ZOTZ | KLIMAS
MVZ Düsseldorf-Centrum GbR

Stand: 15.07.2025

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, V1	ja	
17-OH-Progesteron	Serum	LC-MS/MS	DOK-ID 10932, V1	ja	

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	03.01.2025
Natrium	Serum	ISE (ionenselektive Elektrode)	DOK-ID 10121, V4	ja	03.01.2025

Untersuchungsart:

Ligandenassays [Flex B]

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Androgen-Index, frei	Serum	Berechnung	DOK-ID 11811, V1		01.08.2024
Anti-Müller-Hormon	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
ß-HCG		ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Carcinoembryonales Antigen	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Carcinoma Antigen 15-3	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Dehydroepiandrosteronsulfat	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Estradiol	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Ferritin	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Follikel-stimulierendes Hormon	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Folsäure	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
freies Thyroxin	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
freies Trijodthyronin	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Luteinisierendes Hormon	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Placental like Growth Factor	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Progesteron	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Prolaktin	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Quotienten aus sFlt-1/PlGF	Serum	Berechnung	DOK-ID 11811, V1		01.08.2024
Sexualhormonbindendes Globulin	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Soluble Fms-like thyrosinkinase-1	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Testosteron, gesamt	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Thyreotropin	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Vitamin B12	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Vitamin B12, active	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025

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	26.06.2025
Apolipoprotein A1	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Apolipoprotein B	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Calcium gesamt	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Cholesterin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Cholinesterase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
C-reaktives Protein	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Creatinin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Creatinkinase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Creatinkinase Muscle-Brain type	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Cystatin C	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Eisen	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Ferritin-Index	Serum	Berechnung	DOK-ID 11811, V1		01.08.2024
Gamma-Glutamyltransferase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Gesamteiweiß	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
GFR/glomeruläre Filtrationsrate (CKD-EPI Formel)	Serum	Berechnung	DOK-ID 11811, V1		01.08.2024
GFR/glomeruläre Filtrationsrate (nach Cystatin C)	Serum	Berechnung	DOK-ID 11811, V1		01.08.2024
Glutamat-Oxalacetat-Transaminase/Aspartat-Aminotransferase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Glutamat-Pyruvat-Transaminase/Alanin-Aminotransferase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Hämoglobin A1c	EDTA-Blut	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Haptoglobin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Harnsäure	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Harnstoff	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
HDL-Cholesterin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Homocystein	EDTA-Blut, Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Lactatdehydrogenase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
LDL Friedewald-Formel	Serum	Berechnung	DOK-ID 11811, V1	ja	26.06.2025
LDL-Cholesterin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Lipase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Lipoprotein	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Löslicher Transferrinrezeptor	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Magnesium	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Pankreas-Amylase	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Phosphat anorganisch	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Rheumafaktoren	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Transferinsättigung	Serum	Berechnung	DOK-ID 11811, V1		01.08.2024
Transferrin	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025
Triglyceride	Serum	Turbidimetrie	DOK-ID 11807, V2	ja	26.06.2025

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	27.06.2025
Thyreoperoxidase-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
TSH-Rezeptor-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025

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	NGS (Amplikon-basiert, sequencing by avidity)	DNA-Extraktion: DOK-ID 10119, V4 Thalassämiediagnostik mittels Devyser NGS Kit, DOK-ID: 11435, V1	ja	
Alport-Syndrom (COL4A3, COL4A4, COL4A5, MYH9, CFHR5, NPHS2)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Amyloidose (APOA1, APOA2, APP, B2M, CST3, F10, FGA, GPNMB, GSN, IL31RA, LYZ, MEFV, MVK, NLRP1, NLRP3, OSMR, POLA1, PRNP, RET, TACSTD2, TGFBI, TNFRSF1A, TTR)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Angioödem (ADGRE2, ANGPT1, CPN1, F12, HS3ST6, KNG1, MYOF, NLRP3, PLCG2, PLG, SERPING1, SPINK5, XPNPEP2)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Arthritis (ADA2, ANKH, CCN6, CFI, F8, GALNT3, LACC1, MEFV, NLRP1, NOD2, PHEX, PRG4, PSTPIP1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
autosomal dominante polyzystische Nierenerkrankung (DNAJB11, GANAB, HNF1B, PKD1, PKD2, PAX2, ALG5, ALG9, IFT140, JAG1, NOTCH2, PRKCSH, SEC61A1, SEC63, ALG8, UMOD)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Beta-Thalassämie	EDTA-Blut; DNA	NGS (Amplikon-basiert, sequencing by avidity)	DNA-Extraktion: DOK-ID 10119, V4 Thalassämiediagnostik mittels Devyser NGS Kit, DOK-ID: 11435, V1	ja	
CADASIL/ CARASIL (COL4A1, COL4A2, CTSA, GLA, HTRA1, ITM2B, NOTCH3, TREX1, CSF1R)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Charcot-Marie-Tooth-Krankheit Typ 1A	Gewebe[a], 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	DOK-ID 10456, V3 01.07.2025; DOK-ID 11041, V2 27.06.2025
COMT Polymorphismus c.472G>A (Catechol-O-Methyltransferase)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10218, V2	nein	
CYP19A1 Polymorphismus rs727479 (Aromatase)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10221, V2	nein	
CYP2C19 Allele - Medikamentenunverträglichkeit (Cytochrom P-450 2C19 *2, *3, *4, *5, *6, *7, *8, *17)	EDTA-Blut; DNA	PCR, Strip-Assay	DOK-ID 10219, V2	ja	
CYP2C9 *2 und *3 Allele - Siponimod (Cytochrom P-450 2C9)	EDTA-Blut; DNA	PCR, Strip-Assay	DOK-ID 10210, V1	ja	
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	DOK-ID 10214, V1	ja	
DPYD Polymorphismen - 5-Fluorouracil-Unverträglichkeit (Dihydropyrimidin-Dehydrogenase *2A, *13, p.D949V, HapB3 Typisierung)	EDTA-Blut; DNA	PCR, Strip-Assay	DOK-ID 10213, V2	ja	

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Ehlers-Danlos-Syndrom (ADAMTS2, AEBP1, B3GALT6, B4GALT7, C1R, C1S, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, DSE, FKBP14, PLOD1, PRDM5, SLC39A13, TNXB, ZNF469)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Eizellreifungsdefekte (BTG4, CDC20, KHDC3L, MOS, NLRP2, NLRP5, NLRP7, OOEP, PADI6, PANX1, PATL2, REC114, TLE6, TRIP13, TUBB8, WEE2, ZFP36L2, ZP1, ZP2, ZP3)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Familiäre Adenomatöse Polyposis (APC, BMPR1A, GREM1, MSH3, MUTYH, NTHL1, POLD1, POLE, PTEN, RNF43, SMAD4, STK11)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Faktor V Leiden-Variante (rs6025)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10167, V3	ja	
Faktor V-HR2-Variante (rs1800595)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10265, V3	ja	
Fanconi-Anämie (BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFWD3, SLX4, TOP3A, UBE2T, XRCC2, RFWD3, ATM, NBN, ATR, CENPJ, CEP152, CEP63, DNA2, NIN, NSMCE2, RBBP8, TRAIP)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Fibrinogen-gamma-Variante (rs2066865)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10170, V3	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 prämature Ovarialinsuffizienz mittels AmplideX PCR_CE FMR1 Kit: DOK-ID 10970, V2	ja	DOK-ID 10970, V2 13.05.2024
GAD1 Polymorphismus c.-147G>A (Glutamatdecarboxylase)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10220, V2	nein	
Gastrointestinaler Stromatumor (KIT, MAX, NF1, PDGFRA, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
~~Solide Tumore (AKT1, ALK, BRAF, CTNNB1, EGFR, ERBB2, ERBB3, ESR1, FOXL2, GNA11, GNAQ, IDH1, IDH2, KIT, KRAS, MET, NRAS, PDGFRA, PIK3CA, RAF1, RET, TP53)~~	~~Gewebe[a], EDTA-Blut; DNA~~	~~NGS (sequencing-by-synthesis (Illumina), amplikonbasiert, CLC Genomics Workbench Software (Version 24.0), Translokationsanalyse)~~	~~DNA Extraktion: DOK-ID 10975, V2 Amplifikation: DOK-ID 10854, V3 Sequenzierung: DOK-ID 10986, V3 Auswertung: DOK-ID 10863, V2 Pathologie: DOK-ID 10883, V2 DOK-ID 10112, V2 DOK-ID 10613, V3~~	~~nein~~	gestrichen 13.06.2025
~~Solide Tumore (BRCA1, BRCA2)~~	~~Gewebe[a], EDTA-Blut; DNA~~	~~NGS (sequencing-by-synthesis (Illumina), amplikonbasiert, CLC Genomics Workbench Software (Version 24.0), Mutationssuche)~~	~~DNA Extraktion: DOK-ID 10975, V2 Amplifikation: DOK-ID 10854, V3 Sequenzierung: DOK-ID 10986, V3 Auswertung: DOK-ID 10863, V2 Pathologie: DOK-ID 10883, V2 DOK-ID 10112, V2 DOK-ID 10613, V3~~	~~nein~~	gestrichen 13.06.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Geschlechtsdifferenzierungsstörungen (AR, CYP11A1, CYP17A1, DHH, DHX37, DMRT1, HSD17B3, HSD3B2, LHCGR, MAP3K1, NR0B1, NR5A1, POR, SOX9, SRD5A2, SRY, WT1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Gorlin-Goltz-Syndrom (PTCH1, PTCH2, SUFU)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
GPIa-Variante (rs1126643)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10169, V3	nein	
Habituelle Aborte (ALB, B3GALNT1, BUB1B, C11orf80, F10, F12, F13A1, F13B, F2, F5, FGA, FGB, FGG, HTR1A, KHDC3L, MEI1, MTHFR, NLRP7, PROC, PROS1, SERPINC1, SERPINE1, SYCP3, THBD, TSHR, ANXA5, PIGA, ADAMTS13, HRG, SERPIND1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäre Hauttumore (BAP1, CDK4, CDKN2A, MITF, MLH1, MSH2, MSH6, POT1, PTCH1, PTEN, SUFU, TERT)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Hereditäre neuroendokrine Tumore (AIP, CASR, CDC73, CDKN1B, FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TSC1, TSC2, VHL, PRKAR1A, TP53)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäre Neuropathie mit Neigung zu Druckläsionen	Gewebe[a], 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	DOK-ID 10456, V3 01.07.2025; DOK-ID 11041, V2 27.06.2025
Hereditäre Pankreatitis (CASR, CEL, CFTR, CIDEC, CLDN2, CPA1, CTRC, LPL, PRSS1, SBDS, SLC25A13, SPINK1, TRPV6, UBR1, CELA3B)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditärer Darmkrebs (APC, AXIN2, BMPR1A, CHEK2, GREM1, EPCAM, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, RNF43, SMAD4, STK11, TP53)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Hereditäres Endometriumkarzinom (EPCAM, MLH1, MSH2, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäres Hypophysenadenom (AIP, CDH23, GPR101, MEN1, PRKAR1A, CDKN1B)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäres Magenkarzinom (APC, BMPR1A, CDH1, CHEK2, CTNNA1, EPCAM, KIT, MLH1, MSH2, MSH6, MUTYH, PMS2, SMAD4, STK11, TP53, PDGFRA, PTEN, SDHB, SDHC, SDHD, MAX, SDHA, SDHAF2, TMEM127)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäres Mamma- und Ovarialkarzinom (ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Hereditäres Mammakarzinom bei Männern (ATM, BRCA1, BRCA2, CHEK2, PALB2, PTEN, STK11, TP53)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäres Nierenkarzinom (BAP1, CHEK2, DICER1, DIS3L2, FH, FLCN, GPC3, MAX, MET, PTEN, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMARCA4, SMARCB1, TMEM127, TP53, TSC1, TSC2, VHL, WT1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäres Ovarialkarzinom (ATM, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, SMARCA4, STK11, TP53, BARD1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäres Pankreaskarzinom (APC, ATM, BRCA1, BRCA2, CDKN2A, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PALLD, PMS2, PRSS1, SPINK1, STK11, TP53, VHL)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Hereditäres Paragangliom-/Phäochromozytom-Syndrom (CDC73, CDKN1B, DLST, EPAS1, FH, MAX, MEN1, NF1, PRKAR1A, PTEN, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC25A11, TMEM127, TP53, VHL)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäres Prostatakarzinom (ATM, BRCA1, BRCA2, BRIP1, CHEK2, HOXB13, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, RAD51D, TP53)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hereditäres Schilddrüsenkarzinom (APC, CHEK2, NKX2-1, PRKAR1A, PTEN, RET, MAX, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, TP53, CDKN1B, WRN, MEN1, CDC73, DICER1, SEC23B, FOXE1, SLC5A5)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
HFE-Varianten C282Y / H63D (rs1800562 / rs1799945)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10264, V3	ja	
Hörstörung (AR) Typ IA/IB (GJB2, GJB6)	Gewebe[a], 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	DOK-ID 10456, V3 01.07.2025; DOK-ID 11041, V2 27.06.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Hörstörung nicht syndromal (AR) Typ IA	Gewebe[a], 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	DOK-ID 10224, V3 27.03.2024; DOK-ID 10286, V3 27.03.2024
HPA-I-Ia/1b-Variante (rs5918)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10164, V3	nein	
Hypercholesterinämie (ABCG5, ABCG8, APOA5, APOB, APOC2, APOC3, APOE, CETP, LDLR, LDLRAP1, LIPC, LPL, PCSK9, LIPA, STAP1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hyperlipidämie (ABCG5, ABCG8, APOA5, APOB, APOC2, APOC3, APOE, CETP, CREB3L3, GCKR, GPD1, GPIHBP1, LCAT, LDLR, LDLRAP1, LIPC, LMF1, LPL, LRP6, PCSK9, LIPA, STAP1, APOA5, ALMS1, CYP27A1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hyperparathyreoidismus (AP2S1, CASR, CDC73, CDKN1B, GCM2, GNA11, MEN1, RET)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Hyperthyreose (ALB, SECISBP2, SLC16A2, THRB, TSHR, TTR)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hypertriglyceridämie (APOA5, APOC2, APOC3, APOE, CREB3L3, GCKR, GPD1, GPIHBP1, LCAT, LIPC, LMF1, LPL, LRP6, LIPA)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hypogonadotroper Hypogonadismus (ANOS1, CHD7, FEZF1, FGF17, FGF8, FGFR1, FSHB, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, LHB, NR0B1, NSMF, PROK2, PROKR2, RNF216, SEMA3A, SOX2, SOX10, SPRY4, TAC3, TACR3, WDR11)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Hypokalziurische Hyperkalzämie (AP2S1, CASR, GNA11)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Hypothyreose (DUOX2, DUOXA2, FOXE1, GLIS3, GNAS, HESX1, IGSF1, IRS4, IYD, LHX3, LHX4, NKX2-1, OTX2, PAX8, POU1F1, PRKAR1A, PROP1, SECISBP2, SLC16A2, SLC26A4, SLC26A7, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TRHR, TSHB, TSHR)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Kongenitale Kontrakturale Arachnodaktylie (FBN2)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Legius Syndrom (SPRED1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Li-Fraumeni-Syndrom (CDKN2A, CHEK2, TP53)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Loeys Dietz Syndrom (SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Lynch-Syndrom (MLH1, MSH2, MSH6, PMS2, EPCAM)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Männliche Infertilität (ADGRG2, AK7, AMH, AMHR2, AR, ARMC2, AURKC, CATSPER1, CCDC103, CCDC39, CCDC40, CCNO, CFAP251, CFAP298, CFAP300, CFAP43, CFAP44, CFAP47, CFAP65, CFAP69, DMRT1, DNAAF1 , DNAAF11 , DNAAF2 , DNAAF3, DNAAF4 , DNAAF5, DNAAF6, DNAH1, DNAH17, DNAH2, DNAH5, DNAH8, DNAH9 , DNAI1, DNAI2, DNAL1, DPY19L2, FOXJ1, FSIP2, HYDIN, KLHL10, M1AP, MCIDAS, NEK10, NR5A1, ODAD1, ODAD2, ODAD3, ODAD4, PICK1, QRICH2, RSPH1, RSPH4A, SHOC1, SOHLH1, SPAG1, SPATA16, SPEF2, STAG3, SUN5, SYCE1, TEX11, TEX14, TEX15, TTC21A, TTC29, ZMYND10, ZPBP)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
MAOA uVNTR-Polymorphismus (Monoaminoxidase-A; Neurotransmitter)	EDTA-Blut; DNA	PCR, Sanger Sequenzierung	DOK-ID 10236, V1	nein	

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Marfan-Syndrom (FBN1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Meduläres Schilddrüsenkarzinom (RET)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Mesomele Dysplasie (SHOX)	Gewebe[a], 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	DOK-ID 10456, V3 01.07.2025; DOK-ID 11041, V2 27.06.2025
Molenschwangerschaft (KHDC3L, MEI1, NLRP7, PADI6, REC114, TOP6BL)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
MTHFR Polymorphismen c.667C>T und c.1298A>C (Methylen-Tetrahydrofolat; Methotrexat)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10237, V2	nein	

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Neurofibromatose Typ 1 (NF1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Nicht-obstruktive Azoospermie (AR, DMRT1, KLHL10, M1AP, NR5A1, SHOC1, STAG3, SUN5, SYCE1, TEX11, TEX14, TEX15)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Noonan-Syndrom (BRAF, CBL, CDC42, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Obstruktive Azoospermie (ADGRG2, CFTR)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Osteogenesis imperfecta (BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, MBTPS2, P3H1, PLOD2, PPIB, SEC24D, SERPINF1, SERPINH1, SP7, SPARC, TENT5A, TMEM38B, WNT1, ALPL, FAM46A, KDELR2, MESD, TAPT1)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Ovariale Dysgenesie (BMP15, CLPP, ESR2, ERAL1, FSHR, HARS2, HSD17B4, LARS2, MCM9, MRPS22, NUP107, PSMC3IP, SOHLH1, SPIDR, STAG3, TWNK, ZSWIM7)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
PAI-1-4G/5G-Variante (rs1799889)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10168, V3	Ja	
PARP-Inhibitoren (BRCA1, BRCA2, PALB2)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Polyzystisches Ovarialsyndrom (CAPN10, CYP11A1, INS, INSR)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Prämature Ovarialinsuffizienz (BMP15, C14orf39, CLPP, CYP17A1, DIAPH2, ERCC6, ESR1, FIGLA, FOXL2, FSHB, FSHR, GALT, GDF9, HARS2, HFM1, HSD17B4, HSF2BP, INHA, KASH5, KHDRBS1, LARS2, LHCGR, MCM8, MCM9, MSH4, MSH5, NOBOX, NR5A1, POF1B, POLG, PSMC3IP, SOHLH1, STAG3, SYCE1, TP63, TWNK)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Primäre ziliäre Dyskinesie (CCDC103, CCDC39, CCDC40, CCNO, CFAP298, CFAP300, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, FOXJ1, HYDIN, MCIDAS, NEK10, ODAD1, ODAD2, ODAD3, ODAD4, RSPH1, RSPH4A, SPAG1, ZMYND10)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Prothrombin-Variante (rs1799963)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10201, V3	ja	
PTEN-Hamartom-Tumorsyndrom und Differentialdiagnosen (AKT1, PIK3CA, PTEN, SDHB, SDHC, SDHD, SEC23B)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
RASopathien (BRAF, CBL, CDC42, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, MAPK1, MRAS, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, RRAS2, SHOC2, SOS1, SOS2, SPRED1, SPRED2)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Schlaganfall oder Schlaganfall-ähnliche Episoden (ABCC6, ADA2, CBS, COL3A1, COL4A1, COL4A2, CST3, FBN1, FLNA, GLA, HTRA1, NOTCH3, RNF213, SLC2A10, TGFB2, TGFBR1, TGFBR2, TREX1, TTR)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Schwannomatose (SMARCB1, LZTR1, NF2)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
SOD2 Polymorphismus Ala16Val (Superoxiddismutase 2)	EDTA-Blut; DNA	Real-Time PCR	DOK-ID 10219, V2	nein	
Spermiendefekte (Form und Beweglichkeit) (AK7, ARMC2, AURKC, CFAP251, CFAP43, CFAP44, CFAP47, CFAP65, CFAP69, DNAH1, DNAH17, DNAH2, DNAH8, DPY19L2, FSIP2, PICK1, QRICH2, SPATA16, SPEF2, TTC21A, TTC29, ZPBP)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Thorakales Aortenaneurysma und Aortendissektion (ACTA2, BGN, COL1A2, COL3A1, COL5A1, COL5A2, EFEMP2, FBN1, FBN2, FLNA, FOXE3, LOX, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SLC2A10, SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2, THSD4)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

Analyt	Untersuchungsmaterial	Untersuchungstechnik	Vorgabedokument, Version	CE-Verfahren	Verfahren geändert am:
Thrombozytopathie/ Thrombozytopenie (ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, ADTB3A, ANKRD26, ANO6, AP3B1, AP3D1, ARPC1B, BLOC1S5, BLOC1S3, BLOC1S6, CD36, CDC42, CYCS, DIAPH1, DTNBP1, ETV6, FERMT3, FLI1, FLNA, FYB1, GATA1, GFI1B, GNE, GP1BA, GP1BA, GP1BB, GP6, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGA2B, ITGB3, ITGB3, IKZF5, KDSR, LYST, MECOM, MPIG6B, MPL, MYH9, NBEA, NBEAL2, P2RY12, PLA2G4A, PLAU, RAP1B, RASGRP2, RBM8A, RUNX1, SLFN14, SRC, STIM1, STXBP2, TBXA2R, TBXAS1, THPO, THPO, TPM4, TUBB1, VIPAS39, VPS33B, VWF, WAS)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
Tuberöse Sklerose (TSC1, TSC2)	EDTA-Blut; DNA, Heparin-Blut; DNA, Mundschleimhaut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025
VKORC1 Polymorphismus c.-1639G>A (Vitamin-K-Epoxid-Reduktase; Cumarinderivate)	EDTA-Blut; DNA	PCR, Strip-Assay	DOK-ID 10211, V1	Ja	
Weibliche Infertilität (BMP15, BTG4, CAPN10, CLPP, CDC20, CYP11A1, C14orf39, DIAPH2, ERCC6, ESR1, ESR2, ERAL1, FIGLA, FOXL2, FSHR, GDF9, HARS2, HFM1, HSD17B4, HSF2BP, INHA, INS, INSR, KASH5, KHDRBS1, KHDC3L, LARS2, LHCGR, MCM8, MCM9, MEI1, MOS, MRPS22, MSH4, MSH5, NLRP2, NLRP5, NLRP7, NOBOX, NR5A1, NUP107, OOEP, PADI6, PANX1, PATL2, POF1B, PSMC3IP, REC114, SOHLH1, SPIDR, STAG3, SYCE1, TLE6, TOP6BL, TP63, TRIP13, TUBB8, TWNK, WEE2, XRCC2, ZFP36L2, ZP1, ZP2, ZP3, ZSWIM7)	Gewebe[a], EDTA-Blut, Heparin-Blut; DNA	NGS (in-solution capture, sequencing by avidity), SNV- und CNV-Pipeline: SequencePilot, Modul SeqNext Version 5.2 (JSI), Varvis version 1.25 (Limbus MedTec)	NGS-Methode: DOK-ID 10885, V2 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)	nein	DOK-ID 11039, V3 07.07.2025; DOK-ID 11393, V2 09.07.2025; DOK-ID 10973, V3 09.04.2025

[a]Einige Proben werden vor Beginn der Bearbeitung durch das Labor histologisch durch Fachärzte für Pathologie bearbeitet und beurteilt. Diese fachfremde Leistung gehört nicht zum Scope der Akkreditierung des Labors.

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	27.06.2025
Toxoplasma gondii Ak-IgG	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Toxoplasma gondii Ak-IgM	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025

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	27.06.2025
Cytomegalievirus IgM-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Hepatitis-A-Virus IgM/IgG-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Hepatitis-B-Virus OberflächenAg	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Hepatitis-B-Virus Oberflächen-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Hepatitis-C-Virus-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
HIV 1/2 p24 screen	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Rötelnvirus IgG-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025
Rötelnvirus IgM-Ak	Serum	ECLIA	DOK-ID 11802, V2	ja	27.06.2025

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	