

Molekulargenetische Analysen mit Referenzbereichen

Molekulargenetik 1 /Pharmakogenetik / Agena MassArray (Angaben von Fa. Agena)				
Gen	cDNA/Mutation	rs-Nummer	Referenz-Transkript	klin. gebräuchliche Bezeichnung
ABCB1	c.3435T>C	rs1045642	NM_000927.4	
ABCB1	c.1236T>C	rs1128503	NM_000927.4	
ABCB1	c.2677T>G	rs2032582	NM_000927.4	
COMT	c.186C>T	rs4633	NM_000754.3	
COMT	c.472G>A	rs4680	NM_000754.3	
COMT	c.408C>G	rs4818	NM_000754.3	
COMT	c.1-98A>G	rs6269	NM_000754.3	
CYP1A2	c.-10+113C>T	rs12720461	NM_000761.4	*1K
CYP1A2	c.-3028G>A	rs2069514	NM_000761.4	*1C
CYP1A2	c.1253+1G>A	rs56107638	NM_000761.4	*7
CYP1A2	c.558C>A	rs72547513	NM_000761.4	*11
CYP1A2	c.-9-154C>A	rs762551	NM_000761.4	*1F
CYP2B6	c.983T>C	rs28399499	NM_000767.4	*18
CYP2B6	c.516G>T	rs3745274	NM_000767.4	*6
CYP2C19	c.-806C>A	rs12248560	NM_000769.3	*17
CYP2C19	c.1A>G	rs28399504	NM_000769.3	*4
CYP2C19	c.358T>C	rs41291556	NM_000769.3	*8
CYP2C19	c.681G>A	rs4244285	NM_000769.3	*2
CYP2C19	c.636G>A	rs4986893	NM_000769.3	*3
CYP2C19	c.1297C>T	rs56337013	NM_000769.3	*5
CYP2C19	c.395G>A	rs72552267	NM_000769.3	*6
CYP2C19	c.819+2T>A	rs72558186	NM_000769.3	*7
CYP2C9	c.1075A>C	rs1057910	NM_000771.3	*3
CYP2C9	c.430C>T	rs1799853	NM_000771.3	*2
CYP2C9	c.1003C>T	rs28371685	NM_000771.3	*11
CYP2C9	c.1080C>A	rs28371686	NM_000771.3	*5
CYP2C9	c.1076T>A	rs56165452	NM_000771.3	*4
CYP2C9	c.269T>C	rs72558187	NM_000771.3	*13
CYP2C9	c.485C>A	rs72558190	NM_000771.3	*15
CYP2C9	c.449G>A	rs7900194	NM_000771.3	*8
CYP2C9	c.818del	rs9332131	NM_000771.3	*6
CYP2C9	c.1465C>T	rs9332239	NM_000771.3	*12
CYP2C9	c.353_362del	rs1304490498	NM_000771.3	*25
CYP2D6	c.1403_1411dup	rs765776661	NM_000106.5	*18 dup4125_4133
CYP2D6	c.100C>T	rs1065852	NM_000106.5	*10
CYP2D6	c.1457G>C	rs1135840	NM_000106.5	*2
CYP2D6	c.886C>T	rs16947	NM_000106.5	*2
CYP2D6	c.320C>T	rs28371706	NM_000106.5	*17
CYP2D6	c.985+39G>A	rs28371725	NM_000106.5	*41
CYP2D6	c.1432C>T	rs28371735	NM_000106.5	*36
CYP2D6	c.775del	rs35742686	NM_000106.5	*3
CYP2D6	c.506-1G>A	rs3892097	NM_000106.5	*4
CYP2D6	c.454del	rs5030655	NM_000106.5	*6
CYP2D6	c.841_843del	rs5030656	NM_000106.5	*9
CYP2D6	c.124G>A	rs5030862	NM_000106.5	*12

Molekulargenetik 1 /Pharmakogenetik / Agena MassArray (Angaben von Fa. Agena)

Gen	cDNA/Mutation	rs-Nummer	Referenz-Transkript	klin. gebräuchliche Bezeichnung
CYP2D6	c.505G>T	rs5030865	NM_000106.5	*8 / *14
CYP2D6	c.971A>C	rs5030867	NM_000106.5	*7
CYP2D6	c.1012G>A	rs59421388	NM_000106.5	*29
CYP2D6	c.765_768del	rs72549353	NM_000106.5	*19
CYP2D6	c.635dup	rs72549354	NM_000106.5	*20
CYP2D6	c.137_138insT	rs72549357	NM_000106.5	*15
CYP2D6	c.181-1G>C	rs5030863 (now known as rs201377835)	NM_000106.5	*11
CYP2D6		CNV	NM_000106.5	
CYP3A4	c.522-191C>T	rs35599367	NM_001202855.2	*22
CYP3A4	c.566T>C	rs4987161	NM_001202855.2	*17
CYP3A4	c.664T>C	rs55785340	NM_001202855.2	*2
CYP3A5	c.1193C>A	rs28365083	NM_000777.4	*2
CYP3A5	c.1035dup	rs41303343	NM_000777.4	*7
CYP3A5	c.624G>A	rs10264272	NM_000777.4	*6
CYP3A5	c.219-237A>G	rs776746	NM_000777.4	*3
CYP4F2	c.1297G>A	rs2108622	NM_001082.4	*3
DPYD	c.1905+1G>C	rs3918290	NM_000110.3	*2
DPYD	c.1679T>G	rs55886062	NM_000110.3	*13
DPYD	c.1129-5923C>G	rs75017182	NM_000110.3	HapB3
DPYD	c.2846A>T	rs67376798	NM_000110.3	
GLP1R	c.780A>C	rs1042044	NM_002062.4	
GLP1R	c.510-1135T>G	rs2300615	NM_002062.4	
GLP1R	c.502G>A	rs6923761	NM_002062.4	
MTHFR	c.1286A>C	rs1801131	NM_005957.4	1298A>C
MTHFR	c.665C>T	rs1801133	NM_005957.5	677C>T
OPRM1	c.118A>G	rs1799971	NM_000914.4	
PNPLA5	c.608-169G>A	rs5764010	NM_001177675.1	
POR	c.1508C>T	rs1057868	NM_000941.2	
SLCO1B1	c.521T>C	rs4149056	NM_006446.4	*5
SULT4A1	c.743-374A>G	rs763120	NM_014351.3	
TPMT	c.719A>G	rs1142345	NM_000367.4	*3A*3C
TPMT	c.460G>A	rs1800460	NM_000367.4	*3A*3B
TPMT	c.238G>C	rs1800462	NM_000367.4	*2
TPMT	c.626-1G>A	rs1800584	NM_000367.4	*4
VKORC1	c.-1639G>T	rs9923231	NM_001311311.1	

Molekulargenetik 2 / Realtime-PCR (LAMP)

Gen	cDNA/Mutation	rs-Nummer/Exon	Referenz-Transkript	klin. gebräuchliche Bezeichnung
MTHFR	c.665C>T	rs1801133	NM_005957.5	C677T
MTHFR	c.1286A>C	rs1801131	NM_005957.4	A1298C
SERPINE1	c.-820_-817G (4_5)	rs1799768	NM_000602.4	4G/5G
Faktor-V-Leiden	c.1601G>A	rs6025	NM_000130.4	G1691A
Prothrombin	c.*97G>A	rs1799963	NM_000506.5	G20210A
HLA-B	-	Exon 3	NM_005514.8	HLA-B27

HFE	c.845G>A	rs1800562	NM_000410.4	C282Y
HFE	c.187C>G	rs1799945	NM_139009.3	H63D

Molekulargenetik 2 / Liquid Biopsy Realtime PCR (Biocartis, Diatech)			
Gen	cDNA/Mutation	Angabe am Befund	Referenz- Transkript
EGFR_Biocartis			
EGFR	p.Gly719Ala c.2156G>C	G719A	NM_005228.5
	p.Gly719Cys c.2155G>T	G719C	
	p.Gly719Cys c.2154_2155delinsTT	G719C	
	p.Gly719Ser c.2155G>A	G719S	
	p.Leu747_Ala750delinsPro c.2238_2248delinsGC c.2239_2248delinsC	Deletion 9, Exon 19	
	p.Leu747_Ala750delinsSer c.2240_2248del		
	p.Leu747_Glu749del c.2239_2247del		
	p.Leu747_Thr751delinsPro c.2239_2251delinsC	Deletion 12, Exon 19	
	p.Leu747_Thr751delinsSer c.2240_2251del		
	p.Glu746_Ala750del c.2235_2249del c.2236_2250del	Deletion 15, Exon 19	
	p.Leu747_Thr751del c.2239_2253del c.2240_2254del c.2238_2252del		
	p.Glu746_Thr751delinsAla c.2237_2251del		
	p.Glu746_Thr751delinsIle c.2235_2252delinsAAT		
	p.Glu746_Thr751delinsVal c.2237_2252delinsT		
	p.Lys745_Ala750delinsThr c.2234_2248del		
	p.Glu746_Thr751delinsLeu c.2236_2253delinsCTA		
	p.Glu746_Thr751delinsVal c.2237_2253delinsTA		
	p.Glu746_Thr751delinsAla c.2235_2251delinsAG		
	p.Glu746_Thr751delinsGln c.2236_2253delinsCAA		
	p.Ile744_Ala750delinsValLys c.2230_2249delinsGTCAA		
	p.Leu747_Pro753delinsSer c.2240_2257del		
	p.Glu746_Ser752delinsVal c.2237_2255delinsT		

	p.Leu747_Ser752del c.2239_2256del		
	p.Glu746_Thr751del c.2236_2253del		
	p.Leu747_Pro753delinsGln c.2239_2258delinsCA		
	p.Glu746_Ser752delinsAla c.2237_2254del		
	p.Glu746_Ser752delinsAsp c.2238_2255del		
	p.Glu746_Ser752delinsIle c.2236_2256delinsATC		
	p.Glu746_Ser752delinsVal c.2237_2256delinsTT c.2237_2256delinsTC c.2235_2255delinsGGT		
	p.Leu747_Pro753del c.2238_2258del	Deletion 21, Exon 19	
	p.Glu746_Ser752del c.2236_2256del		
	p.Ser752_Ile759del c.2253_2276del	Deletion 24, Exon 19	
	p.Thr790Met c.2369C>T	T790M	
	p.Ser768Ile c.2303G>T	S768I	
	p.Asp770_Asn771insGly c.2310_2311insGGT	Insertion Exon 20	
	p.Val769_Asp770insAlaSerVal c.2307_2308insGCCAGCGTG		
	p.Val769_Asp770insAlaSerVal c.2309_2310delinsCCAGCGTGGAT		
	p.Asp770_Asn771insSerValAsp c.2311_2312insGCGTGGACA		
	p.His773_Val774insHis c.2319_2320insCAC		
	p.Leu858Arg c.2573T>G c.2573_2574delinsGT c.2573_2574delinsGA	L858R	
	p.Leu861Gln c.2582T>A	L861Q	
	ESR1_Diatech		
ESR1	p.Lys303Arg c.908A>G	L303R	NM_000125.4
	p.Glu380Gln c.1138G>C	E380Q	
	p.Ser463Pro c.1387T>C	S463P	
	p.Leu536Arg c.1607T>G	L536R	
	p.Leu536His c.1607T>A	L536H	
	p.Leu536Pro c.1607T>C	L536P	
	p.Leu536Gln c.1607_1608delinsAG	L536Q	

p.Tyr537Asn c.1609T>A	Y537N
p.Tyr537His c.1609T>C	Y537H
p.Tyr537Asp c.1609T>G	Y537D
p.Tyr537Cys c.1610A>G	Y537C
p.Tyr537Ser c.1610A>C	Y537S
p.Asp538Gly c.1613A>G	D538G

Molekulargenetik 3 / SSP-PCR

Gen	Referenz- Transkript
HLA-B	Siehe Spezifitätentabelle NM_005514.8

Molekulargenetik 4 / Hybridisierung

Gen	cDNA/Mutation	Referenz- Transkript	klin. gebräuchliche Bezeichnung
HFE	c.187C>G	NM_139009.3	H63D
HFE	c.193A>T	NM_000410.4	S65C
HFE	c.502G>T	NM_139004.3	E168X
HFE	c.845G>A	NM_000410.4	C282Y
HLA-DQA1	-	NM_002122.5	
HLA-DRB1	-	NM_002124.4	
HLA-DQB1	-	NM_002123.5	

Molekulargenetik 5 / NGS (Angaben von Fa. Sophia Genetics)

Humanes Referenzgenom	GRCh37	
Gen	Exons	Referenz-Transkript
CEBPA	alle Exons	NM_004364.5
CSF3R	alle Exons	NM_000760.4
DNMT3A	alle Exons	NM_022552.5
ETV6	alle Exons	NM_001987.5
EZH2	alle Exons	NM_004456.5
JAK2	alle Exons	NM_004972.4
RUNX1	alle Exons	NM_001754.5
TET2	alle Exons	NM_017628.4
TP53	alle Exons	NM_000546.6
ZRSR2	alle Exons	NM_005089.4
ABL1	Exons 4-9	NM_005157.6
ASLX1	Exons 10,12,13	NM_015338.6
BRAF	Exon 15	NM_004333.6
CALR	Exon 9	NM_004343.4
CBL	Exons 8-9	NM_005188.4
FLT3	Exons 13-15, 20	NM_004119.3
HRAS	Exons 2-3	NM_005343.4
IDH1	Exon 4	NM_005896.4

Molekulargenetik 5 / NGS (Angaben von Fa. Sophia Genetics)		
Humanes Referenzgenom	GRCh37	
Gen	Exons	Referenz-Transkript
IDH2	Exon 4	NM_002168.4
KIT	Exons 2, 8-11, 13, 17-18	NM_000222.3
KRAS	Exons 2-3	NM_004985.5
MPL	Exon 10	NM_005373.3
NPM1	Exons 10-11	NM_002520.7
NRAS	Exons 2-3	NM_002524.5
PTPN11	Exons 3, 7-13	NM_002834.5
SETBP1	Exon 4	NM_015559.3
SF3B1	Exons 10-16	NM_012433.4
SRSF2	Exons 1	NM_003016.4
U2AF1	Exons 2, 6	NM_006758.3
WT1	Exons 6-10	NM_024426.6
BCOR	alle Exons	NM_001123385.2
BCORL1	alle Exons	NM_021946.4
CXCR4	alle Exons	NM_003467.3
BIRC3	Exons 6, 8, 9	NM_001165.5
BTK	Exons 2,6,15	NM_000061.3
MYD88	Exons 3-5	NM_001172567.2
NOTCH1	Exon 34	NM_017617.5
NOTCH2	Exons 26,34	NM_024408.4
PLCG2	Exons 19,20, 24, 27	NM_002661.5

Molekulargenetik 5 / NGS Liquid Biopsy NGS (Sophia Genetics) - LBx_Solid Tumor Solution (STS) v1	
Humanes Referenzgenom	hg19
Details siehe Parameterliste NGS Liquid Profiling (Sharepoint Dokument 02-16-20595 - L1195) im Parameterkatalog	

Version	Inhalt der Änderung	Seite, Punkt
8	HFE und HLA-B bei Molekulargenetik 2 eingefügt	S.2 und 3
	Liquid Biopsy – NGS eingefügt	S.4
	Liquid Biopsy – Realtime PCR eingefügt	S.3
	Nicht mehr durchgeführte Analysen- AAT, Hämoglobinopathien, JAK2/Agena entfernt	S.2 und 3
7	NGS: Versionsnummern der Referenz-Transkripte eingefügt	S.3 und 4
	Hybridisierung: LCT entfernt	S.3
	SSP-PCR: siehe Spezifitätentabelle ergänzt	S.3
	Agena MassArray Hämatookologie: Mutationen ausgenommen JAK2 gelöscht	S.2
	Agena MassArray Pharmakogenetik: Aktualisierung bei CYP2C9 und CYP2D6	S.1
6	Änderung Exon ASXL1 und SRSF2	S.5
5	Gene für MOL5/NGS ergänzt	S. 5
4	Übernahme neue Formatvorlage	Gesamtes Dokument
	Änderung der Gen-Bezeichnung: von PAI auf SERPINE1, von Lactase auf LCT, von A1AT auf SERPINA1	S. 4
	Mutation eingefügt für CYP2B6, CYP3A5, DPYD, MTHFR, LCT, HFE und SERPINA1	S. 1,2,4
	Mutation für NPM1 eingefügt	S. 3