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Cryptic Rearrangement of the *KMT2A* Gene in a B-cell Acute Lymphoblastic Leukemia

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Abstract

Background/Aim: A 30-year-old female diagnosed with B cell acute lymphoblastic leukemia (B-ALL) had a normal karyotype at diagnosis.

Case Report: The case was investigated further by fluorescence *in situ* hybridization (FISH), array comparative genomic hybridization (aCGH), and reverse-transcription polymerase chain reaction (RT-PCR) followed by Cycle sequencing. The diagnostic karyotype was normal (46,XX), but FISH studies on tumor cells using a *KMT2A* break-apart probes showed that the proximal part of *KMT2A* was inserted into an apparently normal chromosome 4 with concomitant loss of the distal part of the probe. aCGH identified losses within 11q23.3 and 4q21.3q22.1 with the breakpoints mapping inside the *KMT2A* and *AFF1* loci. The presence of the putative *KMT2A::AFF1* fusion gene was confirmed by FISH analysis and RT-PCR/Cycle sequencing; an in-frame fusion was detected between *KMT2A* (exon 9) and *AFF1* (exon 6). The patient underwent allogenic stem cell transplantation and reached complete remission.

Conclusion: This case highlights the need to supplement banding cytogenetics with appropriate molecular (cyto)genetic techniques whenever the karyotype does not reveal characteristic aberrations. Although *KMT2A* rearrangements in both lymphoblastic and myeloid acute leukemias usually arise through karyotypically visible chromosomal recombinations, this is not always the case.

Keywords: B-lymphoblastic leukemia, cytogenetics, *KMT2A*, fluorescence *in situ* hybridization, array comparative genomic hybridization, fusion gene.

Introduction

B-cell acute lymphoblastic leukemia (B-ALL) is a neoplasia of precursor lymphoid cells committed to the B-cell lineage (1). According to the World Health Classification (5th WHO) (1) and International Consensus Classification (2022 ICC) (2), B-ALL with t(v;11q23.3)/Lysine-specific N-methyl-transferase 2A gene (*KMT2A*) rearrangement is the most

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common genetic leukemia subtype in both infants (<1 year) and adults (>18 years) (2-4). *KMT2A*, also known as mixed-lineage leukemia (*MLL*) and mapping to chromosome band 11q23, is a promiscuous oncogene known to rearrange with more than 100 partners (5). The ALF transcription elongation factor 1 (*AFF1*) gene, in chromosome band 4q21, is the most frequent partner in B-ALL, due to a t(4;11)(q21;q23) translocation (6). The *KMT2A::AFF1* fusion transcript is associated with poor prognosis, as is the case for the set of all *KMT2A*-rearranged lymphoblastic leukemias (3, 7). Here, we report a patient with an apparently normal karyotype but carrying a submicroscopic *KMT2A* rearrangement with *AFF1*.

Case Report

A 30-year-old female was admitted to hospital in August 2021 with leucocytosis (56.6×10^9/l) and a three-day history of pain in her right arm and neck. She had a surgical intervention on the right side of her neck, reportedly due to sialolithiasis, previously that same year. The blood smear was dominated by blasts, and flow cytometric assessment disclosed pre-B ALL with the following immunophenotype: $CD10^-$, $CD11b^-$, $CD13^-$, $CD15^+$, $CD19^+$, $CD20^-$ $cyCD22^+$, $CD24^+$, $CD33^-$, $CD34^+$, $CD38^+$, $CD58^+$, $cyCD79B^+$, $CD133^+$, TdT^+. Treatment was initiated according to the ALLTogether protocol (ClinicalTrials.gov Identifier: NCT04307576). On day 29, the patient was in complete hematological remission, but minimal residual disease [MRD; 1×10^{-2}; (0.7%)] was detected by PCR analysis. MRD remained detectable [3×10^{-4}; (0.03%)] on day 71. Because her disease was classified as high-risk ALL, she was accepted for allogeneic stem cell transplantation in first complete remission (CR1), which took place on day 161 with peripheral blood stem cells from a matched unrelated male donor following conditioning with total body irradiation (TBI; 12 Gy) and etoposide with anti-thymocyte globulin, cyclosporine, and methotrexate as graft-versus-host disease (GvHD) prophylaxis. At transplantation, she was MRD negative. By day +92, she was a complete donor chimera, but she was found to be MRD positive, admittedly below the level allowing quantitation [$<5\times10^{-4}$ (0.05%)]. At three- and six-months post-transplant, she had become MRD negative. As of December 2024, she is in complete remission with no signs of GvHD.

G-banding and fluorescence in situ hybridization (FISH) analyses. Bone marrow was short-term cultured, stained for G-banding analysis, and analyzed cytogenetically as previously described (8). The karyotypic description followed the International System of Cytogenomic Nomenclature (9). FISH investigations (probes from Cytocell, Oxford Gene Technology, OX5 1PF Begbroke, Oxfordshire, Oxford, UK) were used to detect the possible presence of *CDKN2A* (9p21) deletion, fusion genes *ETV6::RUNX1* and *BCR::ABL1*, rearrangements of *ABL1* (9q34), *MYC* (8q24), *KMT2A* (11q23), *TCF3* (19p13), and *IGH* (14q32), *PDGFRB* and *CSF1R* (5q32), and *ABL2* (1q25) as well as additional chromosomes typical of a hyperdiploid karyotype (10). The FISH analyses are those recommended by the WHO, the national health program, and the ALLTogether protocol (ClinicalTrials.gov Identifier: NCT04307576) as standard for the diagnosis of ALL. Additionally, a commercially available *KMT2A::AFF1* double fusion probe was used (Cytocell). The routine FISH analyses were performed on interphase nuclei, after which metaphase FISH for *KMT2A::AFF1* was performed (see below) to determine the chromosomal position of the leukemogenic fusion gene.

DNA and RNA extraction. Fresh frozen bone marrow was used to extract DNA and RNA. DNA was extracted at diagnosis using a Maxwell RSC Instrument together with an RSC Whole Blood DNA Kit (Promega, Madison, WI, USA) according to the manufacturer's recommendations. RNA was extracted using a miRNeasy kit (Qiagen, Hilden, Germany). The concentrations were measured using QIAxel microfluidic UV/VIS spectrophotometer (Qiagen) and Quantus fluorometer (Promega).

Array comparative genomic hybridization (aCGH). aCGH was used to detect genomic imbalances. The CytoSure

Consortium Cancer + SNP arrays (Oxford Gene Technology) were used according to the manufacturer's recommendations. The slides (CytoSure Cancer +SNP array, 4×180k) were scanned in an Agilent SureScan Dx microarray scanner using Agilent Feature Extraction Software (Agilent, Santa Clara, CA, USA; version 12.1.1.1) and data were analyzed using CytoSure Interpret Software (version 4.11.36, Oxford Gene Technology). The copy number aberrations were identified using the Circular Binary Segmentation (CBS) algorithm. A custom-made aberration filter defining imbalances was added. Copy number aberrations (CNA) were defined as a region with a minimum of five gained/lost probes (11). All imbalances were scored, also those smaller than 5 Mb. Annotations were based on the human reference sequence GRCh37/hg19.

Reverse transcription polymerase chain reaction (RT-PCR). The presence of fusion transcripts was confirmed using reverse transcription (RT) polymerase chain reaction (PCR), and Cycle sequencing. One μg of total RNA was reverse-transcribed and cDNA corresponding to 20 ng total RNA was used as a template in subsequent PCR amplification using the primers M13-KMT2A-4117-FW (5′-TGTAAAACGACGGCCAGTCCTCCGGTCAATAAGCAGGAGAA-3′) and M13-AFF1-1419-REV (5′-CAGGAAACAGCTATG ACCAACTTGGATGGCTCAGCTGTACT-3′). The PCR products were subsequently sequenced using the BigDye Direct Cycle Sequencing Kit according to the company's recommendations (ThermoFisher Scientific, Waltham, MA, USA). The basic local alignment search tool software (BLAST) was used for computer analysis of sequence data (12). The BLAT alignment tool and the human genome browser at the University of California, Santa Cruz (UCSC) were also used to map the sequences on the Human GRCh37/hg19 assembly (BLAT) (13).

Results

G-banding analysis of 23 metaphases revealed a normal female karyotype at diagnosis. FISH analyses performed with the routine probes used in ALL showed normal results with all probes except the one for *KMT2A* splitting. Using the *KMT2A* break-apart probe, FISH on interphase nuclei showed one yellow signal (normal *KMT2A*) and two green signals (the proximal part of *KMT2A*) in 186 out of 217 examined nuclei (data not shown). Subsequent metaphase FISH analysis after hybridization with the same probe showed that the yellow signal was on a normal chromosome 11 whereas one of the two green signals hybridized on a derivative chromosome 4 and the other one on a derivative chromosome 11. We assumed the findings to indicate that the proximal *KMT2A* probe (green signal) was split with the part containing the 5'-end of *KMT2A* being inserted into a derivative chromosome 4 with concomitant loss of the distal part of the probe (red signal) (Figure 1A).

aCGH analysis showed losses restricted to chromosome bands 4q23.3 (421.76 Kb) and 11q21.3q22.1 (497.17 Kb) with breakpoints inside the *AFF1* gene (NM_001166693; exons 5-6) and *KMT2A* (NM_005933; exons 9-10), respectively (Figure 1B and C). Considering the diagnosis and the above FISH data indicating that a *KMT2A::AFF1* fusion might be present, additional FISH analysis was then performed on metaphase spreads using the *KMT2A::AFF1* double fusion probe. This showed that a green signal for *KMT2A* had moved to the der(4) and fused with the red signal for *AFF1.* The der(11) did not show signals for any of the probes (Figure 1D). Based on the FISH and aCGH results, a revised karyotype was therefore derived: 46,XX.ish der(4)ins(4;11)(q21;q23q23)(AFF1+,KMT2A+),der(11)ins (4;11)(q21;q23q23)del(11)(q23q23)(AFF1-,KMT2A-).

Finally, RT-PCR followed by direct cycle Sanger sequencing confirmed the existence of a fusion between exon 9 of *KMT2A* and exon 6 of *AFF1* (Figure 2).

Discussion

We present a case of B-ALL in which combined analyses using G-banding, FISH, and aCGH showed a karyotypically cryptic ins(4;11), resulting in a *KMT2A::AFF1* fusion gene identical to the much more common *KMT2A::AFF1* that

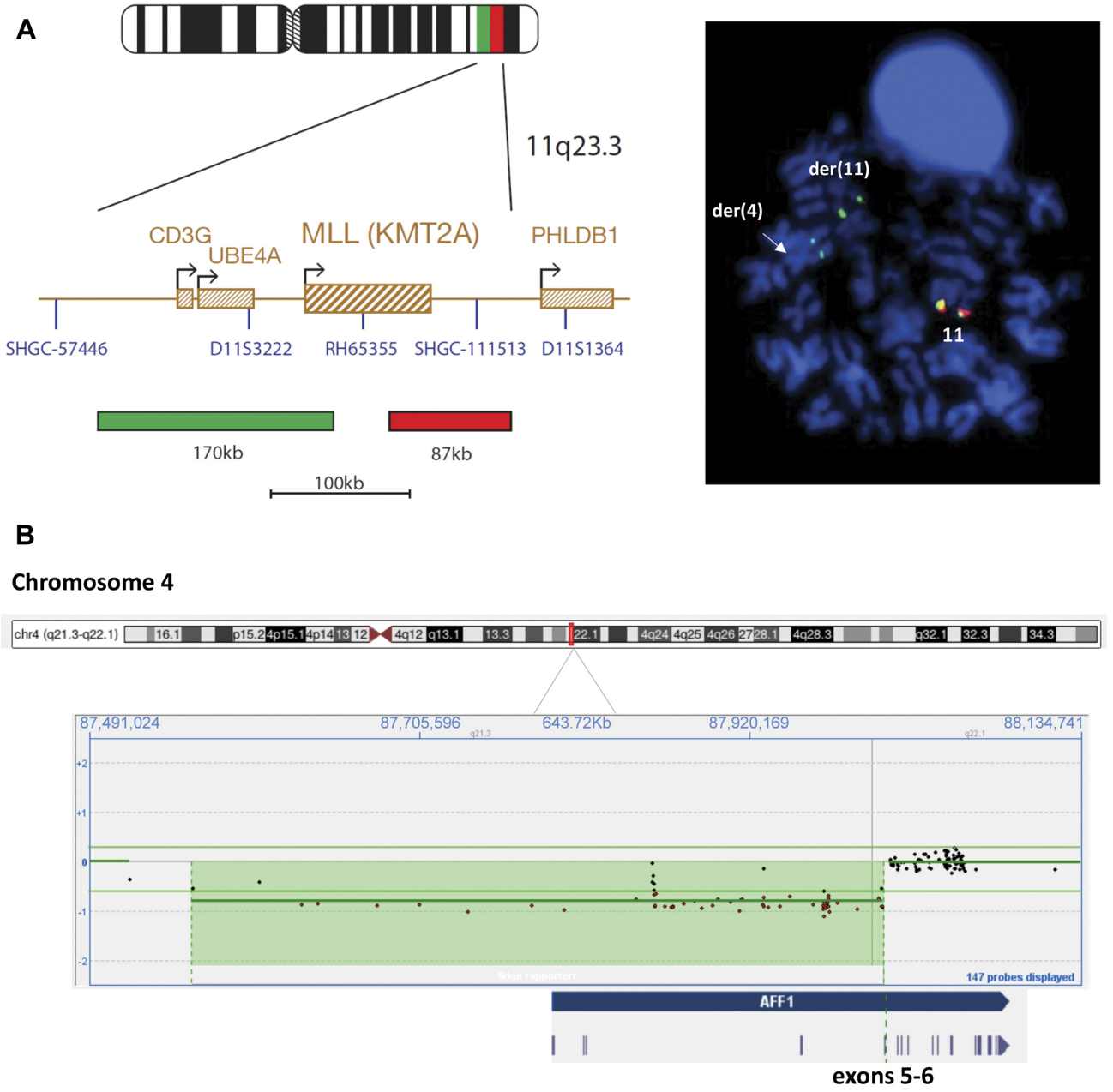

Figure 1. *Continued*

occurs in B-ALL *via* a t(4;11)(q21;q23) translocation. *AFF1* is the most common *KMT2A*-partner in B-ALL, and the detection of this and related *KMT2A*-rearrangements is critical for optimal prognosis assessment and therapy (14). The *KMT2A* gene plays an important role in epigenetic regulation of hematopoietic cell proliferation and differentiation (6). Fusion of the *KMT2A* with numerous partner genes, including *AFF1*, disrupts the KMT2A domain, impairing its normal regulatory functions, and eventually unleashing leukemogenesis (3).

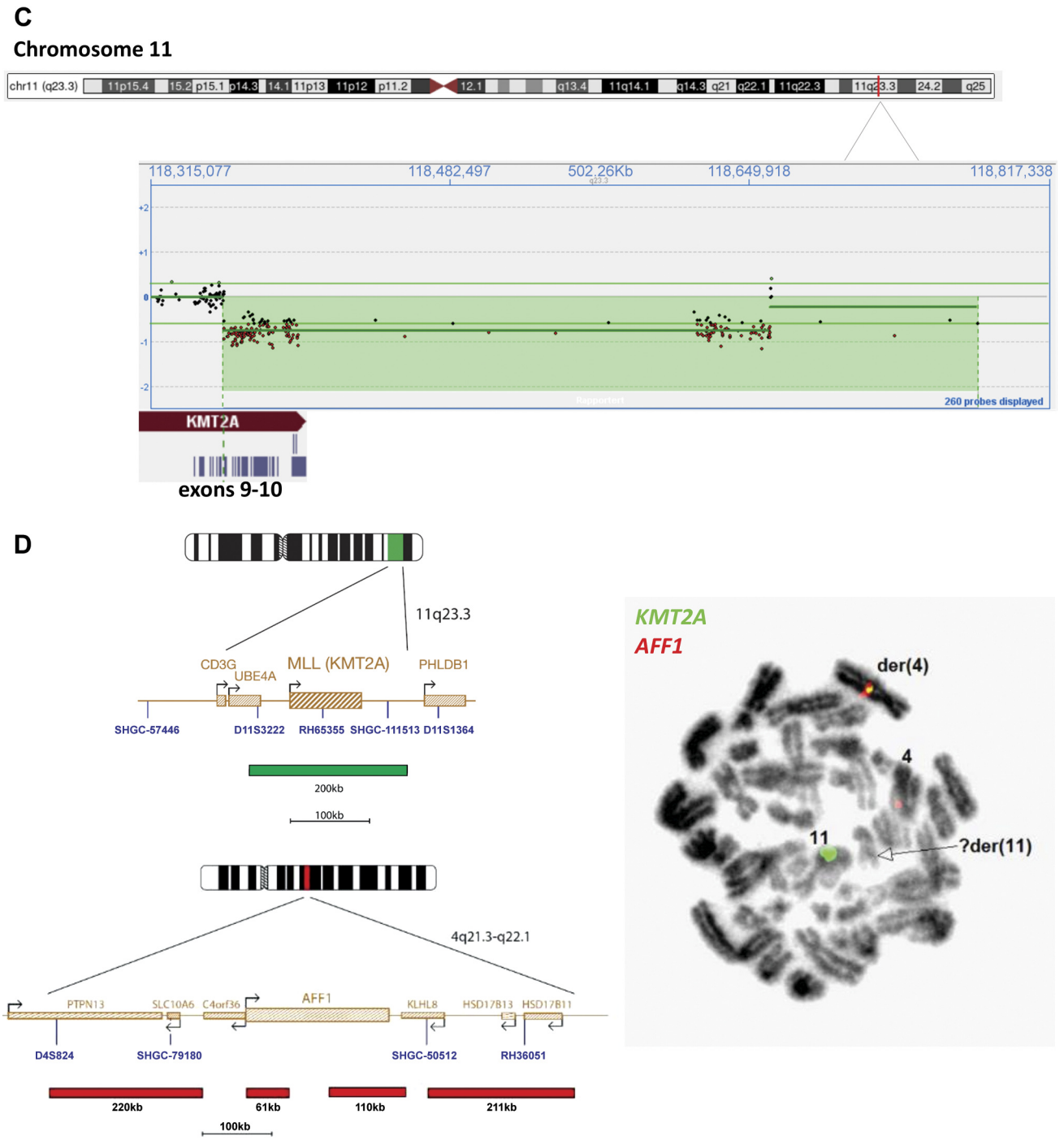

Figure 1. *Fluorescence in situ hybridization (FISH) and array comparative genomic hybridization (aCGH) examination of the B cell acute lymphoblastic leukemia case. (A) Schematic representation of the KMT2A break-apart probe (left); Fluorescence in situ hybridization (FISH) on a metaphase spread with the KMT2A break-apart probe stained with DAPI (right). (B) Overview of aCGH findings for the AFF1 gene mapping on 4q23.3. (C) Overview of aCGH findings for the KMT2A gene mapping on 11q23.3. (D) Schematic representation of the KMT2A::AFF1 probe (left); Fluorescence in situ hybridization (FISH) on metaphase with KMT2A::AFF1 dual fusion probe (right).*

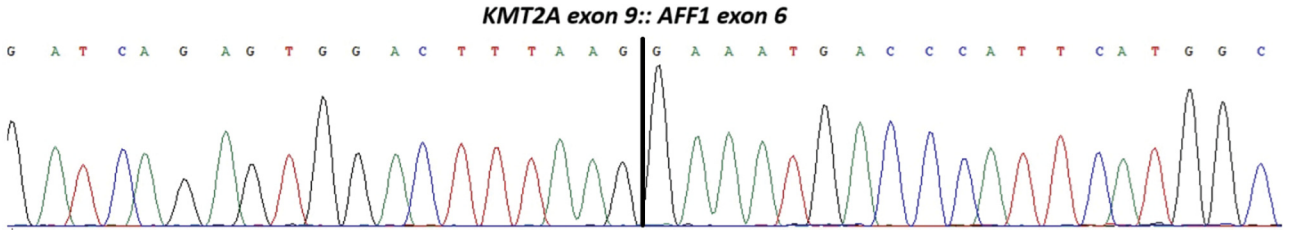

Figure 2. *Molecular genetic analysis of the B cell acute lymphoblastic leukemia case. Partial sequence chromatogram of the cDNA amplified fragment showing the junction of exon 9 of KMT2A with exon 6 of AFF1.*

To the best of our knowledge, only four cases have been reported with a cytogenetically cryptic chromosomal rearrangement leading to *KMT2A::AFF1* (15-18). Von Berg *et al*. (16) described a 30-year-old patient with high-risk ALL and a normal bone marrow karyotype, but having an insertion of 5' *KMT2A* sequences into chromosome 4 resulting in a *KMT2A::AFF1* fusion transcript confirmed by Southern blot analysis and RT-PCR. Tirado *et al.* (15) reported an infant with B-ALL whose diagnostic bone marrow karyotype showing an isochromosome 7q as well as uncertain rearrangements of 9p and 11q. FISH analyses demonstrated insertion of the *KMT2A* gene into chromosome 4. Peterson *et al.* (18) presented a 25-year-old female with B-ALL harboring another cryptic *KMT2A::AFF1* fusion detected by mate-pair sequencing (MPseq), a next-generation sequencing (NGS) strategy. Finally, Othman *et al.* (17) used multitude multicolor banding on a bone marrow sample from a 69-year-old female with B-ALL finding, by dual-color FISH as well as other molecular cytogenetic techniques, and identified another cryptic insertion of 5' *KMT2A* into the *AFF1* locus at 4q21. Additional aberrations were also detected involving at least three chromosomes and five break events.

Ours is thus the fifth case of a cryptic *KMT2A::AFF1* being detected in the bone marrow cells of a B-ALL by a combination of cytogenetic and molecular methods. The *KMT2A* fusion transcript is well known and has important prognostic implications; it is, therefore, fundamental to identify it in a diagnostic screening. The patient's bone marrow was examined several times, and the fusion transcript was always detected. Since *KMT2A* aberrations have poor prognosis, she was accepted for allogeneic stem cell transplantation in CR1, after which she reached complete remission. For now, allogenic hematopoietic cell transplantation (HCT) remains the only curative modality for the management of patients with *KMT2A*-rearranged B-ALL (19). At present, it is not known whether any prognostic differences exist among the many genetic subsets of ALL defined by various partner genes. Ideally, comparative studies should be conducted comparing clinical data on the subgroups, although no evidence is currently at hand indicating prognostic heterogeneity.

Conclusion

The use of different methodological approaches assessing genomic changes at various levels of resolution is necessary to detect cryptic *KMT2A* rearrangements such as *KMT2A::AFF1*. The finding of *KMT2A* alterations in patients with normal karyotypes modifies the prognosis as well as treatment planning and monitoring of the disease. The role of allogeneic hematopoietic stem cell transplantation for B-ALL patients with *KMT2A*-rearrangements seems to have improved clinical outcome significantly.

Conflicts of Interest

The Authors have no conflicts of interest to declare in relation to this study.

Authors' Contributions

Conceptualization, M.B., and F.M.; methodology, M.B., K.A.; software, M.B.; validation, M.B.; formal analysis, M.B; investigation, M.B.; resources, M.B.; data curation, S.S., G.E.T., S.H., and F.M.; writing original draft preparation, M.B, F.M.; writing review and editing, M.B., F.M., S.H.; visualization, M.B., and F.M.; supervision, F.M.; project administration, F.M.; funding acquisition, F.M. All authors have read and agreed to the published version of the manuscript.

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