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Case report

Psychomotor development following early treatment of severe infantile vitamin B12 deficiency and West syndrome – Is everything fine? A case report and review of literature

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Abstract

Background: Severe infantile vitamin B12 deficiency is occasionally reported in developed countries due to maternal nutritional deficiency. The clinical manifestation comprises megaloblastic anemia and neurodevelopmental delay culminating in demyelination and brain atrophy. Few case reports have documented manifestation of West syndrome.

Patient: We report the 8-year long-term follow-up on a 6-month-old exclusively breast-fed girl with serious vitamin B12 deficiency secondary to undiagnosed maternal pernicious anemia. MRI revealed cerebral atrophy and delayed myelination. Strikingly, initial vitamin B12-mediated improvement of neurological and hematological findings was followed by temporary manifestation of infantile spasms requiring anticonvulsive therapy.

Results: Seizures soon dissolved, EEG and MRI scan normalized and developmental catch-up occurred. At the age of 8 years, the girl is symptom-free and visits primary school illustrating remarkable recovery of severe neurodevelopmental delay and symptomatic West syndrome.

Conclusion: Infantile vitamin B12 deficiency has to be considered in the differential diagnosis of mental retardation and infantile spasms, especially if maternal nutritional deficiency might be suspected. Early treatment seems to be crucial for the prevention of irreversible brain damage.

Keywords: Infantile vitamin B12 deficiency; Brain damage; Psychomotor development; West syndrome; Infantile spasms syndrome; Neurodevelopmental delay; Long-term follow-up

1. Introduction

In developed countries, infantile vitamin B12 deficiency usually occurs in children exclusively breast-fed by malnutritioned mothers due to vegetarianism or veganism or malabsorption in a number of gastrointestinal diseases [1–5]. Developmental delay, muscle hypotonia, apathy, and failure to thrive are the typical but non-specific neurological symptoms [6]. Epilepsy has been reported as a rare manifestation of vitamin B12 deficiency. Moreover, few reports have described a causal relationship with manifestation of West syndrome [2,4,7,8]. Although vitamin B12 supplementation often

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Table 1
Admission and follow-up data.

Parameter	At Admission (aged 6 months)	After 2 weeks	After 6 weeks	After 4 months	After 9 months (aged 15 months)	Aged 2 years	Aged 8.5 years
Basic data							
Weight (kg)	6.0 (0.2 kg < 3rd perc.)	6.8 (25th perc.)	9.7 (85th perc.)	10.6 (90th perc.)	11.95 (75th perc.)	14.2 (95th perc.)	30.8 (70th perc.)
Length (cm)	64.0 (3 cm < 3rd perc.)		70 (20th perc.)	73.0 (30th perc.)	84.0 (50th perc.)	85.1 (25th perc.)	134.7 (50th perc.)
OFC (cm)	42.0 (10th perc.)		44.5 (50th perc.)	46.2 (70th perc.)	48.5 (85th perc.)	49.5 (85th perc.)	52.0 (85th perc.)
Clinical findings							
General pediatric status	Severe pallor, apathy, tachycardia 200/min, auscultation normal, moderate splenomegaly	Rosy skin color, adequate reaction, adequate weight gain	Rosy skin color, adequate reaction, adequate weight gain	Adequate reaction, adequate weight gain	No anomaly	No anomaly	No anomaly
Neurological status	Significant delay, comatose, generalized hypotonia, no tendon reflexes, no head support	Significant delay, mild hypotonia, normal tendon reflexes adequate head support, intended movements	Continuous progress, adequate head support, able to turn around, mild hypotonia, infantile spasms	Continuous progress, adequate sitting, able to straighten up mild hypotonia no seizures	Motoric, cognitive + language almost adequate, able to walk, 6–7 words normal muscle tone, no seizures	Motoric, cognitive + language adequate, normal muscle tone, no seizures *BSID II*: MDI + PDI normal	Motoric, cognitive + language adequate, no seizures, deficits in numerical reasoning, *WISC-IV*: PRI 79 (normal 85–114), VCI, WMI, PSI & FSIQ normal
Apparative diagnostics							
EEG	Abnormal, significantly slowed background activity		Hypsarrhythmia, slowed background activity	No hypsarrhythmia, slowed background activity	No hypsarrhythmia, normalized background activity	Normal	Normal
Cranial MRI	Frontal + frontoparietal cranial atrophy, delayed myelination		Regressive cranial atrophy delayed myelination		No structural alterations, myelination almost adequate		
Laboratory findings							
Hb (g/dl) (normal 11.5–15.5)	3.2	10.0		11.7	12.8	12.2	12.6
Hkt (%) (normal 34–40)	10	31		34	37	34	39
Erythrocytes (cells/mm^3) (normal 3.9–5.3 × 10^6)	960,000	3.310.000		3.970.000	4.480.000	4.200.000	4.340.000
MCV (fl) (normal 77–95)	110.1	94.7		86.1	81.8	80.2	89.4
MCH (pg) (normal 23–31)	33.3	30.2		29.5	28.6	36.2	29.0
Platelet count (cells/mm^3) (150–450 x 10^3)	77,000	696.000		354.000	232.000	240.000	220.000
Leukocyte count (cells/mm^3) (normal 5–17 × 10^3)	6,550	10.690		6.040	6.500	5.440	4.930
Reticulocytes (%)	2.6	12.2		3.9	n.a.	1.5	
Peripheral blood smear	Macrocytosis, dysplasia						
Bone marrow examination	Reactive myelodysplasia						