

Leistungsspektrum molekulare Humangenetik (Analysen nach GenDG)	Registrierungsnummer DAkkS D-ML-13304-07-00 Stand 08/2025	Anlage 1 zur VA Humangenetik
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Analyt (Messgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung+Version
Pulmonale Hypertonie (<i>BMPR2, ACVRL1, EIF2AK4, ENG, BMPR1B, CAV1, GDF2, KCNK3, SMAD4, SMAD9, TBX4, KLF2, ABCC8, AQP1, ATP13A3, BMP10, BMPR1A, FBLN2, GGCX, KDR, KLK1, NOTCH3, PDGFD, SMAD1, SOX17, TET2</i>), SNV	EDTA-Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.4.1 (ACMG version 1-6)	2.2.11.2.4 PH / Core Exom Panel (1.23/rev.0)
DPYD-Genotypisierung vor 5 FU Therapie , (Custom made by Qiagen), SNV	EDTA-Blut, DNA; DNA	Amplikon, sequencing by synthesis, Qiagen CLC Genomic Workbench Version 23.0.4	2.2.11.2.2 DPYD Qiaseq (6.25/rev.2)
Polyposissyndrome (<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 2.4.1 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (3.23/rev.2)
Hereditärer Darmkrebs inklusive HNPCC (<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 2.4.1 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (8.24/rev.3)
BRCA1/ BRCA2 bei therapeutischer Indikation (Custom made by Qiagen), SNV	EDTA-Blut, DNA; DNA	Amplikon, sequencing by synthesis, Qiagen CLC Genomic Workbench Version 23.0.4	2.2.11.2.6. Breast-Ca Qiaseq (08.25/rev.0)
HBOC- Hereditäres Mamma- und Ovarialkarzinom (<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 2.4.1 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (8.24/rev.3)

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Rhabdoid-Prädispositionssyndrom Typ I und II (<i>SMARCA4</i> , <i>SMARCB1</i>) TDS V2 Panel (Custom made by TWIST), SNV	EDTA Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.4.1 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (8.24/rev.3)
Neurofibromatose Typ I/Typ II (<i>NF1</i> , <i>NF2</i>) TDS V2 Panel (Custom made by TWIST), SNV	EDTA-Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.4.1 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (8.24/rev.3)
Multiple Endokrine Neoplasie - MEN Typ I und II (<i>MEN1</i> , <i>RET</i>) TDS V2 Panel (Custom made by TWIST), SNV	EDTA-Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.4.1 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (8.24/rev.3)
Medulläres Schilddrüsenkarzinom (<i>RET</i>) TDS V2 Panel (Custom made by TWIST), SNV	EDTA-Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.4.1 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (8.24/rev.3)
Hereditäres Phäochromozytom/Paraganliomsyndrom (<i>SDHA</i> , <i>SDHB</i> , <i>SDHC</i> , <i>SDHD</i> , <i>SDHAF2</i> , <i>MAX</i> , <i>MEN1</i>) TDS V2 Panel (Custom made by TWIST), SNV	EDTA-Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis (ACMG version 1-6)	2.2.11.2.5 TDS Panel (8.24/rev.3)
Hereditäres diffuses Magenkarzinom (<i>CDH1</i> , <i>CTNNA1</i>) TDS V2 Panel (Custom made by TWIST), SNV	EDTA-Blut, DNA; DNA	Hybrid Capture, sequencing by synthesis, Varvis release 2.4.1 (ACMG version 1-6)	2.2.11.2.5 TDS Panel (8.24/rev.3)
BRCA1/BRCA2	EDTA-Blut, DNA; DNA	MLPA, Coffalyser.Net Version 250317.1029	2.2.11.2.1.1 MLPA BRCA1+2 (2.23/rev.1)

Bitte beachten Sie unseren Einsendeschein und Einverständniserklärung nach GenDG