

Untersuchungsgebiet: Humangenetik (Molekulare Humangenetik)
Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Indikation und Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	Ansatztage
Aortenaneurysma (<i>ACTA2, COL3A1, FBN1, MYH11, MYLK, SMAD3, TGFB2, TGFB1, TGFB2</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Arrhythmogene rechtsventrikuläre Kardiomyopathie (ARVD; <i>DSG2, DSP, FLNC, JUP, PKP2, PLN, CDH2, CTNNA3, DES, TGFB3, TMEIM43</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Azoospermie (Azoospermiefaktor (AZF)-Deletion)	EDTA-Blut, DNA; DNA	PCR, Agarosegelelektrophorese	RH-MG-SOP-Multiplex-PCR (LDT) - 3	Thermocycler Gelelektrophoresekammer	wöchentlich
Behçet, Morbus (HLA-B51)	EDTA-Blut, DNA; DNA	PCR, Agarosegelelektrophorese	RH-MG-SOP-HLA-Diagnostik (LDT) - 5	Thermocycler Gelelektrophoresekammer	wöchentlich
BRCA-Diagnostik vor PARP-Inhibitoren-Therapie (<i>BRCA1, BRCA2</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Brugada-Syndrom (<i>SCN5A, CACNA1C, CACNB2, GPD1L, HCN4, KCND3, KCNE3, SCN1B, SCN3B, TRPM4</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Brust- und Ovarialkarzinom, hereditär (HBOC; <i>BRCA1, BRCA2, CHEK2, PALB2, RAD51C, TP53</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Brust- und Ovarialkarzinom, hereditär, erweiterte Diagnostik (<i>ATM, BARD1, BRIP1, CDH1, PTEN, STK11, RAD51D, SMARCA4</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Catecholaminerge polymorphe ventrikuläre Tachykardie (<i>RYR2, CALM1, CALM2, CALM3, CASQ2, KCNJ2, TECL, TRDN</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Cystische Fibrose (Mukoviszidose und CBAVD; <i>CFTR</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig
Dilatative Kardiomyopathie (<i>TTN, LMNA, MYBPC3, MYH6, MYH7, TNNT2, ACTC1, ACTN2, BAG3, DES, DSP, FLNC, JPH2, MYPN, NEXN, PLN, RBM20, SCN5A, TNNC1, TNNI3, TPM1, VCL</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Ehlers-Danlos-Syndrom (<i>COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, TNXB, ADAMTS2, AEBP1, FKBP14, PLOD1, SL39A13</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Ehlers-Danlos-Syndrom (seltene Formen inkl. Brittle-Cornea-Syndrom) (<i>B3GALT6, B4GALT7, C1R, C1S, CHST14, COL12A1, DSE, PRDM5, ZNF469</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Familiäre adenomatöse Polyposis coli (FAP/MAP; <i>APC, MUTYH</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Familiäre Hypercholesterinämie (<i>LDLR, APOB, LDLRAP1, PCSK9</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Familiäre juvenile Polyposis (FJP; <i>BMPR1A, SMAD4</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig

Hintergrundliste ML

geänderte Parameter

nicht mehr durchgeführte Parameter

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Indikation und Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	Ansatztage
Fragiles-X-assoziiertes Tremor-Ataxie-Syndrom (FXTAS, <i>FMR1</i>)	EDTA-Blut, DNA; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (ggf. Southern-Blot)	RH-MG-SOP-Fragmentlängenanalyse (LDT) - 5	ABI310 Thermocycler	monatlich
Fragiles-X-Syndrom (<i>FMR1</i>)	EDTA-Blut, DNA; DNA	größenspezifische DNA-Fragmentanalyse in Gelmatrix (ggf. Southern-Blot)	RH-MG-SOP-Fragmentlängenanalyse (LDT) - 5	ABI310 Thermocycler	monatlich
Gonadendysgenese (<i>SRY</i>)	EDTA-Blut, DNA; DNA	PCR, Agarosegelelektrophorese	RH-MG-SOP-Multiplex-PCR (LDT) - 3	Thermocycler Gelelektrophoresekammer	wöchentlich
Hämoglobin-Genotypisierung (Thalassämie-alpha (<i>HBA1, HBA2</i>) und Thalassämie-beta (<i>HBB</i>))	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger), MLPA	RH-MG-SOP-Sanger-Sequenzierung (LDT) - 4	Thermocycler ABI310, ABI3100	wöchentlich
Hereditäres Non-Polypöses Colon-Karzinom (HNPCC/ Lynch-Syndrom; <i>MLH1, PMS2, MSH6, MSH2</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Hereditäres Periodisches Fieber (HPF, hereditäre Fiebersyndrome; <i>MEFV, ADA2, ELANE, IL1RN, IL36RN, LPIN2, MVK, NLR4, NLRP12, NLRP3, NOD2, PSMB8, PSTPIP1, TNFRSF1A</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq Thermocycler	14-tägig
Hypertrophe Kardiomyopathie (<i>MYBPC3, MYH7, TNNT3, TNNT2, ACTC1, ACTN2, ALPK3, CSRP3, JPH2, MYL2, MYL3, PLN, TNNC1, TPM1</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Kolonkarzinom (<i>TP53, CHEK2, MUTYH, POLE, POLD1, PTEN, STK11</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Kolonkarzinom mit Polyposis (<i>APC, BMPR1A, MUTYH, SMAD4, GREM1, MSH3, NTHL1, PTEN, POLD1, POLE, STK11</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Li-Fraumeni-Syndrom / TPDS (<i>TP53, BAP1, CHEK2, POT1</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Loeys-Dietz-Syndrom (<i>TGFBR1, TGFBR2, SMAD2, SMAD3, TGFBR2, TGFBR3</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Long-QT-Syndrom (<i>KCNH2, KCNQ1, SCN5A, AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNJ2, KCNJ5, SCN4B, SNTA1, TRDN</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Magenkarzinom (<i>CDH1, BMPR1A, CHEK2, MLH1, MSH2, MSH6, PMS2, PTEN, SMAD4, STK11, TP53</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Magenkarzinom, erweiterte Diagnostik (<i>CDH1, APC, ATM, BMPR1A, BRCA1, BRCA2, CHEK2, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, SMAD4, STK11, TP53</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
malignes Melanom (<i>CDKN2A, CDK4, ACD, BAP1, MITF, POT1, TERF2IP, TERT</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Marfan-Syndrom (<i>FBN1, TGFBR1, TGFBR2</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
maternalen Kontaminationsausschluss	EDTA-Blut, DNA, Mundschleimhautabstrich, Gewebe; DNA	Mikro-Satellitenanalyse (Fragmentanalyse inkl. Short Tandem Repeats)	RH-MG-SOP-Mikrosatellitenanalyse (LDT) - 3	ABI310 Thermocycler	wöchentlich
Maturity Onset Diabetes of the Young (MODY; <i>HNF4A, GCK, HNF1A, PDX1, HNF1B, NEUROD1, KLF11, CEL, PAX4, INS, BLK, ABCC8, KCNJ11, APPL1</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig

Hintergrundliste ML

geänderte Parameter
nicht mehr durchgeführte Parameter

Untersuchungsart:

Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Indikation und Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	Ansatztage
Mittelmeerfieber, hereditär (<i>MEFV</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig
Morbus Wilson (<i>ATP7B</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig
Narkolepsie (Haplotyp DQB1*06:02, HLA-DR15/DQ6)	EDTA-Blut, DNA; DNA	PCR, Agarosegelelektrophorese	RH-MG-SOP-HLA-Diagnostik (LDT) - 5	Thermocycler Gelelektrophoresekammer	wöchentlich
Neurofibromatose Typ 1 (<i>NF1</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
NF1, DD Legius-Syndrom (<i>NF1, SPRED1</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Nierenkarzinom (<i>FH, FLCN, MET, CHEK2, PTEN, SMARCB1, TP53, TSC1, TSC2, VHL</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Nierenkarzinom, erweiterte Diagnostik (<i>FH, FLCN, MET, BAP1, CHEK2, DICER1, DIS3L2, MLH1, MSH2, MSH6, PMS2, PTEN, SDHB, SDHD, SMARCB1, TP53, TSC1, TSC2, VHL, WT1</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Non-Compaction Kardiomyopathie (<i>ACTC1, MYH7, PRDM16, TAZ (TAFAZIN), TPM1, ACTN2, DTNA, MIB1, LDB3, MYBPC3, TNNT2</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Noonan-Syndrom (Stufe I: <i>PTPN11</i> , Stufe II: <i>BRAF, KRAS, RAF1, RIT1, SOS1</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig
Ovarialkarzinom (<i>BRCA1, BRCA2, BRIP1, MLH1, MSH2, MSH6, PALB2, RAD51C, RAD51D, STK11</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Ovarialkarzinom - ohne <i>BRCA1 / BRCA2 (BRIP1, MLH1, MSH2, MSH6, PALB2, RAD51C, RAD51D, STK11)</i> ; SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Pankreaskarzinom, PARP-Inhibitoren-Therapie (<i>BRCA1, BRCA2</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Pankreaskarzinom (<i>BRCA1, BRCA2, CDKN2A, PALB2, STK11, TP53</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Pankreaskarzinom, erweiterte Diagnostik (<i>BRCA1, BRCA2, CDKN2A, PALB2, STK11, TP53, APC, ATM, CDC73, CHEK2, MLH1, MSH2, MSH6, PMS2, PTEN</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Pankreatitis, hereditär (<i>PRSS1, SPINK1/PSTI, CFTR, CPA1, CTRC</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig
Papilläres Schilddrüsenkarzinom, familiäres nichtmedulläres Schilddrüsenkarzinom (<i>APC, BAP1, CHEK2, DICER1, PRKAR1A, PTEN, TP53</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig

Hintergrundliste ML

geänderte Parameter

nicht mehr durchgeführte Parameter

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Indikation und Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial, ggf. Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	Ansatztage
Paragangliom-Phäochromozytom-Syndrom (<i>RET, SDHAF2, SDHA, SDHB, SDHC, SDHD, VHL, MAX, MEN1, NF1, TMEM127</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Prämature Ovarialinsuffizienz (POF/FXPOI, <i>FMR1</i>)	EDTA-Blut, DNA; DNA	größen spezifische DNA-Fragmentanalyse in Gelmatrix (ggf. Southern-Blot)	RH-MG-SOP-Fragmentlängenanalyse (LDT) - 5	ABI310 Thermocycler	monatlich
Prostatakarzinom, PARP-Inhibitoren-Therapie (<i>BRCA2, BRCA1</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Prostatakarzinom (<i>BRCA2, ATM, BRCA1</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Prostatakarzinom, erweiterte Diagnostik (<i>BRCA2, ATM, BRCA1, CDH1, CHEK2, FANCA, HOXB13, PALB2</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Protein C-Defizienz (<i>PROC</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig
Protein S-Defizienz (<i>PROS1</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig
Restriktive Kardiomyopathie (<i>TNNI3, FLNC, KIF20A, MYPN, TNNT2</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Short-QT-Syndrom (<i>KCNH2, KCNJ2, KCNQ1, CACNA1C, CACNB2</i>); SNV, CNV [Nextera Custom Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
SHOX-Defizienz (<i>SHOX</i>); SNV, CNV [QiaSeq Targeted DNA Custom Panel, Qiagen]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, amplikonbasiert; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-Qiaseq-Diagnostik (LDT) - 5	MiSeq ABI310 Thermocycler	14-tägig
Sichelzellanämie (HbS-Mutation in <i>HBB</i>)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger)	RH-MG-SOP-Sanger-Sequenzierung (LDT) - 4	Thermocycler ABI3100	wöchentlich
Sotos-Syndrom (<i>NSD1, PTEN, PTCH1, GPC3</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems)); MLPA	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Thalassämie-alpha (<i>HBA1, HBA2</i>)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger), MLPA	RH-MG-SOP-Sanger-Sequenzierung (LDT) - 4	Thermocycler ABI310, ABI3100	wöchentlich
Thalassämie-beta (<i>HBB</i>)	EDTA-Blut, DNA; DNA	DNA Sequenzierung (Sanger), MLPA	RH-MG-SOP-Sanger-Sequenzierung (LDT) - 4	Thermocycler ABI310, ABI3100	wöchentlich
Tuberöse Sklerose (<i>TSC1, TSC2</i>); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig
Tumordiagnostik (individuell); SNV, CNV [TruSight hereditary Cancer-Panel, Illumina]	EDTA-Blut, DNA; DNA	Hochdurchsatzsequenzierung (NGS, Sequence capture; Sequencing-by synthesis; MiSeq Control Software (Illumina); LocalRunManager (Illumina); SeqPilot (JSI medical Systems))	RH-MG-SOP-NGS-TruSight-Diagnostik (LDT) - 6	MiSeq ABI310 Thermocycler	14-tägig

Hintergrundliste ML

geänderte Parameter

nicht mehr durchgeführte Parameter

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Indikation und Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial, ggf. Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	Ansatztage
ABCC8	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P117)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
APC	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P043)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
ATM	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P041, P042, ggf. P190)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
ATP7B	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P098)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
BAG3	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P196)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
BMPR1A	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P158)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
BRAF	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P298, ggf. P370)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
BRCA1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P002, ggf. P087)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
BRCA2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P045, ggf. P077)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
BRIP1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P240)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
CAV3	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P048)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
CDH1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P083)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
CDK4	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P419)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
CDKN2A	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P419)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
CEL	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P357)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
CFTR	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P091)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
CHEK2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P190, ggf. P045/P056)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
COL1A1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P271)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
COL3A1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P155)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
COL5A1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P331, P332)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
CTRC	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P242)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
DiGeorge-Syndrom (Mikrodeletion 22q11.2, CATCH22)	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P250)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
DSC2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P168)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
DSG2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P168)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
DSP	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P168)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
EPCAM	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P003, ggf. P072)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
FBN1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P065, P066)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
FH	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P198)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
FLCN	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P256)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
GCK	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P241)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
GLA	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P159)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
GPC3	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P154)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
GPC4	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P154)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
GREM1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P378)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
HBA1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P140)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	wöchentlich
HBA2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P140)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	wöchentlich
HBB	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P102)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	wöchentlich
HNF1A	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P241)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
HNF1B	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P241, ggf. P357)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
HNF4A	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P241)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
INS	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P357)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
JUP	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P168)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
KCNE1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P114)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
KCNE2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P114)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
KCNH2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P114)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
KCNJ2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P114)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
KCNQ1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P114)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf

Hintergrundliste ML

geänderte Parameter

nicht mehr durchgeführte Parameter

Untersuchungsart:
Molekularbiologische Untersuchungen (Amplifikationsverfahren)

Indikation und Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial, ggf. Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	Ansatztage
KLF11	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P357)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
KRAS	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P298)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
LDLR	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P062)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
LMNA	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P048)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
MEN1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P244)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
MET	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P308)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
MLH1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P003, ggf. P248)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
MSH2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P003, ggf. P248)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
MSH6	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P072)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
MUTYH	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P043, ggf. P378/P072)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
MYBPC3	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P100)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
MYH7	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P418)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
NEUROD1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P357)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
NF1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P081, P082)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
NSD1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P026)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
PALB2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P260)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
PAX4	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P357)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
PDX1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P357)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
PKP2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P168)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
PLOD1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P359)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
PMS2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P008)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
POLD1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P492)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
POLE	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P492)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
PROC	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P265)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
PROS1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P112)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
PRSS1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P242)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
PTCH1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P067)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
PTEN	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P158)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
RAD51C	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P260)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
RAD51D	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P260)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
RYR2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P168)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
SCN5A	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P108)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
SHOX	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P018)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
SMAD4	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P158)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
SPINK1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P242)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	14-tägig
STK11	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P101)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
TGFB3	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P168)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
TGFBR2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P065)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
TNNT2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P196)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
TP53	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P056, ggf. P190)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
TSC1	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P124)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf
TSC2	EDTA-Blut, DNA; DNA	MLPA (MRC Holland P046)	RH-MG-SOP-MLPA-Diagnostik (LDT) - 3	ABI310, Thermocycler	bei Bedarf

Hintergrundliste ML

geänderte Parameter

~~nicht mehr durchgeführte Parameter~~
Untersuchungsart:
Molekularbiologische Untersuchungen (Hybridisierungsverfahren)

Indikation und Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	Ansatztage
Fragiles-X-assoziiertes Tremor-Ataxie-Syndrom (FXTAS, <i>FMR1</i>)	EDTA-Blut, DNA; DNA	Southern-Blot	RH-MG-SOP-Southern Blot, Technik 7a (LDT) - 5	Thermocycler Gelelektrophoresekammer	monatlich
Fragiles-X-Syndrom (<i>FMR1</i>)	EDTA-Blut, DNA; DNA	Southern-Blot	RH-MG-SOP-Southern Blot, Technik 7a (LDT) - 5	Thermocycler Gelelektrophoresekammer	monatlich
Prämature Ovarialinsuffizienz (POF/FXPOI, <i>FMR1</i>)	EDTA-Blut, DNA; DNA	Southern-Blot	RH-MG-SOP-Southern Blot, Technik 7a (LDT) - 5	Thermocycler Gelelektrophoresekammer	monatlich

Untersuchungsgebiet: Humangenetik (Zytogenetik)
Untersuchungsart:
Chromosomenanalyse

Indikation und Analyt (Messgröße)	Untersuchungsmaterial (Eingangsmaterial; ggf. Testmaterial)	Untersuchungstechnik	Anweisung/Version	Gerät	Ansatztage
Angeborener Chromosomensatz	Heparin-Blut, Abortgewebe; kultivierte Zellen	Chromosomenbänderungsanalyse	RH-ZG-SOP-Chromosomenbänderungsanalyse GTG (LDT)- 2	Hellfeld-Mikroskop-Systeme	2x wöchentlich
Angeborener Chromosomensatz	Heparin-Blut, Mundschleimhaut; kultivierte bzw. native Zellkerne	Fluoreszenz-in-situ-Hybridisierung (FISH)	RH-ZG-SOP-FISH an unkultivierten Zellen (LDT)- 4	Fluoreszenz-Mikroskop-System	2x wöchentlich