**Case Report Section**

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**A new case of t(6;8)(q27;p12) with "8p11 myeloproliferative syndrome"**

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**Clinics**

**Age and sex**
19 years old female patient.

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

**Organomegaly**
hepatomegaly, splenomegaly, enlarged lymph nodes (generalized lymphadenopathy), no central nervous system involvement.

**Blood**

WBC : 375.0 X 10^9/l
HB : 9.0 g/dl
Platelets : 244.000 X 10^9/l
Blasts : 0% no blasts

Bone marrow : 8% Hypercellular bone marrow, myeloid hyperplasia with eosinophilia and at least 8% blasts. Erythropoiesis was decreased; megakaryocytes were distributed irregularly with different dysplastic forms.

**Cyto-Pathology Classification**

**Cytology**
T-cell lymphoma / CML

**Immunophenotype**
Not performed.

**Rearranged Ig Tcr**
Not performed.

**Pathology**

Lymph node biopsy showed T-cell lymphoma, CD2, CD3, CD5, Ki-67 positive; CD34 negative and Ki-67 positive in 50% of neoplastic cells. T-cell blasts associated with perivascular eosinophil component.

**Electron microscopy**
Not performed.

**Diagnosis**
Myeloid and lymphoid neoplasms with FGFR1 abnormalities.

**Survival**

**Date of diagnosis:** 07-2009

**Treatment:** Therapy by Hydrea (1000 mg/day) helped to decrease leukocytosis and less lymph node enlargement. The matched unrelated donor allogeneic human stem cell transplantation (MUD allo-HSCT) was performed in March 2011 after 3 courses of chemotherapy according to 7+3 protocol and myeloablative conditioning regimen with busulfan and cyclophosphamide.

**Complete remission:** no. Stable complete hematological and cytogenetic remissions were achieved with hepatomegaly persisting.

**Treatment related death:** There were many complications in post-transplant period, including serum disease, acute and chronic graft versus host diseases, steroid diabetes, CMV-infection.

**Relapse:** no

**Status:** Death. Last follow up: 09-2011

**Survival:** 26 months

**Karyotype**

Sample: Bone marrow aspirate
**Culture time:** 24h without stimulating agents  
**Banding:** GTG

**Results**
Analysis of 20 metaphase cells revealed an abnormal female karyotype in all metaphases.  
46,XX,t(6;8)(q27;p12)[20].

**Other molecular cytogenetics techniques**
Fluorescence in situ hybridisation using the FGFR1 Breakapart probe was performed (Cytocell Aquarius, UK).

**Other molecular cytogenetics results**
One split and one fused signals were observed resulting in 1 yellow, 1 red, 1 green, 2 blue conformation (in all bone marrow nuclei).

**Other Molecular Studies**

**Technics:** RT-PCR on BCR-ABL.  
**Results:** Negative.

**Comments**
The case described here is of a 19-year-old female who was diagnosed with EMS. It is relatively rare condition characterized in its typical form by the occurrence of a bcr/abl-negative myeloproliferative disorder and a lymphoma, usually a precursor T lymphoblastic lymphoma. Cytogenetics revealed a translocation (6;8)(q27;p12) involving the fibroblast growth factor receptor 1 tyrosine kinase gene on chromosome 8p11-12. To our knowledge, only eight cases with the t(6;8)(q27;p12) have been reported previously. Four of these individuals had features at presentation and a clinical course typical of EMS: malignant T-cell lymphoma and CML similar to our case, AML/myeloproliferative disease, CML-like disease with eosinophilia that progressed rapidly to AML and primary AML evolving to EMS following chemotherapy. Of the three remaining individuals, two presented with PV and progressed to AML after a period of 5 years. One case developed PV 2 years after treatment of an MPD and then subsequently progressed to AML. And one patient had features at presentation of FGFR1OP-FGFR1 disease as B-ALL. The myeloid and lymphoid neoplasms with FGFR1 abnormalities are usually ineradicable by conventional chemotherapy but occasional long-term remission patients have been reported following allogeneic bone marrow transplantation.
A new case of t(6;8)(q27;p12) with "8p11 myeloproliferative syndrome" Gindina T, et al.

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