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Hypotonia and Psychomotor Delay Revealing a Vitamin B12 Deficiency: About 2 Cases

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Abstract **Original Research Article**

Vitamin B 12 has important consequences for hematopoiesis and the central nervous system, its deficiency leads to clinical hematological, neurological and psychiatric manifestations. However, most infants with B12 deficiency are born to women with low vitamin B12 levels and have been exclusively breastfed. An early diagnosis and treatment may prevent the progression of neurological damage to irreversible deficits. We report the case of two infants aged 6 months and 10 months, exclusively breastfed, with a significant psychomotor delay, and hypotonia. Physical examination confirmed the pallor, the axial hypotonia, biological examination revealed pancytopenia with a collapsed vitamin B12 level in both infants and both mothers.

Keywords: Hypotonia, Psychomotor delay, Vitamin B12 deficiency, Breastfeeding.

BACKGROUND

A vitamin B12 deficiency in infants is rare, but may sometimes be seen in breastfed babies of strict vegetarian mothers. The aim of this work is to highlight the clinical and biological characteristics of this rare entity.

We report here 2 observations of intake deficiency. The clinical manifestations are not very specific and include a delay in heignt weight and psychomotor development.

OBSERVATION

Case 1:

A 6-month-old infant was hospitalized for pancytopenia. The symptoms began at the age of 2 months with an alteration of the general state, hypotonia, psychomotor regression, apathy, and a slowing of growth, he was under exclusive breastfeeding. The clinical examination showed hypotrophy with lower heignt and weight developement. Weight 5200g (-3DS), height 59 cm (-3DS) and a head circumference: 41 cm (-1DS), very pale, poor eye contact, faint cry, axial hypotonia with slight peripheral hypertonia.

Biological examinations: pancytopenia: Hb at 6.5 g/dl and VGM at 91 fl, a Reticulocytes count 65000, leukocyte count, 6200 cells/mm3 6200 including 600 PNN, platelets count 11000cells/mm3.

Medullogram: very rich marrow with maturation disorders.

Vit B12: 45 pg/ml (197-866) was very low.

Folate: 12, 3 ng/ml (3-19) Nl.

Homocysteine: 136 micromol/l (3.60-21.7).

Amino acid chromatography in blood and urine revealed homocysteinuria.

Brain scan: diffuse cerebral atrophy.

The search for an autoimmune disease associated with the hypothyroidism in the mother was used to make the diagnosis of Biermer's disease. A level of vitamin B12 was very low to 87.55 pg/ml and folate to 12.74 ng/ml, *intrinsic factor antibody* negative and Antiparietal cell antibody positive.

Case 2:

A 10-month-old infant was hospitalized for bicytopenia. The symptoms began at the age of 4 months with an alteration of the general state, hypotonia a psychomotor developmental delay, under exclusive breastfeeding. The clinical examination finds an intense pallor, an axial and peripheral hypotonia.

Biological examinations: pancytopenia: Hb at 6 g/dl and VGM at 75 fl, a Reticulocytes count 55000.

leukocyte count 6200 cells/mm3 including 5500 PNN, platelets count 260000 cells/mm3.

Medullogram: very rich marrow with maturation disorders.

Vit B12: 35 pg/ml (197-866) collapsed.

folates: 15, 3 ng/ml (3-19) normal.

Brain scan: diffuse cortical atrophy.

The mother's vitamin B12 level was very low to 107.55 pg/ml and folate at 12.74 ng/ml.

Treatment with vitamin B12 (Hydroxycobalamin) an injection of 1 mg per day IM for one week, then a weekly injection for one month, then monthly in both patients, with a good clinical and biological improvement and normalization of the vitamin B12 level.

DISCUSSION

Vitamin B12 deficiency in infants has been individualized since the publication of Jadhav *et al*., in 1962, reporting 6 Indian children.

Vitamin B12 in its reduced form (mono- or divalent cobalt) is an essential coenzyme in 2 biochemical reactions (Fig 1). The first is the transformation of homocysteine into methionine by methylation in the cytoplasm; the second transforms methylmalonyl coenzyme A into succinyl coenzyme A in the mitochondria [1]. These two cobalamin-dependent reactions reduce the quantities of two potentially toxic substances: homocysteine, which is responsible for vascular endothelial damage, and methylmalonate, which is responsible for metabolic acidosis [2].

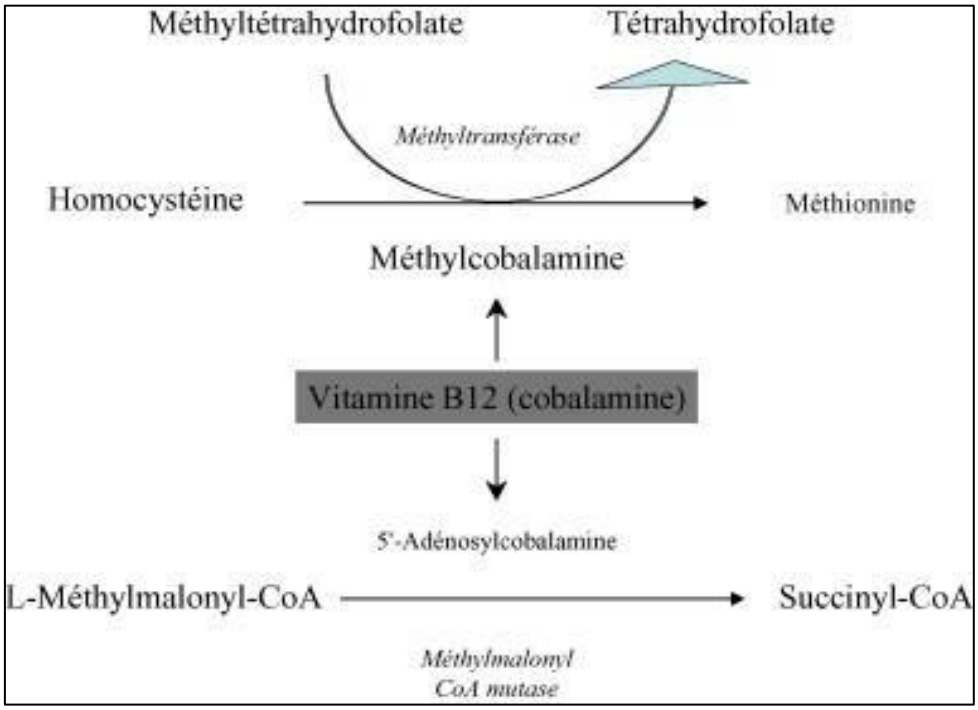


Figure 1: Rôle de coenzyme de la vitamine B12 [4]

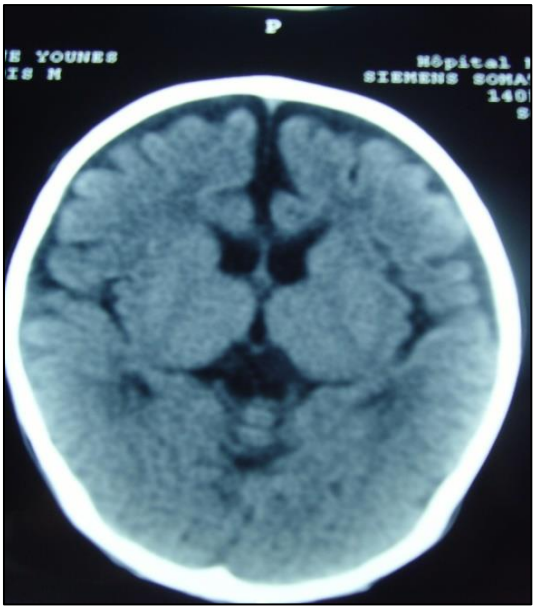
Vitamin B12 is exclusively contained in animal products [1]. Maternal deficiency is most often secondary to Biermer's anemia or to a strict vegetarian diet excluding all animal products, which is transmitted to the infant in case of exclusive maternal feeding.

About a hundred cases of B12 deficient infants have been described in the literature, 64% of which were in vegetarian breastfeeding mothers, 24% in mothers with Biermer's disease, the others were related to digestive pathologies or general deficiencies [3].

The cause may be secondary to a deficiency in intake, an abnormality in absorption, or an inborn error in transport and metabolism [4].

The concentration of vitamin B12 in breast milk parallels that in serum. Normally, a newborn has 25 µg of vit B12 stored in the liver, a quantity supposed to be sufficient until the end of the first year of life [2], these endogenous stocks may be much lower if the mother is deficient as our patient, explaining the clinical signs at the age of 2 months.

The main clinical manifestations of vitamin B12 deficiency are extremely polymorphic and of variable severity [5] occurring in the first year of life, the most common symptoms include failure to thrive, hypotonia, irritability or lethargy, developmental delay and even regression [6, 7], epilepsy or movement disorder [8, 9], brain atrophy, and delayed myelination [10].



Brain scan: diffuse cerebral atrophy

Biologically, serum folate levels are normal, chromatography of amino acids and organic acids reveals an accumulation of homocysteine and plasma and urinary methyl malonic acid confirm vitamin B12 deficiency [11].

The absence of megaloblastosis in our patient is not uncommon as well as the association of hypothyroidism with Biermer's disease in the mother in the context of an autoimmune syndrome.

After treatment with vit B12, the improvement is generally spectacular on the clinical and biological levels [12].

However, the long-term prognosis remains uncertain, with several cases of sequential psychomotor retardation [13], which would be all the more frequent when the diagnosis is made after the age of one year. This highlights the need to prevent vitamin B12 deficiency in pregnant women and nursing mothers with a strict vegetarian diet or Biermer's disease.

CONCLUSION

The clinical presentation of vit B12 deficiency is typical, consisting of *delayed growth*, hypotonia, and delayed or regressed psychomotor development with megaloblastic anemia. The occurrence of such symptoms requires the dosage of vit B12, especially if the infant is exclusively breastfed.

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