

<b>Leistungsspektrum molekulare Humangenetik (Analysen nach GenDG)</b>	Registrierungsnummer DAkKS <b>D-ML-13304-07-00</b> Stand 07/2024
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Analyt (Meßgröße)	Untersuchungs-material (Matrix)	Untersuchungstechnik	Anweisung/ Version
<b>Pulmonale Hypertonie</b> ( <i>BMPT2, ACVRL1, EIF2AK4, ENG, BMPR1B, CAV1, GDF2, KCNK3, SMAD4, SMAD9, TBX4, KLF2, ABCC8, AQP1, ATP13A3, BMP10, BMPR1A, FBLN2, GG CX, KDR, KLK1, NOTCH3, PDGFD, SMAD1, SOX17, TET2</i> ), SNV	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.4 PH / Core Exom Panel (1.23/rev.0)
<b>DPYD-Genotypisierung vor 5 FU Therapie</b> , SNV	EDTA Blut, DNA; DNA	Amplikon, sequencing by synthesis, Qiagen CLC Genomic Workbench Version 23.0.4	2.2.11.2.2 DPYD Qiaseq (1.23/Rev.1)
<b>Polyposissyndrome</b> ( <i>APC, BMPR1A, MUTYH, NTHL1, POLD1, POLE, PTEN, SMAD4, STK11</i> ) TDS V2 Panel (Custom made by TWIST), SNV	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)
<b>Hereditärer Darmkrebs inklusive HNPCC</b> ( <i>MLH1, MSH2, MSH6, PMS2, EPCAM, MSH3, NTHL1, PTEN, POLE, POLD1</i> ) TDS V2 Panel (Custom made by TWIST), SNV	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)
<b>HBOC- Hereditäres Mamma- und Ovarialkarzinom</b> ( <i>ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53</i> ) TDS V2 Panel (Custom made by TWIST), SNV	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)
<b>Rhabdoid-Prädispositionssyndrom Typ I und II</b> ( <i>SMARCA4, SMARCB1</i> ) TDS V2 Panel (Custom made by TWIST), SNV	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)
<b>Neurofibromatose Typ I/Typ II</b> ( <i>NF1, NF2</i> ) TDS V2 Panel (Custom made by TWIST), SNV	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)
<b>Multiple Endokrine Neoplasie - MEN Typ I und II</b> ( <i>MEN1, RET</i> ) TDS V2 Panel (Custom made by TWIST), SNV	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)

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<b>Medulläres Schilddrüsenkarzinom (RET) TDS V2 Panel (Custom made by TWIST)</b>	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)
<b>Hereditäres Phäochromozytom/Paraganliomsyndrom (SDHA, SDHB, SDHC, SDHD, SDHAF2, MAX, MEN1) TDS V2 Panel (Custom made by TWIST), SNV</b>	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)
<b>Hereditäres diffuses Magenkarzinom (CDH1, CTNNA1) TDS V2 Panel (Custom made by TWIST), SNV</b>	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.1.0 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/Rev.2)
<b>BRCA1/BRCA2</b>	EDTA Blut, DNA; DNA	MLPA, Coffalyser.Net Version 220513.1739	2.2.11.2.1.1 MLPA BRCA1+2 (2.23/rev.1)
<b>BRCA1/ BRCA2 Oncomine BRCA Research Assay by Thermo Fisher, SNV</b>	EDTA Blut, DNA; DNA	Amplikon - sequencing by synthesis - Ion Torrent-Server, coverage analysis, V5.12.0.0 und variant caller, V5.12.0.2	2.2.11.2.3 Oncomine BRCA 1+2 für KB (3.23/rev.0)

**Bitte beachten Sie unseren Einsendeschein und Einverständniserklärung nach GenDG**