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Case Report

A Rare Association between Congenital Strabismus and Cortical Malformations: MRI Evidence of Grey Matter Heterotopia

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Abstract

Congenital strabismus is most often attributed to abnormalities of the extraocular muscles or cranial nerves. However, emerging neuroimaging data suggest that cortical developmental anomalies may play an underrecognized role in it's pathogenesis.

We report the case of a 35-year-old male presenting with longstanding, non-progressive strabismus since early childhood, with no history of trauma, perinatal complications, or neuromuscular disease. Brain MRI revealed nodular heterotopia of grey matter located along the lateral ventricular walls in the occipital region, with no other major structural anomalies. This malformation of cortical development was isointense to cortical grey matter across all MRI sequences, confirming the diagnosis of occipital periventricular grey matter heterotopia.

Keywords: *Strabismus; Grey Matter Heterotopia; MRI; Occipital Region*

Introduction

Congenital strabismus is typically linked to extraocular muscle or cranial nerve abnormalities. However, in rare cases, central nervous system malformations may play a role. Grey matter heterotopia, a neuronal migration disorder, is usually associated with epilepsy or cognitive impairment, but its association with oculomotor disturbances is uncommon.

We present the case of an adult male patient with longstanding congenital strabismus, in whom brain MRI revealed periventricular nodular grey matter heterotopia. This case suggests a possible central origin for unexplained strabismus and highlights the value of neuroimaging in selected adult cases.

Case Report

The concurrent presentation of congenital strabismus and periventricular nodular grey matter heterotopia (PNH) in a 35-year-old patient provides a unique insight into strabismus of potential central origin. While grey matter heterotopia is most frequently reported in connection with epilepsy and cognitive impairments [1], its link to oculomotor dysfunction remains largely unexplored.

A pediatric series of 22 patients with heterotopia identified ocular abnormalities-including strabismus-in approximately 27% of cases [2]. Although the cohort primarily involved children and included other neurological anomalies, this finding supports the plausibility of eye movement disturbances arising from disrupted cortical architecture.

From a neuroanatomical standpoint, strabismus-especially in adults-has been associated with structural changes in cortical and subcortical regions responsible for eye movement control. Voxel-based morphometry studies found decreased grey matter volume in visual cortical areas and increased volume in regions involved in eye-movement control (e.g. frontal eye fields, premotor cortex) among adults with comitant strabismus. Likewise, altered cortical thickness and spontaneous neural activity have been reported in children with strabismus in regions such as the precentral gyrus, angular gyrus, and parietal-occipital cortex [3]. These neuroplastic changes underscore the potential impact of cortical malformations on oculomotor control.

Our patient's PNH-situated periventricularly in the occipital region-might disrupt pathways linking subcortical nuclei and cortical areas crucial for binocular alignment and gaze stabilization (Figure 1). Notably, movement disorders have been attributed to heterotopia involvement in proximity to basal ganglia [4], reinforcing the concept that heterotopic grey matter can have functional consequences beyond epilepsy.

Although genetic heterogeneity is established in heterotopia cases, diverse phenotypes including ocular disorders are documented [5]. This suggests that specific genetic or developmental patterns could predispose to ocular motility anomalies when cortical migration fails in regions subserving oculomotor coordination.

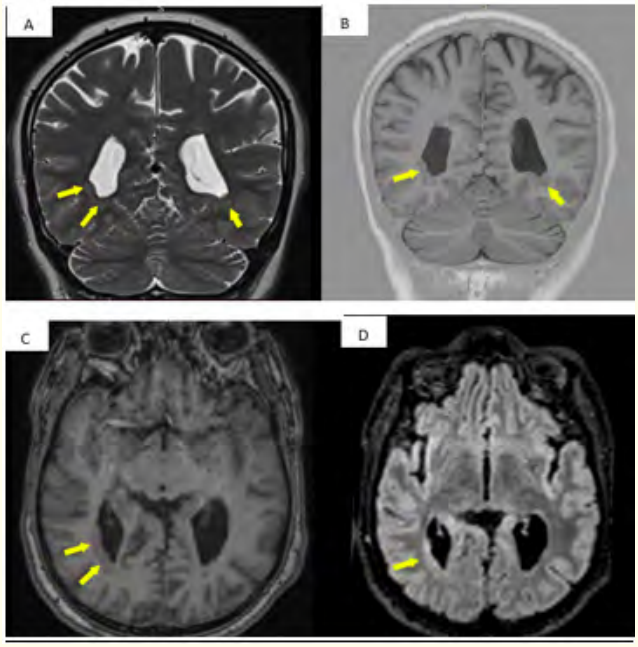

Figure 1: *Brain MRI in coronal T2 (A), coronal TIR (B), axial T1 (C) and axial flair (D) shows the presence of nodular lesions (large yellow arrows) in the occipital periventricular area with a similar signal to that of the grey matter.*

Discussion

Grey matter heterotopia is a rare neurodevelopmental condition resulting from disrupted neuronal migration. Although frequently associated with epilepsy and cognitive deficits, oculomotor symptoms such as strabismus are seldom reported. In this case, the spatial localization of the heterotopia raises the possibility of disrupted cortical-subcortical pathways involved in oculomotor control. This case highlights the potential role of central, cortical factors in otherwise unexplained strabismus and supports the use of brain MRI in atypical or congenital presentations.

We present a 35-year-old patient with longstanding congenital strabismus and MRI-confirmed periventricular nodular grey matter heterotopia (PNH)-a rare finding in the context of oculomotor disorders. Although PNH is classically linked to epilepsy and cognitive issues [6], its role in strabismus is scarcely addressed in literature.

Previous pediatric series revealed ocular disorders in heterotopia cases. For instance, a cohort of 22 children with GMH reported that ≈27% had strabismus, suggesting that cortical migration anomalies might impact eye-muscle coordination, even without seizures. However, adult presentations remain largely undocumented.

In adults with comitant strabismus, voxel-based morphometry (VBM) studies have consistently shown structural changes in the brain. One study identified decreased grey matter volume (GMV) in visual-processing regions (occipital cortex) and increased GMV in oculomotor areas such as the frontal eye fields and premotor cortex [7]. These findings imply that strabismus may arise from cortical reorganization or developmental abnormalities rather than purely ocular-muscular dysfunction.

Anatomically, PNH nodules are adjacent to subcortical and white matter pathways critical for binocular coordination. Disruption in these pathways-whether structural or functional-can undermine gaze alignment. This is consistent with previous cases of movement disorders where heterotopia near basal ganglia precipitated myoclonus or dystonia, which improved after surgical removal [8]. Similarly, our patient's strabismus may be the functional consequence of PNH impinging on oculomotor circuits.

Genetic studies emphasize heterogeneity in GMH, with over 100 implicated genes (FLNA, DCX, etc.) associated with diverse presentations [9]. Variable expressivity means some individuals may exhibit isolated strabismus with minimal or absent epilepsy or cognitive deficits, further supporting the notion that PNH can manifest simply as an oculomotor issue.

Conclusion

This case highlights that grey matter heterotopia-commonly associated with epilepsy and neurodevelopmental disorders-may also manifest solely as longstanding congenital strabismus. Brain MRI can be instrumental in revealing underlying cortical malformations responsible for such ocular misalignment. Documenting additional cases and utilizing advanced imaging techniques could improve our understanding of the neurodevelopmental origins of strabismus with no clear etiology.

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