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A RARE CASE OF MDS WITH 11Q 23 CHROMOSOME DELETION IN A 34 YEAR OLD FEMALE

General Medicine

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ABSTRACT

The myelodysplasias (MDS) are a heterogenous group of hematologic disorders broadly characterised by cytopenias associated with a dysmorphic and usually cellular bone marrow and by consequent ineffective red cell production. Idiopathic MDS is a disease of the elderly, with a slight male preponderance.(1) Cytogenetic abnormalities are found in half of the patients – Loss of all or part of 5,7,20 and trisomy 8 or 11 q 23 deletion following topoisomerase II inhibitors. Most hematological neoplasms with 11q23 rearrangement have been cases of acute

KEYWORDS

MDS, cytopenias

INTRODUCTION

Terminal/subtelomeric/telomeric and interstial deletions of the long arm of chromosome 11[del(11q)] as the only abnormality or as part of a non-complex karyotype are not common in myelodysplastic syndromes (MDS), and currently grouped with other miscellaneous single chromosomal aberrations within the cytogenetic intermediate-risk group according to the International Prognostic Scoring System (IPSS) [1].

In the literature, these cases are generally reported either as a low frequency cytogenetic abnormality (1–5%) in MDS patients in large study cohorts [2], [3], [4], [5]; or reported as one of the "miscellaneous hematological malignancies" with deletions of 11q [6], [7].

A few early case studies described some of the clinicopathologic aspects of this particular alteration in MDS; however, the cases were classified using the French–American–British (FAB) system and cases with del(11q) in a complex karyotype were included. Bain et al. reported 28 cases MDS with abnormalities of 11q23, of which, 12 of which showed deletions of 11q of various lengths. These patients were elderly with age over 50 years, presented with a low white blood cell count (WBC), and had an overall survival (OS) of 19 months [6]. However, the 12 cases reported by Bain and colleagues included refractory anemia with excess blasts in transformation (RAEB-t), unknown MDS subtypes, pediatric MDS, and cases with del(11q) as part of a complex karyotype. series that del(11q) in MDS may be Rearrangements at 11q23 are commonly seen in AML or therapy-related myeloid neoplasms . Many of these rearrangements involve MLL, either through translocations of MLL N-terminus in frame with more than 60 translocation partner genes, or through partial tandem duplications (PTD) . Deletions of 11q close to the telomere, including the region of 11q23, are commonly observed in acute myeloid leukemia (AML). ,. Thus, it is recommended that the presence of del(11)(q23) should be an indication for additional FISH and molecular investigations to exclude or verify the involvement of MLL translocations.

Case Report

A 34 year old female came with the c/o easy fatigueabilty in the form of exertional breathlessness which was progressively increasing in nature and associated with palpitations, there was history of yellowish discoloration of the eyes since 2 months with no history suggestive of hepatic etiology. There was also no history of bleeding tendencies or menorraghia

Findings : Investigations
Hb- 5.3 g/dl
TC - 5130 Plt-96,000 cells
MCV -104
Total bilirubin - 2.5
Indirect bilirubin -2.1
Vitamin b12 levels - normal
Peripheral smear - normocytic normochromic anaemia with thrombocytopenia, reticulocyte count-12%
No schizocytes / fragmented rbcs

Bone marrow aspiration - erythroid hyperplasia with normoblastic picture amd megaloblastoid change. Karyotyping- 11 q 23 deletion coombs test -Positive - Investigations and clinical features were suggestive of autoimmune haemolytic anaemia - Bone marrow examination showed features of MDS- U .

Karyotyping - shows deletion of 11q23 chromosome

CONCLUSIONS

Autoimmune hemolytic anaemia is a rare but commonly overlooked cause of anaemia in patients with MDS. Genetic studies are helpful in ruling out myelodysplastic syndrome.

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