

Liste aller Untersuchungsverfahren im flexiblen Akkreditierungsbereich

Innerhalb der mit * gekennzeichneten Untersuchungsbereiche ist dem medizinischen Laboratorium, ohne dass es einer vorherigen Information und Zustimmung der Deutschen Akkreditierungsstelle GmbH bedarf, die freie Auswahl von normierten oder ihnen gleichzusetzenden Untersuchungsverfahren gestattet. Innerhalb der mit ** gekennzeichneten Untersuchungsbereiche ist dem medizinischen Laboratorium, ohne dass es einer vorherigen Information und Zustimmung der Deutschen Akkreditierungsstelle GmbH bedarf, die Modifizierung sowie Weiter- und Neuentwicklung von Untersuchungsverfahren gestattet.

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)

Untersuchungsart:

Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
SNV-Genotypisierung (Nachweis spezifischer varianter Allele)			
HIV-Wirtsresistenz	EDTA-Blut, DNA; DNA	PCR, Gelelektrophorese	AM-MO-126/D
Chemokininrezeptor 5 (CCR5)-delta 32 Deletion	EDTA-Blut, EDTA-Knochenmark; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
Meulengracht- (Gilbert-) Syndrom (Customized Panel ABA) (UGT1A1*28-, *6- Polymorphismus)	EDTA-Blut, EDTA-Knochenmark; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
Thrombophilie-Panel (Detektion von 11 Mutationen in den Genen F2, F5, F12, F13, HABP2 (alias FSAP), MTHFR, SERPINC1 (alias AT3) und SERPINE1 (alias PAI-1))	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
Alpha-1 Antitrypsin Mangel (AATD) Panel (Detektion von 5 SNPs im Gen SERPINA)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
Leqembi Panel (Detektion von 2 SNPs im Gen APOE)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
Exome-ID-Panel (Detektion von 15 aussagekräftigen SNPs die in der Whole-Exome-Sequenzierung abgedeckt sind)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
5-Fluorouracil-Toxizitäts-Panel (Detektion von 14 Mutationen im Gen DPYD)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
HLFH (Hämochromatose-Laktose-Fruktose-Histamin)-Panel (Detektion von 17 SNPs in den Genen F2, F5, F12, F13, FGB, HABP2 (alias FSAP), MTHFR, SERPINC1 (alias AT3) und SERPINE1 (alias PAI-1))	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
77 SNP PRS-(Polygenetischer Risiko-Score)-Panel zur Vorhersage von Brustkrebs (Detektion von 77 SNPs)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
AML (Akute myeloische Leukämie)-Panel (Detektion von 55 Mutationen in den Genen DNMT3A, FLT3, IDH1, IDH2, KIT und NPM1)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
CMML-(Chronische Myelomonozytäre Leukämie)-Panel (Detektion von 104 Mutationen in den Genen CBL, EZH2, NRAS, SETBP1, SF3B1 und SRSF2)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
JAK2-Panel (Detektion von 5 Mutationen in den Genen CALR, JAK2 und MPL)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version
Lymphom-Panel (Detektion von 87 Mutationen in den Genen BRAF, MYC, MYD88, NRAS und SF3B1)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
MDS (Myelodysplastische Syndrom)-Panel (Detektion von 212 Mutationen in den Genen ultiPlexes containing 87 Assays for the detection of 212 Mutations in the genes CALR, CBL, CSF3R, DDX41, DNMT3A, EZH2, FLT3, IDH1, IDH2, JAK2, KIT, MPL, NPM1, NRAS, SETBP1, SF3B1, SRSF2, U2AF1 und ZRSR2)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
MPN (Myeloproliferativen Neoplasien)-Panel (Detektion von 103 Mutationen in den Genen CALR, CBL, EZH2, IDH1, IDH2, JAK2, MPL, SF3B1, SRSF2 und U2AF1)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
OncoScreen-Panel_4MP (Detektion von 98 Mutationen in den Genen BRAF, EGFR, ERBB2, KRAS, NRAS, PIK3CA und POLE)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
COLON-Panel (Detektion von 149 Mutationen in den Genen BRAF, KRAS, NRAS und PIK3CA)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
POLE-Panel (Detektion von 9 Mutationen im Gen POLE)	EDTA-Blut, DNA; DNA	Multiplex-PCR, matrixunterstützte Laser-Desorptions-Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D
Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version
Nachweis spezifischer chromosomaler Regionen			
cABL/BCR-Translokation t(9;22), qualitativ	peripheres Blut, Knochenmark; RNA	PCR, Gelelektrophorese	AM-MO-112/F
cABL/BCR-Translokation t(9;22), quantitativ	peripheres Blut, Knochenmark; RNA	Realtime-PCR	AM-MO-109/C
CCND1/IGH-Translokation t(11;14)	peripheres Blut, Knochenmark; DNA	PCR, Gelelektrophorese	AM-MO-137/C
IGH/BCL2-Translokation t(14;18)	peripheres Blut, Knochenmark; DNA	PCR, Gelelektrophorese	AM-MO-137/C
MYH11/CBFB-Translokation inv(16)	peripheres Blut, Knochenmark; RNA	PCR, Kapillarelektrophorese	AM-MO-138/F
RUNX1/RUNX1T1-Translokation t(8;21)	peripheres Blut, Knochenmark; RNA	PCR, Kapillarelektrophorese	AM-MO-138/F
Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+Version
Fragmentlängen-Analyse			
Chimärisusanalyse	peripheres Blut, Knochenmark, Mundschleimhautabstrich; DNA	Genotyp zur Chimärisusuntersuchung anhand STR-Analyse	AM-MO-130/D

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Gezielte Kopienzahlanalyse			
Hereditäres Mamma-/ Ovarialkarzinom (HBOC) Vermutetes erhöhtes hereditäres Risiko nach positiver Familienanamnese. (BRCA1 [OMIM *113705], BRCA2 [OMIM *600185])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
HNPCC/Lynch-Syndrom (MLH1 [OMIM *120436], MSH2 [OMIM *609309], MSH6 [OMIM *600678], PMS2 [OMIM *600259], EPCAM [OMIM *185535])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Marfan-Syndrom (FBN1-Gen [OMIM *134797])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Antithrombin-III-Mangel (SERPINC1-Gen [OMIM *107300])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Protein C-Mangel (PROC-Gen [OMIM *612283])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Protein S-Mangel (PROS1-Gen [OMIM *176880])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Von-Willebrand-Syndrom (VWF-Gen [OMIM *613160])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Ehlers-Danlos-Syndrom (COL3A1 [OMIM *120180], COL5A1 [OMIM *120215])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Familiäre Adenomatöse Polyposis (APC [OMIM *611731], MUTYH [OMIM *604933])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Neuropathie (PMP22 [OMIM *601097])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Morbus Osler (ACVRL1 [OMIM *601284], ENG [OMIM *131195])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Maturity Onset Diabetes of the Young (MODY) Typ 1- 10 (GCK [OMIM *138079], HNF1A [OMIM *142410], HNF1B [OMIM *189907], HNF4A [OMIM *600281], KLF11 [OMIM *603301], NEUROD1 [OMIM *601724], PAX4 [OMIM *167413], CEL [OMIM *114840], INS [OMIM *176730], PDX1 [OMIM *600733])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Hereditäre Spastische Paraplegie (SPAST [OMIM *604277], ATL1 [OMIM *606439])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Loeys-Dietz-Syndrom (TGFBRI [OMIM *190181], TGFBRI2 [OMIM *190182])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Morbus Hirschsprung (RET [OMIM *164761])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
F5-Mangel (F5 [OMIM *612309])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
F7-Mangel (F7 [OMIM *613878])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Hämophilie A (F8 [OMIM *300841])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Hämophilie B (F9 [OMIM *300746])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
F10-Mangel (F10 [OMIM *613872])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
F11-Mangel (F11 [OMIM *264900])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F
Zystische Fibrose (CFTR-Gen [OMIM *602421])	EDTA-Blut, DNA; DNA	MLPA	AM-MO-123/F

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Gerichtete NGS-Panel-Analyse bei benannten klinischen Verdachtsdiagnosen			
Familiäre Tumordispositionen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Hereditäres Mamma-/ Ovarialkarzinom (HBOC)	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Mamma-/Ovarialkarzinom; Prostatakarzinom; Pankreaskarzinom fortgeschrittenes Ovarialkarzinom, metastasiertes und HER2-negatives Mammakarzinom, metastasiertes Pankreaskarzinom nach platinbasierter Chemotherapie, metastasiertes, kastrationsresistentes Prostatakarzinom zur Therapieentscheidung (PARP-Inhibitoren) (BRCA1 [OMIM *113705], BRCA2 [OMIM *600185])	EDTA-Blut, DNA; DNA	PCR, "large-amplicon" Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS) (Illumina MiSeq, NextSeq), GATK	AM-MO-141/E NGS GGS Pipeline 1.2
HNPCC/Lynch-Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK, CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2
Carney Complex	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK, CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.3
Familiäres Magenkarzinom	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Familiäres Pankreaskarzinom	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Familiäre adenomatöse Polyposis (FAP)	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
MUTYH- assoziierte Polyposis (MAP)	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Polymerase Proofreading-assoziierte Polyposis (PPAP)	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Polyposis coli	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Familiäre juvenile Polyposis	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Peutz-Jeghers-Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Cowden-Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Li Fraumeni-Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Familiäres Nierenzellkarzinom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
SchilddrüsenCa gesamt (inkl. MTC)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Nicht-medulläres SchilddrüsenCa / PTC	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Gastrointestinale Polyposis	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Fanconi-Anämie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hereditäres Phäochromozytom- und Paragangliomsyndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Kutane Melanome	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Gesamtpanel Tumordisposition	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Neurologische Erkrankungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Amyotrophe Lateralsklerose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Ataxie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
CADASIL	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Demenz	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Dystonie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hereditäre Neuropathien	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hereditäre spastische Paraplegie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Migräne	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hemiplegische Migräne	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Neurodegeneration mit Eisenablagerungen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Parkinson	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Paroxysmale Dyskinesie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Periodische Paralyse	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Small-Fiber-Neuropathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Muskuläre Erkrankungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Gliedergürtelmuskeldystrophie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Kongenitale myasthene Syndrome	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Kongenitale und distale Myopathien	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Metabolische Myopathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Rhabdomyolyse	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Skapuloperoneale Myopathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Spinale Muskelatrophie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Tremor	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Muskuläre Erkrankungen (Gesamt-Panel)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Genetisch bedingte Erkrankungen mit Bezug zur Inneren Medizin (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Cystische Fibrose (CFTR-Gen [OMIM *602421])	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Adipositas	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
ALPS (Autoimmun-Lympho-proliferatives Syndrom)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Amyloidose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Calcium-/Phosphatstoffwechselstörung	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Chronisch-entzündliche Darmerkrankungen (CED)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Fettstoffwechselstörung, nicht-syndromal	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Glykogenose und Differentialdiagnosen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hämochromatose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hepatopathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hereditäres Angioödem (HAE)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hereditäre Pankreatitis	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hyperparathyreoidismus (HPT)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hypogonadotroper Hypogonadismus, syndromal	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hypogonadotroper Hypogonadismus, nicht syndromal (mit und ohne Anosmie)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Immundefekte	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Maturity Onset Diabetes of the Young (MODY)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Mukopolysaccharidose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Multiple Acyl-CoA-Dehydrogenase Defekt (MAAD)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
NASH (Nicht-alkoholische Fettleberhepatitis, NAFLD)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Osteopetrose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Osteoporose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Periodisches Fiebersyndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Phosphatstoffwechselförderung	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Prämature Ovarialinsuffizienz (POI)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Progerie und Lipodystrophie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Pubertas praecox	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Septische Granulomatose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Systemischer Lupus erythematodes (SLE)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Stoffwechselerkrankung (Gesamt-Panel)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Kardiovaskuläre Erkrankungen und RASopathien (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVC)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Dilatative Kardiomyopathie (DCM)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hypertrophe Kardiomyopathie (HCM)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Linksventrikuläre non-compaction Kardiomyopathie (LVNC)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Restriktive Kardiomyopathie (RCM)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Brugada-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Catecholaminerge polymorphe ventrikuläre Tachykardie (CPVT)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Long QT-Syndrom (LQTS)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Short-QT-Syndrom (SQTS)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Isolierte Herzfehler	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Syndromale Herzfehler	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
RASopathien	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Nierenerkrankungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
CAKUT	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Fokale-Segmentale-Glomerulosklerose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hyperaldosteronismus	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Nephrokalzinose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Nephronophthisen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Nephrotisches-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Nierenagenese / Nierenhypoplasie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Zystische Nierenerkrankung	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hauterkrankungen (die Genliste des untenstehenden Panels findet sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Ektodermale Dysplasie Stufe 1	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Ektodermale Dysplasie Stufe 2 (syndromal und DD)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Ichthyose und Differentialdiagnosen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Psoriasis	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Thrombozytopenien/ -pathien (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Isolierte Thrombozytopathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Syndromale/komplexe Thrombozytopenie und Differentialdiagnosen / Thrombozytopathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Isolierte Thrombozytopenie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Hämatologische Erkrankungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Neutropenie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Erweitertes Neutropenie-Panel	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hyper-IgE-Syndrom (HIES)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Sphärozytose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Sphärozytose und DDs (hämolytische Anämie)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Thrombophilie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Epilepsie (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Absence Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Benigne Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Epileptische Enzephalopathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Fieberggebundene Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Fokale Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Frühkindliche Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Gesamtpanel Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Metabolische Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Myoklonische Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Rolando Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Therapie-relevante Epilepsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Entwicklungsstörungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Aicardi-Goutiere-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Anorektale Fehlbildungen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Autismus	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Bardet-Biedl-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
CDG-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Coffin-Siris-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Cornelia-de-Lange Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
46,XY-DSD (nicht-syndromal)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Entwicklungsstörungen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Holoprosenzephalie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Joubert-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Kleinwuchs	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Kortikale Hirnfehlbildung	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Kraniosynostosen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Leukoenzephalopathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Leukodystrophie und Leukoenzephalopathie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Lissenzephalie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Lyosomale Erkrankungen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Makrozephalie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Meckel-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Megalenzephalie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Mikrozephalie und pontocerebelläre Hypoplasie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Senior-Loken-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Skelettfehlbildung	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Zahndurchbruchstörung	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Zellweger-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Bindegewebs- und Gefäßerkrankungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Cutis laxa	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Diverse Bindegewebserkrankungen	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Ehlers-Danlos Syndrom, vaskulärer Typ (Typ IV)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Ehlers-Danlos Syndrom, Gesamtpanel	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Fibromuskuläre Dysplasie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Marfan-Syndrom (FBN1-Gen [OMIM *134797])	EDTA-Blut, DNA; DNA	Sequence capture (Illumina), Sequencing-by-synthesis (Illumina), MiSeq, NextSeq (Illumina), GATK (SNV), CNV-GATK (CNV)	AM-MO-140/F NGS GGS Pipeline 1.2 NGS CNV Pipeline 2.0
Marfan Syndrom-Panel	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Osteogenesis imperfecta (Knochenbrüchigkeit)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Stickler-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Thorakale Aortenaneurysmen und Dissektionen (TAAD)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Augenerkrankungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Achromatopsie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Externe Ophthalmoplegie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Flecked Retina Erkrankung	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Glaukom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hornhautdystrophien	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Katarakt	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Leber'sche kongenitale Amaurose	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Netzhautdystrophie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Okulärer und okulokutaner Albinismus (komplett)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Okulärer und okulokutaner Albinismus (nicht-syndromal)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Okulärer und okulokutaner Albinismus (syndromal)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Optikusatrophie (isoliert, nicht syndromal)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Optikusatrophie Gesamtpanel (syndromal, nicht syndromal)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Retinopathia pigmentosa	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Septo-optische Dysplasie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Usher Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Walker-Warburg-Syndrom	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Zapfen-Stäbchen-Dystrophie	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Hörstörungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Nicht-Syndromale Schwerhörigkeit	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Syndromale Schwerhörigkeit	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0
Atemwegserkrankungen (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Primäre ciliäre Dyskinesie (PCD)	EDTA-Blut, DNA; DNA	Whole Exome Sequencing (WES); SNV, CNV, Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Hämato-Onkologie (die Genlisten der untenstehenden Panels finden sich im Leistungsverzeichnis und in den Untersuchungsanforderungen auf der Homepage)			
Follikuläres Lymphom-Panel	EDTA-Blut, DNA aus Blut; DNA	amplicon-basierte Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS), MiSeq, NextSeq (Illumina), GATK	AM-MO-149/C NGS GGS Pipeline 1.2
Mantelzell-Lymphom (MCL)-Panel	EDTA-Blut, DNA aus Blut; DNA	amplicon-basierte Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS), MiSeq, NextSeq (Illumina), GATK	AM-MO-149/C NGS GGS Pipeline 1.2
Marginalzonen-Lymphom (MZL)-Panel	EDTA-Blut, DNA aus Blut; DNA	amplicon-basierte Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS), MiSeq, NextSeq (Illumina), GATK	AM-MO-149/C NGS GGS Pipeline 1.2
T-Zelllymphom (T-NHL)-Panel	EDTA-Blut, DNA aus Blut; DNA	amplicon-basierte Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS), MiSeq, NextSeq (Illumina), GATK	AM-MO-149/C NGS GGS Pipeline 1.2
Myeloproliferative Neoplasie (MPN)-Panel	EDTA-Blut, DNA aus Blut; DNA	amplicon-basierte Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS), MiSeq, NextSeq (Illumina), GATK	AM-MO-149/C NGS GGS Pipeline 1.2
Myelodysplastisches Syndrom (MDS)-Panel	EDTA-Blut, DNA aus Blut; DNA	amplicon-basierte Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS), MiSeq, NextSeq (Illumina), GATK	AM-MO-149/C NGS GGS Pipeline 1.2
Pan Myelo/Lympho-Panel	EDTA-Blut, DNA aus Blut; DNA	amplicon-basierte Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS), MiSeq, NextSeq (Illumina), GATK	AM-MO-149/C NGS GGS Pipeline 1.2
Solide Tumore			
Mamma-/Ovarialkarzinom 1. Rezidiv eines Ovarialkarzinoms zur Therapieentscheidung (PARP-Inhibitoren) 2. HER2-negatives primäres metastasierendes Mammakarzinom zur Therapieentscheidung (PARP-Inhibitoren) (BRCA1 [OMIM *113705], BRCA2 [OMIM *600185])	Gewebeproben, DNA; DNA	amplicon-basierte Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS), MiSeq, NextSeq (Illumina), GATK	AM-MO-149/C NGS GGS Pipeline 1.2
Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Gerichtete NGS-Analyse bei benannten klinischen Verdachtsdiagnosen ("large-amplicon" Sequenzierung mittels Next-Generation-Sequencing-(NGS))			
Antithrombin-III-Mangel (SERPINC1-Gen [OMIM *107300])	EDTA-Blut, DNA; DNA	PCR, "large-amplicon" Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS) (Illumina MiSeq, NextSeq), GATK	AM-MO-141/E NGS GGS Pipeline 1.2
Protein C-Mangel (PROC-Gen [OMIM *612283])	EDTA-Blut, DNA; DNA	PCR, "large-amplicon" Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS) (Illumina MiSeq, NextSeq), GATK	AM-MO-141/E NGS GGS Pipeline 1.2
Protein S-Mangel (PROS1-Gen [OMIM *176880])	EDTA-Blut, DNA; DNA	PCR, "large-amplicon" Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS) (Illumina MiSeq, NextSeq), GATK	AM-MO-141/E NGS GGS Pipeline 1.2
Von-Willebrand-Syndrom (VWF-Gen [OMIM *613160])	EDTA-Blut, DNA; DNA	PCR, "large-amplicon" Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS) (Illumina MiSeq, NextSeq), GATK	AM-MO-141/E NGS GGS Pipeline 1.2
TP53 Somatisch (TP53-Gen)	EDTA-Blut, DNA; DNA	PCR, "large-amplicon" Sequenzierung mittels Next-Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS) (Illumina MiSeq, NextSeq), GATK	AM-MO-141/E NGS GGS Pipeline 1.2
Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
Ungerichtete NGS-basierende Analyse bei ungerichteter klinischer Indikation			
Whole exome sequencing (WES); SNV, CNV	EDTA-Blut, DNA; DNA	Sequence capture (Twist), Sequencing-by-synthesis (Illumina), NextSeq (Illumina), Congenica (SNV), CNV-GATK (CNV)	AM-MO-152/B NGS CNV Pipeline 2.0

Untersuchungsgebiet: Humangenetik (Zytogenetik)

**Untersuchungsart:
Chromosomenanalyse****

Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung+Version Pipeline/Kit/Panel+ Version
angeborener Chromosomensatz	peripheres Blut, Nabelschnurblut	Chromosomenbänderungsanalyse	AM-ZG-103/P
erworbener Chromosomensatz	Knochenmark, Blut	Chromosomenbänderungsanalyse	AM-ZG-102/P AM-ZG-101/S
Nachweis spezifischer chromosomaler Regionen	Native und/oder kultivierte Zellen aus Knochenmark, Fruchtwasser, Chorionzotten, peripheres Blut, Nabelschnurblut	Fluoreszenz-in-situ-Hybridisierung (FISH): Pränataler Schnelltest Nachweis von Mikrodeletionen, Rearrangements, Translokationen, numerischen Aberrationen	AM-ZG-101/S

Untersuchungsgebiet: Virologie

**Untersuchungsart:
Ligandenassays***

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
anti-HAV IgG	Serum, EDTA- Plasma	CMIA	AM-KC-116/L
HBs-Antigen	Serum, EDTA- Plasma	CMIA	AM-KC-116/L
anti-HBs quantitativ	Serum, EDTA- Plasma	CMIA	AM-KC-116/L
anti-HBc IgG	Serum, EDTA- Plasma	CMIA	AM-KC-116/L
anti-HBe	Serum, EDTA- Plasma	CMIA	AM-KC-116/L
anti-HCV	Serum, EDTA- Plasma	CMIA	AM-KC-116/L
HIV1/2 Antikörper / p24-Antigen	Serum, EDTA- Plasma	CMIA	AM-KC-118/M
CMV IgG + IgM Antikörper	Serum, EDTA- Plasma	CMIA	AM-KC-118/M
HTLV I/II Antikörper	Serum, EDTA- Plasma	CMIA	AM-KC-118/M
Rubella-Virus IgG Antikörper	Serum, EDTA- Plasma	CMIA	AM-KC-118/M
EBV EBNA-1 IgG	Serum, EDTA- Plasma	CMIA	AM-KC-118/M
EBV VCA IgG	Serum, EDTA- Plasma	CMIA	AM-KC-118/M
EBV VCA IgM	Serum, EDTA- Plasma	CMIA	AM-KC-118/M

**Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)****

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
HIV-RNA	Plasma	TMA	AM-MO-106/K
HIV-RNA oder provirale DNA	Plasma	Hochauflösende Resistenztestung mittels Reverser Transkription, nested PCR und Next Generation Sequencing (Illumina MiSeq), NGS-Virologie-Pipeline (in-house)	AM-MO-117/L NGS Virologie Pipeline 1.3
HIV-RNA oder provirale DNA	Plasma	Hochauflösende Tropismusbestimmung mittels Reverser Transkription, nested PCR und Next Generation Sequencing (Illumina MiSeq), NGS-Virologie-Pipeline (in-house)	AM-MO-135/G NGS Virologie Pipeline 1.3
HBV-DNA	Plasma	Realtime-PCR, quantitativ	AM-MO-132/G
HBV-DNA	Plasma	Hochauflösende Resistenztestung/ Genotypisierung mittels PCR und Next Generation Sequencing (Illumina MiSeq), NGS-Virologie-Pipeline (in-house)	AM-MO-133/E NGS Virologie Pipeline 1.3
HCV-RNA	Plasma	TMA	AM-MO-106/K
HCV-RNA	Plasma	Genotypisierung mittels Reverser Transkription, PCR und Sequenzierung	AM-MO-134/H
HCV-RNA	Plasma	Hochauflösende Resistenztestung mittels Reverser Transkription, PCR und Next Generation Sequencing (Illumina MiSeq), NGS-Virologie-Pipeline (in-house)	AM-MO-143/E NGS Virologie Pipeline 1.3
CMV-DNA	Plasma	Realtime-PCR, quantitativ	AM-MO-121/I
HPV-DNA	FFPE	Multiplex-PCR, matrixunterstützte Laser-Desorption/Ionisation mit Flugzeitmassenspektrometer-Detektion (MALDI-TOF-MS)	AM-MO-145/D

Untersuchungsgebiet: Transfusionsmedizin

Untersuchungsart:

Agglutinationsteste *

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
ABO-System	EDTA-Vollblut	AG / AK-Bindung Gelkarte	AM-KC-104/I
Rh-System	EDTA-Vollblut	AG / AK-Bindung Gelkarte	AM-KC-104/I
Kell-Antigen	EDTA-Vollblut	AG / AK-Bindung Gelkarte	AM-KC-104/I
freie irreguläre Antikörper (IgG), AKS	Serum/ EDTA-Plasma	AG / AK-Bindung Gelkarte (indirekter Coombstest)	AM-KC-104/I

Untersuchungsart:

Durchflusszytometrie**

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
HLA-Antikörper (Nachweis und Spezifizierung)	Serum	Durchflusszytometrische Analyse mittels farb-codierter Beads	AM-HL-107/O

Untersuchungsart:

Lysisreaktionen**

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
HLA-Antikörper (Crossmatch)	Serum	LCT	AM-HL-102/I
HLA-Antikörper (Nachweis und Spezifizierung)	Serum	LCT	AM-HL-100/K

Untersuchungsart:

Molekularbiologische Untersuchungen (Amplifikationsverfahren)**

Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version
HLA-A*, B*, C* DRB1*, DRB3*, DRB4*, DRB5* DQA1*, DQB1* DPA1*, DPB1* (Zweifeld-Typisierung)	EDTA-, Citrat-, CPDA-Blut, Mundschleimhautabstrich; DNA	PCR, "large-amplicon" Sequenzierung mittels Next- Generation-Sequencing-(NGS) Technologie ("sequencing-by-synthesis", SBS) (Illumina MiSeq), NGS-HLA-Pipeline (in-house)	AM-HL-109/I NGS HLA Pipeline 1.11

Prüfgebiet: Forensische Genetik (Abstammungsgutachten nach GenDG)

Prüfart:

Polymerase-Kettenreaktion (PCR)

Analyt (Messgröße)	Prüfgegenstände (Matrix)	Prüftechnik	Anweisung/ Version
Genotyp zur Abstammungsfeststellung	Humane DNA aus: Mundschleimhautabstrichen, Blutproben	DNA-Extraktion, STR-Analyse: PCR mit flexiblen PCR-Reagenzien mit anschließender Elektrophorese und allelischer Zuordnung der PCR-Produkte	AM-MO-129/H

Probenahme

Probenahme Forensische Genetik (Abstammungsgutachten nach GenDG)**

Norm / Ausgabedatum Hausmethode / Version	Probenahmeverfahren	Probenmatrix
AA-MO-101 Version G Ausgabedatum: 28.02.2025 AA-IN-102 Version D Ausgabedatum 10.10.2023	Probenahme im Rahmen der Abstammungsfeststellung	Mundschleimhautabstrich, Blut

Prüfgebiet: Nukleinsäureanalytik

Nukleinsäure-Amplifikation [Flex C]

Analyt (Meßgröße)	Prüfgegenstände (Matrix)	Prüftechnik	Anweisung/Version
Nukleinsäuresequenzen	DNA	Genomsequenzierungen (oder "large-amplicon" Sequenzierung) mittels Next-Generation-Sequencing (NGS) Technologie ("sequencing-by-synthesis", SBS) nach vorheriger „DNA-library“-Herstellung (Workflow: DNA-Tag-/Fragmentierung/library"-Herstellung [Nextera/Nextera XT-Library] – SBS – Assemblierung der Sequenzen („reads“) zu „contigs“ (zusammenhängende, große Genomabschnitte) oder „scaffolds“ (Genomgerüst) - (Illumina-Technologie)	AM-MO-122 Version: E
Nukleinsäuresequenzen	DNA	Höchaufösende, ultratiefe Sequenzierung von Genen oder Genabschnitten (PCR-Produkte, „Amplikons“) mittels NGS-Technologie ("sequencing-by-synthesis", SBS) nach vorheriger „DNA-library“-Herstellung (Workflow: Library-Herstellung durch Adaptor-Ligation [TruSeq-Library] oder Verwendung von PCR-Produkten mit P5/P7-Fusionsprimern – SBS [„gespikete“ Hintergrundlibrary phiX] - (Illumina-Technologie)	AM-MO-127 Version: E
Nukleinsäuresequenzen	DNA	Whole Exom oder Panel mittels NGS-Technologie ("sequencing-by-synthesis", SBS) nach vorherigem "Enrichment" mit spezifischen Sonden (Workflow: Fragmentierung - Twist library Herstellung - Enrichment - Mapping).	AM-MO-152 Version: B