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Case Report Section

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Translocation t(X;20)(q13;q13.3) as a secondary chromosome abnormality in a patient with 5q-: a case report

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Clinics

Age and sex
76 years old female patient.

Previous history
Preleukemia. Low risk myelodysplastic syndrome (refractory anemia) diagnosed 32 months before (December, 2009).No previous malignancy, no inborn condition of note.

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

Blood

WBC: 2.7X 10^9/l
HB: 8.5g/dl
Platelets: 146 X 10^9/l
Blasts: 0%
Bone marrow: 5% (Global normocellularity, with erythroid and megakaryocytic hyperplasia and megakaryocytic dysplasia (no-lobated megakaryocytes suggestive of 5q- Syndrome), less than 5% of myeloblasts and 23% of ring sideroblasts.)

Cyto-Pathology Classification

Cytology
Myelodysplastic Syndrome - Refractory anemia with ring sideroblasts (FAB classification).

Diagnosis
Myelodysplastic Syndrome - Refractory citopenia with multilineage dysplasia and ringed sideroblasts (WHO classification).

Survival

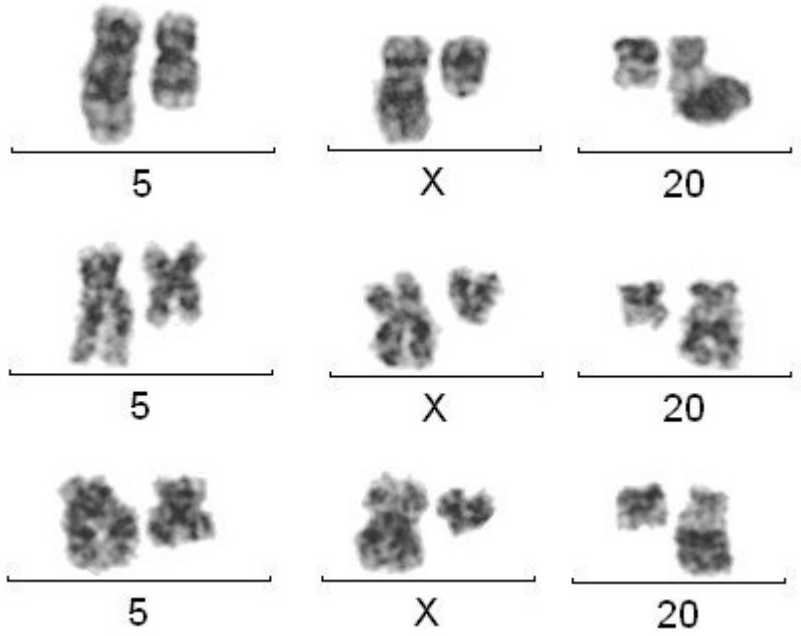
Date of diagnosis: 08-2012

Treatment: Erythropoietin
Complete remission: no
Treatment related death: no
Relapse: no

Status: Alive

Last follow up: 09-2012

Survival: 1 month from the cytogenetic abnormality detection, 30 months from the MDS diagnosis.months



Partial G-banded karyotypes showing the del(5)(q13q31) and the t(X;20)(q13;q13.3)

Karyotype

Sample: Bone marrow

Culture time: 24 and 48 hours without stimulating agents

Banding: GTG

Results
46,XX,del(5)(q13q31)[1]/46,X,t(X;20)(q13;q13.3),del(5(q13q31))[11]/46,XX[8]

Karyotype at Relapse
not done

Comments

The band Xq13 is frequently involved in rearrangements seen in hematological malignancy. The structural rearrangement idic(X)(q13) is associate with the myelodysplastic syndrome with ringed sideroblasts and this region is difficult for mapping because is rich in complex repeats with subregional inversions and high concentration of LINE repeats (boosters of X-inactivation). The chromosome 20q interstitial deletions are well established nonrandom abnormalities in myeloid disorders, particularly in polycythemia vera and myelodysplasia. Very few cases of translocations involving chromosome 20 have been reported in hematological malignancy. The band 20q13 is rich in cancer genes and translocation involving this region has been reported in cases of acute myeloid leukemia. One study shows that breakpoints of X chromosomes associated with myelodysplasia were located in a region of 450 kb next to the gene XIST (Xq13). Another study demonstrated that a critical event in myelodysplasia is loss of tumor suppressor genes present on the long arm of chromosome 20. This suppression can occur when a potential cryptic deletion is associated with a translocation. This phenomenon generates a second mechanism causing inactivation of the X chromosome and derivative 20 resulting loss of function of tumor suppressor genes.

Seven cases of t(X;20)(q13;q13) were described in literature; all affecting women over the age of 57 with myeloid disorders.

We report the detection, by conventional cytogenetic methods, the t(X;20)(q13;q13.3) in one female patient with myelodysplastic syndrome subtype Refractory citopenia with multilineage dysplasia and ringed sideroblasts (classification WHO).

Although this translocation was described as a primary clonal chromosome abnormality, in this case the t(X;20) seems to be a secondary chromosome abnormality following a deletion 5q.

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