Severe encephalopathy with epilepsy in an infant caused by subclinical maternal pernicious anaemia: case report and review of the literature.

Abstract: Vitamin B(12) deficiency is one of the major causes of megaloblastic anaemia with or without neurological symptoms. We report on a patient manifesting acute encephalopathy, epilepsy, microcephaly and megaloblastic anaemia at the age of 4 months. Vitamin B(12) deficiency in the patient was due to subclinical pernicious anaemia of the mother who exhibited neither haematological nor neurological symptoms. Mother and child both had elevated methylmalonic acid in their urine which is a sensitive parameter of vitamin B(12) deficiency. Vitamin B(12) therapy resulted in arrest of convulsions within 24 h. There were no further seizures although the patient showed moderate mental retardation at the age of 7 years but a normal head circumference. Long-term MRI follow-up, performed at the age of 7 years, showed moderate enlargement of the ventricles with reduction of myelin and hypoplasia of the corpus callosum. CONCLUSION: Vitamin B(12) deficiency due to maternal pernicious anaemia should always be considered in the differential diagnosis of neurological symptoms in infants and especially in combination with megaloblastic anaemia. Since the age of onset and the duration of neurological symptoms may contribute to the development of long-term symptoms, early diagnosis and treatment is important for vitamin B(12) deficient children.