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Case report/Kazuistyka

The patient with 5q minus syndrome and *JAK2* V617F mutation with the presence of ringed erythroblasts meeting the criteria of RARS-T effectively treated with lenalidomide – A case report

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ABSTRACT

5q minus syndrome is a form of myelodysplastic syndrome characterized by the presence of an isolated deletion of long arm of the chromosome 5. Patients with 5q minus respond well to the treatment with lenalidomide. The presence of the *JAK2* V617F mutation is a common feature of refractory anemia with ring sideroblasts and marked thrombocytosis. Much less is known about effectiveness of lenalidomide in these patients. We present the patient with 5q minus syndrome and *JAK2* V617F mutation accompanied by the presence of ringed erythroblasts meeting the criteria of RARS-T. We could identify only two such patients reported in the literature; no details were given about effectiveness of lenalidomide in that group. We observed good response to the treatment with lenalidomide with transfusion independence 9 months after starting of the treatment; however, there was no complete eradication of del (5)(q13q31) clone nor the clone with *JAK* V617F mutation.

Introduction

5q minus syndrome (5q minus) is a form of myelodysplastic syndrome (MDS) characterized by the presence of an isolated deletion of long arm of the chromosome 5 [1]. The disease has a relatively good prognosis, the overall survival rate is estimated at approximately 145 months and the risk of transformation to acute myeloid leukemia is less than 10% [2, 3]. In a small percentage of cases, patients with

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5q- syndrome may exhibit *JAK2* V617F mutation [4]. The authors of the fourth edition of WHO Classification of Tumors of Hematopoietic and Lymphoid Tissues (WHO 2008) separated *JAK2* V617F mutated form that emerged from the typical 5q minus syndrome. In their opinion, there is not enough data collected for accurate classification of this subgroup. It was decided that temporarily the diagnosis should be classified as MDS, not as a myeloproliferative disease, with the addition of information about the presence of the *JAK2* V617F mutation [5, 6]. On the other hand, the presence of the *JAK2* V617F mutation is a common feature of refractory anemia with ring sideroblasts and marked thrombocytosis (RARS-T), which was included into "Myelodysplastic/myeloproliferative neoplasm, unclassifiable" as a provisional entity grouping together patients with the diseases having features of myelodysplastic and myeloproliferative disease [7, 8]. Patients with 5q minus syndrome usually respond well to the treatment with lenalidomide [9–11]. Much less is known about its effectiveness in the patients meeting the criteria of RARS-T [12, 13]. In this paper, we present the case of a patient with 5q minus syndrome with *JAK2* V617F mutation features and ringed erythroblasts accompanied by thrombocytosis effectively treated with lenalidomide.

A case report

A 59-year-old female patient was admitted to the department of hematology due to persistent macrocytic anemia with hemoglobin 9.9 g/dl (mean cell volume – MCV – 104.3 fl), white cell count 9.50 G/l, and with platelet count 944 × 10^9/l. Before she was referred to a hematologist, she had been unsuccessfully treated with vitamin B12. At the time of her first presentation, the bone marrow showed 60% cellularity with numerous dysplastic megakaryocytes presenting large hypolobated nuclei and with features of erythroid dysplasia (Fig. 1). Ringed sideroblasts accounted for 18% of her erythroid precursors (Fig. 2). Blast cells expressing CD34 antigen accounted for approximately 3% of

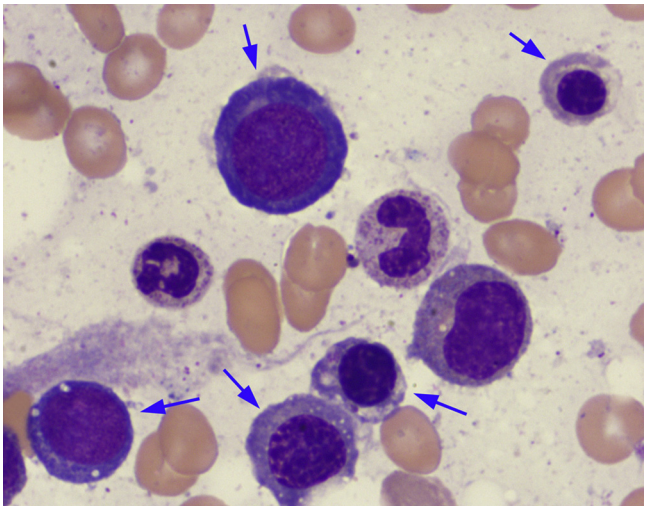

Fig. 1 – Five erythroblasts (arrows), at different stage of maturation, show dysplastic features (megaloid changes, cytoplasmic vacuoles, irregular shape of nucleus, abnormal hemoglobinisation)

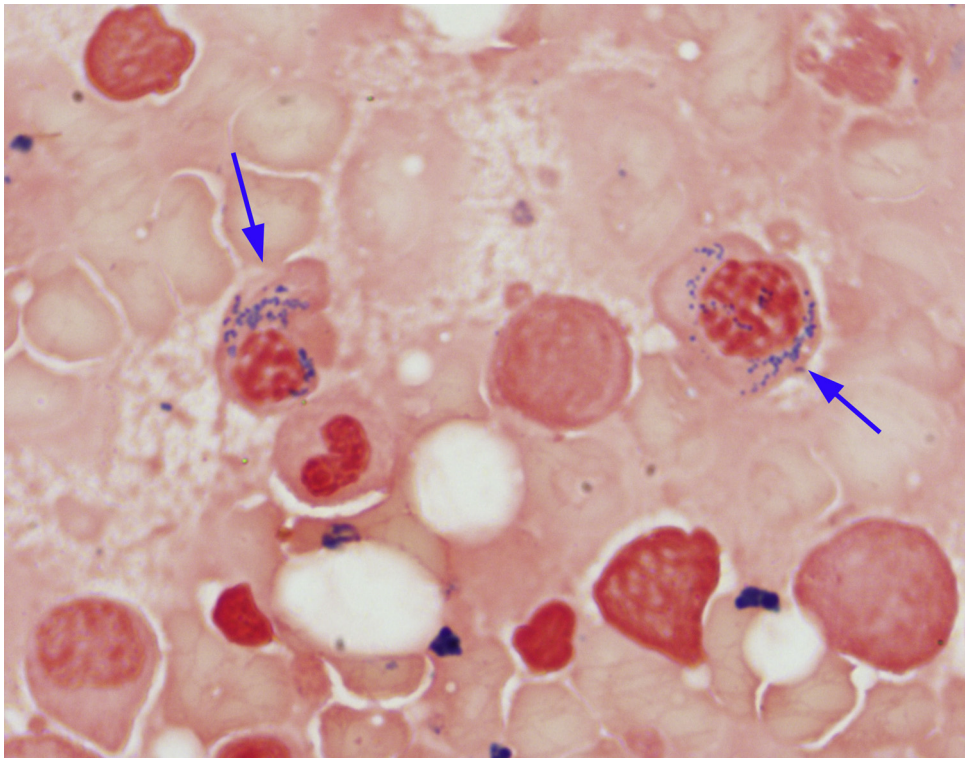

Fig. 2 – Two ringed sideroblasts (arrows)