A Case of Myelodysplastic Syndrome with Bone Marrow Fibrosis and Isochromosomy of 17(q10)

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Abstract

Isolated isochromosome (17q) is reported in patients with myelodysplastic syndrome (MDS)/myeloproliferative disorders. It is a poor prognostic marker. We report clinicopathologic feature of a 26-year-old male a civil engineer with MDS and secondary fibrosis and isochromosomy of 17(q10). He had a history of working in gold mine and an accessory testis in a physical examination.

Keywords: *Accessory testis, fibrosis, gold mine, isochromosome (17q), myelodysplastic syndrome*

Introduction

Isolated isochromosome (17q) is a rare cytogenetic abnormality in Philadelphia-negative myeloid malignancies.

Kanagal-Shamanna *et al*. studied on clinicopathologic, immunophenotypic, and molecular genetic features of 22 myeloid neoplasms with isolated isochromosome 17q. All showed myelodysplastic and myeloproliferative features, including pseudo-Pelger-Huet-like neutrophils, micromegakaryocytic hyperplasia, hypercellularity, fibrosis, and osteosclerosis. They concluded that neoplasms with isolated isochromosome 17q represent a distinct clinicopathologic entity with myelodysplastic and myeloproliferative features, high risk of leukemic transformation, and wild-type TP53.[1]

Abnormalities of chromosome 17p are detected in about 4% of adult patients with *de novo* acute myeloid leukemia (AML) and with higher rates in therapy-related AML.[2] Abnormality of chromosome 17p is a high-risk marker with a negative impact on the complete remission rate, overall survival rate and relapse rate.[3,4]

Here, we report clinicopathologic feature of a case of myelodysplastic syndrome (MDS) and bone marrow fibrosis and isochromosomy of 17(q10) that had a history of working in gold mine.

Case Report

A 26-year-old male a civil engineer with history of working in gold mine was admitted with malaise and pancytopenia. Physical examination revealed pallor and a testicular mass. Abdominal imaging was normal initially. β human chorionic gonadotropin (β-hCG) and lactate dehydrogenase (LDH) were in normal ranges. Testicular Doppler ultrasound revealed an accessory testis. Examination of peripheral blood smear showed pancytopenia, anisopoikilocytosis, hypochromia, many neutrophils with pseudo-Pelger-Huet anomaly, few myeloblasts, and large platelets. Bone marrow aspiration was diluted. Bone marrow biopsy was relatively hypocellular but showed increased in CD34-positive cells (60% of all nucleated marrow cells) as well as some C-kit-positive cells which were indicative of the diagnosis of MDS with excess blasts. Cytogenetic study was done with the standard method: cultures of the patient's bone marrow were established and harvested according to standard laboratory protocols. Chromosome preparations were treated with pancreatin and stained with Giemsa. A total of 50 metaphase cells were analyzed at the 550-band resolution level. The karyotypes were described according to the guidelines of the International System for Human Cytogenetic Nomenclature 2016: 46,XY,i(17)(10)[49]/46,XY[1] [Figure 1].

Chromosomal breakage and flow cytometry for CD55 and CD59 were in normal ranges. His disease was complicated with recurrent

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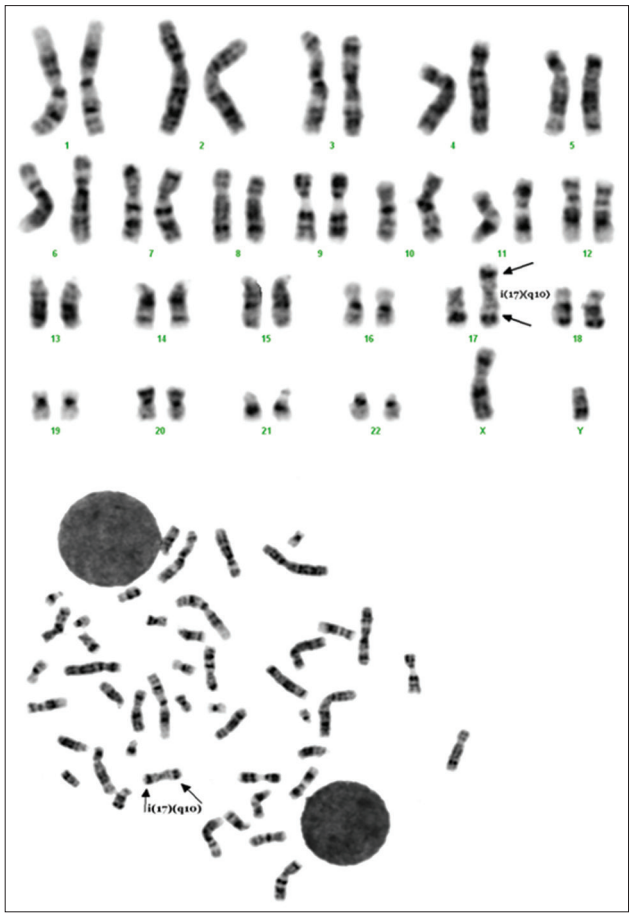

Figure 1: Abnormal male karyotype at diagnosis. Isochromosome of the long arm of chromosome 17 was found in bone marrow cells: 46,XY,i(17)(10)[49]/46,XY[1]

soft-tissue infections that leaded to frequent hospitalization including pyomiositis and abscess formation in the thigh, cellulitis of the forearm, and bacterial infection in submandibular area. Bone marrow aspiration was repeated and was diluted again. Bone marrow re-biopsy was done and showed extensive fibrosis and increased in number of megakaryocytes with many atypical features as well as an increase in the number of blasts in CD34 staining. He had not evidence of splenomegaly in physical examination and abdominal imaging. Polymerase chain reaction study for Jak-2 mutation was negative. According to the findings of peripheral blood smear, bone marrow aspiration and biopsy, cytogenetic and molecular genetic studies, the diagnosis of MDS/myeloproliferative disorders (MPD) was made and he was candidated for allogenic hematopoietic stem cell transplantation. Since he had not any full matched sibling and other related donors and due to high risk features of MDS and active disease which was leaded to frequent infections and blood product transfusions, we decided to start monthly chemotherapy with Vidaza as a bridging agent during searching for matched unrelated donor from world-wide registry human leukocyte antigens bank. His disease was progressed rapidly, and 10 months after presentation, he was admitted with fever, hepatosplenomegaly, and leukocytosis and more than 70% myeloblast in flow cytometry from peripheral blood sample that was consistent with disease transformation to AML non-M3 subtype and he passed away shortly.

Discussion

The diagnosis of MDS/MPD with i17(q10) was made in this young civil engineer with a history of working in gold mine which was presented with malaise and pancytopenia. We found extensive bone marrow fibrosis and rapid progression of MDS to AML in this case of MDS with i17(q10) despite chemotherapy with Vidaza. Myeloid neoplasms with isochromosome 17q have clinicopathologic features of myelodysplastic and myeloproliferative disorders and high risk of leukemic transformation.[1] Coexistence of pneumoconiosis with MDS was reported in a coal miner.[5] Poynter, *et al*. assessed chemical exposures and risk of AML and MDSs and confirmed the well-established risk of MDS and AML associated with benzene exposure and provide risk estimates in a population-based study.[6] Many studies support an association with benzene and other industrial chemicals.[7-12]

We found a new association between MDS and working in gold mine in this young man. In addition, we found a new association between accessory testis and MDS with i17q in this patient.

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Conflicts of interest

There are no conflicts of interest.

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