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Case Report Section

An adult case of biphenotypic acute leukemia with t(6;14)(q25;q32)

Toyotaka Kawamata, Miho Ogawa, Tomomi Takei, Reina Takeda, Kiyosumi Ochi, Kazuaki Yokoyama, Tomofusa Fukuyama, Nobuhiro Ohno, Kaoru Uchimaru, Arinobu Tojo

Department of Hematology/Oncology, Research Hospital (KT, OM, TT, TR, OK, YK, FT, ON, UK, TA); Division of Cell Therapy, Advanced Clinical Research Center (TF); Division of Molecular Therapy, Advanced Clinical Research Center, The Institute of Medical Science, the University of Tokyo (TA), 4-6-1 Shirokanedai, Minato-ku, Tokyo 108-8639, Japan. Toyotaka Kawamata: toyotaka@ims.u-tokyo.ac.jp, Miho Ogawa: miho.ogawa327@gmail.com; Tomomi Takei: t-takei@ims.u-tokyo.ac.jp; Reina Takeda: reina.takeda@gmail.com; Kiyosumi Ochi: equuleus2015@gmail.com; Kazuaki Yokoyama: k-yoko@ims.u-tokyo.ac.jp; Tomofusa Fukuyama: tfukuyam@ims.u-tokyo.ac.jp; Nobuhiro Ohno: nobuohno@ims.u-tokyo.ac.jp; Kaoru Uchimaru: uchimaru@ims.u-tokyo.ac.jp; Arinobu Tojo: a-tojo@ims.u-tokyo.ac.jp

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Clinics

Age and sex

66 years old male patient.

Previous history

no preleukemia
no previous malignancy
no inborn condition of note

Organomegaly

no hepatomegaly , splenomegaly , enlarged lymph nodes (Slightly enlarged lymph nodes (submental, cervical, axial, mediastinal, inguinal) were detected by computed tomography.), no central nervous system involvement

Blood

WBC : 130.4X 10^9/l
HB : 13.9g/dl
Platelets : 40X 10^9/l
Blasts : 93%
Bone marrow : Hypercellular marrow (NCC 497109/l) with 93.8% blast; monotonous and high nuclear-cytoplasm (N/C) ratio blast cells which had a cleaved nuclear were expanded.%

Cyto-Pathology Classification

Phenotype

Mixed phenotype acute leukaemia, T/myeloid, NOS

Immunophenotype

Positve for CD2,cyCD3,CD7,CD13,CD15,CD34,HLA-DR, and dimly positive for CD33,MPO,TdT. Negative for CD1a,CD5,CD11b,CD117,TCR-AB,TCR-GD

Rearranged Ig Tcr

Not performed.

Pathology

Acute leukemia compatible.

Electron microscopy

Not performed.

Diagnosis

Mixed phenotype acute leukemia, T/myeloid, NOS.

Survival

Date of diagnosis
04-2015

Treatment
Japan adult leukemia study group T-ALL213-O induction therapy including vincristine (VCR), cyclophosphamide (CPA), daunorubicin (DNR), L-Asparaginase (L-ASP) and Predonisolone (PSL).
Treatment related death : no
Relapse : no

Status: Alive

Last follow up: 09-2015

Survival: 5 months

Karyotype

Sample: Bone marrow

Culture time: 24-48

Banding: G-banding

Results
46,XY,t(6;14)(q25;q32) [20]

Karyotype at Relapse
not applicable

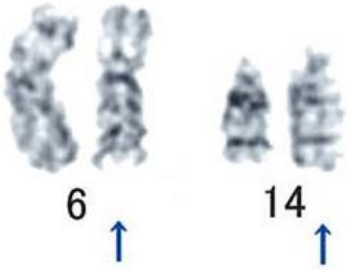
Other molecular cytogenetics technics
fluorescence in situ hybridization(FISH) analysis using IgH 3' flanking region/V probes. 14q32 (IgH) break apart probe is a mixture of two probes, 3'IgH flanking probe and IgH variable region probe as shown in Figure 2a.

Other molecular cytogenetics results
Negative for immunoglobulin heavy chain (IgH) translocation (Figure 2b).

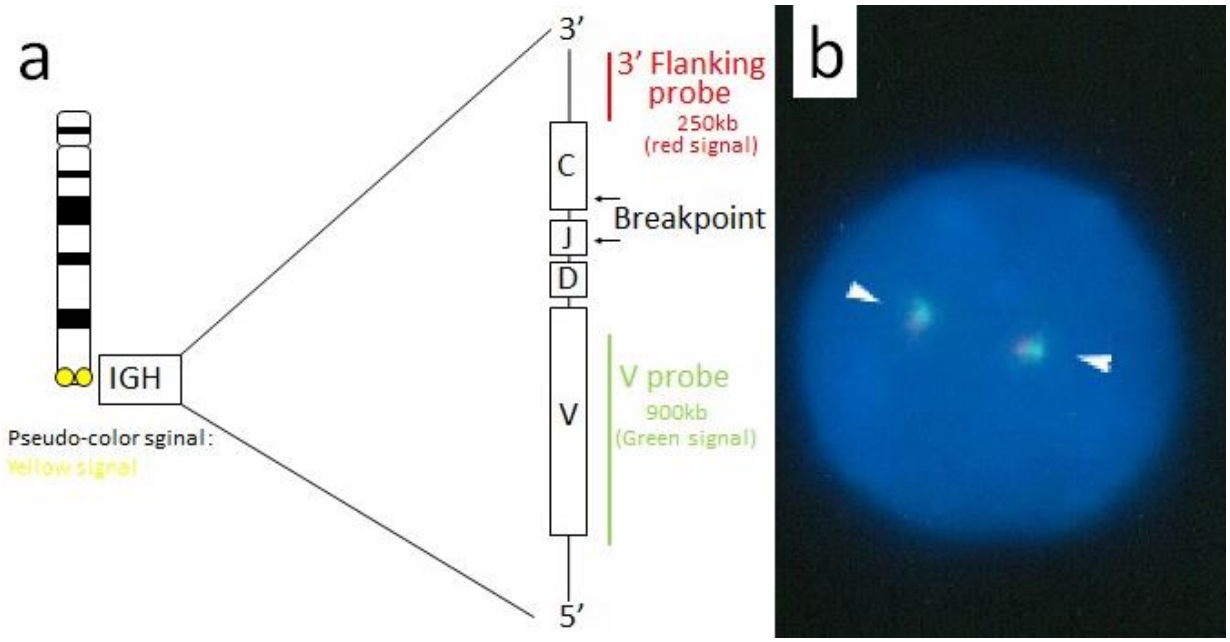
Other Molecular Studies

Technics:
Polymerase chain reaction (PCR) and Sanger sequencing

Results:
Positive for Flt3-internal tandem duplication(ITD). Negative for c-kit mutation.



Partial karyotype from bone marrow cells at the time of diagnosis showing the chromosomal translocation t(6;14)(q25;q32).



FISH analysis was performed by LSI Medience (Tokyo, Japan). (a) A scheme of 3'flanking probe and V probe which is modified from the technical information of LSI Medience corporation (Tokyo, Japan) web site.(b)Negative result for IgH gene rearrangement in this case.

Comments

We present an adult case of biphenotypic acute leukemia with t(6;14)(q25;q32). Chromosome translocations involving 14q32 are generally represented by B cell neoplasms, because the immunoglobulin heavy chain (IgH) gene is located in this region. However, BCL11B gene also located in 14q32 was shown to be involved in this translocation (Bezrookove et al.,2004). BCL11B, a member of the Kruppel family of zinc finger trascription factors, plays a critical role in T cell development and functions as a tumor suppressor (Wakabayashi et al.,2003). The partner gene of this translocation is unknown. The 28S ribosomal DNA (RN28S1) was reported as a candidate fusion partner (Kobayashi et al.,2014), but this gene is not located in 6q25. The phenotype of haematological malignancies with t(6;14)(q25;q32) is variable. These include acute lymphoblastic leukemia (ALL) (Heerema et al.,2002), mixed phenotype acute leukemia (Hayashi et al.,1990, Batanian et al.,1996, Georgy et al.,2008, Kobayashi et al.,2014), acute myeloid leukaemia (AML) (Raimondi et al.,1989, Bezrookove et al.,2004), chronic T cell neoplasm (Inwards et al.,1990) and chronic lymphocytic leukaemia (CLL) (Mayr et al.,2006). In 7 of 9 acute leukaemia cases with this translocation, both myeloid and T-cell lineage markers were detected. No immunophenotype was described in the remaining two cases. This translocation may affect expression of T-cell lineage marker, but the role of BCL11B is unclear.

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