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Case Report

A Rare Presentation of Myelodysplasia

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Abstract

Myelodysplasia usually results in chronic anemia, but can occasionally present with an isolated decrease in other lineages. We report a case in which the manifestation was an isolated thrombocytopenia with cytogenetic and morphologic abnormality.

Key-words: Isolated thrombocytopenia, MDS- unclassified, trisomy 8

Introduction

The myelodysplasia (MDS) encompasses a series of haematologic conditions characterised by chronic cytopenia(anemia, neutropenia, thrombocytopenia) accompanied by abnormal cellular maturation. As a result, patients with MDS are at risk of symptomatic anemia, bleeding and infection, as well as progression to AML, which is often refractory to standard treatment. We report a case where the patient presented with peri anal abscess and was detected to have very low platelet count, and a work up revealed MDS.

Case History

A 26 year old male was admitted in the department of surgery with pain and swelling in the peri anal region of 5 days duration. The patient's vitals were normal. He was not a diabetic. His routine blood reports revealed a very low platelet count of 25000/ cu.mm. However, he did not have any bleeding from mucosal sites. Haemoglobin levels, total count and differential counts were within normal limits. Serum creatinine, LFT, serum electrolytes, chest X ray and USG abdomen and LDH levels were also normal. Dengue serology and smear for malarial parasite were negative. Peripheral smear showed macrocytic anemia with thrombocytopenia. Vitamin B_{12} level was 370pg/ml(211-911 pg/ml). Since the patient was asymptomatic, and without any significant past medical illness and drug history a working diagnosis of ITP was made. Meanwhile the patient was transfused with platelets and peri anal abscess drained when the platelet count was 75000/cu.mm. Anti-platelet ab was negative and bone marrow examination revealed hypercellularity and tri linear hyperplasia. Megakaryocytes were markedly increased with atypia, including micro and macro megakaryocytes, consistent with MDS-unclassified. Chromosomal analysis revealed trisomy 8 which is commonly observed in MDS/MDS-AML with intermediate prognosis. On follow up oftes 2 weeks the platelets count was 50000/cu.mm.

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Discussion

MDS are a group of heterogeneous, hematological malignancies characterised by ineffective hematopoeisis of one or more cell lines with potential to progress to acute leukaemia. MDS diagnosis is usually correlated clinically with refractory anemia. Although other cytopenias are also commonly seen in MDS, isolated thrombocytopenia is, with an incidence estimated at 1-4%.[1] The patient presented with peri anal abscess and was incidentally found to have thrombocytopenia. The born marrow examination features are suggestive of MDS-U, as per WHO classification of myeloid neoplasms.[2,3] Chromosomal analysis showed trisomy 8. Misdiagnosis of ITP is possible in the context of megakaryocytic hyperplasia without identified features of dysplasia and without documented clonal cytogenetics. In order to establish a diagnosis, a careful assessment of potential dysplastic features, as well as cytogenetic studies, should be performed at presentation.[4]

In conclusion, myelodysplasia can occasionally present with isolated thrombocytopenia. After ruling out secondary etiologies, a careful bone marrow examination to look for dysplastic morphological changes has to be done. However, in the absence of proven clonality by cytogenetics or other molecular studies, a definitive diagnosis of myelodysplasia is often difficult to establish.

References

1. Menke DM, Otero GC, Cockerill KJ, Jenkins RB, Noel P, Pierr RV. Refractory thrombocytopenia: a myelodysplastic syndrome that may mimic immune thrombocytopenic purpura, Am. J. Clin. Pathol. 1992; 98: 502-10.

2. Brezinova J, Zemanova Z, Ransdorfova S, Sindelarova L, Siskova M, Neuwirtova R, Cermak J Michalova K. Prognostic significance of del (20q) in patients with hematological malignancies Cancer Genet. Cytogenet. 2005; 160: 188-92.

3. Vardiman JW, Harris NL Brunning RD. WHO classification of myeloid neoplasms Blood. 2002; 100: 2292-2302.

4. Sashida G1,Takaku TI,Shoji N,Nishimaki J,Ito Y,Miyazawa K, Kimura Y,Ohyashiki JH, Ohyashiki K. Clinico-hematologic features of myelodysplastic syndrome presenting as isolated thrombocytopenia: an entity with a relatively favourable prognosis. Leuk Lymphoma.2003; (4):653-58.