

Molekulargenetische Untersuchungen (B5) – verpflichtend nach Rili-BÄK

ctDNA-Untersuchungen

Untersuchung	Ringversuch	Programm 25 (Seiten)	Programm 26 (Seiten)
BRAF proto-oncogene, serine/threonine kinase – BRAF p.V600E (BRAF, NM_004333.6:c.1799T>A, rs113488022)	ctDNA	58	61
Epidermal growth factor receptor – EGFR p.T790M (EGFR, NM_005228.5:c.2369C>T, rs121434569)	ctDNA	58	61
KRAS proto-oncogene, GTPase angebotene Sequenzvarianten: – KRAS p.G12 (KRAS, NM_004985.5:c.34G>T>C>A, rs121913530) – KRAS p.G12 (KRAS, NM_004985.5:c.35G>T>C>A, rs121913529) – KRAS p.G13 (KRAS, NM_004985.5:c.37G>T>C>A, rs121913535) – KRAS p.G13 (KRAS, NM_004985.5:c.38G>T>C>A, rs112445441) – KRAS p.Q61 (KRAS, NM_004985.5:c.181C>G>A, rs121913238) – KRAS p.Q61 (KRAS, NM_004985.5:c.182A>T>G>C, rs121913240) – KRAS p.Q61 (KRAS, NM_004985.5:c.183A>T>C, rs17851045)	ctDNA	58	61
NRAS proto-oncogene, GTPase – NRAS p.Q61 (NRAS, NM_002524.5:c.181C>T>G>A, rs121913254) – NRAS p.Q61 (NRAS, NM_002524.5:c.182A>T>G>C, rs11554290) – NRAS p.Q61 (NRAS, NM_002524.5:c.183A>T>C, rs121913255)	ctDNA	58	61

DNA Isolierung

Untersuchung	Ringversuch	Programm 25 (Seiten)	Programm 26 (Seiten)
Faktor V (Leiden), FV-Leiden (F5, NM_000130.5:c.1601G>A, rs6025)	DI, MG1	59, 60	62, 65
Hereditäre Hämochromatose, HFE, HFE C282Y (HFE, NM_000410.4:c.845G>A, rs1800562), HFE C282Y (HFE, NM_000410.4:c.845G>A, rs1800562)	DI	59	62
Methylenetetrahydrofolate reductase (MTHFR), MTHFR A1298C (MTHFR, NM_005957.5:c.1286A>C, rs1801131) MTHFR C677T (MTHFR, NM_005957.5:c.665C>T, rs1801133)	DI, MG1	59, 60	62, 65
Prothrombin, F2 FII g20210a (F2, NM_000506.5:c.*97G>A, rs1799963)	DI, MG1	59, 60	62, 65

Sequenzvarianten-Analyse

Untersuchung	Ringversuch	Programm 25 (Seiten)	Programm 26 (Seiten)
21-Hydroxylase-Defizienz (Adrenogenitales Syndrom)	MG2	61	66
Alpha-1-Antitrypsin, (serpin family A member1, SERPINA1) – AAT-PI*S (SERPINA1, NM_000295.5:c.863A>T, rs17580) – AAT-PI*Z (SERPINA1, NM_000295.5:c.1096G>A, rs28929474) – AAT – Genotypisierung	MG1	60	65
Angiotensin Converting Enzym Apolipoprotein, ACE, ACE I/D (ACE, NM_000789.3:c.2306-117_2306-116insAF118569.1:g.14094_14382, rs1799752)	MG1	60	65
Antithrombin, SERPINC1, AT3 Cambridge Typ I/II (SERPINC1, NM_000488.4:c.1246G>C>T, rs121909548)	MG1	60	65
Apolipoprotein B 100, APOB, ApoB100 (APOB, NM_000384.3:c.10580G>A, rs5742904)	MG1	60	65
Apolipoprotein E (APOE), – ApoE2 (APOE, NM_000041.4:c.526C>T, rs7412) – ApoE4 (APOE, NM_000041.4:c.388T>C, rs429358) – ApoE – Genotypisierung	MG1	60	65
C-C motif chemokine receptor 5, (CCR5), CCR5-del32bp (CCR5, NM_001394783.1:c.554_585del, rs333)	MG2	61	66
Cytochrom P450 2C19, (CYP2C19), – CYP2C19*2 (CYP2C19, NM_000769.4:c.681G>A, rs4244285) – CYP2C19*3 (CYP2C19, NM_000769.4:c.636G>A, rs4986893) – CYP2C19*17 (CYP2C19, NM_000769.4:c.-806C>T, rs12248560) – CYP2C19 – Genotypisierung	MG2	61	66
Cytochrom P450 2C9, (CYP2C9), – CYP2C9*2 (CYP2C9, NM_000771.4:c.430C>T, rs1799853) – CYP2C9*3 (CYP2C9, NM_000771.4:c.1075A>C, rs1057910) – CYP2C9 – Genotypisierung	MG2	61	66
Cytochrom P450 2D6, (CYP2D6), – CYP2D6*2-296 (CYP2D6*2, NM_000106.6:c.886C>T, rs16947) – CYP2D6*2-486 (CYP2D6*2, NM_000106.6:c.1457G>C, rs1135840) – CYP2D6*3 (CYP2D6*3, NM_000106.6:c.775del, rs35742686) – CYP2D6*4 (CYP2D6*4, NM_000106.6:c.506-1G>A, rs3892097) – CYP2D6*6 (CYP2D6*6, NM_000106.6:c.454del, rs5030655) – CYP2D6*7 (CYP2D6*7, NM_000106.6:c.971A>C, rs5030867) – CYP2D6*8 (CYP2D6*8, NM_000106.6:c.505G>T, rs5030865) – CYP2D6*9 (CYP2D6*9, NM_000106.6:c.841_843del, rs5030656) – CYP2D6*10 (CYP2D6*10, NM_000106.6:c.100C>T, rs1065852) – CYP2D6*17 (CYP2D6*17, NM_000106.6:c.320C>T, rs28371706) – CYP2D6*35 (CYP2D6*35, NM_000106.6:c.31G>A, rs769258) – CYP2D6*41 (CYP2D6*41, NM_000106.6:c.985+39G>A, rs28371725) – CYP2D6*5 (CYP2D6, Deletion) – CYP2D6*xN (CYP2D6, Duplikation/Amplifikation)	MG2	61	66
Dihydropyrimidine dehydrogenase, (DPYD), – DPYD*2A (DPYD, NM_000110.4:c.1905+1G>A, rs3918290) – DPYD*13 (DPYD, NM_000110.4:c.1679T>G, rs55886062) – DPYD p.D949V (DPYD, NM_000110.4:c.2846A>T, rs67376798) – DPYD c.1129-5923C>G (DPYD, NM_000110.4:c.1129-5923C>G, rs75017182) – DPYD c.1236G>A (DPYD, NM_000110.4:c.1236G>A, rs56038477) – DPYD HapB3 (DPYD, NM_000110.4:c.1129-5923C>G, rs75017182, NM_000110.4:c.1236G>A, rs56038477)	MG2	61	66
Duchenne und Becker Muskeldystrophie	MG2	61	66
Erbliche Hörstörungen	MG2	61	66
Faktor V (Leiden), FV-Leiden (F5, NM_000130.5:c.1601G>A, rs6025)	DI, MG1	59, 60	62, 65
Familiäres Brust-/Ovarial- Karzinom (BRCA)	MG2	61	66
Fragiles-X Syndrom	MG2	61	66
Hereditäre Hämochromatose (genotyping and interpretation)	MG2	61	66
Hereditäre Hämochromatose, HFE, HFE C282Y (HFE, NM_000410.4:c.845G>A, rs1800562), HFE C282Y (HFE, NM_000410.4:c.845G>A, rs1800562)	DI	59	62
Hereditäres Kolonkarzinom ohne Polyposis	MG2	61	66
HLA-B*27, (HLA-B), HLA-B*27:01 (HLA-B, NM_005514.8)	MG2	61	66
HLA-B*57:01 (HLA-B, NM_005514.8)	MG2	61	66
Huntington'sche Krankheit	MG2	61	66
Lactase-Phlorizin-Hydrolase, (LCT) LCT c-13910t (LCT, NM_005915.6:c.1917+326C>T, rs4988235)	MG1	60	65
Methylenetetrahydrofolate reductase (MTHFR), MTHFR A1298C (MTHFR, NM_005957.5:c.1286A>C, rs1801131) MTHFR C677T (MTHFR, NM_005957.5:c.665C>T, rs1801133)	DI, MG1	59, 60	62, 65
Prader-Willi und Angelman Syndrome	MG2	61	66
Prothrombin, F2 FII g20210a (F2, NM_000506.5:c.*97G>A, rs1799963)	DI, MG1	59, 60	62, 65
Spinale Muskelatrophie	MG2	61	66
Thiopurine S-methyltransferase, (TPMT), – TPMT*2 (TPMT, NM_000367.5:c.238G>C, rs1800462) – TPMT*3B (TPMT, NM_000367.5:c.460G>A, rs1800460) – TPMT*3C (TPMT, NM_000367.5:c.719A>G, rs1142345) – TPMT – Genotypisierung	MG2	61	66
Uridyl-Glucuronyltransferase-1A, UGT1A1, UGT1A1*28 (UGT1A1, NM_000463.3:c.-41_-40dupTA, rs3064744)	MG2	61	66
Wilson-Krankheit, ATP7B, ATP7B-C3207A (ATP7B, NM_000053.4:c.3207C>A, rs76151636)	MG1	60	65
Y-Chromosom, Mikrodeletionen	MG2	61	66
Zystische Fibrose, Mukoviszidose	MG2	61	66