American Journal of Case Reports

e-ISSN 1941-5923
© Am J Case Rep, 2026; 27: e953926
DOI: 10.12659/AJCR.953926

Received: 2026.04.27
Accepted: 2026.10.21
Available online: 2026.11.06
Published: 2026.XX.XX

Clonal Evolution of *CALR*-Mutated Essential Thrombocythemia With Chromosome 1q Jumping Translocations: A Rare Case Report

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

ABCDEF Noureldien Darwish
ABCDEFG Michael X. Wang

Department of Pathology and Laboratory Medicine, Albany Medical College, Albany, NY, USA

Corresponding Author: Michael X. Wang, Department of Pathology and Laboratory Medicine, Albany Medical Center, 43 New Scotland Ave, Albany, NY 12208, USA
Phone: +1-518-262-8425, e-mail: WangMi@amc.edu

Financial support: None declared
Conflict of interest: None declared

APPROVED GALLEY PROOF

Patient: Male, 77-year-old
Final Diagnosis: Post-ET myelofibrosis arising from calreticulin (CALR)-mutated ET
Symptoms: Splenomegaly
Clinical Procedure: —
Specialty: Hematology • Pathology
Objective: Rare disease

Background: Essential thrombocythemia (ET), a chronic myeloproliferative neoplasm, typically follows an indolent clinical course; however, some patients may undergo clonal evolution to post-ET myelofibrosis or leukemic transformation. Disease progression is frequently associated with acquisition of additional cytogenetic and molecular abnormalities, although the precise molecular mechanisms underlying this evolution remain incompletely understood. We describe a case of post-ET myelofibrosis arising from calreticulin (*CALR*)-mutated ET associated with gain of chromosome 1q21, chromosome 1q jumping translocations, and a *BRIP1* gene mutation.

Case Report: We report a 77-year-old man with *CALR*-mutated ET diagnosed in 2007 and treated with long-term hydroxyurea. After 18 years, he developed progressive anemia, circulating blasts, and splenomegaly. Bone marrow biopsy demonstrated marked hypercellularity (~95% cellularity) with myeloid and megakaryocytic hyperplasia, megakaryocytic atypia, and loose clustering, along with severe reticulin fibrosis (MF-3, myelofibrosis tier 3, WHO grade system) and increased blasts (5%-8% by CD34 immunohistochemistry). Next-generation sequencing (163-gene/40-fusion panel) confirmed *CALR* p.L367Tfs46 mutation persistence and *BRIP1* p.R798* nonsense mutation acquisition. Fluorescence in situ hybridization showed gain of chromosome 1q21. Cytogenetics demonstrated jumping translocations of 1q involving chromosomes 14, 16, and 22.

Conclusions: This report describes the case of a patient with an 18-year history of *CALR*-mutated ET, presenting with leukocytosis, anemia, marked thrombocytosis, splenomegaly, severe myelofibrosis, and acquisition of a *BRIP1 p.R798* mutation, gain of chromosome 1q21, and jumping translocations of the long arm of chromosome 1 (1q) involving recipient chromosomes 14, 16, and 22. This case illustrates late clonal evolution of *CALR*-mutated ET to post-ET myelofibrosis associated with rare cytogenetic and molecular abnormalities. These findings expand the spectrum of genetic events observed during this disease progression.

Keywords: Hematology • Thrombocythemia, Essential • Myelofibrosis • Chromosome Aberrations • Mutation • Case Reports
Full-text PDF: https://www.amjcaserep.com/abstract/index/idArt/953926

1759 2 2 23

Publisher's note: All claims expressed in this article are solely those of the authors and do not necessarily represent those of their affiliated organizations, or those of the publisher, the editors and the reviewers. Any product that may be evaluated in this article, or claim that may be made by its manufacturer, is not guaranteed or endorsed by the publisher

This work is licensed under Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International (CC BY-NC-ND 4.0)

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

APPROVED GALLEY PROOF

Introduction

Essential thrombocythemia (ET) is a Philadelphia chromosome-negative myeloproliferative neoplasm characterized by sustained thrombocytosis and increased numbers of enlarged, mature atypical megakaryocytes in a typically normocellular bone marrow. The megakaryocytes show deeply lobulated and hypersegmented nuclei [1]. Epidemiologic data from the SEER program estimate an incidence of approximately 1.55 cases per 100 000 person-years [2].

At the molecular level, ET is driven by recurrent somatic mutations involving *JAK2, CALR*, or *MPL*. *JAK2* p.V617F is present in approximately 50% to 60% of cases, *CALR* mutations in 25% to 35% [3], and *MPL* mutations in 5% to 10% [4]. *CALR* mutations are frameshift alterations that generate a novel C-terminal protein sequence, most commonly type 1 and type 2 variants. *CALR*-mutated ET is associated with higher platelet counts, younger age at diagnosis, and lower thrombotic risk compared with *JAK2*-mutated disease [5-7].

Despite its generally indolent course, ET may undergo clonal evolution to post-ET myelofibrosis or acute myeloid leukemia, typically over years of disease progression. Transformation to myelofibrosis occurs in approximately 10% of patients, whereas leukemic transformation occurs in approximately 3% [8]. Disease progression is associated with additional somatic mutations and cytogenetic abnormalities reflecting genomic instability and clonal selection. While recurrent mutations in genes such as *TP53, ASXL1*, and *SRSF2* have been well described [9], the chromosome 1q jumping translocations and *BRIP1* mutations remain rare in this context.

Table 1. Jumping chromosome 1q translocations in myeloid neoplasms: key cohorts compared.

Feature	Najfeld 2010 (BJH) [17]	Sanford 2015 (CLML) [18]	Couture 2018 (Case Rep Genet) [19]	Our case
Study design	Retrospective cytogenetics (1984-2009)	Retrospective cytogenetics (2003-2014)	3 institutional cases + literature review	Single case
Total JT / 1q-JT cases	**23**	**10**	**33**	**1**
Disease spectrum	MPN 7, MDS/AML 1**6**	AML **4**, MDS **4**, post-PV-MF **2**	MDS → AML (33 cases, incl. 3 Yale)	ET → post-ET MF
ET cases	0	0	Not included (MDS/AML focus)	**1 (*CALR*+)**
Donor chromosome	1q	Chr 1 in all	1q	1q
Timing (diagnosis → JT)	Mean time to acquire: MPN **47 mo**; MDS 2**1-34 mo**	Median **24.9 mo** (0-248); 1 case at diagnosis	Not summarized as a single median across all 33 cases	**~ 216 mo (18 yrs)**
JT persistence	Often persisted until AML; sometimes cryptic by FISH	Persisted median **6 mo** (4.7-10.8) in those with repeats	Persistent in 3 Yale cases with increasing abnormal%	Not yet known
AML transformation association	High; mean JT → AML **8-9 mo**	High-risk; median OS after JT **9 mo**	All 3 Yale cases progressed to AML and died	Increased blasts (5-8%), not AML
Recipient fusion location	**81% pericentric, 19% telomeric**	Not given as cohort% in pasted text	**82% telomeric, 18% pericentric / interstitial**	Dicentric suggests pericentric involvement
Recipient chromosomes (aggregate)	4,5,6,7,9,13,14,15,16,18, 19,20,21,22,X,Y	Wide; includes 3,5,6,7,8, 9,10,11,12,13,14,15,16,1 7,18,19,20,21,22,X,Y	Frequent: 15p,22p,21p,14p,13p, Yq,17q,18q,21q; also 16q11.2q12, 7q11,20q12q13.1	**14,16,22**
Unique highlight	Defines 1q-JT as marker of imminent AML	Provides patient-level donor sites + survival after JT	Quantifies recipient patterns + acrocentric frequency	CALR-mutated ET progressing to MF-3 with chromosome 1q jumping translocations

Abbreviations: FISH, fluorescence in situ hybridization; JT, jumping translocation; MF, myelofibrosis; MPN, myeloproliferative neoplasms; OS, overall survival; PV, polycythemia vera.

This work is licensed under Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International (CC BY-NC-ND 4.0)

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

Table 2. Hematologic findings at time of disease acceleration.

Laboratory parameter	Result	Units	Reference range	Flag
Hematology				
White blood cell count	**18.2**	× 10^3/μL	3.5-11.0	**H**
Red blood cell count	**2.88**	× 10^6/μL	4.7-6.1	**L**
Hemoglobin	**10.2**	g/dL	13.0-18.0	**L**
Hematocrit	**32.1**	%	41.0-53.0	**L**
Mean corpuscular volume	**111.5**	fL	80.0-95.0	**H**
Red cell distribution width	**21.0**	%	< 14.5	**H**
Platelet count	**868**	× 10^3/μL	150-350	**H**
Mean platelet volume	10.5	fL	9.4-12.3	–
Neutrophils	60.4	%	45.0-78.0	–
Immature granulocytes	**19.9**	%	0.0-1.0	**H**
Lymphocytes	**7.0**	%	15.0-47.0	**L**
Monocytes	11.2	%	0.0-12.0	–
Eosinophils	0.5	%	0.0-7.0	–
Basophils	1.0	%	0.0-2.0	–

Flags: H = above reference range; L = below reference range; – = within reference range.

BRIP1 encodes a DNA helicase involved in homologous recombination and the Fanconi anemia pathway [10]. Although *BRIP1* alterations are well characterized in solid tumors, particularly breast and ovarian carcinomas, they are infrequently reported in hematologic malignancies [11-13]. Their role in cancer remains undefined but may reflect underlying genomic instability.

Chromosome 1q gains and structural rearrangements are more commonly described in plasma cell neoplasms [14] but are also observed in myeloid disorders, particularly in advanced disease such as in acute myeloid leukemia (AML) [15,16]. Jumping translocations of 1q are rare events associated with chromosomal instability and disease progression in myeloid neoplasms such as myeloproliferative neoplasms, including polycythemia vera and primary myelofibrosis, myelodysplastic syndromes (MDS), MDS/AML, and AML (**Table 1**) [17-19].

Here, we describe a case of *CALR*-mutated ET evolving over 18 years into post-ET myelofibrosis with acquisition of *BRIP1* mutation, gain of chromosome 1q21, and chromosome 1q jumping translocations involving recipient chromosomes 14, 16, and 22. The conjunction of a *BRIP1* mutation and chromosome 1q21 abnormalities in post-ET myelofibrosis, to our knowledge, has not been previously reported. This report is primarily descriptive and aims to expand the recognized spectrum of cytogenetic evolution in ET. Recognition of such rare events may improve our understanding of this disease progression.

Case Report

The patient was a 77-year-old man who received a diagnosis of essential thrombocythemia (ET) in 2007 after presenting with marked thrombocytosis (approximately 900 × 10^3/μL). Molecular testing identified a *CALR* p.*L367Tfs*46* mutation, with no *JAK2* or *MPL* mutations or *BCR::ABL1* rearrangement detected. The bone marrow biopsy reportedly confirmed ET, and hydroxyurea therapy was initiated, resulting in good initial disease control. However, the original diagnostic materials, including the peripheral blood smear, bone marrow biopsy, and cytogenetic and molecular testing reports, were unavailable for review.

The patient was lost to regular follow-up after 2017 and subsequently had intermittent hydroxyurea exposure. In 2023, he developed a non-healing skin ulcer and also received a diagnosis of squamous cell carcinoma of the tongue, prompting dose reduction of hydroxyurea.

Beginning in 2023, peripheral blood studies demonstrated persistent thrombocytosis with macrocytic anemia, dysplastic granulocytes, abnormal large hypogranular platelets, and occasional circulating blasts, raising concern for disease progression. Splenomegaly was present. Most recent laboratory values showed a white blood cell count of 18.2 × 10^3/μL, platelet count of 868 × 10^3/μL, and hemoglobin level of 10.2 g/dL (MCV 111.5 fL) (**Table 2**).

APPROVED GALLEY PROOF

This work is licensed under Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International (CC BY-NC-ND 4.0)

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

APPROVED GALLEY PROOF

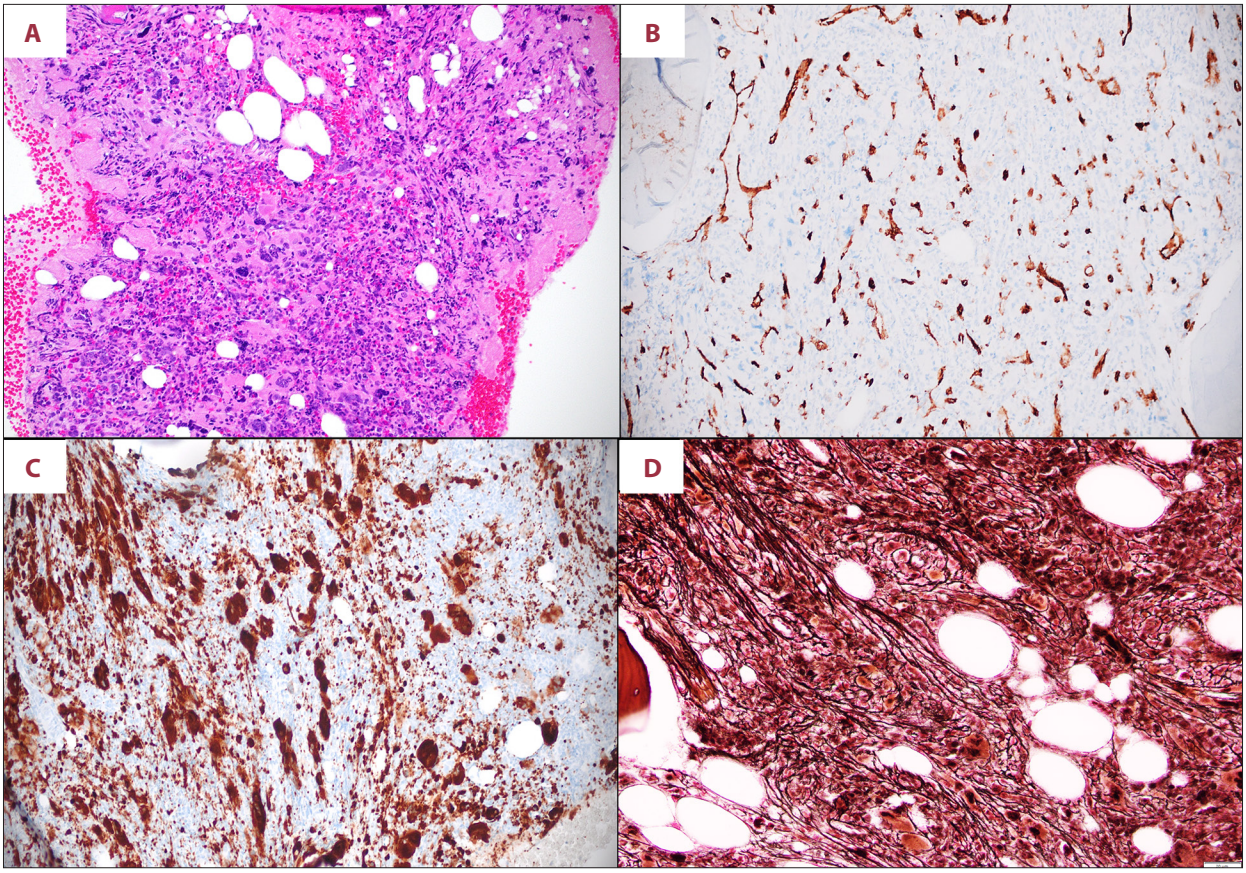


Figure 1. Bone marrow morphology, immunohistochemistry and reticulin stain. (**A**) Hematoxylin and eosin–stained section shows a markedly hypercellular marrow with myeloid hyperplasia and myeloid left-shifted maturation. Megakaryocytes are increased with clustering and display marked atypia, including hyperchromatic, hypolobated, and occasionally hyperlobated nuclei. (**B**) CD34 immunohistochemical stain highlights increased blasts (5%-8% bone marrow nucleated cells) with associated vascular endothelial staining. (**C**) CD61 immunohistochemical stain demonstrates megakaryocytic hyperplasia with prominent clustering and atypia including large and small forms. The platelet aggregates are also stained on the background. (**D**) Reticulin stain shows diffuse and dense reticulin fibrosis with extensive intersections and coarse bundles of thick fibers consistent with MF-3 (severe myelofibrosis, WHO grade system).

A bone marrow biopsy was performed in December 2025, revealing a markedly hypercellular marrow (approximately 95% cellularity) with increased myeloid-to-erythroid ratio (approximately 5: 1). Granulopoiesis showed left-shifted maturation. Megakaryocytes were increased and atypical, showing a spectrum of hyperlobated, hypolobated, and irregular nuclear forms with loose clustering (**Figure 1A**, hematoxylin and eosin microscopy). Immunohistochemistry demonstrated CD34-positive blasts comprising 5% to 8% of nucleated marrow cells (**Figure 1B**). CD117 highlighted 8% to 10% myeloid precursors (not shown). CD61 highlighted increased atypical megakaryocytes (**Figure 1C**). Reticulin staining showed diffuse fibrosis consistent with MF-3 (WHO grade system, **Figure 1D**).

Flow cytometric analysis was performed on the bone marrow aspirate specimen at our institution. It identified approximately 2% aberrant myeloid blasts with partial CD4 expression and reduced expression of CD13 and CD33. Hypogranular granulocytes (71%) showed aberrant CD56 and CD14 expressions. No abnormal B-cell or T-cell populations were identified.

Next-generation sequencing, fluorescence in situ hybridization (FISH), and conventional cytogenetic karyotyping were performed at a Clinical Laboratory Improvement Amendments–certified commercial reference laboratory. The results and the clinical significance were interpreted by pathologists at our institution.

Next-generation sequencing confirmed persistence of *CALR* p.L367Tfs46 (variant allele frequency [VAF] 27.4%) and acquisition of *BRIP1* p.R798 (VAF 26.0%), a pathogenic nonsense mutation predicted to result in a truncated protein. FISH demonstrated gain of chromosome 1q21 using a probe set of 1p32/1q21 including the cyclin-dependent kinase regulatory

This work is licensed under Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International (CC BY-NC-ND 4.0)

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

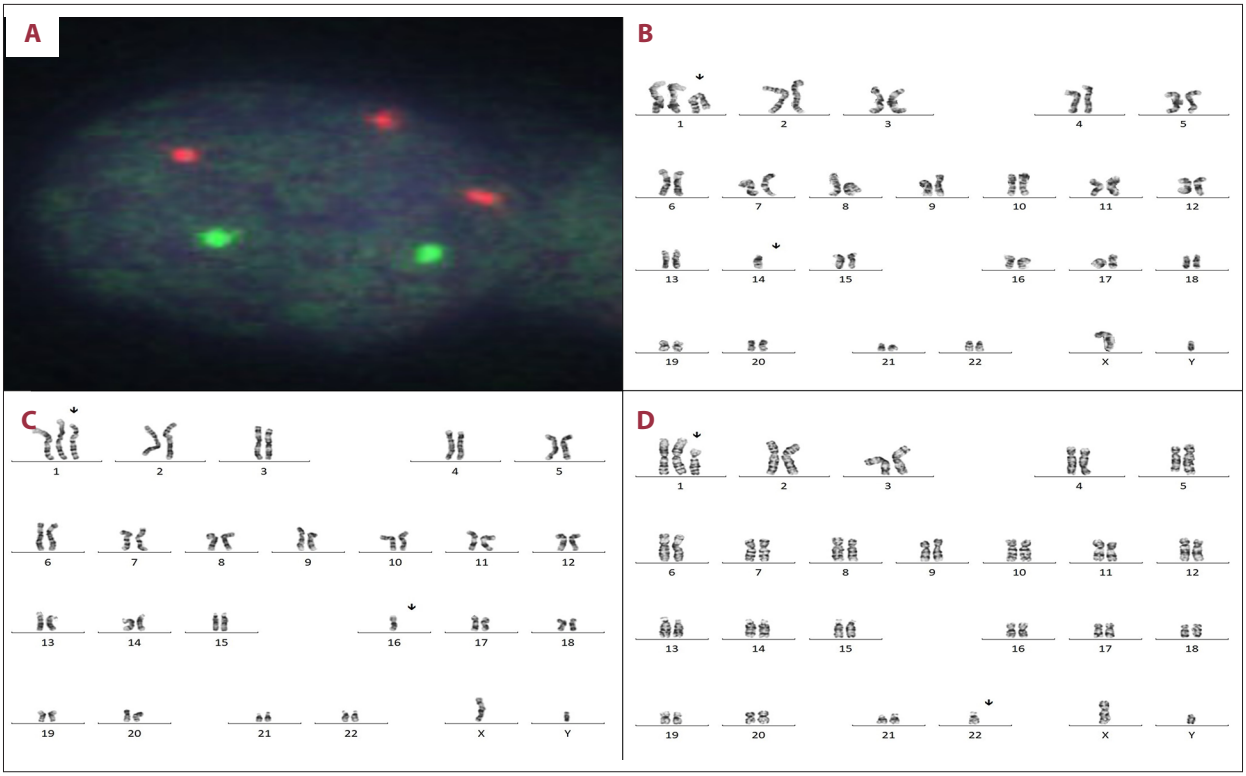


Figure 2. Fluorescence in situ hybridization (FISH) and chromosome cytogenetic findings. The FISH and conventional karyotyping were performed at a Clinical Laboratory Improvement Amendments–certified commercial reference laboratory. The results were interpreted by professionals at the reference laborotory and pathologists at our institution. (**A**) The bone marrow interphase FISH analysis shows gain of chromosome 1q signals (red, 3 dots) relative to control probes (green, 2 dots) using a probe set of 1p32 (green)/1q21, including the *CKS1B* locus, consistent with 1q copy number gain. (**B-D**) G-banded metaphase karyotypes demonstrating unbalanced translocations involving chromosome 1q and multiple recipient chromosomes, including chromosomes 14 (**B**), 16 (**C**), and 22 (**D**). Arrows indicate derivative chromosomes containing additional 1q material, consistent with jumping translocations of chromosome 1q. Collectively, these findings show 1q gain and structural rearrangements of 1q segment to chromosomes 14, 16, and 22 (jumping translocations).

subunit 1 (*CKS1B)* locus (**Figure 2A**). Cytogenetics revealed jumping translocations of the long arm of chromosome 1 (1q) involving recipient chromosomes 14, 16, and 22 (**Figure 2B-2D**). The karyotype showed

46,XY,+1,dic(1;16)(p13;p13.3)[12] / 46,XY,+1,dic(1;22)(p13;p13)[5]/ 46,XY,+1,dic(1;14)(p13;p13)[3]. Together, cytogenetic analysis demonstrated 3 copies of 1q with a small segment of 1p, resulting in small losses of the 16p, 14p, and 22p regions (**Figure 2B-2D**). FISH confirmed these 3 translocation events at the chromosomal level by demonstrating 3 copies of 1q using the 1q21 probe (3 red dots in **Figure 2A**).

These findings were consistent with post-ET myelofibrosis with clonal evolution associated with acquisition of complex cytogenetic abnormalities including 1q jumping translocation involving recipient chromosomes 14, 16, and 22 and *BRIP1* mutation.

Discussion

Prior studies have described 1q jumping translocations primarily in multiple myeloma [14], myelodysplastic syndromes, post-polycythemia vera myelofibrosis, and AML (**Table 1**) [15-19]. Jumping translocations of chromosome 1q represent rare cytogenetic events associated with advanced clonal evolution and genomic instability in myeloid neoplasms [20]. The 1q pericentromeric region is enriched in repetitive DNA sequences and is particularly susceptible to structural instability under conditions of proliferative stress and epigenetic dysregulation [21]. These events typically arise late in disease evolution and are associated with disease progression.

According to the current *WHO Classification of Tumours: Haematolymphoid Tumours,* 5th edition, the diagnosis of multiple myeloma requires 10% or greater clonal bone marrow plasma cells in conjunction with one or more myeloma-defining events, including CRAB features (hypercalcemia, renal insufficiency, anemia, and/or osteolytic bone lesions). In

APPROVED GALLEY PROOF

This work is licensed under Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International (CC BY-NC-ND 4.0)

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

APPROVED GALLEY PROOF

the present case, fewer than 5% of polytypic plasma cells were demonstrated by CD138, kappa, and lambda light chain immunohistochemistry in bone marrow biopsy. Although macrocytic anemia was present, no other myeloma-defining events were documented. Therefore, the findings did not support a diagnosis of multiple myeloma. The diagnosis of AML generally requires 20% or greater blasts in the bone marrow or peripheral blood, except in cases with specific AML-defining genetic abnormalities, including *PML::RARA, RUNX1::RUNX1T1, CBFB::MYH11*, and certain other genetic abnormalities recognized by the current classification. In the present case, flow cytometry identified only 2% aberrant myeloid blasts in the bone marrow aspirate, while CD34 immunohistochemistry demonstrated approximately 5% to 8% myeloid blasts in the bone marrow core biopsy. No AML-defining genetic abnormality was identified by cytogenetic analysis, FISH, or next-generation sequencing. Therefore, the present findings did not meet the diagnostic criteria for AML. Overall, the findings of longstanding thrombocytosis, a myeloproliferative neoplasm–defining *CALR* mutation, absence of *BCR::ABL1*, grade 3 bone marrow fibrosis, anemia, splenomegaly, and increased circulating and bone marrow blasts (but < 20%) meet the diagnostic criteria for post-ET myelofibrosis.

ET as the initial disease context is rarely reported in association with this abnormality. Borri et al reported 1 case of an 86-year-old woman with myelofibrosis evolved from ET who showed unbalanced translocations involving chromosome 1 and the acrocentric chromosomes 14, 15, and 22 [22]. In the present case, chromosome 1q jumping translocations involving chromosomes 14, 16, and 22 were detected after a prolonged disease course of 18 years in a patient with *CALR*-mutated ET. Although these 2 cases highlight the occurrence of 1q jumping translocations in post-ET myelofibrosis, whether these genetic alterations are directly associated with disease progression remains to be determined.

BRIP1 is a DNA repair gene involved in homologous recombination [23]. In the present case, a *BRIP1* mutation was identified at a VAF of 26%, consistent with a somatic event within the neoplastic clone. While disruption of DNA repair pathways may be associated with genomic instability, this case alone does not establish a causal role for *BRIP1* in driving cytogenetic evolution. The significance of the coexistence of *BRIP1* mutation with 1q abnormalities is unclear. Overall, this single case report demonstrates the coexistence of chromosome 1q jumping translocations and a *BRIP1* mutation in post-ET myelofibrosis, thereby expanding the spectrum of genetic alterations described in this disease. However, as an isolated observation, it cannot establish the prevalence, prognostic significance, or molecular mechanistic role of these abnormalities. Furthermore, the prolonged clinical course and intermittent follow-up limit definitive conclusions regarding the temporal sequence and acquisition of these genetic events.

Conclusions

This case documents late clonal evolution of *CALR*-mutated ET to post-ET myelofibrosis associated with *BRIP1* mutation and 1q jumping translocations involving recipient chromosomes 14, 16, and 22. These findings expand the recognized spectrum of genetic abnormalities in advanced ET and illustrate the importance of comprehensive reassessment in patients with disease progression. The coexistence of *BRIP1* mutation and chromosome 1q abnormalities is of interest and may reflect underlying genomic instability; however, causal relationship and mechanistic or prognostic implications remain speculative and require further study in larger cohorts.

Institution Where Work Was Done

Albany Medical Center, Albany, NY, USA.

Declaration About Informed Consent

This case report was approved by the Institutional Review Board of Albany Medical Center. The requirement for informed consent was waived due to the retrospective nature of the study and minimal risk to the patient. All identifiable information has been removed to protect patient confidentiality.

AI Usage

The AI-based language model, ChatGPT-4, was used for spelling and grammar checking of the final version of the manuscript, including the figure legends. Next-generation sequencing, fluorescence in situ hybridization, and conventional cytogenetic karyotyping were technically performed at a CLIA-certified commercial reference laboratory. The results and their clinical significance were interpreted by pathologists at our institution.

Declaration of Figures' Authenticity

All figures submitted have been created by the authors who confirm that the images are original with no duplication and have not been previously published in whole or in part.

This work is licensed under Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International (CC BY-NC-ND 4.0)

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]

References:

1. Khoury JD, Solary E, Abla O et al. The 5th edition of the World Health Organization Classification of haematolymphoid tumours: myeloid and histiocytic/dendritic neoplasms. Leukemia. 2022;36:1703-19
2. Verstovsek S, Yu J, Scherber RM, et al. Changes in the incidence and overall survival of patients with myeloproliferative neoplasms between 2002 and 2016 in the United States. Leuk Lymphoma. 2022;63(3):694-702
3. Klampfl T, Gisslinger H, Harutyunyan AS, et al. Somatic mutations of calreticulin in myeloproliferative neoplasms. N Engl J Med. 2013;369(25):2379-90
4. Pardanani AD, Levine RL, Lasho T, et al. MPL515 mutations in myeloproliferative and other myeloid disorders: A study of 1182 patients. Blood. 2006;108(10):3472-76
5. Gangat N, Karrar O, Al-Kali A, et al. One thousand patients with essential thrombocythemia: The Mayo Clinic experience. Blood Cancer J. 2024;14(1):11
6. Alvarez-Larran A, Sant'Antonio E, Harrison C, et al. Unmet clinical needs in the management of CALR-mutated essential thrombocythaemia: A consensus-based proposal from the European LeukemiaNet. Lancet Haematol. 2021;8(9):e658-65
7. Al Assaf C, Van Obbergh F, Billiet J, et al. Analysis of phenotype and outcome in essential thrombocythemia with CALR or JAK2 mutations. Haematologica. 2015;100(7):893-97
8. Rumi E, Pietra D, Ferretti V, et al. JAK2 or CALR mutation status defines subtypes of essential thrombocythemia with substantially different clinical course and outcomes. Blood. 2014;123(10):1544-51
9. Tefferi A, Gangat N, Loscocco GG, et al. Essential thrombocythemia: A review. JAMA. 2025;333(8):701-14
10. Bridge WL, Vandenberg CJ, Franklin RJ, Hiom K. The BRIP1 helicase functions independently of BRCA1 in the Fanconi anemia pathway for DNA crosslink repair. Nat Genet. 2005;37(9):953-57
11. Ramus SJ, Song H, Dicks E, et al. Germline mutations in BRIP1, BARD1, PALB2, and NBN genes in women with ovarian cancer. J Natl Cancer Inst. 2015;107(11):djv214
12. Moyer CL, Ivanovich J, Gillespie JL, et al. Rare BRIP1 missense alleles confer risk for ovarian and breast cancer. Cancer Res. 2020;80(4):857-67
13. Daly M, Pal T, AlHilli Z. NCCN Clinical Practice Guidelines in Oncology: Genetic/familial high-risk assessment: breast, ovarian, pancreatic, and prostate. Version 2.2025. Available from: https://www.nccn.org [Accessed March 2026]
14. Sawyer JR, Tricot G, Mattox S, Jagannath S, Barlogie B. Jumping translocations of chromosome 1q in multiple myeloma: Evidence for a mechanism involving decondensation of pericentromeric heterochromatin. Blood. 1998;91(5):1732-41
15. Caramazza D, Hussein K, Siragusa S, et al. Chromosome 1 abnormalities in myeloid malignancies: A literature survey and karyotype-phenotype associations. Eur J Haematol. 2010;84(3):191-200
16. Manola KN, Georgakakos VN, Stavropoulou C, et al. Jumping translocations in hematological malignancies: A cytogenetic study of five cases. Cancer Genet Cytogenet. 2008;187(2):85-94
17. Najfeld V, Tripodi J, Scalise A, et al. Jumping translocations of the long arms of chromosome 1 in myeloid malignancies is associated with a high risk of transformation to acute myeloid leukemia. Br J Haematol. 2010;151(3):288-91
18. Sanford D, DiNardo CD, Tang G, et al. Jumping translocations in myeloid malignancies associated with treatment resistance and poor survival. Clin Lymphoma Myeloma Leuk. 2015;15(9):556-62
19. Couture T, Amato K, DiAdamo A, Li P. Jumping translocations of 1q in myelodysplastic syndrome and acute myeloid leukemia: Report of three cases and review of literature. Case Rep Genet. 2018;2018:8296478
20. Halper-Stromberg E, Stinnett V, Morsberger L, et al. 1q jumping translocation as a biomarker in myeloid malignancy: Frequently mutated genes associated with bad prognosis and low survival. Exp Hematol Oncol. 2024;13(1):73
21. Berger R, Bernard OA. Jumping translocations. Genes Chromosomes Cancer. 2007;46(8):717-23
22. Borri D, Ommati LVM, Velloso EDR. Jumping translocation: An unusual cytogenetic finding in myeloid neoplasm. Hematol Transfus Cell Ther. 2022;44(4):612-13
23. Kamal L, Pierce SB, Canavati C, et al. Helicase-inactivating BRIP1 mutation yields Fanconi anemia with microcephaly and other congenital abnormalities. Cold Spring Harb Mol Case Stud. 2020;6(5):a005652

APPROVED GALLEY PROOF

This work is licensed under Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International (CC BY-NC-ND 4.0)

Indexed in: [PMC] [PubMed] [Emerging Sources Citation Index (ESCI)] [Web of Science by Clarivate]