VOLUME - 11, ISSUE - 01, JANUARY - 2022 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Neurology

CHARACTERISTIC UPPER LIMB DYSTONIA PATTERN AS A CORRELATE TO THE 'EYE OF TIGER' MRI SIGN.

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ABSTRACT Childhood onset dystonia are generally genetic. NBIA is one of the commonest cause. We present five cases with a characteristic upper limb dystonia in patients of suspected PKAN with MRI Brain showing classical 'Eye of tiger' sign.

KEYWORDS : Dystonia, PKAN, Eye of tiger sign.

INTRODUCTION:

While evaluating a movement disorder, pattern of phenomenology is studied and the movement classified into hypokinetic or hyperkinetic movement. Hyperkinetic movements are further classified into tremor, dystonia, chorea, myoclonus according to the rhythmicity, periodicity, velocity and predictability of the movement.[1] Studying the characteristics of the movement does help to recognize the pattern further and can help predict the etiology. Few examples are the St Vitus dance quality of chorea in Syndenham's disease,[2] slow negative myoclonus in SSPE[3], pill rolling quality of tremor in IPD,[4] bon bon tongue in tardive dyskinesia,[5] hand writhing stereotypy in Rett syndrome.[6] These are the characterisitcs patterns which when present help to diagnose the condition and is called pattern recognition. Patttern recognition is important in Neurology and more so when we consider movement disorders. Pattern recognition helps us to minimise the investigations to reach a diagnosis. We present five cases with a characteristic upper limb dystonia in patients of suspected PKAN with MRI Brain showing classical 'Eye of tiger' sign.

Clinical Sign

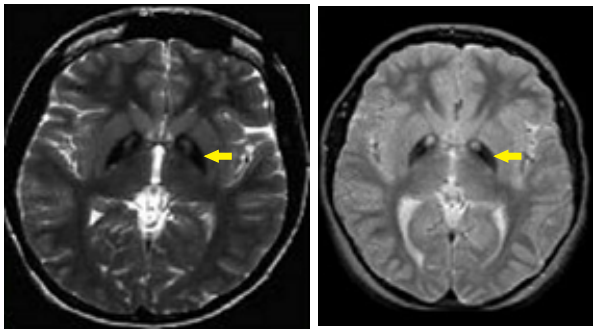
All cases had normal initial development till 2 years of age. Age at symptom onset varied from 2 to 6 years. The symptoms started with change in gait and falls which were due to abnormal foot posturing during walking. Later all were noticed to have characteristic upper limb dystonia which made the upper limb appear as if it is going behind the back. [Figure no.1 a &b]

a b

Figure no.1

Image showing characteristic upper limb dystonia There was extension at shoulder joint, flexion at the elbow and at times pointing index finger. The dystonia was initially task specific and would occur as the child walked, however later appeared at rest too. At times, children would appear to keep the behind going arm over the waist, giving quasi-purposive quality to the dystonia. Patients were examined at 1- 3 years into the illness when all of them had the characteristic upper limb dystonia. All children were found to be playful and happy and they had a smiling face. As the disease advanced, the opposite side was also involved and patient later developed retrocollis and opisthotinus posturing.

MRI brain done on presentation showed bilateral globi pallidi hypo intensity with central hyper intensity on T2 weighted images suggestive of 'Eye of tiger' sign.[Figure no.2 a] SWI images showed iron deposition in bilateral globi pallidi. [Figure no.2 b] MRI showed bilateral symmetric changes even when the dystonia was grossly asymmetric which is very well described in the literature, cause for the same remains unknown.



a b

Figure 2:

MRI Brain: T2 axial (a) shwoing eye of tiger sign

SWI image(b) shwoing iron deposion in b /l globus pallidus

Specificity Of The Sign

The authors examined 5 patients with characteristic upper limb dystonia with MRI correlate of Eye of tiger sign and 25 controls with dystonia due to any cause and MRI not showing 'Eye of tiger'. Only one control group patient who had globus pallidus infarct did show the characteristic dystonia. This means the characteristic upper limb dystonia is highly specific for Eye of tiger MRI correlate.

Pathophysiology-

The pathology of PKAN is largely limit¬ed to the globus pallidus, in contrast to other forms of NBIA. In gross section, the presence of iron is evidenced by a frankly rusty discoloration of the glo¬bus pallidus but not other structures;

this is con¬firmed microscopically with iron-specific stains that reveal the iron to have a perivascular distribution. A central area of neuronal depletion and tissue rar¬efaction corresponds to the "eye of the tiger" seen on MRI. Degenerating ghost neurons are concentrated in globus pallidus abut also seen in putamen and internal capsule. Globi pallidi lesions result in the dystonic syndrome described with PKAN. Despite symmetric pathological affection, the dystonia is grossly asymmetric clinically; pointing to the possibility that functional neurotransmitter deficit is different than structural changes.[7] Why does the pathology lead to characteristic upper limb dystonia needs attention and is a suitable link for future research in PKAN which can have therapeutic implication.

Clinical Significance-

Dystonia in 3-6 year age group children can be due to PKAN, dystonic Cerebral palsy, organic academia, primary genetic dystonia, dopa responsive dystonia and others.[8] In children with dystonia, MRI brain is the standard first investigation of choice. Eye of tiger on T2 weighted images of MRI Brain is seen in PKAN, MPAN, organophosphate poisoning, CO poisoning survivors and MSA.[9] Therefore in children, Eye of tiger sign usually points to NBIA. The characteristic upper limb dystonia had a strong correlation to the Eye of tiger sign. Therefore, the characteristic upper limb dystonia with a happy affect (smiling face) should make the clinician suspect NBIA and MRI should be the first investigation of choice in such cases to look for the 'Eye of tiger' sign. This can show subtle changes of central pallidi T2 hyperintensity in early disease course in the absence of T2 hypointensity which follows later. In such cases the characteristic upper limb dystonia helps to suspect PKAN, which can be proven with genetic testing for PANK2 mutation.

Limitations-

Genetic testing was not done due to affordability issue.

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