

Case Report Section

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der(6)t(1;6)(q21;p21) in myelofibrosis following polycythemia vera

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Clinics

Age and sex

78 years old female patient.

Previous history

No preleukemia, no inborn condition of note.

Organomegaly

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

Blood

WBC : 45 X 10⁹/l**HB** : 6.8 g/dl**Platelets** : 116 X 10⁹/l**Blasts** : 2% (neutrophils 82%, lymphocytes 9%, monocytes 7%, eosinophils 2%). RBC 5.69x10⁹/L, MCHC 310x10⁹/L, RDW 25.9% Smear left shift, platelet clumps, giant platelets, occasional blast cells and few tear drop cells seen.**Bone marrow** : Normocellular, diffusely fibrotic; small to large atypical megakaryocytes with hyperlobulated nuclei seen; normal erythropoiesis, and granulopoiesis.

Cyto-Pathology Classification

Cytology: Myelofibrosis**Immunophenotype**: CD11 95%, CD11c 80%, CD13 100%, CD33 98%, CD15 92%, MPO 82%.**Rearranged Ig Tcr**: NA**Pathology**: NA**Electron microscopy**: NA**Diagnosis**: Post polycythemic myelofibrosis.

Survival

Date of diagnosis: 1998**Treatment**: Plebothomy, hydroxyurea

Complete remission : NA

Treatment related death : no

Relapse : no

Status: Alive**Last follow up**: 09-2009**Survival**: 12 years

Karyotype

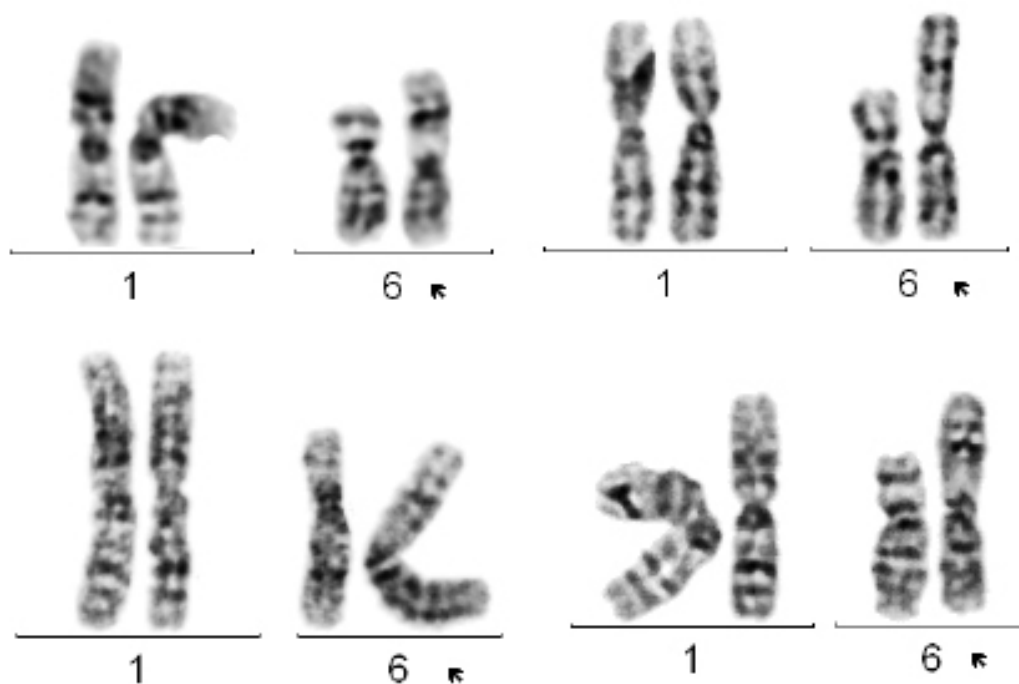
Sample: PB, BM**Culture time**: 24h**Banding**: G-banding**Results**:

PB 46,XX [5]/46,XX,der(6)t(1;6)(q21;p21) [25] ; BM 46,XX,der(6)t(1;6)(q21;p21) [20]

Karyotype at Relapse: -**Other molecular cytogenetics technics**: -**Other molecular cytogenetics results**: -

Other Molecular Studies

Technics: -**Results**: -



G- banded partial karyotypes of the patient from PB cells, showing the translocation der(6)t(1;6)(q21;p21).

Comments

We report a chromosomal translocation der(6)t(1;6)(q21;p21) associated with myelofibrosis following polycythemia vera in a 79 years old female. The patient was diagnosed as having polycythemia vera in 1998 and treated by regular phlebotomy and subsequently with low doses of hydroxyurea. The course of PV was uneventful until 2002 when her myeloproliferative disorder had transformed to myelofibrosis. Due to the unavailability of methods, karyotype analysis and JAK2 V617F analysis was not performed at the time the PV and MF were diagnosed. In 2009 she presented with anemia, fatigue, fever and hepatosplenomegaly. Chromosome analysis performed at that time revealed the karyotypic anomaly 46,XX,der(6)t(1;6)(q21;p21) in bone marrow and blood cultures.

Described as a balanced translocation t(1;6)(q21;p21) in 1 AML case in a 35 years old female with normal karyotype at diagnosis and with cytogenetic polyclonality at relapse (Davidsson et al., 2006) and in 1 case of nodal marginal zone B-cell lymphoma (Ott et al., 2000). More frequently observed as an unbalanced translocation der(6)t(1;6)(q21;p21) resulting in partial trisomy for 1q21 to 1qter, and in loss of 6p21 to 6pter. From the 9 described cases (Najfeld et al., 2007; Shimazaki et al., 1999; Dingli et al., 2005) with der(6)t(1;6)(q21;p21), 8 patients (4 males, 4 females) had myelofibrosis with myeloid metaplasia (Najfeld et al., 2007; Dingli et al., 2005). In addition, several studies have reported heterogeneous 1q and 6p breakpoints resulting in common trisomic region of 1q21-32 and haplo-insufficiency of gene(s) from

different chromosome 6p breakpoints suggesting that gain of gene(s) at 1q21-32 and/or loss of gene(s) from 6p21-6pter may be relevant to the pathogenesis. Our case, together with previously published data demonstrates the association of this rare chromosomal anomaly with primary myeloproliferative disease transformed to myelofibrosis (Dingli et al., 2005, Mertens et al., 1991, Reilly et al., 1997; Andrieux et al., 2003).

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