Clinical Case Reports: Open Access

Yumed Text
OPEN ACCESS JOURNALS

Case Report | Vol 6 Iss 1
ISSN: 2582-5038
https://dx.doi.org/10.46527/2582-5038.243

Pancytopenia as a Presentation of B12 Deficiency

Prinink Purushothaman[1*], Rijas K[1] and Kavya Ravindran[2]

[1]Department of Internal Medicine, Government Medical College Kozhikode, India

[2]Department of Ophthalmology, Government Medical College Kozhikode, India

[*]**Corresponding author:** Prinink Purushothaman, Department of Internal Medicine, Government Medical College Kozhikode, India, Tel: 91-495-2350216; E-mail: prininkroolz@gmail.com

Received: January 17, 2023; **Accepted:** January 28, 2023; **Published:** February 06, 2023

Abstract

We present the case of a 16-year-old boy with hemolytic anemia with investigations showing pancytopenia, which turned out to be due to B12 deficiency. Whenever we see a teenager presenting with pancytopenia the common causes, we think of are hemaltologic malignancies, Drug induced cytopenias, hypersplenism etc. Anemia as a presentation is common in B12 deficiency, yet it is presenting with jaundice is a very uncommon and easily overlooked presentation, same is the case of B12 presenting as pancytopenia which is a well described entity yet omitted initially owing to its uncommonality in this modern developed world.

1. Introduction

Cobalamin deficiency is known to cause megaloblastic anemia and a neurologic affliction in the form of peripheral neuropathy, spastic paralysis with ataxia dementia, psychosis, or a combination of many of these features [1]. Minimal cobalamin deficiency, which may manifest as neurologic symptoms without anemia, seen mainly among the elderly. Vitamin B12 deficiency is a rare cause of hemolytic anemia (approximately 1.5% of cases) [2-3]. Severe hemolytic anemia occurs due to increased folate demand for augmented erythropoiesis and thus may resemble an immune cytopenia also its thought to be related to elevated levels of homocysteine which build up as homocysteine cannot be converted to methionine. In vitro homocysteine was found to cause hemolysis by possible lipid peroxidation and cytoskeletal protein derangement [4].

2. Case Report

Apparently normal 16-year-old teenager presented to the OPD with complaints of easy fatiguability and dyspnoea on exertion which started and progressed over 1 month. There was no history of any palpitations, resting dyspnoea, no decreased appetite with undue weight gain and constipation, decreased urine output, cough with expectoration, or orthopnea. He also started

Citation: Purushothaman P, Rijas K, Ravindran K. Pancytopenia as a Presentation of B12 Deficiency. Clin Case Rep Open Access. 2023;6(1):243.

noticing gradually progressive yellow discoloration of his eyes over 1 week. There was no history of any fever, no development of edema, drug intake. On examination he was conscious, oriented with stable vitals, he was pale with Icterus, hyperpigmented knuckles and had no cyanosis, clubbing, edema, nor any lymph node enlargement. His Systemic examination was within normal limits. He had no past history of recurrent abdominal pain-(gallstones), any cardiac disorders detected in chidhood, or tuberculosis. No significant family history of hematologic disorders. Investigations revealed that he had pancytopenia with Hb-5.5 g/dl, total WBC count-3800 neutrophil predominant, platelet count of 1.3 lacs. His MCV was 100 and his RDW 18%. His Liver function tests revealed indirect bilirubinemia of 3.3 g/dl with AST-156, ALT-48, and a high ESR of 70. His RFT was normal, USG abdomen done showed no hepatosplenomegaly. As his labs revealed a hemolytic pattern, tests were done in this line showed an elevated LDH of 13840 (normal<250), Reticulocyte count 0.8% which makes an RPI <2. Direct and indirect coombs test were negative, ANA-IF -Negative. IRON STUDIES showed normalcy with Serum. Fe-213, Ferritin-89, TIBC-217. A peripheral smear done revealed-Normocytic normochromic red cells with occasional polychromasia and fragmented cells, with spherocytes elliptocytes and tear drop cells, white cells had normal count with neutrophil predominance with occasional hypersegmented neutrophils, platelets were reduced. Hb electrophoresis - All Hb fractions WNL and TSH - 4.3(normal). In view of atypicality of presentation and completion, a bone marrow study was done (with consent) which showed Erythroid series hyperplasia with megaloblastic maturation with a small percentage showing nuclear budding and irregularity and normal Myeloid & Megakaryocytic maturation. He was Managed with 2-pint PRBC transfusion his HB improved to 8. In view of a picture suggestive of megaloblastic anemia, he was given a trial of B12 and assessed for reticulocyte response. His repeat reticulocyte count after 3 weeks was 5.6 and HB was 10 g/dl. To rule out any malabsorption he had undergone endoscopy which was also normal. As to his cause of low B12 levels, his vegetarian lifestyle with his food fussiness was thought to be causative.

3. Discussion

Our 16-year-old patient was presented with symptoms of increasing fatigue and weakness associated with dyspnea on exertion with very low Hb, confirming the presence of anemia. Concomitant jaundice with elevated direct bilirubin and LDH and the presence of multiple scattered erythrocytes seen in the peripheral specimen suggest erythrocyte destruction or hemolytic anemia. Hemolytic anemia could be congenital or acquired. Because the patient has no history of anemia, no blood transfusion, and no previous symptoms related to anemia or gallstone disease, it is unlikely that a family history of anemia is also present. Hb electrophoresis also eliminated the presence of hemoglobinopathy, there is no history that could indicate G6PD deficiency (hematuria, drug use, fever). Screening for viral hepatitis to rule out anemia and concurrent hepatitis. His Coombs test, ANA, and antiphospholipid antibodies were negative, ruling out autoimmune hemolytic anemia. Microangiopathic hemolytic anemia such as thrombotic thrombocytopenic purpura (TTP), hemolytic uremic syndrome (HUS), and disseminated intravascular coagulation (DIC) seemed less likely because the patient had normal coagulation parameters and no renal or neurologic abnormalities. Other intravascular hemolysis, such as valvular heart disease, was also ruled out because he had no murmurs on physical examination and a history of heart disease. The probable cause of hemolytic anemia in this case was vitamin B12 deficiency, as the serum B12 level was very low, and the diagnosis was confirmed by an elevated homocysteine level. Vitamin B12 deficiency is usually associated with macrocytic anemia without jaundice. The increased RDW was consistent with patterns of poikilocytosis and anisocytosis on peripheral blood examination. A low reticulocyte index (<2) also suggests defective erythropoiesis, which can be explained by severe vitamin B12 deficiency and iron deficiency anemia. Intramedullary destruction or hemolysis of fragile and abnormal erythrocyte precursors results from ineffective secondary erythropoiesis. and

cell maturation [1]. The hemolytic picture may resemble microangiopathic hemolytic anemia [1]. If this condition persists, it can deplete iron stores and cause concomitant iron deficiency anemia [5]. Pernicious anemia is the most common cause of cobalamin deficiency worldwide [1]. However, among the elderly, the majority of cases are caused by malabsorption of food and cobalamin due to progressive atrophy of the gastric mucosa and hypochlorhydria [2,3,6]. The syndrome is characterized by an inability to release cobalamin from food for absorption due to decreased gastric acid secretion, but unbound cobalamin can be absorbed normally [2,3]. Folate deficiency can also cause elevated serum homocysteine, but not MMA [1]. Therefore, concomitant folate deficiency should be sought in patients with suspected vitamin B12 deficiency. Due to financial constraints, the concentration of MMA and folate could not be controlled The reason was revealed by further studies. He never had abdominal pain, reflux symptoms, or antacid use, making it less likely that atrophic gastritis or Helicobacter pylori infection was the cause, as shown by EGD evaluation with pathologic biopsy. That said, since his diet consists mostly of greens and vegetables, it's doubtful that a folate deficiency was involved. *Helicobacter pylori* infection is associated with gastric atrophy that can result first in iron deficiency and later lead to cobalamin malabsorption and perhaps even predispose to pernicious anemia [5,6].

REFERENCES

1. Hunt A, Harrington D, Robinson S. Vitamin B12 deficiency. BMJ. 2014;349:g5226.
2. Federici L, Henoun Loukili N, Zimmer J, et al. Update of clinical findings in cobalamin deficiency: Personal data and review of the literature. Rev Med Interne. 2007;28(4):225-31.
3. Vucelić V, Stancić V, Ledinsky M, et al. Combined megaloblastic and immunohemolytic anemia associated - A case report. Acta Clin Croat. 2008;47(4):239-43.
4. Ventura P, Panini R, Tremosini S, et al. A role for homocysteine increase in haemolysis of megaloblastic anaemias due to vitamin B12 and folate deficiency: Results from an in vitro experience. Biochim Biophys Acta. 2004;1739(1):33-42.
5. Bunn HF. Vitamin B12 and pernicious anemia-The dawn of molecular medicine. N Engl J Med. 2014;370(8):773-6.
6. Hershko C, Ronson A, Souroujon M, et al. Variable hematologic presentation of autoimmune gastritis: Age-related progression from iron deficiency to cobalamin depletion. Blood. 2006;107(4):1673-9.