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Case Report

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A Rare t(5;11)(q35;q13) Translocation in an Elderly Patient With Acute Myeloid Leukemia With Maturation: A Case Report

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Abstract

The t(5;11)(q35;q13) reciprocal translocation is a rare chromosomal abnormality that can arise in myeloid neoplasms, mainly in children and younger adults. Here, we report a case of acute myeloid leukemia with maturation, involving an 85-year-old, in which the tumor cells harbored the t(5;11)(q35;q13) chromosomal abnormality. We also address the diagnostic and immunophenotypic characteristics of acute myeloid leukemia involving t(5;11)(q35;q13), along with a review of the literature.

Keywords: t(5; 11)(q35; q13); Acute myeloid leukemia with maturation; Elderly

Introduction

Chromosomal examinations are important for diagnosing hematological neoplasms because of the prognostic significance of their findings. Myeloid malignancies containing the t(5;11)(q35;q13) translocation are very rare, and only four cases involving children or young adults have been reported [1-4]. Here, we present the case of an elderly patient with acute myeloid leukemia (AML) with maturation involving the t(5;11)(q35;q13) translocation.

Case Report

An 85-year-old female was referred to us because of leukocytosis involving the appearance of blasts. She had no symptoms. On admission, her white blood cell count was 10,900/μL (blast cells, 13%; myelocytes, 0.5%; neutrophils, 66.5%; eosinophils, 3.5%; lymphocytes, 13.5%; monocytes, 3%). Her hemoglobin level was 12.2 g/dL, and her platelet count was 378,000/μL. The patient's serum level of lactate dehydrogenase was 272 U/L. A bone marrow examination revealed a hypercellular bone marrow with a blast cell frequency of 54.8% (Fig. 1). Chromosomal analysis of the patient's bone marrow cells showed the following karyotype: 46,XX,t(5;11)(q35;q13)[8]/46,XX[12] (Fig. 2). Surface marker analysis revealed that the blast cells were positive for CD7 (29.3%), CD56 (63.5%), CD13 (26.5%), CD33 (25.2%), CD34 (81.1%), and human leukocyte antigen (HLA)-DR (97.1%) and negative for CD2, CD3, CD4, CD5, CD8, CD10, CD19, CD20, CD16, and CD14. These findings were consistent with AML with maturation. Due to her poor performance status, she was administered supportive care alone and died of intracranial hemorrhaging 6 months after the initial diagnosis.

Discussion

To the best of our knowledge, only four cases of AML involving the t(5;11)(q35;q13) translocation have been reported in the literature [1-4]. All of these cases involved children or young adults (Table 1 [1-5]). Among the reported cases, the present case involves the older patient. It has been shown that translocations involving the breakpoint 5q35, such as t(5;11)(q35;p15) or t(5;17)(q35;q21), mainly occur in AML cases involving children/adolescents/young adults [6]. In addition, it has been reported that the 11q13 translocation is more frequently found in children and adolescents with acute leukemia [7]. Hence, our case seems to be very rare as it involved an 85-year-old patient who harbored the t(5;11)(q35;q13) translocation, which consists of both 5q35 and 11q13.

Concerning the genes involved in this translocation, Wang et al reported that the nuclear receptor-binding SET domain protein (NSD1), which is located at 5q35, is not involved in this translocation, based on experiments involving the fluorescence *in situ* hybridization (FISH) technique [3]. Similarly, using FISH, de Oliveira et al showed that cyclin D1 (CCND1), which is located at 11q13, is also not involved in this translocation [4].

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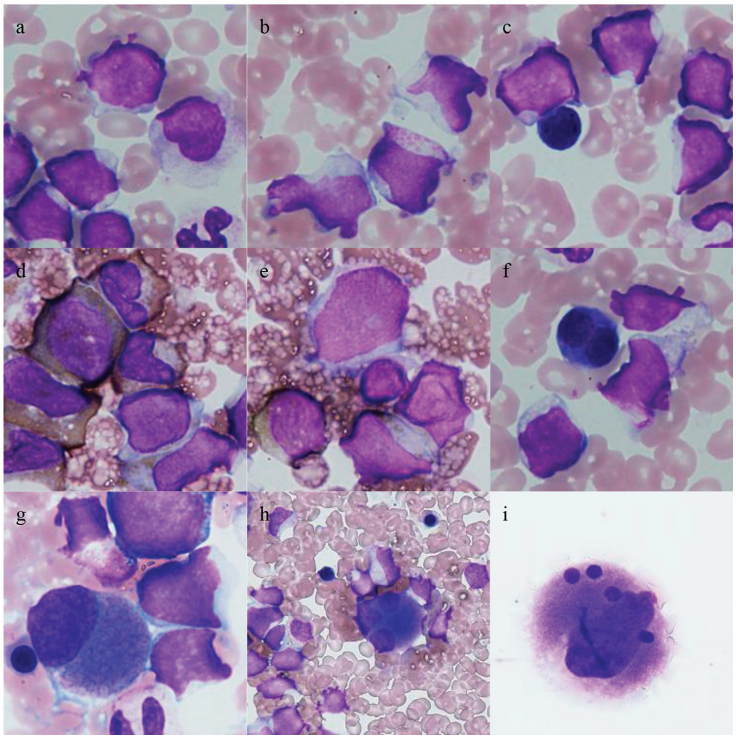

Figure 1. Bone marrow cells seen at the diagnosis of leukemia. Low frequencies of dyserythropoiesis and dysmegakaryopoiesis were observed. (a-c) Leukemic cells (May-Giemsa staining); abundant granules were occasionally observed (b); (d, e) blasts that were positively and negatively stained for myeloperoxidase, respectively; (f) a binuclear erythroblast (May-Giemsa staining); (g-i) megakaryocytes (May-Giemsa staining); (g) a small mononuclear megakaryocyte; (h) a trinuclear megakaryocyte; (i) a megakaryocyte containing multiple separate, round nuclei.

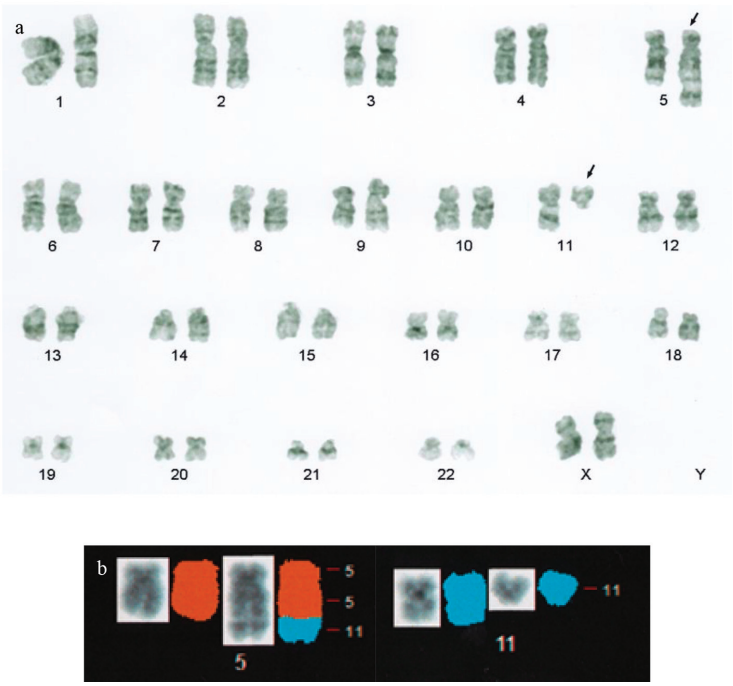

Figure 2. (a) G-band karyotype analysis performed at the diagnosis of leukemia revealed the following karyotype: 46,XX,t(5;11)(q35;q13). The arrows indicate derivative chromosomes. (b) Partial spectral karyotyping of the patient's metaphase spread after spectrum-based classification (left, counterstained with 4',6-diamino-2-phenylindole dihydrochloride; right, spectral karyotyping).

Table 1. Cases of Acute Myeloid Leukemia Involving t(5;11)(q35;q13)

Author	Age/sex	Dx	Karyotype[a]	Immunophenotype	Subsequent therapy and clinical course
Leverger et al [1]	14/M	M5	46,XY,t(5;11)(q35;q13)/46,idem,del(9)(q12q32)	NR	NR
Itoh et al [2]	28/M	M4	46,XY,t(5;11)(q35;q13)	NR	No remission was obtained in spite of various therapy; 18 months of survival
Wang et al [3]	19/M	M2	46,XY,t(5;11)(q35;q13)	$CD13^+$, $CD34^+$, $CD33^+$, $CD19^+$, $CD15^+$, HLA-DR^+	IDR/CA, MIT/ETP/CA (induction failure); returned to the nearest hospital with oral HU
De Oliveira et al [4]	30/M	M2	46,XY,t(5;11)(q35;q13)	$CD13^+$, $CD34^+$, $CD33^+$, $CD19^-$, $CD117^-$, HLA-DR^+	DNR/CA twice (partial remission); received allogeneic SCT from HLA-identical sister
Present case	85/F	M2	46,XX,t(5;11)(q35;q13)[8]/46,XX[12]	$CD13^+$, $CD34^+$, $CD33^+$, $CD7^+$, $CD56^+$, HLA-DR^+	Palliative care alone; died of intracranial hemorrhage (6 months)

NR: not reported; IDR: idarubicin; CA: cytosine arabinoside; ETP: etoposide; DNR: daunorubicin; MIT: mitoxantrone; HU: hydroxyurea; SCT: stem cell transplantation; HLA: human leukocyte antigen. [a]Expect for that for our case, the karyotype descriptions are shown according to the Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer [5].

Although we could not perform a detailed analysis, e.g., ribonucleic acid sequencing, of the genetic rearrangements associated with this translocation due to a lack of specimens, it might be possible to determine the genetic characteristics responsible for this translocation through further case accumulation.

As for the diagnoses of the reported cases of AML involving the t(5;11)(q35;q13) translocation, there were three cases of AML with maturation, including our case (French-American-British classification; M2); one case of acute myelomonocytic leukemia (M4); and one case of acute monoblastic and monocytic leukemia (M5) (Table 1). Although de Oliveira et al reported that the 11q13 abnormality seemed to be associated with differentiation towards the monocytic lineage [4], M2 was the most common diagnosis among the reported cases. Hence, it cannot be assumed that the t(5;11)(q35;q13) translocation is restricted to the monocytic lineage.

Regarding the immunophenotypes of such cases, our case exhibited positivity for CD56. The CD56 antigen is expressed in natural killer/T-cell lymphoma, multiple myeloma, and AML. It was reported that CD56 is associated with a poorer prognosis, induction failure, and worse survival in AML [8-11]. In addition, CD56 expression is associated with recurrent chromosomal abnormalities, such as t(8;16)(p11;p13), t(8;21)(q22;q22), t(15;17)(q22;q21), and t(16;21)(p11;q22) [12]. The positivity rate of CD56 was 63.5% in our case. However, previous reports of t(5;11)(q35;q13)-harboring cases did not mention CD56 expression. Hence, it is unclear whether the CD56 expression seen in our patient is characteristic of t(5;11)(q35;q13) or was due to aberrant expression. The accumulation of further cases of t(5;11)(q35;q13)-harboring AML is necessary to obtain accurate data regarding the frequency of CD56 expression.

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