Aktuelle Liste der Verfahren im flexiblen Akkreditierungsbereich nach DIN EN ISO 15189:2014

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Universitätsklinikum Aachen
Institut für Humangenetik und Genommedizin
Pauwelsstraße 30, 52074 Aachen

Untersuchungen im Bereich:

Medizinische Laboratoriumsdiagnostik

Untersuchungsgebiete:
Humangenetik (Molekulare Humangenetik)

Innerhalb der mit ** gekennzeichneten Untersuchungsbereiche ist dem Laboratorium, ohne dass es einer vorherigen Information und Zustimmung der Deutschen Akkreditierungsstelle GmbH bedarf, die Modifizierung sowie Weiter- und Neuentwicklung von Untersuchungsverfahren gestattet.
Die aufgeführten Untersuchungsverfahren sind beispielhaft. Das Laboratorium verfügt über eine aktuelle Liste aller Untersuchungsverfahren im flexiblen Akkreditierungsbereich.

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Untersuchungsart:
Molekularbiologische Untersuchungen (Ampflifikationsverfahren)**

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät
H19/LIT1 (Beckwith-Wiedemann-Syndrom/Silver-Russell-Syndrom)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MS-MLPA	QMH_VA_MLPA: V10 QMH_AA_Imprintingerkrankungen: V09	Thermocycler AB3500
UBE3A (Angelman-Syndrom)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MS-MLPA	QMH_VA_MLPA: V10 QMH_AA_Imprintingerkrankungen: V09	Thermocycler AB3500
SNRPN (Prader-Willi-Syndrom)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MS-MLPA	QMH_VA_MLPA: V10 QMH_AA_Imprintingerkrankungen: V09	Thermocycler AB3500

MEG3 (Temple-Syndrom, KOS14)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MS-MLPA	QMH_VA_MLPA: V10 QMH_AA_Imprintingerkrankungen: V09	Thermocycler AB3500
GNAS (PHPIb)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MS-MLPA	QMH_VA_MLPA: V10 QMH_AA_Imprintingerkrankungen: V09	Thermocycler AB3500
GRB10 (Silver-Russell-Syndrom)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MS-MLPA	QMH_VA_MLPA: V10 QMH_AA_Imprintingerkrankungen: V09	Thermocycler AB3500
MEST (Silver-Russell-Syndrom)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MS-MLPA	QMH_VA_MLPA: V10 QMH_AA_Imprintingerkrankungen: V09	Thermocycler AB3500
PLAGL1 (TNDM)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MS-MLPA	QMH_VA_MLPA: V10 QMH_AA_Imprintingerkrankungen: V09	Thermocycler AB3500
BRCA1/2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10 QMH_AA_HBOC_Tumor-Diagnostik: V10	Thermocycler AB3500
DMD/BMD	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_AA_Muskeldystrophie: V01	Thermocycler AB3500
HSP	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_AA_NMD_divers: V01	Thermocycler AB3500
Marfan	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10; QMH_AA_EDS_MFS: V01	Thermocycler AB3500
NPHP1 (Joubert-Syndrom, Nephronophthise)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler AB3500
VHL	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler AB3500
PMP22 (CMT1A, HNPP)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_AA_CMT: V01	Thermocycler AB3500

SMN1 (SMA)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_AA_SMA: V04	Thermocycler AB3500
PKHD1 (ARPKD)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler AB3500
HNF1beta (RCAD, MODY5)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler AB3500
MFN2 (CMT)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA, QMH_AA_CMT	Thermocycler AB3500
MPZ (CMT)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_AA_CMT: V01	Thermocycler AB3500
PKD1 (ADPKD)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler AB3500
PKD2 (ADPKD)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler AB3500
SHOX	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	MLPA	QMH_VA_MLPA: V10, QMH_AA_SHOX: V01	Thermocycler AB3500
PABY/X	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	PCR	QMH_VA_PCR: V09, QMH_AA_Kerngeschlechtsbestimmung mittels PABY-PCR: V02	Thermocycler AB3500
DMPK (Myotone Dystrophie Typ I)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Myopathie: V01	Thermocycler AB3500
FMR1-Gen (fraX, POF, FXTAS)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalyse	QMH_AA_FraX: V06	Thermocycler AB3500
ZNF9 (DM2/PROMM)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Myopathie: V01	Thermocycler AB3500

ATXN2 (SCA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Ataxie: V01	Thermocycler AB3500
ATXN3 (SCA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Ataxie: V01	Thermocycler AB3500
CACNA1A (SCA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Ataxie: V01	Thermocycler AB3500
ATXN7 (SCA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Ataxie: V01	Thermocycler AB3500
ATXN8oS (SCA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Ataxie: V01	Thermocycler AB3500
TBP (SCA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR, QMH_AA_Ataxie	Thermocycler AB3500
FXN (FRDA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Ataxie: V01	Thermocycler AB3500
AR (SBMA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_VA_SMA: V04	Thermocycler AB3500
ATXN1 (SCA)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Ataxie: V01	Thermocycler AB3500
c9orf72 (ALS)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_ALS: V03	Thermocycler AB3500
PABPN1 (OPMD)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Muskeldystrophie: V01	Thermocycler AB3500
RFC1 (CANVAS)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/Fragmentanalytik	QMH_VA_PCR: V09, QMH_AA_Ataxie: V01	Thermocycler AB3500

SMN1 (SMA)	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	PCR/RFLP/Fragmentanalytik/ DNA Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_SMA: V04	Thermocycler AB3500
UGT1A1	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	PCR/Fragmentanalytik/DNA Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_UGT1A: V01	Thermocycler AB3500
SLC3A1 (Cystinurie)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_Cystinurie: V02	Thermocycler AB3500
SLC7A9 (Cystinurie)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_Cystinurie: V02	Thermocycler AB3500
VHL	EDTA-Blut, Mundschleimhaut, CVS, Fruchtwasser, Fibroblasten; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_ZN_Diagnostik: V04	Thermocycler AB3500
IGHMBP2 (SMARD)	EDTA-Blut, Fruchtwasser, CVS; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_SMA: V04	Thermocycler AB3500
SHOX (Kleinwuchs)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_SHOX: V01	Thermocycler AB3500
HFE (Hämochromatose: rs1800562, rs1799945)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_Hämochromatose: V04	Thermocycler AB3500
F2 (Prothrombin: rs1799963)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_Faktor II (Prothrombin): V04	Thermocycler AB3500

F5 (Faktor-V-Leiden: rs6025)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_Faktor V Leiden: V04	Thermocycler AB3500
DPYD (rs3918290, rs55886062, rs67376798, rs75017182)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_AA_DPYD_DPD: V08	Thermocycler AB3500
Analyt (Meßgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**
Amylotrophe Lateralsklerose (ALS): ALS2, ANG, ANXA11, C9orf72, CHCHD10, FIG4, FUS, SETX, SOD1, TARDBP, TUBA4A, UBQLN2, VAPB	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_ALS: V01	Thermocycler; AB3500; NovaSeq
Aorten-Aneurysma: ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB1, TGFB2, TGFBR1, TGFBR2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_EDS_MFS: V01	Thermocycler; AB3500; NovaSeq
Arrhythmias: CACNA1C, CALM1, CALM2, CAV3, HCN4, KCNE1, KCNE2, KCNH2, KCNQ1, MYH6, SCN5A	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Kardiomyopathie: V01	Thermocycler; AB3500; NovaSeq
Arthrogryposis: FBN2, MYH3, PIEZO2, TNNI2, TNNT3, TPM2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Ataxie (episodisch): CACNA1A, CACNB4, KCNA1, PRRT2, SCN2A, SLC1A3, SLC2A1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH-AA_Ataxie: V02	Thermocycler; AB3500; NovaSeq
Autismus: ARID1A, ARID1B, CACNA1C, CDKL5, DEPDC5, DYRK1A, FOXP1, KCNQ2, KCNT1, MBD5, MECP2, PRRT2, PTEN, SCN1A, SCN2A, SCN8A, SHANK3, SLC2A1, STXBP1, SYNGAP1, TCF4, UBE3A, ZEB2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Autosomal-dominate Zystennierenerkrankung (ADPKD): ALG5, ALG9, DNAJB11, GANAB, HNF1B, IFT140, PAX2, PKD1, PKD2, UMOD	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Autosomal-rezessive Zystennierenerkrankung (ARPKD): DZIP1L , HNF1B, INVS, NPHP3, PKHD1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler; AB3500; NovaSeq
Bardet-Biedel-Syndrom (BBS): ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, MKKS, MKS1, TRIM32, TTC8	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler; AB3500; NovaSeq

Beckwith-Wiedemann-Syndrom-Spektrum (BWSp): CDKN1C, DIS3L2, EED, EZH2, GPC3, GPC4, HRAS, NFIX, NSD1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Imprintingerkrankungen_ V09	Thermocycler; AB3500; NovaSeq
Branchiootorenales Syndrom/Renal agenesis: EYA1, FGF20, FRAS1, FREM2, GFRA1, GREB1L, HNF1B, IFT27, ITGA8, KIF14, PAX2, RET	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
CAKUT: BMP4, CHD1L, DSTYK, EYA1, GATA3, HNF1B, PAX2, ROBO2, SALL1, TBX18	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler; AB3500; NovaSeq
Charcot-Marie-Tooth-Erkrankung (CMT): AARS1, BSCL2, DNM2, EGR2, GARS1, GDAP1, GJB1, HSPB1, IGHMBP2, LITAF, LMNA, MFN2, MME, MPZ, MTMR2, NEFL, PMP22, PRX, RAB7A, SH3TC2, TRPV4	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_CMT_ V01	Thermocycler; AB3500; NovaSeq
Cholestase: ABCB11, ABCB4, ABCG5, ABCG8, ATP8B1, NR1H4, TJP2, TRMU, UGT1A1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Colonkarzinom (MS-stabil): BMPR1A, BUB1B, MSH3, MUTYH, POLD1, POLE, PTEN, SMAD4, STK11, TP53	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Colon-Karzinom (FAP)/GIST: APC, EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Demenz: APP, CHCHD10, CHMP2B, CSF1R, FUS, GRN, MAPT, NOTCH3, PRNP, PSEN1, PSEN2, SQSTM1, TARDBP, TBK1, TREM2, VCP	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH-AA-Demenz: V01	Thermocycler; AB3500; NovaSeq
Diamond blackfan anemia (DBA): GATA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, SBDS, TSR2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Dyskeratosis congenita (DKC): ACD, AK2, DKC1, FERMT1, LIG4, NHP2, NOP10, PARN, RTEL1, TERT, TINF2, USB1, WRAP53	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Dystrophien (DYS): ANO5, CAPN3, CAV3, COL6A1, COL6A2, COL6A3, DMD, DYSF, EMD, FKRP, LMNA, SGCA, SGCB, SGCD, SGCG, SYNE1, TMEM43	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Muskeldystrophien: V01	Thermocycler; AB3500; NovaSeq
Ehlers-Danlos-Syndrom (EDS): ADAMTS2, B4GALT7, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, FKBP14, FLNA, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_CMT: V01	Thermocycler; AB3500; NovaSeq
Epilepsie: DEPDC5, KCNQ2, KCNT1, PRRT2, SCN1A, SCN2A, SCN8A, SLC2A1, STXBP1, SYNGAP1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Epilepsie: V01	Thermocycler; AB3500; NovaSeq
Focal_Epilepsy: CHRNA2, CHRNA4, CHRNB2, DEPDC5, KCNQ2, KCNQ3, KCNT1, LGI1, MICAL1, NPRL2, NPRL3, PRIMA1, PRRT2, RELN, SCN2A, SCN8A, TSC1, TSC2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Epilepsie: V01	Thermocycler; AB3500; NovaSeq
Epileptic_Encephalopathy: CDKL5, GNAO1, GRIN2A, KCNA2, KCNB1, KCNQ2, KCNT1, PCDH19, SCN1A, SCN2A, SCN8A, SLC2A1, SLC6A1, STXBP1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Epilepsie: V01	Thermocycler; AB3500; NovaSeq

Erythrocytose: BHLHE41, BPGM, EGLN1, EGLN2, EGLN3, EPAS1, EPO, EPOR, GFI1B, HBA1, HBA2, HBB, HIF1A, HIF1AN, HIF3A, JAK2, KDM6A, OS9, SH2B3, VHL, ZNF197	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Familial_Hemiplegic_Migraine: ATP1A2, ATP1A3, CACNA1A, KCNK18	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Epilepsie: V01	Thermocycler; AB3500; NovaSeq
Fiebersyndrome: ELANE, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRP12, NLRP3, NOD2, PLCG2, PSTPIP1, TNFRSF1A	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Genetic_Generalized_Epilepsy: GABRD, GABRG2, HCN1, HCN2, KCNMA1, SCN1A, SCN1B, SCN2A, SLC2A1, STX1B	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Epilepsie: V01	Thermocycler; AB3500; NovaSeq
Gerinnungsstörung: F10, F13B, F2, F5, F9, FGG, PROC, PROS1, SERPINC1, SERPIND1, THBD, VWF	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Herditäre Kardiomyopathie (HCM): ACTC1, ACTN2, MYBPC3, MYH7, MYL2, MYL3, PLN, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTN	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Kardiomyopathien: V02	Thermocycler; AB3500; NovaSeq
Dilatative Kardiomyopathie (DCM): ACTC1, ACTN2, DES, LMNA, MYBPC3, MYH7, PLN, RBM20, TNNC1, TNNI3, TNNT2, TPM1, TTN	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Kardiomyopathien: V01	Thermocycler; AB3500; NovaSeq
Hereditäre Leukämie (HerLeuk): ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, CBL, CDKN2A, CEBPA, CHEK2, DDX41, DKC1, EPCAM, ETV6, FANCA, GATA1, GATA2, HRAS, IKZF1, JAK2, KRAS, MAP2K1, MAP2K2, MLH1, MSH2, MSH6, NBN, NF1, NRAS, PAX5, PMS2, PTPN11, RBBP6 , RIT1, RUNX1, SAMD9, SAMD9L, SBDS, SOS1, SRP72, TERT, TINF2, TP53	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Hereditäre myeloproliferative Neoplasie (HerMPN): BLM, BRCA2, CHEK2, JAK2, RBBP6 , SH2B3, TERT, TP53	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Hereditäre sensorisch und autonome Neuropathie (HSAN): ATL1, ATL3, DNMT1, DST, ELP1, GLA, KIF1A, NGF, NTRK1, PRDM12, RAB7A, RETREG1, SCN10A, SCN11A, SCN9A, SPTLC1, SPTLC2, STING1, TRPA1, TTR, WNK1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Sensible Neuropathien: V01	Thermocycler; AB3500; NovaSeq
Hereditäre spastische Paraparesen (HSP): ABCD1, ALS2, ATL1, CYP7B1, FA2H, KIF5A, PLP1, PNPLA6, REEP1, SPAST, SPG11, SPG7, WASHC5	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Hereditärer Brust- und Einerstockkrebs (HBOC): ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_HBOC_Tumor-Diagnostik: V10	Thermocycler; AB3500; NovaSeq
Hörstörung (autosomal-rezessiv): CDH23, GJB2, GJB6, MYO15A, MYO7A	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Hörstörung (autosomal-dominant): ACTG1, CCDC50, COCH, GJB2 , GJB6, KCNQ4, MYH9, POU3F4, PRPS1, TECTA, TMC1, WFS1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Immundefekte: ADA, CD247, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CORO1A, DCLRE1C, IL2RG, IL7R, JAK3, LIG4, NFKB1, PNP, RAG1, RAG2, ZAP70	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Inherited Bone Marrow Failure (IBMF): ACD, AK2, DDX41, DKC1, FANCA, GATA2, LIG4, NBN, NHP2, NOP10, PARN, RTEL1, SAMD9L, SRP72, TERT, TINF2, TP53, USB1, WRAP53	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Intellectual Disability (X-linked): AFF2, ARX, ATRX, CUL4B, DLG3, FTSJ1, GDI1, NEXMIF, PHF8, SLC6A8, USP9X	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Intellectual Disability: CTNNB1, KPTN, MAN1B1, MED23, PHF21A, PTEN, SCN2A, ST3GAL3, STXBP1, SYNGAP1, TRAPPC9	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Joubert-Syndrom (JBTS): AHI1, CC2D2A, CEP290, CPLANE1, TMEM67	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_VA_ZN_Diagnostik: V04	Thermocycler; AB3500; NovaSeq

Leukenzephalopathie: AARS2, COL4A1, CSF1R, EIF2B4, EIF2B5, NOTCH3	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Low-phospholipid-associated cholelithiasis (LPAC): ABCB11, ABCB4, ABCC2, ABCG5, ABCG8, ATP8B1, NR1H4, TJP2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Lymphom: ACAN, ATM, B2M, BLM, CHEK2, DICER1, ETV6, FAS, KDR, KLHDC8B, LIG1, LIG4, NBN, NFKBIA, NPAT, PIK3CD, POT1, SH2D1A, SOCS1, TNFAIP3, WAS	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Makrozephalie: AKT3, DNMT3A, EZH2, GCDH, GPC3, NFIX, NSD1, PTCH1, PTEN	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Makuladystrophie: ABCA4, BEST1, CNGB3, ELOVL4, IMPG1, IMPG2, MAPKAPK3, MFSD8, PROM1, RPE65, TIMP3	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Meckel-Gruber-Syndrom(MKS): B9D1, B9D2, CC2D2A, CEP290, MKS1, RPGRIP1L, TCTN2, TMEM216, TMEM67	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Melanom: BRAF, BRCA1, BRCA2, CDK4, CDKN2A, MC1R, MITF, POT1, STK11, TERT, TP53, TYR, XRCC3	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Mikrozephalie: ASPM, CASK, CENPJ, CHD7, LIG4, MCPH1, NBN, NIPBL, PAFAH1B1, TUBA1A, WDR62	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Mitochondropathien (nukleär): DGUOK, DNA2, FLAD1, HADHA, HADHB, ISCU, MGME1, OPA1, POLG, POLG2, RNASEH1, RRM2B, TK2, TMEM126B, TWNK, TYMP	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Mukopolysaccharidose: NAGLU, GLB1, HGSNAT, IDUA, GUSB, GNS, IDS, ARSB, GALNS, SGSH, HYAL1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Myopathien (MYO): BAG3, BIN1, CRYAB, DES, DNAJB6, DNM2, FHL1, FLNC, GNE, LDB3, MTM1, MTMR14, MYH2, MYOT, NEB, RYR1, SELENON, SQSTM1, TPM3, VCP	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Osteogenesis imperfecta: BMP1, CREB3L1, CRTAP, FKBP10, P3H1, PLOD2, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TENT5A, TMEM38B, WNT1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Pankreas-Tumor: ATM, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Pankreatitis: CASR, CEL, CELA3B, CFTR, CLDN2, CPA1, CTRC, KRT8, LPL, PRSS1, SPINK1, TRPV6	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Paragangliome: AIP, BUB1B, CASR, CDC73, CDKN1B, FH, IDH1, KIF1B, MAX, MEN1, RET, SDHA, SDHB, SDHC, SDHD, STK11, TMEM127, VHL	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Podozythopathien: ACTN4, CD2AP, COL4A1, COL4A3, COL4A4, COL4A5, COQ6, LAMB2, MYH9, MYO1E, NPHS2, PLCE1, TRPC6, WT1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Prämature Ovarial Insuffizienz (POI): AARS2, BMP15, CLPP, CYP17A1, CYP19A1, EIF2B5, FANCM, FOXL2, FSHB, FSHR, GALT, GGPS1, HARS2, HSD17B4, LARS2, MCM8, MCM9, MSH4, NOBOX, NR5A1, PMM2, POLG, POLG2, SOHLH1, TWNK	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Polydaktylie: CIBAR1, GLI1, GLI3, HOXD13, IQCE, LMBR1, MIPOL1, PITX1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Pontocerebelläre Hypoplasie 1b (PCH1B): EXOSC3, EXOSC8, PRDM13, TSEN54, VRK1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Rasopathie: A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MRAS, NF1, NRAS, PPP1CB , PTPN11, RAF1, RASA2 , RIT1, RRAS, RRAS2, SHOC2, SOS1, SOS2, SPRED1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Retinitis pigmentosa (Usher): EYS, MYO7A, PRPF31, PRPH2, RHO, RP1, RP2, RPGR, USH2A	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Schwannomatose: DICER1, KRAS, LZTR1, NF1, NF2, NRAS, PTPN11, SMARCB1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Silver-Russell-Syndrom-like (SRS): ANKRD11, BLM, CDKN1C, CUL7, HMGA2, IGF1, IGF2, MBTPS1, PCNT, PIK3R1, PLAG1, SRCAP, TRIM37	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_210624	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Imprintingerkrankungen_ V09	Thermocycler; AB3500; NovaSeq
Skelettdysplasie: COL1A1, COL1A2, COL2A1, COL9A1, FGFR3, SHOX, SLC26A2, SOX9, TRAPPC2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Small Fiber Neuropathie (SFN): ATL1, ATL3, GLA, SCN10A, SCN11A, SCN9A, SPTLC1, SPTLC2, TRPA1, TTR	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq

Spinale Muskelatrophie (SMA): BICD2, BSCL2, CHCHD10, DNAJB2, GARS1, HSPB8, IGHMBP2, REEP1, TRPV4, UBA1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR, QMH_VA_Sangersequenzierung, QMH_VA_NGS, QMH_VA_ZN	Thermocycler; AB3500; NovaSeq
Spinocerebelläre Ataxien (SCA): APTX, ATM, COQ8A, FGF14, ITPR1, POLG, PPP2R2B, PRKCG, SACS, SETX, SPG7, SPTBN2, TTBK2	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Transienter Neonataler Diabetes Mellitus (TNDM): ABCC8, GCK, HNF1B, INS, KCNJ11, PDX1	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	PCR/DNA-Sequenzierung; Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10, QMH_AA_Imprintingerkrankungen_ V09	Thermocycler; AB3500; NovaSeq
Whole Exome (IDT, xGen Exome Research Panel v2.0)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	NGS: Sequence capture (IDT, xGen Exome Research Panel v2.0); Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq
Whole Genome (Illumina DNA PCR-Free Prep)	EDTA-Blut, Fibroblasten, Mundschleimhaut; genomische DNA	Illumina DNA PCR-Free Prep; Sequencing-by-synthesis; Auswertesoftware: in-house-Pipeline NGS-HG-AC_V1.2	QMH_VA_PCR: V09, QMH_VA_Sangersequenzierung: V16, QMH_VA_NGS: V10	Thermocycler; AB3500; NovaSeq