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Case Report Section

A pediatric case of acute lymphoblastic leukemia with t(2;9)(q12;q34) (RANBP2/ABL1 fusion)

Marc De Braekeleer, Nadia Guéganic, Alexandra Schifferli, Joëlle Tchinda

Cytogenetics Laboratory, Faculty of Medicine, University of Brest, France marc.debraekeleer@univ-brest.fr (MdeB, NG)) Department of Hematology/Oncology, University Children's Hospital Basel, Switzerland (AS) University Children's Hospital Zurich, Switzerland (JT).

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Clinics

Age and sex 21 months old female patient.

Previous history
no preleukemia
no previous malignancy
no inborn condition of note

Organomegaly
No hepatomegaly , no splenomegaly , no enlarged lymph nodes , no central nervous system involvement (t)

Blood

WBC : 77.5 (N: 6-17.5)X 10^9/l
HB : 29 (N: 105-135)g/dl
Platelets : 69 (N: 150-450)X 10^9/l
Blasts : 76%
Bone marrow : Hypercellular marrow, with 93.7% blasts (small to middle-sized cells with large nucleus and minimal cytoplasm).%

Cyto-Pathology Classification

Phenotype
Pre-B acute lymphoblastic leukemia.

Immunophenotype
cTDT, cCD79a, cIgM, CD19 and CD20 positive.

Rearranged Ig Tcr not performed.

Diagnosis Pre-B acute lymphoblastic leukemia.

Survival

Date of diagnosis 01-2014

Treatment
Protocol AIEOP-BFM ALL 2009 high risk.
Complete remission :
Treatment related death : no
Relapse : no

Status A

Last follow up 12-2015

Survival 23 +months

Karyotype

Sample bone marrow.

Banding G banding.

Results
46,XX,t(2;9)(q12-14;q34),add(5)(p14)[5]/46,sl,-7,+mar[2]/46,XX[3]

Other molecular cytogenetics technics
fluorescence in situ hybridization(FISH) analysis using ETV6-RUNX1, 5'MLL-3'MLL, CEP4, CEP10, CEP17, 5'IGH-3'IGH, 3'TCF3-5'TCF3, BCR-ABL1.

FISH experiments with BAC clones located in bands 2q12.1 to 2q14.2.

Other molecular cytogenetics results

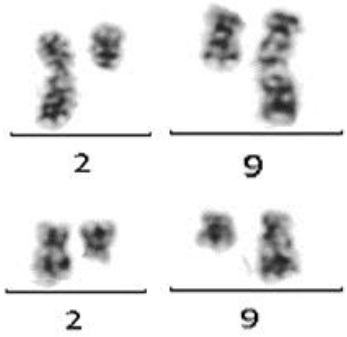
All negative, except for BCR-ABL1 with 3 ABL1 signals and 2 BCR signals.

A split signal on der(2) and der(9) was found with RP11-622D1, RP11-347H10, RP11-259O12, RP11-348G16 and RP11-953L12. These BAC clones overlap the RANBP2 gene and allow refinement of the breakpoint to a 25kb region covering the 5' end and the first three exons of RANBP2.

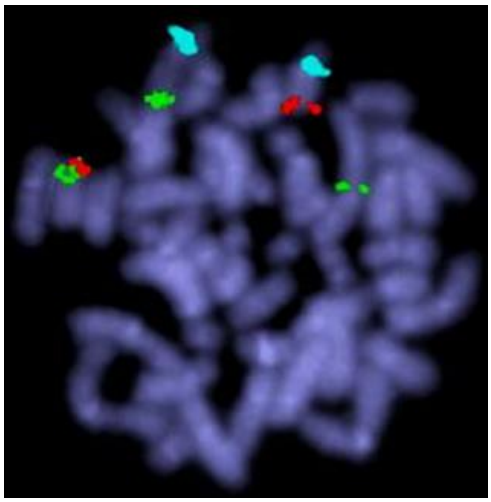
Other Molecular Studies

Technics: MLPA.

Results: Negative.



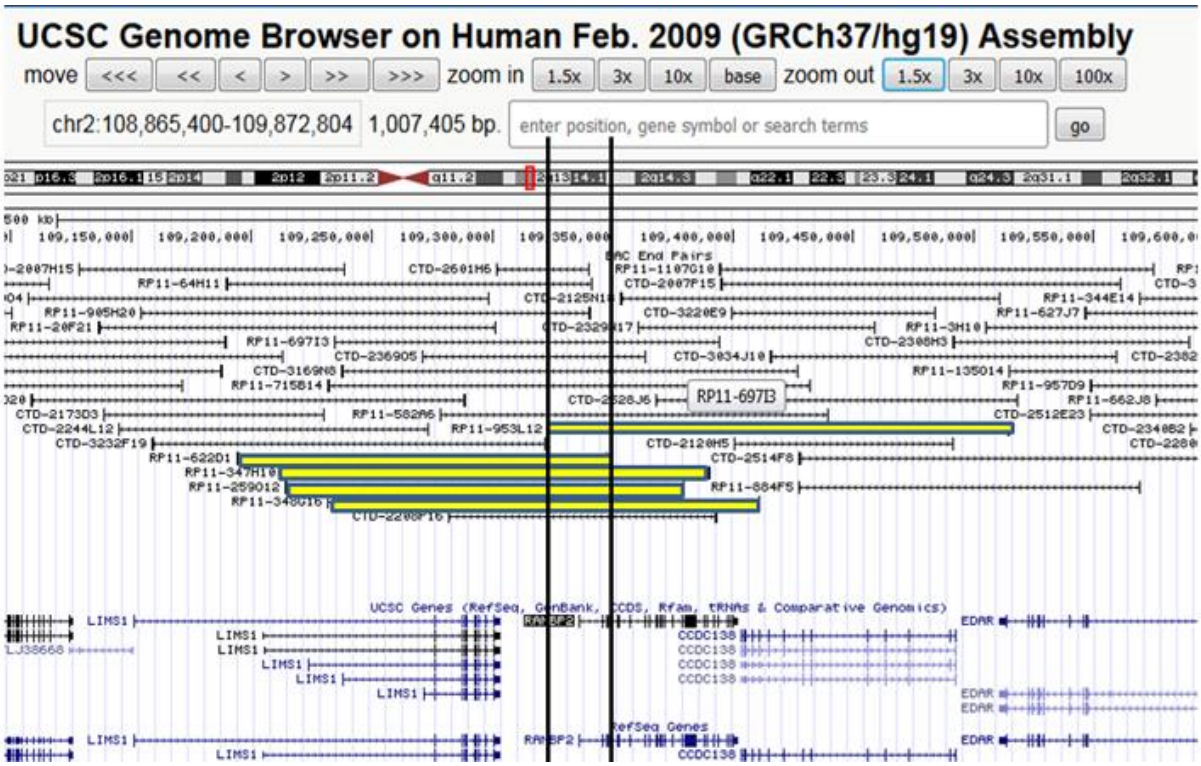
GTG banding showing chromosomes 2 and 9 and the derivatives der(2) and der(9).



FISH with BACs RP11-953L12 (spectrum green, located in 2q12 and containing RANBP2) and RP11-83J21 (spectrum orange, located in 9q34 and containing the 3' part of ABL1) and CEP9 (in aqua) showing one fusion signal on der(2). No fusion is detected on der(9) because RP11-83J21 does not cover the 5' part of ABL1.

Comments

We present here a unique case of pediatric acute lymphoblastic leukemia. This fusion gene was identified in another case by RNA-sequencing (Roberts et al., 2012).



References

Roberts KG, Morin RD, Zhang J, Hirst M, Zhao Y, Su X, Chen SC, Payne-Turner D, Churchman ML, Harvey RC, Chen X, Kasap C, Yan C, Becksfort J, Finney RP, Teachey DT, Maude SL, Tse K, Moore R, Jones S, Mungall K, Birol I, Edmonson MN, Hu Y, Buetow KE, Chen IM, Carroll WL, Wei L, Ma J, Kleppe M, Levine RL, Garcia-Manero G, Larsen E, Shah NP, Devidas M, Reaman G, Smith M, Paugh SW, Evans WE, Grupp SA, Jeha S, Pui CH, Gerhard DS, Downing JR, Willman CL, Loh M, Hunger SP, Marra MA, Mullighan CG. Genetic alterations activating kinase and cytokine receptor signaling in high-risk acute lymphoblastic leukemia. Cancer Cell. 2012 Aug 14;22(2):153-66

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