

A Rare Case of Severe Vitamin B12 Deficiency Presenting as Pancytopenia and Neuropsychiatric Manifestations in a Young Adult: A Case Report

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Abstract

Background

Vitamin B12 deficiency commonly presents with megaloblastic anemia and neurological symptoms; however, severe pancytopenia with neuropsychiatric manifestations in young adults is relatively uncommon and may mimic hematological malignancies or bone marrow failure syndromes.

Case Presentation

A 28-year-old male presented with progressive generalized weakness, exertional breathlessness, tingling and numbness of both lower limbs, memory disturbances, and loss of appetite for three months. Clinical examination revealed pallor, mild icterus, impaired vibration sense, and gait instability. Laboratory investigations demonstrated severe pancytopenia with hemoglobin 5.8 g/dL, total leukocyte count 2,100/mm³, platelet count 72,000/mm³, and macrocytosis with mean corpuscular volume of 118 fL. Peripheral smear showed macro-ovalocytes and hypersegmented neutrophils. Serum vitamin B12 level was markedly reduced (78 pg/mL). Bone marrow examination revealed megaloblastic erythropoiesis. The patient was treated with parenteral vitamin B12 therapy, folic acid supplementation, and supportive care. Significant hematological and neurological improvement was observed within six weeks.

Conclusion

Vitamin B12 deficiency should be considered in the differential diagnosis of pancytopenia and neuropsychiatric manifestations, particularly in young adults. Early diagnosis and prompt treatment can result in complete clinical recovery and prevent irreversible neurological damage.

Keywords: *Vitamin B12 deficiency; Pancytopenia; Megaloblastic anemia; Neuropsychiatric manifestations*

Introduction

Vitamin B12 deficiency is a common nutritional disorder, especially in developing countries. It predominantly affects the hematological and nervous systems. The classical presentation includes macrocytic anemia, glossitis, and peripheral neuropathy. However, severe pancytopenia associated with neurological and psychiatric manifestations is uncommon and may resemble bone marrow disorders such as aplastic anemia, myelodysplastic syndrome, or acute leukemia ⁽¹⁾.

The deficiency results in impaired DNA synthesis leading to ineffective hematopoiesis and neurological dysfunction due to demyelination of the dorsal columns and corticospinal tracts ⁽²⁾. Early recognition is important because timely treatment can reverse most hematological and neurological abnormalities.

We report a rare case of severe vitamin B12 deficiency presenting with pancytopenia and neuropsychiatric manifestations in a young adult male who showed dramatic improvement following replacement therapy.

Case Presentation

A 28-year-old unmarried male presented to the Department of General Medicine with complaints of progressive generalized weakness, easy fatigability, exertional breathlessness, tingling sensation in both lower limbs, difficulty in walking, irritability, and poor memory for the past three months. He also reported decreased appetite and weight loss of approximately 4 kg over two months.

There was no history of fever, bleeding manifestations, chronic diarrhea, tuberculosis, alcohol intake, drug abuse, or exposure to radiation/toxic chemicals. The patient followed a strict vegetarian diet for many years.

Clinical Examination

On examination, the patient was conscious and oriented but appeared pale and mildly icteric. Vital signs were stable:

- Pulse rate: 96/min
- Blood pressure: 110/70 mmHg
- Respiratory rate: 20/min
- Temperature: Afebrile

Neurological examination revealed impaired vibration and position sense in both lower limbs with positive Romberg sign. Deep tendon reflexes were diminished at the ankles. Mild cognitive impairment and irritability were noted.

Systemic examination of cardiovascular, respiratory, and abdominal systems was otherwise unremarkable.

Investigations

Hematological Parameters

Parameter	Result	Reference Range
Hemoglobin	5.8 g/dL	13–17 g/dL
Total leukocyte count	2,100/mm ³	4,000–11,000/mm ³
Platelet count	72,000/mm ³	150,000–450,000/mm ³
Mean corpuscular volume	118 fL	80–100 fL
Reticulocyte count	0.5%	0.5–2.5%
ESR	30 mm/hr	<20 mm/hr

Peripheral smear showed macro-ovalocytes, anisopoikilocytosis, and hypersegmented neutrophils.

Biochemical Parameters

Investigation	Result
Serum Vitamin B12	78 pg/mL
Serum Folate	Normal
LDH	980 U/L
Total Bilirubin	2.1 mg/dL
Thyroid Profile	Normal
Liver Function Tests	Normal
Renal Function Tests	Normal
HIV, HBsAg, HCV	Negative

Nerve conduction studies revealed sensory neuropathy.

Bone marrow aspiration demonstrated hypercellular marrow with megaloblastic erythropoiesis.

Upper gastrointestinal endoscopy was normal. Anti-intrinsic factor antibody testing could not be performed due to financial constraints.

Differential Diagnosis

The following differential diagnoses were considered:

1. Aplastic anemia
2. Myelodysplastic syndrome
3. Acute leukemia
4. Megaloblastic anemia due to vitamin B12 deficiency

5. Hypersplenism

The presence of macrocytosis, hypersegmented neutrophils, low vitamin B12 levels, and megaloblastic marrow favored vitamin B12 deficiency.

Treatment

The patient received:

- Intramuscular hydroxocobalamin 1000 mcg daily for one week
- Weekly injections for four weeks followed by monthly maintenance therapy
- Oral folic acid 5 mg once daily
- Two units packed red blood cell transfusion
- Nutritional counseling and dietary advice

Outcome and Follow-Up

The patient showed symptomatic improvement within one week. Reticulocyte response was observed by day 5.

At six-week follow-up:

Parameter	Baseline	After 6 Weeks
Hemoglobin	5.8 g/dL	11.2 g/dL
Total leukocyte count	2,100/mm ³	5,800/mm ³
Platelet count	72,000/mm ³	220,000/mm ³
Mean corpuscular volume	118 fL	94 fL

Neurological symptoms improved significantly, and gait became normal. Cognitive symptoms also improved.

Discussion

Vitamin B12 deficiency remains an important cause of reversible hematological and neurological disease worldwide. Pancytopenia occurs due to ineffective hematopoiesis and intramedullary destruction of blood cell precursors⁽³⁾. Severe deficiency may clinically mimic bone marrow failure syndromes or hematological malignancies.

Neurological manifestations may include peripheral neuropathy, subacute combined degeneration of the spinal cord, cognitive dysfunction, depression, and psychiatric disturbances⁽⁴⁾. Interestingly, neurological involvement may occur even in the absence of severe anemia.

In the present case, the patient had severe pancytopenia associated with sensory neuropathy and cognitive changes. Strict vegetarian diet was considered the major predisposing factor. Similar

findings were reported by Andr s et al., who observed pancytopenia and neurological manifestations among patients with severe cobalamin deficiency.⁽⁵⁾

Macrocytosis and hypersegmented neutrophils on peripheral smear remain important clues toward diagnosis. Bone marrow findings of megaloblastic erythropoiesis further support the diagnosis. Elevated LDH and indirect bilirubin reflect ineffective erythropoiesis and hemolysis. Early treatment with parenteral vitamin B12 usually results in rapid hematological recovery. Neurological recovery depends on duration and severity of symptoms before treatment initiation.⁽⁶⁾ Delayed diagnosis may lead to irreversible neurological deficits.

This case highlights the importance of considering vitamin B12 deficiency in patients presenting with pancytopenia and neuropsychiatric manifestations, particularly in regions where vegetarian dietary practices are common.

Conclusion

Vitamin B12 deficiency can present with severe pancytopenia and neuropsychiatric manifestations, mimicking serious hematological disorders. Careful clinical evaluation along with peripheral smear examination and vitamin B12 estimation are essential for early diagnosis. Prompt treatment results in excellent hematological and neurological recovery.

Author contribution

All authors have contributed equally

Conflict of Interest

Nil

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