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Case Report Section

t(14;20)(q11.2;q13.3) involving the T-cell receptor α/δ gene in a pediatric acute lymphoblastic leukemia B-cell type

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Abstract

Case report on t(14;20)(q11.2;q13.3) involving the T-cell receptor α/δ gene in a pediatric acute lymphoblastic leukemia B-cell type.

Previous History

Preleukemia: no preleukemia
Previous malignancy: no previous malignancy
Inborn condition: no inborn condition of note

Clinics

Age and sex: 1 year 4 month female patient.

Organomegaly: hepatomegaly, splenomegaly, enlarged lymph nodes, no central nervous system involvement

Blood

WBC: 82.9X 10^9/l
HB: 8,6g/dl
Platelets: 43X 10^9/l
Blasts: 83% (neutrophils 2, lymphocytes 10, monocytes 1, myelocytes 2, metamyelocytes 2)
Bone marrow: Bone marrow biopsy and aspirate revealed a hypercellular marrow with 90 % blast cells.

Cyto-Pathology Classification

Phenotype B-ALL

Immunophenotype
Positive for CD 10 (71%), CD19 (80%), CD20 (41%), CD22 (72%), CD79a (81%), CD45 (98%), HLADR (94%), CD34 (33%), TdT (62%).

Survival

Date of diagnosis 02-2016

Status Alive

Karyotype

Sample Bone marrow

Culture time 24 h

Banding G-banding

Results
46,XX,?del(9)(p21),t(14;20)(q11.2;q13.3)[5]/46,XX,der(9)?del(9)(p11?p23)t(9;20) (p11;q11.2), t(14;20)(q11.2;q13.3)[5]/ 46,XX [10]

Other molecular cytogenetics technics
Fluorescence in situ hybridization (FISH) for LSI CDKN2A (9p21)/CEP9 Dual Color, LSI 20q, LSI IGH Dual Color, Break Apart (BA), WCP 20 and

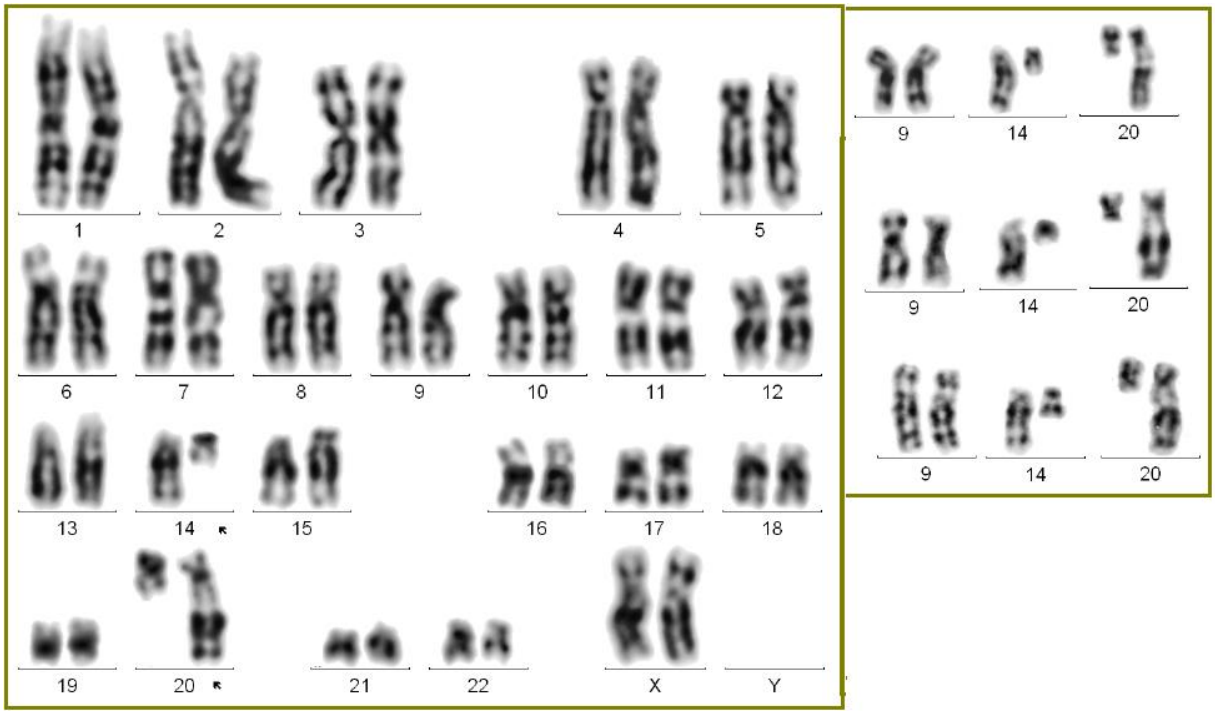
LSI TCR alpha/delta Dual Color, Break Apart Rearrangement probes (Vysis/Abbott Molecular, Des Plaines, IL, USA) according to standard techniques.

Other molecular cytogenetics results

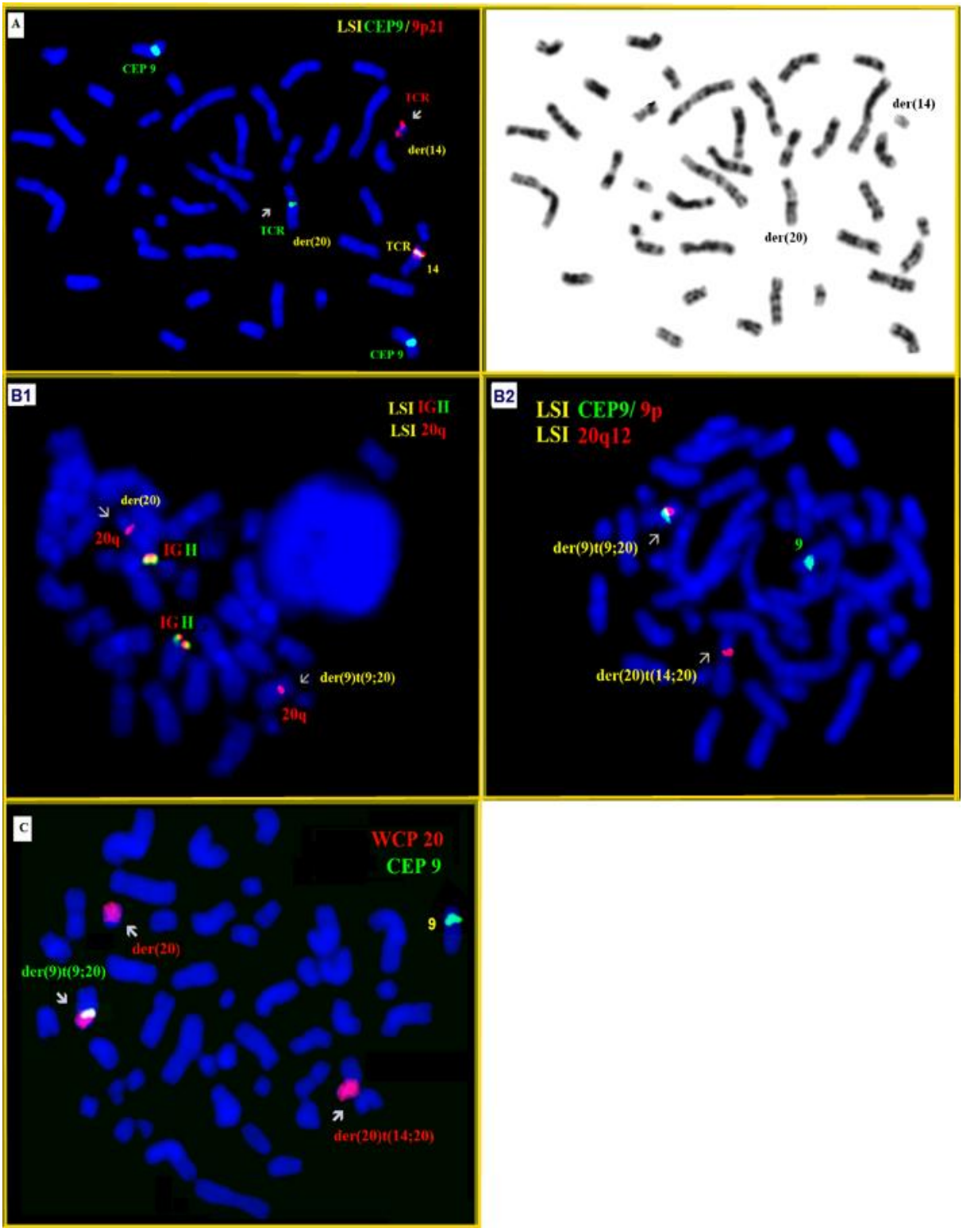
Applying the LSI TCR A/D-specific BA probe on interphase cells revealed 1 red, 1 green and 1 fusion signal pattern in 80% of cells, indicative of TCR α/δ rearrangement. Applying the LSI CDKN2A (9p21)/CEP9 probe on interphase cells revealed 2 signals (green) for chromosome 9 centromere and no red signal for CDKN2A (9p21) in 70% of cells. FISH analysis with combination of LSI TCR A/D BA and LSI CDKN2A (9p21)/CEP9 probes revealed juxtaposition of telomeric TCR sequences (green) to the der(20) chromosome as a result of t(14;20)(q11.2;q13) and 2 green chromosome 9 centromere signals, while no red signal for CDKN2A (9p21) was detected on 5 metaphases (Figure 2A). Applying the LSI 20q probe located on 20q12 revealed 2 normal signals in 98% of interphase cells. Hybridization with LSI 20q12 probe (red signals) on metaphases revealed one signal of 20q12 on der(20)t(14;20) chromosome and the other 20q12 signal on der(9) chromosome. Hybridization with LSI 20q probe on metaphases confirmed the presence of 20q12 signals (red) on der(20)t(14;20) and der(9)t(9;20) chromosomes (Figure 1B). Additional FISH analyses on 5 metaphases by using the WCP 20 and CDKN2A (9p21)/CEP9 probes (Figure 2C) confirmed the presence of chromosome 20 sequences in der(20)t(14;20) and der(9)t(9;20) chromosomes with simultaneous deletion of 9p21 sequences (no red signal was detected on chromosomes 9)

Comments

Rearrangements of the T-cell receptor α/δ genes has been only rarely observed in B-cell malignancies. We describe a B-cell ALL patient with t(14;20)(q11.2;q13.3) and t(9;20)(p11;q11) in whom FISH studies confirmed TCR α/δ disruption and homozygous deletions of 9p21. The t(14;20)(q11.2;q13) have been reported only in 2 (Erikson et al., 1986; Douet-Guilbert et al., 2004), while translocations between 9p11-21 and 20q have been found only in 4 patients (Horiike et al., 1988; UKCCG., 1992; Gardiner et al., 2012; Le Noir et al., 2012). Among them, Le Noir et al., described a T-ALL case with simultaneous t(9;20)(p21;q12) and TCR α/δ disruption. The coexistence of 9p21 deletion and TCR α/δ rearrangement in our patient may indicate cooperative oncogenesis between gene inactivation and oncogene activation in ALL.



G-banded karyotype of the patient with t(14;20)(q11.2;q13.3) and partial karyotypes with t(9;20)(p11;q11),t(14;20)(q11.2;q13.3).



FISH analysis was performed to determine the breakpoints on 14q using a T-cell receptor alpha delta (TCR A/D) DNA Probe, revealing split signal as a result of t(14;20)(q11.2;q13.3) and hybridization with the LSI CDKN2A (9p21)/CEP9) probe showed homozygous deletions of 9p21 indicative of biallelic deletion of genes from 9p21 (Figure 1A). Simultaneous hybridization with LSI IGH and 20q12 probes revealed one signal of IGH on der(20)t(14;20) chromosome distal to the 20q12 signal (red) and the other 20q12 signal on der(9) chromosome (Figure 1B1). Alternatively: hybridization with LSI 20q12 probe revealed one signal of 20q12 on der(20)t(14;20) chromosome (red) and the other 20q12 signal on der(9) chromosome (Figure 1B2). Hybridization with WCP 20 and CDKN2A (9p21)/CEP9 probes confirmed the juxtaposition of chromosome 20 sequences to der(20)t(14;20) and der(9)t(9;20) chromosomes with simultaneous deletion of 9p21 sequences (Figure 1C).

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