

## Deutsche Akkreditierungsstelle

### Anlage zur Akkreditierungsurkunde D-ML-22009-01-01 nach DIN EN ISO 15189:2024

**Gültig ab:** 13.07.2026

Ausstellungsdatum: 13.07.2026

**Diese Urkundenanlage ist Bestandteil der Akkreditierungsurkunde D-ML-22009-01-00.**

Inhaber der Akkreditierungsurkunde:

**MVZ Düsseldorf-Centrum GbR**  
**Immermannstraße 65A, 40210 Düsseldorf**

mit dem Standort

**MVZ Düsseldorf-Centrum GbR**  
**Immermannstraße 65A, 40210 Düsseldorf**

Das Medizinische Laboratorium erfüllt die Anforderungen gemäß DIN EN ISO 15189:2024, um die in dieser Anlage aufgeführten Konformitätsbewertungstätigkeiten durchzuführen. Das Medizinische Laboratorium erfüllt gegebenenfalls zusätzliche gesetzliche und normative Anforderungen, einschließlich solcher in relevanten sektoralen Programmen, sofern diese nachfolgend ausdrücklich bestätigt werden.

Die Anforderungen an das Managementsystem in der DIN EN ISO 15189 sind in einer für medizinische Laboratorien relevanten Sprache verfasst und stehen insgesamt in Übereinstimmung mit den Prinzipien der DIN EN ISO 9001.

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**Untersuchungen im Bereich:**

Medizinische Laboratoriumsdiagnostik

**Untersuchungsgebiete:**

Klinische Chemie

Immunologie

Humangenetik

Mikrobiologie

Virologie

Transfusionsmedizin

**Flexibler Akkreditierungsbereich:**

Dem Medizinischen Laboratorium ist innerhalb der gekennzeichneten Untersuchungsbereiche, ohne dass es einer vorherigen Information und Zustimmung der DAkkS bedarf,

[Flex B] die freie Auswahl von genormten oder ihnen gleichzusetzenden Untersuchungsverfahren gestattet.

[Flex C] die Modifizierung sowie Weiter- und Neuentwicklung von Untersuchungsverfahren gestattet.

Die aufgeführten Untersuchungsverfahren sind beispielhaft. Das Medizinische Laboratorium verfügt über eine aktuelle Liste aller Untersuchungsverfahren im flexiblen Akkreditierungsbereich. Die Liste ist öffentlich verfügbar auf der Webpräsenz des Medizinischen Laboratoriums.

## Untersuchungsgebiet: Klinische Chemie

### Untersuchungsart:

#### Chromatographie (UV/VIS-, Massenspektrometrische, Fluoreszenz-Detektion) <sup>[Flex B]</sup>

| Analyt (Messgröße) | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|--------------------|--------------------------------|----------------------|
| Vitamin A          | Serum                          | HPLC-UV              |
| Vitamin E          | Serum                          | HPLC-UV              |
| Vitamin C          | Serum                          | HPLC-UV              |
| Vitamin B2 (FAD)   | EDTA-Blut                      | HPLC-FLD             |
| Vitamin B1         | EDTA-Blut                      | HPLC-FLD             |
| Vitamin B6         | EDTA-Blut                      | HPLC-FLD             |
| Methylmalonsäure   | Serum                          | LC-MS/MS             |
| 17-OH-Progesteron  | Serum                          | LC-MS/MS             |

### Untersuchungsart:

#### Elektrochemische Untersuchungen <sup>[Flex B]</sup>

| Analyt (Messgröße) | Untersuchungsmaterial (Matrix) | Untersuchungstechnik           |
|--------------------|--------------------------------|--------------------------------|
| Kalium             | Serum                          | ISE (ionenselektive Elektrode) |
| Natrium            | Serum                          | ISE (ionenselektive Elektrode) |

### Untersuchungsart:

#### Ligandenassays <sup>[Flex B]</sup>

| Analyt (Messgröße)             | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|--------------------------------|--------------------------------|----------------------|
| Anti-Müller-Hormon             | Serum                          | ECLIA                |
| β-HCG                          | Serum                          | ECLIA                |
| Carcinoembryonales Antigen     | Serum                          | ECLIA                |
| Carcinoma Antigen 15-3         | Serum                          | ECLIA                |
| Dehydroepiandrosteronsulfat    | Serum                          | ECLIA                |
| Estradiol                      | Serum                          | ECLIA                |
| Ferritin                       | Serum                          | ECLIA                |
| Follikel-stimulierendes Hormon | Serum                          | ECLIA                |
| Folsäure                       | Serum                          | ECLIA                |
| freies Thyroxin                | Serum                          | ECLIA                |
| freies Trijodthyronin          | Serum                          | ECLIA                |
| Luteinisierendes Hormon        | Serum                          | ECLIA                |
| Placental like Growth Factor   | Serum                          | ECLIA                |
| Progesteron                    | Serum                          | ECLIA                |
| Prolaktin                      | Serum                          | ECLIA                |
| Sexualhormonbindendes Globulin | Serum                          | ECLIA                |

| Analyt (Messgröße)                | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|-----------------------------------|--------------------------------|----------------------|
| Soluble Fms-like thyrosinkinase-1 | Serum                          | ECLIA                |
| Testosteron, gesamt               | Serum                          | ECLIA                |
| Thyreotropin                      | Serum                          | ECLIA                |
| Vitamin B12                       | Serum                          | ECLIA                |
| Vitamin B12, active               | Serum                          | ECLIA                |

**Untersuchungsart:**

**Spektrometrie (UV- /VIS-Photometrie)** <sup>[Flex B]</sup>

| Analyt (Messgröße) | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|--------------------|--------------------------------|----------------------|
| Bilirubin, direkt  | Serum                          | VIS-Photometrie      |
| Bilirubin, gesamt  | Serum                          | VIS-Photometrie      |
| Glucose            | Serum, Citrat-Fluorid-Blut     | UV-Photometrie       |

**Untersuchungsart:**

**Spektrometrie (Turbidimetrie)** <sup>[Flex B]</sup>

| Analyt (Messgröße)                                                 | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|--------------------------------------------------------------------|--------------------------------|----------------------|
| Alkalischen Phosphatase                                            | Serum                          | Turbidimetrie        |
| Apolipoprotein A1                                                  | Serum                          | Turbidimetrie        |
| Apolipoprotein B                                                   | Serum                          | Turbidimetrie        |
| Calcium gesamt                                                     | Serum                          | Turbidimetrie        |
| Cholesterin                                                        | Serum                          | Turbidimetrie        |
| Cholinesterase                                                     | Serum                          | Turbidimetrie        |
| C-reaktives Protein                                                | Serum                          | Turbidimetrie        |
| Creatinin                                                          | Serum                          | Turbidimetrie        |
| Creatinkinase                                                      | Serum                          | Turbidimetrie        |
| Creatinkinase Muscle-Brain type                                    | Serum                          | Turbidimetrie        |
| Cystatin C                                                         | Serum                          | Turbidimetrie        |
| Eisen                                                              | Serum                          | Turbidimetrie        |
| Gamma-Glutamyltransferase                                          | Serum                          | Turbidimetrie        |
| Gesamteiweiß                                                       | Serum                          | Turbidimetrie        |
| Glutamat-Oxalacetat-<br>Transaminase/Aspartat-<br>Aminotransferase | Serum                          | Turbidimetrie        |
| Glutamat-Pyruvat-<br>Transaminase/Alanin-<br>Aminotransferase      | Serum                          | Turbidimetrie        |
| Hämoglobin A1c                                                     | EDTA-Blut                      | Turbidimetrie        |

| Analyt (Messgröße)            | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|-------------------------------|--------------------------------|----------------------|
| Haptoglobin                   | Serum                          | Turbidimetrie        |
| Harnsäure                     | Serum                          | Turbidimetrie        |
| Harnstoff                     | Serum                          | Turbidimetrie        |
| HDL-Cholesterin               | Serum                          | Turbidimetrie        |
| Homocystein                   | EDTA-Blut, Serum               | Turbidimetrie        |
| Lactatdehydrogenase           | Serum                          | Turbidimetrie        |
| LDL-Cholesterin               | Serum                          | Turbidimetrie        |
| Lipase                        | Serum                          | Turbidimetrie        |
| Lipoprotein                   | Serum                          | Turbidimetrie        |
| Löslicher Transferrinrezeptor | Serum                          | Turbidimetrie        |
| Magnesium                     | Serum                          | Turbidimetrie        |
| Pankreas-Amylase              | Serum                          | Turbidimetrie        |
| Phosphat anorganisch          | Serum                          | Turbidimetrie        |
| Rheumafaktoren                | Serum                          | Turbidimetrie        |
| Transferrin                   | Serum                          | Turbidimetrie        |
| Triglyceride                  | Serum                          | Turbidimetrie        |

## Untersuchungsgebiet: Immunologie

### Untersuchungsart:

#### Ligandenassays <sup>[Flex B]</sup>

| Analyt (Messgröße)  | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|---------------------|--------------------------------|----------------------|
| Anti-CCP-Ak         | Serum                          | ECLIA                |
| Thyreoperoxidase-Ak | Serum                          | ECLIA                |
| TSH-Rezeptor-Ak     | Serum                          | ECLIA                |

## Untersuchungsgebiet: Humangenetik

### Untersuchungsart:

#### Molekularbiologische Untersuchungen <sup>[Flex C]</sup>

| Analyt (Messgröße)             | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                                     |
|--------------------------------|-------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------|
| ACE I-/ D-Variante (rs1799752) | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                                            |
| Alpha-Thalassämie              | EDTA-Blut; DNA                                                    | Amplikonbasierte Anreicherung,<br>Sequencing by Avidite Base<br>Chemistry (SNV, CNV), custom<br>pipeline |

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| Analyt (Messgröße)                                                                                                         | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                                     |
|----------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------|
| Alport-Syndrom                                                                                                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |
| Amyloidose                                                                                                                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |
| Angioödem                                                                                                                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |
| Angelman-Syndrom,<br>Methylierungsstatus und<br>Deletions/Duplikationsanalyse<br>(15q11-q13 region, SNRPN,<br>MAGEL2 DMRs) | EDTA-Blut; DNA                                                    | Methylierungs-sensitive Multiplex<br>Ligation dependent Probe<br>Amplification (MS-MLPA)                 |
| Anterior-Segment-Dysgenese                                                                                                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |
| Arthritis                                                                                                                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |
| Ataxie                                                                                                                     | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |
| Autoinflammatorische Syndrome                                                                                              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |
| autosomal dominante polyzystische<br>Nierenerkrankung                                                                      | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |
| Beta-Thalassämie                                                                                                           | EDTA-Blut; DNA                                                    | Amplikonbasierte Anreicherung,<br>Sequencing by Avidite Base<br>Chemistry (SNV, CNV), custom<br>pipeline |
| Bernard-Soulier-Syndrom                                                                                                    | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline              |

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| Analyt (Messgröße)                                                                                                                                                                                                                                                                                                      | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| CADASIL/ CARASIL                                                                                                                                                                                                                                                                                                        | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Charcot-Marie-Tooth-Krankheit Typ<br>1A                                                                                                                                                                                                                                                                                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | Multiplex Ligation dependent Probe<br>Amplification (MLPA)                                  |
| COMT Polymorphismus c.472G>A<br>(Catechol-O-Methyltransferase)                                                                                                                                                                                                                                                          | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| CYP19A1 Polymorphismus rs727479<br>(Aromatase)                                                                                                                                                                                                                                                                          | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| CYP2C19 Allele -<br>Medikamentenunverträglichkeit<br>(Cytochrom P-450 2C19 *2, *3, *4,<br>*5, *6, *7, *8, *17)                                                                                                                                                                                                          | EDTA-Blut; DNA                                                    | PCR, Strip-Assay                                                                            |
| CYP2C9 *2 und *3 Allele -<br>Siponimod (Cytochrom P-450 2C9)                                                                                                                                                                                                                                                            | EDTA-Blut; DNA                                                    | PCR, Strip-Assay                                                                            |
| CYP2C9 *2 und *3 Allele -<br>Siponimod (Cytochrom P-450 2C9)                                                                                                                                                                                                                                                            | EDTA-Blut; DNA                                                    | PCR, Sanger Sequenzierung                                                                   |
| CYP2D6 Polymorphismen<br>(Cytochrom P-450 2D6<br>Polymorphismen c.-1584C>G,<br>c.31G>A, c.100C>T, c.124G>A,<br>c.137_138insT, c.181-1G>C,<br>c.320C>T, c.454delT, c.505G>T,<br>c.505G>A, c.506-1G>A,<br>c.522_523ins(10nt), c.755delA,<br>c.841_843delAAG, c.886C>T,<br>c.971A>C, c.985+39G>A,<br>c.1012G>A, c.1457G>C) | EDTA-Blut; DNA                                                    | PCR, Strip-Assay                                                                            |
| Cystische Fibrose/CFTR-assozierte<br>Erkrankungen                                                                                                                                                                                                                                                                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| DPYD Polymorphismen - 5-<br>Fluorouracil-Unverträglichkeit<br>(Dihydropyrimidin-Dehydrogenase<br>*2A, *13, p.D949V, HapB3<br>Typisierung)                                                                                                                                                                               | EDTA-Blut; DNA                                                    | PCR, Strip-Assay                                                                            |
| Ehlers-Danlos-Syndrom                                                                                                                                                                                                                                                                                                   | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

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| Analyt (Messgröße)                | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|-----------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Eizellreifungsdefekte             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Eisenrefraktäre Eisenmangelanämie | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Faktor V Leiden-Variante (rs6025) | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| Faktor II-Mangel                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Faktor V-HR2-Variante (rs1800595) | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| Faktor V-Mangel                   | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Faktor VII-Mangel                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Faktor X-Mangel                   | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Faktor XI-Mangel                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Faktor XII-Mangel                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Faktor XIII-Mangel                | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| FVIII Erhöhung                    | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

| Analyt (Messgröße)                                       | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                               |
|----------------------------------------------------------|-------------------------------------------------------------------|----------------------------------------------------------------------------------------------------|
| Familiäre Adenomatöse Polyposis                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline        |
| Familiäres Mittelmeerfieber                              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline        |
| Fanconi-Anämie                                           | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline        |
| Favismus/ G6PD-Mangel                                    | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline        |
| FGFR3 assoz. Erkrankungen                                | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline        |
| Fibrinogen-gamma-Variante<br>(rs2066865)                 | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                                      |
| Fibrinogen-Störung                                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline        |
| Fragiles-X-Syndrom                                       | EDTA-Blut; DNA                                                    | Repeat-primed PCR                                                                                  |
| GAD1 Polymorphismus c.-147G>A<br>(Glutamatdecarboxylase) | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                                      |
| Gastrointestinaler Stromatumor                           | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline        |
| Tumordiagnostik (Mutation)                               | Gewebeproben <sup>a</sup> ; DNA                                   | Amplikonbasierte Anreicherung,<br>Sequencing by Avidite Base<br>Chemistry (SNV), in-house pipeline |
| Tumordiagnostik (Translokation)                          | Gewebeproben <sup>a</sup> ; RNA, DNA                              | Amplikonbasierte Anreicherung,<br>Sequencing by Avidite Base<br>Chemistry (SNV), in-house pipeline |
| Mammakarzinom (Mutation)                                 | Blut-Plasma; cfDNA                                                | Amplikonbasierte Anreicherung,<br>Sequencing by Avidite Base<br>Chemistry (SNV), in-house pipeline |

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| Analyt (Messgröße)                                     | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|--------------------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Geschlechtsdifferenzierungsstörungen                   | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Gorlin-Goltz-Syndrom                                   | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| GPIa-Variante (rs1126643)                              | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| Habituelle Aborte                                      | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hämochromatose seltene Formen                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hämophilie A                                           | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hämophilie B                                           | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hämophilie                                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäre Hauttumore                                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäre neuroendokrine Tumore                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäre Neuropathie mit<br>Neigung zu Druckläsionen | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | Multiplex Ligation dependent Probe<br>Amplification (MLPA)                                  |
| Hereditäre Pankreatitis                                | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

| Analyt (Messgröße)                                    | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|-------------------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Hereditärer Darmkrebs                                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Endometriumkarzinom                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Hypophysenadenom                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Magenkarzinom                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Mamma- und<br>Ovarialkarzinom             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Mammakarzinom bei<br>Männern              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Nierenkarzinom                            | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Ovarialkarzinom                           | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Pankreaskarzinom                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Paragangliom-<br>/Phäochromozytom-Syndrom | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hereditäres Prostatakarzinom                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

Anlage zur Akkreditierungsurkunde D-ML-22009-01-01

| Analyt (Messgröße)                                    | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|-------------------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Hereditäres Schilddrüsenkarzinom                      | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| HFE-Varianten C282Y / H63D<br>(rs1800562 / rs1799945) | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| Hörstörung (AR) Typ IA/IB (GJB2,<br>GJB6)             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | Multiplex Ligation dependent Probe<br>Amplification (MLPA)                                  |
| Hörstörung nicht syndromal (AR)<br>Typ IA             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | PCR, Sanger Sequenzierung                                                                   |
| HPA-I-Ia/1b-Variante (rs5918)                         | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| Hyperbilirubinämie                                    | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hypercholesterinämie                                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hyperlipidämie                                        | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hyperparathyreoidismus                                | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hyperthyreose                                         | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hypertriglyceridämie                                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hypogonadotroper<br>Hypogonadismus                    | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hypokalziurische Hyperkalzämie                        | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

| Analyt (Messgröße)                           | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|----------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Hypophosphatasie                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Hypothyreose                                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Klinisches Exom                              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Komb. FVIII-/ Faktor V-Mangel                | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Kongenitale Kontrakturale<br>Arachnodaktylie | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Legius Syndrom                               | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Leiomyomatosis                               | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Li-Fraumeni-Syndrom                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV)<br>custom pipeline  |
| Loeys Dietz Syndrom                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Lynch-Syndrom                                | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Männliche Infertilität                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

Anlage zur Akkreditierungsurkunde D-ML-22009-01-01

| Analyt (Messgröße)                                                                       | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|------------------------------------------------------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| MAOA uVNTR-Polymorphismus<br>(Monoaminoxidase-A;<br>Neurotransmitter)                    | EDTA-Blut; DNA                                                    | PCR, Sanger Sequenzierung                                                                   |
| Marfan-Syndrom                                                                           | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Meduläres Schilddrüsenkarzinom                                                           | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Mesomele Dysplasie (SHOX)                                                                | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | Multiplex Ligation dependent Probe<br>Amplification (MLPA)                                  |
| MODY                                                                                     | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Molenschwangerschaft                                                                     | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Morbus Osler                                                                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Morbus Wilson                                                                            | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| MTHFR Polymorphismus c.665C>T<br>(aka C677T) (Methylen-<br>Tetrahydrofolat; Methotrexat) | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| Muir-Torre-Syndrom                                                                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Multiple Endokrine Neoplasie Typ 1                                                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Multiple Endokrine Neoplasie Typ 2                                                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

| Analyt (Messgröße)                   | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|--------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Muskeldystrophie                     | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Muskeldystrophie Duchenne/<br>Becker | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Nephroblastom/ Wilms-Tumor           | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Neurofibromatose Typ 1               | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Neurofibromatose Typ 2               | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Neutropenie                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Nicht-obstruktive Azoospermie        | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Nierenerkrankungen                   | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Noonan-Syndrom                       | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Obstruktive Azoospermie              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Osteogenesis imperfecta              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

Anlage zur Akkreditierungsurkunde D-ML-22009-01-01

| Analyt (Messgröße)                                                                                                          | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|-----------------------------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Ovariale Dysgenese                                                                                                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| PAI-1-4G/5G-Variante (rs1799889)                                                                                            | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| PARP-Inhibitoren                                                                                                            | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Pendred-Syndrom                                                                                                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Periodische Fiebersyndrome                                                                                                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Peutz-Jeghers                                                                                                               | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Phenylketonurie                                                                                                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Plasminogen-Mangel                                                                                                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Polyzystisches Ovarialsyndrom                                                                                               | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Prader-Willi-Syndrom (15q11-q13<br>region, SNRPN, MAGEL2 DMRs),<br>Methylierungsstatus und<br>Deletions/Duplikationsanalyse | EDTA-Blut; DNA                                                    | Methylierungs-sensitive Multiplex<br>Ligation dependent Probe<br>Amplification (MS-MLPA)    |
| Prämatüre Ovarialinsuffizienz                                                                                               | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

Anlage zur Akkreditierungsurkunde D-ML-22009-01-01

| Analyt (Messgröße)                                      | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|---------------------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Primäre Erythromelalgie                                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Primäre ziliäre Dyskinesie                              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Prothrombin-Variante (rs1799963)                        | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| Protein C-Mangel                                        | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Protein C-Rezeptordefizienz                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Protein S-Mangel                                        | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Protein Z-Mangel                                        | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| PTEN-Hamartom-Tumorsyndrom<br>und Differentialdiagnosen | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| RASopathien                                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Schilddrüsenhormonresistenz                             | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Schlaganfall oder Schlaganfall-<br>ähnliche Episoden    | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Schwannomatose                                          | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

Anlage zur Akkreditierungsurkunde D-ML-22009-01-01

| Analyt (Messgröße)                                                                     | Untersuchungsmaterial<br>(Eingangsmaterial; ggf.<br>Testmaterial) | Untersuchungstechnik                                                                        |
|----------------------------------------------------------------------------------------|-------------------------------------------------------------------|---------------------------------------------------------------------------------------------|
| Small Fiber Neuropathie                                                                | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Smith-Lemli-Opitz Syndrom                                                              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| SOD2 Polymorphismus Ala16Val<br>(Superoxiddismutase 2)                                 | EDTA-Blut; DNA                                                    | Real-Time PCR                                                                               |
| Spermiendefekte (Form und<br>Beweglichkeit)                                            | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Thorakales Aortenaneurysma und<br>Aortendissektion                                     | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Thrombozytopathie/<br>Thrombozytopenie                                                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Thrombomodulin-Defekt                                                                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| TSH-Rezeptordefizienz                                                                  | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| Tuberöse Sklerose                                                                      | EDTA-Blut, Heparin-Blut,<br>Mundschleimhaut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| VKORC1 Polymorphismus c.-<br>1639G>A (Vitamin-K-Epoxid-<br>Reduktase; Cumarinderivate) | EDTA-Blut; DNA                                                    | PCR, Strip-Assay                                                                            |
| Von Hippel-Lindau-Syndrom                                                              | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |
| von Willebrand-Syndrom                                                                 | Mundschleimhaut, EDTA-Blut,<br>Heparin-Blut; DNA                  | in-solution capture, Sequencing by<br>Avidite Base Chemistry (SNV, CNV),<br>custom pipeline |

| Analyt (Messgröße)           | Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial) | Untersuchungstechnik                                                                  |
|------------------------------|-------------------------------------------------------------|---------------------------------------------------------------------------------------|
| Weibliche Infertilität       | Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA               | in-solution capture, Sequencing by Avidite Base Chemistry (SNV, CNV), custom pipeline |
| Whole exome sequencing (WES) | Mundschleimhaut, EDTA-Blut, Heparin-Blut; DNA               | in-solution capture, Sequencing by Avidite Base Chemistry (SNV, CNV), custom-Pipeline |

<sup>a</sup>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.

### Untersuchungsart:

#### Chromosomenanalyse <sup>[Flex C]</sup>

| Analyt (Messgröße)          | Untersuchungsmaterial (Matrix)           | Untersuchungstechnik                                                                             |
|-----------------------------|------------------------------------------|--------------------------------------------------------------------------------------------------|
| angeborener Chromosomensatz | Mundschleimhaut, EDTA-Blut, Heparin-Blut | molekulare Karyotypisierung, in-solution capture, sequencing by synthesis (CNV), custom-Pipeline |

### Untersuchungsgebiet: Mikrobiologie

#### Untersuchungsart:

#### Ligandenassays <sup>[Flex B]</sup>

| Analyt (Messgröße)        | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|---------------------------|--------------------------------|----------------------|
| Treponema pallidum (TPHA) | Serum                          | ECLIA                |
| Toxoplasma gondii Ak-IgG  | Serum                          | ECLIA                |
| Toxoplasma gondii Ak-IgM  | Serum                          | ECLIA                |

### Untersuchungsgebiet: Virologie

#### Untersuchungsart:

#### Ligandenassays <sup>[Flex B]</sup>

| Analyt (Messgröße)               | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|----------------------------------|--------------------------------|----------------------|
| Cytomegalievirus IgG-Ak          | Serum                          | ECLIA                |
| Cytomegalievirus IgM-Ak          | Serum                          | ECLIA                |
| Hepatitis-A-Virus IgM/IgG-Ak     | Serum                          | ECLIA                |
| Hepatitis-B-Virus Oberflächen-Ag | Serum                          | ECLIA                |
| Hepatitis-B-Virus Oberflächen-Ak | Serum                          | ECLIA                |
| Hepatitis-C-Virus-Ak             | Serum                          | ECLIA                |

| Analyt (Messgröße) | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|--------------------|--------------------------------|----------------------|
| HIV 1/2 p24 screen | Serum                          | ECLIA                |
| Rötelnvirus IgG-Ak | Serum                          | ECLIA                |
| Rötelnvirus IgM-Ak | Serum                          | ECLIA                |

## Untersuchungsgebiet: Transfusionsmedizin

### Untersuchungsart:

#### Molekularbiologische Untersuchungen <sup>[Flex B]</sup>

| Analyt (Messgröße)            | Untersuchungsmaterial (Matrix) | Untersuchungstechnik |
|-------------------------------|--------------------------------|----------------------|
| Typisierung der Rhesus-Formel | EDTA-Blut                      | SSP-PCR              |
| Rhesus weak D Typisierung     | EDTA-Blut                      | SSP-PCR              |
| Rhesus partial D Typisierung  | EDTA-Blut                      | SSP-PCR              |
| ABO Typisierung               | EDTA-Blut                      | SSP-PCR              |
| KEL Typisierung               | EDTA-Blut                      | SSP-PCR              |
| Fetales Rhesus D              | fetale ccfDNA, EDTA-Blut       | Real-Time PCR        |