Open Journal of
Clinical & Medical
Case Reports

Volume 4 (2018)
Issue 11

ISSN 2379-1039

Amyloidosis involving bone marrow without monoclonal gammopathy: A rare entity

Shahzad Ali Jiskani*; Humaira Rizwan; Lubna Naseem

***Shahzad Ali Jiskani**

Department of Pathology, Pakistan Institute of Medical Sciences, Islamabad
Email: Shahzadbaloach289@gmail.com

Abstract

Amyloidosis is extracellular deposition of fibrillar proteins in various tissues of body. Bone marrow involvement is common and presents as multiple myeloma. We present case of 54 year old male who present with infiltration of amyloidosis in bone marrow but without any monoclonal gammopathy having no evidence of plasma cells on bone marrow aspirate or trephine biopsy, normal protein and urine electrophoresis.

Keywords

amyloidosis; bone marrow; monoclonal gammopathy; multiple myeloma

Case Report

A 54 year old male presented to Department of Medicine with complaints of easy fatigability, myalgia and lower limb weakness since 4 months. There were no any other active complaints. He was smoker since 30 years. He had myocardial infarction 6 years ago. Patient had history of vertigo, syncope attacks, weakness in both legs and myalgia 4 years ago. Patient was labeled as chronic inflammatory demyelinating polyradiculopathy with vitamin B12 deficiency. Patient was given vitamin B12 therapy but did not show any response to the treatment. Family history was not significant. Patient did not get any transfusion for blood products. Patient was treated with steroids, heparin and vitamin B12 therapy recently.

On general physical examination, he was pallor. There were no any signs of jaundice, koilonychia or clubbing. Lymph nodes were not palpable. Liver was mildly elevated (1cm). Spleen was not palpable.

On laboratory investigations, his white blood cells was 7.6 X 10^9/L (4 – 10 X 10^9/L), red blood cell count 4.44 million/µL (4.5 – 5.5 million/µL), hemoglobin 11.1g/dL (13 – 17 g/dL), hematocrit 38.5% (40–50 %), mean corpuscular volume (MCV) 86.7 fL (81 – 95 fL), Mean Corpuscular Hemoglobin (MCH) 29.3 pg (27-32 pg), Mean Corpuscular Hemoglobin Concentration (MCHC) 33.8 g/dL (31.5 – 34.5 g/dL), platelet count 245 X 1000/µL (150 – 410 X 1000/µL) and Red cell Distribution Width (RDW) of 12.4% (11 – 14%). His peripheral film was normocytic normochromic showing neutrophils (47%), lymphocytes (45%), monocytes (7%) and eosinophil (1%). His reticulocyte count was 2.0 % (0–2%). There was no evidence of any atypical cells. His Erythrocyte Sedimentation Rate (ESR) was 8 mm/hr (0 - 22 mm/hr). His Prothrombin Time (PT) and Activated Partial Thromboplastin Time (APTT) were 11

seconds and 40 seconds respectively. His International Normalized Ratio (INR) was 0.99. Serum biochemistry was done. His ALT was 22 U/L(7 – 55 U/L), AST 36 U/L (8 – 48 U/L), alkaline phosphatase 94 U/L (45 – 115 U/L), serum albumin 3.9 g/dL (3.5 – 5 g/dL), total protein 7.1 g/dL (6.3 – 7.9 g/dL), serum bilirubin 0.5 mg/dL (0.1 – 1.2 mg/dL), serum urea 31 mg/dL (10 – 50 mg/dL), serum creatinine 0.9 mg/dL (0.5 – 1.4 mg/dL), serum uric acid 3.5 mg/dL (3.4 – 7.0 mg/dL), serum sodium 141 mmol/L (135 – 155 mmol/L), serum potassium 3.8 mmol/L (3.5 – 5.5 mmol/L), serum calcium 9.1 mg/dL (8.5 – 10.2 mg/dL), thyroid stimulating hormone 20 µIU/mL (9 – 30 µIU/mL), free T3 412 pg/d (230 – 619 pg/d) and T4 7.6 µg/dL (4.6 – 12 µg/dL). His serum Prostate Specific Antigen (PSA) level was 0.21 ng/mL (<4.0 ng/mL). His vitamin D level was 11 ng/mL (20 – 50 ng/mL). His serum folate level was 17 ng/mL (2 – 20 ng/mL) and serum vitamin B12 level was 393 pg/mL (200 – 500 pg/mL). His 24 hour urinary protein level was 901 mg/day (<100 mg/day). His urinary protein electrophoresis was normal showing selective proteinuria, although there was no evidence of any adventitious band. His serum protein electrophoresis was also normal. His echocardiogaph showed evidence of amyloid deposits.

His bone marrow biopsy was done. Bone marrow aspirate was moderate cellular. Erythropoiesis was moderate with normal maturation. Myelopoiesis was moderate showing all stages of cells. Megakaryocytes were adequate. Lymphocytes and histiocytes were normal on aspirate. Plasma cells were not increased on aspirate. There was no evidence of atypical cells on bone marrow aspirate. Histological sections of trephine biopsy showed portion of bone marrow with depressed erythroid, myloid and megakaryocytic series cells. There was an area of hyalinization suggesting extracellular deposits. So Congo Red Stain was applied on specimen and was observed under polarized microscope, which showed apple–green birefringence areas upon contacting with polarized light (Figure 1, 2).

Discussion

Amyloidosis is a heterogeneous group of diseases which occurs due to extracellular accumulation of autologous fibrillar proteins, which are arranged into 3 – dimensional β – plated form which disturbs normal function of organs [1,2]. Amyloid can be found in almost any part of the body. Amyloid is localized if amyloid fibrils are present in one organ. Although, if multiple organs are involved then it is called systemic amyloidosis. Amyloid is classified into various categories such as light chain (AL), Amyloid – Associated protein (AA) or Familial (AF) [3]. Some cases are found to be familial as there are involvement of genetic mutations that affect production of amyloid precursor protein and ultimately enhance the production of amyloid fibrils which are insoluble [4]. Deposition of amyloid in bone marrow is common in patients having AL amyloidosis [5]. Amyloidosis involving bone marrow often present as plasma cell dyscrasia. Bone marrow aspirate and trephine examination in these cases is performed to see the infiltration of plasma cells in bone marrow [6].

This patient was found to have bone marrow involvement of amyloid, but there was no evidence of plasma cell dyscrais on bone marrow aspirate or trephine along with normal serum and urine protein electrophoresis. Saoji et al reported two cases in India having amyloid present in bone marrow with no infiltration of plasma cells and with normal urine and protein electrophphoresis [7]. There were no other cases reported with this condition. Amyloid involving bone marrow can also be present as various unusual presentations. Dwivedi et al reported a case of primary systemic amyloidosis in India, having waxy lesions in periorbital area associated with macroglossia. Serum electrophoresis protein was

normal but bone marrow showed increase in the number of plasma cells with prominent binucleated and immature plasma cells [8].

Conclusion

Bone marrow being common involvement in primary AL amyloidosis, can present with atypical presentations. Noninvasive investigations cannot disclose full picture of organ damaging diseases like amyloidosis. Rare entities should be in mind while evaluating the patient.

Figures

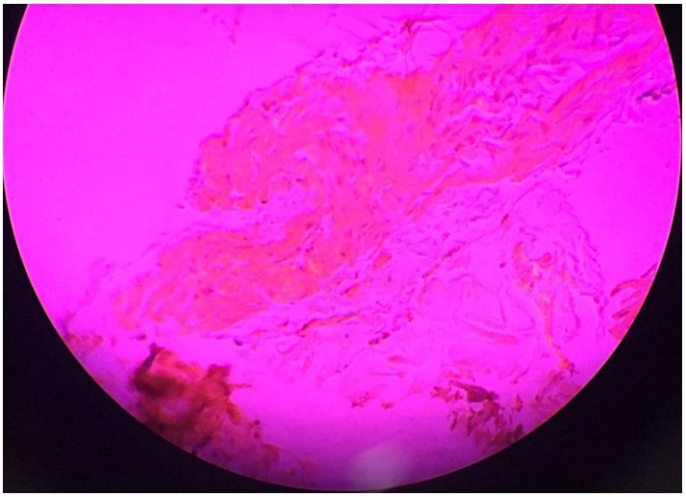

Figure 1: Specimen of bone marrow trephine stained with Congo Red Stain showing area of hyalinization

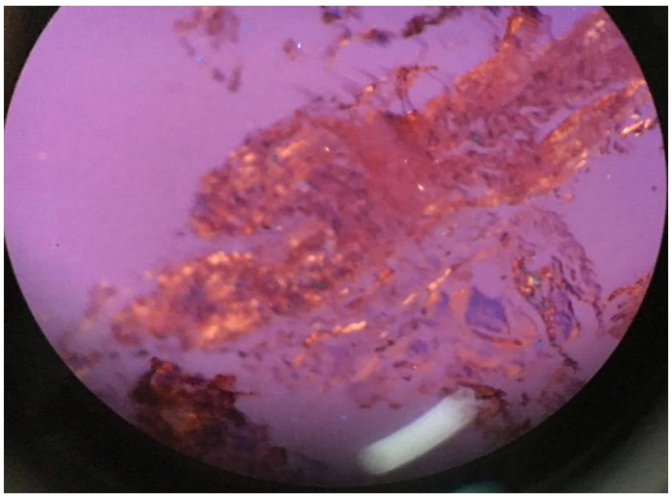

Figure 2: Specimen of bone marrow trephine showing apple - green Birefringence areas upon contacting with polarized light

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Manuscript Information: Received: May 02, 2018; Accepted: June 12, 2018; Published: June 15, 2018

Authors Information: Shahzad Ali Jiskani*; Humaira Rizwan; Lubna Naseem

Department of Pathology, Pakistan Institute of Medical Sciences, Islamabad, Pakistan

Citation: Jiskani SA, Rizwan H, Naseem L. Amyloidosis involving bone marrow without monoclonal gammopathy: A rare entity. Open J Clin Med Case Rep. 2018; 1425.

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