

# Molecular genetic analyses (B5) – in addition to Rili-BÄK

## cfDNA analyses

Investigation	EQA scheme	Program 25 (pages)	Program 26 (pages)
cfDNA isolation	ctDNA	58	61

## Sequence variant analysis

Investigation	EQA scheme	Program 25 (pages)	Program 26 (pages)
ABCB1 c.3435T>C (ABCB1, NM_001348946.2:c.3435T>C, rs1045642)	MG2	61	66
Aldolase, fructose-bisphosphate B, (ALDOB), - AldoB 149 (ALDOB, NM_000035.4:c.448G>C, rs1800548) - AldoB 174 (ALDOB, NM_000035.4:c.524C>A, rs76917243) - AldoB 334 (ALDOB, NM_000035.4:c.1005C>G, rs78340951)	MG1	60	65
beta-fibrinogen g-455a (FGB, NM_005141.4:c.-463G>A, rs1800790)	MG1	60	65
BRAF p.V600 (BRAF, NP_004324.2:p.V600E/K), - BRAF V600E (BRAF, NM_004333.6:c.1799T>A, rs113488022) - BRAF V600K (BRAF, NM_004333.6:c.1798_1799delinsAA, rs121913227)	MG2	61	66
c-KIT p.D816V (KIT, NM_000222.3:c.2447A>T, rs121913507)	MG2	61	66
CETP B1/B2 (CETP, NM_000078.3:c.118+279G>A, rs708272)	MG1	60	65
Col1A1 SP1 (Col1A1, NM_000088.4:c.104-441G>T, rs1800012)	MG1	60	65
CYP2C8*3 (CYP2C8, NM_000770.3:c.1196A>G, rs10509681)	MG2	61	66
CYP3A4*22 (CYP3A4, NM_017460.6:c.522-191C>T, rs35599367)	MG2	61	66
CYP3A5*3 (CYP3A5, NM_000777.5:c.219-237A>G, rs776746)	MG1	60	65
Cytochrome P450 2B6, (CYP2B6), - CYP2B6*4 (CYP2B6, NM_000767.5:c.785A>G, rs2279343) - CYP2B6*9 (CYP2B6, NM_000767.5:c.516G>T, rs3745274) - CYP2B6*6 (CYP2B6, NM_000767.5:c.785A>G, rs2279343 + NM_000767.5:c.516G>T, rs3745274)	MG2	61	66
DPD activity score according to CPIC	MG2	61	66
DPD activity score according to DGHO	MG2	61	66
ESR1 (estrogen receptor 1), PIK3CA (phosphatidylinositol-4,5-bisphosphate-3-kinase catalytic subunit alpha) - p.E380Q (ESR1, NM_000125.4:c.1138G>C, rs1057519827) - p.L536R (ESR1, NM_000125.4:c.1607T>G, rs1057519717) - p.Y537N (ESR1, NM_000125.4:c.1609T>A, rs2152506534) - p.Y537C (ESR1, NM_000125.4:c.1610A>G, rs2152506541) - p.Y537S (ESR1, NM_000125.4:c.1610A>C, rs2152506541) - p.D538G (ESR1, NM_000125.4:c.1613A>G, rs1585254649) - ESR1 exons 3-7 (ESR1, NM_000125.4), specifically covering at least codons 310-547. - p.E542K (PIK3CA, NM_006218.4:c.1624G>A, rs121913273) - p.E545K (PIK3CA, NM_006218.4:c.1633G>A, rs104886003) - p.H1047R (PIK3CA, NM_006218.4:c.3140A>G, rs121913279) - PIK3CA exons 8, 10, and 21 (PIK3CA, NM_006218.4), often referred to as exons 7, 9, and 20	Lbcbp2	-	64
ESR1 (estrogen receptor 1), PIK3CA (phosphatidylinositol-4,5-bisphosphate-3-kinase catalytic subunit alpha) - p.E380Q (ESR1, NM_000125.4:c.1138G>C, rs1057519827) - p.L536R (ESR1, NM_000125.4:c.1607T>G, rs1057519717) - p.Y537N (ESR1, NM_000125.4:c.1609T>A, rs2152506534) - p.Y537C (ESR1, NM_000125.4:c.1610A>G, rs2152506541) - p.Y537S (ESR1, NM_000125.4:c.1610A>C, rs2152506541) - p.D538G (ESR1, NM_000125.4:c.1613A>G, rs1585254649) - p.E542K (PIK3CA, NM_006218.4:c.1624G>A, rs121913273) - p.E545K (PIK3CA, NM_006218.4:c.1633G>A, rs104886003) - p.H1047R (PIK3CA, NM_006218.4:c.3140A>G, rs121913279)	Lbcbp1	-	63
Factor VII (R353Q) (F7, NM_019616.4:c.1172G>A, rs6046)	MG1	60	65
FSAP Marburg-I (HABP2, NM_004132.5:c.1601G>A, rs7080536)	MG1	60	65
FV-Cambridge (F5, NM_000130.5:c.1001G>C, rs118203906)	DI	59	62
FV-HI299R (F5, NM_000130.5:c.3980A>G, rs1800595)	DI	59	62
FV-Hong Kong (F5, NM_000130.5:c.1000A>G, rs118203905)	DI	59	62
FXII c46t (F12, NM_000505.4:c.-4T>C rs1801020)	MG1	60	65
FXIII V34L (F13A1, NM_000129.4:c.103G>T, rs5985)	MG1	60	65
GP1IIa (ITGB3, NM_000212.3:c.176T>C, rs5918)	MG1	60	65
IL28B (C/T polymorphism) (IFNL4, NM_001276254.2:c.151-152G>A, rs12979860)	MG2	61	66
IL6 G(-174)C (IL6, NM_000600.4:c.-237C>G, rs1800795)	MG2	61	66
ITGA2 GpIIa C807T (ITGA2, NM_002203.4:c.759C>T, rs1126643)	MG1	60	65
KRAS proto-oncogene, GTPase (KRAS), KRAS p.G12/p.G13 (KRAS, NP_004976.2:p.G12/G13) - 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.G12 (KRAS, NP_004976.2:p.G12) - 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, NP_004976.2:p.G13) - 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, NP_004976.2:p.Q61) - 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)	MG2	61	66
NRAS (NRAS, NP_002515.1:p.Q61) - NRAS (NRAS, NM_002524.5:c.181C>T>G>A, rs121913254) - NRAS (NRAS, NM_002524.5:c.182A>T>G>C, rs11554290) - NRAS (NRAS, NM_002524.5:c.183A>T>C, rs121913255)	MG2	61	66
Nucleotide binding oligomerization domain containing 2, (NOD2), - NOD R702W (NOD2, NM_001370466.1:c.2023C>T, rs2066844) - NOD G908R (NOD2, NM_001370466.1:c.2641G>C, rs2066845) - NOD L1007finsC (NOD2, NM_001370466.1:c.2938dup, rs2066847)	MG1	60	65
PAI-1 4G5G (SERPINE1, NM_000602.5:c.-820G[(4_5)], rs1799762)	MG1	60	65
TNF alpha 238 (TNF, NM_000594.3:c.-418G>A, rs361525)	MG2	61	66
TNF alpha 308 (TNF, NM_000594.3:c.-488G>A, rs1800629)	MG2	61	66
Vitamin D receptor, (VDR), - VDR BsmI (VDR, NM_000376.3:c.1024+283G>A, rs1544410) - VDR ApaI (VDR, NM_000376.3:c.1025-49G>T, rs7975232) - VDR TaqI (VDR, NM_000376.3:c.1056T>C, rs731236)	MG1	60	65
Vitamin K epoxide reductase complex subunit 1 (VKORC1) - VKORC1 G-1639 (VKORC1, NM_024006.6:c.-1639G>A, rs9923231) - VKORC1 C1173T (VKORC1, NM_024006.6:c.174-136C>T, rs9934438)	MG1	60	65

## DNA sequencing

Investigation	EQA scheme	Program 25 (pages)	Program 26 (pages)
DNA sequencing	SQ	63	68

## Sequence variant analysis with medical interpretation

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ESR1 (estrogen receptor 1), PIK3CA (phosphatidylinositol-4,5-bisphosphate-3-kinase catalytic subunit alpha) - p.E380Q (ESR1, NM_000125.4:c.1138G>C, rs1057519827) - p.L536R (ESR1, NM_000125.4:c.1607T>G, rs1057519717) - p.Y537N (ESR1, NM_000125.4:c.1609T>A, rs2152506534) - p.Y537C (ESR1, NM_000125.4:c.1610A>G, rs2152506541) - p.Y537S (ESR1, NM_000125.4:c.1610A>C, rs2152506541) - p.D538G (ESR1, NM_000125.4:c.1613A>G, rs1585254649) - ESR1 exons 3-7 (ESR1, NM_000125.4), specifically covering at least codons 310-547. - p.E542K (PIK3CA, NM_006218.4:c.1624G>A, rs121913273) - p.E545K (PIK3CA, NM_006218.4:c.1633G>A, rs104886003) - p.H1047R (PIK3CA, NM_006218.4:c.3140A>G, rs121913279) - PIK3CA exons 8, 10, and 21 (PIK3CA, NM_006218.4), often referred to as exons 7, 9, and 20	Lbcbp2	-	64
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