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CASE REPORT

Clinical Characteristics and Diagnosis of Myelodysplastic Syndrome Accompanied by Eosinophilia and Basophilia

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ABSTRACT

Background: **The aim was to explore the clinical characteristics, diagnostic methods and prognosis of Myelodysplastic Syndromes (MDS) accompanied by eosinophilia and basophilia.**
Methods: **The clinical data of a patient with MDS accompanied by eosinophilia and basophilia were retrospectively analyzed, including the patient's clinical manifestations, blood routine, bone marrow image, flow cytometry, and molecular biological examination results, and discussed in combination with relevant literature.**
Results: **The patient is a middle-aged female who was admitted to the hospital due to fatigue and dizziness for more than two months. The blood routine test indicated pancytopenia, an increased percentage of eosinophils, and an increased percentage of basophils. The bone marrow image showed active proliferation of nucleated cells, obvious pathological hematopoiesis can be seen, and the proportion of eosinophils and basophils increased. Flow cytometry results showed that the proportion of myeloid primitive naive cells increased, the expression of CD34 was enhanced, the expression of CD117 in some cells was weakened, the phenotype was abnormal. The proportion of granulocytes decreased, the proportion of eosinophils and basophils increased, and the expression of CD36 in some erythroid cells was weakened. Molecular biology examination revealed mutations in the IDH1 gene, SRSF2 gene, SETBP1 gene, ETV6 gene, ASXL1 gene and DNMT3A gene. Based on the patient's clinical manifestations and various examination results, the diagnosis was myelodysplastic syndrome with increased eosinophils and basophils. After the corresponding treatment was given, the patient's symptoms were relieved and the blood routine indicators improved compared with before.**
Conclusions: **MDS accompanied by eosinophilia and basophilia is a rare type of leukemia. Clinical manifestations are not specific. Diagnosis requires a combination of blood routine, bone marrow image, flow cytometry, and molecular biological examination. This type of patient has a relatively poor prognosis, especially those with gene mutations. Early and clear diagnosis and the selection of appropriate treatment plans based on the stratification of patients' risk levels, are of great significance for improving the prognosis of patients.**
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KEYWORDS

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INTRODUCTION

MDS is a group of heterogeneous myeloid clonal diseases characterized by bone marrow hematopoietic fail-

Table 1. The detection results of hot spot mutation sites closely related to diseases.

Mutant gene	Chromosome	Mutation location	Nucleotide alteration	Amino acid alteration
IDH1 gene	2q34	exon4	c.394C>T	p.R132C
SRSF2 gene	17q25.1	exon1	c.284C>T	p.P95L
SETBP1 gene	18q12.3	exon4	c.2608G>A	p.G870S
ETV6 gene	12p13.2	exon5	c.811dupA	p.S271Kfs*29
ASXL1 gene	20q11.21	exon13	c.3700C>T	p.Q1234*

Table 2. The detection results of mutation sites that are possibly related to the disease.

Mutant gene	Chromosome	Mutation location	Nucleotide alteration	Amino acid alteration
DNMT3A gene	2p23.3	exon14	c.1598A>G	p.Y533C

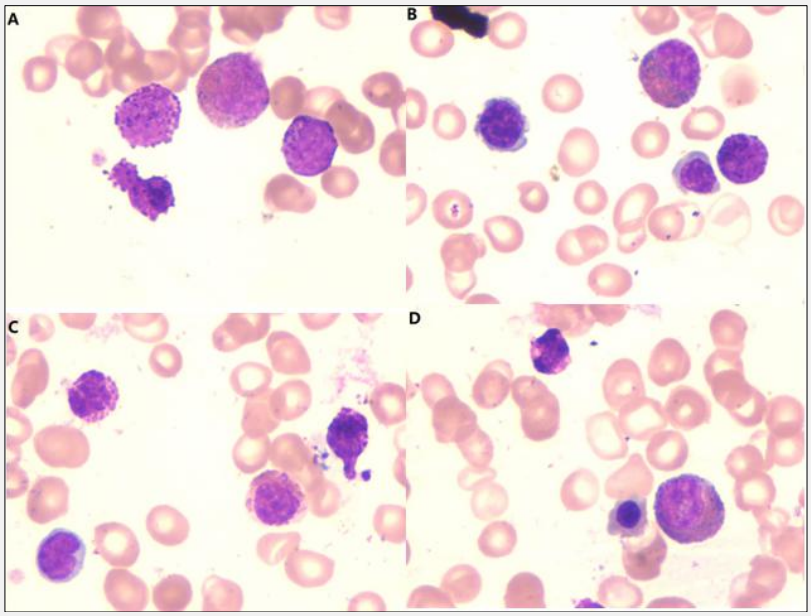

Figure 1. Bone marrow images (Giamsa stain 10*100).

ure, peripheral blood cytopenia, and pathological hematopoiesis of one or more bone marrow series. Some patients are at risk of transforming into acute myeloid leukemia (AML) [1-3]. In the clinical practice of MDS, most patients present with single or multiple series of cytopenia. It is relatively rare for patients to have a significant increase in eosinophils and basophils in peripheral blood and bone marrow simultaneously. The pathogenesis of this condition has not been fully elucidated. It is easily confused with other hematological diseases accompanied by eosinophilic/basophilic cell proliferation, such as chronic myeloid leukemia and idiopathic eosinophilia syndrome, during diagnosis, which brings certain challenges to clinical diagnosis and treatment [4].

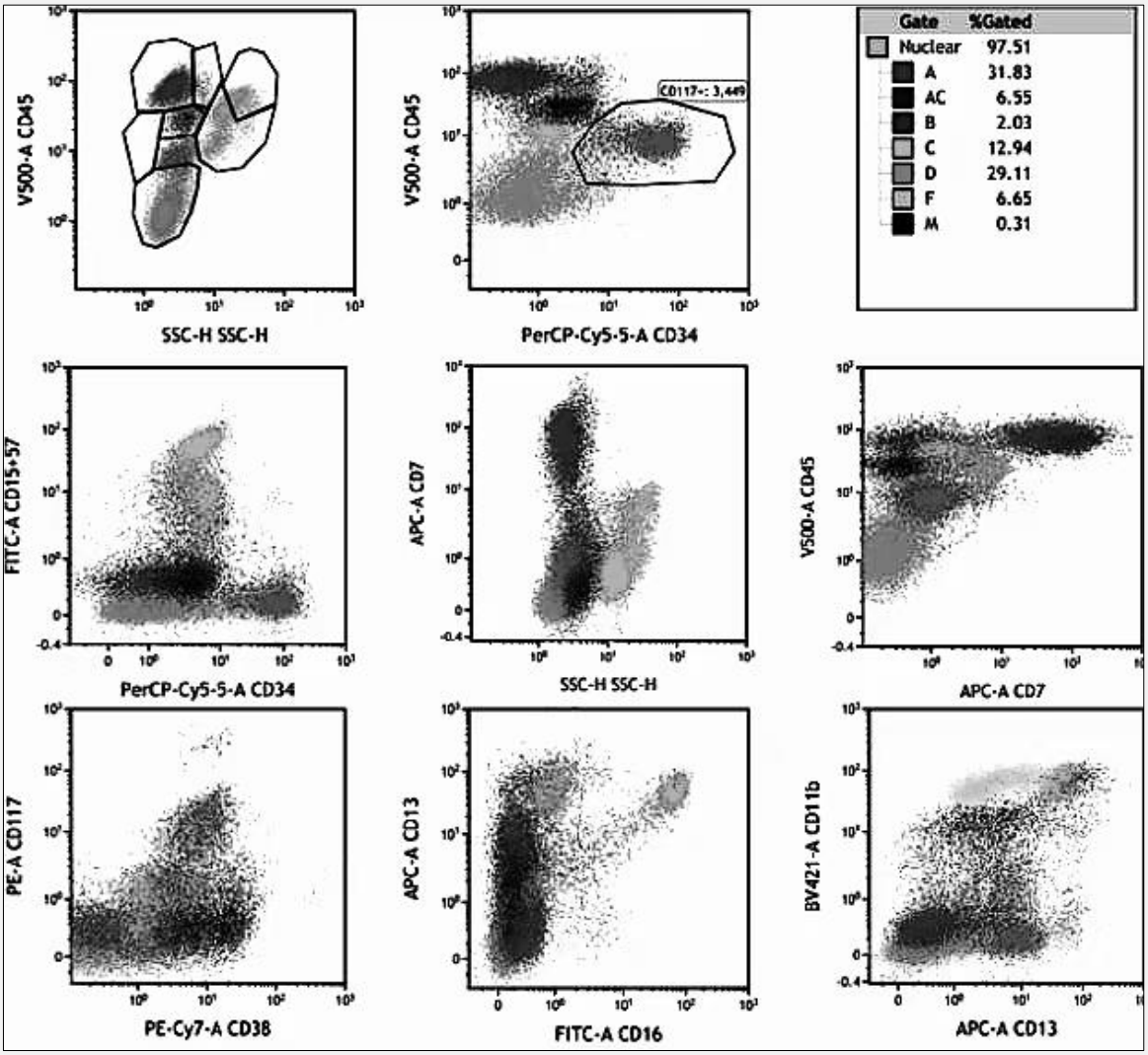

Figure 2. Flow cytometry.

The proportion of myeloid primitive naive cells increases, the expression of CD34 is enhanced, the expression of CD117 in some cells is weakened, the phenotype is abnormal. The proportion of granulocytes decreases, the proportion of eosinophils and basophils increases, and the expression of CD36 in some erythroid cells is weakened.

This article reports a case of a patient who was eventually diagnosed with MDS accompanied by increased eosinophils and basophils, with recurrent dizziness and fatigue as the main manifestations. It elaborates on the clinical features, laboratory test results, diagnostic basis, and treatment response in detail, aiming to provide a reference for the clinical diagnosis and treatment of such rare cases, and at the same time enrich the clinical research data of related diseases.

CASE REPORT

The patient is a woman, 47 years old. Main cause was fatigue and dizziness for more than two months. The patient's skin all over the body is pale yellow, and the lips and eyelids are pale. Blood routine test: WBC: 1.33 x 10^9/L, RBC: 2.13 x 10^{12}/L, Hb: 67g/L, Plt: 11 x 10^9/L. Bone marrow smear: The proportion of primitive cell increase was 13%, eosinophilia was 12.5%, and basophilia was 13.5% (Figure 1). Flow cytometry results show that the proportion of myeloid primitive naive

cells increases, the expression of CD34 is enhanced, the expression of CD117 in some cells is weakened, the phenotype is abnormal. The proportion of granulocytes decreases, the proportion of eosinophils and basophils increases, and the expression of CD36 in some erythroid cells is weakened (Figure 2). Molecular biology examination revealed mutations in the IDH1 gene, SRSF2 gene, SETBP1 gene, ETV6 gene, ASXL1 gene and DNMT3A gene (Table 1 - 2). Diagnosis and treatment process: Upon admission, the patient was scheduled for red blood cell and platelet transfusion, and relevant examinations were completed. The patient had no contraindications to chemotherapy. The patient was given a chemotherapy regimen of azacitistine +CAG, and at the same time, symptomatic and supportive treatments such as antiemetic, liver protection, and stomach protection were provided. The patient's tolerance was acceptable. During the bone marrow suppression period after chemotherapy, the patient was intermittently given symptomatic and supportive treatments such as red blood cell and platelet transfusions. Based on the patient's clinical manifestations and various examination results, the diagnosis was myelodysplastic syndrome with increased eosinophils and basophils. After the corresponding treatment was given, the patient's symptoms were relieved and the blood routine indicators improved compared with before. Blood routine test: WBC: 3.11 x 10^9/L, RBC: 2.23 x 10^{12}/L, Hb: 66g/L, Plt: 44 x 10^9/L. The patient's condition is stable and they have been discharged to recuperate at home.

DISCUSSION

MDS is mainly characterized by single or multiple series of cytopenia and pathological hematopoiesis in the bone marrow. Subtypes accompanied by an increase in both eosinophils and basophils in the peripheral blood and bone marrow are extremely rare. A search of domestic and foreign literature reveals that there are less than 100 such cases reported, accounting for only 0.3% to 0.5% of the total number of MDS cases [5,6]. Among the common subtypes of MDS, eosinophilia or basophilia often occur alone, but in this case, both types of cell proliferation are present simultaneously, which is truly rare. The report of this case further enriches the clinical database of rare MDS subtypes and provides an important example for exploring the clonal evolution patterns of such diseases.

The bone marrow morphology of this case shows significant particularity: The proportion of primitive cell increase was 13%, eosinophilia was 12.5%, and basophilia was 13.5%. The results of flow cytometry in this case further confirmed the diagnosis of Myelodysplastic syndrome accompanied by eosinophilia and basophilia. Molecular biology examination revealed mutations in the IDH1 gene, SRSF2 gene, SETBP1 gene, ETV6 gene, ASXL1 gene and DNMT3A gene. A p.R132C mutation was detected in the IDH1 gene, which is a hotspot mutation. IDH1 mutations are mainly found in AML, MDS, and MPN and are rare in lymphoid tumors. The main mutation site is R132. IDH1 mutation indicates poor OS (Overall Survival) and EFS (Event-Free Survival). A p.P95L mutation was detected in the SRSF2 gene, which is a hot spot mutation. SRSF2 mutations are found in MDS, MDS/MPN, AML, and PMF. SRSF2 mutations occurring in MDS and MF suggest a poor prognosis, shortened OS and EFS, and an increased risk of transformation to AML. It can be used as an independent prognostic indicator. Mutations in SRSF2 in MDS are often accompanied by mutations in genes such as RUNX1, IDH1, IDH2, and ASXL1. A p.G870S mutation was detected in the SETBP1 gene, which is a hotspot mutation. In MDS, patients with SETBP1 mutations have a shortened OS. A p.S271Kfs*29 mutation was detected in the ETV6 gene, which is a truncated mutation. ETV6 is a tumor suppressor gene. The survival rate of MDS patients with ETV6 mutations is reduced. A p.Q1234* mutation was detected in the ASXL1 gene, which is a truncated mutation. Literature reports that ASXL1 mutations in myeloid tumors such as AML, MDS, MDS/MPN, and MPN all indicate poor chemotherapy efficacy and poor prognosis. They can be used as an independent factor for poor prognosis in patients with MDS and PMF. A p.Y533C mutation was detected in the DNMT3A gene, which was predicted to be a somatic mutation. In MDS patients, this gene mutation indicates a poor prognosis and an increased risk of transformation to AML [7-9]. In the future, it is necessary to clarify the mutation spectrum through gene sequencing to guide individualized treatment [10].

In conclusion, the rarity of this case, as well as the particularity of its morphological and flow cytometry features, provide a new perspective for the study of heterogeneity in MDS. In clinical diagnosis, it is necessary to combine multi-dimensional detection including morphology, immunology and cytogenetics to avoid missed diagnosis or misdiagnosis. In terms of treatment, demethylating drugs may be an effective option, but the treatment plan needs to be optimized in combination with the results of gene mutations.

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Declaration of Interest:
All authors declare: 1. All views and data in this paper are supported by references and data. The manuscript has not been published before and is not being considered for publication elsewhere. 2. All authors have contributed to the creation of this manuscript for important intellectual content and read and approved the final manuscript. We declare there is no conflict of interest. 3. This paper is published with the consent of patients, in line with ethical requirements.

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