der(1;18)(q10;q10) in a patient with AML following essential thrombocythemia

Adriana Zamecnikova, Soad Al Bahar, Ramesh Pandita
Kuwait Cancer Control Center, Dep of Hematology, Laboratory of Cancer Genetics, Kuwait (AZ, SA, RP)

Clinical

Age and sex
52 years old female patient.

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

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

Blood
WBC: $3.9 \times 10^9/l$
HB: 7.7g/dl
Platelets: $168 \times 10^9/l$
Blasts: 8%
Bone marrow: 20% (myeloblasts)

Cyto-Pathology Classification

Cytology
AML following essential thrombocythemia

Immunophenotype
of the blast cells performed by flowcytometry was positive for CD7 (46%), CD13 (80%), CD15 (58%), CD33 (49%), CD34 (50%), CD45 (68%), and HLDR (76%).

Rearranged Ig Tcr
Not done.

Pathology
Bone marrow biopsy shows moderate to marked fibrosis with fibroblastic proliferation involving all marrow spaces, there is marked megakaryocytic hyperplasia, few lymphoid cells mixed with erythroid precursors.
The megakaryocytes are variable in morphological appearance, many mononuclear and few with hyperlobulated nuclei and are distributed singly or in tiny clusters.
The normal erythroid and granulocytic cell lines are suppressed.

Electron microscopy
Not done.

Diagnosis
ET transformed to AML.

Survival
Date of diagnosis: 03-2012
Treatment: Allogeneic bone marrow transplant on 11/07/2012
Complete remission: after BMT
Treatment related death: no
Relapse: no
Status: Alive
Last follow up: 11-2012
Survival: 12 months

Karyotype
Sample: Bone marrow, peripheral blood
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(A) Partial karyotype of the patient showing a whole-arm chromosome translocation between chromosomes 1 and 18 associated with a 20q deletion. (B) C-banded metaphase showing the der(1;18)(q10;q10) (arrow). (C) Fluorescence in situ hybridization studies with LSI D20S108 (Abbott) and LSI MALT1 Break Apart (Abbott) probes showing the loss of a D20S108 signal and the presence of a MALT1 signal on the der(1;18)(q10;q10). (D) Hybridization with LSI 1p36/1q25 and LSI 20q12 probes showing one red (20q12) signal on chromosome 20 and an extra green signal for 1q25 on the der(1;18)(q10;q10).

**Culture time:** 24h, direct

**Banding:** G-banding

**Results**

46,XX,+1,der(1;18)(q10;q10),del(20)(q11q13) [20]

bone marrow; 46,XX,+1,der(1;18)(q10;q10),del(20)(q11q13) [20]

blood

**Other molecular cytogenetics techniques**

Fluorescence in situ hybridization applying the LSI D20S108, LSI 1p36/1q25 and LSI MALT1 Break Apart probes (Abbott).

**Other molecular cytogenetics results**

One signal of D20S108 and 3 signals 1q25 in 80% of bone marrow cells.

**Comments**

A 52-years old female patient, with a history of thrombocythemia of 2 years duration was presented with symptomatic anemia and hepatosplenomegaly on March, 2012. Blood film showed evidence of leukoerythroblastic picture with 8% blast cells and bone marrow biopsy confirmed myelofibrosis with megakaryocytic hyperplasia demonstrated the presence of 20% myeloblast cells. Conventional cytogenetic analysis of blood and bone marrow samples revealed a rare chromosome abnormality: der(18)(t(1;18)(q10;q10)) associated with a deletion of the long arm of chromosome 20 in all metaphases.
Unbalanced translocations involving the long arm of chromosome 1 are recurrent chromosome aberrations in patients with various myeloid neoplasms, including myeloproliferative disorders. The centromeric fusion between chromosomes 1 and 18, leading to a normal chromosome 18 substituted with a der(1;18) chromosome observed in our patient has been described in only 5 patients. 4 patients were diagnosed with chronic myeloproliferative disorders (MPD) and 1 patient with complex karyotype with multiple myeloma. Among the 4 patients with MPD, additional chromosome anomaly was detected only in 1 patient (+22), indicating that the der(1;18)(q10;q10) is a primary chromosome anomaly in myeloproliferative disorders. However; as deletion of the long arm of chromosome 20 is a known primary anomaly in myeloid disorders, we cannot exclude the possibility that the der(1;18)(q10;q10) is a secondary anomaly in our case; possibly involved in disease transformation. The unbalanced nature of the rearrangement indicates that gain of 1q and/or loss of 18p might be pathogenetically relevant for neoplastic transformation in this group of patients.

References


Wan TS, Ma SK, Au WY, Chan LC. Derivative (1;18)(q10;q10): a recurrent and novel unbalanced translocation involving 1q in myeloid disorders. Cancer Genet Cytogenet. 2001 Jul 1;128(1):35-8


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