Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Untersuchungsart: Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren	Analyt in der Anlage zur Akkreditierungsurkunde D-ML-21655-01 aufgeführt
Asplenie (RPSA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
3-MGA Aciduria, Typ VII, with cataracts, neurologic involvement and neutropenia (CLPB)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
3-MGA-aciduria Typ iV (SERAC1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
3-M-Syndrom 1 (CUL7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
3-M-Syndrom 2 (OBSL1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
3-M-Syndrom 3 (CCDC8)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Achondrogenesis Typ 1A, Typ 1B, Typ 2 (TRIP11, SLC26A2, COL2A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Achondrogenesis Typ iB (DTDST)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Achondrogenesis Typ II (COL2A1),	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Achondroplasie (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Acromesomelic Dysplasia, Maroteaux Type (AMDM) (NPR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Adipositas: Gen-Panel (8 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Adrenogenitales Syndrom (CYP21A2, CYP11B1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
AGAT- Defizienz (GATM)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Alagille-Syndrom (JAG1, NOTCH2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	

Albinismus (OCA1, OCA2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Allan-Herndon-Dudley-Syndrom; AHDS (SLC16A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Alpa1-Antitrypsin-Mangel A1-AT (SERPINA1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Alpha-Methylacetoacetic Aciduria (ACAT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Alport-Syndrom (COL4A3, COL4A4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Alveolokapilläre Dysplasie (FOXF1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Amelogenesis Imperfecta: Gen-Panel (9 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Anämie: Diamond-Blackfan-Anämie Gen-Panel (13 Gene), Sideroblastische Anämie Gen-Panel (302 Gene),	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Androgen-Resistenz (AR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	

Anemie und Spinocerebrale Ataxie (ABCB7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Angeborene Glykosylierungsstörungen – CDG-Syndrome Gen-Panel (107 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Angelman-Syndrom (UBE3A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Antithrombin-III-Mangel (SERPINC1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Apert-Syndrom (FGFR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
ARC Syndrom (Arthrogryposis, renal dysfunction and cholestasis) (VIPAS39)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Argininosuccinase Mangel (ASL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Aromatase Defizienz (CYP19A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Arrhythmogene rechtsventrikuläre Kardiomyopathie (TMEM43, JUP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Ataxie (APTX)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Atelosteogenesis I –III (FLNB, SLC26A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Augenerkrankungen: Morbus-Stargardt Gen-Panel (6 Gene), Kongenitale extraokuläre Muskelfibrose (Kongenitale Ophthalmoplegie) Gen-Panel (4 Gene), Retinitis pigmentosa Gen-Panel (8 Gene), Glaukom Gen-Panel (13 Gene), Lebersche kongenitale Amaurose (LCA) Gen-Panel (22 Gene), Netzhautdystrophie Gen-Panel (216 Gene),	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Autismus-Spektrum-Störungen: Gen-Panel (101 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Autosomal-dominante polyzystische Nierenerkrankung/ ADPKD (PKD1 und PKD2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Autosomal-rezessive polyzystische Nierenerkrankung/ ARPKD (PKHD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Azoospermiefaktor (SY84, SY86, SY127, SY134, SY254, SY255, SY160, SYCP3, SRY)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Balkenagenesie (Agenesie des Corpus callosum) (Gen-Panel, 44 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Baraitser-Winter- Syndrom (ACTB, ACTG1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Bardet-Biedl-Syndrom (BBS1, BBS2, BBS10)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Batten Disease/Ceroid Lipofuscinosis Neuronal 3 (CLN3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Beckwith-Wiedemann Syndrom (Deletionsanalyse)	genomische DNA	MS-MLPA	**MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018**	Genetic Analyzer ABI3500	MS-MLPA		X
Bindegewebsschwäche/here-ditäre Gefäßerkrankungen: Loeys-Dietz-Syndrom Gen-Panel (7 Gene), Ehlers-Danlos-Syndrom Gen-Panel (3 Gene), Marfan-Syndrom Gen-Panel (1 Gen)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Biotinidase Mangel (BTD)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Biotin-responsive Basalganglienerkrankung (BBGD) (SLC19A3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Blepharophimosis-Epicanthus-inversus-Syndrom BPES (FOXL2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Borjeson-Forssman-Lehmann Syndrom (PHF6)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Brachydactyly type E (PTHLH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Brachydactyly-Syndactyly Syndrom (HOXD13)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Branchio-oto-renales Syndrom (Bor-Syndrom) (EYA1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Brown-Vialetto-van-Laere-Syndrom (SLC52A3, SLC52A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Brust- und Eierstockkrebs (BRCA Diagnostik) Gen Panel (18 Gene), Brust- und Eierstockkrebs (erweitertes NGS Tumor Panel) Gen-Panel (93 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Campomele Dysplasie (SOX9)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Carney complex, type 1; CNC1 (PRKAR1A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Carnitine deficiency (SLC22A5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Carpenter Syndrom Typ2 (MEGF8)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Carpenter-Syndrom (RAB23)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Catel-Manzke Syndrom/Desbuqois dysplasia (CANT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Catel-Manzke-Syndrom (IMPAD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
CBAVD (Congenital Bilateral Aplasia of Vas Deferens)(CFTR-Gen)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Cenani-Lenz-Syndaktylie Syndrom (LRP4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Cerebral cavernous malformation (CCM) (KRIT1, CCM2, PDC10)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Cerebrale Amyloidangiopathie (CAA) (CST3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Cerebrotendinous xanthomatosis (CYP27A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Charcot-Marie-Tooth -Syndrom (CMT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	

CHARGE-Syndrom (CHD7, SEMA3E)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Chondrocalcinose 2 (ANKH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Chondrodysplasia punctata AR (PEX7, GNPAT, AGPS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Chondrodysplasia punctata XD (EBP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Chondrodysplasie punctata XR (ARSE)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Chorea, hereditary benign (NKX2-1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Chronisch entzündliche Darmerkrankungen (Morbus Crohn), Gen-Panel (62 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	

Chylomicron retention disease; CMRD (SAR1B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
CiliopathienGen-Panel (Gen-Panel, 138 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
CK-Syndrom (CHILD-Syndrom) (NSDHL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Coffin-Lowry-Syndrom (RPS6KA3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Cold-induced sweating syndrome (CLCF1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Cornelia de Lange-Syndrom 1 (CDLS1) (NIPBL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Cowden-Syndrom (PTEN)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Cystische Fibrose (CFTR): Herkunftsland (Ethnie)	genomische DNA	**PCR, Fragmentanalyse, MLPA**	**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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA, Fragmentanalyse		

DFNB1A (GJB2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
DFNB1B (GJB6)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
DFNB4 (SLC26A4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Diastrophe Dysplasie (SLC26A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Diastrophie Dysplasie (DTDST)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Dihydopyrimidinasemangel (DPYS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Duane-radial ray syndrome (SALL4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Ellis-van-Creveld-Syndrom (EvC/EvC2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Empty follicle syndrome (EFS) (LHCGR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Epilepsie, generalisiert mit Fieberkrämpfen plus, Typ 2(SCN1A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Epilepsien: Funkindliche epileptische Enzephalopathie Gen-Panel (9 Gene), Epileptische Enzephalopathie Gen-Panel (16 Gene), Familiäre nächtliche Frontallappenepilepsie (ADNFLE) Gen-Panel (3 Gene), Benigne familiäre Neugeborenenkrämpfe (BFNS) Gen-Panel (2 Gene), Generalisierte	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Epilepsy, progressive myoclonic, 4, with or without renal failure; EPM4 (SCARB2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Epileptische Enzephalopathie (SLC25A22, ALDH7A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Episodische Ataxie Typ 2 (CACNA1A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Extremitätenfehlbildung (Gen-Panel, 47 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Familiäre Adenomatöse Polyposis (FAP2) (APC, MUTYH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Familiäre Hypokalziurische Hyperkalzämie (FHH) (CASR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	

Familiäres Meduläres Schilddrüsen-Karzinom (FMTC) und MEN Syndrome (RET, MEN1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Fetale Akinesie (Gen-Panel, 412 Gene; >25kb)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Fetale Herzrythmusstörungen (Arrhythmien): Long-QT-Syndrom (LQTS) Gen Panel (15 Gene), Short-QT-Syndrom (LQTS) Gen Panel (3 Gene), Brugada-Syndrom (BrS) Gen Panel (17 Gene), Sick-Sinus-Syndrom Gen Panel (3 Gene), Katecholaminerge polymorphe ventrikuläre Tachykardie (CPVT) Gen Panel (3 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Floppy Infant Syndrom: Gen-Panel (58 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Fraser-Syndrom (FRAS1, FREM2,GRIP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Frühkindliche X-gebundene Epilepsie mit geistiger Behinderung (PCDH19)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Fruktose-Intoleranz (ALDOB, FBP1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
G6PD-Mangel (Favismus) (G6PD)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Galaktosämie (GALT)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Gastrointestinale und weitere Tumorerkrankungen: Pankreaskarzinom Gen-Panel (16 Gene), Kolonkarzinom Gen-Panel (14 Gene), Magenkarzinom Gen-Panel (10 Gene), Gastrointestinaler Stromatumor (GIST) Gen-Panel (11	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Geschlechtsdifferenzierungs-störung (Gen-Panel, 21 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Gilbert Syndrom (Morbus Meulengracht) (UGT1A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Glass Syndrom (SATB2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
GLI3-assoziierte Krankheitsbilder (GLI3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Glykogenose Typ 5/ Morbus Mc Ardle (PYGM)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Glykogenose Typ1A (G6PC)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Glykogen-Speicherkrankheit durch hepatischen Glykogensynthase-Mangel (GYS2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Gorlin-Goltz Syndrom (PTCH1, PTCH2, SUFU)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Großwuchs-Syndrom: Gen-Panel (18 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Hämochromatose (HFE)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Hämolytische Anämie (CD59)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hämophillie (Faktor 7, Faktor 8, Faktor 9, Faktor 11)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Hereditäre Angioödeme (SERPING1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Hereditäre Angioödeme Typ 3 (HAE 3) (F12)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hereditäre Lymphödeme (LMPH1A), Gen-Panel (29 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hereditäre Pankreatitis, Gen-Panel (13 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	

Hereditäres Magenkarzinom (CDH1, CTNNA1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Hereditäres nicht-polypöses kolorektales Karzinom (HNPCC-Diagnostik): MLH1, MSH2, MSH6, PMS2 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Hereditäres Paragangliom/Phäochromozytom (PGL/PCC): Gen-Panel (23 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Heterotaxie (Gen-Panel, 22 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hirnfehlbildungen: Balkenagenesie (Corpus-callosum-Agenesie) Gen-Panel (44 Gene), Dandy Walker Malformation Gen-Panel (7 Gene), Encephalozele Gen-Panel (6 Gene), Holoprosencephalie Gen-Panel (9 Gene), Hydrocephalus Gen-Panel (11	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Holocarboxylase Synthetase Defizienz (HLCS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Holt-Oram-Syndrom (TBX5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Homozystinurie/CBS-Defizienz (CBS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hydrops fetalis: Gen-Panel (145 Gene; >25kb)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Hypercholesterinämie: Gen-Panel (10 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Hyperekplexie (SLC6A5, GLRB, GPHN)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hyper-IgE-Syndrom: Gen-Panel (5 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Hyperinsulinismus (KCNJ11, ABCC8)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Hyperoxalurie (AGXT)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hypochondroplasie (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Hypokalziurische Hyperkalzämie (Typ II) (GNA11)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hypophosphatämie: Gen-Panel (10 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Hypophosphatasie (ALPL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hypophyseninsuffizienz (POU1F1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Hypopituitarismus Gen-Panel (19 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Hypothyreose Gen-Panel (17 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Idiopathische infantile Hyperkalzämie (CYP24A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Immundefizienz/Immundefekt, Allgemeine variable Immundefizienz (CVID) Gen-Panel (14 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Intelectual Disability : ABCD1, ACBD6, ACSL4, ACY1, ADCK3, ADK, ADRA2B, ADSL, AFF2 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Interstitielle Lungenerkrankungen im Kindesalter / diffuse parenchymatische Lungenerkrankungen, Gen-Panel (12 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Intrakranielle Hämorrhagie (COL4A1, COL4A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Intrauterine Wachstumsretardierung und Kleinwuchs Gen-Panel (158 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Isolierte Brachydaktylie der Finger IV/V (BMPR1B, IHH, GDF5, NOG, ROR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Isolierte Wachstumsfaktor Defizienz TYP 1A und 1B (GH1, GHRHR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	

Jeune Syndrom (WDR34)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Joubert Syndrom 12/Acrocallosal Syndrom (KIF7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Juveniles Parkinson Syndrom (PARK2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Kabuki-Syndrom (MLL2, KDM6A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Kapilläre Fehlbildung-arteriovenöse Fehlbildung (RASA1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Kardiomyopathien: ABCC9, ACTC1, ACTN2, BAG3, CSRP3, DES, DMD, DSG2 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Kardiomyopathien: Hypertrophe Kardiomyopathie (HCM) Gen Panel (25 Gene), Dilatative Kardiomyopathie (DCM) Gen Panel (35 Gene), Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD) Gen Panel (8 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Kartagener Syndrom (DNAH5/DNAI1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Katarakt 1, multiple types (GJA8)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kingsmore Panel Diagnostik (552 Gene)	genomische DNA	**NGS Target Enrichment by Hybrid Capture**	**Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021**	**NextSeq550 (Sequencing by Synthesis (SBS)“ , Illumina)**		**NGS**	
Kleefstra-Syndrom (EHMT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kleidocraniale Dysplasie (CCD)(RUNX2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Kleinwuchs: AKT1, ANKH, ANO5, ARSE, COL10A1, COMP, IGF1, IGF1R, IGF2, IGFALS, IHH, SOX9, SHOX und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Klinische Exome und Trio Analysen (5230 Gene; Agilent; Custom Constitutional Panel)	genomische DNA	**NGS Target Enrichment by Hybrid Capture**	**Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021**	**NextSeq550 (Sequencing by Synthesis (SBS)“ , Illumina)**		**NGS**	
Klippel Feil-Syndrom (GDF3, GDF6, PAX1, MEOX1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kollagenopathie Typ II (COL2A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Kombinierter schwerer Immundefekt (Severe combined immunodeficiency; SCID) Gen-Panel (25 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kongenitale Fasertypdisproportion (CFTD) (MYH7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kongenitale Herzerkrankungen (Panel: NKX2.5, GATA4, GJA1, FOXP1, NOTCH1, NOTCH2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kongenitaler Klumpfuss; CCF (PITX1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kongenitales myasthenisches Syndrom /CMS (MUSK,CHAT, CHRNE, COLQ, GFPT1, CHRNA1, CHRNB1, CHRND)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kongenitales zentrales Hypoventilationssyndrom (CCHS) (PHOX2B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kraniosynostose Typ II (MSX-2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Kraniosynostose Typ III (TCF12)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Kurzrippen-Polydaktylie-Syndrom (EvC1, EvC2, IFT80, WDR19)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Langketten 3 hydroxyacyl CoA Dehydrogenase (LCHAD) Defizienz (HADHA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Leigh Syndrom/ Paragangliomas 5 (SDHA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Leri-Weill Dyschondrosteose LWD / Kleinwuchs (SHOX)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Leukodystrophie (TUBB4a, ABCD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Leukodystrophien: ABCD1, ALDH3A2, ARSA, DGUOK, HSD17B4, SLC16A2, SLC17A5,SPG20 und weitere Gene (135 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Li-Fraumeni-Syndrom(TP53, CHEK2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Limb-Pelvis-Syndrom (WNT7A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Lippen-Kiefer-Gaumenspalte (MSX1, IRF6,TBX22)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Long QT Syndrom (KCNQ1, KCNH2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Lowe-Syndrom (OCRL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Lymphangiom (AA) (PGM1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Marshall Smith-Syndrom (NFIX)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
MCAD-Defizienz (ACADM)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
McKusick-Kaufmann-Syndrom/ Bardet-Biedl-Syndrom6 (MKKS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Meckel-Gruber Syndrom (TMEM216, MKS1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrom (MMIH), Berdon Syndrom (ACTG2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , 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	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Metachromatic Leukodystrophy due to saposin B deficiency (PSAP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Metachromatische Leukodystrophie (ARSA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Miller Syndrom (DHODH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Mitochondriale Erkrankungen (Mitochondriopathien), Mitochondrial kodierte Erkrankungen (Gen-Panel: 37 mitochondrial kodierte Gene), Zugehörige Erkrankungen und Syndrome	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Möbius Syndrom (REV3L)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Monogener Diabetes (MODY), MODY Core Gen-Panel (14 Gene), Monogener Diabetes Gen-Panel erweitert (50 Gene), Diabetes Gen-Panel Comprehensive (79 Gene),	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Morbus Crouzon (FGFR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Morbus Krabbe (GALC)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Morbus Muenke (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Morbus Pfeiffer (FGFR1, FGFR2, FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Morbus Wilson (ATP7B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
MTDPS4B und PEOA1 (POLG1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Mukopolysaccharidosis TypIVB (GLB1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Multiple Epiphysäre Dysplasie Typ I / Pseudoachondroplasie (COMP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Multiple Epiphysäre Dysplasie Typ V (MATN3, COL2A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Multiple kartilaginäre Exostosen Typ 1 und 2 (EXT1, EXT2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	

Muskeldystrophie Duchenne/Becker (DMD)	genomische DNA	MLPA	**MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018**	Genetic Analyzer ABI3500	MLPA		
Muskeldystrophie Typ Fukuyama (FCMD) (FKTN)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Nagel-Patella-Syndrom (LMX1b)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Nager-Syndrom (SF3B4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Nephronophthise 1 (NPHP1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Nephrotisches Syndrom: ACTN4, CD2AP, COQ6, INF2, LAMB2, LMX1B, NPHS1, NPHS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, WT1, Gen-Panel (14 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Netherton-Syndrom (SPINK5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Neurodegeneration mit Eisenablagerung im Gehirn (c19orf12)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Neurofibromatose (NF1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	

Neuromuskuläre Erkrankungen : Myopathie-Panel Gen-Panel (34 Gene), Neuromuskuläre Erkrankungen-Panel Gen-Panel (373 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
NGS Whole Exom und Trio Analysen (23.585 Gene)	genomische DNA	**NGS Target Enrichment by Hybrid Capture**	**Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021**	**NextSeq550 (Sequencing by Synthesis (SBS)“ , Illumina)**		**NGS**	
Nicht-autoimmune Hypertyreose (TSHR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Nierenerkrankungen: Gen-Panel (108 Gene), Nierenerkrankungen und Ciliopathien Gen-Panel (513 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Nierenzysten und Diabetes-Syndrom (HNF1B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Nocturnal frontal lobe epilepsy (ENFL) (CHRNA2, CHRNA4, CHRNB2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Noonan-Syndrom (PTPN11, SOS1, RAF1, KRAS, RIT1, BRAF)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	X
Opsismodysplasie (INPPL1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Ostegenesis imperfecta Typ IX (PPIB)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Osteogenesis imperfecta 17 (SPARC)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Osteogenesis imperfecta Typ IIb / VII (CRTAP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Osteogenesis imperfecta Typ I-IV (COL1A1 / COL1A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Osteogenesis imperfecta Typ VIII (LEPRE1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Osteogenesis imperfecta TypV (IFITM5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Osteogenesis imperfecta TypV (TMEM38B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Osteopathie mit Sklerose (OSCS) (AMER1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
P450 Oxidase Mangel (POR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Parkinson: Gen-Panel (65 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	

Periodische Fieber Syndrome Gen-Panel (21 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Periventrikuläre noduläre Heterotopien (FLNA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Peters-Plus-Syndrom (PPS) (B3GALTL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Peutz-Jeghers Syndrom (STK11)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Phenylketonurie (PKU) (PAH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Pitt-Hopkins-Syndrom (TCF4, NRXN1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Pituitary Stalk Interruption Syndrome (PSIS) = angeborene Variante des Pickardt-Fahlbusch Syndrom--> Tertiäre Hypothyreose (GPR161)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
PNKD (Paroxysmale nicht-kinesiogene Dyskenesie) Myofibrillogenesis Regulator 1 (PNKD)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Polycystische Lebererkrankung (PLCD) (SEC63, PRKCSH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Polymikrogyrie (TUBB2B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	X
Porphyrie Gen-Panel (9 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Primäre Immundefizienz (Primary immunodeficiency) Gen-Panel (408 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Primäre/vorzeitige Ovarialinsuffizienz /POF (FMR1, BMP15, FSHR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Progressive familiäre intrahepatische Cholestase Gen-Panel (90 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Protein C-Defizienz (PROC)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Protein S-Mangel (PROS1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Pseudoachondroplasie (COMP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Pseudohypoaldosteronismus Typ1 (PHA1A) (NR3C2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Pyridoxamine 5 -Phosphat –Mangel (PNPO)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Rasopathien: Gen-Panel (16 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Renal tubular acidosis with deafness (ATP6V1B1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Restriktive Dermatopathie (ZMPSTE24)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Rett-Syndrom (MECP2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Rolando-Epilepsie (SRPX2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Saethre-Chotzen-Syndrom (FGFR2-3, TWIST)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Schlaganfall (early onset): Gen-Panel (33 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Schwerhörigkeit: Gen-Panel (169 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	

Septo-optische Dysplasie (HESX1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Sex Reversal (SRY)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Shwachman-Diamon Syndrom (SBDS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Simpson-Golabi-Behmel-Syndrom Typ1 (GPC3, GPC4, OFD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Sinusknotenerkrankungen (HCN4, SCN5A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Skelettdysplasie: Skelettdysplasie/IUGR Gen Panel (106 Gene), Skelettdysplasie Gen-Panel (291 Gene; bei komplexen Syndromen mit Skelettbeteiligung)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Smith-Lemli-Opitz-Syndrom (DHCR7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
SMMCI Syndrom Gen-Panel (5 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
SOTOS-Syndrom (NSD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Sotos-Syndrom/Großwuchs (NSD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Spastische Paraplegie 4 (SPAST)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Spastische Paraplegien (SPG): Hereditäre Spastische Paraplegien autosomal dominant Gen-Panel (10 Gene), Hereditäre Spastische Paraplegien autosomal rezessiv Gen-Panel (11 Gene), Hereditäre Spastische Paraplegien X-	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Spinale Muskelatrophie (SMN1)	genomische DNA	**MLPA**	**MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018**	Genetic Analyzer ABI3500	MLPA		X
Spondylocostale Dysostostis Typ 4 (DLL3, MESP2, HES7, LFNG)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Spondylocostale Dysostostis Typ 5 (SCDO5) (TBX6)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
ß-Thalassämie (HBB)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Startle-Syndrom (GLRA1, GLRB, GPHN, SLC6A5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Stickler-Syndrom (COL2A1, COL11A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Stoffwechselstörungen und seltene endokrinologische Erkrankungen (90 Gene)	genomische DNA	**NGS Target Enrichment by Hybrid Capture**	**Agilent SureSelect QXT Target Enrichment-NextSeq550 (AA)Version: 005/04.2020, Magnis Dx NGS Prep System (VA) Version: 001/03.2021**	**NextSeq550 (Sequencing by Synthesis (SBS)“ , Illumina)**		**NGS**	
Strukturelle Herzfehler: Kongenitale Herzerkrankungen Gen-Panel (51 Gene), Erweitertes Herzpanel Gen-Panel (325 Gene),	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Succinyl-CoA Transferase Defizienz (SCOT) (OXCT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Suszeptibilität für Mykobakteriosen durch kompletten IFN-gamma-R1-Defekt (IFNGR1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Syndaktylie Typ IV (LMBR1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
TAR-Syndrom (RBM8A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Tay-Sachs (HEXA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
TDP-glucose 4,6,- Dehydratase (TGDS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Tetraamelie (WNT3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	

Thanatophore Dysplasie Typ I (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Thanatophore Dysplasie Typ II (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Thrombophilie (Faktor-II-Mutation, Faktor-V-Mutation, MTHFR C677T, MTHFR A1298C)	genomische DNA	**Real-time PCR (VA)**	**Real-time PCR (VA)**	7300 PCR Real Time System	Real-time PCR		
Thrombozytose/Thrombozythämie: Thrombozytose Gen-Panel (24 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **NGS**	
Treacher-Collins-Franceschetti-Syndrom (TCOF1,POLR1C, POLR1D)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Truncus arteriosus isoliert (PLXND1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Tuberöse Sklerose (TSC1, TSC2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Tumor Panel (gesamt) Gen-Panel (93 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , 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	

VACTERL Assoziation: VACTERL Gen-Panel (59 Gene)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
Von Hippel Lindau Syndrom (VHL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	MLPA	PCR , SEQ , **NGS**	
Hämophillie (Faktor 7, Faktor 8, Faktor 9, Faktor 11)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Von Willebrand Jürgens Syndrom (VWF)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Katarakt 1, multiple types (GJA8)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Retinopathie pigmentosa (RP2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Startle-Syndrom (GLRA1, GLRB, GPHN, SLC6A5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Ehlers-Danlos-Syndrom (PLOD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Ehlers-Danlos Syndrom Typ IV AD (COL3A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Marfan Syndrom (FBN1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Cystische Fibrose (CFTR): Herkunftsland (Ethnie)	genomische DNA	**PCR, Fragmentanalyse, MLPA**	**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)**	MLPA, Fragmentanalyse		
Netherton-Syndrom (SPINK5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Restriktive Dermatopathie (ZMPSTE24)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Alagille-Syndrom (JAG1, NOTCH2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Borjeson-Forssman-Lehmann Syndrom (PHF6)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Blepharophimosis-Epicanthus-inversus-Syndrom BPES (FOXL2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Branchio-oto-renales Syndrom (Bor-Syndrom) (EYA1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
CK-Syndrom (CHILD-Syndrom) (NSDHL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Coffin-Lowry-Syndrom (RPS6KA3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Cold-induced sweating syndrome (CLCF1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Cornelia de Lange-Syndrom 1 (CDLS1) (NIPBL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Fraser-Syndrom (FRAS1, FREM2,GRIP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Glass Syndrom (SATB2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
GLI3-assoziierte Krankheitsbilder (GLI3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Kabuki-Syndrom (MLL2, KDM6A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Kleefstra-Syndrom (EHMT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Klippel Feil-Syndrom (GDF3, GDF6, PAX1, MEOX1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Lippen-Kiefer-Gaumenspalte (MSX1, IRF6,TBX22)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Lowe-Syndrom (OCRL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Nager-Syndrom (SF3B4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Marshall Smith-Syndrom (NFIX)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Periventrikuläre noduläre Heterotopien (FLNA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Peters-Plus-Syndrom (PPS) (B3GALTL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Pitt-Hopkins-Syndrom (TCF4, NRXN1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
PNKD (Paroxysmale nicht-kinesiogene Dyskenesie) Myofibrillogenesis Regulator 1 (PNKD)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Rett-Syndrom (MECP2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
SOTOS-Syndrom (NSD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Treacher-Collins-Franceschetti-Syndrom (TCOF1,POLR1C, POLR1D)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Webb-Dattani-Syndrom (ARNT2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hypophyseninsuffizienz (POU1F1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Kongenitaler Klumpfuss; CCF (PITX1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Kongenitales myasthenisches Syndrom /CMS (MUSK,CHAT, CHRNE, COLQ, GFPT1, CHRNA1, CHRNB1, CHRND)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Cerebral cavernous malformation (CCM) (KRIT1, CCM2, PDC10)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	

Cerebrale Amyloidangiopathie (CAA) (CST3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hereditäre Angioödeme (SERPING1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Hereditäre Angioödeme Typ 3 (HAE 3) (F12)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hereditäre Lymphödeme (LMPH1A) (VEGFC, FLT4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Lymphangiom (AA) (PGM1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Intrakranielle Hämorrhagie (COL4A1, COL4A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Kapilläre Fehlbildung-arteriovenöse Fehlbildung (RASA1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Angelman-Syndrom (UBE3A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Amish frühkindliches Epilepsie-Syndrom (ST3GAL5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Nocturnal frontal lobe epilepsy (ENFL) (CHRNA2, CHRNA4, CHRNB2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Epilepsie, generalisiert mit Fieberkrämpfen plus, Typ 2(SCN1A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Epileptische Enzephalopathie (SLC25A22, ALDH7A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
West-Syndrom (FOXG1, MEF2C, STXBP1, CDKL5, ARFGEF2, ARX)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Frühkindliche X-gebundene Epilepsie mit geistiger Behinderung (PCDH19)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Rolando-Epilepsie (SRPX2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Epilepsy, progressive myoclonic, 4, with or without renal failure; EPM4 (SCARB2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
DFNB1A (GJB2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
DFNB1B (GJB6)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	

DFNB4 (SLC26A4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Schwere kombinierte Immundefizienz (SCID) (ADA, IL2RG)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Immundefizienz (CARD9)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Suszeptibilität für Mykobakteriosen durch kompletten IFN-gamma-R1-Defekt (IFNGR1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Beckwith-Wiedemann Syndrom (Deletionsanalyse)	genomische DNA	MS-MLPA	**MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018**	Genetic Analyzer ABI3500	MS-MLPA		
Cowden-Syndrom (PTEN)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Simpson-Golabi-Behmel-Syndrom Typ1 (GPC3, GPC4, OFD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Sotos-Syndrom/Großwuchs (NSD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	

ß-Thalassämie (HBB)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Diamond-Blackfan Anämie (RPS19, RPL15, RPL5, RPL26, DBA6, TSR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Hämolytische Anämie (CD59)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Sideroblastische Anämie (GLRX5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Long QT Syndrom (KCNQ1, KCNH2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Sinusknotenerkrankungen (HCN4, SCN5A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Arrhythmogene rechtsventrikuläre Kardiomyopathie (TMEM43, JUP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Brust- und Eierstockkrebs (BRCA-Diagnostik): ATM, BRCA1, BRCA2, CHEK2, CDH1, PALB2, RAD51D und TP53	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Hereditäres nicht-polypöses kolorektales Karzinom (HNPCC-Diagnostik): MLH1, MSH2, MSH6, PMS2 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	

Gastrointestinale und weitere Tumorerkrankungen: KIT, MAX, MEN1, NF1, PRKAR1A, SDHAF2, SDHB, SDHC, SDHD, SMARCB1, TMEM127 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Carney complex, type 1; CNC1 (PRKAR1A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Familiäre Adenomatöse Polyposis (FAP2) (APC, MUTYH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Familiäres Meduläres Schilddrüsen-Karzinom (FMTC) und MEN Syndrome (RET, MEN1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Gorlin-Goltz Syndrom (PTCH1, PTCH2, SUFU)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Hereditäres Magenkarzinom (CDH1, CTNNA1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Li-Fraumeni-Syndrom(TP53, CHEK2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Multiple kartilaginäre Exostosen Typ 1 und 2 (EXT1, EXT2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	

Paragangliom (SDHA, SDHB, MEN)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Peutz-Jeghers Syndrom (STK11)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Tuberöse Sklerose (TSC1, TSC2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Von Hippel Lindau Syndrom (VHL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Asplenie (RPSA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
CHARGE-Syndrom (CHD7, SEMA3E)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Holt-Oram-Syndrom (TBX5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Kongenitale Herzerkrankungen (Panel: NKX2.5, GATA4, GJA1, FOXP1, NOTCH1, NOTCH2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Truncus arteriosus isoliert (PLXND1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Hypertrophe Kardiomyopathie (ALPK3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Baraitser-Winter- Syndrom (ACTB, ACTG1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Holoprosenzephalie (SHH, ZIC2, SIX3, TGIF1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Hydrozephalus (X-linked) (L1CAM)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Lissenzephalie / subkortikale Bandheterotopie/ Double-cortex-Syndrom (LIS1, DCX, TUBA1A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Polymikrogyrie (TUBB2B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Schizenzephalie (SIX3, SHH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Septo-optische Dysplasie (HESX1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Kongenitales zentrales Hypoventilationssyndrom (CCHS) (PHOX2B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , 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**	
Polycystische Lebererkrankung (PLCD) (SEC63, PRKCSH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Chylomicron retention disease; CMRD (SAR1B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , 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	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
MTDPS4B und PEOA1 (POLG1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Alport-Syndrom (COL4A3, COL4A4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Autosomal-dominante polyzystische Nierenerkrankung/ ADPKD (PKD1 und PKD2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	

Autosomal-rezessive polyzystische Nierenerkrankung/ ARPKD (PKHD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
McKusick-Kaufmann-Syndrom/ Bardet-Biedl-Syndrom6 (MKKS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Nierenzysten und Diabetes-Syndrom (HNF1B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrom (MMIH), Berdon Syndrom (ACTG2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Nephronophthise 1 (NPHP1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
ARC Syndrom (Arthrogryposis, renal dysfunction and cholestasis) (VIPAS39)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Anemie und Spinocerebrale Ataxie (ABCB7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Ataxie (APTX)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Batten Disease/Ceroid Lipofuscinosis Neuronal 3 (CLN3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren	
Biotin-responsive Basalganglienerkrankung (BBGD) (SLC19A3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Brown-Vialetto-van-Laere-Syndrom (SLC52A3, SLC52A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Chorea, hereditary benign (NKX2-1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hyperekplexie (SLC6A5, GLRB, GPHN)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Juveniles Parkinson Syndrom (PARK2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Leigh Syndrom/ Paragangliomas 5 (SDHA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Spastische Paraplegie 4 (SPAST)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Neurodegeneration mit Eisenablagerung im Gehirn (c19orf12)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Kongenitale Fasertypdisproportion (CFTD) (MYH7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Muskeldystrophie Duchenne/Becker (DMD)	genomische DNA	MLPA	**MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018**	Genetic Analyzer ABI3500	MLPA		
Muskeldystrophie Typ Fukuyama (FCMD) (FKTN)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Charcot-Marie-Tooth -Syndrom (CMT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Möbius Syndrom (REV3L)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Bardet-Biedl-Syndrom (BBS1, BBS2, BBS10)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Kartagener Syndrom (DNAH5/DNAI1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Meckel-Gruber Syndrom (TMEM216, MKS1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Joubert Syndrom 12/Acrocallosal Syndrom (KIF7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Balkenagenesie (Agenesie des Corpus callosum): AKT3, ARID1A, ARID1B, ARX, B3GALTL, DISC1, DYNC1H1, EOMES, FOXG1, GCSH und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Ciliopathien: DNAI1, DNAH5, DNAH11, RPGR, RSPH4A, RSPH9, CCDC40, CCDC39 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Fetale Akinesie: CHRNA1, CHRNB1, CHRND, RASPN1, SYN1 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Fetale Herzrythmusstörungen (Arrhythmien): AKAP9, ANK2, CACNA1C, CALM1, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NOS1AP, SCN4B, SCN5A, SNTA und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Heterotaxie: ACVR2B, CCDC39, CCDC40, CFC1 , CITED2, CRELD1 , DNAAF1 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hirnfehlbildungen: AP1S2, CDKN1C, FOXC1, LAMC1, NID1, ZIC1 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Intrauterine Wachstumsretardierung: OBSL1, CCDC8, CDKN1C, CUL7, DHCR7, FGFR3, PIK3R1, RNU4ATAC, SHOX, SRCAP und TRIM37	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Mikrozephalie: ASPM, CDK5RAP2, CDK6, CENPE, CENPJ, CEP135, CEP152, KNL1, MCPH1, MFSD2A, PCNT, PHC1, SASS6, STIL, WDR62, ZNF335 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Makrozephalie: BRWD3, CUL4B, FMR1, KIF7, RAB39B und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Nierenerkrankungen: EYA1, HNF1B, PAX2, PKD2, PKHD1, SIX5, UMOD und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Rasopathien: BRAF, CBL, HRAS, MAP2K1, MAP2K2, NF1, NRAS, SPRED1, PTPN11, KRAS, RAF1, SHOC2, SOS1, RIT1	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Skelettdysplasie: COL10A1, COL9A1, COMP, DHODH, DLL3, EOGT, ESCO2, FGFR1,FGFR2,FGFR3, FIG4, FLNA, FLNB und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Strukturelle Herzfehler: GJA1, NKX2-5, GATA4, FOXP1 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Autismus-Spektrum-Störungen: CHD2, CHD8, CTTNBP2, DIP2A, IQGAP3, KCNQ3, LAMC3 und RELN	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Epilepsien: AMT, ARX, CDKL5, GCSH, GLDC, KCNQ2, SCN2A, SLC25A22, STXBP1 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Floppy Infant Syndrom: ACTA1, AGRN, ALG2, BIN1, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	

Großwuchs-Syndrom: CDKN1C, DIS3L2, DNMT3A, EZH2, GPC3, NFIX, NSD1, OFD1 und SHANK3	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Intelectual Disability : ABCD1, ACBD6, ACSL4, ACY1, ADCK3, ADK, ADRA2B, ADSL, AFF2 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Kardiomyopathien: ABCC9, ACTC1, ACTN2, BAG3, CSRP3, DES, DMD, DSG2 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , 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	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Kleinwuchs: AKT1, ANKH, ANO5, ARSE, COL10A1, COMP, IGF1, IGF1R, IGF2, IGFALS, IHH, SOX9, SHOX und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Leukodystrophien: ABCD1, ALDH3A2, ARSA, DGUOK, HSD17B4, SLC16A2, SLC17A5,SPG20 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Nephrotisches Syndrom: ACTN4, CD2AP, COQ6, INF2, LAMB2, LMX1B, NPHS1, NPHS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, WT1	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Neuromuskuläre Erkrankungen : ACTA1, ANO5, BAG3, BIN1, CAPN3, MYH7, MYOT, RYR1, SEPN1 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Parkinson: ADH1C, ATP13A2, ATP1A3, ATP6AP2,MAPT, PARK2 , STXBP1, TAF1, TBP und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	

Periodische Fiebersyndrome: CARD14, ELANE, IL10, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, MEFV, MVK und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , 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 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Spastische Paraplegien (SPG): ATL1, BSCL2, HSPD1, KIF5A, NIPA1, REEP1, RTN2, SLC33A1, SPAST, ZFYVE27 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
X-chromosomale mentale Retardierung: ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CCDC22, CDKL5 und weitere Gene	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	NGS	PCR , SEQ , **NGS**	
Alveolokapilläre Dysplasie (FOXF1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Noonan-Syndrom (PTPN11, SOS1, RAF1, KRAS, RIT1, BRAF)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Neurofibromatose (NF1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Achondrogenesis Typ iB (DTDST)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Achondrogenesis Typ II (COL2A1),	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Achondrogenesis TypI 1A (TRIP11)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Achondroplasie (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Acromesomelic Dysplasia, Maroteaux Type (AMDM) (NPR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Apert-Syndrom (FGFR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Atelosteogenesis I –III (FLNB, SLC26A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Brachydactyly type E (PTHLH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Brachydactyly-Syndactyly Syndrom (HOXD13)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Campomele Dysplasie (SOX9)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Carpenter-Syndrom (RAB23)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Carpenter Syndrom Typ2 (MEGF8)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Catel-Manzke-Syndrom (IMPAD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Catel-Manzke Syndrom/Desbuqois dysplasia (CANT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Cenani-Lenz-Syndaktylie Syndrom (LRP4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Chondrocalcinose 2 (ANKH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Chondrodysplasia punctata AR (PEX7, GNPAT, AGPS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Chondrodysplasia punctata XD (EBP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Chondrodysplasie punctata XR (ARSE)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Diastrophe Dysplasie (SLC26A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Diastrophie Dysplasie (DTDST)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Duane-radial ray syndrome (SALL4)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Ellis-van-Creveld-Syndrom (EvC/EvC2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Episodische Ataxie Typ 2 (CACNA1A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Hypochondroplasie (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hypophosphatasie (ALPL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Isolierte Brachydaktylie der Finger IV/V (BMPR1B, IHH, GDF5, NOG, ROR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Isolierte Wachstumsfaktor Defizienz TYP 1A und 1B (GH1, GHRHR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Jeune Syndrom (WDR34)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Kleidocraniale Dysplasie (CCD)(RUNX2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Kollagenopathie Typ II (COL2A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Kraniosynostose Typ II (MSX-2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Kraniosynostose Typ III (TCF12)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Kurzrippen-Polydaktylie-Syndrom (EvC1, EvC2, IFT80, WDR19)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Limb-Pelvis-Syndrom (WNT7A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	

Miller Syndrom (DHODH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Morbus Crouzon (FGFR2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Morbus Pfeiffer (FGFR1, FGFR2, FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Morbus Muenke (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Multiple Epiphysäre Dysplasie Typ I / Pseudoachondroplasie (COMP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Multiple Epiphysäre Dysplasie Typ V (MATN3, COL2A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Nagel-Patella-Syndrom (LMX1b)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Opsismodysplasie (INPPL1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Ostegenesis imperfecta Typ IX (PPIB)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Osteogenesis imperfecta Typ IIb / VII (CRTAP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Osteogenesis imperfecta Typ I-IV (COL1A1 / COL1A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Osteogenesis imperfecta Typ VIII (LEPRE1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Osteogenesis imperfecta TypV (IFITM5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Osteogenesis imperfecta TypV (TMEM38B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Osteogenesis imperfecta 17 (SPARC)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Osteopathie mit Sklerose (OSCS) (AMER1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Pseudoachondroplasie (COMP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Saethre-Chotzen-Syndrom (FGFR2-3, TWIST)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Spondylocostale Dysosostis Typ 4 (DLL3, MESP2, HES7, LFNG)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Spondylocostale Dysosostis Typ 5 (SCDO5) (TBX6)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Stickler-Syndrom (COL2A1, COL11A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Syndaktylie Typ IV (LMBR1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
TAR-Syndrom (RBM8A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Tetraamelie (WNT3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Thanatophore Dysplasie Typ I (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Thanatophore Dysplasie Typ II (FGFR3)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Spinale Muskelatrophie (SMN1)	genomische DNA	**MLPA**	**MLPA / MS-MLPA Analyse (VA) Version: 006/06.2018**	Genetic Analyzer ABI3500	MLPA		
Adrenogenitales Syndrom (CYP21A2, CYP11B1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Androgen-Resistenz (AR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Aromatase Defizienz (CYP19A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Azoospermiefaktor (SY84, SY86, SY127, SY134, SY254, SY255, SY160, SYCP3, SRY)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
CBAVD (Congenital Bilateral Aplasia of Vas Deferens)(CFTR-Gen)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Empty follicle syndrome (EFS) (LHCGR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Primäre/vorzeitige Ovarialinsuffizienz /POF (FMR1, BMP15, FSHR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Sex Reversal (SRY)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

AGAT- Defizienz (GATM)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Albinismus (OCA, OCA2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Allan-Herndon-Dudley-Syndrom; AHDS (SLC16A2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Alpa1-Antitrypsin-Mangel A1-AT (SERPINA1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Alpha-Methylacetoacetic Aciduria (ACAT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Argininosuccinase Mangel (ASL)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Biotinidase Mangel (BTD)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Carnitine deficiency (SLC22A5)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Cerebrotendinous xanthomatosis (CYP27A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Dihydopyrimidinasemangel (DPYS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Familiäre Hypokalziurische Hyperkalzämie (FHH) (CASR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Fruktose-Intoleranz (ALDOB, FBP1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
G6PD-Mangel (Favismus) (G6PD)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Galaktosämie (GALT)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Gilbert Syndrom (Morbus Meulengracht) (UGT1A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Glykogenose Typ1A (G6PC)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Glykogenose Typ 5/ Morbus Mc Ardle (PYGM)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Glykogen-Speicherkrankheit durch hepatischen Glykogensynthase-Mangel (GYS2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hämochromatose (HFE)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Holocarboxylase Synthetase Defizienz (HLCS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Homozystinurie/CBS-Defizienz (CBS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hyperinsulinismus (KCNJ11, ABCC8)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Hypokalziurische Hyperkalzämie (Typ II) (GNA11)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hyperoxalurie (AGXT)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Hypothyreose (IGSF1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Idiopathische infantile Hyperkalzämie (CYP24A1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Langketten 3 hydroxyacyl CoA Dehydrogenase (LCHAD) Defizienz (HADHA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Leukodystrophie (TUBB4a, ABCD1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
MCAD-Defizienz (ACADM)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
MODY Syndrom Typ 1 und 3 (HNF1A, HNF4A)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Morbus Krabbe (GALC)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Morbus Wilson (ATP7B)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Analyt (Messgröße)	**Untersuchungsmaterial (Matrix)**	**Untersuchungstechnik**	**Anweisung/Version**	**Gerät**	**CE-Verfahren**	**in Haus-Verfahren**	
Metachromatische Leukodystrophie (ARSA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Metachromatic Leukodystrophy due to saposin B deficiency (PSAP)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Mukopolysaccharidosis TypIVB (GLB1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Nicht-autoimmune Hypertyreose (TSHR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Phenylketonurie (PKU) (PAH)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Pituitary Stalk Interruption Syndrome (PSIS) = angeborene Variante des Pickardt-Fahlbusch Syndrom--> Tertiäre Hypothyreose (GPR161)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Porphyrie (UROS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Pseudohypoaldosteronismus Typ1 (PHA1A) (NR3C2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Pyridoxamine 5 -Phosphat –Mangel (PNPO)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
P450 Oxidase Mangel (POR)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Renal tubular acidosis with deafness (ATP6V1B1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	

Smith-Lemli-Opitz-Syndrom (DHCR7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Succinyl-CoA Transferase Defizienz (SCOT) (OXCT1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Shwachman-Diamon Syndrom (SBDS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Tay-Sachs (HEXA)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
TDP-glucose 4,6,- Dehydratase (TGDS)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
3-MGA-aciduria Typ iV (SERAC1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
3-MGA Aciduria, Typ VII, with cataracts, neurologic involvement and neutropenia (CLPB)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Thrombophilie (Faktor-II-Mutation, Faktor-V-Mutation, MTHFR C677T, MTHFR A1298C)	genomische DNA	**Real-time PCR (VA)**	**Real-time PCR (VA)**	7300 PCR Real Time System	Real-time PCR		
Protein S-Mangel (PROS1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	in Haus-Verfahren	
Protein C-Defizienz (PROC)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Antithrombin-III-Mangel (SERPINC1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , 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	
3-M-Syndrom 1 (CUL7)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
3-M-Syndrom 2 (OBSL1)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
3-M-Syndrom 3 (CCDC8)	genomische DNA	PCR, DNA-Sequenzierung, **NGS**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**		PCR , SEQ , **NGS**	
Leri-Weill Dyschondrosteose LWD / Kleinwuchs (SHOX)	genomische DNA	PCR, DNA-Sequenzierung, **NGS,** MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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)**	MLPA	PCR , SEQ , **NGS**	
Von Willebrand Jürgens Syndrom (VWF)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**, MLPA	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)" , Illumina)**	MLPA	PCR , SEQ , **NGS**	

Webb-Dattani-Syndrom (ARNT2)	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**		PCR , SEQ , **NGS**	
X-chromosomale mentale Retardierung: Gen-Panel (118 Gene), X-chromosomale Gene (komplett) Gen Panel (1007 Gene), X-chromosomale Gene (komplett) Gen Liste (1007 Gene),	genomische DNA	PCR, DNA-Sequenzierung, **NGS Target Enrichment by Hybrid Capture**	**Polymerasekettenreaktion (VA) Version: 003/12.2019, DNA-Sequenzierung (VA) Version: 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 (Sequencing by Synthesis (SBS)“ , Illumina)**	NGS	PCR , SEQ , **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		
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 (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	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 (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	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 (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	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 (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	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 - Version 005/08.2021**	Fluoreszenzmikroskop (Zeiss, manuelle Aufnahme) mit Auswertung mittels Isis der Fa. MetaSystems	x		