Neurocutaneous Syndrome of Infantile B12 Deficiency: A Case Series from India (ID: 652)

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Introduction

- Neurocutaneous syndrome of infantile B12 deficiency, is commonly known as Infantile Tremor syndrome (ITS)
- It is a treatable cause of developmental delay and neuro-regression
- Characterized by skin and hair changes, developmental delay or regression of milestones, termers, anemia and a cherubic appearance

Methodology

- All children diagnosed as ITS based on clinical profile and laboratory features were included for the study from July 2023 to March 2024
- Diagnosis of ITS was based on typical cutaneous findings with or without tremors, in children with developmental delay or regression, and a laboratory picture suggestive of megaloblastic anemia.

Results

- A total of 48 children (21 females) in the age range (6-36 months) were enrolled.
- The mean age at diagnosis was 12 months
- Pigmentary skin and hair changes were present in all children (48/48).
- Tremors were present in only 19% at diagnosis
- Most common reason for hospital visit was infection followed by developmental delay/regression.

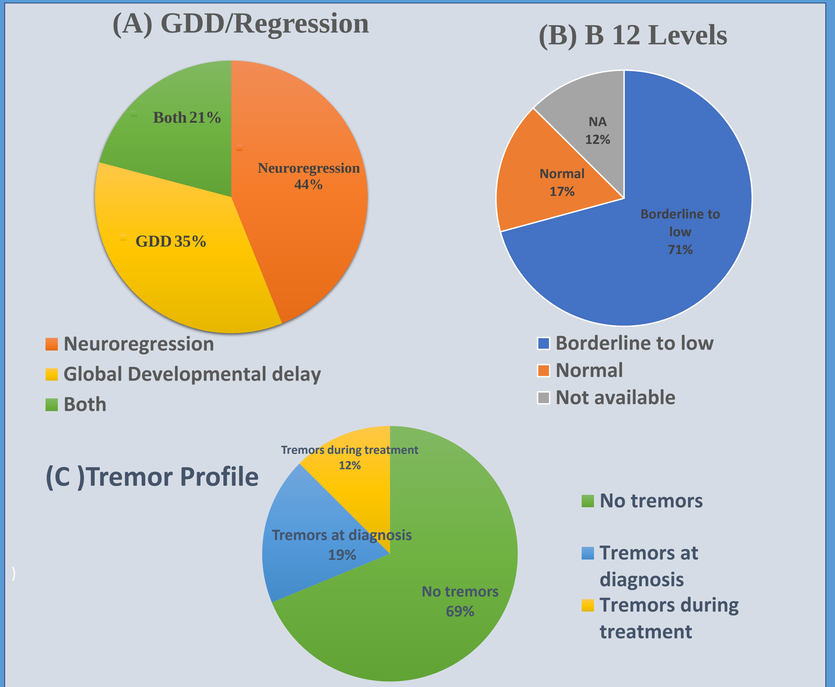

Figure 1: Distribution of Global Developmental Delay (GDD) vs Regression at Presentation (A), Serum B12 Levels (B), and Tremor Profile at Diagnosis (C) in the study population

- Imaging was done in 17 cases Most common neuroimaging finding was diffuse cortical atrophy (12/17; 66.6%), followed by thinning of corpus callosum(6/17; 35%)
- All the children showed a good response to injectable/oral B12 replacement therapy, with a resolution of dullness, and gradual gain in milestones

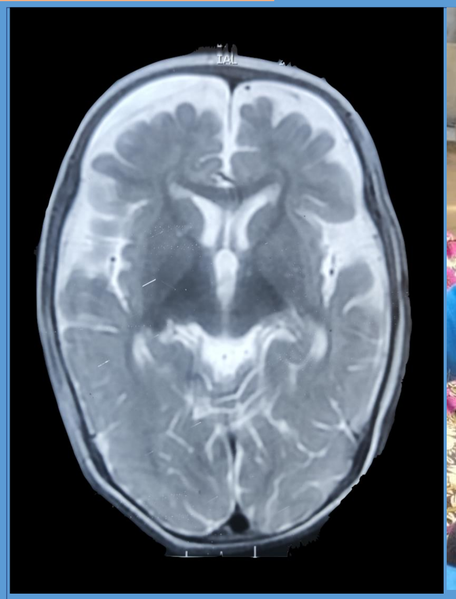

Figure 2: T2-weighted axial MRI brain showing frontal predominant diffuse cortical atrophy

Figure 3: A case showing hypopigmented sparse hair, and cherubic appearance

Conclusion

- The classical features of tremors, low serum B12 levels may not be present in all cases.
- A high index of suspicion is required to diagnose this treatable cause of developmental delay/regression in infants and toddlers.

References.

1. Goraya JS, Kaur S. Infantile tremor syndrome: A review and critical appraisal of its etiology. J Pediatr Neurosci. 2016 Oct-Dec;11(4):298-304. doi: 10.4103/1817-1745.199475. PMID: 28217150; PMCID: PMC5314841.
2. Chaudhary H, Verma S, Bhatia P, Vaidya PC, Singhi P, Sankhyan N. Infantile Tremor Syndrome or a Neurocutaneous Infantile B12 Deficiency (NIB) Syndrome? Indian J Pediatr. 2020 Mar;87(3):179-184. doi: 10.1007/s12098-019-03117-w. Epub 2020 Jan 27. PMID: 31984470.