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Case Study

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CASE REPORT ON SEVERE VITAMIN B12 DEFICIENCY IN YOUNG CHILD: PRESENTATION WITH PANCYTOPENIA AND SPLENOMEGALY

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ABSTRACT

Vitamin B12 deficiency is a well-recognized cause of nutrition deficiency anaemia. The presentation of vitamin B12 deficiency ranges from being asymptomatic to affecting multiple organ systems. Vitamin B12 deficiency manifests as macrocytic anaemia and presenting symptoms often include signs of anemia, such as fatigue and visible pallor, generalized weakness and hyperpigmentation. We report a case of a 15-year-old patient with severe vitamin B12 deficiency who presented to our clinic with easy fatiguability and hyperpigmentation of knuckles. Patient had dramatic improvement after initiation of treatment with vitamin B12 supplementation. This case highlights the necessity of early diagnosis and treatment of vitamin B12 deficiency especially in vegetarians when clinician encounters vague complaints like skin hyperpigmentation and easy fatiguability.

INTRODUCTION

Vitamin B12 is an important water-soluble vitamin which is required for the development, myelination, and function of the central nervous system and DNA synthesis. Its deficiency is associated with hematologic, neurologic, psychiatric, cutaneous, gastrointestinal, and cardiovascular disorders. B12 deficiency manifests as macrocytic anaemia and presenting symptoms often include signs of anemia, such as fatigue and visible pallor. The deficiency of vitamin B12 can cause specific skin manifestations, such as hyperpigmentation of knuckles and terminal phalanges, angular stomatitis, and hair and nail changes. Because of diversity in sign and symptoms of vitamin B12 deficiency it is often overlooked and misdiagnosed in clinical practice.

CASE REPORT

A 15-year-old boy born to non-consanguineous parents presented with complaints of paleness and easy fatiguability which had been noted by the parents for about 2 years, although these findings became more obvious within the last few weeks. Patient also noted progressive development of hyperpigmentation on his knuckles which later involved dorsal aspect of interphalangeal joints and distal phalanges. Patient was strict vegetarian who primarily ate vegetables, rice and pulses. He denied any history of bleeding per rectum, change in bowel habits, weight loss, and fever. His previous medical and medication history was unremarkable. He had never undergone any surgical procedure. On the physical examination, patient had severe pallor and mild icterus present. He was afebrile and his heart rate was 90 bpm and 2/6 systolic murmur at all auscultatory foci was heard. On systemic examination, he was found to have mild splenomegaly. The rest of the examination was unremarkable.

Investigations

Laboratory investigations revealed severe anaemia (haemoglobin 5.8 g/dL; macrocytosis with mean corpuscular volume (MCV), 118 fL), leucopenia (white cell count 3200/µL; neutrophil count 1140/µL) and thrombocytopaenia (121000/mm^3). A peripheral smear shows features of megaloblastic anemia (hypersegmented neutrophils) with pancytopenia and leukoerythroblastic reaction. No abnormal cells were found. Serum vitamin b12 levels were found to be severely low <83pmol/L. Total bilirubin was 3.8 mg/dL with normal alanine aminotransferase (ALT), aspartate aminotransferase (AST), and alkaline phosphatase (ALP). Renal function tests and other chemistry labs were normal. Serum Lactate dehydrogenase (LDH) levels were 630 U/L. C reactive protein, direct Coombs and testing for glucose-6-phosphate dehydrogenase (G6PD) deficiency were all negative. The autoimmune workup which included anti-nuclear antibody, anti Ds DNA and anti-transglutaminase IgG and IgA antibodies were unremarkable. Serum iron levels, serum ferretin, total iron binding capacity and percentage saturation was found to be normal. Hemoglobin electrophoresis showed a normal pattern. Bone marrow aspiration revealed hypercellular bone marrow with macrocytosis hypersegmented neutrophils. no obvious myelodysplasia or malignancy was noted.

Treatment

Diagnosis of severe vitamin B12 was established. Patient had severe anaemia which was caused by nutritional deficiency and no sign of congestive heart failure was seen. So blood transfusion was deferred. He was then initiated on daily intramuscular injections of vitamin B12 at 1000µg for two week, followed by weekly injections for the next four weeks, and he was subsequently prescribed monthly injections to be continued indefinitely. Oral iron (4 mg/kg/day of elemental iron) and folic acid (5 mg/day) were also initiated. The patient was advised to increase their dietary intake of vitamin B12 from animal or fortified foods.

Outcome and Follow-UP

Following 2 week of vitamin B_{12} supplementation, patient's symptoms were resolved and knuckle hyperpigmentation was also improved. His haemoglobin levels are 9.5 g/dL, haematocrit 31%, WBC count 3460, platelets 138 000/µL and MCV 98fL.

DISCUSSION

Vitamin B12 deficiency of treatable but often unrecognized in clinical practice due to diversity in sign and symptoms of its deficiency. Its symptoms can be clinically subtle ranging from asymptomatic to multiorgan involvement. Its deficiency is associated with hematologic, neurologic, psychiatric, cutaneous, gastrointestinal, and cardiovascular disorders. Other presenting complaints may include peripheral neuropathy, glossitis, stomatitis, hair changes, diarrhea, headaches, and neuropsychiatric disturbances.

Reversible skin and mucosal hyperpigmentation are the most commonly found skin manifestations of vitamin B12 deficiency.[1] It has been observed that up to 1 in 5 patients with a deficient B12 level may have cutaneous hyperpigmentation.[2] Hematological abnormalities in vitamin B12 deficiency ranges from an incidentally discovered increase in mean corpuscular volume (MCV) in asymptomatic patients to full-blown megaloblastic anemia.[3]

Other hematological abnormalities include pancytopenia and hemolytic anemia. Vitamin B12 deficiency may cause bone marrow aplasia, which as a result causes extramedullary haematopoiesis resulting in a rise in the splenic red cell volume, which is usually associated with cytopenic concentrations in the splenic red pulp as a compensatory mechanism, causing splenomegaly.[4]

A serum B12 level <200 pg/mL along with clinical evidence of disease is consistent with deficiency, and no further confirmatory testing is needed. However, in symptomatic patients with borderline B12 levels from 200 to 300 pg/mL, deficiency is possible and verification with serum methylmalonic acid and/or serum homocysteine level may be necessary.[5] It does not ordinarily require bone marrow aspiration or biopsy for diagnosis of vitamin b12 deficiency. However, many patients still undergo this procedure as vitamin B_{12} deficiency may present with findings mimicking acute leukaemia like pancytopenia and organomegaly.

The treatment consists of oral and/or parenteral vitamin B12 depending on the severity of symptoms and the level of deficiency and therapeutic doses of folate. Parentral vitamin B12 at the dose of 1000 µg intramuscular injection daily for two week, followed by weekly injections for the next four weeks, and then monthly for life.

Learning point

1. Vitamin B12 deficiency is common cause of nutritional anaemia and seen predominantly in strict vegetarians
2. Vitamin B12 deficiency may present with findings mimicking acute leukaemia like pancytopenia and splenomegaly.
3. Parentral administration of vitamin B12 and folic acid supplementation results in rapid resolution of associated symptoms and of abnormal laboratory findings.

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