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A case of vitamin B12 deficiency in an exclusively breastfed child.

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Abstract

Vitamin B12 deficiency in infants is a serious concern due to its role in neurological development and hematopoiesis. This deficiency often arises from inadequate intake, necessitating early diagnosis to prevent enduring neurological consequences.

We present the case of a 14-month-old girl exclusively breastfed until her presentation with symptoms of asthenia and psychomotor regression. Clinical examination revealed macrocephaly, peripheral hypotonia, and abolished reflexes. Laboratory tests identified macrocytic anemia, neutropenia, and elevated homocysteine levels. Vitamin B12 levels were significantly low. Investigations on the mother showed no anaemia and a vitamin B12-level at the lower range of normal. The child received intramuscular vitamin B12 and well-diversified diet. The evolution was favourable with normal examination at the age of 18 months old.

This case underscores the importance of considering vitamin B12 deficiency in breastfed infants with neurological and hematological abnormalities, even when maternal vitamin B12 levels appear normal. Long-term neurological outcomes remain uncertain, emphasizing the need for vigilance in diagnosis and management.

Keywords: Vitamin B12 deficiency, Children, Breast-milk

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Introduction

Rare and severe, vitamin B12 deficiency in infants represents a serious and potentially life-threatening condition. Vitamin B12, also known as cobalamin, plays a pivotal role in various critical physiological processes, with its absence having particularly dire consequences in the early stages of life.

One of the most vital functions of vitamin B12 is its integral role in the hematopoiesis and the development of the central nervous system.

Vitamin B12 deficiency in infants primarily arises from inadequate intake, particularly if they are exclusively breastfed by mothers with a deficiency or following a strict vegetarian or vegan diet devoid of animal products [1].

Thus, routine monitoring of vitamin B12 levels in at-risk children is crucial to support their healthy growth and neurological development.

We present here the case of a 14-month-year old girl with vitamin B12 deficiency presenting with asthenia and psychomotor regression and evolved favorably after vitamin B12 supplementation.

Case Presentation

We report a 14-month-old girl presented with asthenia and psychomotor regression. Medical history revealed that she was born at term, after a pregnancy complicated by gestational diabetes and an uncomplicated delivery. No parental consanguinity was observed. She was born to a mother aged thirty-five with no previous medical history.

The infant was exclusively breastfed until the age of 14 months.

Psychomotor development was normal, with unsupported standing acquired at the age of 1 year. Walking was not yet acquired.

The mother reported a 2-day febrile episode the past month that resolved spontaneously, followed two weeks later by hypotonia with loss of the ability to stand.

On examination, the infant was eutrophic but had macrocephaly. She had no facial dysmorphia. She was placid and looks pale. On neurological examination, the child was unable to stand up, she had peripheral hypotonia, and symmetrically abolished osteotendinous reflexes, preserved sensitivity, no muscle atrophy, no signs of intracranial hypertension and no organomegaly.

Laboratory Investigations demonstrated a normochromic macrocytic anaemia (haemoglobin 8.3 g/dl, mean corpuscular volume 103fl), nonregenerative (reticulocyte 28200el/mm 3) with neutropenia (PNN 1410el/mm3) and no biological inflammatory syndrome.

No rhabdomyolysis was found (The Creatine phosphokinase 41 UI/L and Lactate dehydrogenase 261UI/L). A lumbar puncture showed no albuminocytological dissociation (1 element/mm3,

albuminorrachia 0.16 g/L, glugorrachia 0.8 g/l). The renal and hepatic function was normal. A chest X-ray, an electro-cardiogram and an electromyogram were performed with no abnormalities. Cerebral Magnetic resonance imaging was normal.

Considering peripheral hypotonia, spinal muscular atrophy, Guillain-Barré syndrom, metabolic and genetic myopathy was eliminated.

Given the association of macrocytosis and neurological signs, vitamin B12 deficiency was suspected. We then completed with the vitamin B12 assay, which was very low 65 pmol/L (reference range: 145-569 pmol/L) while folic acid was normal 19,88 nmol/L (reference range: 11-34 nmol/L). The homocysteine assay showed an elevated level of 91 micromol/L [normal <10 micromol/L]. Metylmalonine chromatography was normal.

As part of the etiological work-up, investigations on the mother showed no anaemia and a vitamin B12-level at the lower range of normal (154 nmol/L-reference range: 145-569 pmol/L). Intrinsic factor antibodies were negative however anti-stomach parietal cell antibodies were positive. In view of these results, Biremer's disease was not considered in the mother.

Our patient was diagnosed with a vitamin B12 deficiency, due to a vitamin deficiency in breast milk (The assay of vitamin B12 in the maternal milk was not performed).

The child received intramuscular vitamin B12 and well-diversified diet. The dose of vitamin B12 was 1 mg/day for one week, then 1 mg/week for 1 month

Three days after the first injection, she was smiling again and was neither lethargic nor placid. She was able to stand up with support. The osteotendinous reflexes were present but weak. Biologically, the level of haemoglobin, neutrophils and vitaminB12 has normalised after one month (Table1).

At the last follow-up, the child was 18 months old. On examination she was reactive and able to walk. The head circumference was normal (48 cm) and the osteotendinous reflexes were present.

Results and Discussion

Vitamin B12 deficiency in infants is a rare and potentially serious condition. Jadhav et al., described the first observation of vitamin B12 deficiency in 1962 [1]. However its prevalence isn't well elucidated in the literature. In a referral center for inherited metabolic disorders, Honzik reported that vitamin B12 deficiency in breastfed children represent 1% in all children referred for suspected metabolic disease in 5 years [2].

Our patient is the second Tunisian paediatric case to be published. In a study by Honzik et al., [2], it was reported that the average age at diagnosis was 4.4 months (range 1–19.5 months). Our patient was diagnosed at the age of 14 months. The diagnostic delay can be explicated with the mild deficiency of the mother [2,3].

No clinical signs are constant or pathognomonic and the deficit may be asymptomatic [2].

This disorders is responsible for neurological signs such as peripheral neuropathy developmental delay or regression, hypotonia, apathy, anorexia, epilepsy and microcephaly [2,4].

The neurological signs may occur due to the role of this vitamin in the development and the myelination of the nervous system; the accumulation of neurotoxic substance like homocysteine and reduced tetrahydrofolate formation required for methylation reactions.

Microcephaly was reported in 23% of the cases, however in our case the infant presented macrocephaly with a normal cerebral MRI and metabolic assessment. Our patient was diagnosed with idiopathic macrocephaly.

In our case the infant presented neurological manifestations associated with haematological signs like macrocytic anaemia and neutropenia.

Actually the macrocytic anaemia was present in 63% of the cases and a neutropenia was reported in one case by Cariou et al., No additional haematological signs were documented in the literature [2-4].

	On the admission	**Evolution after 1 month**
Psychomotor development	Unable to stand up	Walk after 3 months
Osteotendinous reflexes	Symmetrically abolished	Present but weak
Head circumference (cm)	48 (>90th percentile)	48
Haemoglobin (g/dl)	8,2	10,2
MCV*(fl)	103	72
Absolute neutrophil count (/mm3)	1410	2310
Serum vitamin B12 concentrations (pmol/l)	65	518

Table 1: Clinical and biological data on admission and after one month. **Note:** Mean Corpuscular Volume (MCV).

When neurological and haematological signs are combined, vitamin B12 deficiency should be considered as the main cause but after ruling out other conditions such as Spinal Muscular Atrophy, Guillain-Barre Syndrom and myopathy.

The absence of abnormal findings in the electromyogram and the concurrent presence of megaloblastic anemia guide the diagnosis toward a potential etiology of Vitamin B12 deficiency. This diagnosis was confirmed by a low vitamin B12 concentration in the infant.

The most common cause of vitamin B12 deficiency in breastfeed infant is the mother's vitamin deficiency. Maternal deficiency was due to malnutrition, strict vegetarian diet or Bireme's disease, leading to a reduction in neonatal reserves and the onset of vitamin B12 deficiency.

A deficiency in the mother's milk was the only possible etiology to explain the vitamin B12 deficiency in our infant. McPHEE, et al., reported a similar case in which the investigations showed a normal maternal serum vitamin B12 concentration while the concentration of vitamin B12 in the breast milk were very low [5]. Thus, a decreased level can exist in breast milk even if the blood level is normal.

In our case, the family history reviled that the mother had an unbalanced diet and she excluded many types of animal protein.

Treatment consists of the diet's diversification and replacement therapy by administrating vitamin B12.

There is neither consensus on the dose nor on the duration of the treatment. In a case reported by Cariou et al., [4] the infant was given intramuscular injection of 1 mg/day, three times a week then the substitute treatment was quickly administrated orally for a total duration of four months. However side effect like tremor was reported by Cariou et al., with good spontaneous improvement.

After receiving the vitamin supplementation, in our patient the evolution was marked by clinical and biological improvement both on the hematological and neurological levels. The other reported cases show a rapidly favourable improvement [4-6].

However the long-term prognosis remains uncertain with several cases of irreversible neurological sequelae [7].

Conclusion

The diagnosis of vitamin B12 deficiency should be considered when macrocytic anaemia is associated with recent-onset neurological symptoms in an exclusively breastfed child.

The primary reason for vitamin B12 deficiency in breast-fed infants is typically due to a deficiency in the mother.

This diagnosis is not ruled out, even in the presence of a normal vitamin B12 level in maternal blood. The long-term neurological outcome after treatment is unpredictable.

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