Archer® FusionPlex

Radboudumc

Assay beschrijving

De Archer® FusionPlex is een targeted NGS methode, gebaseerd op anchored multiplex PCR, welke wordt uitgevoerd t.b.v. detectie van RNA fusietranscripten. Fusietranscripten kunnen worden aangetoond als 1 van beide partners van het fusietranscript is opgenomen in het panel, en in de juiste oriëntatie wordt geanalyseerd. De sensitiviteit van deze analyse voor de detectie van specifieke fusietranscripten is vooralsnog onbekend, maar is o.a. afhankelijk van met de kwaliteit van het RNA, het percentage tumorcellen en de expressie van zowel het fusietranscript als het wildtype transcript (in omliggend normaal weefsel). Sequencing wordt uitgevoerd op een Illumina MiniSeq of NextSeq 500. Data analyse wordt uitgevoerd met behulp van Archer® Analysis software. Weefsels met minder dan 30% neoplastische cellen hebben mogelijk een te lage tumorcontent voor detectie van een fusietranscript met een relatief laag expressieniveau. De kwaliteitsbepaling van de analyse berust op detectie van controle transcripten en percentage RNA reads.

Er wordt gebruik gemaakt van het commerciële Lung panel of het Radboudv1 custom panel.

Fusionplex Lung

Gen	NM nummer	Coverage (exonen)
ALK	NM_004304	exon 2, 4, 6, 10, 16, 17, 18, 19, 20, 21, 22, 23, 26
BRAF	NM_004333	exon 1, 2, 3, 7, 8, 9, 10, 11, 12, 13, 15, 16
EGFR	NM_005228	exon 1, 7, 8, 9, 16, 19, 20, 24, 25
FGFR1	NM_015850	exon 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 17
FGFR2	NM_000141	exon 2, 5, 7, 8, 9, 10, 16, 17
FGFR3	NM_000142	exon 3, 5, 8, 9, 10, 16, 17
MET	NM_000245	exon 2, 4, 5, 6, 13, 14, 15, 16, 17, 21
NRG1	NM_013962	exon 1,
NRG1	NM_004495	exon 1, 2, 3, 6
NRG1	NM_013957	exon 1, 4, 8
NTRK1	NM_002529	exon 2, 4, 6, 8, 10, 11, 12, 13
NTRK2	NM_006180	exon 7, 9, 11, 12, 13, 14, 15, 16, 17
NTRK3	NM_002530	exon 4, 7, 10, 12, 13, 14, 15, 16
NTRK3	NM_001007156	exon 15
RET	NM_020975	exon 8, 9, 10, 11, 12, 13, 14
RET	NM_020630	exon 2, 4, 6
ROS1	NM_002944	exon 2, 4, 7, 31, 32, 33, 34, 35, 36, 37

FusionPlex RadboudV1

Gen	NM nummer	Coverage (exonen)
ABL1	NM_005157	exon 1, 2, 3, 4
ABL2	NM_007314	exon 2, 3, 4, 5, 6
ALK	NM_004304	exon 2, 4, 6, 10, 16, 17, 18, 19, 20, 21, 22, 23, 26
BCOR	NM_001123385 NM_017745	exon 6, 7, 8, 12, 14, 15 exon 8
BRAF	NM_004333	exon 1, 2, 3, 7, 8, 9, 10, 11, 12, 13, 15, 16
CAMTA1	NM_015215	exon 3, 8, 9, 10
CIC	NM_015125	exon 18, 19, 20
EGFR	NM_005228	exon 1, 7, 8, 9, 16, 19, 20, 24, 25
ERBB2	NM_004448	exon 4, 5, 23, 24, 25, 26
ERG	NM_004449	exon 2, 3, 4, 5, 6, 7, 8, 9, 10, 11
ETV6	NM_001987	exon 1, 2, 3, 4, 5, 6, 7
EWSR1	NM_005243	exon 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14
FGFR1	NM_015850	exon 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 17

FGFR2	NM_000141	exon 2, 5, 7, 8, 9, 10, 16, 17
FGFR3	NM_000142	exon 3, 5, 8, 9, 10, 16, 17
FOS	NM_005252	exon 1, 2, 3, 4
FOSB	NM_006732	exon 1, 2, 3
	NM_001114171	exon 1, 2, 3
FOXO1	NM_002015	exon 1, 2, 3
FUS	NM_004960	exon 3, 4, 5, 6, 7, 8, 9, 10, 11, 13, 14
GLI1	NM_005269	exon 4, 5, 6, 7
HMGA2	NM_003483	exon 1, 2, 3, 4, 5
JAZF1	NM_175061	exon 2, 3, 4
MALT1	NM_006785	exon 2, 3, 4, 5, 6, 7, 9, 10
MAML2	NM_032427	exon 2, 3
MET	NM_000245	exon 2, 4, 5, 6, 13, 14, 15, 16, 17, 21
MKL2	NM_014048	exon 11, 12, 13
MYB	NM_001130173	exon 7, 8, 9, 11, 12, 13, 14, 15, 16
MYBL1	NM_001080416	exon 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16
	NM_001144755	exon 8
NCOA1	NM_147223	exon 12, 13, 14, 15
NCOA2	NM_006540	exon 11, 12, 13, 14, 15, 16
NFIB	NM_005596	exon 5, 6, 7, 8, 9
	NM_001190737	exon 9, 10
NR4A3	NM_173200	exon 3, 4
NR4A3	NM_006981	exon 4
NRG1	NM_013962	exon 1
	NM_004495	exon 1, 2, 3, 6
	NM_013957	exon 1, 4, 8
NTRK1	NM_002529	exon 2, 4, 6, 8, 10, 11, 12, 13, 14, 15
NTRK2	NM_006180	exon 5, 7, 9, 11, 12, 13, 14, 15, 16, 17
NTRK3	NM_002530	exon 4, 7, 10, 12, 13, 14, 15, 16
	NM_001007156	exon 15
PDGFB	NM_002608	exon 2, 3
PDGFRB	NM_002609	exon 8, 9, 10, 11, 12, 13, 14
PHF1	NM_024165	exon 1, 2, 3, 4, 5, 6, 7, 8
PLAG1	NM_002655	exon 1, 2, 3, 4
PPARG	NM_015869	exon 1, 2, 3
PRKD1	NM_002742	exon 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17
PRKD2	NM_016457	exon 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17
PRKD3	NM_005813	exon 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17
RAD51B	NM_133509	exon 11
	NM_002877	exon 11
	NM_133510	exon 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11
RAF1	NM_002880	exon 4, 5, 6, 7, 8, 9, 10, 11, 12
RET	NM_020975	exon 8, 9, 10, 11, 12, 13, 14
	NM_020630	exon 2, 4, 6
ROS1	NM_002944	exon 2, 4, 7, 31, 32, 33, 34, 35, 36, 37
SS18	NM_001007559	exon 4, 5, 6, 8, 9, 10, 11
STAT6	NM_001178078	exon 1, 2, 3, 4, 5, 6, 7, 15, 16, 17, 18, 19, 20
TFE3	NM_006521	exon 2, 3, 4, 5, 6, 7, 8, 10
THADA	NM_022065	exon 24, 25, 26, 27, 28, 29, 30, 36, 37
TMPRSS2	NM_005656	exon 1, 3, 4, 5, 6
	NM_001135099	exon 1, 2
USP6	NM_004505	exon 1, 2, 3
YWHAE	NM_006761	exon 5