

Liste der Verfahren im flexiblen Scope der Akkreditierung

Akkreditierungsnummer: D-ML-22009-01

Urkundeninhaber:

MVZ Düsseldorf-Centrum GbR

Immermannstr. 65A, 40210 Düsseldorf

Untersuchungsgebiet: Klinische Chemie

Untersuchungsart:

Chromatographie (UV/VIS-, Massenspektrometrische, Fluoreszenz-Detektion) ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Vitamin A	Serum	HPLC-UV	DOK-ID 11151, V2	ja	
Vitamin E	Serum	HPLC-UV	DOK-ID 11151, V2	ja	
Vitamin C	Serum	HPLC-UV	DOK-ID 11045, V1	ja	
Vitamin B2 (FAD)	EDTA-Blut	HPLC-FLD	DOK-ID 11219, V1	ja	
Vitamin B1	EDTA-Blut	HPLC-FLD	DOK-ID 11235, V2	ja	
Vitamin B6	EDTA-Blut	HPLC-FLD	DOK-ID 11235, V2	ja	
Methylmalonsäure	Serum	LC-MS/MS	DOK-ID 11329, V2	ja	04.09.2026
17-OH-Progesteron	Serum	LC-MS/MS	DOK-ID 10932, V2	ja	04.09.2026

Untersuchungsart:

Elektrochemische Untersuchungen ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Kalium	Serum	ISE (ionenselektive Elektrode)	DOK-ID 10121, V4	ja	10.01.2025
Natrium	Serum	ISE (ionenselektive Elektrode)	DOK-ID 10121, V4	ja	10.01.2025
Clorid	Serum	ISE (ionenselektive Elektrode)	DOK-ID 10121, V4	ja	10.01.2025

Untersuchungsart:

Ligandenassays ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Androgen-Index, frei	Serum	Berechnung	DOK-ID 11811, V1	ja	01.08.2024
Anti-Müller-Hormon	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
ß-HCG	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Carcinoembryonales Antigen	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Carcinoma Antigen 15-3	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025

Dehydroepiandrosteronsulfat	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Estradiol	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Ferritin	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Follikel-stimulierendes Hormon	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Folsäure	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
freies Thyroxin	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
freies Trijodthyronin	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Luteinisierendes Hormon	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Placental like Growth Factor	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Progesteron	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Prolaktin	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Quotienten aus sFlt-1/PlGF	Serum	Berechnung	DOK-ID 11811, V1	ja	01.08.2024
Sexualhormonbindendes Globulin	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Soluble Fms-like thyrosinkinase-1	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Testosteron, gesamt	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Thyreotropin	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Vitamin B12	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Vitamin B12, active	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025

Untersuchungsart:
Spektrometrie (UV- /VIS-Photometrie) ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Bilirubin, direkt	Serum	VIS-Photometrie	DOK-ID 11809, V1	ja	01.08.2024
Bilirubin, gesamt	Serum	VIS-Photometrie	DOK-ID 11809, V1	ja	01.08.2024
Glucose	Serum, Citrat-Blut (+Inhibitor)	UV-Photometrie	DOK-ID 11809, V1	ja	01.08.2024

Untersuchungsart:
Spektrometrie (Turbidimetrie) ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Alkalischen Phosphatase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Apolipoprotein A1	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Apolipoprotein B	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Calcium gesamt	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Cholesterin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Cholinesterase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
C-reaktives Protein	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Creatinin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Creatinkinase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Creatinkinase Muscle-Brain type	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Cystatin C	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025

Eisen	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Ferritin-Index	Serum	Berechnung	DOK-ID 11811, V1	ja	01.08.2024
Gamma-Glutamyltransferase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Gesamteiweiß	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
GFR/glomeruläre Filtrationsrate (CKD-EPI Formel)	Serum	Berechnung	DOK-ID 11811, V1	ja	01.08.2024
GFR/glomeruläre Filtrationsrate (nach Cystatin C)	Serum	Berechnung	DOK-ID 11811, V1	ja	01.08.2024
Glutamat-Oxalacetat-Transaminase/Aspartat-Aminotransferase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Glutamat-Pyruvat-Transaminase/Alanin-Aminotransferase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Hämoglobin A1c	EDTA-Blut	Turbidimetrie	DOK-ID 11807, V2	ja	11.12.2025
Haptoglobin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Harnsäure	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Harnstoff	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
HDL-Cholesterin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Homocystein	EDTA-Blut, Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Lactatdehydrogenase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
LDL Friedewald-Formel	Serum	Berechnung	DOK-ID 11811, V1	ja	01.08.2024
LDL-Cholesterin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Lipase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Lipoprotein	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Löslicher Transferrinrezeptor	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Magnesium	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Pankreas-Amylase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Phosphat anorganisch	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Rheumafaktoren	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Transferinsättigung	Serum	Berechnung	DOK-ID 11811, V1	ja	26.11.2025
Transferrin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025
Triglyceride	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.11.2025

Untersuchungsgebiet: Immunologie

Untersuchungsart:

Ligandenassays ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Anti-CCP-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	
Thyreoperoxidase-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	
TSH-Rezeptor-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Untersuchungsart:

Molekularbiologische Untersuchungen (Amplifikationsverfahren) ^[Flex C]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
ACE I-/ D-Variante (rs1799752)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10166, V3	nein	
Alpha-Thalassämie	EDTA-Blut; DNA	Amplikonbasierte Anreicherung, Sequencing by Avidite Base Chemistry (SNV, CNV), custom pipeline	DNA-Extraktion: DOK-ID 10119, V4 Thalassämiediagnostik mittels Devyser NGS Kit, DOK-ID: 11435, V2	ja	
Alport-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Amyloidose	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Angioödem	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Angelman-Syndrom (15q11-q13 region und SNRPN and MAGEL2 DMRs), Methylierungsstatus und Deletions/Duplikationsanalyse	EDTA-Blut; DNA	Methylierungs-sensitive Multiplex Ligation dependent Probe Amplification (MS-MLPA)	DNA-Extraktion: DOK-ID 10119, V4 Multiplex ligation-dependent probe amplification (MLPA®): DOK-ID 10456, V3 Auswertung: DOK-ID 11041, V2	ja	
Anterior-Segment-Dysgenese	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Arthritis	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Ataxie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Autoinflammatorische Syndrome	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

autosomal dominante polyzystische Nierenerkrankung	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Beta-Thalassämie	EDTA-Blut; DNA	Amplikonbasierte Anreicherung, Sequencing by Avidite Base Chemistry (SNV, CNV), custom pipeline	DNA-Extraktion: DOK-ID 10119, V4 Thalassämiediagnostik mittels Devyser NGS Kit, DOK-ID: 11435, V2	ja	
Bernard-Soulier-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
CADASIL/ CARASIL	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Charcot-Marie-Tooth-Krankheit Typ 1A	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	Multiplex Ligation dependent Probe Amplification (MLPA)	DNA-Extraktion: DOK-ID 10119, V4 Multiplex ligation-dependent probe amplification (MLPA®): DOK-ID 10456, V1 Auswertung: DOK-ID 11041, V1	ja	
COMT Polymorphismus c.472G>A (Catechol-O-Methyltransferase)	EDTA-Blut; DNA	Real-Time PCR	DNA-Extraktion: DOK-IDs 10119, V4; DOK-ID 11352, V1 Real-Time PCR: DOK-ID 10217, V3	nein	
CYP19A1 Polymorphismus rs727479 (Aromatase)	EDTA-Blut; DNA	Real-Time PCR	DNA-Extraktion: DOK-IDs 10119, V4; DOK-ID 11352, V1 Real-Time PCR: DOK-ID 10217, V3	nein	

CYP2C19 Allele - Medikamentenunverträglichkeit (Cytochrom P-450 2C19 *2, *3, *4, *5, *6, *7, *8, *17)	EDTA-Blut; DNA	PCR, Strip-Assay	DNA-Extraktion: DOK-ID 10119, V4 Strip-Assay: DOK-ID 10213, V3	ja	
CYP2C9 *2 und *3 Allele - Siponimod (Cytochrom P-450 2C9)	EDTA-Blut; DNA	PCR, Strip-Assay	DNA-Extraktion: DOK-ID 10119, V4 Strip-Assay: DOK-ID 10213, V3	ja	
CYP2C9 *2 und *3 Allele - Siponimod (Cytochrom P-450 2C9)	EDTA-Blut; DNA	PCR, Sanger Sequenzierung	DNA-Extraktion: DOK-ID 10119, V4 PCR: DOK-ID 10224, V3 Sanger-Sequenzierung: DOK-ID 10286, V3 Kapillarelektrophorese: DOK-ID 10287, V2 Auswertung: DOK-ID 10488, V2	nein	
CYP2D6 Polymorphismen (Cytochrom P-450 2D6 Polymorphismen c.-1584C>G, c.31G>A, c.100C>T, c.124G>A, c.137_138insT, c.181-1G>C, c.320C>T, c.454delT, c.505G>T, c.505G>A, c.506-1G>A, c.522_523ins(10nt), c.755delA, c.841_843delAAG, c.886C>T, c.971A>C, c.985+39G>A, c.1012G>A, c.1457G>C)	EDTA-Blut; DNA	PCR, Strip-Assay	DNA-Extraktion: DOK-ID 10119, V4 Strip-Assay: DOK-ID 10213, V3	ja	
Cystische Fibrose/CFTR-assoziierte Erkrankungen	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
DPYD Polymorphismen - 5-Fluorouracil-Unverträglichkeit (Dihydropyrimidin-Dehydrogenase *2A, *13, p.D949V, HapB3 Typisierung)	EDTA-Blut; DNA	PCR, Strip-Assay	DNA-Extraktion: DOK-ID 10119, V4 Strip-Assay: DOK-ID 10213, V3	ja	
Ehlers-Danlos-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Eizellreifungsdefekte	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Eisenrefraktäre Eisenmangelanämie (IRIDA)	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Faktor V Leiden-Variante (rs6025)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10167, V3	ja	
Faktor II-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Faktor V-HR2-Variante (rs1800595)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10265, V3	ja	
Faktor V-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Faktor VII-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Faktor X-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Faktor XI-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Faktor XII-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Faktor XIII-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
FVIII Erhöhung	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Familiäre Adenomatöse Polyposis	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Familiäres Mittelmeerfieber	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Fanconi-Anämie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Favismus/ G6PD-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
FGFR3 assoz. Erkrankungen	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Fibrinogen-gamma-Variante (rs2066865)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10170, V3	nein	
Fibrinogen-Störung	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Fragiles-X-Syndrom	EDTA-Blut; DNA	Repeat-primed PCR	DNA-Extraktion: DOK-ID 10119, V4 Nachweis Fragiles-X-Syndrom, Fragile-X-assoziierte Tremor_Ataxie Syndrom, Fragile-X-assoziierte prämatüre Ovarialinsuffizienz mittels AmpliEx PCR_CE FMR1 Kit: DOK-ID 10970, V2	ja	
GAD1 Polymorphismus c.-147G>A (Glutamatdecarboxylase)	EDTA-Blut; DNA	Real-Time PCR	DNA-Extraktion: DOK-IDs 10119, V4; DOK-ID 11352, V1 Real-Time PCR: DOK-ID 10217, V3	nein	
Gastrointestinaler Stromatumor (KIT, MAX, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127)	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Tumordiagnostik (Mutation)	Gewebeproben ^a ; DNA	Amplikonbasierte Anreicherung, Sequencing by Avidite Base Chemistry (SNV, CNV), custom pipeline	DNA Extraktion: DOK-ID 10975, V3 Amplifikation: DOK-ID 10854, V5 Sequenzierung: DOK-ID 10986, V4 Auswertung: DOK-ID 10863, V4 Pathologie: 10883, V2	nein	
Tumordiagnostik (Translokation)	Gewebeproben ^a ; RNA, DNA	Amplikonbasierte Anreicherung, Sequencing by Avidite Base Chemistry (SNV, CNV), custom pipeline	DNA Extraktion: DOK-ID 10975, V3 Amplifikation: DOK-ID 10854, V5 Sequenzierung: DOK-ID 10986, V4 Auswertung: DOK-ID 10863, V4 Pathologie: 10883, V2	nein	
Mammakarzinom (Mutation)	Blut-Plasma; cfDNA	Amplikonbasierte Anreicherung, Sequencing by Avidite Base Chemistry (SNV, CNV), custom pipeline	DNA Extraktion: DOK-ID 10975, V3 Amplifikation: DOK-ID 10854, V5 Sequenzierung: DOK-ID 10986, V4 Auswertung: DOK-ID 10863, V4 Pathologie: 10883, V2	nein	
Geschlechtsdifferenzierungsstörungen	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Gorlin-Goltz-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
GP1a-Variante (rs1126643)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10169, V3	nein	
Habituelle Aborte	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hämochromatose seltene Formen	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hämophilie A	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Hämophilie B	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hämophilie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäre Hauttumore	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäre neuroendokrine Tumore	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäre Neuropathie mit Neigung zu Druckläsionen	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	Multiplex Ligation dependent Probe Amplification (MLPA)	DNA-Extraktion: DOK-ID 10119, V4 Multiplex ligation-dependent probe amplification (MLPA®): DOK-ID 10456, V3 Auswertung: DOK-ID 11041, V2	ja	

Hereditäre Pankreatitis	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditärer Darmkrebs	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäres Endometriumkarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäres Hypophysenadenom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Hereditäres Magenkarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäres Mamma- und Ovarialkarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäres Mammakarzinom bei Männern	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäres Nierenkarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Hereditäres Ovarialkarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäres Pankreaskarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäres Paragangliom- /Phäochromozytom-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hereditäres Prostatakarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Hereditäres Schilddrüsenkarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
HFE-Varianten C282Y / H63D (rs1800562 / rs1799945)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10264, V3	ja	
Hörstörung (AR) Typ IA/IB (GJB2, GJB6)	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	Multiplex Ligation dependent Probe Amplification (MLPA)	DNA-Extraktion: DOK-ID 10119, V4 Multiplex ligation-dependent probe amplification (MLPA®): DOK-ID 10456, V3 Auswertung: DOK-ID 11041, V2	ja	
Hörstörung nicht syndromal (AR) Typ IA	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	PCR, Sanger Sequenzierung	DNA-Extraktion: DOK-ID 10119, V4 PCR: DOK-ID 10224, V3 Sanger-Sequenzierung: DOK-ID 10286, V3 Kapillarelektrophorese: DOK-ID 10287, V2 Auswertung: DOK-ID 10488, V2	nein	
HPA-I-la/1b-Variante (rs5918)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10164, V3	nein	
Hyperbilirubinämie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hypercholesterinämie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Hyperlipidämie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hyperparathyreoidismus	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hyperthyreose	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hypertriglyceridämie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Hypogonadotroper Hypogonadismus	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hypokalziurische Hyperkalzämie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hypophosphatasie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Hypothyreose	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Klinisches Exom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Komb. FVIII-/ Faktor V-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Kongenitale Kontrakturale Arachnodaktylie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Legius Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Leiomyomatosis	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Li-Fraumeni-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Loeys Dietz Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Lynch-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Männliche Infertilität	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
MAOA uVNTR-Polymorphismus (Monoaminoxidase-A; Neurotransmitter)	EDTA-Blut; DNA	PCR, Sanger Sequenzierung	DOK-ID 10236, V1	nein	
Marfan-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Meduläres Schilddrüsenkarzinom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Mesomele Dysplasie (SHOX)	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	Multiplex Ligation dependent Probe Amplification (MLPA)	DNA-Extraktion: DOK-ID 10119, V4 Multiplex ligation-dependent probe amplification (MLPA®): DOK-ID 10456, V3 Auswertung: DOK-ID 11041, V2	ja	
MODY	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Molenschwangerschaft	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Morbus Osler	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Morbus Wilson	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
MTHFR Polymorphismus c.665C>T (aka C677T) (Methylen-Tetrahydrofolat; Methotrexat)	EDTA-Blut; DNA	Real-Time PCR	DNA-Extraktion: DOK-IDs 10119, V4, DOK-ID 11352, V1 Real-Time PCR: DOK-ID 10217, V3	nein	
Muir-Torre-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Multiple Endokrine Neoplasie Typ 1	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Multiple Endokrine Neoplasie Typ 2	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Muskeldystrophie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Muskeldystrophie Duchenne/ Becker	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Nephroblastom/ Wilms-Tumor	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Neurofibromatose Typ 1	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Neurofibromatose Typ 2	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Neutropenie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Nicht-obstruktive Azoospermie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Nierenerkrankungen	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Noonan-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Obstruktive Azoospermie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Osteogenesis imperfecta	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Ovariale Dysgenese	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
PAI-1-4G/5G-Variante (rs1799889)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10168, V3	ja	
PARP-Inhibitoren	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Pendred-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Periodische Fiebersyndrome	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Peutz-Jeghers	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Phenylketonurie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Plasminogen-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Polyzystisches Ovarialsyndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Prader-Willi-Syndrom (15q11-q13 region, SNRPN, MAGEL2 DMRs), Methylierungsstatus und Deletions/Duplikationsanalyse	EDTA-Blut; DNA	Methylierungs-sensitive Multiplex Ligation dependent Probe Amplification (MS-MLPA)	DNA-Extraktion: DOK-ID 10119, V4 Multiplex ligation-dependent probe amplification (MLPA®): DOK-ID 10456, V3 Auswertung: DOK-ID 11041, V2	ja	
Prämatüre Ovarialinsuffizienz	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Primäre Erythromelalgie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)		
Primäre ziliäre Dyskinesie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Prothrombin-Variante (rs1799963)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10201, V3	ja	

Protein C-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Protein C-Rezeptordefizienz	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Protein S-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Protein Z-Mangel	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

PTEN-Hamartom-Tumorsyndrom und Differentialdiagnosen	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
RASopathien	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Schilddrüsenhormonresistenz	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Schlaganfall oder Schlaganfall-ähnliche Episoden	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Schwannomatose	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Small Fiber Neuropathie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Smith-Lemli-Opitz Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
SOD2 Polymorphismus Ala16Val (Superoxiddismutase 2)	EDTA-Blut; DNA	Real-Time PCR	DNA-Extraktion: DOK-IDs 10119, V4; DOK-ID 11352, V1 Real-Time PCR: DOK-ID 10217, V3	nein	
Spermiendefekte (Form und Beweglichkeit)	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Thorakales Aortenaneurysma und Aortendissektion	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Thrombozytopathie/ Thrombozytopenie	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Thrombomodulin-Defekt	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
TSH-Rezeptordefizienz	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Libraray Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Tuberöse Sklerose	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
VKORC1 Polymorphismus c.-1639G>A (Vitamin-K-Epoxid-Reduktase; Cumarinderivate)	EDTA-Blut; DNA	PCR, Strip-Assay	DOK-ID 10211, V1	ja	
von Hippel-Lindau-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
von Willebrand-Syndrom	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
Weibliche Infertilität	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	

Whole exome sequencing (WES)	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	in-solution capture, sequencing by Avidite Base Chemistry (SNV, CNV) custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI), DOK-ID 10973 V3 (Varvis) Modul SeqNext in aktueller Version (JSI), Varvis in aktueller Version (Limbus MedTec)	nein	
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Untersuchungsart:
Chromosomenanalyse ^[Flex C]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
angeborener Chromosomensatz	Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA	molekulare Karyotypisierung, in-solution capture, sequencing by synthesis (CNV), custom pipeline	NGS-Methode: DOK-ID 10885, V3 DNA-Extraktion: DOK-ID 10119, V4 Library Präparation: DOK-ID 11039, V3 Laufplanung (Pooling, Barcodes, Samplesheet): DOK-ID 11393, V2 Auswertung: DOK-ID 10966, V2 (JSI)	nein	

Untersuchungsgebiet: Mikrobiologie
Untersuchungsart:
Ligandenassays ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Treponema pallidum (TPHA)	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Toxoplasma gondii Ak-IgG	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Toxoplasma gondii Ak-IgM	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025

Untersuchungsgebiet: Virologie

Untersuchungsart:

Ligandenassays ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Cytomegalievirus IgG-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Cytomegalievirus IgM-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Hepatitis-A-Virus IgM/IgG-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Hepatitis-B-Virus Oberflächen-Ag	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Hepatitis-B-Virus Oberflächen-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Hepatitis-C-Virus-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
HIV 1/2 p24 screen	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Rötelnvirus IgG-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025
Rötelnvirus IgM-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	26.11.2025

Untersuchungsgebiet: Transfusionsmedizin

Untersuchungsart:

Molekularbiologische Untersuchungen ^[Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Typisierung der Rhesus-Formel	EDTA-Blut	SSP-PCR	DNA Extraktion: DOK-ID 10119, V4 Amplifikation: DOK-ID 11332, V1	ja	
Rhesus weak D Typisierung	EDTA-Blut	SSP-PCR	DNA Extraktion: DOK-ID 10119, V4 Amplifikation: DOK-ID 11332, V1	ja	
Rhesus partial D Typisierung	EDTA-Blut	SSP-PCR	DNA Extraktion: DOK-ID 10119, V4 Amplifikation: DOK-ID 11332, V1	ja	
AB0 Typisierung	EDTA-Blut	SSP-PCR	DNA Extraktion: DOK-ID 10119, V4 Amplifikation: DOK-ID 11332, V1	ja	
KEL Typisierung	EDTA-Blut	SSP-PCR	DNA Extraktion: DOK-ID 10119, V4 Amplifikation: DOK-ID 11332, V1	ja	
Fetales Rhesus D	fetale ccfDNA, EDTA-Blut	Real-Time PCR	DNA Extraktion: DOK-ID 11276, V3 Amplifikation: DOK-ID 10666, V1	ja	