

SQSTM1::RARA fusion in acute promyelocytic leukemia: the first case report and literature review

Acute promyelocytic leukemia (APL) with the typical *PML::RARA* fusion gene caused by t(15;17)(q24;q21) has been distinguished from other types of acute myeloid leukemia (AML).¹ However, a few cases that cannot be identified as having a *PML::RARA* fusion gene by conventional methods have abnormal promyelocytes that are fully in accordance with the morphology, cytochemistry, and immunophenotype of APL.^{2,3} In this report, we describe *SQSTM1::RARA* and reciprocal *RARA::SQSTM1* fusions, identified for the first time in a patient lacking the t(15;17)(q24;q21)/*PML::RARA* fusion. This patient proved resistant to induction therapy based on all-*trans* retinoic acid (ATRA). To date, a comprehensive literature review has identified 21 novel *RARA* fusion partner genes distinct from *PML::RARA*. The *SQSTM1::RARA* reported in our study represents the 22nd such fusion. Among these variant fusions, *ZBTB16::RARA*, *TTMV::RARA*, and *STAT5B::RARA* are the most frequently reported and are associated with clinical features that differ from those of *PML::RARA*-positive APL.

The index case was an 82-year-old man initially admitted to a local hospital because of persistent fatigue for over 10 days. His clinical course is summarized in Figure 1A. Blood tests showed a white blood cell count of 0.66x10⁹/L, and absolute neutrophil count of 0.07x10⁹/L, hemoglobin level of 93 g/L, and platelet count of 93x10⁹/L. Bone marrow (BM) examination revealed abnormal promyelocytes accounting for 88.5% of the cellularity. These cells were characterized by round nuclei and abundant small azurophilic granules, but no Auer rods were observed (Figure 1B). Flow cytometry identified 85.8% myeloblasts with an immunophenotype positive for CD13, CD33, CD45, CD117, CD64, and MPO, while negative for CD34, CD11b and HLA-DR. Notably, the absence of CD38 expression presented a distinctive feature compared to classic *PML::RARA*-positive APL (Figure 1C). The presumptive initial diagnosis of this patient was APL. However, both reverse transcription polymerase chain reaction and fluorescence *in situ* hybridization (FISH) using the standard *PML::RARA* dual-color dual-fusion probe failed to detect the classic *PML::RARA* fusion. Instead, FISH analysis revealed an atypical signal pattern in 71.5% of cells, characterized by three *RARA* signals (red) and two *PML* signals (green) (Figure 1D). In the context of the karyotype findings, this pattern suggests that the t(5;17) breakpoint did not split the *RARA* locus. Instead, the intact *RARA* gene was translocated to the derivative chromosome 5. The presence of an extra derivative chromosome 5, as confirmed by karyotyping, accounts for the three *RARA* signals observed. The karyotype analysis confirmed this: 46,X,-Y,t(5;17)(q35;q21),der(5)t(5;17),+8[2]/46,id

em,r(1),add(12)(p13)[14]/46,XY[4] (Figure 1E). Whole transcriptome sequencing was subsequently performed, revealing a novel *SQSTM1::RARA* fusion gene involving *SQSTM1* (5q35) and *RARA* (17q21). The fusion results from the joining of exon 5 of *SQSTM1* to exon 3 of *RARA*. Bioinformatic analysis of the sequencing data confirmed that this fusion is in-frame and is predicted to generate a functional chimeric protein. The reciprocal *RARA::SQSTM1* fusion was also detected (Figure 2A). Whole transcriptome sequencing analysis revealed downregulated *CD38* transcript levels and upregulated *BCL2* transcript levels (Figure 2B). These findings may explain the absence of surface protein detection by flow cytometry and the marked sensitivity to subsequent venetoclax therapy. Furthermore, comparative transcriptomic analysis between our patient, patients with classical APL, and patients with non-APL AML identified numerous differentially expressed genes. Enrichment analysis demonstrated that, compared to patients with classical APL and those with non-APL AML, our patient exhibited upregulation of multiple pathways related to protein translation (Figure 2C). We subsequently validated this result using polymerase chain reaction and Sanger sequencing (Figure 2D, E). Additionally, next-generation sequencing mutation analysis revealed *TET2* and *BRCA2* mutations. *TET2* p.Y592Lfs*46 was detected at a variant allele frequency (VAF) of 41.0%; this mutation induces premature translational termination, generating a truncated protein. A *BRCA2* p.P512R missense mutation, with a VAF of 46.5%, was also found.

Given the high clinical suspicion of APL, the patient promptly initiated dual induction therapy. The specific regimen included arsenic trioxide (ATO) at a dose of 0.15 mg/kg daily (administered as 10 mg/day) and ATRA at a dose of 25 mg/m² daily (administered as 20 mg twice daily). The patient's body mass index was 23.4 kg/m². Steroid prophylaxis was not administered during the induction phase. However, ATO and ATRA were discontinued on day 5 due to a negative *PML::RARA* fusion gene result and a poor early response showing no signs of differentiation. A follow-up BM examination 33 days after induction chemotherapy showed a predominance of promyelocytes (88%) with 1% myeloblasts, and MPO staining was strongly positive. The patient was then started on a regimen of venetoclax (100 mg orally on day 1) and azacitidine (100 mg subcutaneously). On the first day of dosing, the patient developed dyspnea and decreased oxygen saturation and was transferred to the Intensive Care Unit. Two weeks later, a repeat BM aspirate showed 84.5% of promyelocytes. The patient subsequently began a 9-day course of combination therapy with venetoclax and ATRA. The venetoclax dose was escalated as

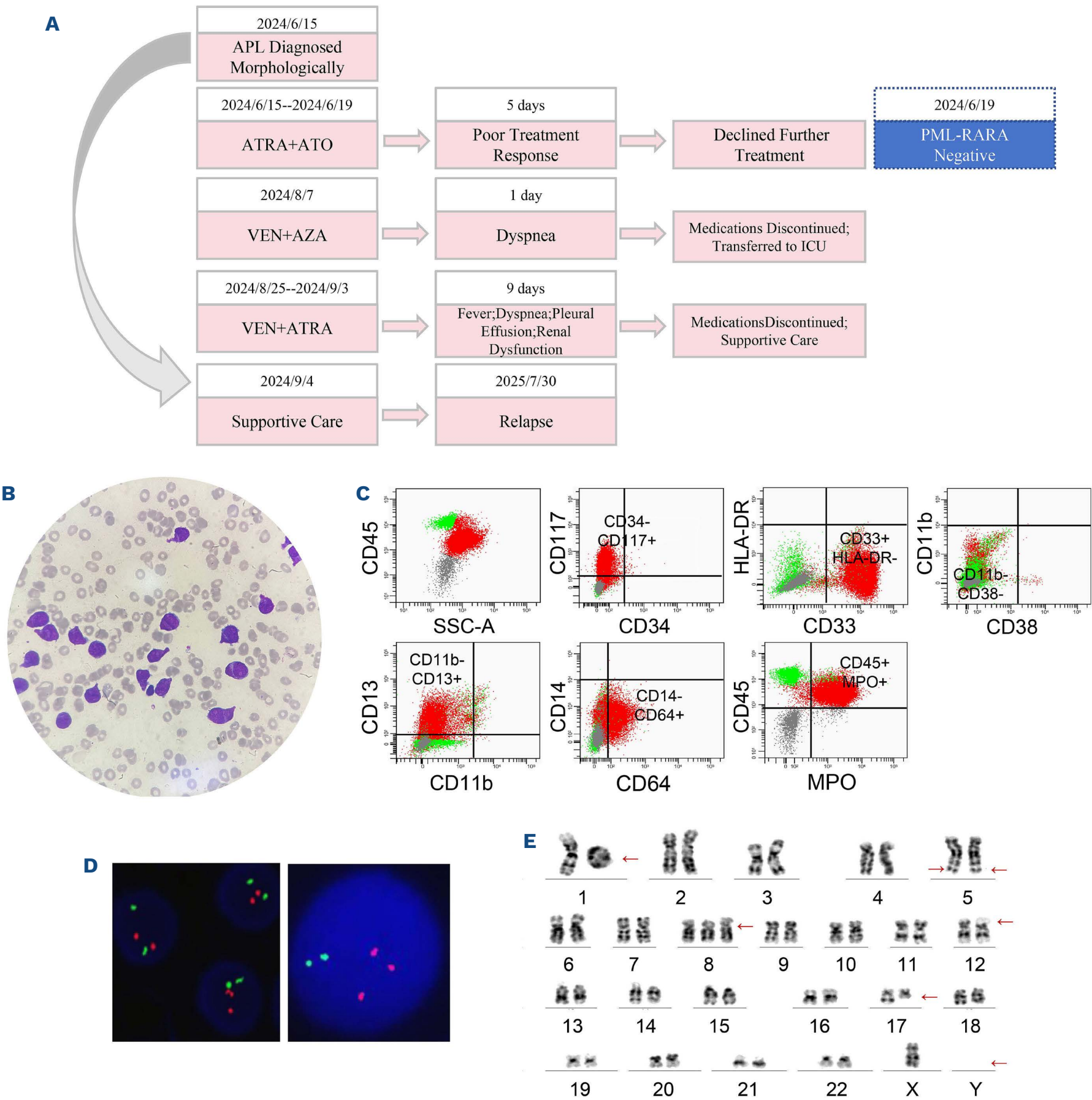


Figure 1. Clinical, morphological, immunophenotypic, and cytogenetic features of the index case at diagnosis. (A) Clinical course of the patient. (B) High-power view of the bone marrow aspirate smear at initial diagnosis (Wright-Giemsa stain; original magnification x1,000). The leukemic cells exhibit round nuclei and abundant small azurophilic granules, consistent with hypergranular promyelocytes. Note the absence of Auer rods. (C) Flow cytometry analysis of the bone marrow aspirate at initial diagnosis. Representative scatter plots show the distinct clustering of the abnormal cell population (highlighted in red), distinguishable from internal normal lymphocytes (green population) and erythroblasts (gray population). Standard quadrant or boundary gates demarcate positive and negative populations, with thresholds defined by internal controls. (D) Fluorescence *in situ* hybridization analysis of bone marrow cells using a *PML* (15q24, green)/*RARA* (17q21.1, red) dual-color, dual-fusion probe. Left panel. A normal control interphase nucleus exhibiting the expected wild-type pattern of two *PML* and two *RARA* signals (2G2R). Right panel. A representative interphase nucleus from the patient exhibiting an atypical signal pattern consisting of two *PML* signals and three *RARA* signals (2G3R) (original magnification x1,000). (E) G-banded karyotype of the bone marrow aspirate at initial diagnosis. A complex abnormal karyotype was identified: 46,X,-Y,t(5;17)(q35;q21),der(5)t(5;17),+8[2]/46,idem,r(1),add(12)(p13)[14]/46,XY[4]. APL: acute promyelocytic leukemia; ATO: arsenic trioxide; ATRA: all-*trans* retinoic acid; AZA: azacitidine; ICU: intensive care unit; VEN: venetoclax.

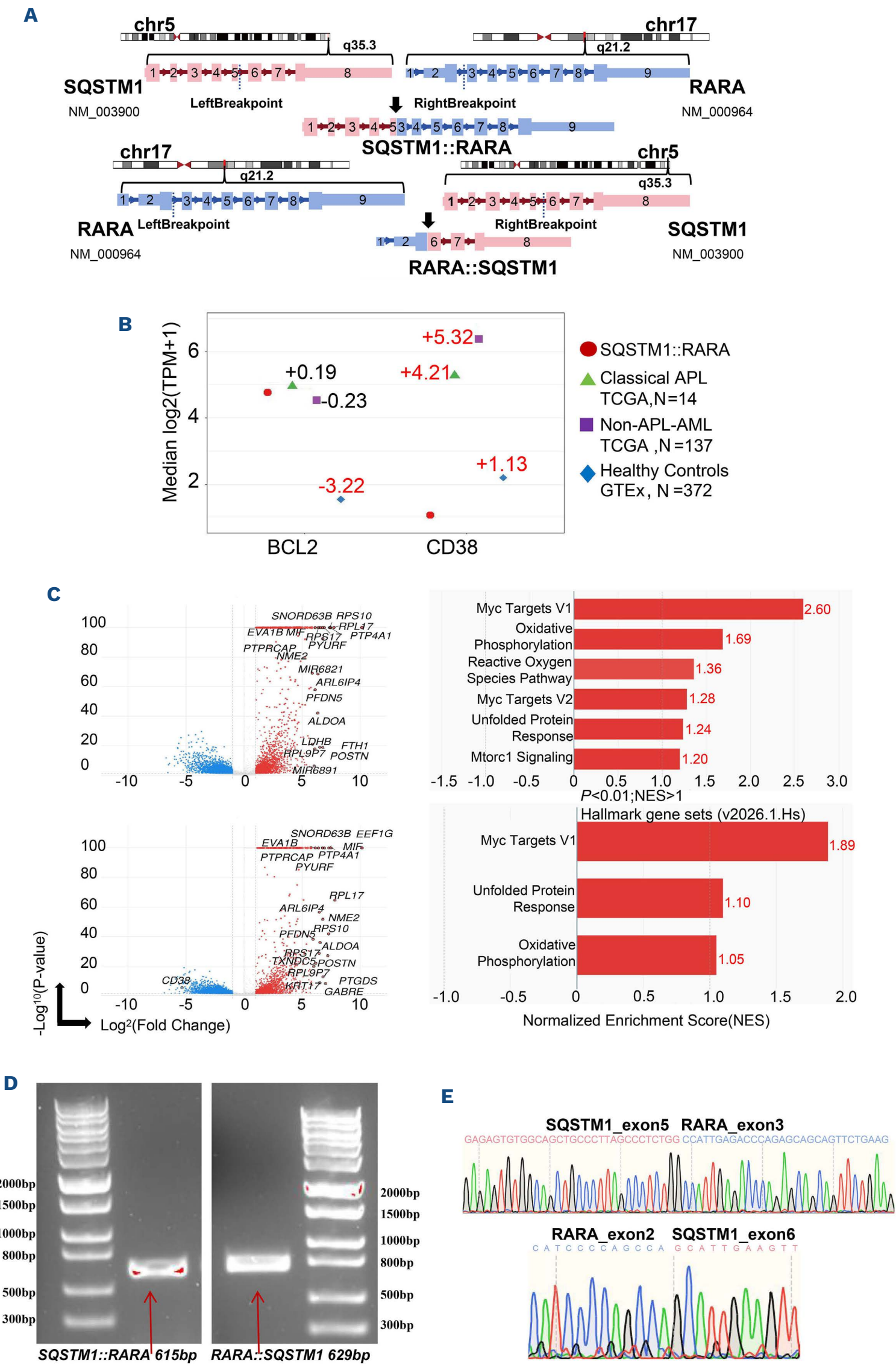


Figure 2. Molecular and transcriptomic characterization of the *SQSTM1::RARA* and reciprocal *RARA::SQSTM1* fusions. (A) Schematic representation of the *SQSTM1::RARA* and reciprocal *RARA::SQSTM1* fusions detected by whole transcriptome sequencing in bone marrow aspirate smears at initial diagnosis and relapse. (B) Median *BCL2* expression ($\log_2(\text{TPM}+1)$) in our patient, TCGA classical APL (N=14), TCGA non-APL-AML (N=137), and a GTEx healthy control group (N=372). (C) Volcano plots of differentially expressed genes in our patient versus classical APL (top left) and non-APL AML (bottom left). Bar charts of upregulated pathways versus those in classical APL (top right) and non-APL AML (bottom right). (D) Detection of the partial *SQSTM1::RARA* (left, 615 bp) and *RARA::SQSTM1* (right, 629 bp) fusions transcripts in cDNA. The specific primer sequences used were: *SQSTM1::RARA*: 5'-CTTGTGTAGCGTCTGCGAGGGAAAG-3' (forward) and 5'-CGGTCGTTTCTCACAGACTCCTTGG-3' (reverse); *RARA::SQSTM1*: 5'-CATGGCCAGCAACAGCAGCTC-3' (forward) and 5'-GCATCTGGGA-GAGGGACTCAATCAG-3' (reverse). (E) Sanger sequencing validation of *SQSTM1*-exon 5 fused to *RARA*-exon 3 (left), and the reciprocal *RARA*-exon 2 fused to *SQSTM1*-exon 6 (right) in a bone marrow aspirate at initial diagnosis. AML: acute myeloid leukemia; APL: acute promyelocytic leukemia; GTEx: Genotype-Tissue Expression; TCGA: The Cancer Genome Atlas; TPM: transcripts per million.

follows: 50 mg daily (days 1-3), 100 mg daily (days 4 and 5), and 200 mg daily (days 6-9). Concomitant ATRA was maintained at 20 mg twice daily throughout this period (days 1-9). During treatment, the patient experienced adverse events – including fever, dyspnea, pleural effusion, and renal dysfunction – which prompted the concurrent administration of intravenous dexamethasone (5 mg daily from day 1 to day 7). On day 10, the patient refused to continue oral venetoclax and ATRA therapy. Management was subsequently limited to supportive care only. Forty-three days after discontinuation of therapy, a blood test at our institution revealed a white blood cell count of $10.36 \times 10^9/\text{L}$, hemoglobin of 86 g/L, and platelet count of $253 \times 10^9/\text{L}$. A BM morphology examination showed only 1% promyelocytes. Most notably, an assessment of minimal residual disease by flow cytometry was negative. The patient was subsequently transferred to the Urology Department for a minimally invasive procedure to address benign prostatic hyperplasia. The patient presented to our hospital for a follow-up visit 1 year later and a BM smear showed relapse. RNA sequencing of the BM aspirate detected the *SQSTM1::RARA* and the reciprocal *RARA::SQSTM1* fusions, with exon breakpoints identical to those identified at the initial diagnosis. This finding confirms the role of *SQSTM1::RARA* as the driver gene in this case.

The human *SQSTM1* gene, located on chromosome 5, spans approximately 16 kilobases and consists of eight exons.^{4,5} It encodes the multifunctional p62/SQSTM1 protein, which is primarily recognized as a selective autophagy receptor that targets cellular components for lysosomal degradation.⁶ Beyond its role in autophagy, p62 functions as a pivotal signaling hub due to its capacity to interact with diverse partners. It critically regulates key pathways including Nrf2-mediated oxidative stress response,⁷ mTORC1-dependent nutrient sensing,⁸ and NF- κ B-driven inflammation.⁹ Notably, p62 dysregulation is implicated in tumorigenesis; gene copy gains in the 5q locus are linked to clear cell renal cell carcinoma, partly through p62 overexpression,¹⁰ and p62 accumulation is a common feature observed in numerous cancer types.^{7,11,12}

This case presents a unique therapeutic trajectory that offers insights into venetoclax biology. The patient showed clear resistance to conventional ATRA/ATO therapy, con-

sistent with the absence of the *PML::RARA* rearrangement. However, the rapid development of dyspnea, hypoxemia, and renal dysfunction within 24 hours of a single dose of 100 mg venetoclax – in combination with azacitidine – strongly suggests incipient tumor lysis syndrome, providing compelling *in vivo* evidence of exceptional venetoclax sensitivity. Despite this early toxicity, subsequent treatment with low-dose venetoclax (escalated to 200 mg) combined with ATRA for only 9 days was sufficient to induce deep remission. Most remarkably, 43 days after all therapy had been discontinued, BM examination revealed only 1% promyelocytes with negative minimal residual disease by flow cytometry. This durable response following limited treatment exposure suggests that even brief BCL-2 inhibition may trigger sustained leukemic clearance in genetically susceptible subsets. We hypothesize that the novel genetic abnormality identified in this case may confer profound BCL-2 dependence, rendering the clone exquisitely sensitive to venetoclax despite resistance to standard APL-directed therapy. The concurrent *TET2* and *BRCA2* pathogenic variants may provide additional context for this patient's atypical course. *TET2* mutations are associated with clonal hematopoiesis and enhanced sensitivity to hypomethylating agents such as azacitidine, which the patient received. The *BRCA2* variant, involving the homologous recombination repair pathway, may contribute to genomic instability and facilitate accumulation of multiple genetic lesions. Together, these variants highlight the value of comprehensive genomic profiling in understanding complex treatment responses. Unlike the classic *PML::RARA* fusion, which is highly sensitive to ATRA and ATO, most rare *X::RARA* variants are known to confer resistance to standard differentiation therapy. Clinical management of these resistant cases remains challenging; however, current literature suggests potential benefits from intensive chemotherapy or novel targeted agents. Notably, BCL-2 inhibitors such as venetoclax and the antibody-drug conjugate gemtuzumab ozogamicin have emerged as promising salvage therapies. Beyond therapeutic responses, distinct clinicopathological features have been observed in specific variants. *TTMV::RARA* cases show a higher incidence in juveniles (<18 years: 75.8%), while *FIP1L1::RARA* occurs primarily in young children (≤ 3 years: 62.5%) with a female

Table 1. Characteristics of 22 *X::RARA* fusions.

<i>X::RARA</i>	Total N	Age, years, range (median)	Gender N (%)	Cytogenetics	Sensitive regimens N (%)	Detection method N (%)	Common mutations N (%)
<i>ZBTB16::RARA</i>	39	7-83 (53)	M: 28 (71.8) F: 10 (25.6) Unknown: 1	t(11;17) (q23;q12)	①ATRA+Chemo: 6 (15.4) ②Chemo: 5 (12.8) ③VEN+DAC: 1 (2.6) ④GO+DA: 1 (2.6) ⑤VEN: 1 (2.6) ⑥ATRA+ATO: 1 (2.6) ⑦IDA+ATO: 1 (2.6)	RT-PCR: 20 (51.3) WTS: 3 (7.7) Southern blot: 1 (2.6)	<i>TET2</i> : 5 (12.8) <i>ARID1A</i> : 5 (12.8) <i>IDH2</i> : 3 (7.7) <i>SRSF2</i> : 3 (7.7) <i>CEBPA</i> : 3 (7.7) <i>FLT3</i> : 3 (7.7) <i>RUNX1</i> : 2 (5.1) <i>CSF3R</i> : 2 (5.1) <i>NRAS</i> : 1 (2.6) <i>SMARCA4</i> : 1 (2.6) <i>KDM6A</i> : 1 (2.6) <i>SAMD9</i> : 1 (2.6) <i>ZRSR2</i> : 1 (2.6) <i>KMT2C</i> : 1 (2.6)
<i>TTMV::RARA</i>	33	2-49 (8) (<18 yrs: N=25, 75.8%)	M: 19 (57.6) F: 14 (42.4)	-	①ATRA+Chemo: 8 (24.2) ②ATRA+ATO: 4 (12.1) ③Chemo: 3 (9.1) ④ATRA;ATO (or ATRA)+VEN: 2 (6.1) ⑤ATRA;ATO (or ATRA)+VEN+Chemo: 2 (6.1) ⑥VEN+AZA: 1 (3.0)	WTS: 33 (100)	<i>NRAS</i> : 4 (12.1) <i>FLT3</i> : 3 (9.1) <i>WT1</i> : 2 (6.1) <i>ARID1B</i> : 1 (3.0) <i>SAMD9</i> : 1 (3.0) <i>TCF3</i> : 1 (3.0) <i>ARID1A</i> : 1 (3.0) <i>KMT2C</i> : 1 (3.0) <i>DHX15</i> : 1 (3.0) <i>KRAS</i> : 1 (3.0) <i>BRAF</i> : 1 (3.0) <i>DNMT3A</i> : 1 (3.0)
<i>STAT5b::RARA</i>	19	17-67 (41)	M: 16 (84.2) F: 3 (15.8)	der(17)	①Chemo: 8 (42.1) ②ATRA;ATO (or ATRA)+ Chemo: 4 (21.1) ③VEN+Chemo: 2 (10.5) ④ATRA: 1 (5.3) ⑤GO: 1 (5.3)	RT-PCR: 14 (73.7) WTS: 3 (15.8) 5'-RACE: 1 (5.3)	<i>RARA</i> : 1 (5.3) <i>STAT5b</i> : 1 (5.3)
<i>NPM1::RARA</i>	13	0.5-74 (12)	M: 9 (69.2) F: 4 (30.8)	t(5;17) (q35;q12)	①ATRA;ATO(or ATRA)+ Chemo: 10 (77.0) ②Chemo: 2 (15.4) ③ATRA: 1 (7.7) ④ATO alone or ATO- based therapy: 1 (7.7)	RT-PCR: 8 (61.5) Southern blot: 1 (7.7)	-
<i>IRF2BP2::RARA</i>	8	19-76 (48.5)	M: 4 (50) F: 4 (50)	t(1;17) (q42;q21)	①ATRA;ATO(or ATRA alone)+Chemo: 3 (37.5) ②ATRA+ATO+GO: 1 (12.5) ③ATRA: 1 (12.5) ④GO: 1 (12.5)	WTS: 6 (75) RT-PCR: 1 (12.5) 5'-RACE: 1 (12.5)	<i>WT1</i> : 2 (25) <i>FLT3</i> : 1 (12.5) <i>NRAS</i> : 1 (12.5) <i>PHF6</i> : 1 (12.5) <i>KIT</i> : 1 (12.5) <i>ARID1B</i> : 1 (12.5)
<i>FIP1L1::RARA</i>	8	0.75-90 (2.65) (≤3 yrs: N=5, 62.5%)	F: 6 (75) M: 2 (25)	t(4;17) (q12;q21)	①ATRA;ATO(or ATRA)+ Chemo: 3 (37.5) ②ATRA: 1 (12.5) ③Chemo: 1 (12.5)	WTS: 2 (25) RT-PCR: 2 (25) 5'-RACE: 2 (25) Conventional karyotype analysis: 1 (12.5)	<i>KRAS</i> : 1 (14.3) <i>NRAS</i> : 1 (14.3) <i>MAP2K2</i> : 1 (14.3) <i>NF1</i> : 1 (14.3) <i>FLT3</i> : 1 (14.3)
<i>TNRC18::RARA</i>	4	50-61 (58.5)	M: 3 (75) F: 1 (25)	t(7;17) (p22; q21)	①ATRA+VEN: 1 (25) ②VEN+ATRA+AZA: 1 (25) ③VEN+AZA: 1 (25) ④ATRA+IDA+Ara-C: 1 (25)	WTS: 3 (75) FISH: 1 (25)	<i>TET2</i> : 1 (25) <i>WT1</i> : 1 (25) <i>ATG2B</i> : 1 (25)

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<i>X::RARA</i>	Total N	Age, years, range (median)	Gender N (%)	Cytogenetics	Sensitive regimens N (%)	Detection method N (%)	Common mutations N (%)
<i>TBL1XR1::RARA</i>	4	-	-	t(3;17) (q26;q21)	①ATO+MTZ: 1 (25) ②Chemo: 1 (25)	WTS: 1 (25) 5'-RACE: 1 (25)	-
<i>BCOR::RARA</i>	3	45-71 (47)	M: 3 (100)	t(X;17) (p11;q21) and all cases showed -Y	①ATRA alone or ATRA- based therapy: 2 (66.7) ②IDA+Ara-C: 1 (33.3)	WTS: 2 (66.7) 5'-RACE: 1 (33.3)	<i>NRAS</i> : 1 (33.3) <i>KRAS</i> : 1 (33.3) <i>FLT3</i> : 1 (33.3)
<i>STAT3::RARA</i>	3	24-45 (26)	M: 2 (66.7) F: 1 (33.3)	t(17;17) (q21;q12)	-	WTS: 3 (100)	-
<i>TFG::RARA</i>	3	16-56 (56)	F: 2 (66.7) M: 1 (33.3)	t(3;14;17) (q12;q11;q21)	①VEN+ATRA: 2 (66.7) ②ATRA: 1 (33.3)	WTS: 2 (66.7) Targeted RNA-seq: 1 (33.3)	<i>SBDS</i> : 1 (33.3)
<i>PRKAR1A::RARA</i>	2	66-67 (66.5)	F: 1 (50) M: 1 (50)	t(17;17) (q24;q12)	ATRA;ATO(or ATRA)+ Chemo: 2 (100)	WTS: 1 (50)	<i>KRAS</i> : 1 (50)
<i>NUP98::RARA</i>	2	31-70 (50.5)	M: 2 (100)	t(11;17) (p15;q21)	IDA+Ara-C: 1 (50)	WTS: 2 (100)	-
<i>HNRNPC::RARA</i>	2	29-41 (35)	M: 1 (50) F: 1 (50)	t(14;17) (q11;q21)	VEN+AZA: 1 (50)	WTS: 2 (100)	PHF6: 1 (50)
<i>IRF2BP1::RARA</i>	1	51	F	t(19;17) (q13;q21)	Sensitive to ATRA	WTS	-
<i>THRAP3::RARA</i>	1	34	M	t(1;17) (q3;q21)	VEN+DAC	WTS	-
<i>NuMA::RARA</i>	1	0.5	M	t(11;17) (q13;q21)	Sensitive to ATRA	-	-
<i>FNDC3B::RARA</i>	1	36	M	t(1;17) (q42;q21)	Ara-C+DNR	5'-RACE	-
<i>OBFC2A::RARA</i>	1	59	M	t(2;17) (q32;q21)	ATRA+IDA+Ara-C	Array CGH analysis	-
<i>NAB2::RARA</i>	1	73	M	t(12;17) (q13;q21)	-	WTS	<i>TET2</i> ; <i>ZRSR2</i> ; <i>SRSF2</i> ; <i>IDH2</i>
<i>GTF2I::RARA</i>	1	35	M	t(7;17) (q11;q21)	-	5'-RACE	-
<i>SQSTM1::RARA</i>	1	82	M	t(5;17) (q35;q21)	-	WTS	<i>TET2</i> ; <i>BRCA2</i>

Data are presented as the number of cases out of the total reported number of cases for each fusion subtype. Ara-C: cytarabine; ATO: arsenic trioxide; ATRA: all-*trans* retinoic acid; AZA: azacitidine; Chemo: chemotherapy; DA: daunorubicin and cytarabine; DAC: decitabine; DNR: daunorubicin; F: female; FISH: fluorescence *in situ* hybridization; GO: gemtuzumab ozogamicin; IDA: idarubicin; M: male; MTZ: mitoxantrone; 5'RACE: 5'-rapid amplification of cDNA ends; RNA-seq: RNA-sequencing; RT-PCR: reverse transcription polymerase chain reaction; WTS: whole transcriptome sequencing; VEN: venetoclax.

predominance (75%). Additionally, all reported *BCOR::RARA* cases were male and exhibited loss of the Y chromosome. From a diagnostic perspective, whole transcriptome sequencing is instrumental in discovering novel fusions. For rare fusions, such as *SQSTM1::RARA*, which show suboptimal responses, early molecular identification and transition to alternative potent therapies are crucial for improving outcomes. The characteristics of the 22 *X::RARA* cases are summarized in Table 1. Ethical review and approval were not required for this case report in accordance with local legislation and in-

stitutional requirements. The study was conducted in accordance with the ethical standards of the People's Republic of China. Written informed consent was obtained from the patient for publication of this case report and accompanying images.

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<https://doi.org/10.3324/haematol.2025.300329>

Received: December 4, 2025.

Accepted: April 10, 2026.

Early view: April 23, 2026.

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Disclosures

No conflicts of interest to disclose.

Contributions

XY, JA, and HL collected the clinical data. NL, ZC, and SS analyzed the genetic results. H-HZ and NL drafted the manuscript. All authors approved the final version.

Funding

This work was supported by the National Natural Science Foundation of China (82450101, 82370169), the “Dengfeng” Talent Training Program of Beijing Hospitals Authority (DFL20240301), and funds from the Chinese Institutes for Medical Research, Beijing (CIMR) to H-HZ.

Data-sharing statement

All data generated or analyzed during this study are included in this published article.

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