VOLUME - 15, ISSUE - 04, APRIL - 2026 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Paediatrics

WHEN CLINICAL EXAMINATION MAKES THE DIFFERENCE: A TEEN WITH PANCYTOPENIA AND A SURPRISING DIAGNOSIS - AN INTERESTING CASE REPORT

Dr Chelamkuri Pardha Venkateshwar Rao — Junior Resident, Department of Paediatrics, SDUAHER

Dr. Krishnappa J — Professor, Department of Paediatrics, SDUAHER

Dr. Lavanya R A — Assistant Professor, Department of Paediatrics, SDUAHER

ABSTRACT Nutritional anemia is common in Indian adolescents, yet vitamin B12 deficiency is often underdiagnosed. Case: A 16-year-old male presented with fatigue, weakness, and abdominal discomfort. Examination showed severe pallor, icterus, and splenomegaly. Labs revealed pancytopenia and dimorphic anemia. Investigations: Peripheral smear showed macrocytic cells and hypersegmented neutrophils. Bone marrow aspiration confirmed megaloblastic changes. Management: Stabilized with PRBC transfusion. Serum B12 levels confirmed deficiency. Conclusion: This case highlights the importance of clinical and smear examination in diagnosing B12 deficiency, advocating for its inclusion in adolescent health programs.

KEYWORDS : Vitamin B12 Deficiency, Megaloblastic Anemia, Pancytopenia, Peripheral Neuropathy, Hepatosplenomegaly, Adolescent.

INTRODUCTION

Megaloblastic anemia due to vitamin B12 deficiency is a reversible but often underdiagnosed condition in adolescents. It presents with a wide spectrum ranging from fatigue and gastrointestinal symptoms to life-threatening pancytopenia and neurological complications. Due to overlapping features with hematologic malignancies or aplastic anemia, it can be misdiagnosed without a high index of suspicion [1,2].

In resource-constrained settings, a thorough physical examination remains a critical and low-cost tool that often provides essential diagnostic clues—like mucocutaneous signs, organomegaly, and subtle neurologic findings [3,4]. Here, we report a case where hyperpigmented knuckles and splenomegaly, coupled with suggestive peripheral smear findings, directed us toward the correct diagnosis.

Case Study

A 16-year-old boy presented with a 6-month history of progressive fatigue, generalized weakness, poor appetite, intermittent dull abdominal pain, associated with vomiting – non projectile non bilious for 1 week. and significant weight loss (8-9kg). No h/o fever, bleeding, joint pain, or infections. Child had good scholastic performance.

Dietary history : 3 meal per day with no major snacks
Common meals: rice/ chapathi, dhal, vegetables .
No animal -source foods.
On initial assessment, Vitals: Tachycardia, Tachypnea, and raised JVP, Collapsing pulse, suggestive of Congestive Cardiac Failure.

Anthropometry :
Weight- 35.5 kg (3^{rd}-10^{th} centile) normal
Height -173 cm (@75^{th} centile) normal
BMI – 11.8 KG/m2 -underweight

On General Physical Examination: he appeared pale and Icterus noted. hyperpigmented knuckles +, no lymphadenopathy, pedal oedema

No glossitis or mucosal ulcers.

On Per Abdomen Examination: liver was palpable 7 cm below the right costal margin, and the spleen 3.5 cm below the left costal margin, both firm, smooth, and non-tender.

Neurologically, he had bilateral lower limb sensory loss (vibration and position sense), in the lower limbs as compared to upper limbs, though power was preserved.

These diverse systemic findings prompted a detailed hematologic workup:
Hematological Parameters:
Hemoglobin: 3.9 g/dl, WBC: 2,400/cu.mm, Platelets: 87,000/cu.mm
MCV: 107fl, (macrocytic), MCH: 36pg, MCHC- 34.2, Retic count-1
Peripheral Blood Smear: Severe Anisopoikilocytosis, Macro ovulocytes, target cells, Hypersegmented neutrophils

LFT - Total bilirubin: 2.2 mg/dL, Direct bilirubin -0.4, SGPT-64, SGOT- 33, ALP- 7.2, Total Protein- 7.2, Albumin- 4.5, Globulin- 2.7, A/G- 1.6, GGT-10, Vit B12 levels -159 Folic acid -2.51 Ca-8.9 /Po4-4.7/Uric Acid- 26/ SE- Na – 137, K – 4.

USG abdomen-SPLENOMEGALY (enlarged spleen 12.0 x 5.4 cm) with normal echotexture.

In view of organomegaly, a bone marrow aspiration and biopsy were done to rule out infiltrative or malignant processes, despite suggestive peripheral findings. Bone Marrow showed erythroid hyperplasia with megaloblastic changes, no atypical or dysplastic cells confirming the diagnosis.

Nerve Conduction Studies: Sensory neuropathy, more in the lower limbs, with normal motor conduction.

Given the constellation of findings—pancytopenia, dimorphic anemia, hypersegmented neutrophils, hepatosplenomegaly, and peripheral neuropathy—a working diagnosis of megaloblastic anemia (Vitamin B12 deficiency) was established.

In view of Severe Anemia in Congestive Cardiac Failure Packed red cell transfusion (2 units at 5 mL/kg over 4 hours) was given and started on Intramuscular methylcobalamin 1000 mcg, thrice weekly for two weeks and folic acid 5mg OD and continued orally at same doses. Diet chart was provided to ensure dietary modifications and he was compliant.

The child showed marked clinical improvement, and repeat labs at discharge were: Hb: 7.7 g/dL, WBC: 3.83 x 10^3/L, Platelets: 165 x 10^3/L

Child was followed up after 2 weeks – Clinically improved with good appetite, weight gain noted currently weighing 40 kg and his CBC on follow up revealed –HB- 10.5, RBC- 3.66, WBC- 6.66 , PLT- 358.

DISCUSSION

Vitamin B12 deficiency can present with varied manifestations including anemia, pancytopenia, neuropathy, and organomegaly. In adolescents, it is often underdiagnosed, especially in resource-limited settings. The presence of hypersegmented neutrophils and macroovulocytes on peripheral smear typically suggests a megaloblastic process [1,2]. However, the presence of hepatosplenomegaly, as seen in our patient, raises the possibility of infiltrative disorders or hematological malignancies, necessitating a bone marrow examination to rule out conditions like leukemia or lymphoma [3].

Bone marrow in our case showed erythroid hyperplasia with megaloblastic maturation, confirming vitamin B12 deficiency as the etiology. The observed dimorphic anemia also suggests co-existing nutritional deficiencies, possibly iron, which is common in adolescents from low socioeconomic backgrounds [4]. The accompanying sensory neuropathy, a hallmark of B12 deficiency, further supported the diagnosis.

Prompt recognition and treatment with parenteral vitamin B12 led to significant hematologic and clinical improvement, reinforcing that early intervention can prevent irreversible neurological sequelae [5].

CONCLUSION

Early identification and prompt treatment of vitamin B12 deficiency are crucial to prevent permanent neurological damage and restore normal hematologic function. This case highlights how comprehensive clinical examination, integrated with targeted investigations, can accurately diagnose treatable causes of pancytopenia like megaloblastic anemia.

REFERENCES

1. Stabler SP. Vitamin B12 deficiency. N Engl J Med. 2013;368(2):149–60.
2. Bhatia P, Kulkarni KP. Vitamin B12 deficiency in children: Often missed diagnosis. Indian Pediatr. 2015;52(8):613–7.
3. Tiwari A, Chaudhary N, Singh A. Clinical and hematological profile of pancytopenia in children. Indian J Hematol Blood Transfus. 2019;35(1):35–40.
4. Chhabra R, Kumar P. Reversible bone marrow failure secondary to vitamin B12 deficiency in adolescents. Indian J Pediatr. 2018;85(11):1009–11.
5. Osifo OD, Evbuomwan I. Megaloblastic anemia masquerading as leukemia in a teenager. J Trop Pediatr. 2016;62(1):69–71.