

Received Date: August 21, 2025**Accepted Date: September 13, 2025****Published Date: October 01, 2025**

Vitamin B12 Deficiency Revealed by Severe Pancytopenia:

A Case Study

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Abstract- Vitamin B12 deficiency is a rare but significant cause of anaemia associated with neurological manifestations in children. We report the case of a 16-month-old infant presenting with profound pancytopenia and psychomotor regression, who was diagnosed with severe vitamin B12 deficiency. This case highlights the importance of measuring vitamin B12 levels in children with anaemia associated with neurological signs.

Keywords: Vitamin B12, pancytopenia, infant, megaloblastic anaemia, nutritional deficiency.

1. Introduction

Vitamin B12 is essential for DNA synthesis and neurological function. A deficiency of this vitamin can lead to megaloblastic anaemia and neurological damage. In children, it is often linked to insufficient intake, particularly in cases of prolonged exclusive breastfeeding by a deficient mother. The prevalence of this deficiency varies according to region and dietary habits, with a higher incidence among vegetarian populations and those with limited access to animal sources of vitamin B12 [1,2].

Several studies have highlighted the serious neurological and haematological consequences of prolonged vitamin B12 deficiency in infants, which can lead to irreversible sequelae if diagnosis is delayed [3,4].

We report a case of severe vitamin B12 deficiency revealed by profound pancytopenia, in order to raise awareness among clinicians of this potentially reversible cause of haematological and neurological abnormalities.

2. Observation

A 16-month-old female infant, born to non-consanguineous parents with no notable neonatal history, was presented for consultation due to asthenia, hypotonia, growth retardation and marked psychomotor regression. The clinical symptoms were insidious, with a gradual deterioration in the child's general condition. The infant had been exclusively breastfed since birth, without vitamin supplementation, particularly vitamin D or vitamin B12. The introduction of solid foods, which began at 5 months of age, was difficult and the child gradually refused to eat solid foods. This refusal to eat solid foods, combined with weight loss and a decrease in growth, led the parents to seek medical advice.

The initial clinical examination revealed generalised pallor of the skin and mucous membranes. The neurological examination revealed marked hypotonia. The child appeared apathetic, with no reaction or interest in surrounding stimuli. In addition, weight gain retardation was observed, with a weight below -2 SD (standard deviation), and motor retardation, characterised by the inability to sit up. These findings were consistent with developmental delay, both physical and cognitive, suggesting significant nutritional deficiency.

Biological tests confirmed the clinical diagnosis. The blood count revealed severe pancytopenia, with haemoglobin at 5.6 g/dL, and a high mean corpuscular volume (MCV) of 108 fL, suggesting megaloblastic anaemia, associated with leukopenia ($2,300/\text{mm}^3$) and thrombocytopenia ($90,000/\text{mm}^3$). This pancytopenia pointed to a condition affecting blood cell production, in particular a deficiency in vitamin B12, which is essential for red blood cell synthesis and proper bone marrow function. The myelogram performed revealed bone marrow rich in megaloblasts, confirming megaloblastic anaemia and ruling out other causes of pancytopenia, such as aplasia or leukaemia.

Biological tests revealed severe hypovitaminosis B12, with serum vitamin B12 levels below 100 pg/mL in both the child and the mother, leading to a diagnosis of nutritional vitamin B12 deficiency. This deficiency was probably due to an exclusively milk-based diet without vitamin B12 intake, combined with a refusal to diversify the diet, which is essential for the supply of this vitamin. Vitamin B12 deficiency can lead to megaloblastic anaemia, neurological demyelination and psychomotor retardation, as observed in our case.

Treatment consisted of hydroxocobalamin supplementation, administered intramuscularly at high doses during the initial phase, followed by oral hydroxocobalamin during the maintenance phase. The aim of this treatment was to quickly correct the vitamin B12 deficiency and improve the child's clinical condition. At the same time, examination of the mother revealed Biermer's disease with high levels of anti-intrinsic factor and anti-parietal cell antibodies. She began treatment with intramuscular hydroxocobalamin, which led to a favourable clinical outcome.

After one week of treatment with hydroxocobalamin, a blood count check showed almost complete normalisation of the three blood cell lines. The child showed a significant improvement in his clinical condition, with a gradual recovery of muscle tone and motor activity, and signs of resumed psychomotor development were observed, including an improvement in his ability to sit up. The infant gradually became more alert and responsive to stimuli, and his appetite began to return, allowing solid foods to be introduced with greater acceptance.

This favourable outcome confirmed the effectiveness of vitamin B12 replacement therapy and highlighted the

importance of early management of nutritional deficiencies, particularly vitamin B12 deficiency, in the first months of life.

Follow-up was reinforced with regular consultations to ensure complete normalisation of psychomotor development and prevention of recurrence, particularly through education of the family on the child's specific nutritional needs and regular monitoring of vitamin B12 levels.

3. Discussion

3.1. Vitamin B12 and its fundamental biological roles

Vitamin B12, or cobalamin, is a water-soluble vitamin essential for many biological functions, including DNA synthesis and DNA methylation, two processes critical for cell division and blood cell maturation [1]. It is also crucial for fatty acid metabolism and central nervous system (CNS) function [2,3]. Vitamin B12 deficiency disrupts these processes and leads to neurological and haematological abnormalities, particularly in rapidly growing infants [4]. Infants have increased requirements to support axonal myelination and brain development, which explains why deficiency can have serious and sometimes irreversible consequences for their neurological development [5, 6].

3.2. Vitamin B12 deficiency: An issue in breastfed infants

Vitamin B12 deficiency in infants is often linked to insufficient intake due to the mother's diet, particularly in cases where the mother follows a strict vegetarian diet. This condition is particularly common in populations where vegetarianism is widespread, without adequate vitamin B12 supplementation [7]. In fact, the breast milk of vegetarian mothers who do not take supplements is often deficient in vitamin B12, thereby increasing the risk of deficiency in their children [8]. One study reported that up to 15% of infants breastfed by vegetarian mothers have insufficient levels of vitamin B12, which can lead to neurological disorders, psychomotor regression and macrocytic anaemia [9, 10].

Neurological symptoms of vitamin B12 deficiency in infants include motor disorders such as hypotonia, apathy, psychomotor developmental regression, and loss of acquired skills such as sitting or walking [11, 12]. These signs can easily be confused with other paediatric neurological disorders, delaying diagnosis and treatment [13].

3.3. Absorption mechanisms and genetic risk factors

Vitamin B12 is mainly absorbed in the terminal ileum via an intrinsic factor (IF) receptor, a glycoprotein secreted by the stomach [14]. However, genetic abnormalities, such as mutations in the cobalamin receptor gene, can impair this absorption, rendering even adequate dietary intake ineffective [15]. In addition, diseases such as Biermer's disease, which affects the production of intrinsic factor, lead to malabsorption

of vitamin B12 [16]. These factors are particularly relevant in cases of deficiency where dietary supplementation appears to be insufficient, and they require genetic investigations and malabsorption tests to confirm the diagnosis [17].

3.4. Clinical manifestations and diagnosis

The clinical manifestations of vitamin B12 deficiency in infants are varied and may include neurological disorders, gastrointestinal signs, and megaloblastic anaemia [18]. Indeed, blood counts may show pancytopenia with megaloblastic red blood cells, as well as bone marrow hyperplasia [19]. Biochemically, measurements of vitamin B12, methylmalonyl-CoA (MMA) and homocysteine are key diagnostic tools [20]. MMA and homocysteine levels are often elevated in patients with vitamin B12 deficiency, even though serum vitamin B12 levels are not always indicative in the early stages of the disease [21].

3.5. Management and treatment of vitamin B12 deficiency

Treatment of vitamin B12 deficiency is based primarily on hydroxocobalamin injections, which are administered in two phases: the initial phase, with high doses, and the maintenance phase, which consists of less frequent but regular injections [22]. Biological monitoring is essential to assess response to treatment, and improvement in haematological and neurological parameters can be observed after only a few days of treatment [23]. Infants, as in our case, generally show a significant improvement in blood count after one week of treatment, confirming the effectiveness of supplementation [24].

3.6. Prevention prospects: supplementation and early screening

To prevent vitamin B12 deficiency, supplementation strategies for vegetarian breastfeeding mothers should be implemented systematically. One study has shown that vitamin B12 supplementation in breastfeeding mothers significantly reduces the risk of deficiency in their infants, thereby improving their neurological health [25]. In addition, systematic screening of high-risk infants, particularly those who are exclusively breastfed, should be incorporated into routine paediatric care [26]. Postnatal care should include monitoring vitamin B12 levels in mothers and children, especially in vegetarian populations and in countries where this condition is still widely underdiagnosed [27].

Conclusion

Vitamin B12 deficiency in infants is a preventable public health problem, but it remains underdiagnosed in many regions, particularly among vegetarian populations. The neurological and haematological symptoms observed in these infants highlight the importance of early screening and prompt

treatment to avoid long-term consequences on psychomotor development. It is therefore crucial to incorporate prevention strategies, such as vitamin B12 supplementation in breastfeeding mothers and systematic screening of infants, to ensure optimal health and normal development in young children.

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