Case Report

Infantile Tremor Syndrome: A Case Report with Review of Current Evidence

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Abstract

Background: Infantile Tremor Syndrome (ITS) is a rare yet clinically significant nutrition-related neurological disorder that is observed in most infants and young children in developing nations. Tremors, neurodevelopmental delay or deterioration, anemia, pigmented skin, and shaggy hair are characteristic feature. Increasing evidence suggests a strong association with vitamin B12 deficiency, particularly in exclusively breastfed infants born to nutritionally deficient mothers. We report a case of a 10-month-old female infant exhibited typical features of Infantile Tremor Syndrome with markedly low vitamin B12 levels despite normal growth, and showed significant clinical improvement after treatment. ITS should be considered in infants with tremors and developmental delay even with normal anthropometry, as early detection and vitamin B12 supplementation can prevent long-term neurodevelopmental complications.

Keywords: Infantile tremor syndrome; Vitamin B12 deficiency; Nutritional rehabilitation; Neurodevelopmental delay.

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Introduction

Infantile Tremor Syndrome (ITS) is a rare nutritional disease that mainly affects children aged 5 months to three years.[1] It is typified by a classical tetrad of pallor, developmental retardation or regression, skin hyperpigmentation, and sparse, hypopigmented scalp hair with or without involuntary tremors.[2] The syndrome has been reported in the Indian subcontinent, Southeast Asia, and a certain segment of Africa, with male children showing a higher incidence.[3]

The exact etiology of ITS is still unclear. Still, there is growing evidence to implicate the condition with vitamin B12 deficiency, especially in those infants who are only breastfed and are born to mothers with subclinical or overt deficiency of nutrients.[4] Early recognition and immediate nutritional response are important, as failure to control the disease promptly can lead to long-term neurodevelopmental disabilities.

Case Presentation

A 10-month-old female infant was referred with a history of an increasing number of tremors during the last month, loss of appetite, and fatigue. The shaking was mostly in the upper extremities, rough in character, and increasing in frequency. There is also the observation of a change in the child's voice in response to the mother.

There was no history of fever, seizures, trauma, or chronic systemic disease. Historical development showed delays across all areas. The infant was born at term with a normal perinatal history and was exclusive breastfed. Age was appropriate for immunization.

The anthropometric measurements were within normal limits for age and sex.

Upon general observation, the child was apathetic and pale. The scalp hair was thin, coarse, and hypopigmented in appearance. Hyperpigmentation was observed in the knuckles. Neurological physical examination revealed coarse tremors and generalized hypotonia. Other systemic examination was normal.

Investigations

Laboratory testing showed considerably low levels of serum vitamin B12. All other hematological and biochemical parameters were abnormal.

Management and Outcome

Nutritional rehabilitation was conducted in accordance with the Integrated Management of Neonatal and Childhood Illness (IMNCI) and World Health Organization requirements. Parenteral vitamin B12 treatment was initiated, and oral supplementation was provided.

A slow improvement in clinical performance has been reported. The number of tremors decreased significantly during treatment, appetite improved, and the child became more active and engaged in communication with his mother. The patient was advised to continue supplementation and nutritional counselling in a stable condition.

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Discussion

ITS is a rare diagnosis that goes under-diagnosed because of similar symptoms with other neurodevelopmental disorders (5). The current case reinforces the emerging literature regarding the association between ITS and a lack of vitamin B12, among infants with normal anthropometric indices.[6] This demonstrates the weakness of relying solely on growth parameters as a measure of nutritional adequacy.

Recent research has shown that cerebral atrophy and other brain changes in Infantile Tremor Syndrome are reversible with timely administration of vitamin B12. This shows that the disorder is manageable when detected early.[5-8] The role of inappropriate maternal nutrition, particularly exclusive breastfed babies, is yet another factor that is supported by our case, even in infants with normal growth parameters but with tremors and retarded development. An accurate maternal dietary history and simple biochemical tests can help with early diagnosis and prevent the development of long-term neurological and developmental difficulties.

Conclusion

Infantile tremor syndrome should be considered in any young child aged 6 months to 5 years with pallor, tremors, neurodevelopmental delay, or regression, along with typical changes in skin and hair, despite normal anthropometry. ITS is a possibly reversible but potentially severe disorder that has the potential of causing considerable neurodevelopmental impairment when not addressed. The management ought to pay attention to early correction of nutritional deficiencies, such as vitamin B12, and to elaborate parental nutritional counselling. Standardised screening and management for early detection and prevention are also unknown and require additional research to develop standardised protocols.

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Conflicts of interest

There are no conflicts of interest.

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