL9A2, COL18A1, GZF1, LRP2, IRX5, P3H2, SLITRK6		
Stickler syndrome (STL): 4 genes (14,7 kb)		
COL11A1, COL2A1, COL9A1, COL9A		
Nystagmus (NYS) *	4 - 6 w	E
Gene panel: ID331.00		
Nystagmus (NYS): 44 genes (105,8 kb)		
ATF6, CACNA1A, CACNA1F, CASK, CEP290, CNGA3, CNGB3, CRB1, CRX, DCT, FRMD7, GJC2, GNAT2, GPR143, GUCY2D, HPS1, HPS5, IMPDH1, KCNJ13, KIDINS220, LCA5, LRAT, LRMDA, LYST, MAG, NMNAT1, NYX, OCA2, OPN1LW, OPN1MW, PAX6, PDE6C, PDE6H, PLP1, PRPH2, RPE65, RPGRIP1, SLC24A5, SLC38A8, SLC45A2, TULP1, TYR, TYRP1, USP45		
Nystagmus, nonsyndromic (NYS): 2 genes (3,4 kb)		
FRMD7, GPR143		
Foveal hypoplasia, nonsyndromic (FVH): 2 genes (2,6 kb)		
PAX6, SLC38A8		
Albinism (OCA): 11 genes (29,2 kb)		
DCT, GPR143, HPS1, HPS5, LRMDA, LYST, OCA2, SLC24A5, SLC45A2, TYR, TYRP1		
Achromatopsia (ACHM): 8 genes (12,6 kb)		
ATF6, CNGA3, CNGB3, GNAT2, OPN1LW, OPN1MW, PDE6C, PDE6H		
Leber congenital amaurosis (LCA): 14 genes (33,0 kb)		
CEP290, CRB1, CRX, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2, RPE65, RPGRIP1, TULP1, USP45		
Pelizaeus-Merzbacher disease (PMD): 3 genes (4,0 kb)		
PLP1, GJC2, MAG		
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		
Optic atrophy (OPA) *	3 - 5 w	E
Gene panel ID081.04		
Optic atrophy (OPA): 36 genes (57,9 kb)		
ACO2, AFG3L2, ATP1A3, C19ORF12, CISD2, DNAJC30, DNM1L, EPRS1, FDXR, ISCA2, KLC2, MECR, MFF, MFN2, 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: 10 genes (14,8 kb)		
ACO2, AFG3L2, DNM1L, OPA1, OPA3, RTN4IP1, SPG7, 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		

Diseases/Diagnostics	TAT	Material
Eye Diseases		
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
Retinitis pigmentosa (RP), autosomal dominant * Gene panel: ID053.02, 25 genes (54,6 kb) AIPL1, ARL3, BEST1, FSCN2, GUCA1B, HK1, IMPDH1, KIF3B, KLHL7, NR2E3, NRL, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RGR, RHO, RP1, RP9, RPE65, SEMA4A, SNRNP200, TOPORS	3 - 5 w	E
Retinitis pigmentosa (RP), autosomal recessive * Gene panel: ID050.03, 57 genes (153,2 kb) ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2, C8ORF37, CDHR1, CERKL, CLCC1, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, IDH3A, IDH3B, IFT43, IFT140, IFT172, IMPG2, KIAA1549, KIZ, LRAT, MAK, MERTK, NEK2, NR2E3, PCARE, PDE6A, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, RBP3, RDH12, REEP6, RGR, RHO, RP1, RP1L1, RPE65, SAG, SEMA4A, SLC7A14, SPATA7, TTC8, TULP1, USH2A, ZNF408, 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
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
Stargardt disease (STGD) * Gene panel: ID102.00, 5 genes (13,8 kb) ABCA4, CNGB3, 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

Diseases/Diagnostics	TAT	Material
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
Gastric cancer * Gene panel: ID090.03, 24 genes (71,9 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, 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
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
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	2 - 4 w	E
Pancreatic cancer * Gene panel: ID089.03, 17 genes (55,2 kb) APC, ATM, BRCA1, BRCA2, CDKN2A, CTRC, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL	3 - 5 w	E
Pancreatitis (PCTT) * Gene panel: ID141.02, 18 genes (29,6 kb) APOA5, APOC2, CASR, CEL, CFTR, CPA1, CLDN2, CELA3B, CTRC, CTSB, GPIHBP1, LMF1, LPL, PNLIP, PRSS1, SPINK1, TRPV6, UBR1	3 - 5 w	E
Polycystic liver disease (PCLD) * Gene panel: ID305.01 Polycystic liver disease (PCLD): 11 genes (45,6 kb) ALG5, ALG8, DNAJB11, DZIP1L, GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63 Polycystic liver disease with or without kidney cysts (PCLD): 4 genes (10,3 kb) ALG8, LRP5, PRKCSH, SEC63 Polycystic kidney disease with polycystic liver disease (PKD): 7 genes (35,3 kb) ALG5, DNAJB11, DZYP1L, GANAB, PKD1, PKD2, PKHD1	3 - 5 w	E
Polyposis syndrome (PS, FAP) * Gene panel: ID005.05, 14 genes (37,6 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 w	E
Visceral neuropathy and myopathy, comprehensive diagnostics * Gene panel: ID238.02 Visceral neuropathy (VSCN) and myopathy (VSCM): 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		
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		
Bone marrow failure (BMF) *	3 - 5 w	E
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		
Erythrocytosis (ECYT) *	3 - 5 w	E
Gene panel: ID138.02, 11 genes (13,9 kb)		
BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB, JAK2, SH2B3, VHL		
Fanconi anemia (FANC) *	3 - 5 w	E
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		
Hemochromatosis (HFE) and Hemosiderosis *	3 - 5 w	E
Gene panel: ID114.04, 10 genes (14,6 kb)		
BMP6, CP, FTH1, FTL, HAMP, HFE, HJV, SLC40A1, TF, TFR2		
Hemophilia *	3 - 5 w	E
Gene panel: ID154.00, 3 genes (16,9 kb)		
F8, F9, VWF		
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		
Myelodysplastic syndrome (MDS) and Acute myeloid leukemia (AML) *	4 - 6 w	E
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		
Neutropenia *	3 - 5 w	E
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		

Diseases/Diagnostics	TAT	Material
Hematological Diseases		
Platelet disorders, comprehensive diagnostics * Gene panel: ID274.01 Platelet disorders, comprehensive diagnostics: 64 genes (142,3 kb) ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S3, BLOC1S5, BLOC1S6, CD36, CDC42, CYCS, DIAPH1, DTNBP1, EPHB2, ETV6, FERMT3, FLI1, FYB1, GATA1, GFI1B, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, IKZF5, ITGA2, ITGA2B, ITGB3, JAK2, LYST, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, TBXA2R, TBXAS1, THPO, TUBB1, WAS, WDR1, WIPF1 Thrombocytopenia, nonsyndromic (THC): 9 genes (17,7 kb) ANKRD26, CYCS, ETV6, FYB1, IKZF5, MASTL, SRC, TUBB1, WAS Thrombocythemia (THCYT): 3 genes (6,4 kb) JAK2, MPL, THPO Bleeding disorder, platelet-type (BDPLT): 23 genes (52,8 kb) ACTN1, ANO6, CD36, EPHB2, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, P2RY12, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1 Hermansky-Pudlak syndrome (HPS): 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	4 - 6 w	E
Porphyria * Gene panel: ID153.01, 10 genes (12,8 kb) ALAD, ALAS2, CLPX, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	3 - 5 w	E
Sideroblastic anemia (SIDBA) * Gene panel: ID355.00, 10 genes (14,9 kb) ABCB7, ALAS2, GLRX5, HSCB, HSPA9, LARS2, PUS1, SLC25A38, TRNT1, YARS2	3 - 5 w	E
Spherocytosis (SPH) and Elliptocytosis (EL) * Gene panel: ID203.01 Spherocytosis (SPH) and Elliptocytosis (EL) : 6 genes (26,8 kb) ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB Spherocytosis (SPH): 5 genes (24,2 kb) ANK1, EPB42, SLC4A1, SPTA1, SPTB Elliptocytosis (EL): 4 genes (18,4 kb) EPB41, SLC4A1, SPTA1, SPTB	3 - 5 w	E
Thrombocytopathy (BDPLT, HPS) * Gene panel: ID119.01 Thrombocytopathy (BDPLT, HPS): 37 genes (91,0 kb) ACTN1, ANO6, AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, CD36, DTNBP1, EPHB2, FERMT3, FLI1, GFI1B, GP1BA, GP1BB, GP6, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGA2B, ITGB3, LYST, MYH9, NBEAL2, P2RY12, PLA2G4A, PLAU, PRKACG, PTGS1, RASGRP2, SLFN14, TBXA2R, TBXAS1 Bleeding disorder, platelet-type (BDPLT), with thrombocytopenia: 15 genes (37,9 kb) ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14 Bleeding disorder, platelet-type (BDPLT), without thrombocytopenia: 10 genes (20,3 kb) ANO6, EPHB2, GP6, ITGA2B, ITGB3, P2RY12, PTGS1, RASGRP2, TBXA2R, TBXAS1 Hermansky-Pudlak syndrome (HPS): 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 w	E
Thrombocytopenia * Gene panel: ID104.02 Thrombocytopenia (THC): 40 genes (84,7 kb) ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CD36, CDC42, CYCS, DIAPH1, ETV6, FLI1, FYB1, GATA1, GFI1B, GP1BA, GP1BB, GP9, HOXA11, IKZF5, ITGA2, ITGA2B, ITGB3, MASTL, MECOM, MPIG6B, MPL, MYH9, NBEAL2, PLAU, PRKACG, RBM8A, RUNX1, SLFN14, SRC, STIM1, TUBB1, WAS, WDR1, WIPF1 Thrombocytopenia, nonsyndromic (THC): 9 genes (17,7 kb) ANKRD26, CYCS, ETV6, FYB1, IKZF5, MASTL, SRC, TUBB1, WAS Bleeding disorder, platelet-type (BDPLT): 15 genes (37,9 kb) ACTN1, CD36, FLI1, GFI1B, GP1BA, GP1BB, GP9, ITGA2, ITGA2B, ITGB3, MYH9, NBEAL2, PLAU, PRKACG, SLFN14	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

Diseases/Diagnostics	TAT	Material
Immunological Diseases		
Atypical hemolytic uremic syndrome (AHUS) *		
Gene panel: ID163.04, 20 genes (42,1 kb)	3 - 5 w	E
ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN		
Autoinflammatory syndromes *		
Gene panel: ID087.04	3 - 5 w	E
Autoinflammatoy syndromes: 37 genes (61,0 kb)		
ADA2, AP1S3, ACP5, ARPC1B, CARD14, ELANE, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NTRK1, OTULIN, PLCG2, POMP, PSMA3, PSMB4, PSMB8, PSMB9, PSMB10, PSMG2, PSTPIP1, RBCK1, RIPK1, SLC29A3, SOCS1, STING1, TNFAIP3, TNFRSF1A, TRAF3IP2, TRNT1, WDR1		
Recurrent fever syndromes: 12 genes (24,4 kb)		
ADA2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NTRK1, OTULIN, RIPK1, TNFRSF1A, TRNT1, WDR1		
Bone marrow failure (BMF) *		
Gene panel ID357.00	3 - 5 w	E
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		
Hyper-IgE syndrome mit recurrent infections (HIES) *		
Gene panel: ID240.01, 10 genes (29,5 kb)	3 - 5 w	E
DOCK8, DSG1, IL6R, IL6ST, PGM3, SPINK5, STAT3, STAT6, TYK2, ZNF341		
Neutropenia *		
Gene panel: ID189.02	3 - 5 w	E
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		
Periodic fever syndromes (HPF) *		
Gene panel: ID088.03, 7 genes (18,1 kb)	3 - 5 w	E
MEFV, MVK, NLRC4, NLRP12, NLRP3, PLCG2, TNFRSF1A		
Thrombotic microangiopathy (TMA) *		
Gene panel: ID707.00	3 - 5 w	E
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		
Kidney Diseases		
Alport syndrome (ATS) *		
Gene panel: ID099.00, 4 genes (21,0 kb)	3 - 5 w	E
COL4A3, COL4A4, COL4A5, MYH9		
Aminoaciduria *		
Gene panel: ID318.00	3 - 5 w	E
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) *		
Gene panel: ID163.04, 20 genes (42,1 kb)	3 - 5 w	E
ADAMTS13, C1GALT1C1, C2, C3, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR4, CFHR5, CFI, DGKE, MMACHC, MTHFD1, MTR, MTRR, THBD, VTN		
Bardet-Biedl syndrome (BBS) *		
Gene panel: ID093.02, 21 genes (39,0 kb)	3 - 5 w	E
ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP290, IFT27, IFT74, LZTFL1, MKKS, MKS1, SDCCAG8, TRIM32, TTC8, WDPCP		
Bartter syndrome (BARTS) *		
Gene panel: ID156.01, 8 genes (17,7 kb)	3 - 5 w	E
BSND, CASR, CLCNKA, CLCNKB, KCNJ1, MAGED2, SLC12A1, SLC12A3		

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
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: ID100.07		
Cystic kidney diseases, comprehensive diagnostics: 49 genes (159,8 kb)		
ALG5, ALG8, ALG9, ANKS6, BICC1, CEP83, CEP164, CEP290, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, LRP5, MAPKBP1, MUC1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, PRKCSH, REN, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, SLC41A1, TSC1, TSC2, TTC21B, TMEM67, UMOD, VHL, WDR19, XPNPEP3, ZNF423		
Polycystic kidney disease (PKD): 7 genes (35,3 kb)		
ALG5, DNAJB11, DZIP1L, GANAB, PKD1, PKD2, PKHD1		
Polycystic liver disease with kidney cysts (PCLD): 4 genes (10,3 kb)		
ALG8, LRP5, PRKCSH, SEC63		
Medullary cystic kidney disease (MCKD, ADTKD): 5 genes (7,7 kb)		
HNF1B, MUC1, REN, SEC61A1, UMOD		
Nephronophthisis (NPHP): 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		
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		
Fanconi renotubular syndrome (FRTS) *	3 - 5 w	E
Gene panel: ID359.00, 7 genes (10,4 kb)		
CTNS, EHHADH, GATM, HNF4A NDUFAF6, SLC2A2, SLC34A1		
Focal segmental glomerulosclerosis (FSGS) and Nephrotic syndrome (SRNS, NPHS) *	4 - 6 w	E
Gene panel: ID098.05		
Focal segmental glomerulosclerosis (FSGS) and Nephrotic syndrome (SRNS, NPHS): 40 genes (126,0 kb)		
ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CD2AP, COL4A3, COL4A4, COL4A5, COQ8B, CRB2, DAAM2, DGKE, EMP2, INF2, KANK2, KIRREL1, LAMA5, LAMB2, LMX1B, MAGI2, MYO1E, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PAX2, PLCE1, PTPRO, SGPL1, SMARCAL1, TBC1D8B, TRPC6, WT1		
Nephrotic syndrome (SRNS, NPHS): 30 genes (100,6 kb)		
ARHGAP24, ARHGDIA, AVIL, COQ8B, COL4A3, COL4A4, COL4A5, DAAM2, DGKE, EMP2, KANK2, KIRREL1, LAMA5, LAMB2, PTPRO, MAGI2, NOS1AP, NPHS1, NPHS2, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, PLCE1, SGPL1, SMARCAL1, TBC1D8B, WT1		
Focal-segmental glomerulosclerosis (FSGS): 13 genes (40,6 kb)		
ACTN4, ANLN, APOL1, COL4A3, COL4A4, COL4A5, CRB2, CD2AP, INF2, LMX1B, MYO1E, PAX2, TRPC6		
Fraser syndrome (FRASRS) *	3 - 5 w	E
Gene panel: ID317.00, 3 genes (24,8 kb)		
FRAS1, FREM2, GRIP1		
Galloway-Mowat syndrome (GAMOS) *	3 - 5 w	E
Gene panel: ID251.01, 10 genes (12,5 kb)		
GON7, LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73, YRDC		

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
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, TOGAGRAM1, 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
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
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
Polycystic kidney disease (PKD) * Gene panel: ID295.01 Polycystic kidney disease (PKD): 7 genes (35,3 kb) ALG5, DNAJB11, DZIP1L, GANAB, PKD1, PKD2, PKHD1 Polycystic kidney disease, autosomal dominant (PKD, ADPKD): 5 genes (20,8 kb) ALG5, DNAJB11, GANAB, 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: ID305.01 Polycystic liver disease (PCLD): 11 genes (45,6 kb) ALG5, ALG8, DNAJB11, DZIP1L, GANAB, LRP5, PKD1, PKD2, PKHD1, PRKCSH, SEC63 Polycystic liver disease with or without kidney cysts (PCLD): 4 genes (10,3 kb) ALG8, LRP5, PRKCSH, SEC63 Polycystic kidney disease with polycystic liver disease (PKD): 7 genes (35,3 kb) ALG5, DNAJB11, DZYP1L, GANAB, PKD1, PKD2, PKHD1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
Pseudoaldosteronism (LIDLS) and Pseudohypoaldosteronism (PHA) *	3 - 5 w	E
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		
Renal amyloidosis *	3 - 5 w	E
Gene panel: ID320.00, 8 genes (11,8 kb)		
APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TTR		
Renal cancer *	3 - 5 w	E
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		
Renal hypodysplasia, aplasia and agenesis *	3 - 5 w	E
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		
Renal tubular acidosis (RTA) *	3 - 5 w	E
Gene panel: ID297.00, 9 genes (18,5 kb)		
ATP6V0A4, ATP6V1B1, CA2, FOXI1, SLC4A1, SLC4A4, VIPAS39, VPS33B, WDR72		
Renal tubular dysgenesis (RTD) *	2 - 4 w	E
Gene panel: ID316.00, 4 genes (7,7 kb)		
ACE, AGT, AGTR1, REN		
Senior-Loken syndrome (SLSN) *	3 - 5 w	E
Gene panel: ID029.01, 8 genes (27,9 kb)		
CEP290, IQCB1, NPHP1, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19		
Thrombotic microangiopathy (TMA) *	3 - 5 w	E
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		
Tubulointerstitial kidney disease, autosomal dominant (ADTKD) *	3 - 5 w	E
Gene panel: ID296.00, 6 genes (8,8 kb)		
DNAJB11, HNF1B, MUC1, REN, SEC61A1, UMOD		
Urolithiasis, nephrolithiasis and nephrocalcinosis *	3 - 5 w	E
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		
Urothelial cancer *	4 - 6 w	E
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		

Diseases/Diagnostics	TAT	Material
Kidney Diseases		
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
Wilms tumor (WT) * Gene panel: ID335.00, 21 genes (70,4 kb) AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CEP57, CTR9, DICER1, DIS3L2, GPC3, GPC4, NSD1, PALB2, POU6F2, REST, TRIM28, TRIM37, TRIP13, WT1	3 - 5 w	E
Lipometabolic Disorders		
Combined hyperlipidemia * Gene panel: ID025.06, 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: ID095.02, 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 Hyperchylomicronemia: 6 genes (6,1 kb) APOC2, APOE, APOA5, GPIHBP1, LMF1, LPL Lipodystrophy: 12 genes (19,0 kb) AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEC, LIPE, LMNA, PLIN1, POLD1, PPARG, ZMPSTE2	3 - 5 w	E
Lipodystrophy (CGL, FPLD)* Gene panel: ID055.03, 12 genes (19,0 kb) AGPAT2, AKT2, BSCL2, CAV1, CAVIN1, CIDEC, LIPE, LMNA, PLIN1, POLD1, PPARG, ZMPSTE24 Congenital generalized lipodystrophy (CGL): 4 genes (3,7 kb) AGPAT2, BSCL2, CAV1, CAVIN1 Familial partial lipodystrophy (FPLD): 6 genes (9,6 kb) CAV1, CIDEC, LIPE, LMNA, 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: ID044.02, 56 genes (107,1 kb) ABCA1, ABCG5, ABCG8, 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, PLIN1, PNPLA5, POLD1, PPARG, SAR1B, SCARB1, SMPD1, SORT1, ZMPSTE24	4 - 6 w	E
Statin-associated myopathy * Gene panel: ID097.00, 10 genes (35,2 kb) ACADM, ACADS, ACADVL, AMPD1, CACNA1S, CAV3, CPT2, LPIN1, PYGM, RYR1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Metabolic Diseases		
Aminoaciduria * Gene panel: ID318.00 Aminoaciduria: 13 genes (21,1 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC1A1, SLC2A2, SLC3A1, SLC6A19, SLC6A20, SLC7A7, SLC7A9, SLC34A1, SLC36A2 Cystinuria: 2 genes (3,5 kb) SLC3A1, SLC7A9 Hyperglycinuria: 3 genes (5,2 kb) SLC6A19, SLC6A20, SLC36A2 Fanconi renotubular syndrome (FRTS): 5 genes (7,7 kb) EHHADH, GATM, HNF4A, NDUFAF6, SLC34A1	3 - 5 w	E
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: ID287.00, 51 genes (67,7 kb) AARS2, AIFM1, ATP5F1A, C1QBP, CARS2, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL44, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NSUN3, QRSL1, PNPT1, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	3 - 5 w	E
Congenital disorder of glycosylation (CDG) * Gene panel ID035.02 Congenital disorder of glycolysation (CDG): 51 genes (74,7 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6AP1, ATP6AP2, ATP6V0A2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, EDEM2, GALNT2, MAGT1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1, SLC37A4, SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199, TUSC3 Congenital disorder of glycolysation, type I (CDG1): 29 genes (39,6 kb) ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, ALG13, ATP6V0A2, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, MAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3, SSR4, STT3A, STT3B, TUSC3 Congenital disorder of glycolysation, type II (CDG2): 22 genes (35,1 kb) ATP6AP1, ATP6AP2, B4GALT1, CCDC115, COG1, COG2, COG4, COG5, COG6, COG7, COG8, EDEM2, GALNT2, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A8, TMEM165, TMEM199	3 - 5 w	E
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 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

Diseases/Diagnostics	TAT	Material
Metabolic Diseases		
Hyperphosphatasia with impaired intellectual development syndrome (HPMRS) * Gene panel: ID292.00, 6 genes (8,2 kb) PGAP2, PGAP3, PIGO, PIGV, PIGW, PIGY	2 - 4 w	E
Hypoglycemia, hyperinsulinism and ketone metabolism * Gene panel: ID280.00 Hypoglycemia, hyperinsulinism and ketone metabolism: 44 genes (85,9 kb) ABCC8, ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, OXCT1, PC, PCCA, PCCB, PCK1, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC16A1, SLC2A2, SLC37A4 Glycogen storage disease (GSD): 24 genes (48,4 kb) AGL, ALDOA. ENO3, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, SLC2A2, SLC37A4 Hyperinsulinemic hypoglycemia (HHF): 9 genes (18,8 kb) ABCC8, KCNJ11, GCK, HADH, INSR, GLUD1, SLC16A1, HNF1A, HNF4A	3 - 5 w	E
Hypomagnesemia (HOMG)* Gene panel: ID054.02, 14 genes (30,9 kb) ATP1A1, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, EGF, 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
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
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 epilepy 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
Mucopolysaccharidosis (MPS) * Gene panel: ID308.00, 12 genes (21,2 kb) ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, VPS33A	3 - 5 w	E

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		
Obesity *	4 - 6 w	E
Gene panel: ID183.02		
Obesity: 54 genes (130,1 kb)		
ADCY3, ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS2, BBS4, BBS5, BBS7, BBS9, BBS12, CARTPT, CELA2A, CEP19, CEP290, CFAP418, CPE, CUL4B, DYRK1B, FFAR4, FTO, IFT27, IFT74, IFT172, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PHF6, PHIP, POMC, PPARG, RAB23, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP		
Obesity, nonsyndromic: 19 genes (27,1 kb)		
ADCY3, CARTPT, CELA2A, CEP19, DYRK1B, FFAR4, FTO, LEP, LEPR, MC3R, MC4R, MRAP2, NR0B2, PCSK1, POMC, PPARG, SIM1, SH2B1, UCP3		
Obesity, syndromic: 36 genes (103,6 kb)		
ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CFAP418, CPE, CUL4B, IFT172, IFT27, IFT74, KSR2, LZTFL1, MAGEL2, MEGF8, MKKS, MKS1, MYT1L, NTRK2, 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		
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		
Renal amyloidosis *	3 - 5 w	E
Gene panel: ID320.00, 8 genes (11,8 kb)		
APOA1, B2M, FGA, GSN, LYZ, MEFV, NLRP3, TTR		
Storage diseases with cardiac involvement *	3 - 5 w	E
Gene panel: ID149.01, 13 genes (21,5 kb)		
ATP7B, FTH1, GAA, GLA, GSN, HAMP, HFE, HJV, LAMP2, PRKAG2, SLC40A1, TFR2, 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		
Intellectual developmental disorder and macrocephaly *	4 - 6 w	E
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		
Intellectual developmental disorder and microcephaly *	4 - 6 w	E
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		
Hydrocephaly *	3 - 5 w	E
Gene panel: ID221.04		
Hydrocephaly: 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		
Pontocerebellar hypoplasia (PCH) *	3 - 5 w	E
Gene panel: ID071.02, 26 genes (60,5 kb)		
AMPD2, CASK, CDC40, CHMP1A, CLP1, COASY, EXOSC1, EXOSC3, EXOSC8, EXOSC9, MINPP1, PCLO, PPIL1, RARS2, RELN, SEPSECS, SLC25A46, TBC1D23, TOE1, TSEN2, TSEN15, TSEN34 TSEN54, VPS51, VPS53, VRK1		
Primary microcephaly, autosomal recessive (MCPH) *	4 - 6 w	E
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		
Rubinstein-Taybi syndrome (RSTS) *	3 - 5 w	E
Gene panel: ID142.01, 3 genes (24,3 kb)		
CREBBP, EP300, SRCAP		
Seckel syndrome (SCKL) *	3 - 5 w	E
Gene panel: ID113.00, 9 genes (33,5 kb)		
ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP		
Sotos syndrome (SOTOS) and similar malformation syndromes	3 - 5 w	E
Gene panel: ID181.01, 4 genes (18,8 kb)		
APC2, EZH2, NFIX, NSD1		

Diseases/Diagnostics	TAT	Material
Mitochondrial Diseases		
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
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: ID287.00, 51 genes (67,7 kb) AARS2, AIFM1, ATP5F1A, C1QBP, CARS2, EARS2, ELAC2, FARS2, FASTKD2, GATB, GATC, GFM1, GFM2, GTPBP3, LYRM4, MICOS13, MIEF2, MIPEP, MRPL3, MRPL12, MRPL44, MRPS2, MRPS7, MRPS14, MRPS16, MRPS22, MRPS23, MRPS25, MRPS34, MRPS28, MTFMT, MTO1, MTRFR, MARS2, NARS2, NSUN3, QRSL1, PNPT1, PTCD3, RMND1, SFXN4, SLC25A26, TARS2, TIMM22, TRIT1, TRMT5, TRMT10C, TSFM, TUFM, TXN2, VARS2	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 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 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 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
Multisystemic Malformation Syndromes		
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: ID118.01, 10 genes (34,2 kb) ARID1A, ARID1B, ARID2, DPF2, SMARCC2, SMARCA4, SMARCB1, 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
FG syndrome (FGS) * Gene panel: ID215.00, 3 genes (17,2 kb) CASK, FLNA, MED12	3 - 5 w	E
Kabuki syndrome (KABUK) #,* Gene panel: ID127.00, 2 genes (20,8 kb) KDM6A, KMT2D	3 - 5 w	E
Marfan syndrome (MFS) #,* Gene panel: ID022.00, 3 genes (11,8 kb) FBN1, TGFBR1, TGFBR2	2 - 4 w	E
Malformation syndromes with predominantly facial involvement * Gene panel: ID279.00 Malformation syndromes with predominantly facial involvement: 25 genes (90,9 kb) C2CD3, COL11A2, CPLANE1, DDX59, FGFR1, FGFR2, FGFR3, FRAS1, FREM2, GRIP1, IFT57, INTU, KIAA0753, MEGF8, MYH3, MYMK, MYT1, OFD1, RAB23, RBM10, TCTN3, TGDS, TMEM107, 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
Rubinstein-Taybi syndrome (RSTS) * Gene panel: ID142.01, 3 genes (24,3 kb) CREBBP, EP300, SRCAP	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Multisystemic Malformation Syndromes		
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
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
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
Centronuclear myopathy (CNM) * Gene panel: ID257.00, 7 genes (21,4 kb) CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1, SPEG	3 - 5 w	E
Charcot-Marie-Tooth disease, axonal type (CMT, HMSN) * Gene panel: ID052.02 Charcot-Marie-Tooth disease, axonal type (CMT, HMSN): 41 genes (98,5 kb) AARS1, AIFM1, ATP1A1, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PRPS1, RAB7A, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth disease, axonal, autosomal dominant (CMT2): 22 genes (60,2 kb) AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, TRPV4, VCP Charcot-Marie-Tooth disease, axonal, autosomal recessive (CMT2): 10 genes (25,1 kb) GDAP1, IGHMBP2, LMNA, LRSAM1, MED25, MFN2, MME, MPV17, 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
Muscular Diseases		
Charcot-Marie-Tooth disease, demyelinating type (CMT, HMSN) *	3 - 5 w	E
Gene panel: ID051.02		
Charcot-Marie-Tooth disease, demyelinating type (CMT, HMSN): 26 genes (56,7 kb)		
CNTNAP1, COX6A1, DNM2, EGR2, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1		
Charcot-Marie-Tooth disease, demyelinating, autosomal dominant (CMT1): 7 genes (6,3 kb)		
EGR2, GDAP1, LITAF, MPZ, NEFL, PMP2, PMP22		
Charcot-Marie-Tooth disease, demyelinating, autosomal recessive (CMT4): 12 genes (33,8 kb)		
EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1		
Charcot-Marie-Tooth disease, intermediate (CMTDI, CMTRI): 11 genes (18,5 kb)		
COX6A1, DNM2, GDAP1, GJB1, GNB4, INF2, KARS1, MPZ, NEFL, PLEKHG5, YARS1		
Neuropathy of Déjerine-Sottas (CMT3, DSS): 4 genes (7,1 kb)		
EGR2, MPZ, PMP22, PRX		
Congenital hypomyelinating neuropathy (CHN): 3 genes (6,3 kb)		
EGR2, MPZ, CNTNAP1		
Charcot-Marie-Tooth disease (CMT, HMSN), comprehensive diagnostics *	4 - 6 w	E
Gene panel: ID312.01		
Charcot-Marie-Tooth disease (CMT, HMSN), comprehensive diagnostics: 62 genes (146,3 kb)		
AARS1, AIFM1, ARHGEF10, ATP1A1, CNTNAP1, COX6A1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PRPS1, PRX, RAB7A, SBF1, SBF2, SH3TC2, SLC25A46, SORD, SPG11, SPTLC1, SURF1, TRIM2, TRPV4, VCP, YARS1		
Charcot-Marie-Tooth disease, demyelinating, dominant (CMT1): 7 genes (6,3 kb)		
EGR2, GDAP1, LITAF, MPZ, NEFL, PMP2, PMP22		
Charcot-Marie-Tooth disease, demyelinating, recessive (CMT4): 12 genes (33,8 kb)		
EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1		
Charcot-Marie-Tooth disease, axonal (CMT2): 29 genes (79,8 kb)		
AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, RAB7A, 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		
Charcot-Marie-Tooth disease with optic atrophy (CMT6): 3 genes (4,5 kb)		
MFN2, PDXK, SLC25A46		
Hypertrophic neuropathy of Déjerine-Sottas (CMT3, DSS): 4 genes (7,1 kb)		
EGR2, MPZ, PMP22, PRX		
Congenital hypomyelinating neuropathy (CHN): 3 genes (6,3 kb)		
EGR2, MPZ, CNTNAP1		
Congenital myasthenic syndrome (CMS) *	3 - 5 w	E
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		
Congenital structural myopathy *	3 - 5 w	E
Gene panel: ID212.01		
Congenital structural myopathy: 20 genes (78,8 kb)		
ACTA1, BIN1, CCDC78, CFL2, DNM2, KBTBD13, KLHL40, KLHL41, LMOD2, MAP3K20, MTM1, MTMR14, MYPN, NEB, RYR1, SELENON, SPEG, TNNT1, TPM2, TPM3		
Core myopathy: ID212.01, 4 genes (18,9 kb)		
ACTA1, RYR1, SELENON, TPM3		
Nemaline myopathy (NEM): ID212.01, 11 genes (40,6 kb)		
ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, TNNT1, TPM2, TPM3, MYPN, NEB		
Centronuclear myopathy (CNM): ID212.01, 7 genes (20,3 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 myopathy (MPD) *	4 - 6 w	E
Gene panel: ID328.00, 18 genes (169,4 kb)		
ACTN2, ADSS1, ANO5, CAV3, CRYAB, DES, DNAJB6, DYSF, FLNC, GNE, LDB3, MATR3, MYH7, MYOT, NEB, TIA1, TCAP, TTN		

Diseases/Diagnostics	TAT	Material
Muscular Diseases		
Distal hereditary motor neuronopathy (HMN, DSMA) *	3 - 5 w	E
Gene panel: ID254.00		
Distal hereditary motor neuronopathy (HMN, DSMA): 17 genes (38,7 kb)		
ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, TRPV4, WARS1		
Distal hereditary motor neuronopathy, dominant type (HMN, DHMN): 12 genes (26,7 kb)		
BSCL2, DCTN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, TRPV4, WARS1		
Distal hereditary motor neuronopathy, recessive type (DSMA): 5 genes (12,0 kb)		
ATP7A, DNAJB2, IGHMBP2, PLEGHG5, SIGMAR1		
Emery-Dreifuss muscular dystrophy (EDMD) *	3 - 5 w	E
Gene panel: ID121.00, 6 genes (51,8 kb)		
EMD, FHL1, LMNA, SYNE1, SYNE2, TMEM43		
Fetal akinesia deformation sequence (FADS) *	3 - 5 w	E
Gene panel: ID201.00, 10 genes (30,3 kb)		
CHRNA1, CHRND, CHRNG, DOK7, GBE1, MUSK, MYOD1, NUP88, RAPSN, RYR1		
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		
Limb-girdle muscular dystrophy (LGMD) *	4 - 6 w	E
Gene panel: ID122.03		
Limb-girdle muscular dystrophy (LGMD): 37 Gene (196,8 kb)		
ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DNAJB6, DPM3, DYSF, FKRP, FKTN, GMPPB, HNRNPDL, JAG2, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN		
Limb-girdle muscular dystrophy, autosomal recessive (LGMDR): 34 Gene (191,8 kb)		
ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, CRPPA, DAG1, DES, DPM3, DYSF, FKRP, FKTN, GMPPB, JAG2, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC3, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TRAPPC11, TRIM32, TT		
Limb-girdle muscular dystrophy, autosomal dominant (LGMDD): 7 genes (23,2 kb)		
CAPN3, COL6A1, COL6A2, COL6A3, DNAJB6, HNRNPDL, TNPO3		
Limb-girdle muscular dystrophy-dystroglycanopathy (MDDGC): 11 genes (17,6 kb)		
CRPPA, DAG1, DPM3, FKRP, FKTN, GMPPB, POMGNT1, POMGNT2, POMK, POMT1, POMT2		
Ullrich muscular dystrophy (UCMD): 3 genes (15,7 kb)		
COL6A1, COL6A2, COL6A3		
Bethlem myopathy (BTHLM): 3 genes (15,7 kb)		
COL6A1, COL6A2, COL6A3		
Nemaline myopathy (NEM) *	3 - 5 w	E
Gene panel: ID199.00, 11 genes (40,6 kb)		
ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MYPN, NEB, TNNT1, TPM2, TPM3		
Muscular diseases, comprehensive diagnostics *	4 - 6 w	E
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		

Diseases/Diagnostics	TAT	Material
Muscular Diseases		
Muscular diseases with cardiac involvement * Gene panel: ID123.02 Muscular diseases with cardiac involvement: 34 genes (220,8 kb) BAG3, BVES, CAV3, CRYAB, DES, DMD, DPM3, EMD, FHL1, FKRP, FKTN, FLNC, JAG2, KY, LDB3, LAMA2, LIMS2, LMNA, MYL2, MYOT, POMT1, PYROXD1, SGCA, SGCB, SGCD, SGCG, SVIL, SYNE1, SYNE2, TCAP, TMEM43, TOR1AIP1, TTN, UNC45B Myofibrillar myopathy (MFM): 12 genes (130,7 kb) BAG3*, CRYAB, DES*, FLNC, KY, LDB3, MYL2, MYOT, PYROXD1, SVIL, TTN, UNC45B Limb-girdle muscular dystrophy (LGMD): 17 genes (131,7 kb) BVES, CAV3, DES, DPM3, FKRP, FKTN, JAG2, LAMA2, LIMS2, POMT1, SGCA, SGCB, SGCD, SGCG, TCAP, TOR1AIP1, TTN 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
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
Periodic paralysis * Gene panel: ID253.00, 7 genes (16,6 kb) CACNA1S, KCNE3, KCNJ2, KCNJ5, KCNJ12, KCNJ18, SCN4A	3 - 5 w	E
Sensory and autonomic neuropathy (HSAN, HSN) * Gene panel: ID086.01, 15 genes (52,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, WNK1	3 - 5 w	E
Spinal muscular atrophy (SMA) * Gene panel: ID152.01 Spinal muscular atrophy (SMA): 31 genes (69,5 kb) ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB, VRK1, WARS1 Proximal spinal muscular atrophy (SMA): 10 genes (26,6 kb) ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, SMN1, SMN2, TRIP4, UBA1, VAPB Distal spinal muscular atrophy (DSMA, HMN): 18 genes (41,9 kb) ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, SIGMAR1, PLEKHG5, REEP1, SETX, SLC5A7, TRPV4, UBA1, WARS1	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: ID209.02 Amyotrophic lateral sclerosis (ALS): 31 genes (71,4 kb) ALS2, ANG, ANXA11, CCNF, CHCHD10, CHMP2B, CYLD, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VAPB, VCP Amyotrophic lateral sclerosis, juvenile type: 4 genes (21,0 kb) ALS2, SETX, SIGMAR1, SPG11 Amyotrophic lateral sclerosis and frontotemporal dementia (FTDALS): 13 genes (21,2 kb) CCNF, CHCHD10, CHMP2B, CYLD, FUS, OPTN, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP	3 - 5 w	E
Basal ganglia calcification (IBGC) * Gene panel: ID327.00, 6 genes (11,2 kb) JAM2, MYORG, PDGFB, PDGFRB, SLC20A2, XPR1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
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
Cerebellar ataxia, autosomal dominant * Gene panel: ID236.02, 29 genes (95,6 kb) AFG3L2, ATP1A3, CACNA1A, CACNA1G, CCDC88C, DAB1, DNMT1, EEF2, ELOVL4, ELOVL5, FAT2, FGF12, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, PDYN, PLD3, PRKCG, PUM1, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2, TUBB4A	4 - 6 w	E
Cerebellar ataxia, autosomal recessive * Gene panel: ID213.03 Cerebellar ataxia, autosomal recessive: 50 genes (164,7 kb) ANO10, APTX, ATCAY, ATG5, ATM, ATP8A2, CA8, COA7, COQ8A, CWF19L1, CYP27A1, FXN, GDAP2, GRID2, GRM1, PEX7, PHYH, PIK3R5, PMPCA, PNKP, PNPLA6, POLG, RNF216, RUBCN, SCYL1, SACS, SETX, SIL1, SLC52A2, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP1, TDP2, THG1L, TPP1, TTPA, TWNK, UBA5, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1 Spinocerebellar ataxia (SCAR): 24 genes (87,8 kb) ANO10, ATG5, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PMPCA, RUBCN, SCYL1, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP2, THG1L, TPP1, UBA5, VPS13D, VWA3B, WWOX, XRCC1 Spinocerebellar ataxia with axonal neuropathy (SCAN): 3 genes (10,6 kb) COA7, SETX, TDP1 Cerebellar ataxia with oculomotor apraxia (AOA): 4 genes (13,3 kb) APTX, PIK3R5, PNKP, SETX Cerebellar ataxia with mental retardation (CAMRQ): 4 genes (12,9 kb) ATP8A2, CA8, VLDLR, WDR81	4 - 6 w	E
Cerebellar ataxia, X-linked * Gene panel: ID273.00, 7 genes (15,9 kb) ABCB7, AIFM1, ATP2B3, CASK, OPHN1, PRPS1, SLC9A6	3 - 5 w	E
Charcot-Marie-Tooth disease (CMT, HMSN), comprehensive diagnostics * Gene panel: ID312.01 Charcot-Marie-Tooth disease (CMT, HMSN), comprehensive diagnostics: 62 genes (146,3 kb) AARS1, AIFM1, ARHGEF10, ATP1A1, CNTNAP1, COX6A1, DHTKD1, DNAJB2, DNM2, DYNC1H1, EGR2, FGD4, FIG4, GAN, GARS1, GDAP1, GJB1, GNB4, HARS1, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LITAF, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, NAGLU, NDRG1, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PMP2, PMP22, PRPS1, PRX, RAB7A, SBF1, SBF2, SH3TC2, SLC25A46, SORD, SPG11, SPTLC1, SURF1, TRIM2, TRPV4, VCP, YARS1 Charcot-Marie-Tooth disease, demyelinating, dominant (CMT1): 7 genes (6,3 kb) EGR2, GDAP1, LITAF, MPZ, NEFL, PMP2, PMP22 Charcot-Marie-Tooth disease, demyelinating, recessive (CMT4): 12 genes (33,8 kb) EGR2, FGD4, FIG4, GDAP1, HK1, MTMR2, NDRG1, PRX, SBF1, SBF2, SH3TC2, SURF1 Charcot-Marie-Tooth disease, axonal (CMT2): 29 genes (79,8 kb) AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, IGHMBP2, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, RAB7A, 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 Charcot-Marie-Tooth disease with optic atrophy (CMT6): 3 genes (4,5 kb) MFN2, PDXK, SLC25A46 Hypertrophic neuropathy of Déjerine-Sottas (CMT3, DSS): 4 genes (7,1 kb) EGR2, MPZ, PMP22, PRX Congenital hypomyelinating neuropathy (CHN): 3 genes (6,3 kb) EGR2, MPZ, CNTNAP1	4 - 6 w	E

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Charcot-Marie-Tooth disease, axonal type (CMT, HMSN) *	4 - 6 w	E
Gene panel: ID052.02		
Charcot-Marie-Tooth disease, axonal type (CMT, HMSN): 41 genes (98,5 kb)		
AARS1, AIFM1, ATP1A1, COX6A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, GJB1, GNB4, HARS1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1B, LMNA, LRSAM1, MARS1, MED25, MFN2, MME, MORC2, MPV17, MPZ, NAGLU, NEFH, NEFL, PDK3, PDXK, PLEKHG5, PRPS1, RAB7A, SLC25A46, SPG11, TRIM2, TRPV4, VCP, YARS1		
Charcot-Marie-Tooth disease, axonal, autosomal dominant (CMT2): 22 genes (60,2 kb)		
AARS1, ATP1A1, DHTKD1, DNM2, DYNC1H1, GARS1, GDAP1, HARS1, HSPB1, HSPB8, KIF1B, LRSAM1, MARS1, MFN2, MORC2, MPZ, NAGLU, NEFH, NEFL, RAB7A, TRPV4, VCP		
Charcot-Marie-Tooth disease, axonal, autosomal recessive (CMT2): 10 genes (25,1 kb)		
GDAP1, IGHMBP2, LMNA, LRSAM1, MED25, MFN2, MME, MPV17, 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 (CMT, HMSN) *	3 - 5 w	E
Gene panel: ID051.02		
Charcot-Marie-Tooth disease, demyelinating type (CMT, HMSN): 26 genes (56,7 kb)		
CNTNAP1, COX6A1, DNM2, EGR2, FGD4, FIG4, GDAP1, GJB1, GNB4, HK1, INF2, KARS1, LITAF, MPZ, MTMR2, NDRG1, NEFL, PLEKHG5, PMP2, PMP22, PRX, SBF1, SBF2, SH3TC2, SURF1, YARS1		
Charcot-Marie-Tooth disease, demyelinating, autosomal dominant (CMT1): 7 genes (6,3 kb)		
EGR2, GDAP1, LITAF, MPZ, NEFL, 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		
Choreatiform dyskinesia *	3 - 5 w	E
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		
Distal hereditary motor neuronopathy (HMN, DSMA) *	3 - 5 w	E
Gene panel: ID254.00		
Distal hereditary motor neuronopathy (HMN, DSMA): 17 genes (38,7 kb)		
ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, TRPV4, WARS1		
Distal hereditary motor neuronopathy, dominant type (HMN, DHMN): 12 genes (26,7 kb)		
BSCL2, DCTN1, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, TRPV4, WARS1		
Distal hereditary motor neuronopathy, recessive type (DSMA): 5 genes (12,0 kb)		
ATP7A, DNAJB2, IGHMBP2, PLEGHG5, SIGMAR1		
Dystonia (DYT) *	3 - 5 w	E
Gene panel: ID128.03		
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: 9 genes (27,8 kb)		
ANO3, AOPEP, COL6A3, GNAL, HPCA, KMT2B, THAP1, TOR1A, TUBB4A		
Dystonia (DYT), combined type: 16 genes (28,1 kb)		
ACTB, ADCY5, ATP1A3, ECHS1, GCH1, KCTD17, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SPR, TAF1, TH; VPS16		
Episodic ataxia (EA) *	3 - 5 w	E
Gene panel: ID184.02, 6 genes (20,6 kb)		
ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3		
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		
Fetal akinesia deformation sequence (FADS) *	3 - 5 w	E
Gene panel: ID201.00, 10 genes (30,3 kb)		
CHRNA1, CHRND, CHRNG, DOK7, GBE1, MUSK, MYOD1, NUP88, RAPSN, RYR1		

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Frontotemporal dementia (FTD) * Gene panel: ID310.00 Frontotemporal dementia (FTD): 18 genes (27,9 kb) CCNF, CHCHD10, CHMP2B, CYLD, FUS, GRN, HNRNPA1, HNRNPA2B1, MAPT, OPTN, PSEN1, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Frontotemporal dementia and Amyotrophic Lateralsklerosis (FTDALS): 13 genes (21,2 kb) CCNF, CHCHD10, CHMP2B, CYLD, FUS, OPTN, SQSTM1, TARDBP, TBK1, TIA1, TUBA4A, UBQLN2, VCP Frontotemporal dementia, inclusion body myopathy and Paget disease (IBMPFD): 3 genes (4,6 kb) HNRNPA1, HNRNPA2B1, VCP	3 - 5 w	E
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
Hereditary ataxia, comprehensive diagnostics * Gene panel: ID276.01, 114 genes (334,2 kb) ABCB7, ABHD12, ACO2, AFG3L2, AIFM1, ANO10, APTX, ATCAY, ATG5, ATM, ATP1A3, ATP2B3, ATP8A2, CA8, CACNA1A, CACNA1G, CACNB4, CAPN1, CASK, CCDC88C, CHP1, CLCN2, CLN5, COA7, 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, OPHN1, PCDH12, PCNA, PDYN, PEX7, PHYH, PIK3R5, PLD3, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POLR3B, PRKCG, PRPS1, PTF1A, PUM1, RNF216, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC1A3, SLC25A46, SLC52A2, SLC9A1, SLC9A6, SNX14, SPTBN2, SQSTM1, STUB1, SYNE1, SYT14, TDP1, TDP2, TGM6, THG1L, TMEM240, TPP1, TRPC3, TSFM, TTBK2, TTPA, TUBB4A, TWNK, TXN2, UBA5, VAMP1, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1 Episodic ataxia (EA): 6 genes (20,5 kb) ATP1A3, CACNA1A, CACNB4, KCNA1, SCN2A, SLC1A3 Spastic ataxia (SPAX): 6 genes (24,7 kb) AFG3L2, CHP1, KIF1C, MARS2, MTPAP, NKX6-2, SACS, VAMP1 Spinocerebellar ataxia, autosomal dominant (SCA): 25 genes (85,6 kb) AFG3L2, CACNA1A, CACNA1G, CCDC88C, DAB1, EEF2, ELOVL4, ELOVL5, FAT2, FGF14, GRM1, ITPR1, KCNC3, KCND3, MME, PDYN, PLD3, PRKCG, PUM1, SPTBN2, STUB1, TGM6, TMEM240, TRPC3, TTBK2 Spinocerebellar ataxia, autosomal recessive (SCAR): 24 genes (87,8 kb) ANO10, ATG5, COQ8A, CWF19L1, GDAP2, GRID2, GRM1, PMPCA, RUBCN, SCYL1, SLC9A1, SNX14, SPTBN2, STUB1, SYNE1, SYT14, TDP2, THG1L, TPP1, UBA5, VPS13D, VWA3B, WWOX, XRCC1 Spinocerebellar ataxia with axonal neuropathy (SCAN): 3 genes (10,6 kb) COA7, SETX, TDP1 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	4 - 6 w	E
Hyperekplexia (HKPX) * Gene panel: ID216.00, 9 genes (16,0 kb) ACTL6B, ARHGEF9, ASNS, ATAD1, GLRA1, GLRB, GPHN, SLC6A5, TRAK1	3 - 5 w	E
Hypomyelinating leukodystrophy (HLD) * Gene panel: ID277.00, 22 genes (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11	3 - 5 w	E
Neurodegeneration with brain iron accumulation (NBIA) * Gene panel: ID264.00, 11 genes (20,0 kb) ATP13A2, C19ORF12, CP, COASY, CRAT, FA2H, FTL, PANK2, PLA2G6, REPS1, WDR45	3 - 5 w	E
Neurodevelopmental disorders (NED), comprehensive diagnostics * Gene panel: ID358.00, 169 genes (431,8 kb) ADARB1, ADAT3, ADCY5, AFG2A, AFG2B, AGO1, ANAPC7, ARHGEF2, ATP6V0A1, ATP9A, BCAS3, BPTF, BRAT1, C18ORF32, CACNA1B, CACNA1C, CACNA1I, CAPN15, CDC42BPB, CHAMP1, CHD5, CHKA, CLCN3, COPB1, CPSF3, CSNK2A1, CSNK2B, CTNNB1, CUL3, DEAF1, DHPS, DHX30, DHX37, DLL1, DOHH, DYNC1I2, EMC10, 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
Neuronal ceroid lipofuscinosis (CLN) * Gene panel: ID132.01, 15 genes (20,2 kb) ASAH1, ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, NHLRC1, PPT1, TPP1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Leukodystrophy and Leukoencephalopathy, comprehensive diagnostics * Gene panel: ID204.04 Leukodystrophy and Leukoencephalopathy, comprehensive diagnostics : 122 genes (208,5 kb) AARS2, ABCD1, ACBD5, ACER3, ACOX1, ADAR, AIFM1, AIMP1, AIMP2, ALDH3A2, ARSA, ASPA, AUH, BOLA3, CLCN2, CLDN11, CNP, COA7, COA8, COL4A1, COL4A2, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS1, DARS2, DEGS1, EARS2, EIF2AK1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS1, FA2H, FAM126A, FDX2, FOLR1, FOXRED1, FUCA1, GALC, GBE1, GCDH, GFAP, GFM1, GJC2, GLB1, HEPACAM, HIKESHI, HMGCL, HSD17B4, HSPD1, HTRA1, IBA57, IFIH1, ISCA1, ISCA2, KARS1, KCNT1, L2HGDH, LMNB1, LYRM7, MARS2, MCOLN1, MLC1, MTFMT, NAXD, NAXE, NFU1, NKX6-2, NOTCH3, NUBPL, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PLAA, PLEKHG2, PLP1, PMPCB, POLR1C, POLR3A, POLR3B, POLR3K, PSAP, PYCR2, RARS1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SAMHD1, SCP2, SDHAF1, SLC13A3, SLC16A2, SLC17A5, SOX10, SPTAN1, STN1, SUMF1, TMEM106B, TMEM63A, TREM2, TREX1, TUBB4A, TYMP, TYROBP, UFM1, VPS11, ZFYVE26 Leukodystrophy with hypomyelination (HLD): 22 genes (35,0 kb) AIMP1, AIMP2, CLDN11, CNP, DEGS1, EPRS1, FAM126A, GJC2, HIKESHI, HSPD1, PLP1, POLR1C, POLR3A, POLR3B, POLR3K, PYCR2, RARS1, TMEM63A, TMEM106B, TUBB4A, UFM1, VPS11 Leukodystrophy with peroxisome biogenesis disorder (PBD): 15 genes (20,8 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26, PHYH Orthochromatic leukodystrophy: 10 genes (14,6 kb) ASPA, CSF1R, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, GFAP, HEPACAM, MLC1 Metachromatic leukodystrophy: 3 genes (4,2 kb) ARSA, PSAP, SUMF1 Aicardi-Goutières syndrome (AGS): 7 genes (11,9 kb) ADAR, IFIH1, RNASE2A, RNASE2B, RNASE2C, SAMHD1, TREX1 CADASIL, CARASIL: 2 genes (8,4 kb) HTRA1, NOTCH3	4 - 6 w	E
Parkinson disease (PARK) * Gene panel: ID077.02 Parkinson disease (PARK): 37 genes (93,8 kb) ADH1C, ATP13A2, ATP1A3, ATP6AP2, CHCHD2, DCTN1, DNAJC6, EIF4G1, FBXO7, GBA1, GCH1, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, PARK7, PINK1, PLA2G6, POLG, PRKRA, PRKN, PSAP, RAB39B, SLC18A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SYNJ1, TAF1, TH, TRPM7, UCHL1, UQCRC1, VPS13C, VPS35 Parkinson disease (PARK), early-onset: 9 genes (30,3 kb) ATP13A2, DNAJC6, FBXO7, PARK7, PINK1, PLA2G6, PRKN, SYNJ1, VPS13C Parkinson disease (PARK), late-onset: 12 genes (27,3 kb) ADH1C, CHCHD2, EIF4G1, GBA1, GIGYF2, GLUD2, HTRA2, LRRK2, MAPT, SNCA, UCHL1, VPS35 Dystonia-parkinsonism (DYT): 9 genes (18,3 kb) ATP1A3, GCH1, PRKRA, SLC6A3, SLC18A2, SLC30A10, SLC39A14, TAF1, TH	3 - 5 w	E
Paroxysmal dyskinesia * Gene panel: ID286.00, 14 genes (38,9 kb) ADCY5, ATP1A2, ATP1A3, CACNA1A, GCH1, KCNA1, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2, SCN8A, SLC2A1, TBC1D24	3 - 5 w	E
Peroxisome biogenesis disorder (PBD) * Gene panel: ID083.01 Peroxisome biogenesis disorder (PBD): 14 genes (19,9 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Zellweger syndrome (PBD, type A): 12 genes (18,3 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26 Neonatal adrenoleukodystrophy/ Infantile refsum syndrome (PBD, type B): 11 genes (17,0 kb) PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX11B, PEX13, PEX16, PEX26 Heimler syndrome (PBD, type C): 2 genes (6,8 kb) PEX1, PEX6	3 - 5 w	E
Sensory and autonomic neuropathy (HSAN, HSN) * Gene panel: ID086.01, 15 genes (52,5 kb) ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A, SCN11A, SPTLC1, SPTLC2, WNK1	3 - 5 w	E
Spinal muscular atrophy (SMA) * Gene panel: ID152.01 Spinal muscular atrophy (SMA): 31 genes (69,5 kb) ASAH1, ASCC1, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DYNC1H1, EXOSC3, EXOSC8, EXOSC9, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, SMN1, SMN2, TRIP4, TRPV4, UBA1, VAPB, VRK1, WARS1 Proximal spinal muscular atrophy (SMA): 10 genes (26,6 kb) ASAH1, ASCC1, BICD2, CHCHD10, DYNC1H1, SMN1, SMN2, TRIP4, UBA1, VAPB Distal spinal muscular atrophy (DSMA, HMN): 18 genes (41,9 kb) ATP7A, BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2, SIGMAR1, PLEKHG5, REEP1, SETX, SLC5A7, TRPV4, UBA1, WARS1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Neurodegenerative Diseases		
Spastic paraplegia (HSP, SPG) *	4 - 6 w	E
Gene panel: ID148.03		
Spastic paraplegia (HSP, SPG): 58 genes (117,9 kb)		
ALDH18A1, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C19ORF12, CAPN1, CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, HSPD1, IBA57, KIF1A, KIF5A, L1CAM, MAG, MTRFR, NIPA1, NT5C2, PCYT2, PLP1, PNPLA6, REEP1, REEP2, RTN2, SELENOI, SLC33A1, SPART, SPAST, SPG7, SPG11, SPG21, TECPR2, TFG, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26, ZFYVE27		
Spastic paraplegia (SPG), autosomal dominant: 17 genes (33,9 kb)		
ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1, KIF1A, KIF5A, NIPA1, REEP1, REEP2, RTN2, SLC33A1, SPAST, SPG7, UBAP1, WASHC5, ZFYVE27		
Spastic paraplegia (SPG), autosomal recessive: 44 genes (92,3 kb)		
AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATP13A2, B4GALNT1, C19ORF12, CAPN1, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2, HPDL, IBA57, KIF1A, L1CAM, MAG, MTRFR, NT5C2, PCYT2, PLP1, PNPLA6, REEP2, SELENOI, SPART, SPG7, SPG11, SPG21, TECPR2, TFG, UCHL1, VPS37A, ZFYVE26		
Spastic paraplegia (SPG), X-linked: 2 genes (4,6 kb)		
L1CAM, PLP1		
Spastic ataxia (SPAX) *	3 - 5 w	E
Gene panel: ID228.00, 12 genes (34,8 kb)		
AFG3L2, CAPN1, CHP1, GJC2, KIF1C, MARS2, MTPAP, NKX6-2, POLR3A, SACS, SPG7, VAMP1		
Tuberous sclerosis (TSC) *	2 - 4 w	E
Gene panel: ID332.00, 2 genes (8,9 kb)		
TSC1, TSC2		
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		
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		
Primary ciliar dyskinesia with or without situs inversus (PCD, CILD) *	4 - 6 w	E
Gene panel: ID085.02, 42 genes (132,9 kb)		
ARMC4, CCDC103, CCDC39, CCDC40, CCDC65, CCDC114, CCDC151, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH5, DNAH9, DNAH11, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, HYDIN, LRRC6, LRRC56, MCIDAS, NEK10, NME8, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, TTC12, TTC25, ZMYND10		
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		

Diseases/Diagnostics	TAT	Material
Reproductive Disorders		
Adrenogenital syndrome (AGS, CAH) * Gene panel: ID111.02, 7 genes (10,1 kb) CYP11A1, CYP11B1, CYP17A1, CYP21A2, HSD3B2, POR, STAR	3 - 5 w	E
Disorder of sex development (DSD) * Gene panel: ID117.03 Disorder of sex development (DSD): 49 genes (96,7 kb) AKR1C2, AMH, AMHR2, ANOS1, AR, ARX, ATRX, CBX2, CDKN1C, CHD7, CTU2, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DHX37, DMRT1, GATA4, HHAT, HOXA13, HSD17B3, HSD3B2, LHCGR, MAMLD1, MAP3K1, MYRF, 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	4 - 6 w	E
Female infertility (PREMBL, OOMD) * Gene panel: ID239.01, 12 genes (18,0 kb) BTG4, PADI6, PANX1, PATL2, REC114, TLE6, TRIP13, TUBB8, WEE2, ZP1, ZP2, ZP3	3 - 5 w	E
Hypogonadotropic hypogonadism with or without anosmia (KAL, HH) * Gene panel: ID170.04, 37 genes (62,5 kb) ANOS1, CHD7, CPE, DUSP6, FEZF1, FGFR1, FGF8, FGF17, FLRT3, FSHB, GNRH1, GNRHR, HESX1, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NDNF, NHLH2, NSMF, PNPLA6, POLR3B, PROK2, PROKR2, RNF216, SEMA3A, SOX10, SOX2, SOX11, SPRY4, TAC3, TACR3, TCF12, WDR11	3 - 5 w	E
Male infertility (SPGF, CBAVD) * Gene panel: ID192.04 Male infertility (SPGF, CBAVD): 81 genes (311,0 kb) ACTL9, ADGRG2, AK7, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC62, CEP112, CFAP43, CFAP44, CFAP47, CFAP58, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, CFTR, DNAH1, DNAH2, DNAH8, DNAH10, DNAH17, DNHD1, DPY19L2, DZIP1, FANCM, FBXO43, FSIP2, IFT74, GCNA, GGN, KLHL10, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPAG17, SPATA16, SPEF2, SPINK2, STAG3, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TERB1, TERB2, TEX11, TEX14, TEX15, TDRD9, TSGA10, TTC21A, TTC29, USP9Y, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Spermatogenic failure (SPGF): 79 genes (303,5 kb) ACTL9, AK7, ARMC2, AURKC, BRDT, C14ORF39, C2CD6, CATIP, CATSPER1, CCDC62, CEP112, CFAP43, CFAP44, CFAP47, CFAP58, CFAP65, CFAP69, CFAP70, CFAP91, CFAP251, DNAH1, DNAH2, DNAH8, DNAH10 DNAH17, DNHD1, DPY19L2, DZIP1, FANCM, FBXO43, FSIP2, IFT74, GCNA, GGN, KLHL10, M1AP, MEIOB, MOV10L1, MSH4, MSH5, NANOS1, NR5A1, PDHA2, PLCZ1, PMFBP1, PNLDC1, PPP2R3C, QRICH2, RNF212, RPL10L, SEPTIN12, SHOC1, SLC26A8, SOHLH1, SPAG17, SPATA16, SPEF2, SPINK2, STAG3, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TERB1, TERB2, TEX11*, TEX14, TEX15, TDRD9, TSGA10, TTC21A, TTC29, USP9Y, WDR19, XRCC2, ZMYND15, ZPBP, ZSWIM7 Congenital bilateral aplasia of vas deferens (CBAVD): 2 genes (7,5 kb) ADGRG2, CFTR	4 - 6 w	E
Ovarian dysgenesis (ODG) * Gene panel: ID293.01 Ovarian dysgenesis (ODG): 16 Gene (27,8 kb) BMP15, CLPP, ERAL1, ESR2, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, TWNK, ZSWIM7 XX gonadal dysgenesis (ODG): 10 Gene (17,1 kb) BMP15, ESR2, FSHR, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, ZSWIM7 XX gonadal dysgenesis with deafness (PRLTS): 6 genes (10,7 kb) CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK	3 - 5 w	E
Premature ovarian failure (POF) * Gene panel: ID078.05 Premature ovarian failure (POF): 23 Gene (51,1 kb) BMP15, BNC1, C14ORF39, DIAPH2, ERCC6, FSHR, GDF9, FANCM, FIGLA, FMR1, FOXL2, HFM1, HSF2BP, INHA, LHCGR, MCM8, MSH5, NOBOX, NR5A1, POF1B, STAG3, SYCE1, XRCC2 Premature ovarian failure (POF), X-linked: 4 genes (8,2 kb) BMP15, DIAPH2, FMR1, POF1B Premature ovarian failure (POF), autosomal recessive: 10 genes (25,0 kb) C14ORF39, GDF9, FANCM, HFM1, HSF2BP, MCM8, MSH5, STAG3, SYCE1, XRCC2 Premature ovarian failure (POF), autosomal dominant: 6 genes (12,7 kb) BNC1, ERCC6, FIGLA, FOXL2, NOBOX, NR5A1	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Acrocephalosyndactyly (ACS) *		
Gene panel: ID311.00, 6 genes (17,0 kb)	3 - 5 w	E
FGFR1, FGFR2, FGFR3, MEGF8, RAB23, TWIST1		
Adams-Oliver syndrome (AOS) *		
Gene panel: ID259.00, 6 genes (23,2 kb)	3 - 5 w	E
ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ		
Arachnodactyly *		
Gene pnel: ID124.00, 13 genes (35,9 kb)	3 - 5 w	E
CHST14, CTSC, DSE, EFEMP2, FBN1, FBN2, SCARF2, SKI, TGFB2, TGFB3, TGFBR1, TGFBR2, SMAD3		
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)	4 - 6 w	E
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		
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	3 - 5 w	E
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		
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	3 - 5 w	E
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		
Distal arthrogryposis (DA) *		
Gene panel: ID196.01, 11 genes (40,5 kb)	3 - 5 w	E
ECEL1, FBN2, MYBPC1, MYH3, MYH8, MYLPF, PIEZO2, TNNI2, TNNT3, TPM2, UBA1		
Fetal akinesia deformation sequence (FADS) *		
Gene panel: ID201.00, 10 genes (30,3 kb)	3 - 5 w	E
CHRNA1, CHRND, CHRNG, DOK7, GBE1, MUSK, MYOD1, NUP88, RAPSN, RYR1		
Fraser syndrome (FRASRS)		
Gene panel: ID317.00, 3 genes (24,8 kb)	3 - 5 w	E
FRAS1, FREM2, GRIP1		
Frontonasal dysplasia (FND) *		
Gene panel: ID339.00, 11 genes (22,1 kb)	3 - 5 w	E
ALX1, ALX3, ALX4, ANKH, EFNB1, FGFR1, FGFR2, FGFR3, GLI3, TWIST1, ZSWIM6		
Heart-hand syndrome *		
Gene panel: ID165.01, 9 genes (23,8 kb)	3 - 5 w	E
DACT1, GATA6, LMNA, RBM8A, RECQL4, SALL1, SALL4, TBX3, TBX5		

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Hand malformations, comprehensive diagnostics * Gene panel: ID298.00, 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 Weill-Marchesani syndrome (WMS): 4 genes (20,7 kb) ADAMTS10, ADAMTS17, FBN1, LTBP2	4 - 6 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
Klippel-Feil syndrome (KFS) * Gene panel: ID207.00, 5 genes (12,5 kb) GDF3, GDF6, MEOX1, MYO18B, PAX1	3 - 5 w	E
Lethal congenital contracture syndrome (LCCS) * Gene panel: ID197.00, 12 genes (34,6 kb) ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42	3 - 5 w	E
Mandibulofacial dysostosis (MFD) * Gene panel: ID188.01 Mandibulofacial dysostosis (MFD): 11 genes (22,0 kb) DHODH, EDNRA, EFTUD2, POLR1A, POLR1B, POLR1C, POLR1D, RPS28, SF3B4, TCOF1, TSR2 Treacher-Collins syndrome (TCS): 4 genes (9,4 kb) POLR1B, POLR1C, POLR1D, TCOF1	3 - 5 w	E
Multiple epiphyseal dysplasia (EDM) * Gene panel: ID202.01, 10 genes (25,9 kb) CANT1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, EIF2AK3, KIF7, MATN3, SLC26A2	3 - 5 w	E
Multiple pterygium syndrome * Gene panel: ID158.01, 8 genes (16,8 kb) CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4	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

Diseases/Diagnostics	TAT	Material
Skeletal and Bone Diseases		
Orofacial cleft (OFC) *	4 - 6 w	E
Gene panel: ID266.00		
Orofacial cleft (OFC): 41 genes (129,7 kb)		
ARHGAP29, BMP4, C2CD3, CDH1, CHD7, CPLANE1, CTNND1, DDX59, DHCR7, DLX4, ESCO2, FGFR1, FGFR2, FLNB, GRHL3, HDAC8, IFT57, INTU, IRF6, KDM6A, KIAA0753, KMT2D, MEIS2, MID1, MSX1, NECTIN1, NIPBL, OFD1, RAD21, RIPK4, SEMA3E, SLC26A2, SMC1A, SMC3, SPECC1L, SUMO1, TBX22, TCTN3, TGDS, TMEM107, TP63		
Orofacial cleft (OFC), nonsyndromic: 10 genes (15,4 kb)		
ARHGAP29, BMP4, DLX4, GRHL3, IRF6, MSX1, NECTIN1, SUMO1, TBX22, TP63		
Orofaciodigital syndrome (OFD): 9 genes (29,7 kb)		
C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107		
Cornelia de Lange syndrome (CDLS): 5 genes (18,8 kb)		
HDAC8, NIPBL, RAD21, SMC1A, SMC3		
Kabuki syndrome (KABUK): 2 genes (20,8 kb)		
KDM6A, KMT2D		
CHARGE syndrome: 3 genes (12,9 kb)		
CHD7, SEMA3E, TBX22		
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: ID115.01, 13 genes (29,4 kb)		
CALCR, COL1A1, COL1A2, ESR1, LRG4, LRP5, PLS3, SGMS2, SLC34A1, SLC9A3R1, UGT2B17, VDR, WNT1		
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)	3 - 5 w	E
ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1, RBPJ		
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	3 - 5 w	E
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		
Amelogenesis imperfecta (AI) *		
Gene panel: ID232.01, 17 genes (29,4 kb)	3 - 5 w	E
ACP4, AMBN, AMELX, AMTN, DLX3, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, MMP20, ODAPH, RELT, SLC24A4, WDR72		
Cowden syndrome (CWS) *		
Gene panel: ID075.01, 8 genes (12,8 kb)	3 - 5 w	E
AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1		
Cutaneous malignant melanoma (CMM) *		
Gene panel: ID193.01, 12 genes (26,4 kb)	3 - 5 w	E
BAP1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, PTEN, TERT, TP53, TYR, XRCC3		
Cutis laxa (ARCL, ADCL) *		
Gene panel: ID109.02, 11 genes (27,9 kb)	3 - 5 w	E
ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, EFEMP2, ELN, FBLN5, LTBP1, LTBP4, PYCR1		
Dyskeratosis congenita (DKC) *		
Gene panel: ID347.01, 15 genes (24,1 kb)	3 - 5 w	E
ACD, CTC1, DCLRE1B, DKC1, ENOSF1, NHP2, NOP10, PARN, RTEL1, TERC, TERT, TINF2, TYMS, USB1, WRAP53		
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)	4 - 6 w	E
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		
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)	4 - 6 w	E
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		

Diseases/Diagnostics	TAT	Material
Skin and Dental Diseases		
Ectodermal dysplasia (ECTD) * Gene panel: ID136.04 Ectodermal dysplasia (ECTD): 44 genes (74,6 kb) AP1B1, CDH3, CHUK, CST6, DLX3, DSG4, EDA, EDAR, EDARADD, GJA1, GJB2, GJB6, GRHL2, HOXC13, IKBKG, KDF1, KREMEN1, KRT14, KRT16, KRT17, KRT74, KRT81, KRT83, KRT85, KRT86, LEF1, LRP6, MSX1, NECTIN1, NECTIN4, NFKBIA, PKP1, PORCN, PRKD1, RIPK4, SMARCAD1, SREBF1, TBX3, TP63, TRAF6, TRPS1, TSPEAR, TWIST2, WNT10A Ectodermal 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	3 - 5 w	E
Gorlin-Goltz syndrome (BCNS) * Gene panel: ID174.00, 3 genes (9,4 kb) PTCH1, PTCH2, SUFU	2 - 4 w	E
Hereditary angioedema (HAE) * Gene panel: ID345.00, 7 genes (16,4 kb) ANGPT1, F12, HS3ST6, KNG1, MYOF, PLG, SERPING1	3 - 5 w	E
Hermansky-Pudlak syndrome (HPS) * Gene panel: ID289.00, 11 genes (22,6 kb) AP3B1, AP3D1, BLOC1S3, BLOC1S5, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6	3 - 5 w	E
Hydrops fetalis * Gene panel: ID370.00, 148 genes (404,7 kb) ACAD9, AHCY, ALG1, ALG12, ALG8, ALG9, ALPK3, ANGPT2, ARSB, ASAH1, ATP1A2, BRAF, CALCRL, CBL, CCBE1, CDAN1, CEP55, CFH, CHD7, CHRNA1, CHRND, 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	3 - 5 w	E
Hypotrichosis, nonsyndromic type (HYPT) * Gene panel: ID146.01, 12 genes (20,1 kb) APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE	3 - 5 w	E
Ichthyosis * Gene panel: ID217.01 Ichthyosis: 38 genes (73,0 kb) ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, AP1B1, AP1S1, ASPRV1, CASP14, CERS3, CLDN1, CLDN10, CSTA, CYP4F22, ELOVL4, ERCC2, FLG, GJB2, KRT1, KRT10, KRT2, LIPN, LORICRIN, MBTPS2, NIPAL4, PEX7, PHYH, PNPLA1, POMP, SDR9C7, SLC27A4, SNAP29, SPINK5, SREBF1, ST14, STS, SULT2B1, TGM1 Congenital ichthyosis, autosomal recessive (ARCI): 14 genes (28,7 kb) ABCA12, ALOX12B, ALOXE3, CASP14, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, SDR9C7, SLC27A4, ST14, SULT2B1, TGM1 Ichthyosis, autosomal dominant or X-linked: 7 genes (21,3 kb) ASPRV1, FLG, GJB2, KRT1, KRT2, KRT10, STS	3 - 5 w	E
Multiple pterygium syndrome * Gene panel: ID158.01, 8 genes (16,8 kb) CHRNA1, CHRNB1, CHRND, CHRNG, IRF6, LMX1B, MYH3, RIPK4	3 - 5 w	E
Neurofibromatosis (NF) #,* Gene panel: ID210.00, 3 genes (11,6 kb) NF1, NF2, SPRED1	3 - 5 w	E
Oculocutaneous albinism (OCA) * Gene panel: ID082.02, 9 genes (13,3 kb) DCT, GPR143, LRMDA, MC1R, OCA2, SLC24A5, SLC45A2, TYR, TYRP1	3 - 5 w	E
Orofaciodigital syndrome (OFD) * Gene panel: ID265.00, 9 genes (29,7 kb) C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1, TCTN3, TMEM107	3 - 5 w	E
Pachyonychia congenita (PC) * Gene panel: ID120.01, 13 genes (30,1 kb) AAGAB, ATP2A2, DSG1, DSP, GJB6, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, MBTPS2, TRPV3	3 - 5 w	E
Peeling skin syndrome (PSS) * Gene panel: ID309.00, 6 genes (13,6 kb) CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Skin and Dental Diseases		
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		
Progeria and progeroid syndromes *	3 - 5 w	E
Gene panel: ID147.00, 27 genes (67,0 kb)		
ALDH18A1, ANAPC1, B3GALT6, B4GALT7, BANF1, BLM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, FBN1, GORAB, LMNA, MDM2, MTX2, NAA10, POLD1, POLR3A, POLR3GL, PYCR1, RECQL4, SLC25A24, WRN, ZMPSTE24		
Selective tooth agenesis (STHAG) *	3 - 5 w	E
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		
Systemic diseases with multiple café-au-lait spots *	3 - 5 w	E
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		
Tuberous sclerosis (TSC) *	2 - 4 w	E
Gene panel: ID332.00, 2 genes (8,9 kb)		
TSC1, TSC2		
Xeroderma pigmentosum (XP) *	3 - 5 w	E
Gene panel: ID282.00, 10 genes (23,5 kb)		
DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC		
Tumor Syndromes		
BRCA1 and BRCA2-associated cancer #,*	2 - 4 w	E
Gene panel: ID001.00, 2 genes (15,8 kb)		
BRCA1, BRCA2		
Breast cancer *	2 - 4 w	E
Gene panel: ID021.02, 12 genes (41,0 kb)		
ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53		
Breast and ovarian cancer (HBOC) *	3 - 5 w	E
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		
Breast and ovarian cancer, comprehensive diagnostics *	4 - 6 w	E
Gene panel: ID068.01, 50 genes (151,5 kb)		
ABRAXAS1, ATM, BARD1, BLM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, MLH1, MRE11A, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, POLD1, POLE, PTEN, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RECQL, RECQL4, SDHB, SDHC, SDHD, SLX4, SMARCA4, STK11, TP53, XRCC2		
Chromosomal instability syndromes *	4 - 6 w	E
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		

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
Colorectal cancer (CRC) * Gene panel: ID049.01, 14 genes (40,4 kb) ATM, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11	3 - 5 w	E
Colorectal cancer and polyposis * Gene panel: ID006.08 Colorectal cancer and polyposis: 22 genes (62,3 kb) APC, ATM, AXIN2, BMPR1A, CHEK2, EPCAM, FLCN, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53 Colorectal polyposis: 14 genes (37,6 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11 Colorectal cancer: 14 genes (40,4 kb) ATM, CHEK2, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11	3 - 5 w	E
Colorectal cancer, comprehensive diagnostics * Gene panel: ID285.01, 33 genes (87,9 kb) APC, ATM, AXIN2, BLM, BMPR1A, CDH1, CHEK2, EPCAM, EXO1, FLCN, GALNT12, GREM1, MLH1, MLH3, MSH2, MSH3, MSH6, MUTYH, NBN, NTHL1, PLA2G2A, PMS2, POLD1, POLE, PTEN, RFC1, RPA1, RNF43, RPS20, SMAD4, STK11, TGFBR2, TP53	3 - 5 w	E
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
Constitutional mismatch repair deficiency syndrome (CMMRDS, MMRCS) * Gene panel: ID362.00, 5 genes (12,7 kb) EPCAM, MLH1, MSH2, MSH6, PMS2	3 - 5 w	E
Cowden syndrome (CWS) * Gene panel: ID075.01, 8 genes (12,8 kb) AKT1, PIK3CA, PTEN, SEC23B, SDHB, SDHC, SDHD, WWP1	3 - 5 w	E
Cutaneous maligne 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: ID090.03, 24 genes (71,9 kb) APC, ATM, BMPR1A, BRCA1, BRCA2, CDH1, CHEK2, CTNNA1, EPCAM, KIT, 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

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
Gorlin-Goltz syndrome (BCNS) * Gene panel: ID174.00, 3 genes (9,4 kb) PTCH1, PTCH2, SUFU	2 - 4 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	2 - 4 w	E
Medulloblastoma * Gene panel: ID205.02, 22 genes (75,7 kb) APC, BRCA2, CHEK2, DICER1, ELP1,EPCAM, ERCC2, FANCM, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTCH1, PTCH2, PTEN, SMARCB1, SMARCA4, SUFU, TP53, VHL	3 - 5 w	E
Myelodysplastic syndrome (MDS) and Acute myeloid leukemia (AML) * Gene panel: ID321.01 Myelodysplastic syndrome (MDS) and Acute myeloid leukemia (AML): 121 genes (244,1 kb) ACD, ADA2, ADH5, ALDH2, ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, BRIP1, CBL, CEBPA, CHEK2, CLPB, CSF3R, CTC1, DCLRE1B, DDX41, DKC1, 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
Neurofibromatosis (NF) #,* Gene panel: ID210.00, 3 genes (11,6 kb) NF1, NF2, SPRED1	3 - 5 w	E
Ovarian cancer * Gene panel: ID004.04, 14 genes (45,5 kb) BRCA1, BRCA2, BRIP1, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, SMARCA4, STK11, TP53	3 - 5 w	E
Pancreatic cancer * Gene panel: ID089.03, 17 genes (56,2 kb) APC, ATM, BRCA1, BRCA2, CDKN2A, CTRC, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PRSS1, SPINK, STK11, TP53, VHL	3 - 5 w	E
Pheochromocytoma/paraganglioma syndrome (PPGL) * Gene panel: ID042.02, 16 genes (29,4 kb) DLST, FH, KIF1B, GDNF, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, VHL	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: ID005.05, 14 genes (37,6 kb) APC, AXIN2, BMPR1A, FLCN, GREM1, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11	3 - 5 w	E
Prostate cancer * Gene panel: ID140.01, 28 genes (91,1 kb) AR, ATM, ATR, BAP1, BRCA1, BRCA2, CYP3A43, CDH1, CHEK2, EHBP1, ELAC2, EPCAM, HOXB13, MLH1, MRE11A, MSR1, MSH2, MSH6, MSMB, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, RNASEL, TP53, TRRAP	3 - 5 w	E

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
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: ID223.01, 37 genes (107,5 kb) APC, ATM, ATR, BLM, CDKN1C, CDKN2A, CHEK2, DICER1, EPCAM, ERCC2, EXT1, EXT2, FH, HRAS, KIT, MLH1, MSH2, MSH6, MTAP, NBN, NF1, PDGFRA, PMS2, PRKAR1A, PTCH1, RB1, RECQL4, SDHA, SDHB, SDHC, SDHD, SQSTM1, SUFU, TNFRSF11A, TP53, WRN, ZNF687	4 - 6 w	E
Tumor syndromes, comprehensive diagnostics * Gene panel: ID018.03, 190 genes (454,1 kb) ABRAXAS1, ACD, AIP, AKT1, ALK, ANAPC1, ANKRD26, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CCND1, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CYLD, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, EFL1, EGFR, ELAC2, ELP1, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ETV6, EXT1, EXT2, EZH2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, FOXE1, GALNT12, GATA2, GPC3, GREM1, HABP2, HAVCR2, HNF1A, HNF1B, HOXB13, HRAS, IKZF1, KIF1B, KIT, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MAX, MC1R, MEN1, MET, MINPP1, MITF, MLH1, MLH3, MRAS, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NDUFA13, NF1, NF2, NKX2-1, NOP10, NHP2, NRAS, NSD1, NTHL1, PALB2, PALLD, PARN, PAX5, PCNA, PDGFRA, PHOX2B, PIK3CA, PMS1, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PTCH1, PTCH2, PTEN, PTPN11, RABL3, RAD50, RAD51, RAD51C, RAD51D, RAF1, RASA2, RB1, RECQL, RECQL4, REST, RET, RFWD3, RHBDF2, RINT1, RIT1, RNASEL, RNF139, RNF43, RPS20, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SOS1, SOS2, SPRED1, SRGAP1, STK11, SUFU, TERT, TINF2, TMEM127, TOP3A, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2, XRCC3 Tumor syndromes in childhood: 128 genes (317,3 kb) ACD, ALK, APC, ATM, BAP1, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, BUB1B, CBL, CDC73, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, DDB2, DDX41, DICER1, DIS3L2, DKC1, DLST, EFL1, ELP1, EPCAM, 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, MAP2K1, MAP2K2, MAPK1, MRAS, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NKX2-1, NOP10, NHP2, NRAS, NSD1, PALB2, PARN, PAX5, PHOX2B, PMS2, POLH, PRKAR1A, PTCH1, PTCH2, PTEN, PTPN11, RAD51, RAD51C, RAF1, RB1, RECQL4, REST, RET, RIT1, RRAS2, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, SOS2, STK11, SUFU, TERT, TMEM127, TINF2, TP53, TRIM28, TRIM37, TRIP13, TSC1, TSC2, UBE2T, VHL, WRAP53, WRN, WT1, XPA, XPC Tumor syndromes in adulthood: 131 genes (309,8 kb) AKT1, APC, ATM, BAP1, BARD1, BMPR1A, BRAF, BRCA1, BRCA2, BRIP1, CBL, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, DDB2, DICER1, DLST, EGFR, ELAC2, EPCAM, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FH, FLCN, FOXE1, GALNT12, GPC3, GREM1, HABP2, HNF1A, HNF1B, HOXB13, HRAS, KIF1B, KIT, KRAS, LZTR1, MAD2L2, MAP2K1, MAP2K2, MAPK1, MAX, MC1R, MEN1, MET, MINPP1, MITF, MLH1, MLH3, MRAS, MSH2, MSH3, MSH6, MUTYH, NF1, NF2, NKX2-1, NRAS, NTHL1, PALB2, PDGFRA, PIK3CA, PMS1, PMS2, POLD1, POLE, POLH, POT1, PTCH1, PTCH2, PTEN, PTPN11, RABL3, RAD51, RAD51C, RAD51D, RAF1, RB1, RET, RFWD3, RHBDF2, RIT1, RNASEL, RNF139, RNF43, RRAS2, RTEL1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SHOC2, SLC25A11, SLX4, SMAD4, SMARCA4, SMARCB1, SOS1, SOS2, SRGAP1, STK11, SUFU, TERT, TMEM127, TP53, TSC1, TSC2, UBE2T, VHL, WRAP53, WT1, XPA, XPC, XRCC2, XRCC3	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

Diseases/Diagnostics	TAT	Material
Tumor Syndromes		
Thyroid cancer *	3 - 5 w	E
Gene panel: ID220.01, 26 genes (54,7 kb)		
AKT1, APC, CDC73, CDKN1B, CHEK2, DICER1, FOXE1, HABP2, MAP2K5, MEN1, MET, MINPP1, NDUFA13, NKX2-1, NTRK1, PIK3CA, PRKAR1A, PTEN, RET, SDHA, SDHB, SDHC, SDHD, SEC23B, SRGAP1, TP53		
Urothelial cancer *	4 - 6 w	E
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		
Wilms tumor (WT) *	3 - 5 w	E
Gene panel: ID335.00, 21 genes (70,4 kb)		
AMER1, ASXL1, BLM, BRCA2, BUB1B, CDC73, CDKN1C, CEP57, CTR9, DICER1, DIS3L2, GPC3, GPC4, NSD1, PALB2, POU6F2, REST, TRIM28, TRIM37, TRIP13, WT1		
Xeroderma pigmentosum (XP) *	3 - 5 w	E
Gene panel: ID282.00, 10 genes (23,5 kb)		
DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC		
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		

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, 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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		

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