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OBM Genetics

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Case Report

Jumping Translocation of Chromosome 11q in *De Novo* Acute Myeloid Leukemia: A Rare Chromosomal Aberration

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Academic Editors: Ying S. Zou and Akbar Dorgalaleh

Collection: Genetic Testing

OBM Genetics
2026, volume 10, issue 3
doi:10.21926/obm.genet.2603351

Received: April 15, 2026
Accepted: August 03, 2026
Published: August 10, 2026

Abstract

Acute myeloid leukemia (AML) represents a clinically and genetically heterogeneous group of hematologic malignancies. Among the less frequently encountered cytogenetic abnormalities are jumping translocations (JTs), in which a segment from a donor chromosome relocates to multiple recipient chromosomes. Although uncommon, these events have been described in association with clonal evolution and, in some cases, more aggressive disease behavior. Here, we describe a 49-year-old man who presented with fatigue and generalized weakness and was subsequently diagnosed with AML harboring an inv(16)(p13.1q22)/*CBFB::MYH11* rearrangement. In addition to the primary leukemic clone, cytogenetic evaluation identified three sideline clones demonstrating a jumping translocation involving chromosome 11 with the breakpoint at band 11q13. The 11q13 segment was observed on different recipient chromosomes, namely 16p13.1, 19p13.3, and 21q22, indicating a notable degree of clonal heterogeneity. While AML with inv(16) is typically associated with a favorable prognosis, the concurrent presence of a chromosome 11q13 jumping translocation in this case suggests

added biological complexity. Such secondary cytogenetic changes may have implications for disease evolution and risk stratification. This case underscores the importance of integrating conventional cytogenetic analysis with next-generation sequencing to delineate clonal architecture better and support clinical decision-making in AML.

Keywords
Jumping translocations (JTs); Acute myeloid leukemia (AML); Core binding factor (CBF)

1. Introduction

Jumping translocations (JTs) are rare cytogenetic aberrations characterized by unbalanced non-reciprocal translocations of a single donor chromosome segment to two or more different recipient chromosomes in distinct cell lines within the same patient [1]. JTs often involve regions with repetitive DNA (telomeres, centromeres, and nucleolar organizer regions), and their presence is associated with trisomy for the donor chromosome segment and partial deletions of the recipient chromosomes [2, 3].

The breakpoint in the donor chromosome is often characterized by an interstitial telomeric sequence which facilitates fusion with recipient chromosomes. JTs were first described in 1979 in a patient with Prader–Willi syndrome and later, in 1988, in a case of squamous cell carcinoma of the skin in a patient with Xeroderma pigmentosum [4, 5].

JTs are uncommon events occurring in constitutional chromosome abnormalities, solid tumors, and hematological malignancies including acute lymphoblastic leukemia (ALL), myeloproliferative neoplasms (MPN), myelodysplastic neoplasms (MDS), acute myeloid leukemia (AML), multiple myeloma, lymphomas, and chronic myeloid leukemia (CML) [6-10]. Several studies have associated JTs with increased disease progression, poor prognosis, and reduced overall survival [11-13]. Although the exact mechanisms underlying JTs remain unclear, proposed contributing factors include telomere shortening and instability, pericentric heterochromatin decondensation, viral infection, chromosome instability, and homologous recombination between telomere repeat sequences and interstitial telomeric sequences within the donor chromosome [14]. More recently, pathogenic variants/deletion involving the *TP53* gene have also been implicated in the development of JTs [15]. Chromosomes 1 and 3 are the most frequently reported donor chromosomes in JTs. Additionally, there have been several other reports of JTs of chromosomes 6, 11, 15, and 21 as donor chromosomes [15-18]. Therefore, JTs can lead to copy number gains of donor chromosomal material and losses from recipient chromosomes, contributing to genomic imbalance and clonal evolution.

AML is a genetically heterogeneous hematological malignancy arising from the clonal proliferation of hematopoietic stem cells, characterized by chromosomal anomalies, genetic mutations, epigenetic factors affecting chromatin structure, and microRNA deregulation. The 5th edition of the World Health Organization (WHO) and the International Consensus Classification of Myeloid Neoplasms and Acute Leukemias stratify AML into distinct entities based on recurrent genetic abnormalities, each associated with specific clinicopathologic and prognostic features [19-21]. Chromosomal abnormalities detectable by karyotyping are present in approximately 55% of

adults with *de novo* AML [22]. Core binding factor (CBF) AML which is defined by t(8;21)(q22;q22.1) and inv(16)(p13q22) represents one of the most frequent subtypes in both children and adults, constituting around 15% of all AML and is associated with favorable prognosis [23]. AML with inv(16) results in *CBFB::MYH11* fusion and is generally associated with a favorable prognosis. However, the prognostic impact of additional cytogenetic abnormalities in this subgroup is still debatable.

To date, 33 cases of AML and 28 cases of MDS with JTs have been described in the literature [24-26]. The present study describes the genomic landscape of a patient with AML harboring inv(16)(p13.1q22)/*CBFB*::*MYH11* fusion and a jumping translocation involving chromosome 11q as the donor segment. We have performed cytogenetic and molecular studies and delineated the clinicopathological characteristics, clinical course, clonal evolution, disease progression, and treatment response associated with this rare and complex cytogenetic rearrangement.

2. Case Report

A 49-year-old male presented with a 25-day history of joint pain, fever, and lethargy, along with lymphadenopathy noted approximately one month prior to presentation. He was initially treated for a febrile illness; however, due to lack of clinical improvement, further evaluation was undertaken to rule out an underlying hematologic malignancy. The patient did not have similar presentation in his past medical history, nor any co-morbidities, or known drug allergies. Initial laboratory evaluation demonstrated anemia (hemoglobin 9.2 g/dL), leukocytosis (white blood cell count 32.7 × 10^9/L), and thrombocytopenia (platelet count 28 × 10^9/L), with 72% circulating blasts. Bone marrow aspirate showed a hypercellular marrow with marked reduction in normal trilineage hematopoiesis and approximately 60% myeloblasts. Flow cytometric analysis confirmed that 92% of the cells were blasts expressing CD34, CD117, HLA-DR, CD33, CD13, CD38, CD15, CD73, and cytoplasmic myeloperoxidase (cMPO), consistent with a diagnosis of AML. The patient was given the 7 + 3 regimen for cytarabine/daunorubicin and received a transient hematologic response. In the absence of allogeneic HSCT, the disease relapsed morphologically at 7 months, with recurrence of circulating blasts and leukocytosis.

Conventional cytogenetic analysis (karyotyping) was performed on the bone marrow aspirate collected at the time of presentation by setting up a 24 hours unstimulated culture [27].

G-banded chromosome analysis was conducted at a resolution of 450-550 bands, and karyotyping and imaging were performed on 30 metaphases using CytoVision 7.7 image analysis software (Leica Biosystems, Germany). Karyotypes were described according to the International System for Human Cytogenomic Nomenclature (ISCN) 2024 [28] as 46,XY,inv(16)(p13.1q22)[14]/46,XY,der(16)inv(16)(p13.1q22),t(11;16)(q13;p13.1)[7]/46,XY,inv(16)(p13.1q22),der(19)t(11;19)(q13;p13.3)[6]/46,XY,inv(16)(p13.1q22),der(21)t(11;21)(q13;q22)[3]. The karyotype suggested that the Jumping Translocation involve the transfer of the 11q13 to 11qter segment to chromosomes 16p13.1, 19p13.3 and 21q22, with apparently normal copies of chromosome 11 homologues. In the der(16) clone, the insertion of the 11q13 donor segment at 16p13.1 on the inv(16) chromosome is likely to result in the gain of 11q13 to 11qter along with the loss of the chromosomal segment distal to the 16p13.1 breakpoint, potentially involving the sequences from the rearranged chromosomes generated by inv(16)(p13.1q22). Therefore, Jumping Translocation is expected to result in partial trisomy of the donor 11q13 segment. In addition, the

derivative recipient chromosomes are expected to have a loss of the chromosomal segment distal to the respective breakpoint regions namely 16p13.1, 19p13.3 and 21q22.

Fluorescence in situ hybridization (FISH) studies were conducted on bone marrow cytogenetic specimens. Dual-color FISH was performed using dual-color *CBFB* break-apart probe [5' end (centromeric); red and 3' end (telomeric); green], and images were generated using Isis FISH imaging software (MetaSystems GmbH, Altussheim, Germany). FISH analysis showed a 1F1R1G signal pattern consistent with *CBFB* rearrangement associated with the inv(16)(p13.1q22) abnormality (Figure 1 and Figure 2).

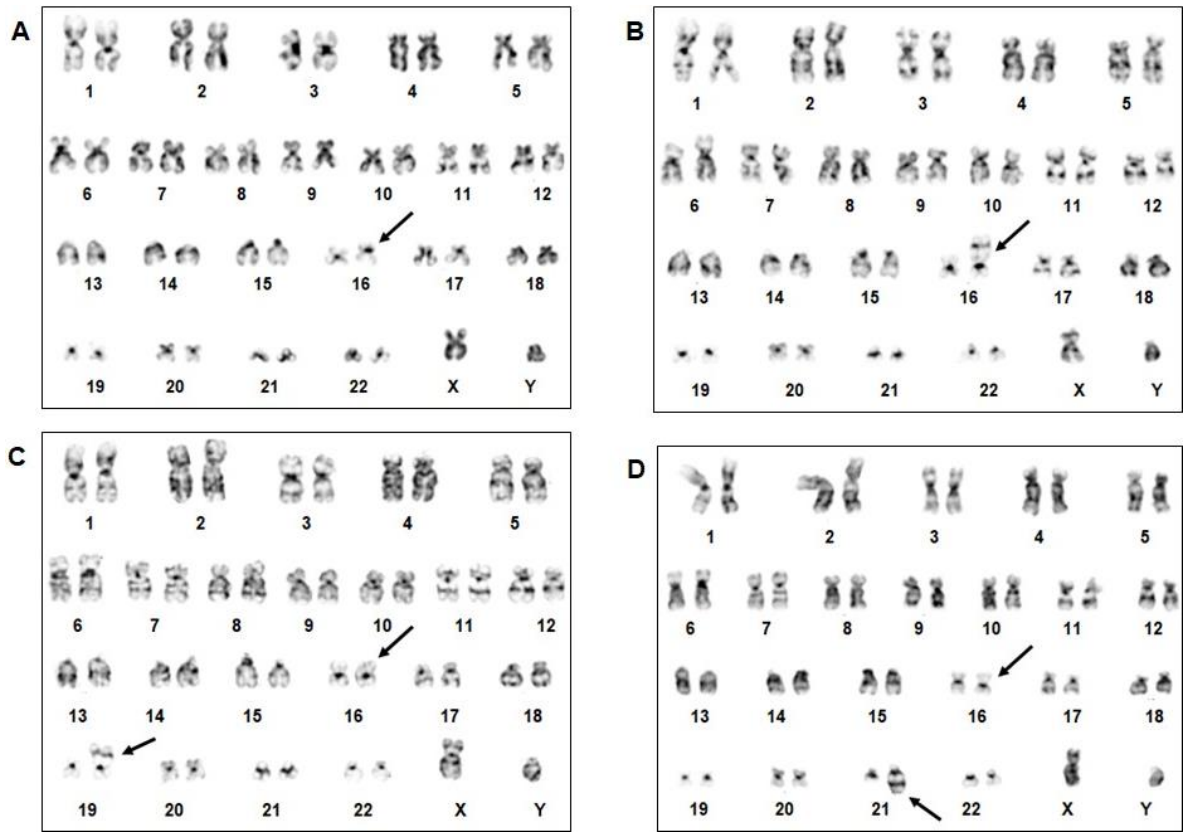

Figure 1 Representative G banded Karyogram showing (a) 46,XY,inv(16)(p13.1q22) as a sole abnormality and co-occurrence of (b) 46,XY,der(16)inv(16)(p13.1q22),t(11;16)(q13p13.1) (c) 46,XY,inv(16)(p13.1q22),der(19)t(11;19)(q13;p13.3) and (d) 46,XY,inv(16)(p13.1q22),der(21)t(11;21)(q13;q22). The arrows indicate the rearranged chromosomes.

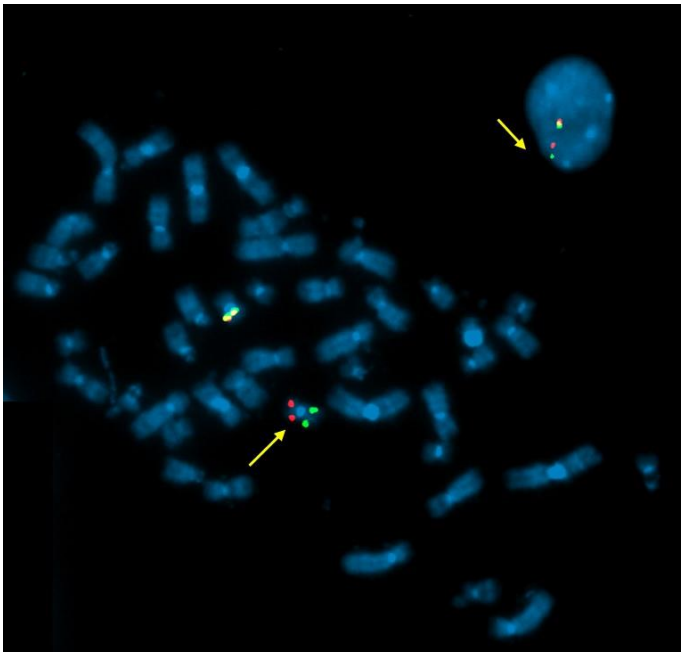

Figure 2 FISH performed on metaphases using *CBFB* (16q22) break-apart probe, one fusion signal along with separate red (5' end, centromeric side of chromosome) and green (3' end, telomeric side of chromosome) signals on interphase and metaphase cells, indicative of inv(16) (arrows).

Molecular profiling was performed using next-generation sequencing (NGS) with the Oncomine™ Myeloid Research Assay (Thermo Fisher Scientific, USA), targeting 129 myeloid-associated genes. Approximately 80 ng of DNA/RNA extracted from the diagnostic bone marrow specimen using the RNeasy Mini Kit (Qiagen, Hilden, Germany) was used as input material. Amplicon libraries were prepared using the Ion AmpliSeq custom panel, followed by multiplex PCR amplification with the Ion AmpliSeq Library Kit 2.0 and custom Ion AmpliSeq primers according to the manufacturer's instructions. Sequencing data were aligned to the human reference genome (hg19) and analyzed using the Oncomine™ Ion Torrent Suite Software (version 4.4). Variant calls were visualized and manually reviewed using Integrated Genomics Viewer (IGV), version 2.3.8 (https://igv.org/). NGS analysis identified a *FLT3* (V592G) missense mutation in addition to the *CBFB*::*MYH11* fusion transcript (data not shown).

2.1 Ethics Approval

All methods performed in this study were in accordance with the ethical standards of the institutional committee. The Ethics Committee of Dr. Lal Path Labs approved the study. The committee's reference number: EC/NEW/INST/2021/1702.

3. Discussion

AML is characterized by the accumulation of undifferentiated blast cells in the peripheral blood and bone marrow [29]. Conventional cytogenetic analysis (karyotyping) remains a key method of choice for AML work-up and prognostication because of the strong correlation between chromosomal aberrations and particular biological variants as well as the identification of additional

abnormalities that targeted testing such as FISH and other molecular techniques may not detect. While certain chromosomal aberrations may characterize a specific leukemia variant, it is rare to find similar chromosomal abnormalities across leukemias from different hematopoietic lineages. Among chromosome 11 alterations, the 11q23 and 11q13 regions are most frequently involved in hematologic malignancies [18]. The *KMT2A* gene, a crucial regulator of hematopoiesis, is located in the 11q23 region. The aberrant chromosomal fragment is transferred to several recipient chromosomes, including 6, 10, 17, and 19, in a Jumping Translocation event using 11q23 as a donor segment, reflecting genomic instability and clonal evolution. The 11q23 JTs are usually acquired secondary abnormalities in AML linked to treatment resistance, complex karyotypes, poor clinical outcomes, and disease progression. Their presence may dysregulate *KMT2A* and other genes located within the transferred 11q23 segment, contributing to leukemogenesis and aggressive disease behavior [30, 31]. Additional genes within the 11q23 and 11q13.3 regions include *CBL* and *CCND1* genes, involved in hematopoietic signalling pathways and cell cycle progression, respectively. Thus, gain of the 11q13-11q23 segment via JTs may increase gene dosage, thereby promoting leukemic proliferation and clonal evolution [32].

The present study provides insight into a rare and complex genomic rearrangement characterized by a jumping translocation involving the 11q13 donor segment occurring in the context of inv(16)(p13.1q22)/*CBFB*::*MYH11*–positive AML (Figure 3). Inversion 16 a core-binding factor abnormality is traditionally associated with a favourable prognosis, high complete remission rates, and prolonged overall survival. In the present study, retention of both copies of chromosome 11 homologs together with the translocation of the 11q13 segment to three recipient chromosomes suggest a gain of 11q13 to 11qter. Additionally, the derivative chromosomes 16, 19 and 21 are expected to harbor terminal losses distal to their expected breakpoints. Therefore, in the proband, JT resulted in a complex karyotype with multiple concurrent cytogenetic alterations, potentially abrogating the favourable prognostic impact of inv(16), thereby driving clonal evolution and disease progression and shifting the patient into a higher-risk category [13, 26]. Furthermore, the identification of an activating *FLT3* missense mutation likely contributed to disease aggressiveness through activation of downstream signalling pathways, including *STAT5*, *MAPK*, and *AKT*, promoting cellular proliferation and resistance to apoptosis and conferring an unfavourable prognosis [33].

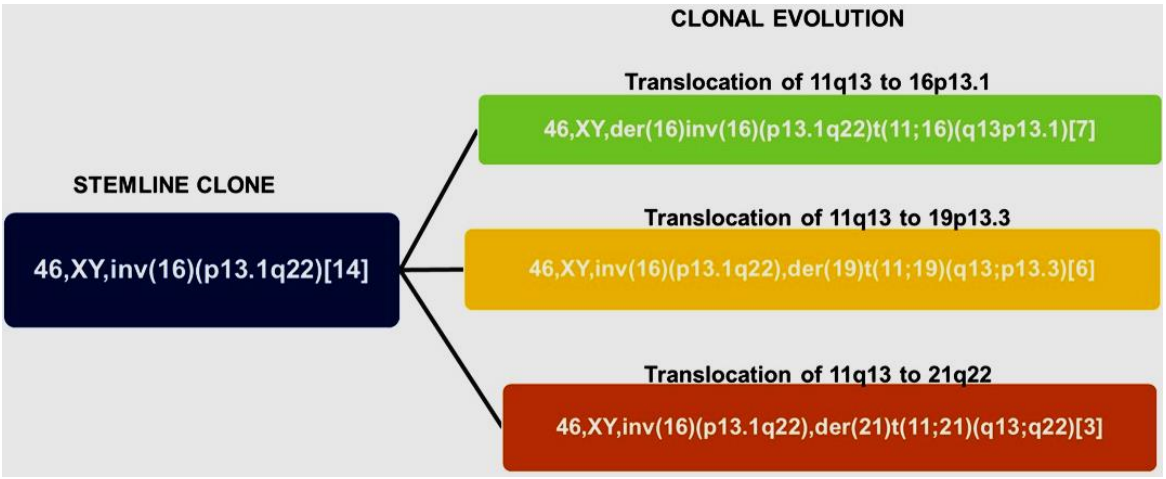

Figure 3 The diagrammatic representation of the major clone and three sideline subclones identified at diagnosis.

Collectively, the cytogenetic and molecular findings in this patient are consistent with the observed poor clinical outcome, as the patient demonstrated a suboptimal response to therapy and ultimately succumbed to the disease. Given the adverse prognostic implications of JTs in hematologic malignancies, further investigation into the underlying mechanisms driving these complex genomic events and their clinical impact is warranted. Improved understanding of these processes may facilitate refined risk stratification and the development of more effective, targeted therapeutic strategies.

4. Conclusions

JTs are rare events in hematologic neoplasms, and chromosome 11–associated JTs are particularly uncommon, as donor abnormalities are fewer than those described in AML to date. In the present study, the coexistence of a jumping translocation with the *FLT3*(V592G) mutation may have contributed to resistance to chemotherapy, aggressive disease behavior, and reduced overall survival. While the *FLT3* mutation is potentially targetable with specific kinase inhibitors, it has been associated with adverse clinical outcomes. Thus, the findings support the role of jumping translocation as a marker of clonal evolution and unfavorable prognosis in AML. Although genomic studies are increasingly used for the diagnosis and classification of AML, comprehensive cytogenetic studies remain indispensable for accurate risk stratification. They are among the most important prognostic tools for predicting therapeutic response and disease progression.

Acknowledgments

Authors are grateful to the patients and their family members for their support for the case study. We acknowledge the efforts of all the healthcare professionals involved in the clinical care and diagnostic evaluation of this case. We would also like to thank the management of Dr. Lal Path Labs for providing excellent diagnostic facilities.

Author Contributions

LR conceptualized, performed literature survey, data collection, interpretation and manuscript preparation. VKT revised the manuscript critically for important intellectual content. All authors read and approved the final manuscript.

Competing Interests

The authors declare that they have no competing interests.

Data Availability Statement

Data sharing is not applicable to this article due to patient confidentiality and institutional data protection policies.

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