
SYNLAB
Human Genetic
NGS Panel Diagnostics

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Brain Malformations

☐ **Cerebellar dysgenesis, X-linked**
Panel-ID: ID219.00 (small)
11 genes (24540 kb): ABCB7, CASK,**, DKC1, FMR1, L1CAM*,**, MECP2*,**, MID1*,**, OFD1*, OPHN1, SLC9A6, ZIC3*

☐ **Complex cortical dysplasia (CDCBM)**
Panel-ID: ID271.00 (small)
11 genes (24030 kb): APC2, CTNNA2, KIF2A, KIF5C, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B*, TUBB3*, TUBG1*
Basis-Panel (Tubulinopathies):
7 gene (9426 bp): TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3*, TUBG1*

☐ **Holoprosencephaly (HPE)**
Panel-ID: ID169.02 (small)
17 genes (48792 kb): CDON, CNOT1, DLL1, DISP1, FGF8, FGFR1,**, GAS1, GLI2**, PRRX1, PTCH1*,**, SHH*,**, SIX3**, STAG2, STIL*,**, TGIF1**, WDR62,***, ZIC2***

☐ **Lissencephaly (LIS)**
Panel-ID: ID133.00 (small)
12 genes (46416 kb): ARX,**, CDK5, CEP85L, DCX*,**, KATNB1, LAMB1, MACF1, NDE1, PAFAH1B1*,**, RELN, TMTC3, TUBA1A*

☐ **Polymicrogyria**
Panel-ID: ID176.01 (small)
15 genes (41952 kb): ADGRG1, AKT3, CCND2, COL3A1*,**, FIG4, KIFBP, OCLN*, RTTN, PI4KA, PIK3CA*, PIK3R2, TUBA1A, TUBA8, TUBB2B*, WDR62*,***

☐ **Schizencephaly**
Panel-ID: ID173.00 (small)
7 genes (19737 kb): COL4A1, COL4A2, COLGALT1, EMX2, SHH,**, SIX3, WDR62*,***

☐ **Neuronal migration disorders, comprehensive diagnostics**
Panel-ID: ID180.00 (large)
80 genes (253977 kb): ACTB, ACTG1, ADGRG1, AKT3, APC2*, ARF1, ARFGEF2, ARX*,**, B3GALNT2*, B4GAT1, CCND2, CDK5, CEP85L, COL3A1*,**, COL4A1, COL4A2, COLGALT1, CRADD, CTNNA2, DAG1, DCHS1, DCX*,**, DYNC1H1, EML1, EMX2*, ERMARD, FAT4, FH*,**, FIG4, FKRP*, FKTN*, FLNA*,**, GMPPB, ISPD*, KATNB1, KIF1BP, KIF2A, KIF5C, 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*,***

Basis-Panel I (Periventricular nodular heterotopia (PVNH)):
5 genes (24099 bp): ARF1, ARFGEF2, ERMARD, FLNA,**, MAP1B, NEDD4L*

Basis-Panel II (Complex cortical dysplasia (CDCBM)):
11 genes (24030 bp): APC2, CTNNA2, KIF2A, KIF5C, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B*, TUBB3*, TUBG1*

Basis-Panel III (Lissencephaly (LIS):
10 genes (24762 bp): ARX,**, CDK5, CEP85L, DCX*,**, KATNB1, NDE1, PAFAH1B1*,**, RELN, TMTC3, TUBA1A*

Basis-Panel IV (Polymicrogyria):
11 genes (25311 bp): ADGRG1, AKT3, CCND2, KIFBP, OCLN*, RTTN, PIK3R2, TUBA1A, TUBA8, TUBB2B*, WDR62*,***

Basis-Panel V (Schizencephaly):
7 genes (19737 bp): COL4A1, COL4A2, COLGALT1, EMX2, SHH,**, SIX3, WDR62*,***

Basis-Panel VI (Walker-Warburg syndrome (MDDGA)):
14 genes (23682 bp): B3GALNT2, B4GAT1, DAG1, FKRP*, FKTN*, GMPPB, ISPD*, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1*

Epilepsy and Migraine

☐ **Aicardi-Goutìeres syndrome (AGS)**
Panel-ID: ID058.00 (small)
7 genes (11898 kb): ADAR, IFIH1*, RNASEH2A*,**, RNASEH2B*,**, RNASEH2C*,**, SAMHD1*,**, TREX1*,***

Microcephaly and Macrocephaly

☐ **Pontocerebellar hypoplasia (PCH)**
Panel-ID: ID071.01 (small)
19 genes (42006 kb): AMPD2, CHMP1A, CLP1, COASY, EXOSC3, EXOSC8, EXOSC9, PCLO, RARS2, SEPSECS, TBC1D23, TOE1, TSEN2, TSEN15, TSEN34*, TSEN54*, VPS51, VPS53, VRK1*

Cardiovascular Diseases

☐ **Arrhythmogenic right ventricular dysplasia (ARVD, ARVC)**
Panel-ID: ID010.00 (small)
9 genes (38295 kb): DES, DSC2*,**, DSG2*,**, DSP*,**, JUP*,**, PKP2*,**, RYR2*, TGFB3*,**, TMEM43*

☐ **Atrial fibrillation (ATFB)**
Panel-ID: ID016.01 (small)
14 genes (24231 kb): ABCC9, GJA5, KCNA5, KCNE1,**, KCNE2*,**, KCNH2*,**, KCNJ2*,**, KCNQ1*,**, NPPA, SCN1B*, SCN2B, SCN3B*, SCN4B, SCN5A*,***

☐ **Atrial septal defect and Ventricular septal defect (ASD, VSD)**
Panel-ID: ID143.00 (small)
12 genes (21066 kb): ACTC1, CITED2, CRELD1, GATA4*, GATA5*, GATA6, GJA1*, MYH6*, NKX2-5*, NR2F2*, TBX20, TLL1*

☐ **Brugada syndrome (BRGDA)**
Panel-ID: ID014.01 (small)
15 genes (34553 kb): CACNA1C, CACNA2D1, CACNB2*, GPD1L*, HCN4*, KCND3, KCNE3*, KCNJ8*, PKP2*,**, RANGRF, SCN1B*, SCN2B, SCN3B*, SCN5A*,**, TRPM4*

☐ **CADASIL and CARASIL**
Panel-ID: ID167.00 (small)
2 genes (8403 kb): HTRA1, NOTCH3*,***

☐ **Cardiac arrhythmia, comprehensive diagnostics**
Panel-ID: ID026.01 (large)
49 genes (132609 kb): ABCC9, AKAP9, ANK2, CACNA1C, CACNA2D1, CACNB2*, CALM1, CASQ2*, CAV3, DES*, DSC2*,**, DSG2*,**, DSP*,**, EMD*, GJA5, GPD1L*, HCN4*, JPH2, JUP*,**, KCNA5, KCND3, KCNE1*,**, KCNE2*,**, KCNE3*, KCNH2*,**, KCNJ2*,**, KCNJ5, KCNJ8*, KCNQ1*,**, LMNA*,**, MYH6*, NKX2-5*, NPPA, PKP2*,**, PRKAG2*, RANGRF, RYR2*, SCN1B*, SCN2B*,**, SCN3B*, SCN4B, SCN5A*,**, SNTA1, TBX5, TGFB3*,**, TMEM43*, TNNI3*, TRDN, TRPM4*

☐ **Cardiomyopathy, comprehensive diagnostics**
Panel-ID: ID027.00 (large)
96 genes (316812 kb): ABCC9, ACTA1, ACTC1*, ACTN2*, ALMS1*, ANKRD1, BAG3, BRAF*, CALR3, CASQ2*, CAV3, CBL, COX15, CRYAB, CSRP3, DES, DMD*,**, DNAJC19, DOLK, DSC2*,**, DSG2*,**, DSP*,**, DTNA, EMD*, EYA4, FHL1*, FHL2*, FKRP, FKTN, FXN, GAA*,**, GATAD1, GLA*,**, HADHA, HFE*,**, HRAS*, ILK, JPH2*, JUP*,**, KLF10, KRAS*, LAMA2, LAMA4, LAMP2*, LDB3, LMNA*,**, MAP2K1*, MAP2K2*, MIB1, MURC, MYBPC3*,**, MYH6*, MYH7*,**, MYL2*, MYL3*, MYLK2, MYO6, MYOZ2, MYPN*, NEXN, NPPA, NRAS*, PDLIM3, PKP2*, PLN, PRDM16, PRKAG2*, PTPN11*, RAF1*, RBM20*, RYR2*, SCN2B, SCN5A*,**, SCO2, SDHA*, SGCB, SGCD, SGCG, SHOC2*, SLC25A4, SOS1*, TAZ*, TBX20*, TCAP, TGFB3*,**, TMEM43*, TMPO, TNNC1*, TNNI3*, TNNT2*,**, TPM1*, TRIM63, TRPM4, TTN, TXNRD2, VCL*

☐ **Catecholaminergic polymorphic ventricular tachycardia (CPVT)**
Panel-ID: ID012.01 (small)
6 genes (31884 kb): ANK2, CALM1, CASQ2, KCNJ2*,**, RYR2*, TRDN*

☐ **Congenital heart defects, nonsyndromic**
Panel-ID: ID017.02 (small)
34 genes (80270 kb): ACTC1, ACVR2B, CFAP53, CFC1, CITED2, CRELD1, ELN, GATA4*, GATA5*, GATA6, GDF1*, GJA1*, JAG1*,**, MMP21, MED13L, MYH6*, ISL1, NKX2-5*, NKX2-6, NR2F2*, NODAL, NOTCH1*, PKD1L1, PLD1, PRDM6, ROBO4, SMAD6*, TAB2, TBX1*,**, TBX20*, TFAP2B, TLL1, ZFPM2*, ZIC3*

Basis-Panel I (Atrial/Ventrikular septal defect):
12 genes (21060 bp): ACTC1, CITED2, CRELD1, GATA4*, GATA5*, GATA6, GJA1*, MYH6*, NR2F2*, NKX2-5*, TBX20, TLL1*

Basis-Panel II (Tetralogy of Fallot):
11 genes (19351 bp): GATA4,GATA5*, GATA6, GDF1*, ISL1, JAG1*,**, NKX2-5*, NR2F2*, TAB2, TBX1*,**, ZFPM2**

Basis-Panel III (Heterotaxy):
9 genes (18819 bp): ACVR2B, CFAP53, CFC1, CRELD1, GDF1, MMP21, NODAL, PKD1L1, ZIC3*

Basis-Panel IV (Conotruncal heart malformations):
10 genes (19509 bp): CFC1, GATA5, GATA6, GDF1*, MED13L, NKX2-5*, NKX2-6, TBX1*, ZFPM2*, ZIC3*

Basis-Panel V (Aortic valve disease):
7 genes (15999 bp): ELN, GATA5, NOTCH1*, NR2F2*, ROBO4, SMAD6*, TAB2*

Basis-Panel VI (Hypoplastic left heart syndrome):
4 genes (5939 bp): GDF1, GJA1*, NKX2-5*, NR2F2**

Basis-Panel VII (Multiple congenital heart defects):
6 genes (11717 bp): GATA5, GDF1*, NR2F2*, PLD1, TAB2, ZIC3*

☐ **Congenital heart defects, syndromic**
Panel-ID: ID252.00 (large)
47 genes (210192 kb): ADAMTS10, ADAMTS17, AFF4, ARHGAP31, CDK13, CHD4, CHD7,**, CREBBP*,**, DLL4, DOCK6*, DTNA, EHMT1, EOGT, EP300*,**, EVC, EVC2, FBN1*,**, FBN2*, FLNA*, FOXC1*,**, GPC3*,**, JAG1*,**, KDM6A*,**, KMT2D*,**, LTBP2, MED12*, MGP, MYH11*, NIPBL*,**, NOTCH1*, NOTCH2*, NSD1*,**, PITX2*,**, PRKAR1A, PRKD1, RBM10, RBPJ, SALL1, SALL4, SEMA3E, TBX3, TBX5*, TGFBR1*,**, TGFBR2*,**, TMEM260, WDPCP*, ZEB2*,***

Basis-Panel I (CHARGE syndrome):
2 genes (11316 bp): CHD7,**, SEMA3E*

Basis-Panel II (Weill-Marchesani syndrome):
4 genes (20670 bp): ADAMTS10, ADAMTS17, FBN1,**, LTBP2*

Basis-Panel III (Adams-Oliver syndrome):
6 genes (23232 bp): ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1*, RBPJ*

Basis-Panel IV (Kabuki syndrome):
2 genes (20814 bp): KDM6A,**, KMT2D*,***

Basis-Panel V (Alagille syndrome):
2 genes (11067 bp): JAG1,**, NOTCH2**

Basis-Panel VI (Marfan syndrome):
3 genes (according to tariff EBM11444/11445): FBN1,**, TGFBR1*,**, TGFBR2*,***

Basis-Panel VII (Rubinstein-Taybi syndrome):
2 genes (14568 bp): CREBBP,**, EP300*,***

☐ **Conotruncal heart malformations (CTHM)**
Panel-ID: ID160.00 (small)
10 genes (19617 kb): CFC1, GATA5, GATA6, GDF1*, MED13L, NKX2-5*, NKX2-6, TBX1*, ZFPM2*, ZIC3*

☐ **Dilated cardiomyopathy (CMD, DCM)**
Panel-ID: ID008.01 (large)
39 genes (202365 kb): ABCC9, ACTC1, ACTN2*, BAG3, CRYAB, CSRP3, DES*, DMD*,**, DSG2*,**, DSP*,**, EYA4, FKTN*, GATAD1, JUP*,**, LAMA4, LAMP2*, LDB3, LMNA*,**, MYBPC3*,**, MYH6*, MYH7*,**, MYPN, NEXN*, PLN, PRDM16*, RAF1*, RBM20*, SCN5A*,**, SDHA*, SGCD, TAZ*, TMPO, TNNC1*, TNNI3*, TNNT2*,**, TPM1*, TTN*, TTR, VCL**

☐ **Hypertrophic cardiomyopathy (CMH, HCM)**
Panel-ID: ID007.01 (large)
31 genes (151684 kb): ACTC1, ACTN2*, CALR3, CAV3, COX15, CSRP3*, GAA*,**, GLA*,**, JPH2*, LAMP2*, LDB3, MYBPC3*,**, MYH6*, MYH7*,**, MYL2*, MYL3*, MYLK2, MYOZ2, MYPN*, NEXN*, PLN, PRKAG2*, SCO2, TCAP, TNNC1*, TNNI3*, TNNT2*,**, TPM1*, TTN*, TTR, VCL**

☐ **Left ventricular noncompaction (LVNC, NCCM)**
Panel-ID: ID011.00 (small)
11 genes (25956 kb): ACTC1, ACTN2, DTNA, LDB3, MIB1, MYBPC3*,**, MYH7*,**, PRDM16, TAZ*, TNNT2*,**, TPM1**

☐ **Long QT syndrome (LQT)**
Panel-ID: ID013.00 (small)
14 genes (47952 kb): AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1*,**, KCNE2*,**, KCNH2*,**, KCNJ2*,**, KCNJ5, KCNQ1*,**, SCN4B, SCN5A*,**, SNTA1*

☐ **Neuromuscular diseases with cardiac involvement**
Panel-ID: ID123.01 (large)
14 genes (132213 kb): CAV3, DES, DMD*,**, EMD*, FHL1*,*

*FKRP**, FKTN*, LAMA2, LMNA*,**, SGCB*,**, SGCD**, SGCG*,**, TCAP, TTN**

☐ **Restrictive cardiomyopathy (RCM)**
Panel-ID: ID105.00 (small)
10 genes (17460 kb): ACTC1, BAG3, DES*, MYH7*,**, MYL2*, MYL3*, MYPN, TNNI3*, TNNT2*,**, TPM1**

☐ **Short QT syndrome (SQT)**
Panel-ID: ID233.00 (small)
6 genes (18306 kb): CACNA1C, CACNA2D1, CACNB2*, KCNH2*,**, KCNJ2*,**. KCNQ1*,***

☐ **Sick sinus syndrome (SSS)**
Panel-ID: ID107.00 (small)
3 genes (15474 kb): HCN4, MYH6*, SCN5A*,***

☐ **Susceptibility to hemorrhagic or ischemic stroke**
Panel-ID: ID234.01 (large)
39 genes (120948 kb): ACE, ACTA2, ADA2, ALOX5AP, APOE*, APP, CBS*, CCM2*,**, COL3A1*,**, COL4A1, COL4A2, CST3, F2, F5, FBN1*,**, FLNA*,**, GLA*,**, GUCY1A1, HTRA1*, ITM2B, JAG1*,**, KRIT1*,**, MTHFR, MYH11*, MYLK*, NOS3, NOTCH3*,**, OTC, PDCD10*,**, POLG*,**, PRKCH, RNF213, SLC2A10*, SMAD3*, TGFB2*, TGFBR1*,**, TGFBR2*,**, TREX1*,**, TTR*

Basis-Panel I:
17 genes (25080 bp + according tariff EBM 11448: 31527 bp): ACTA2, APP, CBS*, COL3A1*,**, COL4A1, COL4A2, FBN1*,**, GLA*,**, HTRA1*, MYH11*, MYLK*, NOTCH3*,**, SMAD3*, TGFB2*, TGFBR1*,**, TGFBR2*,**, TREX1*,***

Basis-Panel II (Intracerebral hemorrhage):
5 genes (16815 bp): ACE, APP, COL4A1, COL4A2, CST3

Basis-Panel III (Moyamoya disease):
4 genes (22179 bp): ACTA2, GUCY1A1, JAG1, RNF213*

Basis-Panel IV (Aneurysm):
9 genes (according tariff EBM 11448: 31527 bp): ACTA2, COL3A1*,**, FBN1*,**, MYH11*, MYLK*, SMAD3*, TGFB2*, TGFBR1*,**, TGFBR2*,***

Basis-Panel V (CADASIL, CARASIL):
2 genes (8403 bp): HTRA1, NOTCH3*,***

Basis-Panel VI (Cerebral cavernous malformations):
3 genes (4176 bp): CCM2,**, KRIT1*,**, PDCD10*,***

☐ **Tetralogy of Fallot (TOF)**
Panel-ID: ID144.00 (small)
11 genes (19365 kb): GATA4, GATA5*, GATA6, GDF1*, ISL1, JAG1*,**, NKX2-5*, NR2F2*, TAB2, TBX1*,**, ZFPM2**

☐ **Visceral heterotaxy (HTX)**
Panel-ID: ID145.00 (small)
9 genes (18826 kb): ACVR2B, CFAP53, CFC1, CRELD1, GDF1, MMP21, NODAL, PKD1L1, ZIC3*

Developmental and Growth Disorders

☐ **RAS-related disorders**
Panel-ID: ID015.03 (small)
21 genes (44709 kb): A2ML1, BRAF,**, CBL, HRAS*,**, KRAS*,**, LZTR1*,**, MAP2K1*, MAP2K2*, MRAS*, NF1*,**, NRAS*,**, PTPN11*, RAF1*, RASA2, RRAS2, PPP1CB, RIT1*, SHOC2*, SOS1*, SOS2*, SPRED1**

Kidney Diseases, Endocrine Disorders

☐ **Endocrine arterial hypertension**
Panel-ID: ID270.00 (small)
24 genes (66759 kb): CACNA1H, CACNA1D, CLCN2, CUL3, CYP11B1*, CYP11B2, CYP17A1*, HSD11B2, KCNJ5, KLHL3, NR3C2, PDE3A, NF1*,**, SCNN1A, SCNN1B, SCNN1G, SDHA*,** SDHAF2**, SDHB*,**, SDHC**, SDHD*,**, WNK1, WNK4, YY1AP1*

Hyperaldosteronism (HALD):
7 genes (20589 bp, 7 genes: CACNA1H, CACNA1D, CLCN2, CYP11B1*, CYP11B2, KCNJ5*

Pseudohypoaldosteronism (PHA2):
4 genes (15708 bp): CUL3, KLHL3, WNK1, WNK4

Liddle syndrome (LIDLS):
3 genes (5883 bp): SCNN1A, SCNN1B, SCNN1G

Pheochromocytoma and Paragangliomas:
5 genes (4329 bp): SDHA,**, SDHAF2**, SDHB*,**, SDHC**, SDHD*,***

Metabolic Diseases

☐ **Storage diseases with cardiac involvement**
Panel-ID: ID149.00 (small)
12 genes (19158 kb): ATP7B,**, FTH1*, GAA*,**, GLA*,**, HAMP*,**, HFE*,**, HJV*,**, LAMP2*, PRKAG2*, SLC40A1*,**, TFR2*,**, TTR**

Pulmonary and Respiratory Diseases

☐ **Pulmonary arterial hypertension (PAH)**
Panel-ID: ID281.00 (small)
22 genes (45870 kb): ABCC8, ACVRL1*,**, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG*,**, FOXF1*,**, G6PC3, GDF2*, KCNA5*, KCNK3, NFU1, NOTCH3*,**, SARS2, SMAD1, SMAD4*,**, SMAD9, SOX17, TBX4*

Skeletal and Bone Diseases

☐ **Heart-hand syndrome**
Panel-ID: ID165.01 (small)
9 genes (23871 kb): DACT1, GATA6, LMNA*,**, RBM8A, RECQL4, SALL1**, SALL4**, TBX3, TBX5*,***

Ciliopathies

Kidney Diseases, Brain Malformations

☐ **Meckel syndrome (MKS)**
Panel-ID: ID032.02 (small)
13 genes (35055 kb): B9D1, B9D2, CC2D2A*, CEP290*, KIF14, MKS1, NPHP3, RPGRIP1L*, TCTN2, TMEM107, TMEM216*, TMEM231, TMEM67**

☐ **Joubert syndrome (JBTS)**
Panel-ID: ID028.02 (small)
35 genes (91859 kb): AHI1, ARL13B, ARL3, ARMC9, B9D1, B9D2*, CC2D2A*, CEP104, CEP120, CEP290*, CEP41, CPLANE1, CSPP1, INPP5E*, KIAA0556, KIAA0586, KIF7, NPHP1*,**, MKS1*, OFD1*, PDE6D, PIBF1, RPGRIP1L*, SUFU*, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216*, TMEM231, TMEM237, TMEM67*, TTC21B, ZNF423*

Kidney Disease, Eye Diseases

☐ **Senior-Loken syndrome (SLSN)**
Panel-ID: ID029.01 (small)
8 genes (27936 kb): CEP290, IQCB1, NPHP1*,**, NPHP3, NPHP4, SDCCAG8, TRAF3IP1, WDR19*

Kidney Diseases, Multisystemic Malformation Syndromes

☐ **Bardet-Biedl syndrome (BBS)**
Panel-ID: ID093.02 (small)
21 genes (39018 kb): ARL6, BBIP1, BBS1, BBS2*, BBS4, BBS5, BBS7, BBS9*, BBS10*, BBS12*, C8ORF37, CEP290*, IFT27, IFT74, LZTFL1, MKKS*, MKS1*, SDCCAG8, TRIM32, TTC8*, WDPCP*

Pulmonary and Respiratory Diseases

☐ **Primary ciliary dyskinesia with or without situs inversus (PCD, CILD)**
Panel-ID: ID085.02 (large)
42 genes (132917 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*

Connective Tissue Diseases

☐ **Cutis laxa (ARCL, ADCL)**
Panel-ID: ID109.01 (small)
10 genes (22728 kb): ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A,**, EFEMP2, ELN, FBLN5, LTBP4*, PYCR1*

☐ **Ehlers-Danlos syndrome (EDS)**
Panel-ID: ID039.05 (small)
20 genes (79572 kb): ADAMTS2, AEBP1*, B3GALT6*, B4GALT7, C1R, C1S*, CHST14, COL1A1*,**, COL1A2*,**, COL3A1*,**, COL5A1*,**, COL5A2*, COL12A1, DSE, FKBP14, PLOD1*,**, PRDM5, SLC39A13*, TNXB*,**, ZNF469*

Basis-Panel I (EDS, dominant):
7 genes (22680 bp + according to tariff EBM 11446/11447 4398 bp): C1R, C1S, COL1A1*,**, COL1A2*,**, COL3A1*,**, COL5A1*,**, COL5A2**

Basis-Panel II (EDS, rezessive):
11 genes (23496 bp): ADAMTS2, AEBP1*, B3GALT6*, B4GALT7, CHST14, COL1A2*,**, DSE, FKBP14, PLOD1*,**, PRDM5, SLC39A13**

Cardiovascular Diseases

☐ **Connective tissue diseases with aortic involvement**
Panel-ID: ID137.02 (large)
60 genes (192024 kb): ABCC6,**, ACTA2*, ADAMTS2*, AEBP1*, ALDH18A1, ATP6V0A2, ATP6V1, ATP6V1E1, ATP7A*,**, B3GALT6*, B4GALT7, BGN, C1R, C1S*, CBS*, CHST14, COL1A1*,**, COL1A2*,**, COL3A1*,**, COL4A1, COL5A1*,**, COL5A2*, COL12A1, DSE, EFEMP2, ELN, FBLN5, FBN1*,**, FBN2*, FKBP14, FLNA*,**, FOXE3*, ITPKC, LOX, LTBP4*, MAT2A, MFAP5, MYH11*, MYLK*, NOTCH1*, PLOD1*,**, PRDM5, PRKG1, PLOD3, PYCR1, RIN2, ROBO4, SKI*, SLC2A10*, SLC39A13*, SMAD2, SMAD3*, SMAD4*,**, SMAD6*, TGFB2*, TGFB3*,**, TGFBR1*,**, TGFBR2*,**, TNXB*,**, ZNF469*

Basis-Panel I (Thoracic aortic aneurysm):
9 genes (according to tariff EBM 11448): ACTA2, COL3A1*,**, FBN1*,**, MYH11*, MYLK*, SMAD3*, TGFB2*, TGFBR1*,**, TGFBR2*,***

Basis-Panel II (Marfan syndrome):
3 genes (according to tariff EBM 11444/11445): FBN1,**, TGFBR2*,**, TGFBR1*,***
Basis-Panel III (Ehlers-Danlos syndrome, dominant):
7 genes (22680 bp + according to tariff EBM 11446/11447): C1R, C1S, COL1A1*,**, COL1A2*,**, COL3A1*,**, COL5A1*,**, COL5A2**

Basis-Panel IV (Ehlers-Danlos syndrome, rezessive):
11 genes (23496 bp): ADAMTS2, AEBP1*, B3GALT6*, B4GALT7, CHST14, COL1A2*,**, DSE, FKBP14, PLOD1*,**, PRDM5, SLC39A13**

Basis-Panel V (Cutis laxa):
10 genes (22722 bp): ALDH18A1, ATP6V0A2, ATP6V1E1, ATP6V1A, ATP7A,**, EFEMP2, ELN, FBLN5, LTBP4*, PYCR1*

Basis-Panel VI (Loeys-Dietz syndrome):
8 genes (11178 bp): BGN, SLC2A10, SMAD2, SMAD3*, TGFB2*, TGFB3*, TGFBR1*,**, TGFBR2*,***

Basis-Panel VII (Aortic aneurysm, nonsyndromic):
10 genes (22179 bp): ACTA2, FOXE3*, LOX, MAT2A, MFAP5, MYH11*, MYLK*, PRKG1, TGFBR1*,**, TGFBR2*,***

Basis-Panel VIII (Aortic valve disease):
3 genes (12177 bp): NOTCH1, ROBO4, SMAD6**

Cardiovascular Diseases

☐ **Hereditary hemorrhagic telangiectasia (HHT)**
Panel-ID: ID155.00 (small)
5 genes (9540 kb): ACVRL1,**, BMPR2, ENG*,**, GDF2*, SMAD4*,***

☐ **Loeys-Dietz syndrome (LDS) and similar aortic diseases**
Panel-ID: ID009.05 (small)
31 genes (95208 kb): ACTA2, BGN, COL1A2*,**, COL3A1*,**, COL5A1*,**, COL5A2*, EFEMP2, ELN, FBLN5, FBN1*,**, FBN2*, FLNA*,**, FOXE3*, LOX, MAT2A, MFAP5, MYH11*, MYLK*, NOTCH1, PLOD1*,**, PRKG1, ROBO4, SKI*, SLC2A10*, SMAD2, SMAD3*, SMAD6*, TGFB2*, TGFB3*,**, TGFBR1*,**, TGFBR2*,***

Basis-Panel I:
21 genes (25089 bp + according to tariff EBM 11448): ACTA2, BGN, COL3A1*,**, FBN1*,**, FOXE3*, LOX, MAT2A, MFAP5, MYH11*, MYLK*, NOTCH1, PRKG1, ROBO4, SLC2A10*, SMAD2, SMAD3*, SMAD6, TGFB2*, TGFB3*,**, TGFBR1*,**, TGFBR2*,***

Basis-Panel II (Thoracic aortic aneurysm):
9 genes (according to tariff EBM 11448): ACTA2, COL3A1*,**, FBN1*,**, MYH11*, MYLK*, SMAD3*, TGFB2*, TGFBR1*,**, TGFBR2*,***

Basis-Panel III (Aortic aneurysm, nonsyndromic):
10 genes (22179 bp): ACTA2, FOXE3*, LOX, MAT2A, MFAP5, MYH11*, MYLK*, PRKG1, TGFBR1*,**, TGFBR2*,***

Basis-Panel IV (Loeys-Dietz syndrome):
8 genes (11178 bp): BGN, SLC2A10, SMAD2, SMAD3*, TGFB2*, TGFB3*, TGFBR1*,**, TGFBR2*,***

Basis-Panel V (Aortic valve disease):
3 genes (12177 bp): NOTCH1, ROBO4, SMAD6**

☐ Marfan syndrome and similar syndromes
Panel-ID: ID194.01 (large)
42 genes (141546 kb): ABCD4, ACTA2, ADAMTS10, ADAMTS17, ADAMTSL4, BGN, CBS*, COL1A2, COL2A1*,**, COL3A1*,**, COL5A1**,*, COL5A2*, COL9A1, COL9A2, COL11A1*,**, FBN1*,**, FBN2*, FKBP14, FLNA*, LIG4, LMBRD1, LOX, LTBP2, PLOD1*,**, MED12*, MMACHC, MMADHC, MTHFR, MTR, MTRR, MYH11*, MYLK*, PRDM5, SKI*, SLC2A10*, SMAD2, SMAD3*, TGFB2*, TGFB3*,**, TGFBR1*,**, TGFBR2*,**, ZNF469*

Basis-Panel I:
18 genes (24651 bp + according to tariff EBM 11448): ACTA2, BGN, CBS, COL3A1*,**, FBN1*,**, FBN2*, LOX, LTBP2, MYLK, MYH11, SKI*, SLC2A10*, SMAD2, SMAD3*, TGFB2*, TGFB3*,**, TGFBR1*,**, TGFBR2*,***

Basis-Panel II (Marfan syndrome):
3 genes (according to tariff EBM 11444/11445): FBN1,**, TGFBR1*,**, TGFBR2*,***

Basis-Panel III (Aortopathy):
9 genes (according to tariff EBM 11448): ACTA2, COL3A1*,**, FBN1*,**, MYH11*, MYLK*, SMAD3*, TGFB2*, TGFBR1*,**, TGFBR2*,***

Basis-Panel IV (Homocystinuria):
8 genes (14763 bp): ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR*

Basis-Panel V (Weill-Marchesani syndrome):
4 genes (20670 bp): ADAMTS10, ADAMTS17, FBN1,**, LTBP2*

Basis-Panel VI (Stickler syndrome):
4 genes (14709 bp): COL2A1,**, COL9A1, COL9A2, COL11A1*,***

Basis-Panel VII (Ehlers-Danlos syndrome):
6 genes (18813 bp + according to tariff EBM 11446/11447): COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, PRDM5

Basis-Panel VIII (Brittle-Cornea syndrome):
2 genes (13665 bp): PRDM5, ZNF469

☐ Thoracic aortic aneurysm with or without aortic dissection (TAA/D) - EBM 11448
Panel-ID: ID020.00 (small)
9 genes (31527 kb): ACTA2, COL3A1*,**, FBN1*,**, MYH11*, MYLK*, SMAD3*, TGFB2*, TGFBR1*,**, TGFBR2*,***

Cardiovascular Diseases, Multisystemic Malformation Syndromes

☐ Marfan syndrome - EBM 11444/11445
Panel-ID: ID022.00 (small)
3 genes (11823 kb): FBN1,**, TGFBR2*,**, TGFBR1*,***

Multisystemic Malformation Syndromes, Eye Diseases

☐ Stickler syndrome (STL)
Panel-ID: ID062.00 (small)
6 genes (21048 kb): COL2A1,**, COL9A1, COL9A2, COL9A3, COL11A1*,**, COL11A2**

Developmental and Growth Disorders

☐ Autism
Panel-ID: ID076.02 (large)
95 genes (327561 kb): ADNP, ADSL, AFF2, ALDH5A1, ARX,**, ASH1L, ASTN2, AUTS2, BCL11A, CACNA1C*, CASK*,**, CC2D1A, CDKL5*,**, CHD2, CHD7*,**, CHD8, CNOT3, CNTN4, CNTNAP2*, CTNNB1, DHCR7*,**, DLGAP2, DPP6, DYRK1A, EHMT1, EIF4E, FMR1*, FOXG1*,**, FOXP1*,**, FOXP2*,**, FRMPD4, GRIA3, GRIN2A, GRIN2B, HERC2, IL1RAPL1, IQSEC2, KDM5C, KMT2C, MAOA, MBD5, MECP2*,**, MED12*, MEF2C*,**, MYT1L, NAA15, NAGLU, NEXMIF, NHS, NLGN1, NLGN3, NLGN4X, NR1I3, NRXN1*,**, NRXN2, NRXN3, OPHN1, PAH, PCDH19*,**, PHF6*, PHF8, POGZ, PQBP1, PTCHD1, PTEN*,**, RAB39B, RAI1, RPL10, SCN1A*,**, SCN2A, SEMA5A, SETD2, SGSH, SHANK2, SHANK3, SLC6A8, SLC9A6, SLC9A9, SLC35A3, SMARCB1*,**, SYN1, SYNGAP1*, TBL1XR1, TBR1, TCF4, TMLHE, TSC1*,**, TSC2*,**, UBE2A, UBE3A*,**, UPF3B, VPS13B*,**, ZEB2*,**, ZIC1, ZMYND11*

Basis-Panel I (autosomal dominant):
6 genes (25233 bp): CHD8, CNTNAP2, NLGN1, NRXN1, PTEN*,**, SHANK3*

Basis-Panel II (X-linked):
13 genes (24177 bp): CASK, CDKL5,**, IL1RAPL1, MAOA, MECP2*,**, NLGN3, NLGN4X, PTCHD1, RAB39B, RPL10, SYN1, TMLHE, UPF3B*

☐ FG syndrome (FGS)
Panel-ID: ID215.00 (small)
3 genes (17211 kb): CASK,**, FLNA*, MED12**

☐ Mental retardation, aurosomal dominant (MRD)
Panel-ID: ID036.02 (large)
71 genes (247922 kb): ADNP, AHDC1, AP2M1, ARID1A, ARID1B*, ARID2, ASH1L, AUTS2, CACNG2, CAMK2A, CAMK2B, #CAMK2G, CDH15, CHAMP1, CERT1, CIC, CLTC, CTCF, CTNNB1*, DEAF1, DLG4, DPF2, DPP6, DYNC1H1, DYRK1A*, EEF1A2, EHMT1, EPB41L1, GATAD2B, GNB1, GRIN1, GRIN2B*,**, HIVEP2, KANSL1, KAT6A, KCNQ5, KIF1A, KMT2C*, KMT5B, MBD5*, MED13, MEF2C*,**, MYT1L, NAA15, NR1I3, NUS1, PACS1, POGZ, PPP2R1A, PPP2R5D, PURA*, RAC1, SET, SETBP1*, SETD5, SMARCA2*, SMARCA4*, SMARCB1*,**, SMARCC2, SMARCE1, SOX4, SOX11, STAG1, SYNGAP1*, TBL1XR1, TLK2, TRIO, TRIP12, ZBTB18*, ZEB2*,**, ZMYND11*

Basis-Panel I:
10 genes (24803 bp): CTNNB1, DYRK1A*, GNB1, GATAD2B, GRIN2B*,**, MBD5*, MEF2C*,**, SET, SYNGAP1*, ZBTB18**

Basis-Panel II (Coffin-Siris syndrome):
8 genes (24945 bp): ARID1A, ARID1B*, DPF2, SMARCA4*, SMARCB1*,**, SMARCE1, SOX4, SOX11*

Basis-Panel III (Kleefstra syndrome):
5 genes (25355 bp): EHMT1, KMT2C, NR1I3, MBD5*, SMARCB1*,***

☐ **Mental retardation, aurosomal recessive (MRT)**
Panel-ID: ID037.01 (large)
52 genes (129152 kb): ADAT3, ADK, ALKBH8, ANK3, C12ORF4, CAMK2A, CC2D1A, CRADD, CRBN, EDC3, EIF3F, ELP2, FBXO31,FMN2, GPT2, GRIK2, HERC2, HNMT, IMPA1, KDM5B, KPTN, LINGO1, LINS1, LMAN2L, MAN1B1, MBOAT7, MED23, METTL23, METTL5, NDST1, NSUN2*, PGAP1, PGAP2, PIGC, PIGG, PRSS12, PUS3, RSRC1, RUSC2, SLC6A17, ST3GAL3, TAF2, TAF13, TECR, TNIK, TRAPPC9, TRMT1, TTI2, TUSC3, WASHC4, ZBTB11, ZC3H14*

☐ **Mental retardation, X-linked (MRX, MRXS)**
Panel-ID: ID038.03 (large)
71 genes (182931 kb): ACSL4, AFF2*, AP1S2, ARX*,**, ATP6AP2, ATRX*,**, BRWD3, CASK*,**, CLCN4, CLIC2, CNKSR2, CUL4B*, CXORF56, DDX3X, DLG3, FGD1*,**, EIF2S3, FMR1*, FRMPD4, FTSJ1, GDI1, GRIA3*, HCFC1, HNRNPH2, HSD17B10, HS6ST2, HUWE1*, IGBP1, IL1RAPL1*, IQSEC2*, KDM5C, KIF4A, KLHL15, L1CAM*,**, LAS1L, MECP2*,**, MED12*, MID2, MSL3, NEXMIF, NKAP, NONO, OGT, OPHN1, PAK3, PHF6*, PHF8, POLA1, PQBP1, PRPS1, RAB39B, RLIM, RPL10, RPS6KA3*,**, SHROOM4, SLC16A2, SLC6A8*,**, SLC9A6, SLC9A7, SMS, SYP, TAF1, THOC2, TSPAN7, UBE2A*, UPF3B*, USP9X, USP27X, ZC4H2*, ZDHHC9, ZNF711*

Basis-Panel I (MRX, nonsyndromic):
12 genes (24849 bp): ACSL4, ARX*,**, DLG3, FTSJ1, GDI1, IL1RAPL1*,**, IQSEC2*, NEXMIF, PAK3, RAB39B, RPS6KA3*,**, TSPAN7*

Basis-Panel II (MRX, syndromic, rezessive):
15 genes (24759 bp): AP1S2, ARX,**, CUL4B*, FGD1*,**, KDM5C, MECP2*,**, OPHN1, PHF6*, PQBP1, PRPS1, RAB39B, SLC6A8*,**, SMS, UBE2A*, UPF3B**

Basis-Panel III (MRX, syndromic, dominant):
7 genes (24909 bp): CASK,**, CLCN4, DDX3X, HUWE1*, MSL3, SLC16A2, SLC9A6*

☐ **Overgrowth syndromes**
Panel-ID: ID073.03 (small)
17 genes (43887 kb): APC2, CDKN1C*,**, DIS3L2, DNMT3A*, EED, EZH2*, FGFR3*,**, FIBP, GPC3*,**, GPC4*,**, NFIX*,**, NSD1*,**, OFD1*, PDGFRB*, PTEN*,**, RNF125, SUZ12*

Sotos syndrome (SOTOS):
4 genes (18796 bp): APC2, EZH2*, NFIX*,**, NSD1*,**

Simpson-Golabi-Behmel syndrome (SGBS):
3 genes (6440 bp): GPC3,**, GPC4*,**, OFD1**

☐ **Pitt-Hopkins syndrome (PTHS)**
Panel-ID: ID106.00 (small)
3 genes (10635 kb): CNTNAP2, NRXN1*,**, TCF4*,**

☐ **Rett syndrome (RTT) and similar syndromes**
Panel-ID: ID125.01 (small)
16 genes (45345 kb): CDKL5,**, FOXG1*,**, GABBR2, GABRA2, GABRB2, GRIN2B*, HTT, IQSEC2*, MECP2*,**, MEF2C*,**, NTNG2, SHANK3, STXBP1*,**, TCF4*,**, UBE3A*,**, WDR45*

Microcephaly and Macrocephaly

☐ **Mental retardation with macrocephaly**
Panel-ID: ID131.01 (small)
32 genes (91657 kb): ADK, ALKBH8, BRWD3, CAMK2G, CRADD, CUL4B, DDX3X, DEAF1, FMR1*, GATAD2B, GRIA3*, HEPACAM, HUWE1*, IGBP1, KDM5C, KIF7, KPTN, L1CAM*,**, MECP2*,**, MED12*, MSL3, NONO, OPHN1, PPP2R5D, PTEN, RAB39B, RAC1, SHANK3, SHROOM4, TRIO, UPF3B*, ZDHHC9*

Mental retardation (autosomal dominant) and macrocephaly:
9 genes (24781 bp): CAMK2G, DEAF1, GATAD2B, HEPACAM, PPP2R5D, PTEN, RAC1, SHANK3, TRIO

Mental retardation (autosomal rezessive) and macrocephaly:
5 genes (8985 bp): ADK, ALKBH8, CRADD, KIF7, KPTN

Mental retardation (X-linked) and macrocephaly:
18 genes (57933 bp): BRWD3, CUL4B, DDX3X, FMR1*, GRIA3*, HUWE1*, IGBP1, KDM5C, L1CAM*,**, MECP2*,**, MED12*, MSL3, NONO, OPHN1, RAB39B, SHROOM4, UPF3B*, ZDHHC9*

☐ **Mental retardation with microcephaly**
Panel-ID: ID129.01 (large)
70 genes (202848 kb): ACSL4, ADAT3, ATRX*,**, AUTS2, CAMK2B, CAMK2G, CASK*,**, CERT1, CHAMP1, CTCF, CTNNB1*, DDX3X, DPP6, DYRK1A*, EDC3, EHMT1, EIF2S3, GRIN2B, HCFC1, HIVEP2, HNMT, HNRNPH2, HUWE1*, IQSEC1*, KDM5C, KIF11, KIF1A, L1CAM*,**, LINGO1, LINS1*, MBD5, MBOAT7, MECP2*,**, METTL5, NEXMIF, NSUN2, OGT, PAK3, PGAP1, PHF6*, POGZ, POLA1, PPP2R1A, PQBP1, RAC1, RPL10, RLIM, SET, SHROOM4, SLC16A2, SLC6A8*,**, SLC9A6, SMARCA4*, SMARCB1*, SMARCE1, SOX11, SOX4, SYNGAP1*, TAF1, TAF13, TAF2, THOC2, TLK2, TRAPPC9, TRIO, TRMT1, TTI2, ZBTB18*, ZC4H2*, ZEB2*,**

Mental retardation (autosomal dominant) and microcephaly:
29 genes (88509 bp): AUTS2, CAMK2B, CAMK2G, CERT1, CHAMP1, CTCF, CTNNB1, DPP6, DYRK1A*, EHMT1, GRIN2B, HIVEP2, KIF11, KIF1A, MBD5*, POGZ, PPP2R1A, RAC1, SET, SMARCA4*, SMARCB1*, SMARCE1, SOX11, SOX4, SYNGAP1*, TLK2, TRIO, ZBTB18*, ZEB2*,**

Mental retardation (autosomal rezessiv) and microcephaly:
14 genes (25992 bp): ADAT3, EDC3, HNMT, LINGO1, LINS1, MBOAT7, METTL5, NSUN2*, PGAP1, TAF13, TAF2, TRAPPC9, TRMT1, TTI2*

Mental retardation (X-linked) and microcephaly:
27 genes (88347 bp): ACSL4, ATRX*,**, CASK*,**, DDX3X, EIF2S3, HCFC1, HNRNPH2, HUWE1*, IQSEC1*, KDM5C, L1CAM*,**, MECP2*,**, NEXMIF, OGT, PAK3, PHF6*, POLA1, PQBP1, RPL10, RLIM, SHROOM4, SLC16A2, SLC6A8*,**, SLC9A6, TAF1, THOC2, ZC4H2**

☐ **Seckel syndrome (SCKL)**
Panel-ID: ID113.00 (small)
9 genes (33462 kb): ATR,**, CENPJ*,**, CEP152*, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP*

Microcephaly and Macrocephaly, Multisystemic Malformation Syndromes

☐ **Sotos syndrome (SOTOS)**
Panel-ID: ID181.00 (small)
4 genes (18615 kb): APC2, EZH2, NFIX, NSD1*,**

Multisystemic Malformation Syndromes

☐ **Coffin-Siris syndrome (CSS)**
Panel-ID: ID118.01 (small)
10 genes (34185 kb): ARID1A, ARID1B*, ARID2, DPF2, SMARCC2, SMARCA4*, SMARCB1*,**, SMARCE1, SOX4, SOX11*

☐ **Cornelia de Lange syndrome (CDLS)**
Panel-ID: ID033.02 (small)
8 genes (31959 kb): ANKRD11, BRD4, HDAC8, NIPBL*,**,*

RAD21, SMC1A, SMC3*, SMS*

☐ **Kabuki syndrome (KABUK)**
Panel-ID: ID127.00 (small)
2 genes (20814 kb): KDM6A,**, KMT2D*,***

Skin and Dental Diseases

☐ **Progeria and progeroid syndromes**
Panel-ID: ID147.00 (small)
27 genes (67003 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*

Ear, Nose and Throat Diseases

☐ **Deafness, autosomal dominant (DFNA) or X-linked (DFNX)**
Panel-ID: ID091.02 (large)
50 genes (141771 kb): ACTG1, AIFM1, CCDC50, CD164, CEACAM16, COCH, COL4A6, COL11A1, COL11A2, CRYM, DIABLO, DIAPH1, DMXL2, DSPP, EYA4, GJB2*,**, GJB3*, GJB6*,**, GPRASP2, GRHL2, GSDME*, HOMER2, KCNQ4, KITLG, LMX1A, MCM2, MYH14, MYH9*, MYO6, MYO7A, NLRP3*, OSBPL2, P2RX2, PDE1C, PLS1, POU3F4*, POU4F3, PRPS1, PTPRQ, REST, SIX1, SLC17A8, SLC44A4, SMPX, TBC1D24*, TECTA, TMC1, TNC, TRRAP, WFS1*

Basis-Panel I (DFNA):
24819 bp, 12 genes:
ACTG1, COCH, EYA4, GJB2,**, GJB6*,**, GJB3*, KCNQ4, MYO6, POU4F3, SLC17A8, TECTA, WFS1*

Basis-Panel II (DFNX):
11727 bp, 6 genes:
AIFM1, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX*

☐ **Deafness, autosomal recessive (DFNB) or X-linked (DFNX)**
Panel-ID: ID092.02 (large)
79 genes (218490 kb): ADCY1, AIFM1, BDP1, CABP2, CDC14A, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, 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, PJVK, PNPT1, POU3F4*, PPIP5K2, PRPS1, PTPRQ, RDX, RIPOR2, ROR1, S1PR2, SERPINB6, SLC26A4*, SLC26A5, SMPX, SPNS2, STRC, SYNE4, TBC1D24*, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TPRN, TRIOBP, TSPEAR, USH1C*, WBP2, WHRN**

Basis-Panel I (DFNB):
10 genes (24440 bp + according to tariff EBM 11420): GIPC3, GJB2,**, GJB6*,** (MLPA), LRTOMT, MYO15A, OTOF, SLC26A4, TMC1, TMIE, TMPRSS3*

Basis-Panel II (DFNB):
7 genes (24675 bp + according to tariff EBM 11420): CDH23, CIB2, GJB2,**, GJB6*,** (MLPA), MYO7A, PCDH15, USH1C**

Basis-Panel III (DFNX):
6 genes (11727 bp): AIFM1, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX*

☐ **Deafness, nonsyndromic (DFNA, DFNB, DFNX)**
Panel-ID: ID237.01 (large)
111 genes (310305 kb): ACTG1, ADCY1, AIFM1, BDP1, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, COCH, COL11A1, COL11A2, COL4A6, CRYM, DCDC2, DIABLO, DIAPH1, DMXL2, DSPP, ELMOD3, EPS8, EPS8L2, ESPN, ESRP1, ESRRB, EYA4, GAB1, GIPC3, GJB2*,**, GJB3*, GJB6*,**, 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, SERPINB6, SIX1, SLC17A8, SLC26A4*, SLC26A5, SLC44A4, SMPX, SPNS2, STRC, SYNE4, TBC1D24*, TECTA, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, USH1C*, WBP2, WFS1, WHRN**

Basis-Panel I (DFNA):
24819 bp, 12 genes:
ACTG1, COCH, EYA4, GJB2,**, GJB6*,**, GJB3*, KCNQ4, MYO6, POU4F3, SLC17A8, TECTA, WFS1*

Basis-Panel I (DFNB):
10 genes (24440 bp + according to tariff EBM 11420): GIPC3, GJB2,**, GJB6*,** (MLPA), LRTOMT, MYO15A, OTOF, SLC26A4, TMC1, TMIE, TMPRSS3*

Basis-Panel II (DFNB):
7 genes (24675 bp + according to tariff EBM 11420): CDH23, CIB2, GJB2,**, GJB6*,** (MLPA), MYO7A, PCDH15, USH1C**

Basis-Panel III (DFNX):
6 genes (11727 bp): AIFM1, COL4A6, GPRASP2, POU3F4, PRPS1, SMPX*

☐ **Deafness, syndromic**
Panel-ID: ID190.01 (large)
109 genes (317328 kb): ABHD12, ADGRV1,**, 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*, SPATA5, TCOF1*,**, TFAP2A, TIMM8A*, TWNK, TYR*, USH1C*, USH1G, USH2A*,**, WFS1, WHRN*, ZNF469**

Basis-Panel I (Usher syndrome 1):
*5 genes (24804 bp): CDH23, CIB2, MYO7A, PCDH15, USH1C**

Basis-Panel II (Usher syndrome 2):
3 genes (24738 bp): ADGRV1,**, PDZD7*, WHRN**

Basis-Panel III (Usher syndrome 2, 3 und 4):
5 genes (22125 bp): ARSG, CLRN1, HARS1, USH2A,**, WHRN**

Basis-Panel IV (Stickler syndrome):
6 genes (21048 bp): COL2A1,**, COL9A1, COL9A2, COL9A3, COL11A1*,**, COL11A2*,***

Basis-Panel V (Alport syndrome):
4 genes (21015 bp): COL4A3,**, COL4A4*,**, COL4A5*,**, MYH9**

Basis-Panel VI (Waardenburg syndrome):
8 genes (9342 bp): EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2, SOX10, TYR**

Basis-Panel VII (Perrault syndrome):
6 genes (9288 bp): CLPP, ERAL1, HARS2, HSD17B4, LARS2, TWNK

Basis-Panel VIII (CHARGE syndrome):
2 genes (11319 bp): CHD7,**, SEMA3E*

Eye Diseases, Skin and Dental Diseases

☐ **Albinism, syndromic (HPS, GS, WS)**
Panel-ID: ID175.02 (small)
22 genes (50871 kb): AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, EDN3, EDNRB, HPS1, HPS3, HPS4, HPS5, HPS6, KITLG, LYST, MITF, MLPH, MYO5A, PAX3, RAB27A*, SNAI2*, SOX10*, TYR**

Basis-Panel I (Hermansky-Pudlak syndrome (HPS)):
10 genes (21780 bp): AP3B1, AP3D1, BLOC1S3, BLOC1S6, DTNBP1, HPS1, HPS3, HPS4, HPS5, HPS6

Basis-Panel II (Waardenburg syndrome (WS)):
8 genes (9342 bp): EDN3, EDNRB, KITLG, MITF, PAX3, SNAI2*, SOX10*, TYR**

Basis-Panel III (Griscelli syndrome (GS)):
*3 genes (8028 bp): MLPH, MYO5A, RAB27A**

Endocrine Disorders

Developmental and Growth Disorders

☐ **Growth hormone deficiency**
Panel-ID: ID211.01 (small)
13 genes (18030 kb): BTK,**, GH1*, GHRHR*, GHSR, GLI2, HESX1*,**, LHX3**, LHX4**, OTX2, POU1F1**, PROP1**, RNPC3*, SOX3**

Isolated growth hormone deficiency (IGHD):
6 genes (7902 bp): BTK,**, GH1*,**, GHRHR**, GHSR, RNPC3*, SOX3**

Combined pituitary hormone deficiency (CPHD):
7 genes (10128 bp): GLI2, HESX1,**, LHX3**, LHX4**, OTX2, POU1F1**, PROP1***

Metabolic Diseases

☐ **Diabetes mellitus, neonatal**
Panel-ID: ID162.00 (small)
11 genes (21321 kb): ABCC8, EIF2AK3, GATA6, GCK*,**, GLIS3, INS*,**, KCNJ11*, PDX1*,**, PTF1A, RFX6, ZFP57*

☐ **Hyperinsulinemic hypoglycemia (HHF)**
Panel-ID: ID126.00 (small)
8 genes (16869 kb): ABCC8, KCNJ11*, GCK*,**, HADH, INSR, GLUD1, SLC16A1, HNF4A*,***

☐ **Maturity-onset diabetes of the young (MODY)**
Panel-ID: ID048.01 (small)
14 genes (22986 kb): ABCC8, APPL1, BLK, CEL, GCK*,**, HNF1A*,**, HNF1B*,**, HNF4A*,**, INS*,**, KCNJ11*, KLF11, NEUROD1, PAX4, PDX1*,***

☐ **Glucocorticoid deficiency (GCCD)**
Panel-ID: ID222.00 (small)
16 genes (25056 kb): AAAS, ABCD1, CYP11A1, HSD3B2, MC2R, MCM4, MRAP, NNT, NR0B1*,**, NR3C1*, PCSK1, POMC*,**, PROP1, STAR, TBX19, TXNRD2*

Epilepsy and Migraine

☐ **Absence epilepsy (JAE, CAE)**
Panel-ID: ID057.01 (small)
9 genes (18264 kb): CASR, CLCN2*, EFHC1, GABRA1*, GABRB3, GABRG2*, RORB, SLC2A1*,**, SLC12A5*

☐ **Benign neonatal or infantil seizures (BFNS, BFIS)**
Panel-ID: ID134.01 (small)
6 genes (19797 kb): CHRNA2, KCNQ2,**, KCNQ3*, PRRT2, SCN2A*, SCN8A*

☐ **Early infantile epileptic encephalopathy (EIEE)**
Panel-ID: ID080.01 (large)
76 genes (207030 kb): AARS, ACTL6B, ADAM22, ALG13, AP3B2, ARHGEF9, ARV1, ARX,**, CACNA1A*,**, CACNA1E, CAD, CDKL5*,**, CNPY3, CPLX1, CUX2, CYFIP2, DENND5A, DNM1, DOCK7, EEF1A2, FGF12, FRRS1L, GABBR2, GABRA1*, GABRB1, GABRB3, GABRG2, GLS, GNAO1, GRIN2B*, GRIN2D, GUF1, HCN1, HNRNPU, ITPA, KCNA2, KCNB1, KCNQ2*,**, KCNT1*, KCNT2, MDH2, NECAP1, NEUROD2, NTRK2, PACS2, PARS2, PCDH19*,**, PHACTR1, PIGA, PIGP, PLCB1, PNKP*, RHOBTB2, RNF13, SCN1A*,**, SCN1B*, SCN2A*, SCN3A, SCN8A, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22*, SLC35A2, SPTAN1*, ST3GAL3, STXBP1*,**, SYNJ1, SZT2, TBC1D24*, TRAK1, UBA5, WWOX, YWHAG*

Basis-Panel I (EIEE, dominant):
7 genes (24933 bp): GNAO1, KCNA2, KCNQ2,**, SCN1A*,**, SCN2A*, SCN8A, STXBP1*,***

Basis-Panel II (EIEE, rezessive):
11 genes (25299 bp): AP3B2, FRRS1L, ITPA, NECAP1, MDH2, SLC12A5, SLC13A5, SLC25A22, SZT2, UBA5, WWOX*

Basis-Panel III (EIEE, X-linked):
7 genes (15808 bp): ALG13, ARHGEF9, ARX,**, CDKL5*,**, PCDH19*,**, PIGA, SLC35A2*

☐ Epilepsy with severe developmental disorder
Panel-ID: ID060.00 (small)
20 genes (68364 kb): ARHGEF9, ARX,**, CACNA1A*,**, CDKL5*,**, FOXG1*,**, KCNQ2*,**, MECP2*,**, MEF2C*,**, MTHFR*, PCDH19*,**, SCN1A*,**, SCN2A*, SCN8A, SCN9A*, SLC25A22*, SLC2A1*,**, SPTAN1, STXBP1*,**, SYNGAP1*, UBE3A*,**

☐ Epilepsy, comprehensive disgnostics
Panel-ID: ID061.02 (large)
139 genes (342600 kb): AARS, ACTL6B, ADAM22, ADRA2B, ALDH7A1, ALG13, AP3B2, ARHGEF9, ARV1, ARX*,**, ASAH1, ATP1A2*,**, ATP6V1A, BRAT1, CACNA1A*,**, CACNA1E, CACNA1H*, CACNB4*, CAD, CASR*, CDKL5*,**, CERS1, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2*, CLN8, CNPY3, CNTN2, CNTNAP2*, CPA6*, CPLX1, CSTB, CUX2, CYFIP2, DENND5A, DEPDC5, DNM1, DOCK7, EEF1A2, EFHC1, EPM2A, FGF12, FRRS1L, GABBR2, GABRA1*, GABRB1, GABRB2, GABRB3, GABRD*, GABRG2*, GAL, GLS, GNAO1, GOSR2, GRIN2A*, GRIN2B*, GRIN2D, GUF1, HCN1, HNRNPU, ICK, ITPA, KCNA2, KCNB1, KCNC1, KCNMA1, KCNQ2*,**, KCNQ3*, KCNT1*, KCNT2, KCTD7, LGI1, LMNB2, MDH2, MECP2*,**, MEF2C*,**, MTOR, NECAP1, NEUROD2, NHLRC1, NPRL2, NPRL3*, NTRK2, PACS2, PARS2, PCDH19*,**, PHACTR1, PIGA, PIGP, PLCB1, PNKP*, PNPO*, POLG*,**, PLPBP*, PPP3CA, PRDM8, PRICKLE1, PRRT2, RELN, RHOBTB2, RNF13, ROGDI*, RORB, SAMD12, SCARB2, SCN1A*,**, SCN1B*, SCN2A*, SCN3A, SCN8A, SCN9A*, SIK1, SLC12A5, SLC13A5, SLC1A2, SLC25A12, SLC25A22*, SLC2A1*,**, SLC35A2, SLC6A1, SNIP1, SPTAN1*, SRPX2, ST3GAL3, ST3GAL5, STX1B*, STXBP1*,**, SYN1, SYNGAP1*, SYNJ1, SZT2, TBC1D24*, TCF4, TRAK1, UBA5, WWOX, YWHAG*

Basis-Panel I (Focal epilepsy):
11 genes (24378 bp): CHRNA2, CHRNA4, CHRNB2, CPA6, DEPDC5*, GAL, GRIN2A*, KCNT1*, LGI1, NPRL2, NPRL3**

Basis-Panel II (Absence epilepsy):
10 genes (25011 bp): CACNA1H, CASR*, CLCN2*, EFHC1, GABRA1*, GABRB3, GABRG2*, RORB, SLC2A1*,**, SCL12A5*

Basis-Panel III (Myoclonic epilepsy):
11 genes (23530 bp): ADRA2B, CACNB4, CLCN2*, CNTN2, EFHC1, GABRA1*, GABRD*, ICK, SAMD12, SCN1A*,**, TBC1D24**

Basis-Panel IV (Progressive myoclonic epilepsy):
13 genes (19327 bp): ASAH1, CERS1, CSTB, EPM2A, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG,**, PRDM8, PRICKLE1, SCARB2*

Basis-Panel V (Epilepsy with febrile seizures):
7 genes (18594 bp): GABRD, GABRG2*, HCN, SCN1A*,**, SCN1B*, SCN9A*, STX1B**

Basis-Panel VI (Epileptic encephalopathy, XL):
7 genes (15808 bp): ALG13, ARHGEF9, ARX,**, CDKL5*,**, PCDH19*,**, PIGA, SLC35A2*

Basis-Panel VII (Epileptic encephalopathy, AD):
7 genes (24933 bp): GNAO1, KCNA2, KCNQ2,**, SCN1A*,**, SCN2A*, SCN8A, STXBP1*,**

Basis-Panel VIII (Epileptic encephalopathy, AR):
11 genes (25299 bp): AP3B2, FRRS1L, ITPA, NECAP1, MDH2, SLC12A5, SLC13A5, SLC25A22, SZT2, UBA5, WWOX*

Basis-Panel IX (Benign seizures):
6 genes (19407 bp): CHRNA2, KCNQ2,**, KCNQ3*, PRRT2, SCN2A*, SCN8A*

☐ Epileptic encephalopathy, comprehensive diagnostics
Panel-ID: ID047.02 (large)
99 genes (249327 kb): AARS, ADAR, ACTL6B, ADAM22, ALG13, AMT*, AP3B2, ARHGEF9, ARV1, ARX*,**, ATP6V1A, BRAT1, CACNA1A*,**, CACNA1E, CAD, CDKL5*,**, CHD2, CNPY3, CPLX1, CUX2, CYFIP2, DENND5A, DNM1, DNM1L, DOCK7, EEF1A2, FGF12, FRRS1L, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GCSH*, GLDC*,**, GLS, GNAO1, GRIN2B*, GRIN2D, GUF1, HCN1, HNRNPU, IFIH1*, ITPA, KCNA2, KCNB1, KCNQ2*,**, KCNT1*, KCNT2, MECP2*,**, MDH2, MFF, NECAP1, NEUROD2, NTRK2, PACS2, PARS2, PCDH19*,**, PHACTR1, PIGA, PIGP, PLCB1, PNKP*, PNPO*, PPP3CA, RHOBTB2, RNASEH2A*,**, RNASEH2B*,**, RNASEH2C*,**, RNF13, SAMHD1, SCN1A*,**, SCN1B*, SCN2A*, SCN3A, SCN8A, SCN9A*, SERPINI1, SIK1, SLC1A2, SLC12A5, SLC13A5, SLC2A1*,**, SLC25A12, SLC25A22*, SLC35A2, SPTAN1*, ST3GAL3, STXBP1*,**, SYNJ1, SZT2, TBC1D24*, TCF4, TRAK1, TREX1*,**, UBA5, WWOX, YWHAG*

Basis-Panel I (Encephalopathy, dominant):
7 genes (24912 bp): GNAO1, KCNQ2,**, SCN1A*,**, SCN2A*, SCN8A, SLC2A1*,**, STXBP1*,**

Basis-Panel II (Encephalopathy, rezessive):
11 genes (25299 bp): AP3B2, FRRS1L, ITPA, NECAP1, MDH2, SLC12A5, SLC13A5, SLC25A22, SZT2, UBA5, WWOX*

Basis-Panel III (Encephalopathy, X-linked):
8 genes (17266 bp): ALG13, ARHGEF9, ARX,**, CDKL5*,**, MECP2*,**, PCDH19*,**, PIGA, SLC35A2*

Basis-Panel IV (Aicardi-Goutiéres syndrome):
7 genes (11898 bp): ADAR, IFIH1*, RNASEH2A*,**, RNASEH2B*,**, RNASEH2C*,**, SAMHD1*,**, TREX1*,**

Basis-Panel V (Glycine encephalopathy):
3 genes (4788 bp): AMT, GCSH, GLDC*,**

☐ Febrile seizures with or without epilepsy (FEB, GEFS+)
Panel-ID: ID059.02 (small)
9 genes (38859 kb): ADGRV1,**, CPA6, GABRD*, GABRG2*, HCN1, SCN1A*,**, SCN1B*, SCN9A*, STX1B**

Basis-Panel I (FEB):
5 genes (25076 bp): ADGRV1,** (spec. exons), CPA6, GABRG2*, SCN1A*,**, SCN9A**

Basis-Panel II (GEFS+):
7 genes (18894 bp): GABRD, GABRG2*, HCN1, SCN1A*,**, SCN1B*, SCN9A*, STX1B**

☐ Focal epilepsy
Panel-ID: ID208.01 (small)
17 genes (50919 kb): CHRNA2, CHRNA4, CHRNB2, CNTNAP2, CPA6*, DEPDC5*, GAL, GRIN2A*, KCNT1*, LGI1, NPRL2, NPRL3*, PCDH19*,**, RELN, SCN3A, SRPX2, TBC1D24**

☐ Generalized epilepsy
Panel-ID: ID040.03 (small)
42 genes (77532 kb): ADRA2B, ALDH7A1, ASAH1, CACNB4*, CASR*, CERS1, CLCN2*, CLN8, CNTN2, CSTB, EFHC1, EPM2A, GABRA1*, GABRB3, GABRD*, GABRG2*, GOSR2, HCN1, ICK, KCNC1, KCNMA1, KCTD7, LMNB2, NHLRC1, PLPBP*, POLG*,**, PRDM8, PRICKLE1, ROGDI*, RORB, SAMD12, SCARB2, SCN1A*,**,SCN1B*, SCN9A*, SLC2A1*,**, SLC6A1, SLC12A5, SNIP1, ST3GAL5, STX1B*, TBC1D24**

Basis-Panel I (Epilepsy with febrile seizures):
7 genes (18594 bp): GABRD, GABRG2*, HCN, SCN1A*,**, SCN1B*, SCN9A*, STX1B**

Basis-Panel II (Absence epilepsy):
9 genes (18264 bp): CASR, CLCN2*, EFHC1, GABRA1*, GABRB3, GABRG2*, RORB, SLC2A1*,**, SLC12A5*

Basis-Panel III (Myoclonic epilepsy):
11 genes (23530 bp): ADRA2B, CACNB4, CLCN2*, CNTN2, EFHC1, GABRA1*, GABRD*, ICK, SAMD12, SCN1A*,**, TBC1D24**

Basis-Panel IV (Progressive myoclonic epilepsy):
13 genes (19327 bp): ASAH1, CERS1, CSTB, EPM2A, GABRA1, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG*,**, PRDM8, PRICKLE1, SCARB2*

☐ **Generalized epilepsy with febrile seizures plus (GEFSP, GEFS+)**
Panel-ID: ID235.01 (small)
7 genes (18924 kb): GABRD, GABRG2*, HCN1, SCN1A*,**, SCN1B*, SCN9A, STX1B**

☐ **Hemiplegic migraine (FHM)**
Panel-ID: ID064.00 (small)
3 genes (16572 kb): ATP1A2,**, CACNA1A*,**, SCN1A*,***

☐ **Migraine (MGR)**
Panel-ID: ID065.01 (small)
16 genes (44754 kb): ALPK1, ATP1A2,**, ATP1A3, CACNA1A*,**, CSNK1D, ESR1, HTRA1*, KCNK18, NOTCH3*,**, POLG*,**, PRRT2*, SCN1A*,**, SLC1A3*, SLC2A1*,**, TNF, TREX1*,***

Familial hemiplegic migraine (FHM):
3 genes (16572 bp): ATP1A2,**, CACNA1A*,**, SCN1A*,***

☐ **Myoclonic epilepsy (JME, PME)**
Panel-ID: ID116.01 (small)
24 genes (43020 kb): ADRA2B, ASAH1, CERS1, CLCN2, CNTN2, CSTB, CACNB4*, EFHC1, EPM2A, GABRA1*, GABRD*, GOSR2, ICK, KCNC1, KCTD7, LMNB2, NHLRC1, POLG*,**, PRDM8, PRICKLE1, SAMD12, SCARB2, SCN1A*,**, TBC1D24**

Basis-Panel I:
11 genes (23530 bp): ADRA2B, CACNB4, CLCN2*, CNTN2, EFHC1, GABRA1*, GABRD*, ICK, SAMD12, SCN1A*,**, TBC1D24**

Basis-Panel II (Progressive epilepsy):
13 genes (19327 bp): ASAH1, CERS1, CSTB, EPM2A, GABRA1, GOSR2, KCNC1, KCTD7, LMNB2, NHLRC1, POLG*,**, PRDM8, PRICKLE1, SCARB2*

Metabolic Diseases

☐ **Metabolic disorder with epilepsy in childhood and adolescence**
Panel-ID: ID172.00 (small)
15 genes (28131 kb): ASAH1, ATN1, CLN3, CLN5, CLN6, CSTB, DNAJC5, EPM2A, GBA,**, GOSR2, HTT, NEU1, NHLRC1, PRICKLE1, SCARB2*

☐ **Metabolic disorder with epilepsy in infancy and childhood**
Panel-ID: ID171.00 (small)
18 genes (31542 kb): ALDH5A1, ATP7A,**, BTD*, FOLR1, GAMT*, GATM, HEXA, HEXB, HLCS*, KCTD7, MTHFR, PHGDH, POLG*,**, PPT1*, SLC19A3, SLC2A1*,**, SLC6A8*,**, TPP1**

☐ **Metabolic disorder with neonatal epilepsy**
Panel-ID: ID135.00 (small)
25 genes (39423 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**

Eye Diseases

☐ **Achromatopsia (ACHM)**
Panel-ID: ID164.01 (small)
7 genes (11517 kb): ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, OPN1LW*

☐ **Age-related macular degeneration (ARMD)**
Panel-ID: ID186.00 (small)
16 genes (50970 kb): ABCA4, APOE*, ARMS2**, C2**, C3, C9, CFB, CFH**, CFI*, CST3, CX3CR1, ERCC6, FBLN5, HMCN1, HTRA1*, RAX2*

☐ **Anterior segment dysgenesis (ASGD)**
Panel-ID: ID182.01 (small)
8 genes (17466 kb): CPAMD8, CYP1B1, FOXC1*,**, FOXE3*, PAX6*,**, PITX2*,**, PITX3, PXDN*

☐ **Cataract (CTRCT)**
Panel-ID: ID206.01 (small)
37 genes (59604 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*

Basis-Panel I (autosomal dominant):
24 genes (23655 bp): BFSP1, BFSP2, CHMP4B, CRYAA, CRYAB, CRYBA1, CRYBA2, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGB, CRYGC, CRYGD, CRYGS, EPHA2, GJA3, GJA8, HSF4, MAF, MIP, PITX3, SLC16A12, VIM

Basis-Panel II (autosomal rezessive):
14 genes (25131 bp): AGK, BFSP1, CRYAA, CRYAB, CRYBB1, CRYBB3, DNMBP, FOXE3, FYCO1, GCNT2, LEMD2, LIM2, LSS, TDRD7

☐ **Cone-rod dystrophy (CORD) and Cone dystrophy (COD)**
Panel-ID: ID101.02 (small)
29 genes (72417 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 (63441 bp): 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 (15342 bp): CACNA2D4, GUCA1A, GUCY2D, KCNV2, OPN1LW, PDE6C, PDE6H, RPGR*,***

☐ **Congenital fibrosis of the extraocular muscles (CFEOM)**
Panel-ID: ID063.00 (small)
5 genes (10484 kb): COL25A1, KIF21A, PHOX2A, TUBB2B, TUBB3**

☐ **Fuchs endothelial corneal dystrophy (FECD)**
Panel-ID: ID261.00 (small)
5 genes (13464 kb): AGBL1, COL8A2, SLC4A11, TCF4,**, ZEB1*

☐ **Glaucoma (GLC)**
Panel-ID: ID275.00 (small)
23 genes (51656 kb): ASB10, COL8A2, COL18A1, CPAMD8, CYP1B1, FOXC1*,**, FOXE3*, GPATCH3, LMX1B, LTBP2, MYOC, NTF4, OPA1*,**, OPTN, PAX6*,**, PITX2*,**, PITX3, PXDN, RAMP2, SLC4A11, TBK1, TEK*, WDR36*

☐ **Leber congenital amaurosis (LCA)**
Panel-ID: ID187.01 (small)
19 genes (38769 kb): AIPL1, CEP290*, CRB1, CRX, GDF6*, GUCY2D, IMPDH1, KCNJ13, LCA5, LRAT, NMNAT1, PRPH2*, RD3, RDH12*, RPE65, RPGRIP1*, SPATA7, TULP1, USP45*

☐ **Macular dystrophy (MD)**
Panel-ID: ID139.01 (small)
15 genes (39561 kb): ABCA4, BEST1, CDH3, CNGB3*, CTNNA1, ELOVL4*, IMPG1, IMPG2*, MAPKAPK3, MFSD8, PRDM13, PROM1, PRPH2*, RP1L1, TIMP3*

☐ **Microphthalmia (MCOP) and Coloboma**
Panel-ID: ID263.00 (small)
47 genes (90930 kb): ABCB6, ACTB, ACTG1, ALDH1A3, BCOR, BMP4, CHD7,**, C12ORF57, CC2D2A*, CRYAA, FOXE3*, GDF3, GDF6*, GLI2, HCCS, HMGB3, HMX1, MAB21L2, MAF, MFRP, MITF, NAA10, OTX2, PAX2, PAX6*,**, PDE6D, PITX3, PTCH1*,**, PRSS56, RBP4, RARB, RAX, RPGRIP1L*, SALL2, SEMA3A, SHH*,**, SIX3, SMOC1, SOX2*, SRD5A3, STRA6, TENM3, TFAP2A, TMEM67*, VAX1, VSX2, YAP1*

Basis-Panel I (Microphthalmia):
17 genes (25115 bp): ABCB6, ALDH1A3, BCOR, BMP4, HCCS, HMGB3, MAB21L2, MFRP, NAA10, OTX2, PRSS56, RBP4, RARB, RAX, SOX2, STRA6, VSX2*

Basis-Panel II (Coloboma):
11 genes (23985 bp): ABCB6, GDF3, GDF6, PAX6*,**, RBP4, SALL2, SHH*,**, STRA6, TENM3, VSX2, YAP1*

☐ **Myopia**
Panel-ID: ID079.01 (small)
15 genes (37068 kb): ARR3, CPSF1, COL11A1,**, COL2A1*,**, COL9A1, COL9A2, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLITRK6, ZNF644*

Basis-Panel I:
11 genes (22335 bp): ARR3, CPSF1, LRPAP1, NYX, P3H2, P4HA2, PRIMPOL, SCO2, SLC39A5, SLITRK6, ZNF644

Basis-Panel II (Stickler syndrome):
4 genes (14709 bp): COL11A1,**, COL2A1*,**, COL9A1, COL9A2*

☐ **Night blindness, congenital stationary (CSNB)**
Panel-ID: ID267.00 (small)
14 genes (39210 kb): CACNA1F, GNAT1, GNB3, GPR179, GRK1, GRM6, GUCY2D, LRIT3, NYX, PDE6B, RHO, SAG, SLC24A1, TRPM1*

☐ **Optic atrophy (OPA)**
Panel-ID: ID081.03 (small)
*22 genes (32031 kb): ACO2**, ATP1A3, ANTXR1, C12ORF65, CISD2, DNM1L*, FDXR, MECR, MFN2*,**, NR2F1, OPA1*, OPA3*, PDXK, PRPS1, RTN4IP1, SLC25A46, TBCE, TMEM126A, TIMM8A*, UCHL1, WFS1, YME1L1*

☐ **Retinitis pigmentosa (RP), X-linked or autosomal dominant**
Panel-ID: ID053.01 (small)
28 genes (59718 kb): AIPL1, ARL3, BEST1, CA4, FSCN2, GUCA1B, HK1, IMPDH1, KLHL7*, NR2E3*, NRL, OFD1*, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2*, RGR*, RHO*, RP1*, RP2*, RP9, RPE65, RPGR*, SEMA4A, SNRNP200, TOPORS*

Basis-Panel I (autosomal dominant):
10 genes (25020 bp): GUCA1B, IMPDH1, KLHL7, NR2E3*, PRPF31, PRPF8, PRPH2, RHO*, RP1*, SEMA4A*

Basis-Panel II (X-linked):
3 genes (6531 bp): RP2, RPGR*, OFD1**

☐ **Retinitis pigmentose (RP), X-linked or autosomal recessive**
Panel-ID: ID050.02 (large)
57 genes (150015 kb): ABCA4, AGBL5, AHR, ARHGEF18, ARL2BP, ARL6, BBS2*, C8ORF37, CDHR1, CERKL, CLRN1, CNGA1, CNGB1, CRB1, DHDDS, DHX38, EYS, FAM161A, HGSNAT, IDH3B, IFT43, IFT140, IFT172, IMPG2*, KIAA1549, KIZ, LRAT, MAK, MERTK*, NEK2, NR2E3, OFD1*, PCARE*, PDE6A*, PDE6B, PDE6G, POMGNT1, PRCD, PROM1, RBP3, RDH12, REEP6, RGR*, RHO*, RP1*, RP2*, RPE65, RPGR*, SAG, SEMA4A, SLC7A14, SPATA7, TTC8*, TULP1, USH2A*,**, ZNF408, ZNF513*

Basis-Panel I (autosomal rezessive):
2 genes (25047 bp): EYS, USH2A,**

Basis-Panel II (autosomal rezessive):
9 genes (25039 bp): ABCA4, CERKL, CRB1, MERTK, PDE6A, PDE6B, RHO*, RPE65, TULP1*

Basis-Panel III (X-linked):
3 genes (6531 bp): RP2,**, RPGR*,**, OFD1**

☐ **Stargardt disease (STGD)**
Panel-ID: ID102.00 (small)
5 genes (13821 kb): ABCA4, CNGB3*, ELOVL4*, PROM1, PRPH2*

Ear, Nose and Throat Diseases

☐ **Usher syndrome (USH)**
Panel-ID: ID034.01 (small)
13 genes (70332 kb): ADGRV1,**, ARSG, CDH23, CIB2, CLRN1, HARS1, MYO7A, PCDH15, PDZD7*, USH1C*, USH1G, USH2A*,**, WHRN**

Basis-Panel I (USH1):
*5 genes (24804 bp): CDH23, CIB2, MYO7A, PCDH15, USH1C**

Basis-Panel II (USH2):
3 genes (24738 bp): ADGRV1,**, PDZD7*, WHRN**

Basis-Panel II (USH2, USH3, USH4):
5 genes (22125 bp): ARSG, CLRN1, HARS1, USH2A,**, WHRN**

Gastroenterological Diseases

☐ **Hirschsprung disease (HSCR)**
Panel-ID: ID177.00 (small)
10 genes (16878 kb): ASCL1, ECE1, EDN3, EDNRB, GDNF, KIF1BP, PHOX2B,**, RET*,**, SOX10, ZEB2*,***

☐ **Pancreatitis (PCTT)**
Panel-ID: ID141.02 (small)
18 genes (29559 kb): APOA5, APOC2*, CASR*, CEL, CELA3B, CFTR*,**, CLDN2, CPA1*, CTRC*, CTSB, GPIHBP1*, LMF1*, LPL*,**, PNLIP, PRSS1*,**, SPINK1*,**, TRPV6, UBR1*

☐ **Visceral myopathy (VSCM)**
Panel-ID: ID238.01 (small)
18 genes (43179 kb): ACTA2, ACTG2*, CHRM3*, EDNB, EDNRB, FLNA*,**, GDNF, LMOD1*, MYL9*, MYLK*, MYH11*, POLG*,**, RAD21, RET*,**, RRM2B, SGO1*, SOX10*, TYMP*

Cardiovascular Diseases

☐ **Alagille syndrome (ALGS)**
Panel-ID: ID112.00 (small)
2 genes (11067 kb): JAG1,**, NOTCH2**

Tumor Syndromes

☐ **Gastrointestinal stromal tumor (GIST)**
Panel-ID: ID226.00 (small)
8 genes (18963 kb): KIT,**, NF1*,**, PDGFRA, SDHA*,**, SDHAF2, SDHB*, SDHC, SDHD**

Hematologic Diseases

☐ **Erythrocytosis (ECYT)**
Panel-ID: ID138.02 (small)
11 genes (13854 kb): BPGM, EGLN1, EPAS1, EPO, EPOR, HBA1, HBA2, HBB,**, JAK2, SH2B3, VHL*,***

☐ **Hemophilia**
Panel-ID: ID154.00 (small)
3 genes (16875 kb): F8, F9, VWF

☐ **Platelet disorders, comprehensive diagnostics**
Panel-ID: ID274.00 (small)
47 genes (98583 kb): ACTN1, ADAMTS13, ANKRD26, ANO6, ARPC1B, CD36, CDC42, CYCS, DIAPH1, EPHB2, ETV6, FLI1, FYB1, GATA1, GFI1B, GP1BA, GP1BB*, GP6, GP9*, HOXA11*, IKZF5, ITGA2*, ITGA2B*, ITGB3*, JAK2, MASTL, MECOM, MPIG6B, MPL, MYH9*, NBEAL2*, P2RY12, PLAU, PRKACG, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, TBXA2R, TBXAS1, THPO, TUBB1, WAS*, WDR1, WIPF1*

Basis-Panel I (Thrombocytopenia):
*12 genes (24929 bp): ADAMTS13, ANKRD26, CYCS, ETV6, FYB1, GATA1, GFI1B, MASTL,MPL, RUNX1, SRC, WAS**

Basis-Panel II (Macrothrombocytopenia):
12 genes (22827 bp): ACTN1, CDC42, FLI1, GFI1B, GP1BA, GP1BB*, GP9*, ITGA2B*, ITGB3*, MYH9*, NBEAL2*, PRKACG, RASGRP2, TUBB1*

Basis-Panel III (Thrombocytopathy (BDPLT)):
8 genes (24579 bp): GP1BA, GP1BB*, GP9*, ITGA2B*, ITGB3*, MYH9*, NBEAL2*, RASGRP2*

Basis-Panel IV (Thrombocythemia (THCYT)):
3 genes (6360 bp): JAK2, MPL, THPO

☐ **Spherocytosis (SPH) and Elliptocytosis (EL)**
Panel-ID: ID203.01 (small)
6 genes (26811 kb): ANK1, EPB41, EPB42, SLC4A1, SPTA1, SPTB*

Spherocytosis (SPH):
5 genes (24204) bp: ANK1, EPB42, SLC4A1, SPTA1, SPTB**

Elliptocytosis (EL):
4 genes (18423 bp): EPB41, SLC4A1, SPTA1, SPTB*

☐ **Thrombocytopathy (BDPLT)**
Panel-ID: ID119.00 (small)
22 genes (50955 kb): ACTN1, ANO6, CD36, EPHB2, FLI1, GFI1B, GP1BA, GP1BB*, GP6, GP9*, ITGA2*, ITGA2B*, ITGB3*, MYH9*, NBEAL2*, P2RY12, PLAU, PRKACG, RASGRP2, SLFN14, TBXA2R, TBXAS1*

☐ **Thrombocytopenia (THC)**
Panel-ID: ID104.01 (small)
29 genes (61125 kb): ACTN1, ADAMTS13, ANKRD26, ARPC1B, CYCS, DIAPH1, ETV6, FYB1, GATA1, GFI1B, HOXA11, IKZF5, ITGA2B*, ITGB3*, MASTL, MECOM, MPIG6B, MPL, MYH9*, RBM8A, RUNX1, SLFN14, SRC, STIM1, THPO, TUBB1, WAS*, WDR1, WIPF1*

☐ **Thrombophilia (THPH)**
Panel-ID: ID150.01 (small)
12 genes (25188 kb): F2, F5, F9, F13B, HABP2, HRG, MTHFR, PROC,**, PROS1*,**, SERPINC1*, SERPIND1, THBD*

Metabolic Diseases

☐ **Porphyria**
Panel-ID: ID153.01 (small)
10 genes (12765 kb): ALAD, ALAS2, CLPX, CPOX, FECH, HFE,**, HMBS, PPOX, UROD, UROS*

Immunological Diseases

☐ **Autoinflammatory syndromes, comprehensive diagnostics**
Panel-ID: ID087.03 (small)
34 genes (58716 kb): ADA2, ACP5, AP1S3, ARPC1B, CARD14, ELANE, IL1RN, IL36RN, LPIN2, MEFV,**, MVK*, NLRC4, NLRP1, NLRP12, NLRP3*, NOD2*, NTRK1, OTULIN, PLCG2, POMP, PSMA3, PSMB4, PSMB8, PSMB9, PSTPIP1, RBCK1, RIPK1, SLC29A3, TMEM173, TNFAIP3, TNFRSF1A*, TRAF3IP2, TRNT1*, WDR1*

Basis-Panel (Recurrent fever syndromes):
14 genes (24546 bp): ADA2, ELANE, IL36RN, MEFV,**, MVK*, NLRC4, NLRP12, NLRP3*, NTRK1, OTULIN, RIPK1, TNFRSF1A*, TRNT1*, WDR1*

☐ **Hyper-IgE syndrome**
Panel-ID: ID240.00 (small)
7 genes (22629 kb): DOCK8, DSG1, PGM3, SPINK5, STAT3, TYK2, ZNF341

☐ **Periodic fever syndrome (HPF)**
Panel-ID: ID088.02 (small)
7 genes (18054 kb): MEFV,**, MVK*, NLRC4, NLRP12, NLRP3*, PLCG2, TNFRSF1A**

Hematologic Diseases

☐ **Neutropenia**
Panel-ID: ID189.01 (small)
17 genes (22708 kb): CLPB, CSF3R, CXCR4, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1*, JAGN1, SBDS*, TAZ*, TCIRG1, USB1, VPS45, WAS*, WDR1*

Kidney Diseases

☐ **Bartter syndrome (BARTS)**
Panel-ID: ID156.01 (small)
8 genes (17700 kb): BSND, CASR, CLCNKA, CLCNKB,**, KCNJ1*, MAGED2, SLC12A1*, SLC12A3*

☐ **Congenital anomalies of kidney and urinary tract (CAKUT)**
Panel-ID: ID229.00 (large)
50 genes (171396 kb): ACE, AGT, AGTR1, BICC1, BMP4, BNC2, CDC5L, CHD1L, CRKL, DSTYK, EYA1, FAT4, FGF20, FRAS1, FREM1, FREM2, GATA3, GLI3*,**, GREB1L, GRIP1, HNF1B*,**, HPSE2, ITGA8*, KIF14, LIFR, LRIG2, LRP4, MUC1*, NEK8, NPHP3, NRIP1, PAX2, PBX1, REN, RET*,**, ROBO1, ROBO2, SALL1, SIX1, SIX2, SIX5, SLIT2, SOX17, TBC1D1, TBX18*, TFAP2A, TNXB*,**, TRAP1, UMOD, UPK3A, WNT4**

Basis-Panel I:
11 genes (11 genes): HD1L, DSTYK, HNF1B*,**, NRIP1, PAX2, PBX1, SALL1, SIX2, TBC1D1, TBX18*, TRAP1*

Basis-Panel II (Vesicoureteral reflux):
7 genes (25191 bp): HPSE2, LRIG2, PAX2, ROBO2, SOX17, TNXB,**, UPK3A*

Basis-Panel III (Branchiootorenal syndrome):
4 genes (8751 bp): EYA1, SALL1, SIX1, SIX5

Basis-Panel IV (Renal tubular dysgenesis):
4 genes (7637 bp): ACE, AGT, AGTR1, REN

Basis-Panel V (Fraser syndrome):
3 genes (24771 bp): FRAS1, FREM2, GRIP1

Basis-Panel VI (Renal hypodysplasia/aplasia):
10 genes (22761 bp) DSTYK, FGF20, GATA3, GREB1L, HNF1B, ITGA8*, PAX2, RET*,**, UPK3A, WNT4**

☐ **Focal segmental glomeruloscerosis (FSGS) and Nephrotic syndrome (SRNS, NPHS)**
Panel-ID: ID098.03 (large)
49 genes (142740 kb): ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, AVIL, CD2AP, COL4A3*,**, COL4A4*,**, COL4A5*,**, COQ2*, COQ6, COQ8B, CRB2, CUBN, DGKE*, EMP2, INF2*, ITGA3, KANK1, KANK2, KANK4, LAGE3, LAMB2, LMX1B, MAGI2, MYH9*, MYO1E, NPHS1, NPHS2*, NUP85, NUP93, NUP107, NUP133, NUP160, NUP205, OSGEP, PAX2, PLCE1, PTPRO*, SGPL1, SMARCAL1, TBC1D8B, TP53RK, TPRKB, TRPC6*, WDR4, WDR73, WT1*,**

Basis-Panel I (Nephrotic syndrome (NPHS)):
10 genes (25263 bp): ARHGDIA, COQ8B, EMP2, LAMB2, NPHS1, NPHS2, NUP93, PLCE1, SGPL1, WT1*,**

Basis-Panel II (Focal segmental glomerulosclerosis (FSGS)):
9 genes (25236 bp): ACTN4, ANLN, ARHGAP24, CRB2, CD2AP, INF2*, MYO1E, PAX2, TRPC6**

Basis-Panel III (Alport syndrome (ATS)):
4 genes (21015 bp): COL4A3,**, COL4A4*,**, COL4A5*,**, MYH9**

Basis-Panel IV (Galloway-Mowat syndrome (GAMOS)):
8 genes (11355 bp): LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73

☐ **Glomerulonephritis**
Panel-ID: ID103.01 (small)
18 genes (36942 kb): C1QA, C1QB, C1QC, C2, C3, CD46, CFB, CFI, CFH, CFHR1, CFHR2, CFHR3, CFHR5, DGKE, FN1, PRKCD, SOX18, THBD

☐ **Polycystic kidney disease (PKD)**
Panel-ID: ID100.03 (small)
13 genes (50319 kb): ANKS6, BICC1, DNAJB11, DZIP1L*, GANAB*, HNF1B*,**, MUC1, NEK8, NPHP3, PKD1*,**, PKD2*,**, PKHD1*,**, UMOD*

Basis-Panel I (dominant):
7 genes (24195 bp): DNAJB11, GANAB*, HNF1B*,**, MUC1, PKD1*,**, PKD2*,**, UMOD*

Basis-Panel II (rezessiv):
5 genes (23202 bp): ANKS6, DZIP1L, NEK8, NPHP3, PKHD1*,**

Ciliopathies

☐ **Nephronophthisis (NPHP)**
Panel-ID: ID030.01 (small)
21 genes (69291 kb): ANKS6, CEP83, CEP164, CEP290, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1*,**, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67*, TTC21B, WDR19, XPNPEP3, ZNF423*

Endocrine Disorders

☐ **Pseudohypoaldosteronism (PHA)**
Panel-ID: ID250.00 (small)
8 genes (23790 kb): CUL3, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4

Hematologic Diseases

☐ **Atypical hemolytic uremic syndrome AHUS)**
Panel-ID: ID163.02 (small)
*16 genes (33228 kb): ADAMTS13, C2**, C3, CD46, CFB, CFH**, CFHR1**, CFHR2**, CFHR3**, CFHR4, CFHR5**, CFI*, DGKE*, MMACHC, MTHFD1, THBD*

Metabolic Diseases

☐ **Hypomagnesemia (HOMG)**
Panel-ID: ID054.01 (small)
18 genes (39207 kb): ATP1A1, BSND, CASR*, CLCNKA, CLCNKB*,**, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B*,**, KCNA1*,**, KCNJ1*, KCNJ10, MAGED2, SLC12A1*, SLC12A3*, TRPM6*

Renal hypomagnesemia (HOMG):
11 genes (24582 bp): ATP1A1, CLDN16, CLDN19, CNNM2, EGF, FXYD2, HNF1B,**, KCNA1*,**, KCNJ10, SLC12A3*, TRPM6*

Bartter syndrome (BARTS):
7 genes (14625 bp): BSND, CASR*, CLCNKA, CLCNKB*,**, KCNJ1*, MAGED2, SLC12A1**

☐ **Nephrolithiasis and Nephrocalcinosis**
Panel-ID: ID231.03 (small)
32 genes (58965 kb): AGXT, ALPL,**, APRT, ATP6V0A4, ATP6V1B1, CASR*, CLCN5, CLDN16, CLDN19, CYP24A1*, G6PC, GRHPR, HOGA1, HPRT1, KCNJ1*, MAGED2, MOCOS, OCRL*, SLC2A9, SLC3A1*,**, SLC4A1, SLC4A4, SLC6A19, SLC6A20, SLC7A9*, SLC12A1*, SLC22A12, SLC26A1, SLC34A1*, SLC34A3, SLC36A2, XDH*

Hypercalciuria:
11 genes (21306 bp): CASR, CLCN5, CLDN16, CLDN19, CYP24A1*, KCNJ1*, MAGED2, OCRL*, SLC12A1*, SLC34A1*, SLC34A3*

Hyperoxaluria:
4 genes (5244 bp): AGXT, GRHPR, HOGA1, SLC26A1

Hypouricosuria and Hyperuricosuria:
7 genes (12120 bp): APRT, G6PC, HPRT1, MOCOS, SLC2A9, SLC22A12, XDH

Hyperglycinuria:
3 genes (52374 bp): SLC6A19, SLC6A20, SLC36A2

Hypocitraturia:
4 genes (11108 bp): ATP6V0A4, ATP6V1B1, SLC4A1, SLC4A2

Cystinuria:
2 genes (3516 bp): SLC3A1, SLC7A9*,**

Microcephaly and Macrocephaly, Brain Malformations

☐ **Galloway-Mowat syndrome (GAMOS)**
Panel-ID: ID251.00 (small)
8 genes (11355 kb): LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73

Multisystemic Malformation Syndromes

☐ **Alport syndrome (ATS)**
Panel-ID: ID099.00 (small)
4 genes (21015 kb): COL4A3,**, COL4A4*,**, COL4A5*,**, MYH9**

Lipometabolic Disorders

☐ **Combined hyperlipidemia**
Panel-ID: ID025.05 (small)
14 genes (28344 kb): APOA5, APOB* (Exon 26), APOC2*, APOE*, GCKR*, GPIHBP1*, LDLR*,**, LDLRAP1*, LIPC*, LPL*,**, NPC1L1*, PCSK9*, SORT1*, USF1**

☐ **Disorders of lipid metabolism, comprehensive diagnostics**
Panel-ID: ID044.01 (large)
71 genes (151356 kb): ABCA1, ABCG5*, ABCG8*, ACADM*, ACADS*, ACADVL*, AGPAT2*, AKT2, AMPD1, ANGPTL3, ANGPTL4, APOA1*, APOA2*, APOA4, APOA5*, APOB*, APOC2*, APOC3*, APOE*, BANF1, BSCL2*, CACNA1S, CAV1, CAV3, CETP*, CIDEC, CPT2, CYP27A1*, DHCR24, DHCR7*,**, FBN1*,**, GCKR, GK*, GPD1, GPIHBP1*, KCNJ6, LCAT*, LDLR*,**, LDLRAP1*, LEP*,**, LIPA*, LIPC*, LIPE*, LIPG, LMF1*, LMNA*,**, LPIN1, LPL*,**, MTTP*, NPC1, NPC1L1, NPC2, PCSK9*, PCYT1A, PIK3R1*, PLIN1, POLD1*, PPARG, PSMB8, PTRF, PYGM, RYR1*, SAR1B, SCARB1, SMPD1, SORT1, SPRTN, STAP1*, USF1, WRN, ZMPSTE24*

☐ **Hypercholesterolemia**
Panel-ID: ID024.01 (small)
14 genes (31293 kb): ABCG5, ABCG8*, APOB (Exon 26)*, APOE*, CYP27A1*, DHCR24, DHCR7*,**, LDLR*,**, LDLRAP1*, LIPA*, NPC1L1*, PCSK9*, SORT1, STAP1**

Basis-Panel (FHCL):
4 genes (13521 bp): APOB (Exon 26), LDLR*,**, LDLRAP1*, PCSK9**

☐ **Hypertriglyceridemia**
Panel-ID: ID095.01 (small)
13 genes (20907 kb): APOA5, APOB (Exon 26)*, APOC2*, APOC3*, APOE*, GCKR, GK*, GPD1, GPIHBP1*, LIPC*, LMF1*, LPL*,**, USF1*

☐ **Lipodystrophy**
Panel-ID: ID055.02 (small)
20 genes (38256 kb): AGPAT2, AKT2, BANF1, BSCL2*, CAV1, CIDEC, FBN1*,**, KCNJ6, LEP*,**, LIPE*, LMNA*,**, PIK3R1*, PLIN1, POLD1*, PPARG, PSMB8, PTRF, SPRTN, WRN, ZMPSTE24*

☐ **Lipometabolic disorder due to HDL deficiency**
Panel-ID: ID096.00 (small)
14 genes (24090 kb): ABCA1, ANGPTL4, APOA1*, APOA2*, APOA4, APOC3*, CETP*, LCAT*, LIPC*, LIPG, NPC1, NPC2, SCARB1, SMPD1*

☐ **Lipometabolic disorder due to LDL deficiency**
Panel-ID: ID094.00 (small)
7 genes (25449 kb): ANGPTL3, APOB, APOE*, MTTP*, NPC1L1, PCSK9*, SAR1B*

☐ **Statin-associated myopathy**
Panel-ID: ID097.00 (small)
10 genes (35160 kb): ACADM, ACADS*, ACADVL*, AMPD1, CACNA1S, CAV3, CPT2, LPIN1, PYGM, RYR1**

Metabolic Diseases

☐ **3-Methylglutaconic aciduria (MGCA)**
Panel-ID: ID249.00 (small)
15 genes (14652 kb): ATPAF2, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MD, AUH, CLPB, DNAJC19, ECHS1, HTRA2, OPA3, SERAC1, TAZ*, TIMM50, TMEM70*

☐ **Coenzyme Q10 deficiency (COQ10D)**
Panel-ID: ID225.01 (small)
15 genes (18723 kb): ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, ETFDH, ETFA, ETFB, PDSS1, PDSS2*

☐ **Deficiency of mitochondrial complexes I to V (MCDN)**
Panel-ID: ID074.02 (small)
76 genes (56973 kb): ACAD9, ATP5F1A, ATP5F1D, ATP5F1E, ATP5MD, ATPAF2, BCS1L, COA3, COA5, COA6, COA8, COX4I1, COX6A2, COX6B1, COX8A, COX10, COX14, COX15, COX20, COX5A, 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, UQCRQ, UQCRFS1*

Mitochondrial complex I deficiency, nuclear type (MC1DN):
35 genes (27354 bp): 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 (3666 bp): SDHA,**, SDHAF1, SDHB*, SDHD**

Mitochondrial complex III deficiency, nuclear type (MC3DN):
10 genes (7131 bp): BCS1L, CYC1, LYRM7, TTC19, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, UQCRFS1*

Mitochondrial complex IV deficiency, nuclear type (MC4DN):
21 genes (14667 bp): 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 (4155 bp): ATP5F1A, ATP5F1D, ATP5F1E, ATP5MD, ATPAF2, TMEM70

☐ **Homocystinuria**
Panel-ID: ID191.01 (small)
9 genes (15309 kb): ABCD4, CBS, LMBRD1, MMACHC, MMADHC, MTHFR, MTR, MTRR, PRDX1*

☐ **Hypercalcemia**
Panel-ID: ID262.00 (small)
8 genes (14604 kb): AP2S1, CASR*, CDC73, CYP24A1*, GCM2, GNA11*, SLC34A1*, SLC12A1**

☐ **Hypoglycemia, hyperinsulinism and ketone metabolism**
Panel-ID: ID280.00 (small)
44 genes (85948 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*

Basis-Panel I (Glycogen storage disease (GSD)):
11 genes (24783 bp): AGL, G6PC, GAA,**, GBE1, LDHA, PFKM, PGAM2, PHKA2, PYGL*, PYGM, SLC37A4*

Basis-Panel II (Hyperinsulinemic hypoglycemia (HHF)):
9 genes (18762 bp): ABCC8, KCNJ11*, GCK*,**, HADH, INSR, GLUD1, SLC16A1, HNF1A*,**, HNF4A*,***

Basis-Panel III (Hypoglycemia and ketoacidosis):
6 genes (11694 bp): ACAT1, OXCT1, PC, PCCA,**, PCCB*, SLC16A1*

☐ **Obesity**
Panel-ID: ID183.01 (large)
59 genes (130365 kb): ADCY3, ADRB2, ADRB3, AGRP, ALMS1, ARL6, BBIP1, BBS1*, BBS10*, BBS12*, BBS2*, BBS4, BBS5, BBS7, BBS9*, C8ORF37, CARTPT, CELA2A, CEP19, CEP290*, CUL4B*, DYRK1B, ENPP1, GHRL, FFAR4, FTO, IFT27, IFT74, KSR2, LEP*,**, LEPR*,**, LZTFL1, MAGEL2*, MC3R*,**, MC4R*,**, MEGF8, MKKS*, MKS1*, MRAP2, MYT1L, NR0B2, NTRK2, PCSK1, PHF6*, PHIP, POMC*,**, PPARG*, RAB23, SDC3, SDCCAG8, SIM1, TRIM32, TTC8*, TUB, UCP1, UCP2, UCP3, VPS13B*,**, WDPCP**

Obesity, nonsyndromic:
17 genes (22953 bp): ADCY3, AGRP, CARTPT, FFAR4, FTO, LEP,**, LEPR*,**, MC3R*,**, MC4R*,**, MRAP2, NR0B2, PCSK1, POMC*,**, PPARG*, SIM1, UCP2, UCP3*

Obesity, syndromic:
16 genes (61422 bp): ALMS1, CELA2A, CEP19, CUL4B*, DYRK1B, KSR2, MAGEL2*, MEGF8, MYT1L, NTRK2, PHF6*, PHIP, RAB23, TUB, UCP3, VPS13B*,***

Bardet-Biedl syndrome:
21 genes (39072 bp): ARL6, BBIP1, BBS1, BBS10*, BBS2*, BBS4, BBS5, BBS7, BBS9*, BBS12*, C8ORF37, CEP290*, IFT27, IFT74, LZTFL1, MKKS*, MKS1*, SDCCAG8, TRIM32, TTC8*, WDPCP**

Developmental and Growth Disorders

☐ **Congenital disorder of glycosylation (CDG)**
Panel-ID: ID035.01 (small)
48 genes (70761 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, GALNT2, MAGT1, MGAT2, MOGS, MPDU1, MPI, NUS1, PGM1, PMM2, RFT1,*

SLC35A1, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TMEM199

Basis-Panel I (CDG, Typ I):
23 genes (25257 bp): ALG1, ALG2, ALG3, ALG6, ALG8, ALG9, ALG11, ALG12, DDOST, DHDDS, DPAGT1, DPM2, SSR4, DPM3, MPAGT1, NUS1, DPM1, MPDU1, MPI, PGM1, PMM2, RFT1, SRD5A3*

Basis-Panel II (CDG, Typ II):
16 genes (25170 bp): B4GALT1, CCDC115, COG2, COG4, COG5, COG6, COG7, COG8, MGAT2, MOGS, SLC35A1, SLC35A2, SLC35C1, SLC39A8, TMEM165, TMEM199

Gastroenterological Diseases

☐ **Glycogen storage disease (GSD)**
Panel-ID: ID108.00 (small)
23 genes (46911 kb): AGL, ALDOA, ENO3, G6PC, GAA,**, GBE1, GYG1, GYS1, GYS2, LAMP2*, LDHA, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2*, PYGL*, PYGM, SLC2A2, SLC37A4*

☐ **Intrahepatic cholestasis (PFIC)**
Panel-ID: ID159.01 (small)
6 genes (18582 kb): ABCB11, ABCB4, ATP8B1, NR1H4, SLC25A13, TJP2*

Hematologic Diseases

☐ **Hemochromatosis (HFE)**
Panel-ID: ID114.02 (small)
8 genes (9327 kb): BMP6, FTH1, FTL, HAMP*,**, HFE*,**, HJV*,**, SLC40A1*,**, TFR2*,***

Multisystemic Malformation Syndromes

☐ **Zellweger syndrome (ZWS)**
Panel-ID: ID084.00 (small)
14 genes (22179 kb): ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26

Neurodegenerative Diseases, Multisystemic Malformation Syndromes

☐ **Peroxisome biogenesis disorder (PBD)**
Panel-ID: ID083.01 (small)
14 genes (19743 kb): PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26*

Basis-Panel I (Zellweger syndrome):
12 genes (18285 bp): PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26

Basis-Panel II (Neonatal adrenoleukodystrophy / Infantile Refsum syndrome):
12 genes (17961 bp): PEX1, PEX2, PEX3, PEX5, PEX6, PEX7, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX26

Microcephaly and Macrocephaly

☐ **Hydrocephalus (HYC)**
Panel-ID: ID221.02 (small)
18 genes (53337 kb): AKT3, CCDC88C, CCND2, CFAP43, FANCB, FLVCR2, GPSM2, HYLS1, KIF7, L1CAM*,**, MPDZ, P4HB, PIK3R2, POMT1, SEC24D, TRIM71*, WDR81, ZIC3*

Congenital hydrocephalus, nonsyndromic (HYC):
5 genes (24405 bp): CCDC88C, L1CAM,**, MPDZ, TRIM71*, WDR81*

☐ **Macrocephaly, comprehensive diagnostics**
Panel-ID: (large)
117 genes (329919 kb): ABCC9, AKT1, AKT3*, AMER1, ANKH, APC2*, ASPA, ASXL2, BRAF*,**, BRWD3, CCDC22, CCDC88C, CCND2, CDH2, CDKN1C*,**, CHD1, CHD3, CHD8, CRADD, CSF1R, CUL4B*, DIS3L2, DNMT3A*, DVL1*, DVL3, EED, EML1, EXT2*,**, EZH2*, FGFR3*,**, FIBP, FOXP1*,**, GCDH, GFAP, GLI3*,**, GPC3*,**, GPC4*,**, GRIA3*, GUSB, HEPACAM, HERC1, HRAS*,**, HSD17B4, HUWE1*, IGBP1, INPPL1, KIF7, KPTN, KRAS*,**, L1CAM*,**, LAMB1, LBR, LZTR1*,**, MAN2B1, MAP2K1*, MAP2K2*, MED12*, MITF, MLC1, MPDZ*, MRAS*, MSL3, MTOR, NF1*,**, NFIB, NFIX*,**, NONO, NRAS*,**, NSD1*,**, NXN*, OFD1*, PAK1, PIGA, PIGM, PIGN, PIGT, PIK3CA*, PIK3R2, PKDCC, PPP1CB, PPP2R5D, PTCH1*,**, PTCH2*, PTEN*,**, PTPN11*, RAB39B, RAF1*, RIN2, RIT1*, RNF125, RNF135, ROR2*,**, RRAS2, SEC23B*, SETD2, SHANK3, SHOC2*, SNX10, SOS1*, SOS2*, SPOP, SPRED1*, STRADA, SUFU*, SUZ12, TBC1D7, TCIRG1, TMCO1, TNFRSF11A, TRIO, TRIP12, UPF3B*, WASHC5, WDR81, WNT5A*, ZBTB42, ZDHHC9*

Basis-Panel I (Noonan syndrome):
13 genes (according tariff EBM11355/11356): BRAF,**, KRAS*,**, LZTR1*,**, MRAS*, NRAS*,**, PPP1CB, PTPN11*, RAF1*, RIT1*, RRAS2, SHOC2*, SOS1*, SOS2**

Basis-Panel II (Sotos syndrome):
4 genes (18615 bp): APC2, EZH2*, NFIX*,**, NSD1*,***

Basis-Panel III (Cowden syndrome):
4 genes (8554 bp): AKT1, PIK3CA*, PTEN*,**, SEC23B**

Basis-Panel IV (Robinow syndrome):
5 genes (9507 bp): DVL1, DVL3, NXN*, ROR2*,**, WNT5A**

Basis-Panel V (Overgrowth syndrome):
9 genes (24864 bp): DIS3L2, DNMT3A, EED, EZH2*, GPC3*,**, GPC4*,**, NFIX*,**, NSD1*,**, OFD1**

Basis-Panel VI (Mental retardation syndrome):
5 genes (24588 bp): BRWD3, CUL4B, GRIA3*, HUWE1*, RAB39B*

Basis-Panel VII (Hydrocephalus):
6 genes (24939 bp): CCDC88C, CCND2, L1CAM,**, MPDZ*, PIK3R2, WDR81*

Basis-Panel VIII (RAS-related disorders):
18 genes (according tariff EBM11355/11356 + 25164 bp): BRAF,**, CBL, HRAS*;**, KRAS*,**, LZTR1*,**, MAP2K1*, MAP2K2*, MRAS*, NF1*,**, NRAS*,**, PPP1CB, PTPN11*, RAF1*, RIT1*, RRAS2, SHOC2*, SOS1*, SOS2**

☐ **Microcephaly, comprehensive diagnostics**
Panel-ID: ID069.01 (large)
121 genes (357615 kb): ADARB1, ANKLE2, ARCN1, ARFGEF2, ASPM,**, ATR*,**, BLM, CARS1, CASK*,**, CDK5RAP2*,**, CDK6, CENPE, CENPF, CENPJ*,**, CENPT, CEP135, CEP152*, CEP63, CIT, CKAP2L, COPB2, CRIPT, CTNNB1, CTU2, DIAPH1, DNA2, DNMT3A, DONSON, DPP6, DYNC1I2, DYRK1A, EFTUD2, EIF2S3, ERCC1, ERCC2, ERCC5, ERCC6, EXT2, FOXG1*, GEMIN4, IER3IP1, KAT6A, KATNB1, KCNA4, KIF11*, KIF14, KNL1*,*

LAGE3, MAP11, MCPH1,**, MED17, MFSD2A, MSMO1, MTHFS, MYCN*,**, 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, 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, TRIO, TRMT10A, TUBGCP2, TUBGCP4, TUBGCP6, VARS1, VPS13B*,**, WDFY3, WDR4, WDR62*, WDR73, XRCC4, ZEB2*,**, ZNF335*

Basis-Panel I (Primary microcephaly, MCPH):
5 genes (24813 bp): SPM,**, CDK5RAP2*,**, MCPH1*,**, MFSD2A, WDR62**

Basis-Panel II (Seckel syndrome, SCKL):
7 genes (25089 bp): ATR,**, CENPJ*,**, CEP152*, DNA2, NSMCE2, RBBP8, TRAIP:*

Basis-Panel III (COFS syndrome, COFS):
4 genes (11209 bp): ERCC1, ERCC2, ERCC5, ERCC6

Basis-Panel IV (Warburg Mikro syndrome, WARBM):
4 genes (8877 bp): RAB18, RAB3GAP1, RAB3GAP2, TBC1D20*

Basis-Panel V (Galloway-Mowat syndrome, GAMOS):
8 genes (11355 bp): LAGE3, NUP107, NUP133, OSGEP, TP53RK, TPRKB, WDR4, WDR73

Basis-Panel VI (Microcephaly und chorioretinopathy, MCCRP):
3 genes (10365 bp): PLK4, TUBGCP4, TUBGCP6

Basis-Panel VII (Neurodevelopmental disorder with microcephaly):
13 genes (23843 bp): ADARB1, CTNNB1, DYNC1I2, GEMIN4, MTHFS, PLAA, PRUNE1, TMX2, TRAPPC6B, SARS1, SMPD4, SVBP, VARS1

☐ **Primary microcephaly, autosomal recessive (MCPH)**
Panel-ID: ID031.02 (large)
25 genes (107130 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*

Developmental and Growth Disorders

☐ **Growth abnormalities and macrocephaly**
Panel-ID: ID072.02 (small)
31 genes (87402 kb): AKT1, APC2*, BRAF*,**, BRWD3, CUL4B*, DNMT3A*, EED, EZH2*, GPC3*,**, GPC4*,**, HRAS*,**, HUWE1*, KIF7, KRAS*,**, NF1*,**, NFIX*,**, NRAS*,**, NSD1*,**, OFD1*, PIK3CA*, PPP1CB, PTEN*,**, PTPN11V, RAB39B, RAF1*, RIT1*, RNF125, RRAS, SHOC2*, SOS1*, SPRED1*,**

Overgrowth syndrome and macrocephaly:
10 genes (30012 bp): APC2, DNMT3A, EED, EZH2*, GPC3*,**, GPC4*,**, NFIX*,**, NSD1*,**, OFD1*, RNF125*

Noonan syndrome and macrocephaly:
10 genes (15219 bp): BRAF,**, KRAS*,**, NRAS*,**, PPP1CB, PTPN11*,**, RAF1*, RIT1*, RRAS, SHOC2*, SOS1**

Multisystemic Malformation Syndromes

☐ **Malformation syndromes with predominantly facial involvement**
Panel-ID: ID279.00 (small)
25 genes (90930 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 (17001 bp): FGFR1,**, FGFR2*,**, FGFR3*,**, MEGF8, RAB23, TWIST1*,**

Orofaciodigital syndrome (OFD):
9 genes (29673 bp): C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1*, TCTN3, TMEM107*

Fraser syndrome (FRASRS):
3 genes (24780 bp): FRAS1, FREM2, GRIP1

Pierre-Robin syndrome:
4 genes (9723 bp): COL11A2, MYMK, RBM10, TGDS*

☐ **Noonan syndrome (NS)**
Panel-ID: ID023.05 (small)
14 genes (23409 kb): BRAF,**, KRAS*,**, LZTR1*,**, MAPK1, MRAS*, NRAS*,**, PPP1CB, PTPN11*, RAF1*, RRAS2, RIT1*, SHOC2*, SOS1*, SOS2**

Eye Diseases, Skeletal and Bone Diseases

☐ **Weill-Marchesani syndrome (WMS)**
Panel-ID: ID230.00 (small)
4 genes (20670 kb): ADAMTS10, ADAMTS17, FBN1,**, LTBP2*

Microcephaly and Macrocephaly, Skeletal and Bone Diseases

☐ **Rubinstein-Taybi syndrome (RSTS)**
Panel-ID: ID142.01 (small)
3 genes (24258 kb): CREBBP,**, EP300*,**, SRCAP**

Skeletal and Bone Diseases

☐ **VACTERL association**
Panel-ID: ID258.01 (small)
25 genes (69697 kb): BRCA2,**, CHD7*,**, FANCA, FANCB, FANCC*, FANCD2*, FANCE, FANCF, FANCG, FANCI*, FANCL, FGF8, FOXF1*, GLI3*, HAAO, HOXD13, HSPA6, KYNU*, MNX1, NADSYN1, RECQL4, SALL1, MYCN*,**, TRAP1, ZIC3*

Basis-Panel I:
9 genes (23910 bp): CHD7,**, FANCB, FANCI*, FGF8, FOXF1*, HOXD13, HSPA6, TRAP1, ZIC3*

Basis-Panel II (Fanconi anemia):
9 genes (22816 bp): FANCA, FANCB, FANCC, FANCD2*, FANCE, FANCF, FANCG, FANCI*, FANCL*

Skeletal and Bone Diseases, Skin and Dental Diseases

☐ **Orofaciodigital syndrome (OFD)**
Panel-ID: ID265.00 (small)
9 genes (29673 kb): C2CD3, CPLANE1, DDX59, IFT57, INTU, KIAA0753, OFD1*, TCTN3, TMEM107*

Muscular Diseases

☐ **Centronuclear myopathy (CNM)**
Panel-ID: ID257.00 (small)
7 genes (21348 kb): CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1*,**, SPEG*

☐ **Congenital myasthenic syndrome (CMS)**
Panel-ID: ID130.00 (small)
25 genes (55884 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*

Basis-Panel I (CMS, postsynaptic):
13 genes (24192 bp): CHRNA1, CHRNB1*, CHRND*, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, MUSK, PREPL, RAPSN*, SCN4A*, SNAP25*

Basis-Panel II (CMS, presynaptic):
8 genes (21906 bp): AGRN, CHAT, MYO9A, SLC5A7, SLC18A3, SLC25A1, SYT2, VAMP1

Basis-Panel III (CMS with tubular aggregates):
4 genes (5163 bp): ALG2, ALG14, DPGTA1, GFPT1

Basis-Panel IV (CMS associated with AChR deficiency):
5 genes (8376 bp): CHRNB1, CHRND*, CHRNE, MUSK, RAPSN*,*

☐ **Congenital structural myopathy**
Panel-ID: ID212.01 (small)
20 genes (78774 kb): ACTA1, BIN1, CCDC78, CFL2, DNM2, KBTBD13, KLHL40, KLHL41, LMOD2, MAP3K20, MTM1*,**, MTMR14, MYPN*, NEB, RYR1*, SELENON*, SPEG, TNNT1, TPM2, TPM3*

Basis-Panel I (Core myopathy):
4 genes (18870 bp): ACTA1, RYR1, SELENON*, TPM3*

Basis-Panel II (Nemaline myopathy):
11 genes (24865 bp): ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, TNNT1, TPM2, TPM3, MYPN, NEB (spec. exons)*

Basis-Panel III (Centronuclear myopathy):
7 genes (20292 bp): CCDC78, DNM2, BIN1, MAP3K20, MTMR14, MTM1*,**, SPEG*

☐ **Emery-Dreifuss muscular dystrophy (EDMD)**
Panel-ID: ID121.00 (small)
6 genes (51762 kb): EMD, FHL1*, LMNA*,**, SYNE1, SYNE2, TMEM43**

☐ **Limb-girdle muscular dystrophy (LGMD, MDDGC)**
Panel-ID: ID122.02 (large)
34 genes (187917 kb): ANO5, BVES, CAPN3, COL6A1, COL6A2, COL6A3, DAG1, DNAJB6, DPM3, DYSF, FKRP**, FKTN*, GMPPB, HNRNPDL, ISPD*, LAMA2, LIMS2, PLEC, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SGCA**, SGCB*,**, SGCD**, SGCG*,**, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN*

Basis-Panel I (LGMD, rezessive):
*13 genes (25113 bp): ANO5, CAPN3, DYSF, FKRP**, FKTN*, POMT1, POMT2, SGCA**, SGCB*,**, SGCD**, SGCG*,**, TCAP, TRIM32*

Basis-Panel II (LGMD, dominant):
7 genes (23142 bp): CAPN3, COL6A1, COL6A2, COL6A3, DNAJB6, HNRNPDL, TNPO3*

Basis-Panel III (LGMD and dystroglycanopathy):
11 genes (18528 bp): DAG1, DPM3, FKRP, FKTN*, GMPPB, ISPD*, POMGNT1, POMGNT2, POMK, POMT1, POMT2*

☐ **Myofibrillar myopathy**
Panel-ID: ID161.00 (large)
9 genes (119286 kb): BAG3, CRYAB, DES, FLNC, KY, LDB3, MYOT, PYROXD1, TTN*

☐ **Myotonia**
Panel-ID: ID255.00 (small)
5 genes (25041 kb): ATP2A1, CLCN1,**, HINT1*, HSPG2, SCN4A**

☐ **Nemaline myopathy (NEM)**
Panel-ID: ID199.00 (small)
11 genes (40554 kb): ACTA1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, TNNT1, TPM2, TPM3, MYPN, NEB

☐ **Periodic paralysis**
Panel-ID: ID253.00 (small)
7 genes (16572 kb): CACNA1S, KCNE3, KCNJ5, KCNJ2*,**, KCNJ12, KCNJ18*, SCN4A**

Brain Malformations, Eye Diseases

☐ **Walker-Warburg syndrome (WWS, MDDGA)**
Panel-ID: ID178.00 (small)
14 genes (23682 kb): B3GALNT2, B4GAT1, DAG1, FKRP*,**, FKTN*, GMPPB, ISPD*, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1*

Metabolic Diseases

☐ **Muscular dystrophy-dystroglycanopathy (MDDG)**
Panel-ID: ID179.00 (small)
15 genes (23961 kb): B3GALNT2, B4GAT1, DAG1, DPM3, FKRP*,**, FKTN*, GMPPB, ISPD*, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1*

Basis-Panel I (MDDGA with brain and eye anomalies):
14 genes (22809 bp): B3GALNT2, B4GAT1, DAG1, FKRP*,**, FKTN*, GMPPB, ISPD*, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1*

Basis-Panel II (MDDGB with mental retardation):
7 genes (12687 bp): FKRP,**, FKTN*, GMPPB, LARGE1, POMGNT1, POMT1, POMT2*

Basis-Panel III (MDDGC, limb-girdle muscular dystrophy):
11 genes (18528 bp): DAG1, DPM3, FKRP,**, FKTN*, GMPPB, ISPD*, POMGNT1, POMGNT2, POMK, POMT1, POMT2*

Neurodegenerative Diseases

☐ **Alzheimer disease (AD)**
Panel-ID: ID157.01 (small)
7 genes (16803 kb): ABCA7, ADAM10, APOE, APP, PSEN1*, PSEN2, TF*

☐ **Amyotrophic lateral sclerosis (ALS)**
Panel-ID: ID209.01 (small)
28 genes (64812 kb): ALS2, ANG, ANXA11, CHCHD10, CHMP2B, DCTN1, ERBB4, FIG4, FUS, HNRNPA1, KIF5A, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, SETX, SIGMAR1, SOD1, SPG11, SQSTM1, TARDBP, TBK1, TUBA4A, UBQLN2, VAPB, VCP*

Basis-Panel I:
16 genes (24492 bp): ANG, ANXA11, CHMP2B, FIG4, ERBB4, FUS, HNRNPA1, MATR3, OPTN, PFN1, SOD1, TARDBP, TUB4A4, UBQLN2, VAPB, VCP*

Basis-Panel II (ALS, juvenile):
4 genes (21003 bp): ALS2, SETX, SIGMAR1, SPG11

Basis-Panel III (ALS with frontotemporal dementia):
8 genes (12342 bp): CHCHD10, FUS, SQSTM1, TARDBP, TBK1, TUB4A4, UBQLN2, VCP

☐ **Brain atrophy and demyelinating brain diseases**
Panel-ID: ID278.00 (small)
55 genes (95975 kb): AIMP1, AIMP2, ASPA*,**, B3GALNT2*, B4GAT1, COL4A1, CNP, CRPPA*, DAG1, DARS1, DEGS1, EPRS1, EXOC7, EXOC8, FAM126A, FARSA, FARSB, FKRP*,**, FKTN*, GFAP, GJC2*, GMPPB, GRM7, HIKESHI, HSPD1, LARGE1, MAPT, MAT1A, MED17, MTHFS, PLP1*,**, POLR1C*, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PSEN1, PYCR2, RARS1*, RXYLT1, SLC25A12, SOX10*, TBCD*, TMEM106B, TMEM63A, TRAPPC12, TRAPPC4, TRAPPC6B, TUBB4A, UBTF, UFM1*, VPS11**

Brain atrophy:
13 genes (24252 bp): EXOC7, EXOC8, FARSA, FARSB, GRM7, MAPT, MED17, PSEN1, TBCD, TRAPPC4, TRAPPC6B, TRAPPC12, UBTF*

Hypomyelinating and demyelinating diseases:
27 genes (42983 bp): 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 (23712 bp): B3GALNT2, B4GAT1, CRPPA*, DAG1, FKRP*,**, FKTN, GMPPB, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, RXYLT1*

☐ **Cerebellar ataxia, autosomal dominant (SCA)**
Panel-ID: ID236.02 (small)
30 genes (96369 kb): AFG3L2, ATP1A3, BEAN1, 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*

☐ **Cerebellar ataxia, autosomal recessive (SCAR)**
Panel-ID: ID213.02 (large)
49 genes (163773 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, TPP1*,**, TTPA, TWNK, UBA5, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1*

Basis-Panel I: (Spinocerebellar ataxia, autosomal rezessive (SCAR)):
6 genes (25210 bp): ANO10, COQ8A, GRID2, PMPCA, STUB1, SYNE1 (spec. exons)

Basis-Panel II (Spinocerebellar ataxia with axonal neuropathy (SCAN)):
3 genes (10551 bp): COA7, SETX, TDP1

Basis-Panel III (Spinocerebellar ataxia with mental retardation (CAMRQ)):
4 genes (12876 bp): ATP8A2, CA8, VLDLR, WDR81

Basis-Panel IV (Spinocerebellar ataxia with oculomotor apraxia (AOA)):
4 genes (13263 bp): APTX, PIK3R5, PNKP, SETX*

☐ **Cerebellar ataxia, X-linked**
Panel-ID: ID273.00 (small)
7 genes (15909 kb): ABCB7, AIFM1, ATP2B3, CASK,**, OPHN1, PRPS1, SLC9A6*

☐ **Choreatiform movement disorders**
Panel-ID: ID272.01 (small)
17 genes (40641 kb): ADCY5, ATP1A2,** ATP1A3, FRRS1L, GNAO1, HPRT1, KCNMA1, NKX2-1, PDE2A, PDE10A, PNKD, PRNP*, PRRT2*, RNF216, SLC2A1*,**, VPS13A, XK*

☐ **Dystonia (DYT)**
Panel-ID: ID128.01 (small)
22 genes (47085 kb): ANO3, ACTB, ATP1A3, COL6A3, ECHS1, GCH1*, GNAL*, HPCA, KCTD17, KMT2B, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1*,**, SPR, TAF1, TH*, THAP1*, TOR1A, TUBB4A*

Basis-Panel I (primary DYT):
8 genes (25539 bp): ANO3, COL6A3, GNAL*, HPCA, KMT2B, THAP1*, TOR1A, TUBB4A*

Basis-Panel II (secondary/combined DYT):
14 genes (20655 bp): ACTB, ATP1A3, ECHS1, GCH1, KCTD17, MECR, PNKD, PRKRA, PRRT2, SGCE, SLC2A1*,**, SPR, TAF1, TH**

☐ **Episodic ataxia (EA) (small)**
Panel-ID: ID184.02
6 genes (20526 kb): ATP1A3, CACNA1A,**, CACNB4*, KCNA1*,**, SCN2A*, SLC1A3**

☐ **Episodic pain syndrome (FEPS)**
Panel-ID: ID268.00 (small)
4 genes (20529 kb): SCN9A, SCN10A, SCN11A, TRPA1*

☐ **Hereditary ataxia, comprehensive diagnostics**
Panel-ID: ID276.00 (large)
114 genes (335247 kb): ABCB7, ABHD12, ACO2, AFG3L2, AIFM1, ANO10, APTX, ATCAY, ATG5, ATM*,**, ATP1A3, ATP2B3, ATP8A2, BEAN1, 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, NKX6-2, OPHN1, PCDH12, PCNA, PDYN, PEX7, PHYH, PIK3R5, PLD3, PMPCA, PNKP*,*

PNPLA6, POLG, POLR3A, POLR3B, PRKCG, PRPS1, PTF1A, PUM1, RFC1, RNF216, RUBCN, SACS, SCN2A, SCYL1, SETX, SIL1, SLC1A3*, SLC25A46, SLC52A2, SLC9A1, SLC9A6, SNX14, SPTBN2, SQSTM1, STUB1, SYNE1, SYT14, TDP1, TDP2, TGM6, TMEM240, TPP1*,**, TRPC3, TSFM, TTBK2, TTPA, TUBB4A, TWNK, TXN2, UBA5, VAMP1, VLDLR, VPS13D, VWA3B, WDR73, WDR81, WFS1, WWOX, XRCC1*

Basis-Panel I (Episodic ataxia (EA)):
6 genes (20526 bp): ATP1A3, CACNA1A,**, CACNB4*, KCNA1*,**, SCN2A*, SLC1A3**

Basis-Panel II (Spastic ataxia (SPAX)):
6 genes (24729 bp): AFG3L2, CHP, KIF1C, MARS2, MTPAP, NKX6-2, SACS, VAMP1*

Basis-Panel III (Cerebellar ataxia, X-linked (SCAX)):
7 genes (15909 bp): ABCB7, AIFM1, ATP2B3, CASK,**, OPHN1, PRPS1, SLC9A6*

Basis-Panel IV (Spinocerebellar ataxia, autosomal dominant (SCA)):
8 genes (25347 bp): AFG3L2, FGF14, ITPR1, KCND3*, PRKCG, PDYN, SPTBN2, TGM6*

Basis-Panel V (Spinocerebellar ataxia, autosomal rezessive (SCAR)):
6 genes (25210 bp): ANO10, COQ8A, GRID2, PMPCA, STUB1, SYNE1 (spez. Exons)

Basis-Panel VI (Cerebellar ataxia with axonal neuropathy (SCAN)):
3 genes (10551 bp): COA7, SETX, TDP1

Basis-Panel VII (Cerebellar ataxia with mental retardation (CAMRQ)):
4 genes (12876 bp): ATP8A2, CA8, VLDLR, WDR81

Basis-Panel VIII (Cerebellar ataxia with oculomotor apraxia (AOA)):
4 genes (13263 bp): APTX, PIK3R5, PNKP, SETX*

Basis-Panel IX (Ataxia teleangiectatica (AT)):
3 genes (12078 bp): ATM,**, MRE11, PCNA*

☐ Hereditary essential tremor (ETM)
Panel-ID: ID195.01 (small)
4 genes (16596 kb): DRD3, FUS, SCN4A,**, TENM4*

☐ Hyperekplexia (HKPX)
Panel-ID: ID216.00 (small)
9 genes (15993 kb): ACTL6B, ATAD1, ARHGEF9, ASNS, GLRA1*, GLRB, GPHN, SLC6A5, TRAK1*

☐ Leukodystrophy and leukoencephalopathy, comprehensive diagnostics
Panel-ID: ID204.03 (large)
117 genes (203255 kb): AARS2, ABCD1,**, ACBD5, ACER3, ACOX1, ADAR*, AIFM1, AIMP1*, AIMP2, ALDH3A2, ARSA, ASPA*,**, AUH, BOLA3, CLCN2*, 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, 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, PSAP*, PYCR2, RARS1*, RNASEH2A*,**, RNASEH2B*,**, RNASEH2C*,**, RNASET2, SAMHD1*,**, SCP2*, SDHAF1, SLC13A3, SLC16A2, SLC17A5, SOX10*, SPTAN1*, SUMF1*,TMEM106B, TMEM63A, TREM2, TREX1*,**, TUBB4A, TYMP, TYROBP, UFM1*, VPS11*, ZFYVE26**

Basis-Panel I (Leukodystrophy, adult-onset):
14 genes (25002 bp): AARS2, ABCD1,**, ARSA, CSF1R, CYP27A1*, DARS2, EIF2B5, GALC*,**, GFAP, GJC2*, HEPACAM, LMNB1**, MLC1, PLP1*,**

Basis-Panel II (Leukodystrophy, infantile):
15 genes (25161 bp): AIMP1, ABCD1*,**, ARSA, ASPA, DARS2, GALC*,**, GFAP, GJC2*, HEPACAM, MLC1, PLP1*,**, POLR3A, POLR3B, RNASET2, TUBB4A*

Basis-Panel III (Leukodystrophy, hypomyelinating (HLD)):
14 genes (25245 bp): AIMP1, DEGS1, EPRS1, FAM126A, GJC2*, HIKESHI, HSPD1, PLP1*,**, POLR1C*, POLR3A, POLR3B, RARS1*, TMEM106B, TUBB4A*

Basis-Panel IV (Vanishing white matter leukodystrophy (VWM)):
5 genes (7068 bp): EIF2B1, EIF2B2, EIF2B3*, EIF2B4, EIF2B5*

Basis-Panel V (Neonatal adrenoleukodystrophy (NALD)):
11 genes (16017 bp): PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX26*

Basis-Panel VI (Aicardi-Goutières syndrome (AGS)):
7 genes (10188 bp): ADAR, IFIH1*, RNASE2A*,**, RNASE2B*,**, RNASE2C*,**, SAMHD1*,**, TREX1*,**

Basis-Panel VII (CADASIL, CARASIL):
2 genes (8403 bp): HTRA1, NOTCH3*,**

☐ Leukodystrophy, hypomyelinating (HLD)
Panel-ID: ID277.00 (small)
20 genes (33998 kb): AIMP1, AIMP2, CNP, DEGS1, EPRS1, FAM126A, GJC2*, HIKESHI, HSPD1, PLP1*,**, POLR1C*, POLR3A, POLR3B, PYCR2, RARS1*, TMEM63A, TMEM106B, TUBB4A, UFM1*, VPS11**

☐ Neurodegeneration with brain iron accumulation (NBIA)
Panel-ID: ID264.00 (small)
11 genes (20034 kb): ATP13A2, C19ORF12, CP, COASY, CRAT, FA2H, FTL, PANK2*,**, PLA2G6, REPS1, WDR45*

☐ Parkinson disease (PARK)
Panel-ID: ID077.01 (small)
*21 genes (57537 kb): ADH1C, ATP13A2**, CHCHD2, DNAJC6, EIF4G1, FBXO7, GBA*,**, GIGYF2, GLUD2, HTRA2, LRRK2*,**, MAPT, PARK7**, PINK1**, PLA2G6, PRKN*,**, SNCA*,**, SYNJ1, UCHL1**, VPS13C, VPS35*

Basis-Panel I (PARK, early-onset):
*8 genes (25057 bp): ATP13A2**, DNAJC6, FBXO7, PARK7**, PINK1**, PLA2G6, PRKN*,** VPS13C*

Basis-Panel II (Park, late-onset):
10 genes (24057 bp): CHCHD2, EIF4G1, GBA,**, GIGYF2, HTRA2, LRRK2*,**, MAPT, SNCA*,**, UCHL1**, VPS35*

☐ Paroxysmal dyskinesia
Panel-ID: ID286.00 (small)
14 genes (38934 kb): ADCY5, ATP1A2,**, ATP1A3, CACNA1A*,**, GCH1*,**, KCNA1*,**, KCNMA1, PDE2A, PDE10A, PNKD, PRRT2*, SCN8A, SLC2A1*,**, TBC1D24**

☐ Spastic ataxia (SPAX)
Panel-ID: ID228.00 (small)
12 genes (34743 kb): AFG3L2, CAPN1, CHP1, GJC2*, KIF1C, MARS2, MTPAP, NKX6-2, POLR3A, SACS, SPG7, VAMP1*

☐ Spastic paraplegia (SPG, HSP)
Panel-ID: ID148.02 (large)
55 genes (114447 kb): ALDH18A1, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ATL1, ATP13A2, B4GALNT1, BSCL2, C12ORF65, C19ORF12*, CAPN1,*

CPT1C, CYP2U1, CYP7B1, DDHD1, DDHD2, DSTYK, ENTPD1, ERLIN1, ERLIN2, FA2H, FARS2, GBA2, GJC2*, HSPD1*, IBA57, KIF1A, KIF5A, L1CAM*,**, MAG, NIPA1, NT5C2, PLP1*,**, PNPLA6*, REEP1, REEP2, RTN2, SLC33A1, SPART, SPAST*, SPG7, SPG11, SPG21, TECPR2, TFG, UBAP1, UCHL1, VPS37A, WASHC5, ZFYVE26*, ZFYVE27*

Basis-Panel I (SPG, dominant):
14 genes (25395 bp): ALDH18A1, ATL1, BSCL2, CPT1C, HSPD1*, KIF5A, NIPA1, REEP1, RTN2, SLC33A1, SPAST*, UBAP1, WASHC5, ZFYVE27*

Basis-Panel II (SPG, rezessive):
10 genes (25134 bp): B4GALNT1, C12ORF65, C19ORF12, CYP2U1, CYP7B1, ERLIN2, FA2H, SPG7, SPG11, ZFYVE26*

Basis-Panel III (SPG, X-linked):
2 genes (4602 bp): L1CAM,**, PLP1*,***

Metabolic Diseases

☐ **Neuronal ceroid lipofuscinosis (CLN)**
Panel-ID: ID132.00 (small)
13 genes (17985 kb): ATP13A2, CLN3,**, CLN5*, CLN6**, CLN8**, CTSD*, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1*,**, TPP1*,***

Muscular Diseases

☐ **Charcot-Marie-Tooth disease, axonal (CMT, HMSN)**
Panel-ID: ID052.01 (small)
39 genes (96195 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, PLEKHG5, PRPS1, RAB7A, SPG11, TRIM2, TRPV4*, VCP, YARS1*

Basis-Panel I (CMT2, axonal dominant):
14 genes (24297 bp): AARS1, DNM2, GARS1, GDAP1*, HARS1, HSPB1, HSPB8, LRSAM1, MFN2*,**, MORC2, MPZ*,**, NEFL*, RAB7A, TRPV4*

Basis-Panel II (CMT2, axonal rezessive):
10 genes (25146 bp): GDAP1, IGHMBP2*,**, LMNA*;**, LRSAM1, MED25, MFN2*,**, MME, MPV17, SPG11, TRIM2*

Basis-Panel III (CMTDI / CMTRI, intermediate):
11 genes (18477 bp): COX6A1, DNM2, GDAP1*, GJB1*,**, GNB4, INF2*, KARS1, MPZ*,**, NEFL*, PLEKHG5, YARS1*

☐ **Charcot-Marie-Tooth disease, demyelinating (CMT, HMSN)**
Panel-ID: ID051.01 (small)
25 genes (52530 kb): 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*

Basis-Panel I (CMT1 / CMT4, demyelininating):
15 genes (24500 bp): EGR2, FGD4, FIG4, GDAP1*, HK1, LITAF*, MPZ*,**, MTMR2, NDRG1, NEFL*, PMP2, PMP22*,**, PRX, SH3TC2*, SURF1*

Basis-Panel II (CMT3, Déjerine-Sottas neuropathy):
4 genes (7035 bp): EGR2, MPZ*,**, PMP22*,**, PRX*

Basis-Panel III (CMTDI / CMTRI, intermediate):
11 genes (18477 bp): COX6A1, DNM2, GDAP1, GJB1*,**, GNB4, INF2*, KARS1, MPZ*,**, NEFL*, PLEKHG5, YARS1*

☐ **Distal hereditary motor neuronopathy (HMN, DSMA)**
Panel-ID: ID254.00 (small)
17 genes (38685 kb): ATP7A´**, BSCL2*, DCTN1, DNAJB2, FBXO38, GARS1*, HSPB1, HSPB3, HSPB8, IGHMBP2*,**, PLEKHG5, REEP1, SETX, SIGMAR1, SLC5A7, TRPV4, WARS1*

Basis-Panel I (HMN, dominant):
11 genes (18609 bp): BSCL2, DCTN1, FBXO38, GARS1*, HSPB1, HSPB3, HSPB8, REEP1, SLC5A7, TRPV4, WARS1*

Basis-Panel II (DSMA, rezessive):
5 genes (11997 bp): ATP7A,**, DNAJB2, IGHMBP2*,**, PLEGHG5, SIGMAR1*

☐ **Hereditary sensory and autonomic neuropathy (HSAN, HSN)**
Panel-ID: ID086.01 (small)
15 genes (52455 kb): ATL1, ATL3, DNMT1, DST, ELP1, KIF1A, NGF, NTRK1, PRDM12, RETREG1, SCN9A*, SCN11A, SPTLC1*, SPTLC2, WNK1*

☐ **Spinal muscular atrophy (SMA)**
Panel-ID: ID152.01 (small)
31 genes (69462 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*

Basis-Panel I (SMA, proximal):
7 genes (18843 bp + according to tariff EBM 11410/11411): ASAH1, BICD2, CHCHD10, DYNC1H1, SMN1,**, SMN2*,**, VAPB*

Basis-Panel II (SMA, distal):
15 genes (24915 bp): BSCL2, DCTN1, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, IGHMBP2*,**, SIGMAR1, PLEKHG5, REEP1, SLC5A7, TRPV4*, SMN1*,**, SMN2*,**, VAPB*

Basis-Panel III (SMA, infantile):
12 genes (24705 bp + according to tariff EBM 11410/11411): ASAH1, ASCC1, ATP7A,**, BICD2, IGHMBP2*,**, PLEKHG5, SIGMAR1, SMN1*,**, SMN2*,**, TRIP4, TRPV4*, VRK1*

Basis-Panel IV (SMA, adult):
15 genes (22404 bp + according to tariff EBM 11410/11411): BSCL2, CHCHD10, DNAJB2, FBXO38, GARS1, HSPB1, HSPB3, HSPB8, REEP1, SETX, SLC5A7, SMN1*,**, SMN2*,**, VAPB, WARS1*

Pulmonary and Respiratory Diseases

☐ **Congenital central hypoventilation syndrome (CCHS)**
Panel-ID: ID185.00 (small)
7 genes (8538 kb): ASCL1, BDNF, EDN3, GDNF, MECP2,**, PHOX2B*,**, RET*,***

Metabolic Diseases

☐ **Pulmonary surfactant metabolism dysfunction (SMDP)**
Panel-ID: ID168.01 (small)
6 genes (11958 kb): ABCA3, CSF2RA, CSF2RB, NKX2-1, SFTPB, SFTPC*

Reproductive Disorders

☐ **Female infertility (PREMBL, OOMD)**
Panel-ID: ID239.01 (small)
12 genes (17959 kb): BTG4, PADI6, PANX1, PATL2, REC114, TLE6, TRIP13, TUBB8, WEE2, ZP1, ZP2, ZP3

☐ **Spermatogenic failure (SPGF)**
Panel-ID: ID192.01 (large)
44 genes (168733 kb): AK7, ARMC2, AURKC, BRDT, CATSPER1, CFAP43, CFAP44, CFAP65, CFAP69, CFAP70, DNAH1, DNAH17, DPY19L2, FANCM, FSIP2, KLHL10, MEIOB, NANOS1, NR5A1*,**, PLCZ1, PMFBP1, PPP2R3C, QRICH2, SEPTIN12, SLC26A8, SOHLH1, SPATA16, SPEF2, SPINK2, SUN5, SYCE1, SYCP2, SYCP3, TAF4B, TEX11*, TEX14, TEX15, TDRD9, TSGA10, TTC21A, TTC29, USP9Y, WDR66, ZMYND15*

Basis-Panel I (SPGF, autosomal rezessive):
8 genes (25065 bp): AURKC, CFAP43, DNAH1, DPY19L2, PLCZ1, SPINK2, SUN5, SYCE1*

Basis-Panel II (SPGF, autosomal dominant):
8 genes (11298 bp): KLHL10, NANOS1, NR5A1,**, PPP2R3C, SEPTIN12, SLC26A8, SOHLH1, SYCP3*

Basis-Panel III (SPGF, X- linked or Y-linked):
2 genes (10485 bp): TEX11, USP9Y*

Endocrine Disorders

☐ **Congenital adrenal hyperplasia (CAH)**
Panel-ID: ID111.01 (small)
10 genes (17961 kb): ARMC5, CYP11B1, CYP17A1*, CYP21A2*,**, HSD3B2*, PDE8B, PDE11A, POR, PRKAR1A, STAR*

Adrenogenital syndrome (AGS):
6 genes: (8547 bp): CYP11B1, CYP17A1*, CYP21A2*,**, HSD3B2*, POR, STAR*

☐ **Disorder of sex development (DSD)**
Panel-ID: ID117.01 (small)
37 genes (60108 kb): AKR1C2, AKR1C4, AMH, AMHR2*, AR*,**, BMP15*, CBX2, CYP11A1, CYP11B1, CYP17A1*,**, CYP19A1, CYP21A2*,**, DHH*, ESR2, FSHR*, HSD17B3, HSD17B4, HSD3B2*, LHCGR*, NUP107, MAP3K1, MCM9, MRPS22, NR0B1*,**, NR5A1*,**, POR, PSMC3IP, RSPO1, SOHLH1, SOX3*, SOX9*,**, SRD5A2, SRY*,**, STAR, WNT4*, WT1*,**, ZFPM2*

Basis-Panel I (46,XY-DSD):
14 genes (24789 bp): AKR1C2, AR,**, CBX2, CYP11A1, DHH*, HSD17B3, HSD3B2*, LHCGR, MAP3K1, NR0B1*,**, NR5A1*,**, SRD5A2, SRY*,**, STAR, ZFPM2*

Basis-Panel II (46,XX-DSD):
16 genes (23175 bp): BMP15, CYP19A1, ESR2, FSHR, NR5A1*,**, MCM9, MRPS22, HSD17B4, NUP107, PSMC3IP, RSPO1, SOHLH1, SOX3*, SOX9*,**, SRY*,**, WNT4*

Basis-Panel III (Adrenal hyperplasia):
6 genes (8529 bp): CYP11B1, CYP17A1*, CYP21A2*,**, HSD3B2*, POR, STAR*

☐ **Premature ovarian failure (POF)**
Panel-ID: ID078.03 (small)
17 genes (42186 kb): BMP15, BNC1, DIAPH2, ERCC6, GDF9, FANCM, FIGLA, FMR1**, FOXL2*,**, HFM1*, MCM8*, MSH5, NOBOX*, NR5A1*,**, POF1B*, STAG3, SYCE1*

Basis-Panel I (X-linked):
4 genes (8142 bp): BMP15, DIAPH2, FMR1**, POF1B*

Basis-Panel II (autosomal rezessive):
7 genes (21341 bp): GDF9, FANCM, HFM1, MCM8*, MSH5, STAG3, SYCE1*

Basis-Panel III (autosomal dominant):
6 genes (12703 bp): BNC1, ERCC6, FIGLA, FOXL2,**, NOBOX*, NR5A1*,***

Ear, Nose and Throat Diseases

☐ **Hypogonadotropic hypogonadism with or without anosmia (KAL, HH)**
Panel-ID: ID170.02 (small)
26 genes (41154 kb): ANOS1,**, CHD7*,**, DUSP6, FEZF1, FGFR1*,**, FGF8, FGF17, FLRT3, FSHB, GNRH1*, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R*, LHB, NDNF, NSMF, PROK2, PROKR2, SEMA3A, SPRY4, SOX10*, TAC3*, TACR3, WDR11*

Basis-Panel I (dominant and X-linked):
11 genes (25221 bp): ANOS1,**, CHD7*,**, FGFR1*,**, FGF8, FGF17, HS6ST1, NSMF, PROK2, PROKR2, SEMA3A, WDR11*

Basis-Panel II (rezessive and X-linked):
11 genes (11145 bp): ANOS1,**; FEZF1, FSHB, GNRH1*, GNRHR, IL17RD, KISS1, KISS1R*, LHB, TAC3*, TACR3*

Skeletal and Bone Diseases

☐ **Brachydactyly (BD)**
Panel-ID: ID218.01 (small)
16 genes (26319 kb): BMP2, BMPR1B, CHST11, CHSY1, GDF5, HOXD13, IHH, NOG, PITX1, PRMT7, PDE3A, PTHLH, ROR2,**, RUNX2*,**, TBC1D24*, TRPV4**

Brachydactyly, nonsyndromic:
8 genes (10500 bp): BMP2, BMPR1B, GDF5, HOXD13, IHH, NOG, PTHLH, ROR2,**

Brachydactyly, syndromic:
11 genes (19000 bp): CHST11, CHSY1, GDF5, HOXD13, NOG, PDE3A, PITX1, PRMT7, RUNX2,**, TBC1D24*, TRPV4**

☐ **Craniosynostosis (CRS)**
Panel-ID: ID224.01 (small)
33 genes (75771 kb): ALX4, ASXL1, CD96. COLEC10, COLEC11, CYP26B1, EFNB1, ERF, 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*

Basis-Panel I:
14 genes (25251 bp): ALX4, EFNB1, ERF, FGFR1*,**, FGFR2*,**, FGFR3*,**, GLI3*,**, MSX2, RAB23, SKI*, SMAD6*, TCF12*, TWIST1*,**, ZIC1*

Basis-Panel II (Acrocephalosyndactyly (ACS)):
6 genes (17001 bp): FGFR1,**, FGFR2*,**, FGFR3*,**, MEGF8, RAB23, TWIST1*,**

Basis-Panel III (Cranioectodermal dysplasia (CED)):
4 genes (12084 bp): IFT43, IFT122, WDR19, WDR35

Basis-Panel IV (Trigonocephaly (TRIGNO)):
5 genes (16959 bp): ASXL1, CD96, FGFR1,**, FREM1, PPP3CA*

☐ **Klippel-Feil syndrome (KFS)**
Panel-ID: ID207.00 (small)
5 genes (12522 kb): GDF3, GDF6, MEOX1*, MYO18B, PAX1**

☐ **Mandibulofacial dysostosis (MFD)**
Panel-ID: ID188.01 (small)
11 genes (22047 kb): DHODH, EDNRA, EFTUD2, POLR1A, POLR1B, POLR1C, POLR1D*, RPS28, SF3B4*, TCOF1*,**, TSR2*

Basis-Panel (Treacher-Collins syndrome):
4 genes (9432 bp): POLR1B, POLR1C, POLR1D*, TCOF1*,***

☐ **Multiple epiphyseal dysplasia (EDM)**
Panel-ID: ID202.01 (small)
10 genes (25899 kb): CANT1, COL2A1,**, COL9A1, COL9A2, COL9A3*, COMP, EIF2AK3, KIF7, MATN3, SLC26A2**

☐ **Osteoporosis**
Panel-ID: ID115.01 (small)
13 genes (29370 kb): CALCR, COL1A1,**, COL1A2*,**, ESR1, LGR4, LRP5*, PLS3, SGMS2, SLC34A*1, SLC9A3R1, UGT2B17, VDR, WNT1**

☐ **Polydactyly, nonsyndromic**
Panel-ID: ID166.02 (small)
9 genes (20892 kb): CIBAR1, FBLN1, GLI1, GLI3,**, HOXD13, IQCE, KIAA0825, LMBR1, ZNF141*

☐ **Skeletal dysplasia, severe type**
Panel-ID: ID056.01 (large)
46 genes (144189 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*

Chondrodysplasia, lethal:
12 genes (24543 bp): AGPS, EBP, FLNB, GDF5, GNPAT, GPX4, LBR, PEX5, PEX7*, PTH1R, SLC26A2*, SLC35D1*

Osteogenesis imperfecta (OI), lethal:
5 genes (12564 bp): CRTAP, COL1A1*,**, COL1A2*,**, P3H1*, PPIB*

Short-rip thoracic dysplasia (SRTD), lethal:
9 genes (34983 bp): CEP120, DYNC2H1, IFT52, IFT80, IFT81, KIAA0586, NEK1, WDR34, WDR35

Achondrogenesia (ACG):
4 genes (14130 bp): COL2A1,**, GDF5, SLC26A2*, TRIP11*

Fibrochondrogenesia (FBCG):
2 genes (10631 bp): COL11A1,**, COL11A2**

Thanatophoric Dysplasia (TD):
2 genes (6885 bp): COL1A2,**, FGFR3*,***

☐ **Spondylocostal dysostosis (SCDO)**
Panel-ID: ID227.00 (small)
7 genes (14355 kb): DLL3, FLNB, HES7, LFNG, MESP2, RIPPLY2, TBX6

☐ **Spondyloepiphyseal dysplasia and Spondylometaphyseal dysplasia (SED, SMD, SEMD)**
Panel-ID: ID110.01 (small)
39 genes (90660 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 (25899 bp): ACAN, CHST3, COL2A1,**, COMP, MBTPS1, SMARCAL1, TRAPPC2, TRPV4*, WISP3*

Spondylometaphyseal dysplasia (SMD):
11 genes (30138 bp): ACP5, CFAP410, COL2A1,**, COL10A1, FN1, GPX4, PAM16, PCYT1A, PLCB3, TRIP11, TRPV4**

Spondyloepimetaphyseal dysplasia (SEMD):
21 genes (47547 bp): ACAN, AIFM1, B3GALT6, BGN, COL2A1*,**, DDR2, DDRGK1, EXOC6B, KIF22, MATN3, MMP13, NANS, NEPRO, PAPSS2, PISD, POP1, RPL13, RSPRY1, SIK3, TONSL, UFSP2*

Ciliopathies

☐ **Short-rip polydactyly syndrome and Jeune syndrome (SRTD)**
Panel-ID: ID067.00 (small)
20 genes (68106 kb): CEP120, DYNC2H1, DYNC2LI1, EVC, EVC2, IFT43, IFT52, IFT80, IFT81, IFT140, IFT172, INTU, KIAA0586, NEK1, TCTEX1D2, TTC21B, WDR19, WDR34, WDR35, WDR60*

Basis-Panel I (Jeune syndrome):
5 genes (25218 bp): DYNC2H1, IFT43, FT80, IFT140, WDR19*

Basis-Panel II (Ellis-van Crefeld syndrome):
4 genes (11499 bp): DYNC2LI1, EVC, EVC2, WDR35

Connective Tissue Diseases

☐ **Osteogenesis imperfecta (OI)**
Panel-ID: ID066.02 (small)
21 genes (38243 kb): ANO5, BMP1, COL1A1,**, COL1A2*,**, CREB3L1, CRTAP*, FKBP10, IFITM5*, LRP5*, MBTPS2, MESD, P3H1*, PLOD2*, PPIB, SERPINF1, SERPINH1, SP7, SPARC, TENT5A, TMEM38B, WNT1**

Developmental and Growth Disorders

☐ **Three M syndrome (3M)**
Panel-ID: ID214.00 (small)
*3 genes (12396 kb): CCDC8, CUL7, OBSL1**

Kidney Diseases

☐ **Hypophosphathemic rickets and Hypophosphatasia**
Panel-ID: ID269.01 (small)
14 genes (24990 kb): ALPL, CLCN5, CYP2R1, CYP3A4, CYP27B1, DMP1, ENPP1, FAH, FGF23, KL, PHEX, SLC34A1, SLC34A3, VDR*

Muscular Diseases

☐ Arachnodactyly
Panel-ID: ID124.00 (small)
13 genes (35865 kb): CHST14, CTSC, DSE, EFEMP2, FBN1,**, FBN2*, SCARF2, SKI*, TGFB2*, TGFB3*,**, TGFBR1*,**, TGFBR2*,**, SMAD3**

☐ Arthrogryposis multiplex congenita (AMC)
Panel-ID: ID200.00 (large)
56 genes (202722 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, MAGEL2, MUSK, MYBPC1, MYH3, MYH8, NALCN, NEB, NEK9, NUP88, PIEZO2*, PIP5K1C, PLOD2*, RAPSN*, RYR1*, SYNE1, TNNI2*, TNNT3, TPM2, TRPV4*, UBA1, VIPAS39, VPS33B, ZBTB42, ZC4H2*

Basis-Panel I (Distal arthrogryposis DA):
8 genes (25260 bp): ECEL1, MYH3, MYBPC1, PIEZO2, TNNI2*, TNNT3, TPM2, UBA1*

Basis-Panel II (Lethal congenital contrakture syndrome LCCS):
9 genes (25308 bp): ADGRG6, CNTNAP1, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42

Basis-Panel III (Cerebrooculofacioskeletal syndrome COFS): *4 genes (11208 bp): ERCC1, ERCC2, ERCC5, ERCC6*

Basis-Panel IV (Fetal akinesia FADS):
11 genes (20353 bp): ASCC1, CHRNA1, CHRND*, CHRNG*, DOK7, FLVCR2, GBE1, MAGEL2, MUSK, NUP88, RAPSN**

Basis-Panel V (Nemaline myopathy NEM):
5 genes (25000 bp): ACTA1, KLHL40, KLHL41, LMOD3, NEB (spec. exons)

☐ Distal arthrogryposis (DA)
Panel-ID: ID196.01 (small)
10 genes (39810 kb): ECEL1, FBN2, MYBPC1, MYH3, MYH8, PIEZO2*, TNNI2*, TNNT3, TPM2, UBA1*

☐ Fetal akinesia deformation sequence (FADS)
Panel-ID: ID201.00 (small)
10 genes (30261 kb): CHRNA1, CHRND*, CHRNG*, DOK7, GBE1, MUSK, MYOD1, NUP88, RAPSN*, RYR1**

☐ Lethal congenital contracture syndrome (LCCS)
Panel-ID: ID197.00 (small)
12 genes (34581 kb): ADCY6, ADGRG6, CNTN1, CNTNAP1, DNM2, ERBB3, GLDN, GLE1, MYBPC1, NEK9, PIP5K1C, ZBTB42

Skin and Dental Diseases

☐ Amelogenesis imperfecta (AI)
Panel-ID: ID232.01 (small)
17 genes (29385 kb): ACP4, AMBN, AMELX, AMTN, DLX3, ENAM, FAM20A, FAM83H, GPR68, ITGB6, KLK4, LAMB3, MMP20, ODAPH, RELT, SLC24A4, WDR72*

☐ Ectodermal dysplasia (ECTD)
Panel-ID: ID136.02 (small)
20 genes (27335 kb): CDH3, CST6, EDA, EDAR, EDARADD, GJB6*,**, HOXC13*, IKBKG, KDF1, KRT74, KRT85, MSX1*, KREMEN1, NECTIN1, NECTIN4*, NFKBIA, PKP1, TSPEAR, TP63, WNT10A*,**

☐ Epidermolysis bullosa (EB)
Panel-ID: ID198.01 (large)
28 genes (104439 kb): CD151, CDSN, CHST8, COL17A1, COL7A1, CSTA, DSP*,**, DST, EXPH5, FERMT1, FLG2, ITGA3, ITGA6, ITGB4, JUP*,**, KLHL24, KRT1, KRT10, KRT14*, KRT5, LAMA3, LAMB3*, LAMC2, MMP1, PKP1, PLEC, SERPINB8, TGM5*

Epidermolysis bullosa simplex (EBS):
6 genes (31929 bp): DST, ITGB4, KLHL24, KRT14, KRT5, PLEC*

Epidermolysis bullosa junctionalis (JEB):
6 genes (27592 bp): COL17A1, ITGA6, ITGB4, LAMA3, LAMB3, LAMC2*

Epidermolysis bullosa simplex (EBS):
2 genes (10245 bp): COL7A1, MMP1*

Peeling-Skin syndrome (PSS):
6 genes (13626 bp): CDSN, CHST8, CSTA, FLG2, SERPINB8, TGM5

☐ Hypotrichosis, nonsyndromic (HYPT)
Panel-ID: ID146.01 (small)
*12 genes (20127 kb): APCDD1, CDSN, DSG4, EPS8L3, HR, KRT71, KRT74, LIPH, LPAR6, LSS, RPL21, SNRPE**

☐ Ichthyosis
Panel-ID: ID217.01 (small)
38 genes (72972 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 rezessive (ARCI):
14 genes (28698 bp): ABCA12, ALOX12B, ALOXE3, CASP14, CERS3, CYP4F22, LIPN, NIPAL4, PNPLA1, SDR9C7, SLC27A4, ST14, SULT2B1, TGM1

Ichthyosis, autosomal dominant and X-linked:
7 genes (21261 bp): ASPRV1, FLG, GJB2,**, KRT1, KRT2, KRT10, STS*

☐ Orofacial cleft (OFC)
Panel-ID: ID266.00 (large)
41 genes (129720 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*

Basis-Panel I (Orofacial cleft, nonsyndromic):
10 genes (15297 bp): ARHGAP29, BMP4, DLX4, GRHL3, IRF6,**, MSX1*, NECTIN1, SUMO1, TBX22, TP63*

Basis-Panel II (Orofacial cleft, syndromic):
9 genes (24828 bp): CDH1,**, FLNB, GRHL3, MID1*,**, IRF6*,**, SLC26A2, SPECC1L, TBX22, TP63*

Basis-Panel III (Orofaciodigital syndrome):
7 genes (24651 bp): C2CD3, CPLANE1, DDX59, IFT57, OFD1, TCTN3, TMEM107*

Basis-Panel IV (Kabuki syndrome):
2 genes (20814 bp): KDM6A,**, KMT2D*,***

Basis-Panel V (CHARGE syndrome):
3 genes (12876 bp): CHD7,**, SEMA3E, TBX22*

Basis-Panel VI (Cornelia de Lange syndrome):
5 genes (18801 bp): HDAC8, NIPBL*,**, RAD21, SMC1A*, SMC3**

☐ **Pachyonychia congenita (PC)**
Panel-ID: ID120.01 (small)
13 genes (30129 kb): AAGAB, ATP2A2, DSG1, DSP, GJB6*,**, KRT1, KRT16, KRT17, KRT6A, KRT6B, KRT6C, MBTPS2, TRPV3*

☐ **Selective tooth agenesis (STHAG)**
Panel-ID: ID151.01 (small)
15 genes (24906 kb): EDA, EDAR*, EDARADD, GREM2*, IRF6*,**, KDF1, KREMEN1, LRP6, LTBP3, MSX1*, PAX9, TP63, TSPEAR, WNT10A*,**, WNT10B**

Eye Diseases

☐ **Oculocutaneous albinism (OCA)**
Panel-ID: ID082.01 (small)
8 genes (11562 kb): GPR143, LRMDA, MC1R*, OCA2*,**, SLC24A5, SLC45A2, TYR*,**, TYRP1*

Skeletal and Bone Diseases

☐ **Adams-Oliver syndrome (AOS)**
Panel-ID: ID259.00 (small)
6 genes (23232 kb): ARHGAP31, DLL4, DOCK6, EOGT, NOTCH1*, RBPJ*

☐ **Multiple pterygium syndrome**
Panel-ID: ID158.01 (small)
8 genes (16734 kb): CHRNA1, CHRNB1, CHRND*, CHRNG*, IRF6*,**, LMX1B, MYH3, RIPK4**

Tumor Syndromes

☐ **Basal cell nevus syndrome (BCNS)**
Panel-ID: ID174.00 (small)
3 genes (9402 kb): PTCH1,**, PTCH2*, SUFU**

☐ **Xeroderma pigmentosum (XP)**
Panel-ID: ID282.00 (small)
93 genes (23469 kb): DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, POLH, XPA, XPC

Tumor Syndromes

☐ **Breast and ovarian cancer**
Panel-ID: ID003.02 (small)
17 genes (56508 kb): ATM,**, BARD1*,**, BRIP1*, BRCA1*,**, BRCA2*,**, CDH1*,**, CHEK2*,**, MLH1*,**, MSH2*,**, MSH6*,**, PALB2*,**, PMS2*,**, PTEN*,**, RAD51C*,**, RAD51D*, STK11*,**, TP53*,***

Breast cancer:
10 genes (38892 bp): ATM,**, BARD1*,**, BRCA1*,**, BRCA2*,**, CDH1*,**, CHEK2*,**, PALB2*,**, PTEN*,**, STK11*,**, TP53*,***

Ovarian cancer:
12 genes (39510 bp): BRCA1,**, BRCA2*,**, BRIP1*, MLH1*,**, MSH2*,**, MSH6*,**, PALB2*,**, PMS2*,**, RAD51C*,**, RAD51D*, STK11*,**, TP53*,***

☐ **Breast cancer**
Panel-ID: ID021.01 (small)
10 genes (38892 kb): ATM,**, BARD1*,**, BRCA1*,**, BRCA2*,**, CDH1*,**, CHEK2*,**, PALB2*,**, PTEN*,**, STK11*,**, TP53*,***

☐ **Breast-ovarian cancer - BRCA1/BRCA2**
Panel-ID: ID001.00 (small)
2 genes (15843 kb): BRCA1,**, BRCA2*,***

☐ **Breast-ovarian cancer, comprehensive diagnostics**
Panel-ID: ID068.01 (large)
50 genes (151495 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*

Breast cancer:
10 genes (38892 bp): ATM,**, BARD1*,**, BRCA1*,**, BRCA2*,**, CDH1*,**, CHEK2*,**, PALB2*,**, PTEN*,**, STK11*,**, TP53*,***

Ovarian cancer:
12 genes (39510 bp): BRCA1,**, BRCA2*,**, BRIP1*, MLH1*,**, MSH2*,**, MSH6*,**, PALB2*,**, PMS2*,**, RAD51C*,**, RAD51D*, STK11*,**, TP53*,***

Fanconi anemia:
19 genes (63261 bp): BRCA1,**, BRCA2*,**, BRIP1*, ERCC4, FANCA, FANCB, FANCC*, FANCD2*, FANCE, FANCF, FANCG, FANCI*, FANCL, FANCM, PALB2*,**, RAD51, RAD51C*,**, SLX4, XRCC2*

☐ **Hereditary tumor diseases, comprehensive diagnostics**
Panel-ID: ID018.01 (large)
113 genes (285282 kb): ACD, AIP, AKT1, APC*,**, ATM*,**, BARD1, BAP1, BLM, BMPR1A*,**, BRCA1*,**, BRCA2*,**, BRIP1*,**, CASR, CDC73, CDH1*,**, CDK4*, CDKN1B, CDKN2A*,**, CEBPA,CHEK2*,**,CTCR*, DDB2, DICER1, DIS3L2, EPCAM**, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FAM175A, FANCA, FANCB, FANCC*, FANCD2*, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH*,**, FLCN*,**, GALNT12, GATA2, GPC3*,**, GREM1, HOXB13*, KIF1B, KIT*,**, LZTR1*,**, MAX, MEN1*,**, MET*, MITF, MLH1*,**, MRE11A, MSH2*,**, MSH3, MSH6*,**, MUTYH*,**, NBN*, NF1*,**, NF2*,**, NSD1*,**, NTHL1, PALB2*,**, PDGFRA, PHOX2B*,**,PIK3CA*, PMS2*,**, POLD1*, POLE*, POT1*, PRKAR1A, PTCH1*,**, PTEN*,**, RAD50, RAD51, RAD51B, RAD51C*,**, RAD51D*, RB1, RECQL4, RET*,**, RHBDF2, RINT1, RUNX1, SDHA*, SDHAF2, SDHB*, SDHC, SDHD*, SLX4, SMAD4*,**, SMARCA4, SMARCB1*,**, SMARCE1, SPINK1*,**, SPRED1*, STK11*,**, SUFU*, TERF2IP, TERT, TMEM127, TP53*,**, TSC1*,**, TSC2*,**, VHL*,**,WT1*,**, XPA, XPC, XRCC2*

☐ **Medulloblastoma**
Panel-ID: ID205.00 (small)
17 genes (60924 kb): APC,**, BRCA2*,**, CHEK2*,**, DICER1, ERCC2, FANCM, MLH1*,**, MSH2*,**, MSH6*,**, NBN*, PALB2*,**, PMS2*,**, PTCH1*,**, SMARCB1*,**, SUFU*, TP53*,**, VHL*,**

☐ **Osteosarcoma**
Panel-ID: ID223.00 (small)
8 genes (25383 kb): ATM,**, BLM, CDKN2A*,**, CHEK2*,**, ERCC2, RB1*, RECQL4, TP53*,**

☐ **Ovarian cancer**
Panel-ID: ID004.02 (small)
12 genes (39510 kb): BRCA1,**, BRCA2*,**, BRIP1*, MLH1*,**, MSH2*,**, MSH6*,**, PALB2*,**, PMS2*,**, RAD51C*,**, RAD51D*, STK11*,**, TP53*,**

☐ **Prostata cancer**
Panel-ID: ID140.01 (small)
26 genes (89544 kb): AR,**, ATM*,**, ATR*,**, BAP1, BRCA1*,**, BRCA2*,**, CYP3A43, CDH1*,**, CHEK2*,**, EHBP1, ELAC2, EPCAM**, HOXB13*, MLH1*,**, MSR1, MRE11, MSH2*,**, MSH6*,**, NBN*, PALB2*,**, PMS2*,**, RAD51C*,**, RAD51D*,**, RNASEL, TRRAP, TP53*,**

Endocrine Disorders

☐ **Paragangliomas and Pheochromocytoma**
Panel-ID: ID042.02 (small)
16 genes (29376 kb): FH,**, DLST, GDNF, KIF1B, MAX, MEN1*,**, NF1*,**, RET*,**, SDHA*, SDHAF2, SDHB*, SDHC, SDHD*, SLC25A11, TMEM127, VHL*,**

☐ **Thyroid cancer**
Panel-ID: ID220.01 (small)
25 genes (61881 kb): AKT1, APC*,**, ATM*,**, CDKN1B, CHEK2*,**, DICER1, FOXE1, HABP2, MAP2K5, MEN1*,**, MET*, MINPP1, NKX2-1, NTRK1, PIK3CA*, PRKAR1A, PTEN*,**, RET*,**, SDHA*, SDHB*, SDHC, SDHD*, SEC23B*, SRGAP1, TP53*,**

Gastroenterological Diseases

☐ **Colorectal cancer (CRC)**
Panel-ID: ID049.01 (small)
14 genes (58233 kb): ATM,**, CHEK2*,**, EPCAM**, MLH1*,**, MSH2*,**, MSH6*,**, MUTYH*,**, NTHL1*, PMS2*,**, POLD1*, POLE*, PTEN*,**, SMAD4*,**, STK11*,**

☐ **Colorectal cancer und polyposis**
Panel-ID: ID006.07 (small)
20 genes (58233 kb): ATM,**, CHEK2*,**, EPCAM**, MLH1*,**, MSH2*,**, MSH6*,**, MUTYH*,**, NTHL1*, PMS2*,**, POLD1*, POLE*, PTEN*,**, SMAD4*,**, STK11*,***

Colorectal polyposis:
12 genes (33555 bp): APC,**, AXIN2*, BMPR1A*,**, GREM1, MSH3, MUTYH*,**, NTHL1*, POLD1*, POLE*, PTEN*,**, SMAD4*,**, STK11*,**

Colorectal cancer:
14 genes (40419 bp): ATM,**, CHEK2*,**, EPCAM**, MLH1*,**, MSH2*,**, MSH6*,**, MUTYH*,**, NTHL1*, PMS2*,**, POLD1*, POLE*, PTEN*,**, SMAD4*,**, STK11*,**

☐ **Colorectal cancer with microsatellite instability (MSI-H)**
Panel-ID: ID283.00 (small)
*9 genes (25443 kb): EPCAM**, MLH1*,**, MSH2*,**, MSH6*,**, MUTYH*,**, NTHL1*, PMS2*,**, POLD1*, POLE**

☐ **Colorectal cancer, comprehensive diagnostics**
Panel-ID: ID285.00 (small)
34 genes (90279 kb): APC,**, ATM*,**, AXIN2*, BLM, BMPR1A*,**, CDH1*,**, CHEK2*,**, EPCAM**, EXO1, GALNT12, GREM1, MLH1*,**, MLH3*, MSH2*,**, MSH3, MSH6*,**, MUTYH*,**, NBN*, NTHL1*, PLA2G2A, PMS1*, PMS2*,**, POLD1*, POLE*, PTEN*,**, RFC1, RPA1, RNF43*, RPS20, SMAD4*,**, SMAD7, STK11*,**, TGFBR2*,**, TP53*,**

Colorectal polyposis:
12 genes (33555 bp): APC,**, AXIN2*, BMPR1A*,**, GREM1, MSH3, MUTYH*,**, NTHL1*, OLD1*, POLE*, PTEN*,**, SMAD4*,**, STK11*,**

Colorectal cancer:
14 genes (40419 bp): ATM,**, CHEK2*,**, EPCAM**, MLH1*,**, MSH2*,**, MSH6*,**, MUTYH*,**, NTHL1*, PMS2*,**, POLD1*, POLE*, PTEN*,**, SMAD4*,**, STK11*,**

☐ **Gastric cancer**
Panel-ID: ID090.01 (small)
20 genes (52307 kb): APC,**, ATM*,**, BMPR1A*,**, CDH1*,**, CHEK2*,**, KIT*,**, MLH1*,**, MSH2*,**, MSH6*,**, MUTYH*,**, PDGFRA, SDHA*, PMS2*,**, PTEN*,**, SDHB*, SDHC, SDHD*, SMAD4*,**, STK11*,**, TP53*,**

☐ **Lynch syndrome (HNPCC)**
Panel-ID: ID002.02 (small)
5 genes (12693 kb): MLH1,**, MSH2*,**, MSH6*,**, PMS2*,**, EPCAM**

☐ **Pancreatic cancer**
Panel-ID: ID089.02 (small)
16 genes (54249 kb): APC,**, ATM*,**, BRCA1*,**, BRCA2*,**, CDKN2A*,**, CTRC*, MLH1*,**, MSH2*,**, MSH6*,**, PALB2*,**, PMS2*,**, PRSS1*,**, SPINK*,**, STK11*,**, TP53*,**, VHL*,**

☐ **Pancreatic cancer, comprehensive diagnostics**
Panel-ID: ID284.00 (small)
24 genes (67863 kb): APC,**, ATM*,**, BMPR1A*,**, BRCA1*,**, BRCA2*,**, BUB1B*, CDKN2A*,**, CHEK2*,**, CTRC*, EPCAM**, MLH1*,**, MSH2*,**, MSH6*,**, MUTYH*,**, NBN*, PALB2*,**, PMS2*,**, PRSS1*,**, RABL3, SMAD4*,**, SPINK*,**, STK11*,**, TP53*,**, VHL*,**

☐ **Polyposis syndrome (PS, FAP)**
Panel-ID: ID005.04 (small)
12 genes (33555 kb): APC,**, AXIN2*, BMPR1A*,**, GREM1, MSH3, MUTYH*,**, NTHL1*, POLD1*,**, POLE*,**, PTEN*,**, SMAD4*,**, STK11*,**

Kidney Diseases

☐ **Renal cell cancer**
Panel-ID: ID041.01 (small)
27 genes (59889 kb): BAP1, DIS3L2, FH,**, FLCN*,**, HNF1A*,**, HNF1B*,**, MET*, MITF, MLH1*,**, MSH2*,**, MSH6*,**, PBRM1, PMS2*,**, PTEN*,**, RNF139, SDHA, SDHAF2, SDHB*, SDHC, SDHD*, SLC49A4, SMARCA4, SMARCB1*,**, TSC1*,**, TSC2*,**, VHL*,**, WT1*,**

Pulmonary and Respiratory Diseases

☐ **Lung cancer**
Panel-ID: ID260.02 (large)
33 genes (122509 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*,***

Skin and Dental Diseases

☐ **Cowden syndrome (CWS)**
Panel-ID: ID075.01 (small)
8 genes (12768 kb): AKT1, PIK3CA*, PTEN*,**, SEC23B*, SDHB*,**, SDHC**, SDHD*.**, WWP1*

☐ **Cutaneous malignant melanoma (CMM)**
Panel-ID: ID193.01 (small)
12 genes (26373 kb): BAP1, BRCA2,**, CDK4*, CDKN2A*,**, MC1R*, MITF, POT1*, PTEN*,**, TERT, TP53*,**, TYR, XRCC3*

☐ **Neurofibromatosis (NF)**
Panel-ID: ID210.00 (small)
3 genes (11571 kb): NF1,**, NF2*,**, SPRED1**

Skin and Dental Diseases, Hematologic Diseases

☐ **Fanconi anemia**
Panel-ID: ID043.01 (small)
19 genes (63261 kb): BRCA1,**, BRCA2*,**, BRIP1*,**, ERCC4, FANCA, FANCB, FANCC*, FANCD2*, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, PALB2*,**, RAD51, RAD51C*,**, SLX4, XRCC2*