ARCHER myeloide

Série 1

validation

200914800EB VOL. Mar.

LMC diagnostique en 1999, caryotype pregreffe de 2019

- Caryo

47,XX,+8[6]/45,XX,-7,t(9;22)(q34;q12)[6]

- NGS

mutations d'ASXL1 et SETBP1 (15%)

200914800EB VOL. Mar.

Genes	SS	Reads	%Reads	Strong	Brkpt	Cat	Type	InFrame
BCR → ABL1	167	695	12.73	True	chr22:23632600,chr9:133729451	Fusion		True
BCR → ABL1	75	146	3.01	True	chr22:23631808,chr9:133729451	Fusion		True
BCR → ABL1	16	37	0.77	True	chr22:23632600,chr9:133655756	Fusion		True
BCR → ABL1	6	8	0.17	True	chr22:23632600,chr9:133657191	Fusion		False

Targeted RNA Variant | All Results | New | Edit Columr

Symbol	HGVSp	HGVSc	Depth	AO	AF	Quality Score	TO
ASXL1	p.Gly646TrpfsTer12	c.1934dup	2038	405	0.1987	14239.3	1443
PTPN11	p.Ala72Ser	c.214G>T	477	40	0.0839	1528.94	352
PTPN11	p.Gln79Arg	c.236A>G	530	52	0.0981	2014.63	960
PTPN11	p.Pro491Leu	c.1472C>T	898	59	0.0657	2265.05	454

201605738PM TAI. Léo

- LAM5

- 46,XY,t(11;19)(q23;p13)[12]. ish (11;19)(q23;p13)(5'MLL+;3’MLL+)[20] .nuc ish(MLLx2)(5'MLL sep 3'MLLx1)[180/200]

- Pas de mutation IDH1 ou IDH2, surexpression EVI1, WT1, mutation FLT3

201605738PM TAI. Léo

Genes	SS	Reads	%Reads	Strong	Brkpt	Cat	Type	InFrame
KMT2A → ELL	149	1479	53.7	True	chr11:118355029,chr19:18583692	Fusion		True
KMT2A → ELL	133	1286	47.19	True	chr11:118355690,chr19:18583692	Fusion		True
KMT2A → ELL	24	47	1.94	True	chr11:118355029,chr19:18586743	Fusion		True
KMT2A → ELL	19	28	1.17	True	chr11:118355690,chr19:18586743	Fusion		True

Symbol	HGVSp	HGVSc	Depth	AO	AF	Quality Score	TO
FLT3	p.Asp835Glu	c.2505T>A	4055	880	0.2170	32847.7	223
FLT3	p.Asp835Glu	c.2505T>G	4055	272	0.0671	10167.5	119
FLT3	p.Asp835Tyr	c.2503G>T	4049	312	0.0771	11891.2	297
FLT3	p.Asn676Lys	c.2028C>G	9992	437	0.0437	16828.7	77
NRAS	p.Gln61Arg	c.182A>G	1280	46	0.0359	1735.5	1320
PTPN11	p.Ala72Ser	c.214G>T	316	9	0.0285	342.943	352
PTPN11	p.Gln79Arg	c.236A>G	337	10	0.0297	376.885	960

201611444DW GUI. Fou

- Prélèvement diagnostic d'une LAM2

- 45,X,-Y,t(8;21)(q22;q22)[11]/46,XY[1].ish 11q23(MLL+)[27]

.nuc ish (MLLx2)[198]

BM :

Kit c.2466T>G faiblement positif, pas de délétion de Kit (Lilles)

201611444DW

Genes	SS	Reads	%Reads	Strong
RUNX1 → RUNX1T1	555	3981	14.72	True
RUNX1 → RUNX1T1	248	2086	7.75	True
RUNX1 → RUNX1T1	246	895	3.48	True

Symbol	HGVSp	HGVSc	Depth	AO	AF	Quality Score	TO
KIT	p.Asp419del	c.1255_1257del	1387	59	0.0425	2362.06	23
KIT	p.Asn822Lys	c.2466T>G	1604	69	0.0430	2705.18	153
NRAS	p.Gly12Asp	c.35G>A	335	16	0.0478	604.645	893
NRAS	p.Gly12Cys	c.34G>T	339	24	0.0708	922.471	395
PTPN11	p.Ala72Ser	c.214G>T	325	14	0.0431	536.603	352
PTPN11	p.Gln79Arg	c.236A>G	352	23	0.0653	870.234	960
PTPN11	p.Pro491Leu	c.1472C>T	726	22	0.0303	846.016	454

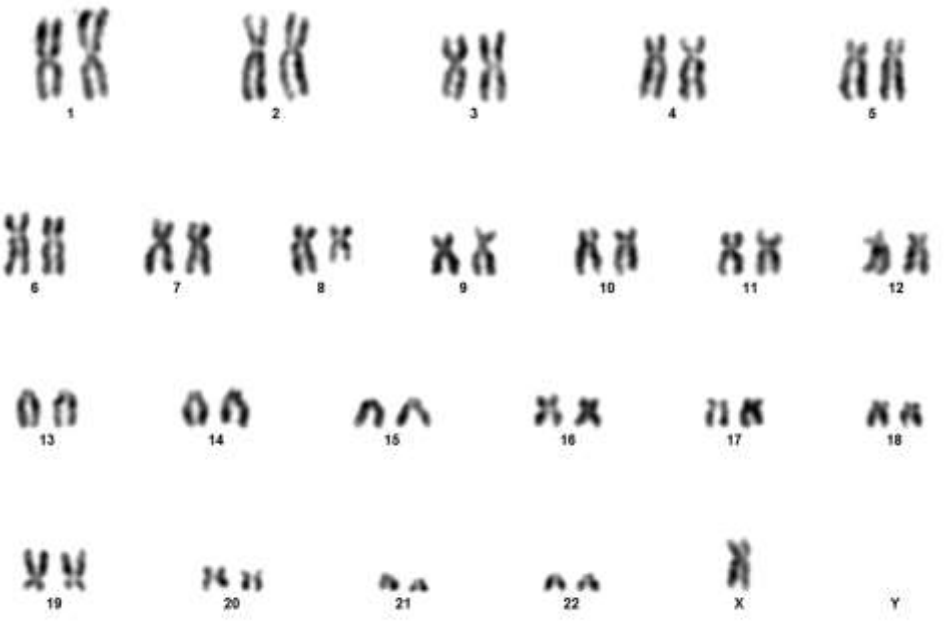
201611995SD

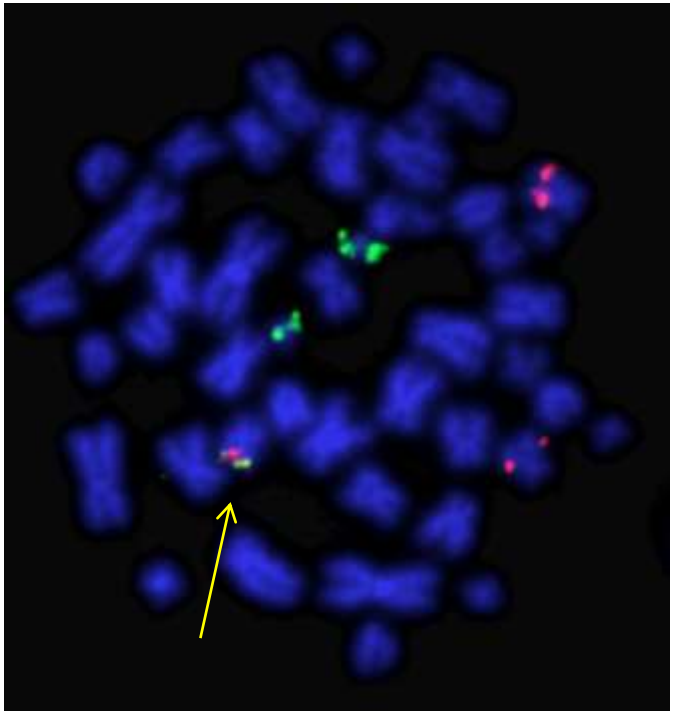
- LAM2
- 45,X,-Y,der(8)t(8;21)(q22;q22),t(8;19)(q22;p13)[1]/45,sl,+t(8;19)(q22;p13),-19[8]/46,XY[8].ish der(8)t(8;21)(q22;q22)(ETO+,AML1+;AML1+,ETO-),t(8;19)(q22;p13)(ETO+;ETO+)[10],t(8;19)(q22;p13)(wcp8;wcp8+,wcp19+)[2]/sl,+t(8;19)(q22;p13) (ETO+;ETO+)[15],+t(8;19)(q22;p13)(wcp8-;wcp8+,wcp19+),-19[12]/8(wcp8+),19(wcp19+)[4],8q22(ETO+),21q22(AML1+)[4],11q23(MLL+)[24].nuc ish(ETOx3),(AML1x3)(ETO con AML1x1)[30]/(ETOx4),(AML1x3)(ETO con AML1x1)[44]/(ETO,AML1)x2[22],(MLLx2)[190]

- BM a Lilles

Mutation ASXL1

201611995SD





- En clair t(8;21) sans le der(21), le der(8) donne une transloc avec le 19 et ce dernier est dupliqué dans un sous clone.

- BM a Lilles

Mutation ASXL1

201611995SD

Genes	SS	Reads	%Reads	Strong	Brkpt
RUNX1 → RUNX1T1	553	4604	22.14	True	chr21:36231771,chr8:93029591
RUNX1 → RUNX1T1	101	204	1.04	True	chr21:36231771,chr8:93029659
RUNX1 → RUNX1T1	101	204	1.04	True	chr21:36231771,chr8:93074937
RUNX1 → RUNX1T1	60	125	18.01	True	chr21:36213292,chr8:93077675
RUNX1 → RUNX1T1	73	99	0.51	True	chr21:36231771,chr8:93077674
RUNX1 → RUNX1T1	60	92	13.24	True	chr21:36213292,chr8:93077675
RUNX1 → RUNX1T1	40	74	0.38	True	chr21:36231771,chr8:93077675
RUNX1 → RUNX1T1	41	63	0.32	True	chr21:36231771,chr8:93077674
RUNX1 → RUNX1T1	32	56	0.29	True	chr21:36231771,chr8:93077675
RUNX1 → RUNX1T1	31	41	0.21	True	chr21:36231771,chr8:93074855
RUNX1 → RUNX1T1	20	26	0.13	True	chr21:36231771,chr8:93077675
RUNX1 → RUNX1T1	15	18	0.14	True	chr21:36265222,chr8:93029591

Symbol	HGVSp	HGVSc	Depth	AO	AF	Quality Score	TO
ASXL1	p.Gly646TrpfsTer12	c.1934dup	1596	496	0.3108	18060.4	1443
PTPN11	p.Ala72Ser	c.214G>T	338	17	0.0503	637.313	352
PTPN11	p.Gln79Arg	c.236A>G	366	21	0.0574	793.891	960
PTPN11	p.Pro491Leu	c.1472C>T	687	26	0.0378	993.291	454

201717085NU

- LAM3
- 46,XX,t(15;17)(q22;q21)[3]/46,XX[17] .ish t(15;17)(PML+,RARA+;RARA+,PML+)[12].nuc ish(PML,RARA)x3(PML con RARAx2)[188/200]
- NGS: mutation FLT3/TKD D835V (39%

201717085NU

Genes	SS	Reads	%Reads	Strong	Brkpt	Cat	Type	InFrame
PML → RARA	275	1489	48.44	True	chr15:74325755,chr17:38504568	Fusion		True
PML → RARA	59	113	3.73	True	chr15:74325751,chr17:38504568	Fusion		False
PML → RARA	10	11	91.67	True	chr15:74325870,chr17:38503849	Fusion		Unknown
PML → RARA	7	8	0.17	True	chr15:74317268,chr17:38504568	Fusion		False
PML → RARA	6	7	0.37	True	chr15:74325763,chr17:38504568	Fusion		False
PML → RARA	7	7	0.26	True	chr15:74325753,chr17:38504341	Fusion		True

Symbol	HGVSp	HGVSc	Depth	AO	AF	Quality Score	TO
ASXL1	p.Gly646TrpfsTer12	c.1934dup	1344	37	0.0275	1161.99	1470
FLT3	p.Asp835Val	c.2504A>T	735	335	0.4558	12717.6	294

201814359RP

- LAL B

- 46,XY,t(4;11)(q21;q23),i(7)(q10)[6]/46,XY[5].ish t(4;11)(3'KMT2A+;5'KMT2A+)[12]/11q23(KMT2A+)[6].nuc ish(KMT2Ax2)(5'KMT2A sep 3'KMT2Ax1)[186/200]

- BM : (St Louis), transcrit KMT2A-AFF1 en MLPA

201814359RP

Genes	SS	Reads	%Reads	Strong	Brkpt	Cat	Type	InFrame
KMT2A → AFF1	127	803	44.32	True	chr11:118355029,chr4:88005275	Fusion		True
KMT2A → AFF1	48	314	17.61	True	chr11:118355029,chr4:88005272	Fusion		True
KMT2A → AFF1	9	16	41.03	True	chr11:118355190,chr4:87989358	Fusion		False

Symbol	HGVSp	HGVSc	Depth	AO	AF	Quality Score	TO	Rept	Tier I
ASXL1	p.Gly646TrpfsTer12	c.1934dup	2225	76	0.0342	2445.11	1470	2	1

201913183KS

- LAM avec inv(16)

- 46,XY,inv(16)(p13q22)[6]/46,XY[6]. Ish 16p13(3'CBFB+), 16q22(5'CBFB+)[20].nuc ish(CBFBx2)(5'CBFB sep 3'CBFBx1)[152/200]

- BM

CBFb-MYH11 type A (161.8490%) sur MO

Surexpression WT1 100.325% sur MO

NGS : mutation NRAS G12D (49%)

201913183KS

Genes	SS	Reads	%Reads	Strong	Brkpt	Cat	Type	InFrame
CBFB → MYH11	222	1419	58.32	True	chr16:67116211,chr16:15814908	Fusion		True
CBFB → MYH11	13	15	0.87	True	chr16:67116242,chr16:15814908	Fusion		False

Symbol	HGVSp	HGVSc	Depth	AO	AF	Quality Score	TO	F
ASXL1	p.Gly646TrpfsTer12	c.1934dup	1160	34	0.0293	1107.1	1470	2
NRAS	p.Gly12Asp	c.35G>A	306	153	0.5000	5779.8	927	

202004131PN

- LAM myelomonocytaire / LAM4

- 47,XY,+8[8]/46,XY[6].ish del(11)(q23)(5'MLL+,3'MLL-)[7] (5'KMT2A+,3'KMT2A-)[16],22q12(BCR+)(17].nuc ish (ABL1,BCR)x2[200],(5'MLLx2,3'MLLx1)[106/200],(5'KMT2Ax2,3'KMT 2Ax1)(5'KMT2A con 3'KMT2Ax1)[90/200]/ (KMT2Ax2)[97/200]

- NGS:

U2AF1 S34F(45%), CBL K389R(79%, perte hétérozygotie),

NRAS Q61P(7%), KRAS(2%)

202004131PN

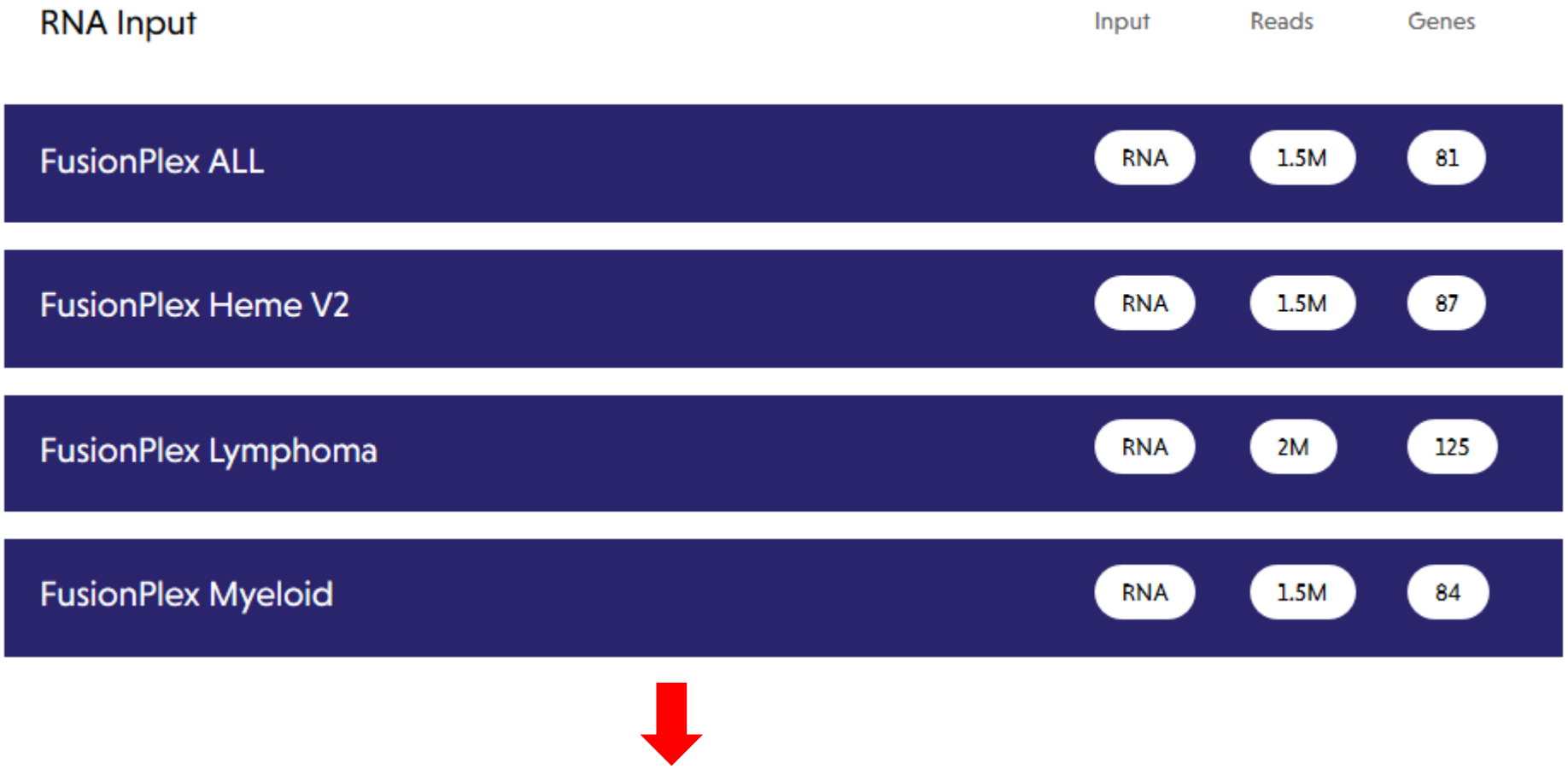
Genes	SS	Reads	%Reads	Strong	Brkpt	Cat	Type	InFrame
KMT2A → CBL	202	661	18.07	True	chr11:118355690,chr11:119149220	Fusion		True
KMT2A → CBL	14	20	0.62	True	chr11:118355029,chr11:119149220	Fusion		True

Symbol	HGVSp	HGVSc	Depth	AO	AF	Quality Score	TO
ASXL1	p.Gly646TrpfsTer12	c.1934dup	914	31	0.0339	956.588	1470
KRAS	p.Gln61Pro	c.182A>C	1761	94	0.0534	3717.65	322
PTPN11	p.Ala72Ser	c.214G>T	175	18	0.1029	685.018	353
PTPN11	p.Gln79Arg	c.236A>G	194	24	0.1237	915.599	969
U2AF1	p.Ser34Phe	c.101C>T	2542	1238	0.4870	49140.6	310

- Manque mutation CBL et NRAS

LEGEND

- SNV/Indel
- Fusion, splicing or exon-skipping
- Expression



CHOIX DU PANEL HEME V2 (MYELOIDE ET LYMPHOIDE, 87 CIBLES)

LEGEND

- SNV/Indel
- Fusion, splicing or exon-skipping
- Expression

- 16 echantillons choisis
  - 8 myeloides
    - 5 complexes avec des points de cassures a proximité de points de cassures connus
    - 3 pour lequel le partenaire manque
  - 8 lymphoides
    - 3 pour lesquels on connait un réarrangement
    - 5 avec problème diagnostic

MYELOIDE

182830799 / 201815580MZ

- LAM secondaire avec dysplasie multilignée
- 47~49,X,del(X)(q25),der(3)t(3;5)(p?21;p?13),del(4)(q21q31),-5,+8,+8,add(11)(p15),del(17)(p12p11),+21[12].ish der(3)t(3;5)(p?;p?)(wcp5+,wcp3+;wcp5-),-5(wcp5-)[20] /3(wcp3+),5(wcp5+)[3],11q23(KMT2A+)[30],17p13(TP53+),17q23(MPO+)[20].nuc ish(KMT2Ax2)[196/200],(TP53,MPO)x2[193/200]

- Archer
  - Rien meme en low confidence

200730612 / 20200430SZ

- LAM5

- 46,XX[20].ish 11q23(MLL+)[20],del(21)(q22)(3'RUNX1+,5'RUNX1-)[20].nuc ish(MLLx2)[191],(3'RUNX1x2,5'RUNX1x1)[192/200]
  - Perte de la partie distale de RUNX1

- Archer
  - Rien meme en low confidence

191770607 /201910210TA

- LAL
- 46~47,XY,der(3)t(3;11)(p12;p11),del(5)(q31),der(11)t(5;3;11)(?;p24;p11),+der(?)t(?;11)(?;qter),del(12)(p12)[cp5]/46,XY[7].ish t(3;11)(wcp11+,wcp3+;wcp3+,wcp11+), der(11)?t(?;11)(wcp11;wcp11+)[7],der(3)t(5;3;11)(wcp5+;wcp3+;wcp11+),del(5)(q31)[6],5p15(TAS2R1+),del(5)(q31)(EGR1dim)[1],add(11)(q24)(KMT2A+)[1],del(12)(p13)(ETV6-)[1]/3(wcp3+),11(wcp11+)[9], 5(wcp5+)[1], 11q23(KMT2A+)[4],12p13(ETV6+)[11].nuc ish(TAS2R1,EGR1dim)x2[46/100],(ETV6x1)[162/200] /(KMT2Ax2)[182/200]

- Archer

SET → NUP214	448	2698	81.46	True	chr9:131456321,chr9:134034770	Fusion
LOC107986169 → TP63	73	168	12.75	True	chr3:189223342,chr3:189455529	Fusion
BCL2 → LINGO1	7	7	63.64	False	chr18:60790585,chr15:77910949	Fusion

190790799 / 201904504FR

- LAM secondaire
- 46,XY,add(11)(q23)[3]/46,XY[9].ish t(?;11)(?p;q23)(5'KMT2A+;3'KMT2A+)[13]/11q23(KMT2A+)[6].nuc ish(KMT2Ax2)(5'KMT2A sep 3'KMT2Ax1)[151/200]

- Archer

KMT2A → MLLT3	172	1946	71.52	True	chr11:118355029,chr9:20365742	Fusion

  - Il y a une t(9;11) impliquant MLLT3 et KMT2A/MLL

TCF3 → ACSL3	15	35	74.47	False	chr19:1619750,chr2:223731482

172990591 / 201713538MU

- LAM
- 46,XX,t(8;5;?)(p23;p15;pter)[10]/46,XX[10].ish t(8;5;?)(D8S504-;D8S504+,EGR1+,CSF1+)[40]
- Archer

SIK1 → IRF4	8	88	13.02	True	chr21:44846890,chr6:394821	Fusion

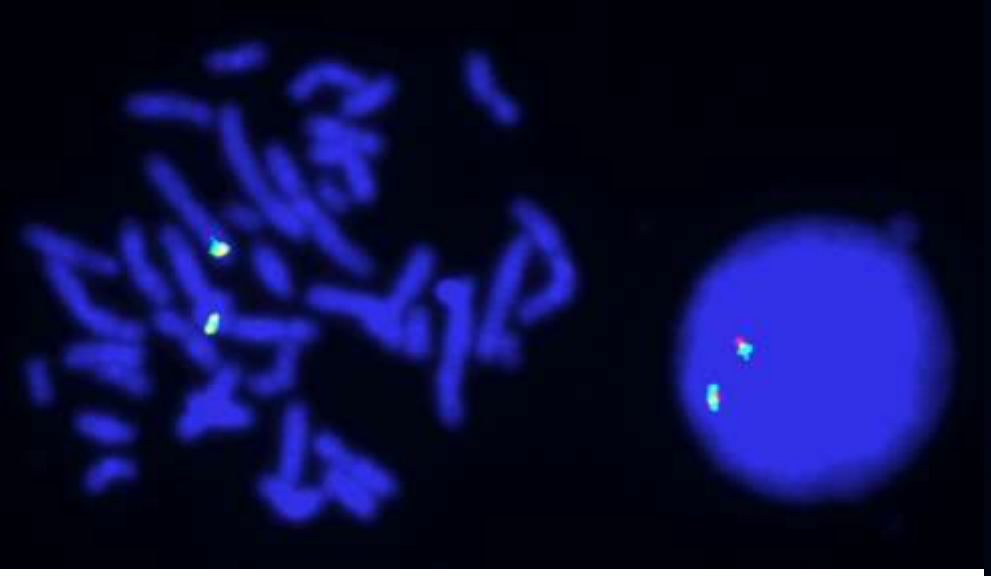
182690591 / 201814755RE

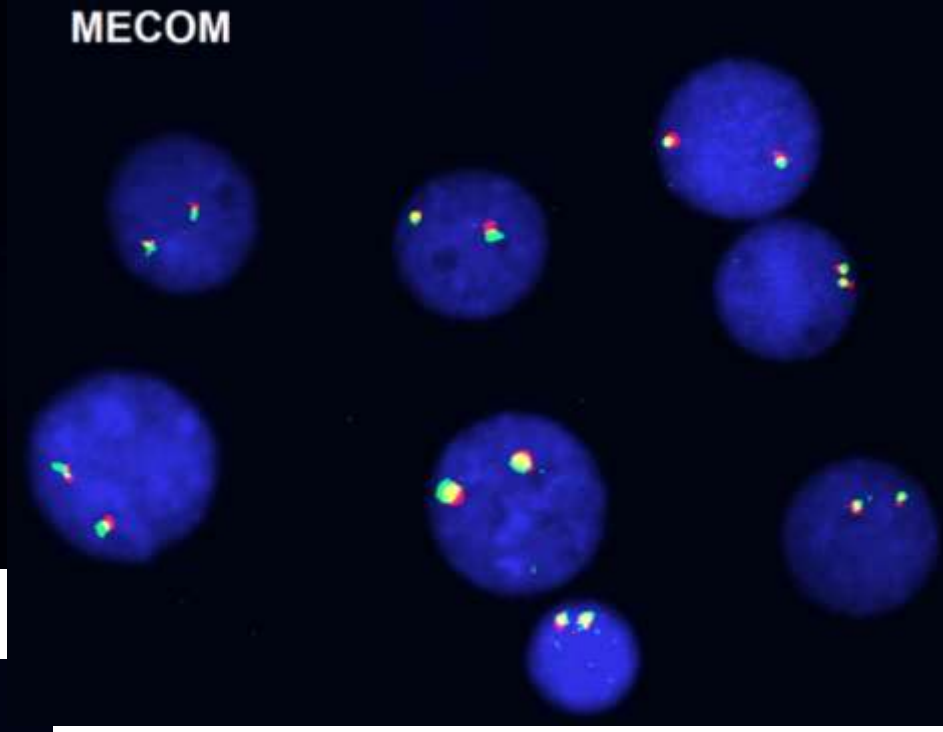
- 41~43,XY,-4,-5,-6,del(8)?(q22),+der(8)t(?;8)(p?;q11),-10,-14,-15,-17,-21x2,-22,+3~5mar[cp15]/46,XY[1].ish 5(wcp5-), del(8)?(q22)(wcp8-),+der(8)t(?;8)(p?;q11)(wcp8-;wcp8+)[17], der(11)t(?;11)(?;KMT2A+)[1],-17(TP53-,MPO-)[20]/ 11q23(KMT2A+)[18].nuc ish(KMT2Ax3)[30], (TP53,MPO)x1[186/200]/(KMT2Ax2)[166/200]

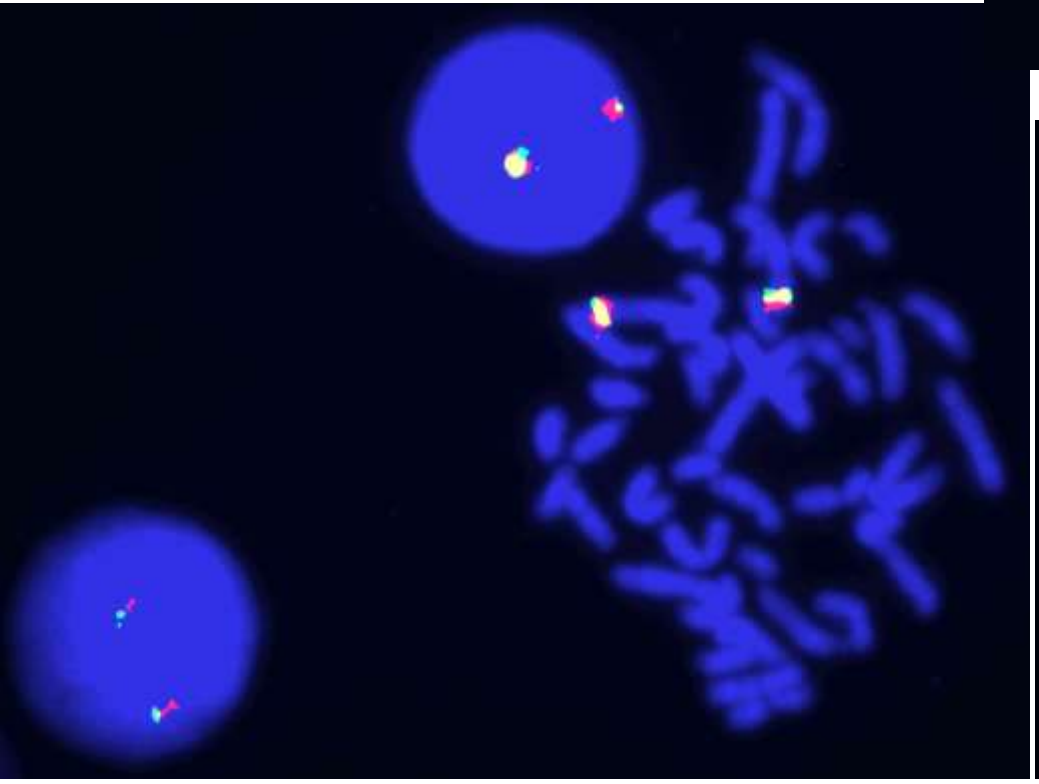
- Archer

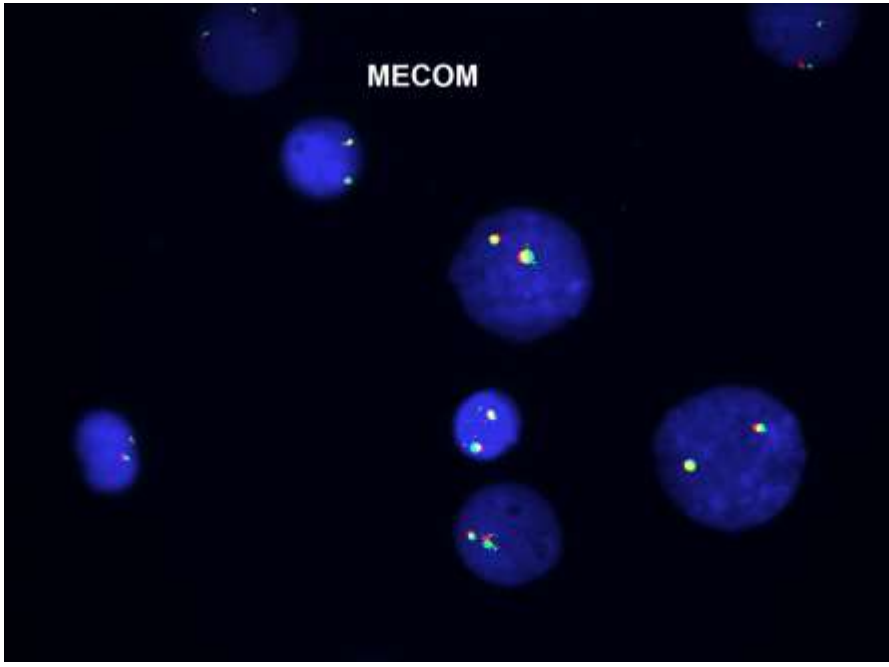
RAB7A → MECOM	22	41	50.62	True	chr3:128445202,chr3:168861620	Fusion

- En FISH
  - Pas de remaniement, spots identiques.
  - RAB7A 3q21: Insertion?




MECOM




MECOM

172920697 / 201715495ST

- LAM secondaire
- 46,X,-Y,?inv(2)(p25q12),t(<u>2</u>;11)(p21;q23),add(4)(p15), t(7;13)(q21;q31),der(11)del(11)(p15)del(11)(q23),t(?;17)(?;q12), +1~2mar[11].ish t(7;13)(q21;q31)(wcp7-;wcp7+)[10],t(?;11) (pter;q14)(MLL+;MLL-)[30],del(11)(q14)(NUP98+)[14], t(?;17)(?;q12)(TP53-,MPO+;TP53-,MPO-)[5]/11p15(NUP98+)[6] .nuc ish(TP53x1,MPOx2)[100]/(MLLx2)[200],(NUP98x2)[200]

- Archer
  - Rien

TLN1 → PRDM16	7	9	37.5	False	chr9:35717143,chr1:2985754

202020732 / 9400657RR

- LAM secondaire
- 46,XX[20].ish t(11;?19)(q23;?p13)(5'KMT2A+;3'KMT2A+)[3]/ 11q23(KMT2A+)[17].nuc ish(KMT2Ax2)(5'KMT2A sep 3'KMT2Ax1)[147/200]

- Archer

TFG → ADGRG7	178	678	41.62	True	chr3:100438902,chr3:100348442	Fusion
KMT2A → EP300	161	581	42.56	True	chr11:118355690,chr22:41547837	Fusion

Décrit dans des pathologies lymphoides et présente dans un autre échantillon de la série…

LYMPHOIDE

04H07072

- LYMPHOME MALIN NON HODGKINIEN DIFFUS A PETITES ET GRANDES CELLULES DE PHENOTYPE T mais doute sur un B au départ
- En 2012, 46,XY[30]
- Archer
  - Rien

PAX5 → ANKRD30BL	10	12	54.55	False	chr9:37006506,chr2:133013143	Fusion
IKZF3 → NOTCH2NL	8	9	40.91	False	chr17:37948937,chr1:145277343	Fusion

19H05011 / 201906174MF

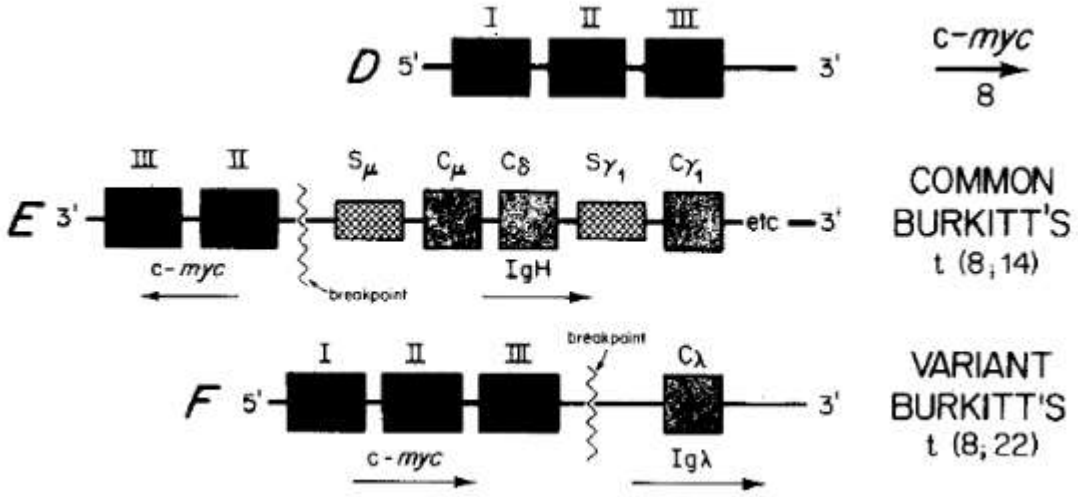
- DLBCL rearrangement MYC
- nuc ish(BCL6x2)[24]/(BCL6x3)[19]/(BCL6x4)[5], (MYC,IGH) x2~4(MYC con IGHx1~2)[60]/(MYC,IGH)x4(MYC con IGHx1)[11] /(MYC,IGH)x2[25],(BCL2x1)[16]/(BCL2x2)[34]/(BCL2x3)[10]

- Archer
  - rien

LINC00486 → NOTCH1	7	7	53.85	False	chr2:33141630,chr9:139400387

Myc et Archer

- Exon 1 et 2



- IGH n’est pas dans le panel.

20H00669 / 8707113LH

- T LAI
- Pas de CytoG
- Archer

TFG → ADGRG7	74	242	48.69	True	chr3:100438902,chr3:100348442	Fusion

Sci Rep. 2019 Mar 26;9(1):5179. doi: 10.1038/s41598-019-41675-3.

Detection of novel fusion-transcripts by RNA-Seq in T-cell lymphoblastic lymphoma

Pilar López-Nieva [1 2 3], Pablo Fernández-Navarro [4 5], Osvaldo Graña-Castro [6], Eduardo Andrés-León [7], Javier Santos [1 2 3], María Villa-Morales [1 2 3], María Ángeles Cobos-Fernández [1 2 3], Laura González-Sánchez [1 2 3], Marcos Malumbres [8], María Salazar-Roa [8], José Fernández-Piqueras [9 10 11]

20H05882 / 202010115NS

- ALCL ALK+

- nuc ish (ALKx2)(5'ALK sep 3'ALKx1)[35]/(ALKx2)[15]

- Archer

ATIC → ALK	129	1226	96.08	True	chr2:216191701,chr2:29446394	Fusion
ATIC → ALK	11	19	2.31	True	chr2:216190861,chr2:29446394	Fusion

20h06463 /201812616HP

- Poursuite évolutive de l'hémopathie lymphoïde T connue et diagnostiquée en 2018 (*18P00368*), répondant à un lymphome T particulier par l'expression forte et homogène du CD30, pouvant faire discuter les hypothèses de PTCL NOS (CD30+) et ALCL (lymphome anaplasique à grandes cellules) ALK-. On complète les investigations par la recherche de réarrangement du gène DUSP22 par technique Fish.

Discuter l'intérêt de la recherche de mutation des gènes STAT3 et JAK 1 et de la translocation de ROS et TYK2, pour progresser dans la classification de cette hémopathie.

- *nuc ish(IRF4,DUSP22)x2[19]/(IRF4,DUSP22)x3[81]*

IKZF1	267	2224	21.08	True	chr7:50367353,chr7:50459427	Oncogenic Isoform	Exon(s) Skipped
IKZF1	66	131	0.74	True	chr7:50367353,chr7:50467616	Oncogenic Isoform	Exon(s) Skipped

20H07290 / 202012610RP

- lymphome B diffus à grandes cellules de type lymphome B primitif du médiastin
- FISH, pas de remaniement de MYC ou de BCL2

- Archer
  - Rien meme en low confidence

20H07395 / 202008624EM

- LYMPHOME B DIFFUS A GRANDES CELLULES, de morphologie anaplasique, d'origine non centro-germinative (CD10-, BCL6-, MUM1-) non double expresseur (C-MYC-, BCL2+), EBV-, chez une patiente transplantée.
- FISH pas de remaniement de BCL2, MYC ou BCL6

- Archer
  - Rien meme en low confidence

20H05351 / 202007920BP

- LNH folliculaire
- 46,XX,del(6)(q12q25),add(7)(p15),del(10)(q21),del(13)(q14),t(14;18)(q32;q21)[9]/46,XX[3]

- Archer

BCL2 → UNALIGNED → IGH	363	2603	97.97	True	chr18:60793441,chr1:1|chr1:10,chr14:106330466	Fusion
BCL2 → UNALIGNED → IGH	15	27	1.15	True	chr18:60793441,chr1:1|chr1:17,chr14:106330058	Fusion
BCL2 → UNALIGNED → IGH	7	9	2.3	True	chr18:60793559,chr1:1|chr1:19,chr14:106330466	Fusion