

Innerhalb der mit \* gekennzeichneten Untersuchungsbereiche ist dem Laboratorium, ohne dass es einer vorherigen Information und Zustimmung der Deutschen Akkreditierungsstelle GmbH bedarf, die freie Auswahl von genormten oder ihnen gleichzusetzenden Untersuchungsverfahren gestattet.

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

**Farblich** markierte Analyte wurden seit der letzten Begutachtung (durch die DAkKS) neu aufgenommen

### Untersuchungsart:

#### Molekularbiologische Untersuchungen (Amplifikationsverfahren)\*\*

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
ABCA4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0397_ABCA4 Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), ViiA 7 Real-Time PCR System (ThermoFisher Scientific)	02	18.04.2024		X
ABCB4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-1757_ABCB4 Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), ViiA 7 Real-Time PCR System (ThermoFisher Scientific)	01	05.11.2020		X
ABCC6	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0680_ABCC6 Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	06.04.2022		X
ABCC8	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0251_ABCC8 Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	01.10.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
ABCD1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0339_ABCD1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	06.05.2024		X
ACAD9	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1273_ACAD9-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.04.2019		X
ACADM	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0399_ACADM-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	20.03.2022		X
ACADVL	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0400_ACADVL-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	02.05.2023		X
ACTA2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0018_ACTA2-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	14.05.2019		X
ACVRL1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0374_ACVRL1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
ADAMTSL4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG- 1367_ADAMTSL4- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	08.04.2019		X
AGL	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0185_AGL- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	04.03.2026		X
ALDOB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0311_ALDOB- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	06.02.2025		X
ALPL	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0503_ALPL- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	04	15.04.2024		X
AMPD1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0389_AMPD1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	19.02.2026		X
ANKRD11	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0677_ANKRD11- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	04	24.06.2022		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
APC	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0022_APC- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
APOB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1388_APOB- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	04.12.2025		X
APOE	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0367_APOE- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	09.02.2021		X
ARID1B	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0998_ARID1B- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	04	07.10.2022		X
ARSA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0023_ARSA- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	06	11.11.2024		X
ASAH1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0484_ASAH1 Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	04	25.11.2021		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
ASL	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1767_ASL-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	29.01.2021		X
ASPA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0691_ASPA-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	08.05.2020		X
ATM	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0290_ATM-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	04	18.04.2024		X
ATP7B	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0265_ATP7B-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	02.01.2025		X
ATRX	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0693_ATRX-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	30.11.2021		X
AUTS2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1115_AUTS2-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	02.04.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
AZF	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	PCR, Elektrophorese	OS-MG-0024_AZF- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	18.07.2019		X
BARD1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 1112_BARD1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	12.06.2024		X
BCHE	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0663_BCHE- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	01.12.2020		X
BCKDHA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0106_BCKDHA- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
BCKDHB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0085_BCKDHB- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	09.03.2026		X
BRAF	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0119_BRAF- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
BRCA1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0026_BRCA1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	04	11.03.2026		X
BRCA2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0027_BRCA2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	22.07.2019		X
BRIP1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1728_BRIP1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	05.12.2019		X
BTD	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0358_BTD- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	05	23.02.2016		X
CACNA1A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0191_CACNA1A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	04	08.11.2019		X
CACNA1S	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0406_CACNA1S- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	08.04.2022		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
CAPN3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0407_CAPN3- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
CASR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1203_CASR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	03.04.2019		X
CBS	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0588_CBS- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	24.02.2026		X
CCM2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0657_CCM2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	08.05.2020		X
CDH1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0098_CDH1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
CDKL5	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0028_CDKL5- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	09.11.2021		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
CDKN2A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0100_CDKN2A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
CEL	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1208_CEL- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	20.02.2025		X
CEP290	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0192_CEP290- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	08.09.2022		X
CFTR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0029_CFTR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	02.04.2019		X
CHD7	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0030_CHD7- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	05	09.06.2021		X
CHEK2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0193_CHEK2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	05	02.10.2023		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
CLCN1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0408_CLCN1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), ViiA 7 Real- Time PCR System (ThermoFisher Scientific)	01	30.03.2019		X
COL3A1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0031_COL3A1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
COL4A1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0123_COL4A1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	04	17.06.2019		X
COL4A2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG- 0669_COL4A2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	20.05.2019		X
COL4A5	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0034_COL4A5- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
COL5A1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0035_COL5A1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahre
COL9A3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG- 0756_COL9A3- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	01.04.2019		X
CPT1A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0907_CPT1A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	26.02.2026		X
CPT2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0414_CPT2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
CTNNB1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0997_CTNNB1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	20.05.2021		X
CTNS	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0264_CTNS- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	09.09.2024		X
CUBN	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0704_CUBN- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	01.04.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
DHCR7	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0617_DHCR7- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	27.05.2021		X
DICER1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 1010_DICER1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	02.04.2023		X
DiGeorge- Syndrom	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	MLPA	AA-MG-011_MLPA	ABI 3500xl DNA Analyzer (ThermoFisher Scientific)	05	27.10.2023		X
DMD	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0039_DMD- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
DSC2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0706_DSC2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	08.06.2021		X
DSP	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0382_DSP- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
DYNC1H1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 1182_DYNC1H1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	29.10.2024		X
DYSF	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0354_DYSF- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	16.10.2024		X
ECM1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0955_ECM1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	30.07.2025		X
EGR2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0416_EGR2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
EHMT1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0206_EHMT1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	21.10.2019		X
ENG	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0207_ENG- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
ENPP1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1539_ENPP1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	08.04.2019		X
ETFDH	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0709_ETFDH-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	05	24.02.2026		X
F2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0129_F2-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	09.05.2023		X
F5	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0130_F5-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	30.06.2020		X
F7	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1560_F7-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	26.01.2022		X
F9	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0806_F9-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	01.04.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
FAH	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0418_FAH-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	04.12.2025		X
Familiärer Brust- und Eierstockkrebs (BRCA1, BRCA2)	EDTA-Blut, eingesandte DNA	Sequence capture (SYC, TWIST), Sequencing-by-Synthesis, Varvis	AA-MG-053_Cancer Panel SYC1 Twist	Twist	01	05.02.2025	X	
Familiäres Tumorsyndrom (ATM, BARD1, BRIP1, CDH1)	EDTA-Blut, eingesandte DNA	Sequence capture (SYC, TWIST), Sequencing-by-Synthesis, Varvis	AA-MG-053_Cancer Panel SYC1 Twist	Twist	01	05.02.2025	X	
FBN1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0040_FBN1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	05	30.10.2023		X
FBP1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0319_FBP1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	23.07.2019		X
FGA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1414_FGA-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	12.08.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
FGB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1439_FGB-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	10.07.2024		X
FGD1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0515_FGD1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	04	04.12.2025		X
FGF10	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0627_FGF10-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	01.04.2019		X
FGFR3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0320_FGFR3-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	13.07.2023		X
FGG	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1460_FGG-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	20.05.2020		X
FH	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0713_FH-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	15.06.2023		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
FLCN	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1086_FLCN- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	02.04.2019		X
FLG	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0134_FLG- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	28.08.2019		X
FLT4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1098_FLT4- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	04	04.05.2022		X
FOXC1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0089_FOXC1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
FOXE3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1019_FOXE3- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	28.08.2019		X
FTL	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0494_FTL- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	12.01.2023		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
G6PC	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0310_G6PC-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	05.05.2020		X
G6PD	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1183_G6PD-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	08.07.2024		X
GAA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0420_GAA-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	12.06.2023		X
GALC	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0091_GALC-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	30.03.2019		X
GALK1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1157_GALK1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	19.02.2026		X
GALT	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0714_GALT-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	27.04.2023		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
GATA1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1646_GATA1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	19.06.2024		X
GBE1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0093_GBE1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	24.02.2026		X
GCDH	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0422_GCDH-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
GCH1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0321_GCH1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	26.08.2024		X
GCK	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0155_GCK-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	05.09.2023		X
GDF1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0911_GDF1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	08.10.2020		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
GJA1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0137_GJA1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	20.05.2020		X
GJB2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0037_GJB2-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	11.03.2026		X
GJB3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0276_GJB3-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
GJB6	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0209_GJB6-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	24.02.2025		X
GLA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0042_GLA-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	25.05.2020		X
GLDC	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1462_GLDC-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	04.12.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
GLMN	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0715_GLMN-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	08.07.2025		X
GNPTAB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0138_GNPTAB-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	04.12.2025		X
HBA1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1714_HBA1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	18.11.2019		X
HBA2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1715_HBA2-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	25.04.2024		X
HBB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0719_HBB-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	23.11.2021		X
HEXA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0487_HEXA-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	24.02.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
HEXB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0335_HEXB-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
HFE	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0043_HFE-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	04.04.2023		X
HGD	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1690_HGD-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	12.07.2019		X
HPRT1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0614_HPRT1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	29.01.2026		X
HTRA1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0428_HTRA1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	09.05.2023		X
IDS	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0213_IDS-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	17.05.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
IDUA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0082_IDUA-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	31.07.2023		X
IGF1R	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0558_IGF1R-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
IKBKG	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1377_IKBKG_Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	03.07.2025		X
INSR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0109_INSR-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	04	24.02.2026		X
ITGB3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1522_ITGB3-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	19.02.2026		X
IVD	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0214_IVD-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	08.07.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
KCNQ1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0847_KCNQ1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	24.02.2025		X
KIF7	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0522_KIF7- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
KIT	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0523_KIT- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	20.02.2025		X
KMT2A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0674_KMT2A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	11.11.2020		X
KRIT1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0633_KRIT1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	12.06.2024		X
KRT5	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0286_KRT5- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
KRT6A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0592_KRT6A-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
LAMB3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0220_LAMB3-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	19.11.2024		X
LDLR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0961_LDLR-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	01.04.2019		X
LEMD3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0221_LEMD3-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	24.02.2026		X
LMX1B	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0150_LMX1B-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
Lynch-Syndrom (HNPCC) (MLH1, MSH2, MSH6,	EDTA-Blut, eingesandte DNA	Sequence capture (SYC, TWIST), Sequencing-by-Synthesis, Varvis	AA-MG-053_Cancer Panel SYC1 Twist	MiSeq (Illumina)	01	05.02.2025	X	

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
MCCC1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-0613_MCCC1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	05.03.2026		X
MCCC2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0621_MCCC2-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	24.06.2024		X
MEFV	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0365_MEFV-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
MEN1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0923_MEN1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	01.04.2019		X
MITO-Genom	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1763_MITO-Genom-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	15.01.2021		X
MLH1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0049_MLH1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	12.10.2021		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
MMACHC	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0493_MMACHC- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	04.12.2025		X
MSH2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0050_MSH2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	23.03.2023		X
MSH6	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0105_MSH6- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	10.10.2023		X
MTHFR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0925_MTHFR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	27.04.2023		X
MT-ND1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0735_MT- ND1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	20.12.2020		X
MT-ND3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0439_MT- ND3- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
MT-ND4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1763_MT- ND4- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	15.01.2021		X
MT-ND6	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1763_MT- ND6- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	15.01.2021		X
MTR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0396_MTR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
MTRR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0858_MTRR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	01.04.2019		X
MT-TL1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0369_MT- TL1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
MUTYH	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0051_MUTYH- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahre
MYBPC3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0739_MYBPC3- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	27.02.2025		X
MYCN	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0052_MYCN- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	02.10.2024		X
MYH11	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0103_MYH11- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	05.03.2026		X
MYH7	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0440_MYH7- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
MYT1L	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1301_MYT1L Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	05.04.2019		X
NAGLU	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0160_NAGLU- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	24.04.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
NBN	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1526_NBN- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	19.06.2024		X
NEB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0331_NEB- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	05.06.2020		X
NF1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0053_NF1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	04	30.04.2025		X
NF2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0360_NF2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	23.04.2025		X
NFKB1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1616_NFKB1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	17.06.2022		X
NOD2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0963_NOD2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	01.04.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
NOTCH3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0226_NOTCH3- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	04	07.12.2023		X
NPC1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0323_NPC1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	19.02.2025		X
NRXN1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0227_NRXN1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	12.07.2019		X
NSD1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0228_NSD1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
OTC	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0631_OTC- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	08.05.2020		X
PABPN1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG- 0309_PABPN1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	24.04.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
PAH	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1039_PAH- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	09.08.2022		X
PALB2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-1126_PALB2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	11.01.2022		X
PCCA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0463_PCCA- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	03.03.2025		X
PCCB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0607_PCCB- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	04.12.2025		X
PCDH19	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0231_PCDH19- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	25.05.2020		X
PHEX	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0598_PHEX- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	03.03.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
PHF8	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0661_PHF8- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	01.04.2019		X
PIEZO2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 1193_PIEZO2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	02.04.2019		X
PKD1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1026_PKD1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	14.10.2020		X
PKD2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1130_PKD2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	16.04.2020		X
PKP2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0383_PKP2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	20.01.2025		X
PMM2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1252_PMM2 Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	09.12.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
PMS2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0101_PMS2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	19.01.2021		X
PNPT1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1674_PNPT1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	08.04.2019		X
POLD1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1055_POLD1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	02.04.2019		X
POLE	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1075_POLE- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	16.10.2024		X
POLR3A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0801_POLR3A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	16.04.2025		X
POMT1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0646_POMT1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	01.04.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo-versionssheet	letzte Änderung	CE-Verfahren	in house-Verfahren
Prader-Willi-Syndrom (PWS) / Angelman-Syndrom (AS)	EDTA-Blut, Mundschleimhautzellen, Gewebeprobe, eingesandte DNA, Amnionzellen, Chorionzellen	methylierungssensitive MLPA	AA-MG-011_MS_MS-MLPA	ABI 3500xl DNA Analyzer (ThermoFisher Scientific)	05	27.10.2023		X
PRKAG2	EDTA-Blut, Mundschleimhautzellen, Gewebeprobe, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0648_PRKAG2-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	06.05.2024		X
PRNP	EDTA-Blut, Mundschleimhautzellen, Gewebeprobe, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0240_PRNP-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	06.02.2025		X
PROC	EDTA-Blut, Mundschleimhautzellen, Gewebeprobe, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real-Time PCR	OS-MG-1158_PROC-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	16.11.2023		X
PROS1	EDTA-Blut, Mundschleimhautzellen, Gewebeprobe, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1356_PROS1-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	16.09.2024		X
PRRT2	EDTA-Blut, Mundschleimhautzellen, Gewebeprobe, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0436_PRRT2-Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
PTCH1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0055_PTCH1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
PTEN	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0242_PTEN- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	03.06.2024		X
PTH1R	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0968_PTH1R- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	02.10.2025		X
PTPN11	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0056_PTPN11- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	01.02.2022		X
PTS	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-1141_PTS- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	26.10.2024		X
QDPR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0791_QDPR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	29.08.2024		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
RAD51C	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0244_RAD51C- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	08.06.2021		X
RAD51D	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0877_RAD51D- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	25.05.2020		X
RASA1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1092_RASA1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	04.04.2023		X
RBM8A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0753_RBM8A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	28.01.2021		X
RET	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0057_RET- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
RNASEH2B	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0019_RNASEH2B- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	04.05.2020		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
RS1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1642_RS1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	08.04.2019		X
RYR1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0328_RYR1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	16.04.2020		X
SALL1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0058_SALL1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	07.08.2019		X
SALL4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0059_SALL4- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	28.09.2023		X
SBDS	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0628_SBDS- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	04	09.03.2021		X
SCN1A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0060_SCN1A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	16.06.2020		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
SCN4A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0455_SCN4A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
SCN5A	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0456_SCN5A- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
SDHA	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1241_SDHA- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	03.04.2019		X
SERPINA1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0612_SERPINA1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	09.06.2021		X
SERPINC1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 1154_SERPINC1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	17.05.2022		X
SERPINE1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG- 1037_SERPINE1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	02.04.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahre
SERPING1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0969_SERPING1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	01.04.2019		X
SETD2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1300_SETD2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	05.04.2019		X
SGSH	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0175_SGSH- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
SHANK3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0937_SHANK3- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	25.04.2023		X
SHOX	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0888_SHOX- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	01.04.2019		X
SLC12A3	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 1045_SLC12A3- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	02.04.2019		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
SLC16A2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0295_SLC16A2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	08.05.2020		X
SLC22A5	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 1378_SLC22A5- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	08.04.2019		X
SLC2A1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0061_SLC2A1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
SLC37A4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0461_SLC37A4- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	02.03.2026		X
SLC40A1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0766_SLC40A1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	24.02.2024		X
SMAD4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 1008_SMAD4- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	02.04.2023		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
SOS1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0063_SOS1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
SPAST	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0465_SPAST- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
SPG7	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0528_SPG7- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	26.02.2025		X
STK11	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0609_STK11- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	30.03.2019		X
STRC	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1586_STRC- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	27.04.2020		X
STXBP1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG- 0384_STXBP1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	19.02.2025		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
SYNGAP1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0971_SYNGAP1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	17.10.2019		X
TGFBR1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG- 0067_TGFBR1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	09.10.2023		X
THRB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0774_THRB- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	19.05.2020		X
TMC6	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1078_TMC6- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	02.04.2019		X
TNXB	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0178_TNXB- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	13.04.2023		X
TP53	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0069_TP53- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	02	01.06.2021		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahre
TREX1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0071_TREX1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	13.06.2024		X
TRPV4	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0550_TRPV4- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	03.03.2025		X
TSC1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0257_TSC1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	03	07.05.2020		X
TSC2	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-0258_TSC2- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	30.03.2019		X
TSEN54	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG- 1615_TSEN54- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	17.06.2021		X
TTR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-0898_TTR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	13.10.2021		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionssheet	letzte Änderung	CE- Verfahren	in house- Verfahren
TUBB2B	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG- 1579_TUBB2B- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	08.04.2019		X
TYR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0479_TYR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	01	30.03.2019		X
UBA1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung	OS-MG-1792_UBA1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	01	06.04.2022		X
UGT1A1	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG- 0391_UGT1A1- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	04	17.07.2023		X
VDR	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; quantitative Real-Time PCR	OS-MG-0777_VDR- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	24.02.2026		X
VHL	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA; quantitative Real- Time PCR	OS-MG-0073_VHL- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina), CFX Opus 384 Real-Time PCR System (BioRad)	03	02.05.2023		X

Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung	Gerät	Oligo- versionsheet	letzte Änderung	CE- Verfahren	in house- Verfahre
VWF	EDTA-Blut, Mundschleimhautzellen, Gewebeproben, eingesandte DNA, Amnionzellen, Chorionzellen	NGS: PCR, Sequencing by synthesis, SEQNEXT; Sanger-Sequenzierung; MLPA	OS-MG-1451_VWF- Oligonucleotide	ABI 3500xl DNA Analyzer (ThermoFisher Scientific), MiSeq (Illumina)	02	05.06.2019		X