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CASE REPORT

Unusual Presentation Of Pediatric Myelodysplastic Syndrome: A Case Report

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ABSTRACT

Myelodysplastic syndrome (MDS) is a clonal hematological disorder characterized by dysplastic hematopoiesis and progression to leukemia. MDS is a rare childhood disorder presenting with nonspecific symptoms and has extremely variable prognosis. It should be considered in the differential diagnosis of all cytopenic disorders in children. The preleukemic phase is usually short and the disease rapidly evolves into overt leukemia. Marrow cellularity is normal or increased in MDS with hypocellular marrow being a rare feature. We report a case of MDS in a child with the rare finding of hypocellular bone marrow for that age.

Keywords: Myelodysplastic syndrome; cellularity; children

Introduction

Myelodysplastic syndrome (MDS) is a clonal hematopoietic stem cell disorder characterized by peripheral cytopenia(s), bone marrow dysplasia and susceptibility to acute leukemia [1]. MDS is rare in childhood [2],[3]. The disease seems to be more frequent in patients who previously received radiotherapy and chemotherapy for a first malignancy [4],[5]. Bone Marrow cellularity is normal or increased in MDS with hypocellular marrow being a rare feature[7]. This case highlights a rare feature of MDS with hypocellular bone marrow in a child.

Case report

A 7 year old boy presented with low grade fever, easy fatigability, and pallor of three months duration which was treated symptomatically considering the diagnosis to be nutritional anemia. General physical examination was unremarkable except for marked pallor with no hepatosplenomegaly or lymphadenopathy. Past history was non contributory with no prior exposure to cytotoxic drugs, radiation or other mutagens.

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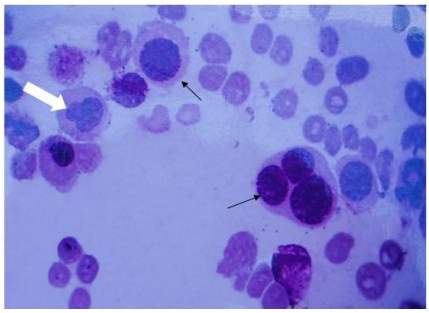
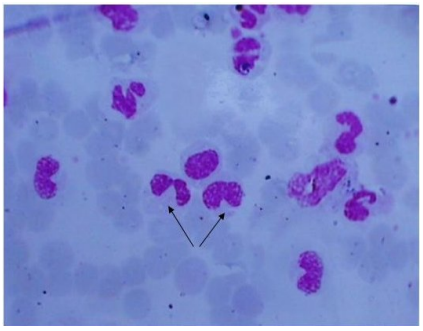
Laboratory picture showed pancytopenia, with hemoglobin 4.7g/dl, hematocrit 14.3%, mean corpuscular volume (MCV) 63 fl, total leukocyte count (TLC) 3.8 X 10^9 /l, with neutrophils 0.8 X10^9 /l, platelet count 75 X 10^9 /l and the proportional circulating blasts were 1%. Morphological diagnosis was based on analysis of peripheral blood smear (PBS), bone marrow aspirate (BMA) and bone marrow biopsy (BMB) with cytochemical staining with periodic acid Schiff (PAS) and Perl's stains. PBS and BMA showed trilineage dysplasia. Dyserythropoiesis was evident principally by macrovalocytes, multinuclearity and nuclear hyperlobation [Tab/Fig 1]. Dysgranulopoiesis was characterized by Pseudo-Pelger-Huët anomaly and hypogranularity [Tab/Fig 2]. Megakaryocytic dysplasia was characterized by hypolobated megakaryocytes of all sizes and hyperlobated widely separated nuclei. [Tab/Fig 3],[Tab/Fig 4]. Perl's reaction on aspirated marrow showed erythrocytes at all stages of maturity loaded with siderotic granules. BMB indicated a hypocellular bone marrow for that age with characteristic trilineage dysplasia. There was no abnormal localization of immature precursors (ALIP) nor was the topography of the bone marrow distorted. However cytogenetic analysis could not be done because of financial constraints. The case was

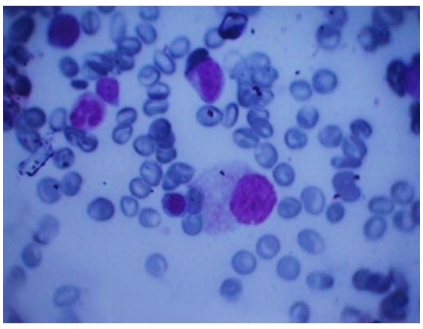
diagnosed as MDS with refractory cytopenia [MDS RC] in accordance with the WHO criteria modified for pediatric age group. The patient received supportive treatment consisting of folate and B12 and blood transfusions. The patient could be followed up to a period of approximately 2 months with routine blood tests to evaluate the hematological outcome of the disease. The patient did not transform to a more aggressive form of MDS or leukemia and was lost to follow up in a satisfactory clinical condition.



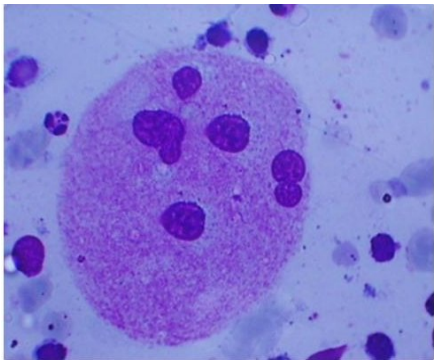
Table/Fig 1:BMA showing megaloblastoid features, multinuclearity and nuclear budding (MGG× 1000)



Table/Fig 2 :BMA showing pseudo-pelger-Huët anomaly (MGG× 1000)



Table/Fig 3 BMA showing agranular micromegakaryocyte (MGG× 1000)



Table/Fig 4: BMA showing hyperlobated megakaryocyte (MGG× 1000)

Discussion

MDS is a rare hematological disorder in children. The symptoms are non specific and the diagnosis may be difficult to make. Prognosis is extremely variable depending on the type of MDS. The pre leukemic phase is usually short in children [12 – 18 months] and the disease rapidly evolves into overt leukemia. This case highlights two uncommon features: First, being the patient's age as the term pediatric MDS is used for < 17 years age considering the latest proposed WHO classification. Second unusual finding is the hypocellularity of the bone marrow for the age of the patient. Bone marrow hypocellularity is a rare feature in MDS [6],[7]. Hypocellularity on BMB is defined when it is below the normal range value adjusted for the age on a semi-quantitative evaluation [8]. Bone marrow cellularity is the critical determinant for the recognition of hypocellular MDS [9]. According to the pediatric modification of the WHO classification [10], MDS patients are classified into three groups: refractory cytopenia (RC), refractory anemia with excess of blasts (RAEB) and RAEB in transformation (RAEB-T). Applying the recently proposed iagnostic criteria for detecting MDS in children, this patient fulfilled at least two of the minimal diagnostic criteria for pediatric MDS as suggested by Hasley et al [10]: sustained unexplained cytopenia and/or at least bilineage morphological myelodysplasia and/or acquired clonal cytogenetic abnormality in hematopoietic cells and/or increased blasts > 5%. The differential diagnosis considered in this case was hypocellular acute myeloid leukemia (AML) with low blast count, aplastic anemia and several congenital bone marrow failure syndromes. The diagnosis was

established by a combination of clinical and laboratory parameters along with BMA and BMB pictures which showed trilineage dysplasia.The diagnosis of hypocellular MDS without excess blasts is to be made with caution especially in the absence of cytogenetics.The sparse number of cells available for evaluation and the subjective grading of qualitative abnormalities account for the diagnostic difficulties. It would have been difficult to diagnose if a monolineage dysplasia was present.

Key message

MDS should be considered in the differential diagnosis of all cytopenic disorders in children. It is important to diagnose MDS at an early stage by critical evaluation of dysplasia particularly in pediatric patients as preleukemic phase is usually short in children and the disease rapidly evolves into overt leukemia.

Conflict of Interest: None declared

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