Liste der Untersuchungsverfahren im flexiblen Akkreditierungsbereich

Untersuchungsgebiet: Humangenetik (Molekulargenetik)

Untersuchungsart: Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Asplenie (RPSA)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
3-MGA Aciduria, Typ VII, with cataracts, neurologic involvement and neutropenia (CLPB)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
3-MGA-aciduria Typ iV (SERAC1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
3-M-Syndrom 1 (CUL7)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
3-M-Syndrom 2 (OBSL1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
3-M-Syndrom 3 (CCDC8)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

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Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Achondrogenesis Typ iB (DTDST)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Achondrogenesis Typ II (COL2A1),	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P214)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Achondrogenesis TypI 1A (TRIP11)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Achondroplasie (FGFR3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Acromesomelic Dysplasia, Maroteaux Type (AMDM) (NPR2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Adrenogenitales Syndrom (CYP21A2, CYP11B1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P050)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
AGAT- Defizienz (GATM)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Alagille-Syndrom (JAG1, NOTCH2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P184)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Albinismus (OCA, OCA2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P325)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Allan-Herndon-Dudley-Syndrom; AHDS (SLC16A2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Alpa1-Antitrypsin-Mangel A1-AT (SERPINA1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Alpha-Methylacetoacetic Aciduria (ACAT1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Alport-Syndrom (COL4A3, COL4A4)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Alveolokapilläre Dysplasie (FOXF1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Amish frühkindliches Epilepsie-Syndrom (ST3GAL5)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Androgen-Resistenz (AR)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P074)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Anemie und Spinocerebrale Ataxie (ABCB7)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Angelman-Syndrom (UBE3A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P336)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Antithrombin-III-Mangel (SERPINC1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Apert-Syndrom (FGFR2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
ARC Syndrom (Arthrogryposis, renal dysfunction and cholestasis) (VIPAS39)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Argininosuccinase Mangel (ASL)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Aromatase Defizienz (CYP19A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Arrhythmogene rechtsventrikuläre Kardiomyopathie (TMEM43, JUP)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Ataxie (APTX)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Atelosteogenesis I –III (FLNB, SLC26A2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P435)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Autismus-Spektrum-Störungen: CHD2, CHD8, CTTNBP2, DIP2A, IQGAP3, KCNQ3, LAMC3 und RELN	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Autosomal-dominante polyzystische Nierenerkrankung/ ADPKD (PKD1 und PKD2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P351, P352)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Autosomal-rezessive polyzystische Nierenerkrankung/ ARPKD (PKHD1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P341, P342)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Azoospermiefaktor (SY84, SY86, SY127, SY134, SY254, SY255, SY160, SYCP3, SRY)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Balkenagenesie (Agenesie des Corpus callosum) (AKT3, ARID1A, ARID1B, ARX, B3GALTL, DISC1, DYNC1H1, EOMES, FOXG1, GCSH)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Baraitser-Winter- Syndrom (ACTB, ACTG1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Bardet-Biedl-Syndrom (BBS1, BBS2, BBS10)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Batten Disease/Ceroid Lipofuscinosis Neuronal 3 (CLN3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Beckwith-Wiedemann Syndrom (Deletionsanalyse)	genomische DNA	MS-MLPA (ME030)	MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500	nein	
Biotinidase Mangel (BTD)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Biotin-responsive Basalganglienerkrankung (BBGD) (SLC19A3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Blepharophimosis-Epicanthus-inversus-Syndrom BPES (FOXL2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Borjeson-Forssman-Lehmann Syndrom (PHF6)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Brachydactyly type E (PTHLH)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Brachydactyly-Syndactyly Syndrom (HOXD13)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Branchio-oto-renales Syndrom (Bor-Syndrom) (EYA1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P153)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Brown-Vialetto-van-Laere-Syndrom (SLC52A3, SLC52A2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Brust- und Eierstockkrebs (BRCA-Diagnostik) (ATM, BRCA1, BRCA2, CHEK2, CDH1, PALB2, RAD51D und TP53)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P002, P045, P077)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Campomele Dysplasie (SOX9)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Carney complex, type 1; CNC1 (PRKAR1A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Carnitine deficiency (SLC22A5)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P076)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Carpenter Syndrom Typ2 (MEGF8)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Carpenter-Syndrom (RAB23)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Catel-Manzke Syndrom/Desbuqois dysplasia (CANT1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Catel-Manzke-Syndrom (IMPAD1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
CBAVD (Congenital Bilateral Aplasia of Vas Deferens)(CFTR-Gen)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Cenani-Lenz-Syndaktylie Syndrom (LRP4)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Cerebral cavernous malformation (CCM) (KRIT1, CCM2, PDC10)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P130, P131)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Cerebrale Amyloidangiopathie (CAA) (CST3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Cerebrotendinous xanthomatosis (CYP27A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Charcot-Marie-Tooth - Syndrom (CMT1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P033)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
CHARGE-Syndrom (CHD7, SEMA3E)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P201)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Chondrocalcinose 2 (ANKH)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Chondrodysplasia punctata AR (PEX7, GNPAT, AGPS)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Chondrodysplasia punctata XD (EBP)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Chondrodysplasie punctata XR (ARSE)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Chorea, hereditary benign (NKX2-1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Chylomicron retention disease; CMRD (SAR1B)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Ciliopathien (DNAI1, DNAH5, DNAH11, RPGR, RSPH4A, RSPH9, CCDC40, CCDC39)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
CK-Syndrom (CHILD-Syndrom) (NSDHL)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Coffin-Lowry-Syndrom (RPS6KA3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Cold-induced sweating syndrome (CLCF1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Cornelia de Lange-Syndrom 1 (CDLS1) (NIPBL)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P141, P142)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Cowden-Syndrom (PTEN)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Cystische Fibrose (CFTR): Herkunftsland (Ethnie)	genomische DNA	PCR, Fragmentanalyse, MLPA (P091)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Fragmentanalyse am ABI3500 (VA) Version: 006/01.2020, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	
DFNB1A (GJB2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P163)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
DFNB1B (GJB6)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P163)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
DFNB4 (SLC26A4)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P280)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Diamond-Blackfan Anämie (RPS19, RPL15, RPL5, RPL26, DBA6, TSR2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P212)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Diastrophe Dysplasie (SLC26A2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Diastrophie Dysplasie (DTDST)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Dihydopyrimidinasemangel (DPYS)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Duane-radial ray syndrome (SALL4)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Ehlers-Danlos Syndrom Typ IV AD (COL3A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Ehlers-Danlos-Syndrom (PLOD1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Ellis-van-Creveld-Syndrom (EvC/EvC2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Empty follicle syndrome (EFS) (LHCGR)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Epilepsie, generalisiert mit Fieberkrämpfen plus, Typ 2(SCN1A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P137)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Epilepsien (AMT, ARX, CDKL5, GCSH, GLDC, KCNQ2, SCN2A, SLC25A22, STXBP1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Epilepsy, progressive myoclonic, 4, with or without renal failure; EPM4 (SCARB2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Epileptische Enzephalopathie (SLC25A22, ALDH7A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Episodische Ataxie Typ 2 (CACNA1A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P279)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Familiäre Adenomatöse Polyposis (FAP2) (APC, MUTYH)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P043)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Familiäre Hypokalziurische Hyperkalzämie (FHH) (CASR)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P177)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Familiäres Meduläres Schilddrüsen-Karzinom (FMTC) und MEN Syndrome (RET, MEN1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P015)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Fetale Akinesie (CHRNA1, CHRNB1, CHRND, RASPN1, SYN1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Fetale Herzrythmusstörungen (Arrhythmien) (AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NOS1AP, SCN4B, SCN5A, SNTA)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Floppy Infant Syndrom (ACTA1, AGRN, ALG2, BIN1, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Fraser-Syndrom (FRAS1, FREM2,GRIP)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Frühkindliche X-gebundene Epilepsie mit geistiger Behinderung (PCDH19)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P330)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Fruktose-Intoleranz (ALDOB, FBP1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
G6PD-Mangel (Favismus) (G6PD)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Galaktosämie (GALT)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P156)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Gastrointestinale und weitere Tumorerkrankungen (KIT, MAX, MEN1, NF1, PRKAR1A, SDHAF2, SDHB, SDHC, SDHD, SMARCB1, TMEM127)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Gilbert Syndrom (Morbus Meulengracht) (UGT1A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Glass Syndrom (SATB2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
GLI3-assoziierte Krankheitsbilder (GLI3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P179)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Glykogenose Typ 5/ Morbus Mc Ardle (PYGM)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Glykogenose Typ1A (G6PC)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Glykogen-Speicherkrankheit durch hepatischen Glykogensynthase-Mangel (GYS2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Gorlin-Goltz Syndrom (PTCH1, PTCH2, SUFU)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P067)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Großwuchs-Syndrom (CDKN1C, DIS3L2, DNMT3A, EZH2, GPC3, NFIX, NSD1, OFD1 und SHANK3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hämochromatose (HFE)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P347)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Hämolytische Anämie (CD59)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hämophillie (Faktor 7, Faktor 8, Faktor 9, Faktor 11)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P207)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Hereditäre Angioödeme (SERPING1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P243-A3, P243-B1)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Hereditäre Angioödeme Typ 3 (HAE 3) (F12)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hereditäre Lymphödeme (LMPH1A) (VEGFC, FLT4)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hereditäres Magenkarzinom (CDH1, CTNNA1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P083)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Hereditäres nicht-polypöses kolorektales Karzinom (HNPCC-Diagnostik) (MLH1, MSH2, MSH6, PMS2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P003, P072, P008)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Heterotaxie (ACVR2B, CCDC39, CCDC40, CFC1 , CITED2, CRELD1 , DNAAF1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hirnfehlbildungen (AP1S2, CDKN1C, FOXC1, LAMC1, NID1, ZIC1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Holocarboxylase Synthetase Defizienz (HLCS)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Holoprosenzephalie (SHH, ZIC2, SIX3, TGIF1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P187)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Holt-Oram-Syndrom (TBX5)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P180)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Homozystinurie/CBS-Defizienz (CBS)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hydrozephalus (X-linked) (L1CAM)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hyperekplexie (SLC6A5, GLRB, GPHN)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hyperinsulinismus (KCNJ11, ABCC8)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P117)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Hyperoxalurie (AGXT)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hypertrophe Kardiomyopathie (ALPK3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Hypochondroplasie (FGFR3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hypokalziurische Hyperkalzämie (Typ II) (GNA11)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hypophosphatasie (ALPL)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hypophyseninsuffizienz (POU1F1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Hypothyreose (IGSF1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Idiopathische infantile Hyperkalzämie (CYP24A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Immundefizienz (CARD9)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Intelectual Disability (ABCD1, ACBD6, ACSL4, ACY1, ADCK3, ADK, ADRA2B, ADSL, AFF2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Intrakranielle Hämorrhagie (COL4A1, COL4A2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Intrauterine Wachstumsretardierung: OBSL1, CCDC8, CDKN1C, CUL7, DHCR7, FGFR3, PIK3R1, RNU4ATAC, SHOX, SRCAP und TRIM37	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Isolierte Brachydaktylie der Finger IV/V (BMPR1B, IHH, GDF5, NOG, ROR2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Isolierte Wachstumsfaktor Defizienz TYP 1A und 1B (GH1, GHRHR)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P216)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Jeune Syndrom (WDR34)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Joubert Syndrom 12/Acrocallosal Syndrom (KIF7)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Juveniles Parkinson Syndrom (PARK1,PARK2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P051, P052)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Kabuki-Syndrom (MLL2, KDM6A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P389, P445)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Kapilläre Fehlbildung-arteriovenöse Fehlbildung (RASA1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P409)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Kardiomyopathien (ABCC9, ACTC1, ACTN2, BAG3, CSRP3, DES, DMD, DSG2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Kartagener Syndrom (DNAH5/DNAI1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P238)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Katarakt 1, multiple types (GJA8)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kingsmore Panel Diagnostik (552 Gene)	genomische DNA	NGS	Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	NextSeq550 (Illumina)		NGS
Kleefstra-Syndrom (EHMT1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kleidocraniale Dysplasie (CCD)(RUNX2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P080)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Kleinwuchs (AKT1, ANKH, ANO5, ARSE, COL10A1, COMP, IGF1, IGF1R, IGF2, IGFALS, IHH, SOX9, SHOX)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Klippel Feil-Syndrom (GDF3, GDF6, PAX1, MEOX1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kollagenopathie Typ II (COL2A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kongenitale Fasertypdisproportion (CFTD) (MYH7)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kongenitale Herzerkrankungen (Panel: NKX2.5, GATA4, GJA1, FOXP1, NOTCH1, NOTCH2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kongenitaler Klumpfuss; CCF (PITX1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kongenitales myasthenisches Syndrom /CMS (MUSK,CHAT, CHRNE, COLQ, GFPT1, CHRNA1, CHRNB1, CHRND)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kongenitales zentrales Hypoventilationssyndrom (CCHS) (PHOX2B)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Kraniosynostose Typ II (MSX-2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kraniosynostose Typ III (TCF12)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Kurzrippen-Polydaktylie-Syndrom (EvC1, EvC2, IFT80, WDR19)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Langketten 3 hydroxyacyl CoA Dehydrogenase (LCHAD) Defizienz (HADHA)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Leigh Syndrom/ Paragangliomas 5 (SDHA)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Leri-Weill Dyschondrosteose LWD / Kleinwuchs (SHOX)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P018)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Leukodystrophie (TUBB4a, ABCD1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P049)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Leukodystrophien: (ABCD1, ALDH3A2, ARSA, DGUOK, HSD17B4, SLC16A2, SLC17A5,SPG20)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Li-Fraumeni-Syndrom(TP53, CHEK2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Limb-Pelvis-Syndrom (WNT7A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Lippen-Kiefer-Gaumenspalte (MSX1, IRF6,TBX22)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P304)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Lissenzephalie / subkortikale Bandheterotopie/ Double-cortex-Syndrom (LIS1, DCX, TUBA1A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Long QT Syndrom (KCNQ1, KCNH2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P114)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Lowe-Syndrom (OCRL)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Lymphangiom (AA) (PGM1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Makrozephalie (BRWD3, CUL4B, FMR1, KIF7, RAB39B)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Marfan Syndrom (FBN1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P65,P66)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Marshall Smith-Syndrom (NFIX)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
MCAD-Defizienz (ACADM)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
McKusick-Kaufmann-Syndrom/ Bardet-Biedl-Syndrom6 (MKKS)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Meckel-Gruber Syndrom (TMEM216, MKS1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrom (MMIH), Berdon Syndrom (ACTG2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Mehrlinge monozygot / dizygot, Kontaminationstest	genomische DNA	PCR, Fragmentanalyse	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Fragmentanalyse am ABI3500 (VA) Version: 006/01.2020	Genetic Analyzer ABI3500		PCR
Mental retardation, autosomal dominant 40; MRD40 (CHAMP1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Metachromatic Leukodystrophy due to saposin B deficiency (PSAP)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Metachromatische Leukodystrophie (ARSA)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Mikrozephalie (ASPM, CDK5RAP2, CDK6, CENPE, CENPJ, CEP135, CEP152, KNL1, MCPH1, MFSD2A, PCNT, PHC1, SASS6, STIL, WDR62, ZNF335)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Miller Syndrom (DHODH)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Möbius Syndrom (REV3L)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
MODY Syndrom Typ 1 und 3 (HNF1A, HNF4A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P241)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Morbus Crouzon (FGFR2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Morbus Krabbe (GALC)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P446)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Morbus Muenke (FGFR3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Morbus Pfeiffer (FGFR1, FGFR2, FGFR3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Morbus Wilson (ATP7B)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P098)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
MTDPS4B und PEOA1 (POLG1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Mukopolysaccharidosis TypIVB (GLB1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Multiple Epiphysäre Dysplasie Typ I / Pseudoachondroplasie (COMP)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Multiple Epiphysäre Dysplasie Typ V (MATN3, COL2A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Multiple kartilaginäre Exostosen Typ 1 und 2 (EXT1, EXT2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P228)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Muskeldystrophie Duchenne/Becker (DMD)	genomische DNA	MLPA (P034, P035)	MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500	ja	
Muskeldystrophie Typ Fukuyama (FCMD) (FKTN)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Nagel-Patella-Syndrom (LMX1b)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Nager-Syndrom (SF3B4)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Nephronophthise 1 (NPHP1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P387)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Nephrotisches Syndrom: ACTN4, CD2AP, COQ6, INF2, LAMB2, LMX1B, NPHS1, NPHS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, WT1	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Netherton-Syndrom (SPINK5)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Neurodegeneration mit Eisenablagerung im Gehirn (c19orf12)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Neurofibromatose (NF1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P081, P082)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Neuromuskuläre Erkrankungen (ACTA1, ANO5, BAG3, BIN1, CAPN3, MYH7, MYOT, RYR1, SEPN1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Nicht-autoimmune Hypertyreose (TSHR)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Nierenerkrankungen (EYA1, HNF1B, PAX2, PKD2, PKHD1, SIX5, UMOD)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Nierenzysten und Diabetes-Syndrom (HNF1B)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Nocturnal frontal lobe epilepsy (ENFL) (CHRNA2, CHRNA4, CHRNB2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Noonan-Syndrom (PTPN11, SOS1, RAF1, KRAS, RIT1, BRAF)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Opsismodysplasie (INPPL1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Ostegenesis imperfecta Typ IX (PPIB)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Osteogenesis imperfecta 17 (SPARC)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Osteogenesis imperfecta Typ IIb / VII (CRTAP)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Osteogenesis imperfecta Typ I-IV (COL1A1 / COL1A2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P271, P272)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Osteogenesis imperfecta Typ VIII (LEPRE1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Osteogenesis imperfecta TypV (IFITM5)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Osteogenesis imperfecta TypV (TMEM38B)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Osteopathie mit Sklerose (OSCS) (AMER1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
P450 Oxidase Mangel (POR)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Paragangliom (SDHA, SDHB, MEN)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P226)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Parkinson (ADH1C, ATP13A2, ATP1A3, ATP6AP2,MAPT, PARK2 , STXBP1, TAF1, TBP)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Periodische Fiebersyndrome (CARD14, ELANE, IL10, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, MEFV, MVK)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Periventrikuläre noduläre Heterotopien (FLNA)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P061)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Peters-Plus-Syndrom (PPS) (B3GALTL)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Peutz-Jeghers Syndrom (STK11)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P101)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Phenylketonurie (PKU) (PAH)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Pitt-Hopkins-Syndrom (TCF4, NRXN1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P75, P379)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Pituitary Stalk Interruption Syndrome (PSIS) = angeborene Variante des Pickardt-Fahlbusch Syndrom--> Tertiäre Hypothyreose (GPR161)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
PNKD (Paroxysmale nicht-kinesiogene Dyskenesie) Myofibrillogenesis Regulator 1 (PNKD)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Polycystische Lebererkrankung (PLCD) (SEC63, PRKCSH)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Polymikrogyrie (TUBB2B)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Porphyrie (UROS)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P412)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Primäre/vorzeitige Ovarialinsuffizienz /POF (FMR1, BMP15, FSHR)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Protein C-Defizienz (PROC)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P265)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Protein S-Mangel (PROS1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P112)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Pseudoachondroplasie (COMP)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Pseudohypoaldosteronismus Typ1 (PHA1A) (NR3C2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Pyridoxamine 5 -Phosphat –Mangel (PNPO)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Rasopathien: BRAF, CBL, HRAS, MAP2K1, MAP2K2, NF1, NRAS, SPRED1, PTPN11, KRAS, RAF1, SHOC2, SOS1, RIT1	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Renal tubular acidosis with deafness (ATP6V1B1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Restriktive Dermatopathie (ZMPSTE24)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Retinopathie pigmentosa (RP2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Rett-Syndrom (MECP2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P015)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Rolando-Epilepsie (SRPX2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Saethre-Chotzen-Syndrom (FGFR2-3, TWIST)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Schizenzephalie (SIX3, SHH)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Schlaganfall (early onset)(ACE, ALOX5AP, ATP1A2, ATP1A3, CACNA1A, COL4A1, COL4A2, F2, F5, HTRA1, NOS3, NOTCH3, OTC, POLG, PRKCH, SCN1A, SLC2A1, TREX1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Schwere kombinierte Immundefizienz (SCID) (ADA, IL2RG)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Schwerhörigkeit (GJB6, OTOA, OTOF, OTOR, TYR, USH1C, USH1G, USH2A, WFS1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Septo-optische Dysplasie (HESX1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Sex Reversal (SRY)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Shwachman-Diamon Syndrom (SBDS)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Sideroblastische Anämie (GLRX5)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Simpson-Golabi-Behmel-Syndrom Typ1 (GPC3, GPC4, OFD1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P154)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Sinusknotenerkrankungen (HCN4, SCN5A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P108)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Skelettdysplasie (COL10A1, COL9A1, COMP, DHODH, DLL3, EOGT, ESCO2, FGFR1,FGFR2,FGFR3, FIG4, FLNA, FLNB)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Smith-Lemli-Opitz-Syndrom (DHCR7)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P457)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
SOTOS-Syndrom (NSD1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Sotos-Syndrom/Großwuchs (NSD1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P026)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Spastische Paraplegie 4 (SPAST)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Spastische Paraplegien (SPG) (ATL1, BSCL2, HSPD1, KIF5A, NIPA1, REEP1, RTN2, SLC33A1, SPAST, ZFYVE27)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Spinale Muskelatrophie (SMN1)	genomische DNA	MLPA (P460)	MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500	ja	
Spondylocostale Dysosostis Typ 4 (DLL3, MESP2, HES7, LFNG)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Spondylocostale Dysosostis Typ 5 (SCDO5) (TBX6)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P463)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
ß-Thalassämie (HBB)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P102)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Startle-Syndrom (GLRA1, GLRB, GPHN, SLC6A5)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P274)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Stickler-Syndrom (COL2A1, COL11A1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Strukturelle Herzfehler (GJA1, NKX2-5, GATA4, FOXP1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Succinyl-CoA Transferase Defizienz (SCOT) (OXCT1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Suszeptibilität für Mykobakteriosen durch kompletten IFN-gamma-R1-Defekt (IFNGR1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Syndaktylie Typ IV (LMBR1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
TAR-Syndrom (RBM8A)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P297)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Tay-Sachs (HEXA)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P199)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
TDP-glucose 4,6,-Dehydratase (TGDS)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Tetraamelie (WNT3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Thanatophore Dysplasie Typ I (FGFR3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Thanatophore Dysplasie Typ II (FGFR3)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Treacher-Collins-Franceschetti-Syndrom (TCOF1,POLR1C, POLR1D)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul MLPA (P310)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Truncus arteriosus isoliert (PLXND1)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Tuberöse Sklerose (TSC1, TSC2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P124, P046)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Uniparentale Disomie (UPD2, UPD7, UPD11, UPD14, UPD15, UPD16)	genomische DNA	PCR, Fragmentanalyse	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Fragmentanalyse am ABI3500 (VA) Version: 006/01.2020	Genetic Analyzer ABI3500		PCR
Von Hippel Lindau Syndrom (VHL)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P016)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja	SEQ , NGS
Von Willebrand Jürgens Syndrom (VWF)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA(P11, P12)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	nein	SEQ , NGS
Webb-Dattani-Syndrom (ARNT2)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
West-Syndrom (FOXG1, MEF2C, STXBP1, CDKL5, ARFGEF2, ARX)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul; MLPA (P075, P189, P395, P138, P015)	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA) Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021, MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018	Genetic Analyzer ABI3500, NextSeq550 (Illumina)	ja (P189, P138, P015)	SEQ , NGS
X-chromosomale mentale Retardierung (ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CCDC22, CDKL5)	genomische DNA	Sanger-Sequenzierung; NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 004/12.2019, Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021	Genetic Analyzer ABI3500, NextSeq550 (Illumina)		SEQ , NGS
Whole Exome Sequencing (WES)	genomische DNA	NGS: Hybrid Capture, Sequencing by Synthesis, JSI medical systems software: NextSeq-Modul	Illumina DNA Prep with Exome 2.0 Plus Enrichment (AA) Version: 001/04.2023, NextSeq2000 (VA) Version: 001/12.2022	NextSeq2000 (Illumina)		NGS

Untersuchungsgebiet: Humangenetik (Zytogenetik)

Untersuchungsart: Chromosomenanalyse**

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Chromosomenregion 22q11.21 (DiGeorge-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit den Vysis DiGeorge Region LSI N25 SpectrumOrange/LSI ARSA SpectrumGreen Probes auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	x	
Chromosomenregion 15q11-13 (Prader-Willi-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit dem Vysis Prader-Willi/Angelman Region SNRPN/CEP 15/PML FISH Probe Kit (CE) auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	x	
Chromosomenregion 15q11-q13 (Angelman-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit dem Vysis Prader-Willi/Angelman Region SNRPN/CEP 15/PML FISH Probe Kit (CE) auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	x	
Chromosomenregion 17p11.2 (Smith-Magenis-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit dem Vysis Smith-Magenis Region LSI SMS SpectrumOrange/LSI RARA SpectrumGreen Probe Set auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	x	
Chromosomenregion 5p15.2 (Cri-du-Chat-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit den Vysis Cri-du-Chat Region Probe - LSI EGR1 SpectrumOrange/ D5S23, D5S721 SpectrumGreen Probes auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	x	

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
Chromosomenregion 4p16.3 (Wolf-Hirschhorn-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit den Vysis Wolf-Hirschhorn Region LSI WHS SpectrumOrange/CEP 4 SpectrumGreen Probes auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop mit Isis, Thermobrite, Zentrifugen	x	
Chromosomenregion 17p13.3 (Miller-Dieker-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit den Vysis Miller-Dieker Region/Isolated Lissencephaly LSI LIS1 SpectrumOrange/RARA SpectrumGreen Probes auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop mit Isis, Thermobrite, Zentrifugen	x	
Chromosomenregion Xp22.3 (Kallmann-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit Vysis Kallmann Region LSI KAL SpectrumOrange/CEP X SpectrumGreen Probes auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop mit Isis, Thermobrite, Zentrifugen	x	
Chromosomenregion 7q11.23 (Williams-Beuren-Syndrom)	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit dem Vysis Williams Region Probe - LSI ELN SpectrumOrange/LSI D7S486, D7S522 SpectrumGreen Probe Kit (CE) auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop mit Isis, Thermobrite, Zentrifugen	x	
Zentromere aller Chromosomen	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit spezifischen Zentromer-Sonden der Firma MetaSystems auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - Version 005/08.2021	Fluoreszenzmikroskop (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	x	
Subtelomere aller Chromosomen	Chromosomenpräparate aller Gewebe (Amnionzellen, Lymphozyten, Chorion- und Plazentazotten, Abortfibroblasten)	Chromosomenanalyse durch FISH mit spezifischen Subtelomer-Sonden der Firma Abbott auf Metaphasen / Zellkernen	VA: Fluoreszenzmikroskopie nach Färbung mit Fluorochromen und markierten DNA-Sonden - 005/08.2021	Fluoreszenzmikroskop (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	x	

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Untersuchungsart: Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren
fetaler Chromosomensatz	EDTA-Blut; cfDNA	nicht-invasive Analyse auf fetale chromosomale Kopienzahlveränderungen sowie copy number variations (CNVs) mittels Next Generation Sequencing (NGS), Hybrid Capture, Sequencing by Synthesis, Bioinformatische Analyse auf chr22q11.2 Deletionssyndrom	M-SOP079 VeriSeq Workflow Version 11, E0072 VeriSeq-NIPT-Solution-v2-Package-insert Version 02 (Dokument-Nr. 1000000078751 v06 DEU) SOP077 Hamilton Version 02 SOP078 Illumina-NextSeq550Dx Version 09 SOP019 Durchfuehrung der Datenanalyse Version 34	Illumina NextSeq 550Dx, Hamilton Microlab STAR, dmap Server	X	
fetales RHD-Gen	EDTA-Blut; cfDNA	nicht-invasive Analyse des fetalen RhD-Status mittels RT-qPCR	SOP104 Nicht-invasive fetale RhD-Bestimmung Version 04, E0102 FetoGnost-Kit-RHD-Gebrauchsanweisung_2021-05 Version 01, SOP106 QuantStudio 7 Pro Version02	QuantStudio 7 Pro	X	