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Case Report Section

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Translocation t(5;6)(q33-34;q23) in an acute myelomonocytic leukemia patient

Adriana Zamecnikova, Soad Al Bahar, Ramesh Pandita

Kuwait Cancer Control Center, Department of Hematology, Laboratory of Cancer Genetics, Kuwait (AZ, SA, RP)

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Abstract

Case report and literature review on translocation t(5;6)(q33-34;q23) in an acute myelomonocytic leukemia patient.

Clinics

Age and sex
68 years old female patient.

Previous history
No preleukemia, no previous malignancy, no inborn condition of note, no main items.

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

Blood

WBC: 104 X 10^9/l
HB: 7.8g/dl
Platelets: 57 X 10^9/l
Blasts: 84%
Bone marrow: Hypercellular marrow with 87% blasts which were PAS diffuse granular positive and SBB (Sudan Black B) positive.

Cyto-Pathology Classification

Cytology
Acute myelomonocytic leukemia

Immunophenotype
Positive for CD13, CD15, CD117, CD33, MPO, CD45, HLDR and dim CD34 (27%)

Diagnosis
Acute myelomonocytic leukemia

Survival

Date of diagnosis: 03-2013

Treatment: Chemotherapy (Daunorubicin & Cytarabine combination therapy; consolidation with high dose Ara-C)
Complete remission: no
Treatment related death: no
Relapse: yes

Phenotype at relapse: Acute myelomonocytic leukemia

Status: Lost

Last follow up: 11-2013

Survival: 8 months

Karyotype

Sample: Bone marrow, blood

Culture time: 24h

Banding: G-banding

Results
46,XX,t(5;6)(q33-34;q23)[25]

Karyotype at Relapse
46,XX,t(5;6)(q33-34;q23)[1]/46,XX,t(5;6)(q33-34;q23),t(7;10)(p22;q23)[19]

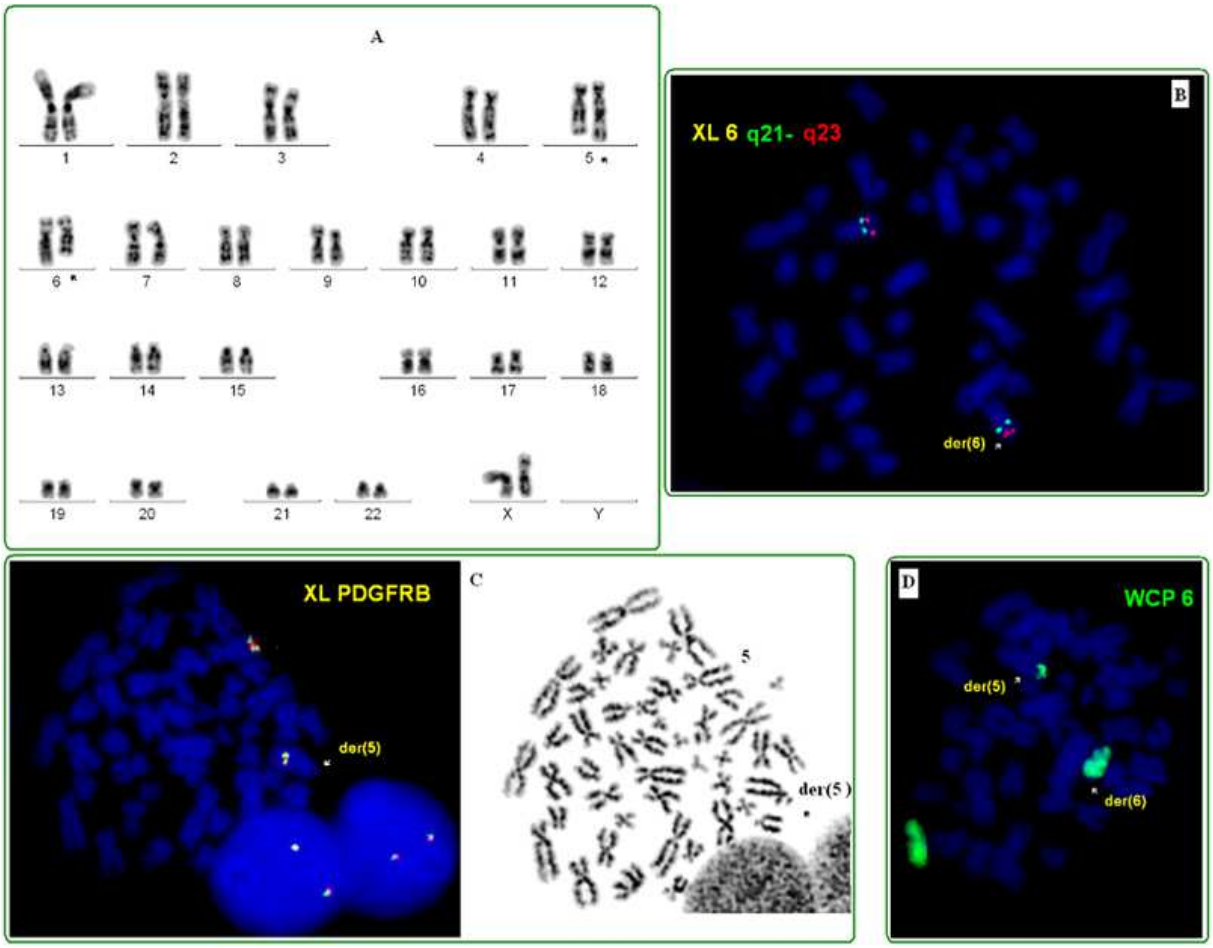

Figure 1. A. Karyotype from the time of diagnosis showing the chromosomal translocation t(5;6)(q33-34;q23). **B.** Fluorescence in situ hybridization studies (FISH) with XL 6q21/6q23 (Metasystem, Germany) probe showing red and green signals on both, normal and der(6) chromosomes. **C.** Applying the XL PDGFR probe (Metasystem, Germany) showed normal signal pattern on both normal and der(5) chromosomes, indicating that PDGFR located on 5q32-33 is not involved in the translocation. **D.** Hybridization with whole chromosome 6 probe (Metasystem, Germany) showing translocation of chromosome 6 sequences to der(5) chromosome.

Other molecular cytogenetics technics

Fluorescence in situ hybridization (FISH) with LSI AML1-ETO, LSI MLL, LSI CBFB/inv(16), LSI EGRI/5q31 (Abbott Molecular, Downers Grove, IL) and XL 6q21/6q23, XL PDGFR, whole chromosome 6 probe Metasystem, Germany).

Other molecular cytogenetics results

Normal signal patterns for LSI AML1-ETO, LSI MLL, LSI CBFB/inv(16), LSI EGRI/5q31, XL 6q21/6q23 and XL PDGFR probes.

Comments

Chromosomal translocations involving 5q33 and 6q23 have been reported in only one patient with T-ALL and an associated myeloproliferative neoplasm and C6ORF204/PDGFRB fusion (Chmielecki et al 2012).

While in this case, the chromosomal translocation appeared to be morphologically identical to our t(5;6)(q33-34;q23), in our patient PDGFRB (5q32-33) is not rearranged and MYB (6q23)is not translocated to chromosome 5 as in a previously described case.

Due to the availability of tyrosine kinase inhibitors for PDGFRB rearranged disorders, our findings emphasize the importance of FISH in precise characterizing of chromosome rearrangements with 5q33-34 breakpoints, especially in suboptimal preparations.

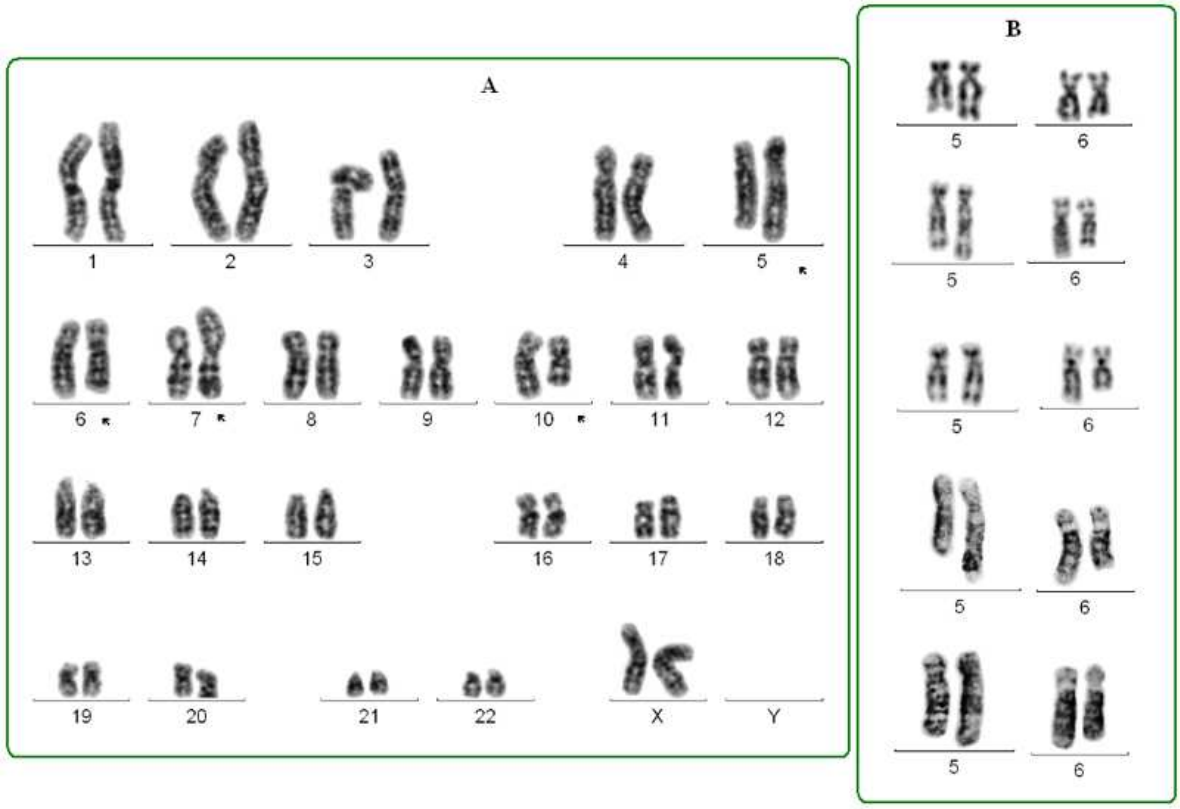

Figure 2. A. Karyotype from blood cell from the time of relapse showing the t(5;6)(q33-34;q23) and a new anomaly t(7;10)(p22;q23). **B.** Partial karyotypes from blood and bone marrow showing the t(5;6)(q33-34;q23).

References

Chmielecki J, Peifer M, Viale A, Hutchinson K, Giltnane J, Socci ND, Hollis CJ, Dean RS, Yenamandra A, Jagasia M, Kim AS, Davé UP, Thomas RK, Pao W. Systematic screen for tyrosine kinase rearrangements identifies a novel C6orf204-PDGFRB fusion in a patient with recurrent T-ALL and an associated myeloproliferative neoplasm. Genes Chromosomes Cancer. 2012 Jan;51(1):54-65

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