

Exercice Nomenclature

Journée du GFCH 03/06/21

Elise Chapiro
Matthieu Decamp
Isabelle Luquet
Christine Terre



Homme
49 ans
LMC



1



2



3



4



5



6



7



8



9



10



11



12



13



14



15



16



17



18



19



20



21



22

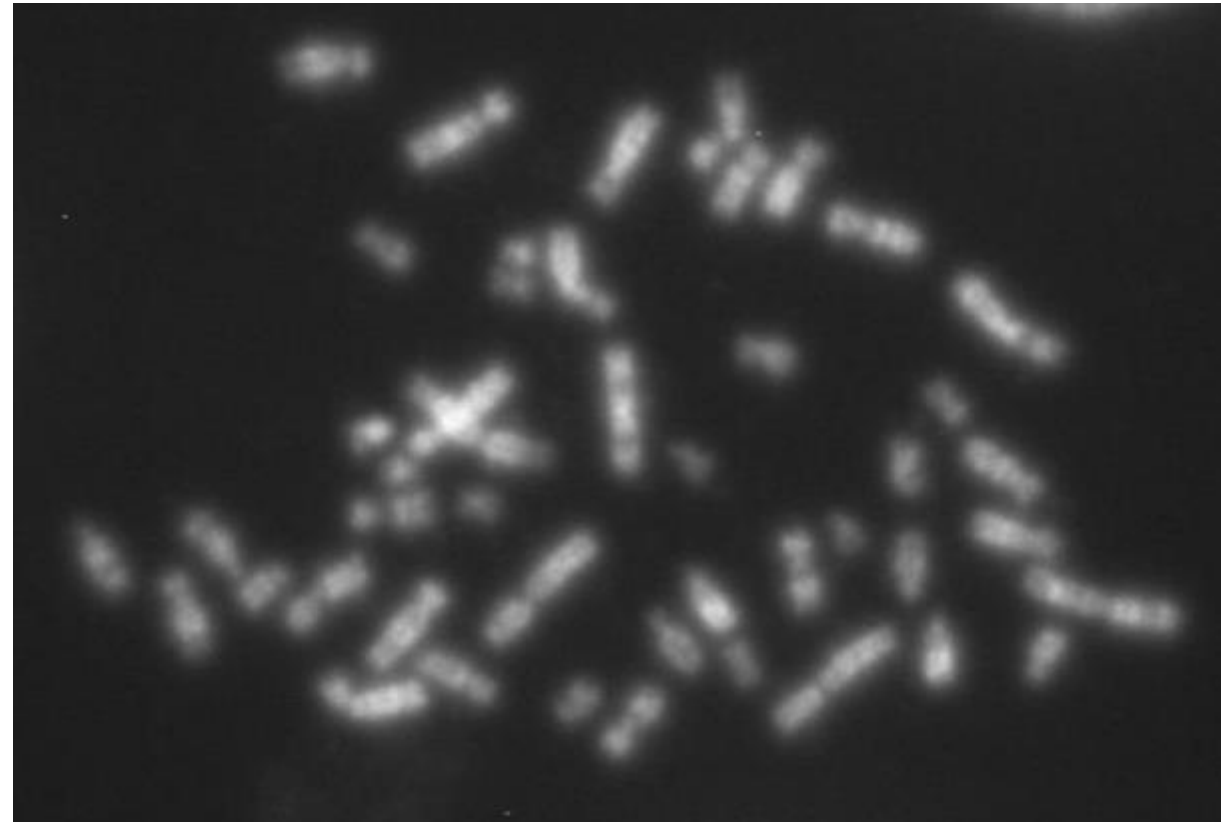
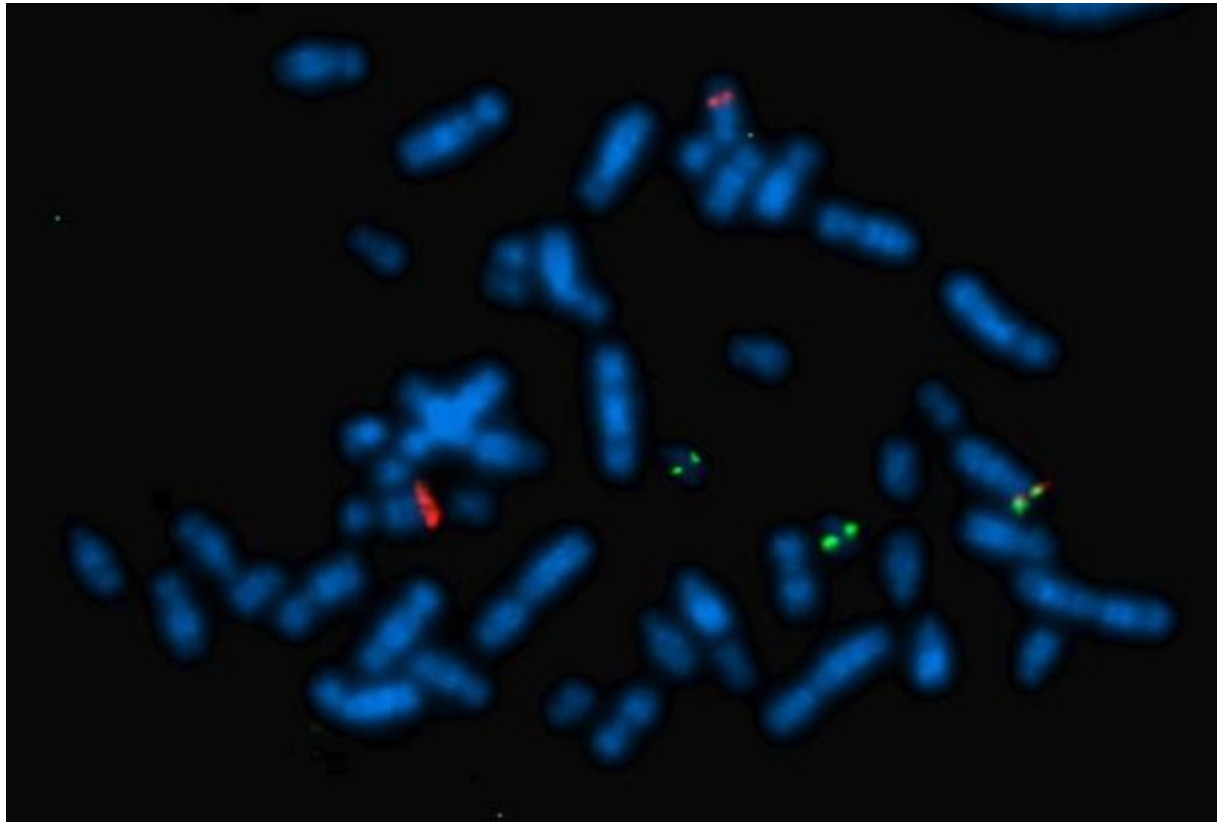
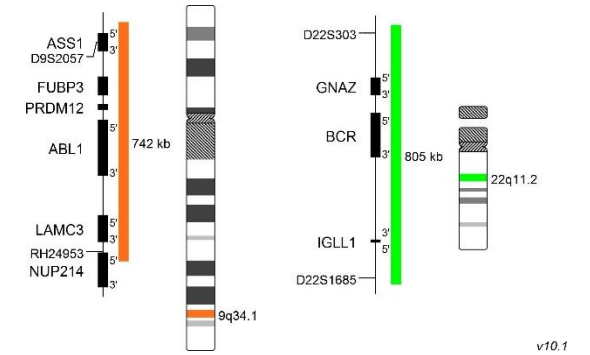


X

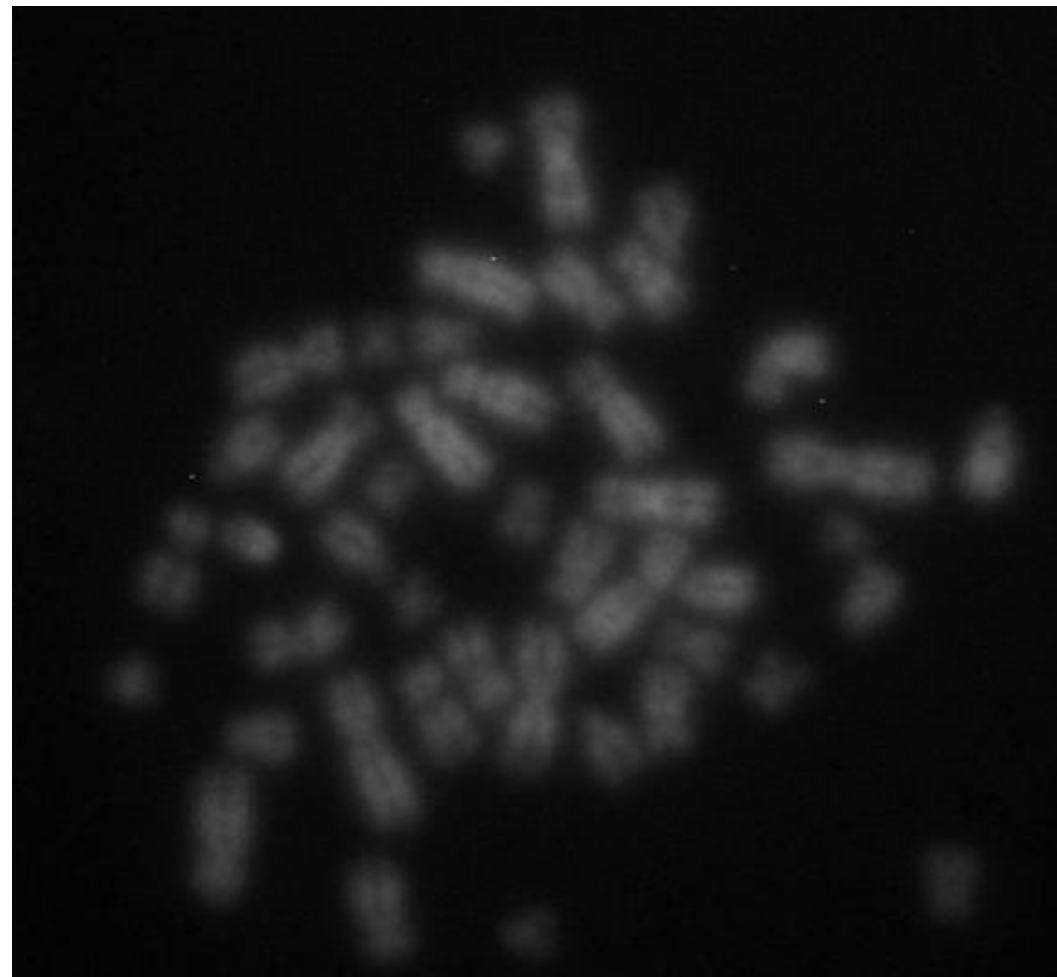
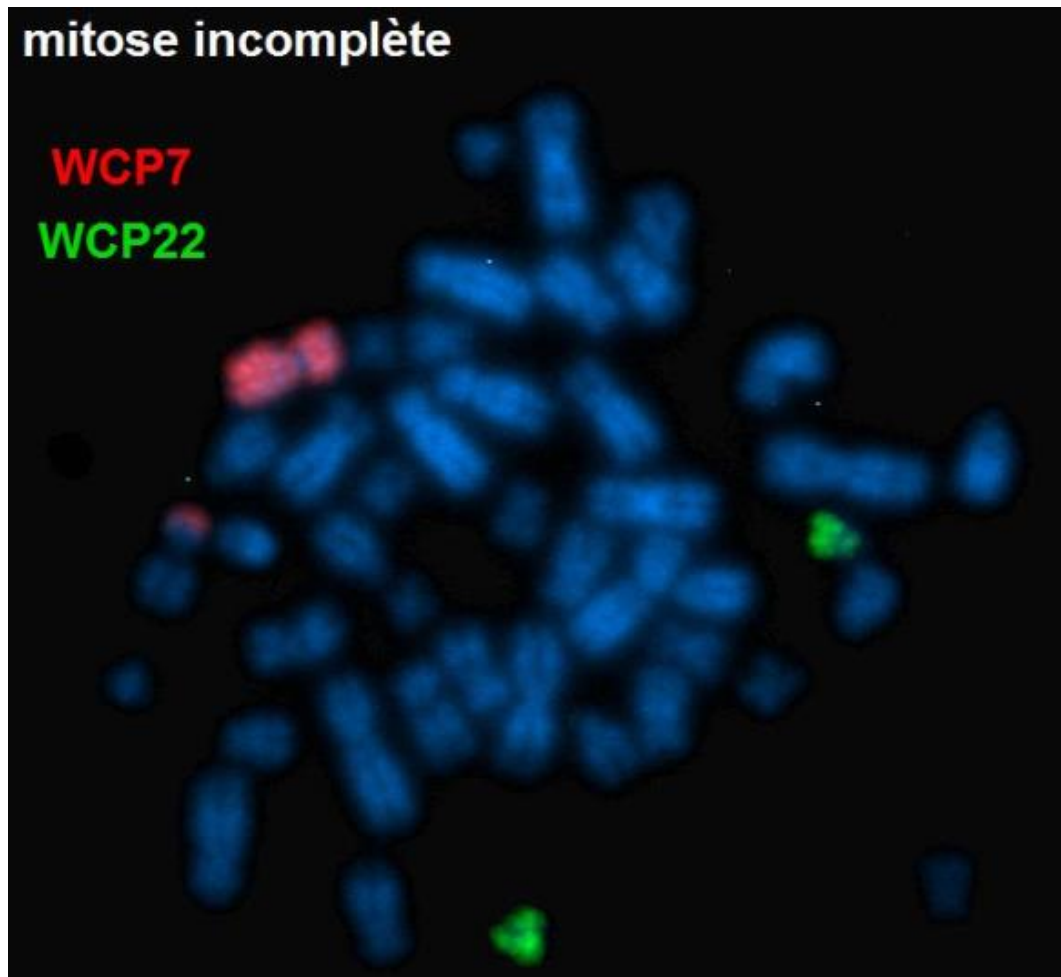


Y

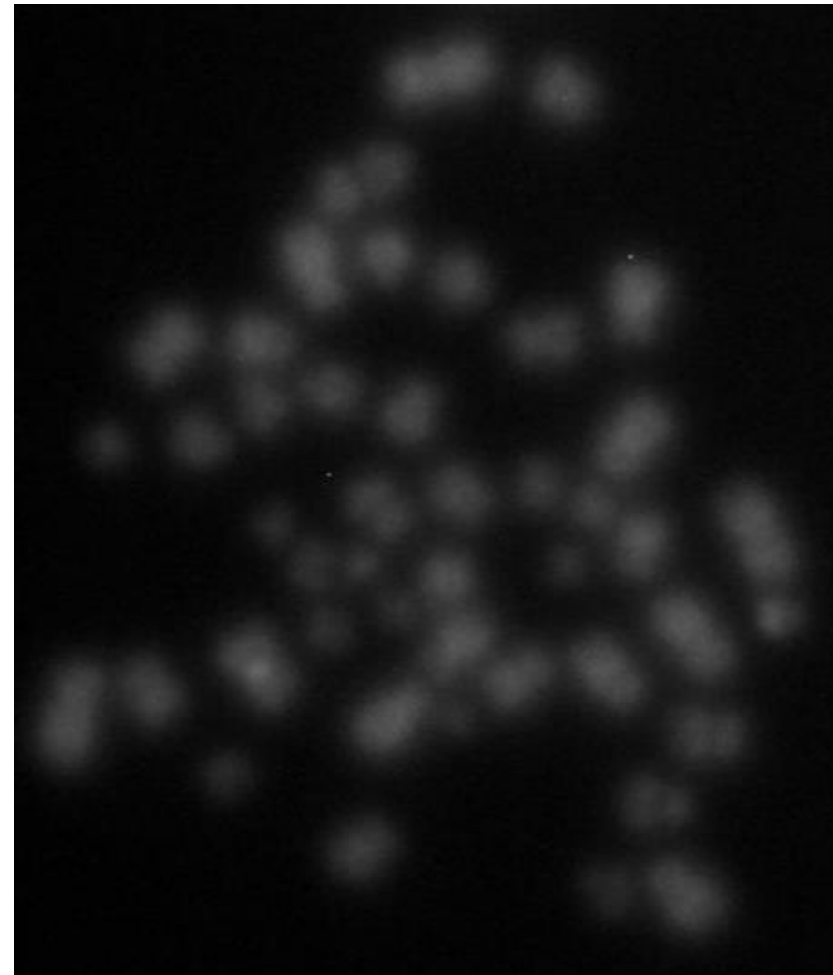
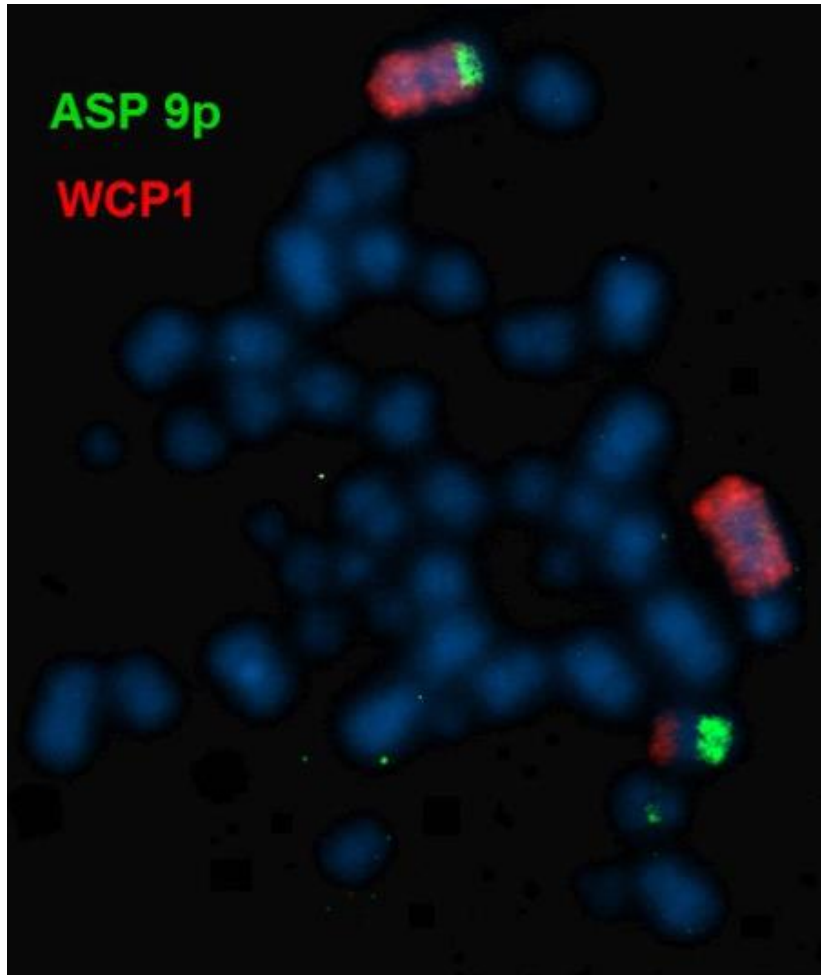
Sonde XL BCR/ABL1 plus (Metasystems)

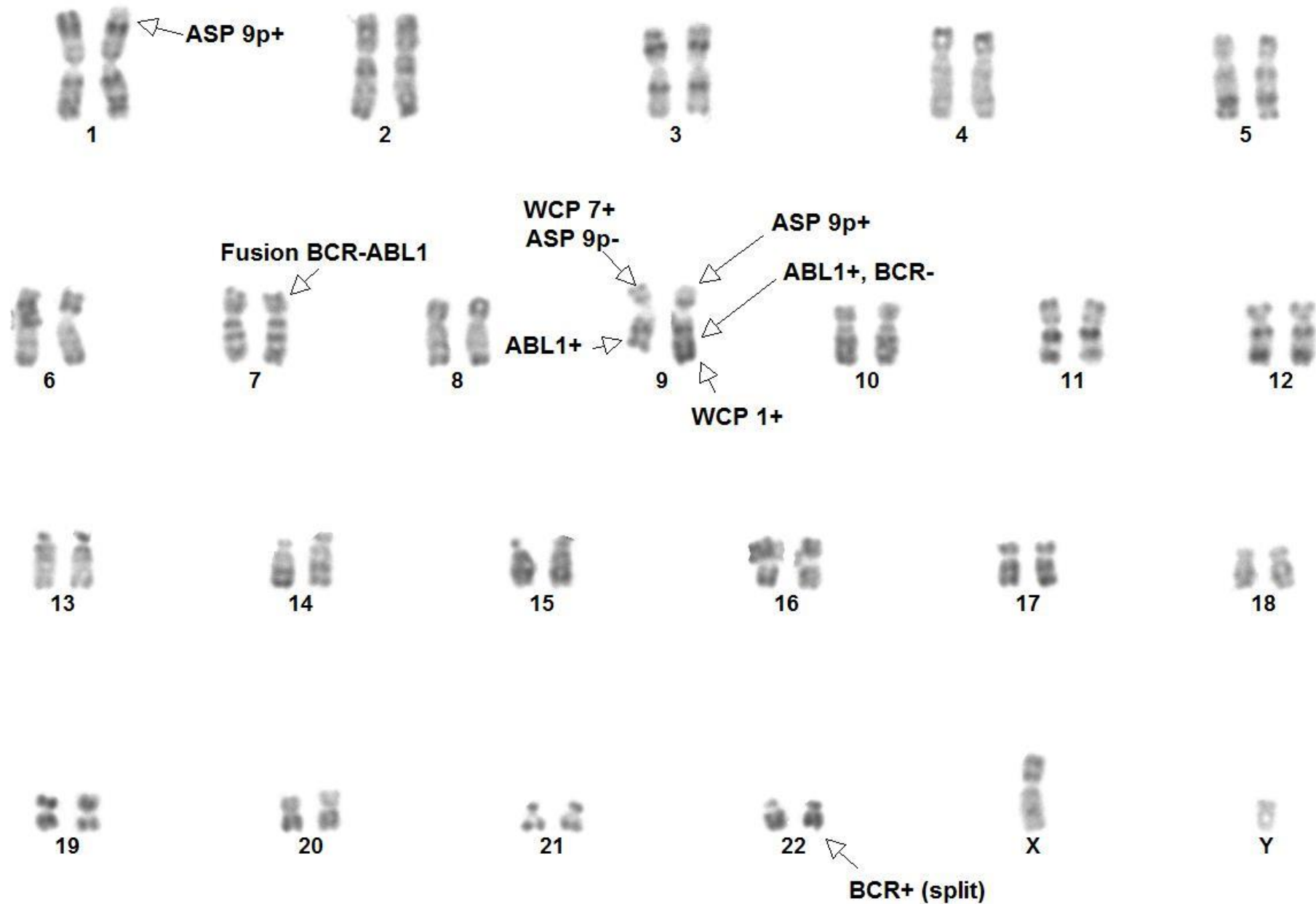


Peinture 7/22



Peinture 1/9p



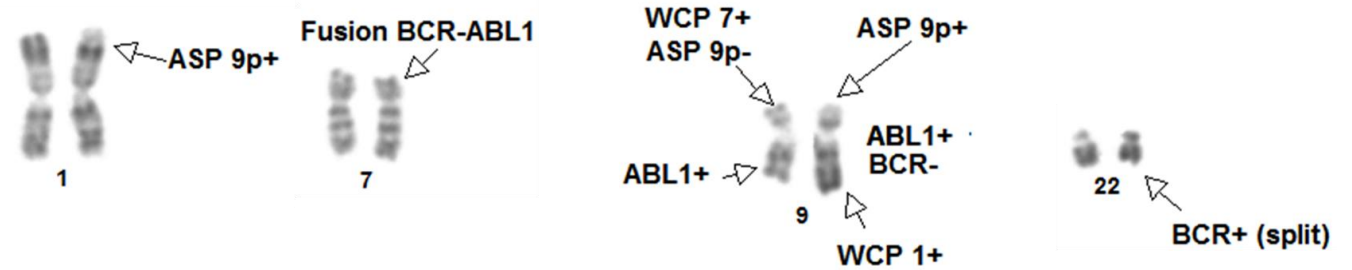


Formule ?

Les 22 semblent entièrement peints

Propositions reçues (Caryotype)

12 réponses



46,XY,add(1)(p33),add(7)(p14),add(9)(q34),add(9)(p13)[2]

46,XY,der(1)t(1;9)(p3?4;p1?3),add(7)(p1?5),add(9)(p1?3),der(9)?t(9;22)(q34;q11)

46,XY,der(1)t(1;9)(p33;p12),del(7)(p13p15),add(9)(p13),der(9)t(1;9)(p33;q34)[2]

46,XY,der(1)(9pter→9p12 ::1p34→1qter),der(7)(9qter→9q34 ::22q11→22q12 ::7p14→7qter),der(9)(7pter→7p14 ::9p12→9qter),der(9)(9pter→9q34 ::1p34→1pter),der(22)(22pter→22q11 ::22q12→22qter)[2]

46,XY,der(1)(9pter→9p1?1::1p3?5→1qter),der(7)(9qter→9q34::22q11→22q11::7p1?4→7qter),der(9)(7pter→7p1?4::9p1?1→9qter),der(9)(9pter→9q34::1p3?5→1pter)

46,XY,der(1)t(9;7;9;1;9)(q34;p21;p22;p36;q22),der(7)ins(9;22)(q34 ;q11q11) t(9;7;9;1;9),der(9) t(9;7;9;1;9),der(9)t(9;7;9;1;9)[2]

46,XY,der(1)t(9;7;9;1)(q34;p21;p13;p34),der(7)ins(9;22)(q34;q11q11)t(9;7;9;1),der(9)ins(9;22)t(9;7;9;1),der(9)t(9;7;9;1),der(22)ins(9;22)t(9;7;9;1)[2]

46,XY,der(1)t(1;9;7;9)(p33;q34;p15;p12),der(7)t(1;9;7;9)ins(7;22)(p15;q11q11),der(9)t(1;9;7;9),der(9)t(1;9;7;9)

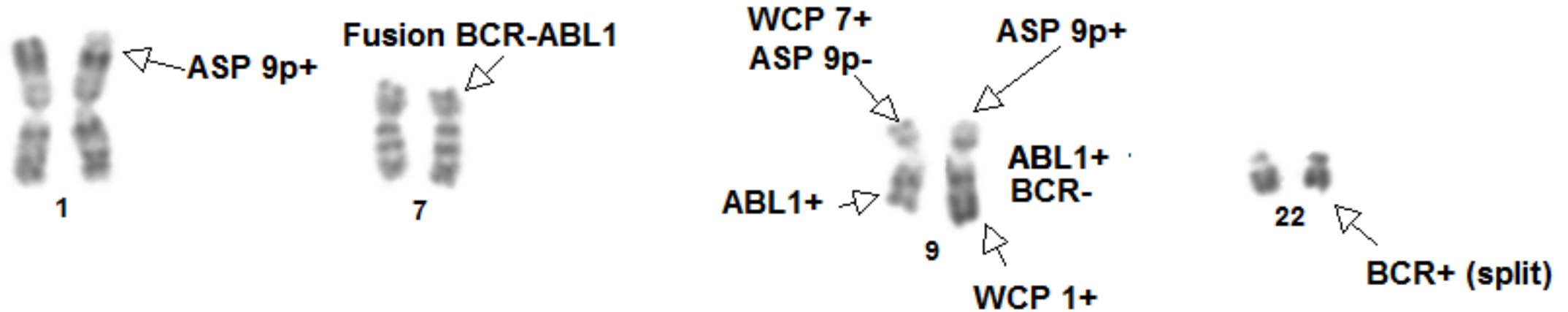
46,XY,t(1;9;7;9)(p33;q34;p25;p13)

46,XY,t(1;9;7;9)(p34;q34;p1?5;p1?3)[2]

46,XY,t(1;9;7;9)(p32;q34;p10;p10),ins(9;22)(q34;q11q11)[2]

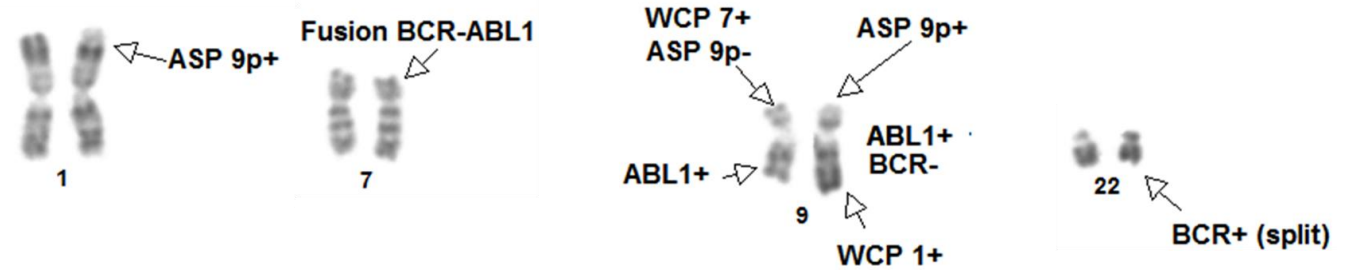
Propositions (Caryotype)

translocation $t(1;9;7;9)$ avec insertion de *BCR* dans *ABL1*



46,XY,der(1)t(1;9)(p33;p12),add(7)(p21),der(9)t(7;9)(p21;p12),der(9)t(1;9)t(1;9)(p33;q34),add(22)(q11)

Propositions (FISH)



Description en 1 temps

ish t(1;9;22;7;9)(p33;q34;q11;p14;p13)(ASP9p+,WCP1+ ;ASP9p+,ABL1+,BCR-,WCP1+;
WCP22+,BCRdim;BCR+,ABL1+;ASP9-,WCP7+,ABL1+)

ish t(1;9;7;9)(ASP9p+,WCP1+;ASP9p+,ABL1+,WCP1+;BCR+,ABL1+,WCP7+;WCP7+,ASP9p-,ABL1+),der(22)ins(9;22)(BCR+)

Description dérivé par dérivé

der(1)t(1;9)(ASP9p+,WCP1+)

der(1)(ASP9+,wcp1+)

der(1)(asp9p+,wcp1+)

der(1)(ASP9p+,WCP1+)

der(1)t(1;9;7;9)(asp9p+,wcp1+)

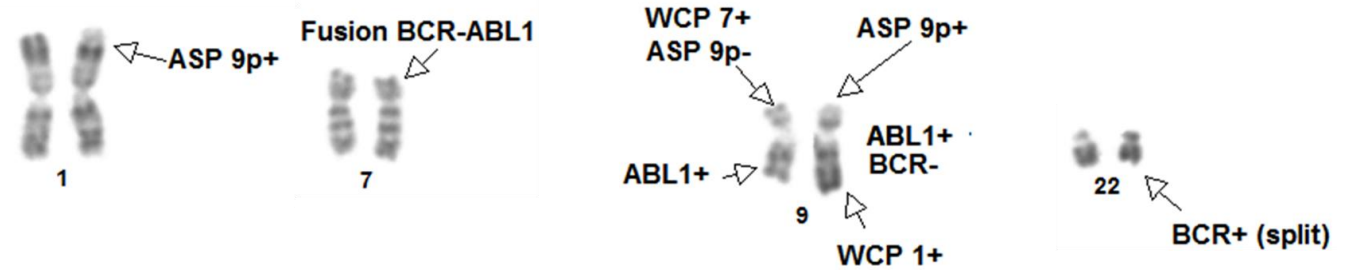
der(1)t(1;9)(p34;p21)(asp9p+,wcp1+)

der(1)(pcp9p+,wcp1+)

pcp9p+ (p464 ISCN2020)

Peintures en minuscules, gènes en majuscules

Propositions (FISH)



Description en 1 temps

ish t(1;9;22;7;9)(p33;q34;q11;p14;p13)(ASP9p+,WCP1+ ;ASP9p+,ABL1+,BCR-,WCP1+;
WCP22+,BCRdim;BCR+,ABL1+;ASP9-,WCP7+,ABL1+)

ish t(1;9;7;9)(ASP9p+,WCP1+;ASP9p+,ABL1+,WCP1+;BCR+,ABL1+,WCP7+;WCP7+,ASP9p-,ABL1+),der(22)ins(9;22)(BCR+)

Description dérivé par dérivé

der(1)t(1;9)(ASP9p+,WCP1+)

der(1)(ASP9+,wcp1+)

der(1)(asp9p+,wcp1+)

der(1)(ASP9p+,WCP1+)

der(1)t(1;9;7;9)(asp9p+,wcp1+)

der(1)t(1;9)(p34;p21)(asp9p+,wcp1+)

der(7)t(9;22;7)(ABL1+,BCR+)

der(7)(BCR+,ABL1+,wcp7+)

der(7)(BCR+,ABL1+,wcp7+)

der(7)(ABL1+,BCR+)

der(7)(9qter->9q34::22q11->22q11::7p25->7qter)(ABL1+,BCR+,wcp7+)

der(7)t(1;9;7;9)(ABL1+,wcp7+)ins(9;22)(q34 ;q11.2q11.2)(BCR+)

der(7)t(7;9)(p21;q34)t(9;22)(q34;q11)(bcr+,abl1+)

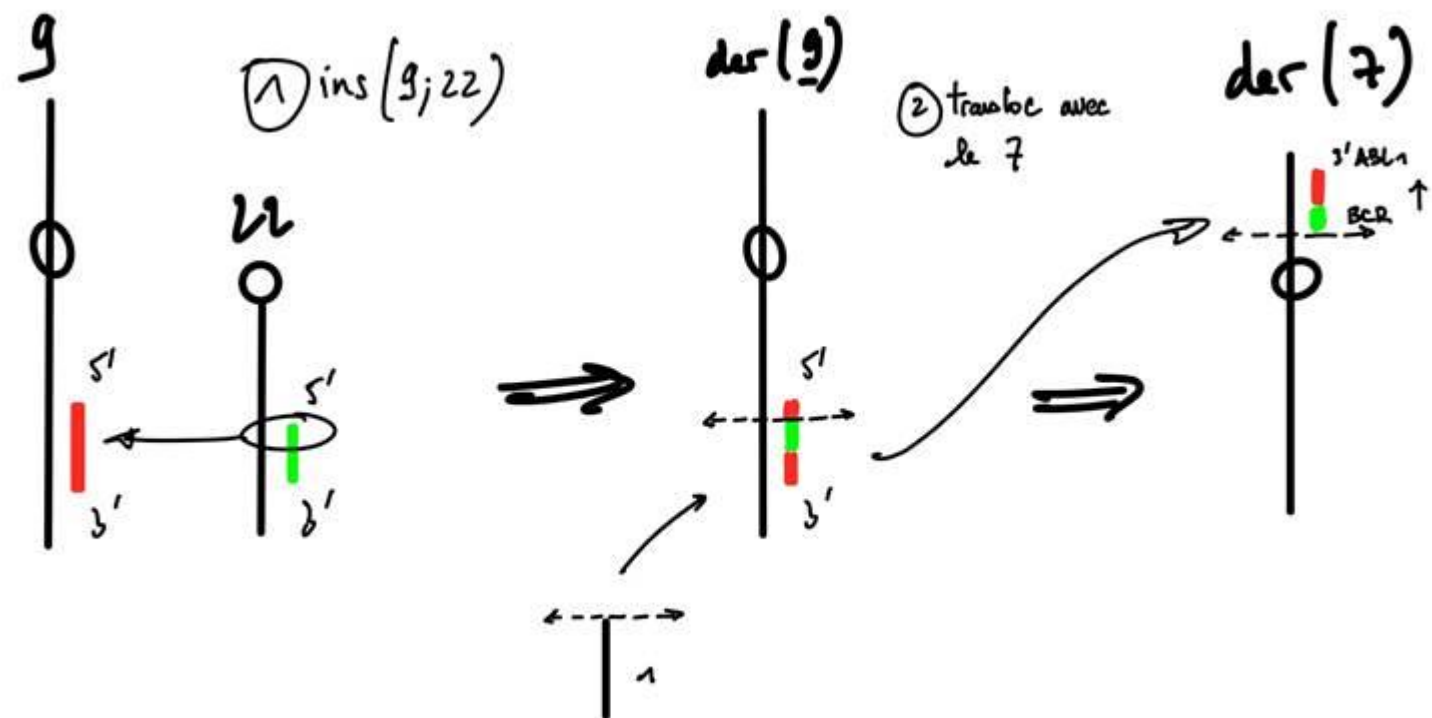
del(7)(ABL1+,BCR+,wcp7+)

der(1)(pcp9p+,wcp1+)

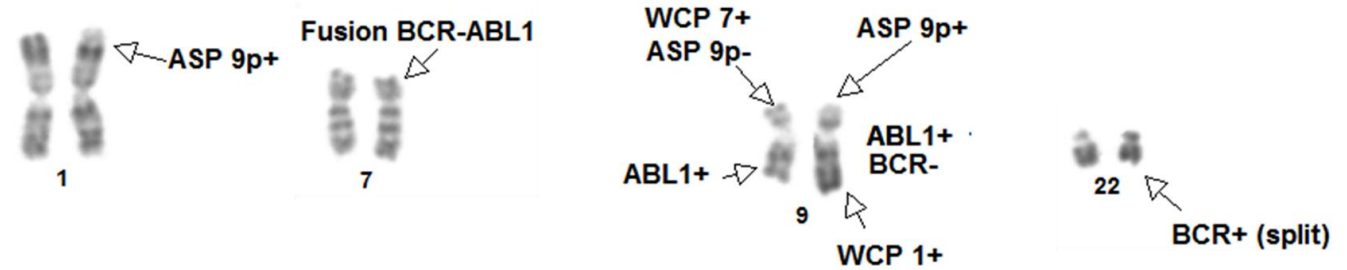
der(7)(9qter→9q34::22q11→22q11::7p15→7qter)(ABL1+,BCR+,wcp7+)

pcp9p+ (p464 ISCN2020)

Peintures en minuscules, gènes en majuscules



Propositions (FISH)



Description en 1 temps

ish t(1;9;22;7;9)(p33;q34;q11;p14;p13)(ASP9p+,WCP1+ ;ASP9p+,ABL1+,BCR-,WCP1+;
WCP22+,BCRdim;BCR+,ABL1+;ASP9-,WCP7+,ABL1+)

ish t(1;9;7;9)(ASP9p+,WCP1+;ASP9p+,ABL1+,WCP1+;BCR+,ABL1+,WCP7+;WCP7+,ASP9p-,ABL1+),der(22)ins(9;22)(BCR+)

Description dérivé par dérivé

der(9)t(7;9)(WCP7+,ASP9p-,ABL1+),der(9)t(1;9)(ASP9p+,ABL1+,BCR-,WCP1+)

der(9)(wcp7+,ASP9p-,ABL1+,BCR-),der(9)(ASP9+,ABL1+,BCR1-,WCP1+)

der(9)(wcp7+,ASP9p-,ABL1+),der(9)(ASP9p+,ABL1+,wcp1+)

der(9)(asp9p+,ABL1+,wcp1+),der(9)(wcp7+,ABL1+)

der(9)(ASP9p+,ABL1+,BCR-,WCP1+),der(9)(WCP7+,ASP9p-,ABL1+)

der(9)t(1;9;7;9)(asp9p+,ABL1+,wcp1+),der(9)t(1;9;7;9)(wcp7+,asp9p+,ABL1+)

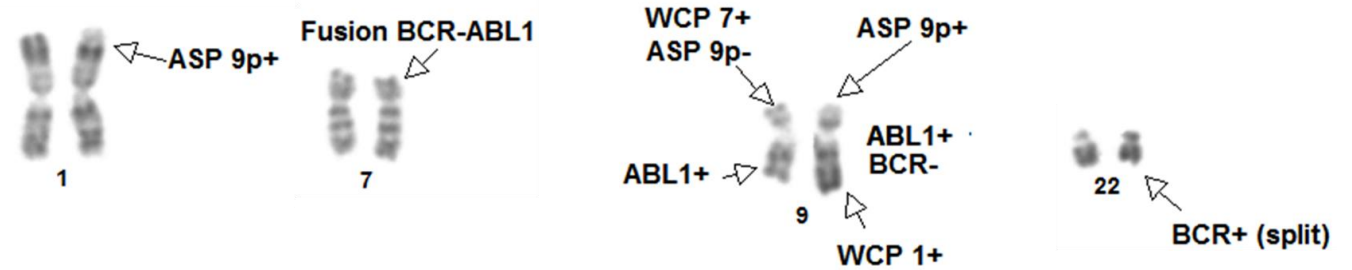
der(9)t(7;9)(p21;p21)(wcp7+,asp9-),der(9)t(1;9)(p34;q34)(abl+,wcp1+)

add(9)(wcp7+,asp9p-,ABL1+),der(9)(asp9p+,ABL1+,wcp1+)

der(9)(7pter→7p15::9p13→9qter)(pcp9p-,wcp7+,ABL1+),

der(9)(9pter→9q34::1p34→1pter)(pcp9p+,ABL1+,wcp1+)

Propositions (FISH)



Description en 1 temps

ish t(1;9;22;7;9)(p33;q34;q11;p14;p13)(ASP9p+,WCP1+ ;ASP9p+,ABL1+,BCR-,WCP1+;
WCP22+,BCRdim;BCR+,ABL1+;ASP9-,WCP7+,ABL1+)

ish t(1;9;7;9)(ASP9p+,WCP1+;ASP9p+,ABL1+,WCP1+;BCR+,ABL1+,WCP7+;WCP7+,ASP9p-,ABL1+),der(22)ins(9;22)(BCR+)

Description dérivé par dérivé

der(9)t(7;9)(WCP7+,ASP9p-,ABL1+),der(9)t(1;9)(ASP9p+,ABL1+,BCR-,WCP1+)
der(9)(wcp7+,ASP9p-,ABL1+,BCR-),der(9)(ASP9+,ABL1+,BCR1-,WCP1+)
der(9)(wcp7+,ASP9p-,ABL1+),der(9)(ASP9p+,ABL1+,wcp1+)
der(9)(asp9p+,ABL1+,wcp1+),der(9)(wcp7+,ABL1+)
der(9)(ASP9p+,ABL1+,BCR-,WCP1+),der(9)(WCP7+,ASP9p-,ABL1+)
der(9)t(1;9;7;9)(asp9p+,ABL1+,wcp1+),der(9)t(1;9;7;9)(wcp7+,asp9p+,ABL1+)
der(9)t(7;9)(p21;p21)(wcp7+,asp9-),der(9)t(1;9)(p34;q34)(abl+,wcp1+)
add(9)(wcp7+,asp9p-,ABL1+),der(9)(asp9p+,ABL1+,wcp1+)

22q11(BCRx2)
22q11(wcp22+,BCRdim+)
22q11.2(wcp22+,BCR dim)
der(22)(wcp22+,BCR+)
der(22)(BCR+)
del(22)(q11.2q11.2)(BCR+)
der(22)ins(9;22)(BCR+)
der(22)(bcr+,wcp22+)

del(22)(q11q11)(wcp22+,BCR+)

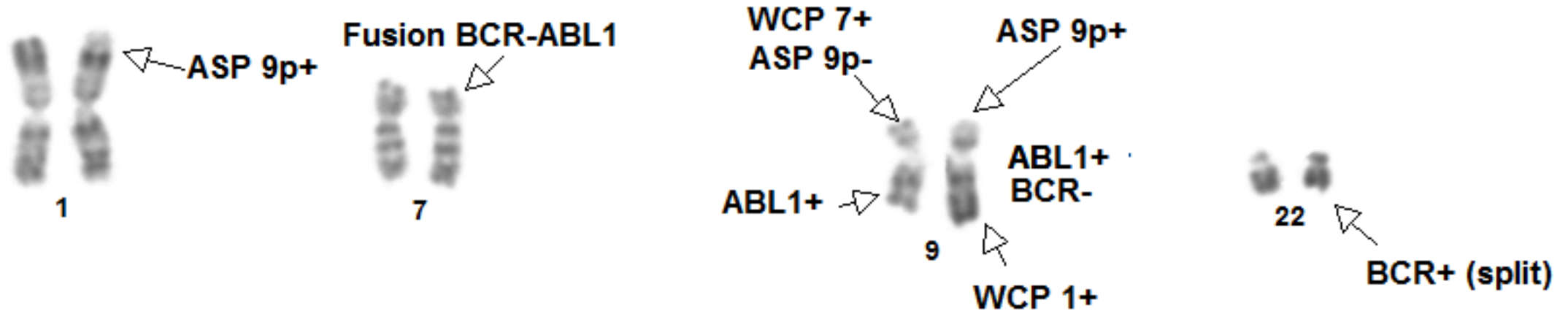
der(9)(7pter→7p15::9p13→9qter)(pcp9p-,wcp7+,ABL1+),
der(9)(9pter→9q34::1p34→1pter)(pcp9p+,ABL1+,wcp1+)

dim 13.2.6 p454 (pas de +/-)

Atelier nomenclature2020 : dim pour anomalies
déséquilibrées

Formule complète

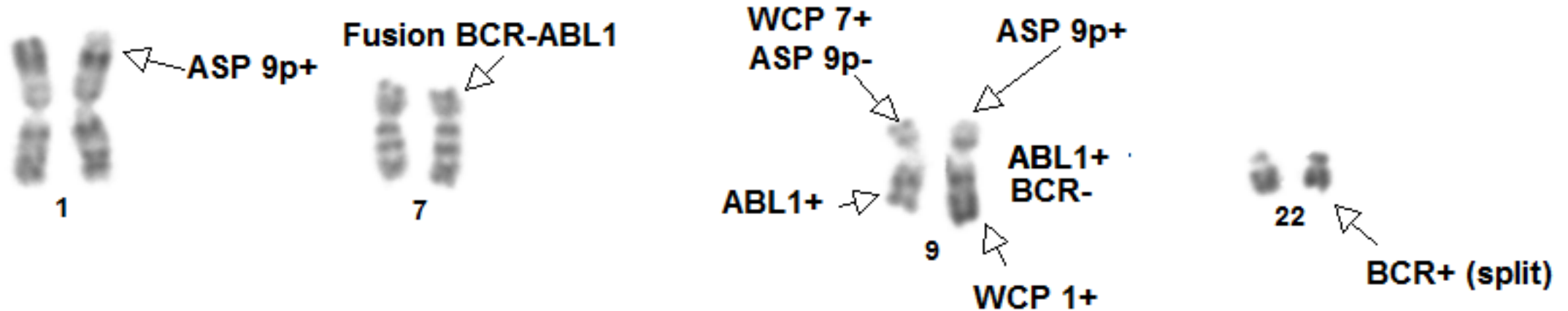
translocation $t(1;9;7;9)$ avec insertion de *BCR* dans *ABL1*



46,XY,der(1)t(1;9)(p33;p12),add(7)(p21),der(9)t(7;9)(p21;p12),der(9)t(1;9)(p33;q34),**add(22)(q11)**[...]
 .ish der(1)(pcp9p+,wcp1+),der(7)(9qter→9q34::22q11→22q11::7p15→7qter)(ABL1+,BCR+,wcp7+),
 der(9)(7pter→7p15::9p13→9qter)(pcp9p-,wcp7+,ABL1+),der(9)(9pter→9q34::1p34→1pter)(pcp9p+,ABL1+,wcp1+),
 del(22)(q11q11)(wcp22+,BCR+)[...]

Formule complète

translocation $t(1;9;7;9)$ avec insertion de *BCR* dans *ABL1*



46,XY,der(1)t(1;9)(p33;p12),add(7)(p21),der(9)t(7;9)(p21;p12),der(9)t(1;9)(p33;q34),**add(22)(q11)**[...]
 .ish der(1)(pcp9p+,wcp1+),der(7)(9qter→9q34::22q11→22q11::7p15→7qter)(ABL1+,BCR+,wcp7+),
 der(9)(7pter→7p15::9p13→9qter)(pcp9p-,wcp7+,ABL1+),der(9)(9pter→9q34::1p34→1pter)(pcp9p+,ABL1+,wcp1+),
 del(22)(q11q11)(wcp22+,BCR+)[...]

Merci !