Benign Paroxysmal Tonic Upgaze of Childhood; a rare syndrome

A.G.A.R. Galappaththi [1] , J. Wanigasinghe [2]

[1]*Registrar in Paediatrics, Lady Ridgeway Hospital*
[2]*Senior Lecturer and Specialist Paediatric Neurologist, Department of Paediatrics, University of Colombo*

ARTICLE INFO

Article history
Submission 02.03.2017
Acceptance 29.05.2017
Available online

DOI:
http://doi.org/10.4038/cjms.v54i1.4820

Introduction

Benign Paroxysmal Tonic Upgaze is an oculomotor syndrome which was first described by Ovurier and Billson [1]. It consists of episodes of conjugate deviation of eyes with the onset in early infancy [2]. Detailed evaluation including metabolic, electroencephalographic and neuroimaging is often normal. The exact pathogenesis is yet unknown. We report a 10 month old boy who presented with persistent tonic upgaze.

Case report

A ten month old boy was presented to us with intermittent upward deviation of the eyes for 3 months duration. These episodes were lasting from one to four hours per days and more during early morning and when the child is fatigued. He is the second child of healthy non consanguineous parents with an uncomplicated antenatal and perinatal period. At 10 months he was able to stand with support with lateral walking and had a mature pincer grasp. He was able to speak few single words. There was no family history of note. He was on Carbamazepine started at the peripheral hospital.

On examination he had upward gaze with down beating nystagmus when attempting to look down. The overall neurological development was otherwise normal. Video electroencephalographic examination was normal. CT brain, electrolytes and blood gases are normal. Ophthalmological assessment performed during the episodes showed binocular fixation with normal horizontal following movements and vertical nystagmus in both eyes. Paroxysmal tonic upward gaze was diagnosed in this child and it was decided not to treat this child and to follow up. Carbamazepine was gradually tailed off and omitted.

Discussion

The clinical characteristics of our patient are consistent with Paroxysmal Tonic Upgaze of childhood with ataxia although our baby didn't yet manifest ataxia. Paroxysmal tonic upgaze is a rare clinical entity. Given its spontaneous outcome it is often under diagnosed. The exact pathogenesis is unknown to date [3]. It is probably mediated by complex interactions between cerebral cortex, cerebellum, basal ganglia, mesencephalon and pons [2]. The observation of similarity between dopamine responsive dystonia and this entity with diurnal fluctuation of symptoms and resolution following levodopa administration, led to the hypothesis that this is a manifestation of neurotransmitter depletion [4]. However subsequent studies have not confirmed the positive effect of levodopa.

The characteristics of the disorder are onset in early infancy, periods of sustained conjugate upward deviation of the eyes, downbeating nystagmus in attempts of down gaze, normal horizontal eye movements, frequent relief by sleep, otherwise normal neurological findings

Corresponding Author:
Dr. A.G.A.R. Galappaththi MBBS, DCH
Registrar in Paediatrics, University Paediatric Unit,
Lady Ridgeway Hospital, Colombo 08, Sri Lanka
rasanjalee17@yahoo.com
http://orcid.org/0000-0002-9557-0238

apart from ataxia, normal metabolic, electroencephalographic and neuro imaging findings and absence of deterioration with spontaneous recovery [2]. Normal neuroimaging helps to differentiate from symptomatic tonic upgaze occurring in tumorous lesion in the pineal region, sylvian aqueductal stenosis, or another organic lesion involving the supranuclear pathways [4].

In conclusion paroxysmal tonic upgaze is a heterogeneous syndrome with respect to outcome.

References

1. Maria Luisa Lispi, Federico Vigevano. Benign paroxysmal tonic upgaze of childhood with ataxia. *Epileptic Disorders. 2002;3(4):203-6*.
2. Robert A. Ouvrier, Frank Billson, Benign Paroxysmal Tonic Upgaze of Childhood. *Brain & Development 27 (2005)185–188*
3. Spalice A, Parisi P, Iannetti P. Paroxysmal tonic upgaze: physiopathological consideration in three additional cases. *Journal of Child Neurology 2000;15: 15-8*
4. Ovrier RA, Billson F. Benign paroxysmal tonic upgaze of childhood. *Journal of Child Neurology 1988; 3:177-80*