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LIFE-THREATENING THROMBOTIC EVENTS REVEALING SYSTEMIC LUPUS ERYTHEMATOSUS IN A PATIENT WITH CHRONIC MYELOMONOCYTIC LEUKAEMIA

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ABSTRACT

Associations between systemic autoimmune and inflammatory diseases and myelodysplastic syndromes have been reported for at least 25 years, suggesting a potential pathogenic link.

We present the case of a 62-year-old woman with chronic myelomonocytic leukaemia, initially diagnosed in August 2022 based on persistent monocytosis, thrombocytopenia, polyclonal hypergammaglobulinemia, *NRAS* and *SCMA1* mutations, and cytogenetic abnormalities (46 chromosomes with a long arm deletion of chromosome 16). In August 2024, she was hospitalized for worsening general condition and dyspnoea, revealing hyperleukocytosis (206 g/l), progressive monocytosis, medullary infiltration, and bilateral pericardial/pleural effusions. A positron emission tomography scan demonstrated bone marrow hypermetabolism and splenomegaly. Treatment with azacitidine and venetoclax was initiated, but weeks later, she developed ST elevation myocardial infarction, ischemic stroke, and deep vein thrombosis, requiring intensive care. Immunological testing showed high-titre antinuclear antibodies (1/1280), anti-Ro52, anti-Sm/RNP, and anti-U1 RNP and complement consumption, leading to a diagnosis of systemic lupus erythematosus (/American College of Rheumatology/ European League Against Rheumatism 2019 criteria). Hydroxychloroquine was started, followed by hydroxycarbamide, azacitidine, and ruxolitinib due to persistent hyperleukocytosis (peaking at 100 g/l), resulting in clinical and biological improvement. The patient underwent allogeneic stem cell transplantation in mid-March 2025.

This case highlights the complex interplay between hematologic malignancies and autoimmune diseases, emphasizing the need for multidisciplinary management.

KEYWORDS

Chronic myelomonocytic leukemia (CMML), systemic lupus erythematosus, thrombotic events, azacitidine, venetoclax, allogeneic stem cell transplantation

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LEARNING POINTS

- There is a two-way relationship between hematologic malignancies and autoimmune diseases, where each can potentially initiate or exacerbate the other.
- Effective management requires close collaboration between haematologists, immunologists/rheumatologists, and intensive care specialists.

INTRODUCTION

For at least 25 years, a number of case report and case series have provided evidence of associations between inflammatory diseases, systemic autoimmune diseases, and myelodysplastic syndromes (MDSs), suggesting a possible pathogenic relationship[1-2].

CASE DESCRIPTION

We report the case of a 62-year-old woman with a history of hypertension and osteoarthritis of the hand. Since August 2022, the patient had been managed in our haematology department for persistent monocytosis, thrombocytopenia and polyclonal hypergammaglobulinemia. The diagnosis of atypical chronic myelomonocytic leukaemia (CMML) was based on molecular biology data (*NRAS* and *SCMA1* mutations), a normal karyotype, flow cytometry data (an Ogata score of 2), and cytogenetic findings (46 chromosomes and deletion of the long arm of chromosome 16).

In August 2024, the patient was admitted to hospital due to worsening of her general condition, a World Health Organization score of 2, and New York Heart Association class 2 dyspnoea on exertion. A clinical examination did not evidence lymph node tumour syndrome or organomegaly. We observed hyperleukocytosis 206 g/l, the progression of monocytosis (peaking at 3.5 g/l) and appearance of granulocytes (but not blasts) in the blood, suggesting an atypical myeloproliferative syndrome (MPS/MDS).

A computed tomography (CT) scan of the chest, abdomen and pelvis showed a 25 mm pericardial effusion, bilateral, moderately pleural effusion, presence of several mediastinal and hilar lymph nodes (size <1 cm) and bilateral axillary lymph nodes (size >1 cm). The pleural effusion was exudative, with no signs of infection or cytological abnormalities.

A positron emission tomography scan revealed homogeneous, diffuse, moderate hypermetabolism in the bone marrow of the axial and appendicular skeleton (but with no focal lesions), homogeneous splenomegaly and inversion of the liver-spleen ratio.

Treatment with a combination of azacitidine and venetoclax was initiated in September 2024. The treatment was well tolerated, and the outcome was favourable after a single course. In October 2024, the patient was admitted to hospital for myocardial infarction with ST segment elevation, a right border-zone ischemic stroke and deep vein thrombosis (DVT).

Coronary angiography showed a non-significant long proximal lesion of the right coronary artery, a neighbouring, large thrombus opposite without any flow slowdown, significant stenosis (60-70%) of the ostium of the second diagonal artery *(Fig. 1)*, and a non-significant short lesion of the ostium of the proximal circumflex.

Transthoracic echocardiography showed preserved left ventricular ejection fraction (LVEF) with hypokinesia of the apical two-thirds of the inferior wall of the left ventricle (LV), absence of valvulopathy, and a dry pericardium.

Brain magnetic resonance imaging (MRI) indicated multiple ischemic emboli in the right junctional territory related to recent ischemia *(Fig. 2)*.

Doppler ultrasound of the supra-aortic trunks showed no abnormalities.

Immunological assessment revealed the presence of anti-nuclear antibodies positive at 1/1280, anti-Ro52 and anti-Sm/RNP complex, anti-U1 RNP associated with complement consumption. Conventional anti-phospholipids were negative. Biological assessment did not reveal hyperhomocysteinemia, and the JAK2 mutation was negative. Proteinuria was positive at 526 mg/24h.

Treatment with antiplatelet therapy (clopidogrel and aspirin) and antithrombotic therapy with heparin was initiated. Given the suspicion of connective tissue disease associated with atypical MPS/MDS syndrome, the patient

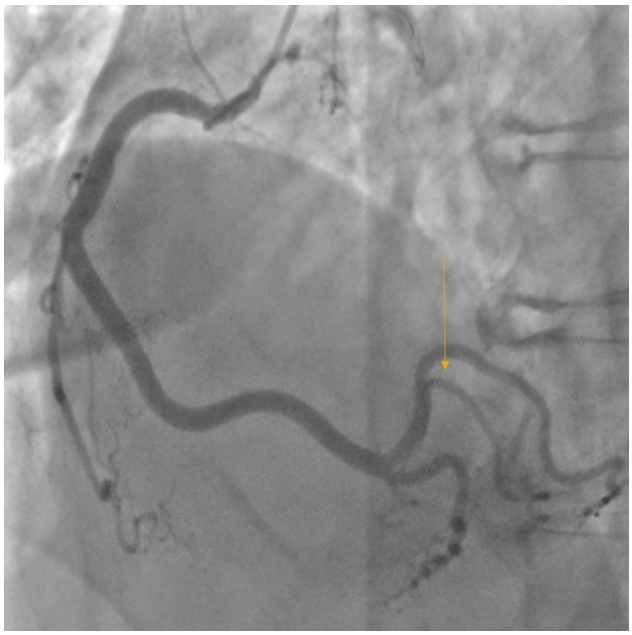


Figure 1. *Coronary angiography (stenose of the ostium of the 2nd diagonal).*

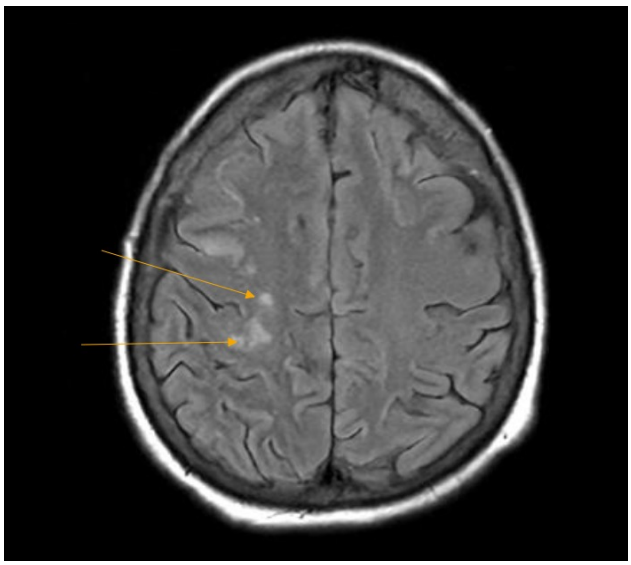


Figure 2. MRI (multiple ischemic emboli).

was transferred to internal medicine for continued care.

During her hospitalization, we observed an increase in hyperleukocytosis (leukocytes up to 100 g/l with major polynucleosis and stable myelemia without circulating blasts). The evolution was favourable under treatment with hydroxycarbamide, subsequently discontinued due to profound thrombocytopenia at 27 g/l with suspension of clopidogrel.

Given the presence of serositis, non-erosive polyarthritis, absence of Raynaud's syndrome, and immunological abnormalities (ANA+, DNA-, Sm-, Sm/RNP+, U1-RNP+, complement consumption), the diagnosis of systemic lupus meeting the American College of Rheumatology/European League Against Rheumatism (ACR/EULAR) 2019 classification criteria was made. The patient then received hydroxychloroquine treatment, which was subsequently suspended.

The patient was transferred to the haematology department for further management. She received treatment with azacitidine 75 mg/m^2 for 7 days and continuous ruxolitinib 10 mg × 2/day then 20 mg × 2/day was started in November 2024. This well-tolerated treatment allowed improvement of the cytopenias. The patient received a total of 5 courses

The patient had a 10/10 photo-identical allograft in March 2025.

Unfortunately, on day 7 post-transplant, the paramedical team found the patient on the floor in cardiorespiratory arrest, with bilateral mydriasis, rigidity, and hypothermia. External cardiac massage was immediately initiated, and the resuscitation team was called, but spontaneous cardiac activity was not restored. No cause of death was identified.

DISCUSSION

Autoimmune or inflammatory manifestations are observed in 10-20% of patients with myelodysplastic syndromes or CMML with a broad clinical spectrum[3]. This clinical case provides a major original contribution to the literature on the association between MDS/CMML and autoimmune diseases. Unlike the large published series[3,4], that primarily document classical autoimmune manifestations or immune thrombocytopenic purpura, our patient presented with an exceptional catastrophic thrombotic triad (ST elevation myocardial infarction [STEMI] + stroke + DVT) revealing a systemic lupus erythematosus like syndrome, meeting the ACR/EULAR 2019 criteria. This severe presentation occurred 2 years after the CMML diagnosis and several weeks after initiating azacitidine-venetoclax therapy, raising important pathophysiological questions about the potential role of clonal evolution and novel therapeutic agents as triggers.

This case significantly expands the known clinical spectrum by documenting an extreme hypercoagulable phenotype not described in existing series, while alerting clinicians to a potentially underestimated thrombotic risk in this population. The polyclonal hypergammaglobulinemia present from the initial diagnosis may represent an early predictive marker warranting enhanced immunological surveillance in these patients.

CONCLUSION

This case illustrates the complexity of atypical CMML, its possible association with connective tissue diseases such as systemic lupus, and the risk of acute thrombotic events. Management required a combined and multidisciplinary therapeutic approach. Allograft represents a potential curative therapeutic option in this context.

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