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Neurodegeneration with Brain Iron Accumulation: A Case Report

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Abstract

Introduction

Neurodegeneration with brain iron accumulation (NBIA) comprises a clinically and genetically heterogeneous group of disorders affecting children and adults. These rare disorders are often first suspected when increased basal ganglia iron is observed on brain magnetic resonance imaging. For the majority of NBIA disorders the genetic basis has been delineated, and clinical testing is available.

Case

A 17 years old female presented in the casualty in the evening hours with complaints of sudden onset stiffness in both of the lower limbs while taking rest after usual household chores and it gradually involved upper limb and muscles of back with complaints of inability to walk since last few days. On examination patient was conscious ,oriented and patient was able to communicate normally. Both the lower limb were rigid with increased tone of all group of muscles, hypertonia was observed in flexor group of muscles. Reflexes of both upper and lower limbs were exaggerated with 3+ grading. Plantar response was mute on both side. Power was nomal in upper limb and patient was not able to walk due to spasm of lower limb muscles. No history of fever ,trauma, abnormal posturing, loss of consciousness or neck rigidity. Family history was

insignificant.On day 2 of admission, patient had generalised tonic clonic seizures.On treatment with antiepileptics,patient regained consciousness approximately 1 hour after onset of convulsion. No post-ictal focal neurological deficit was present after that episode.

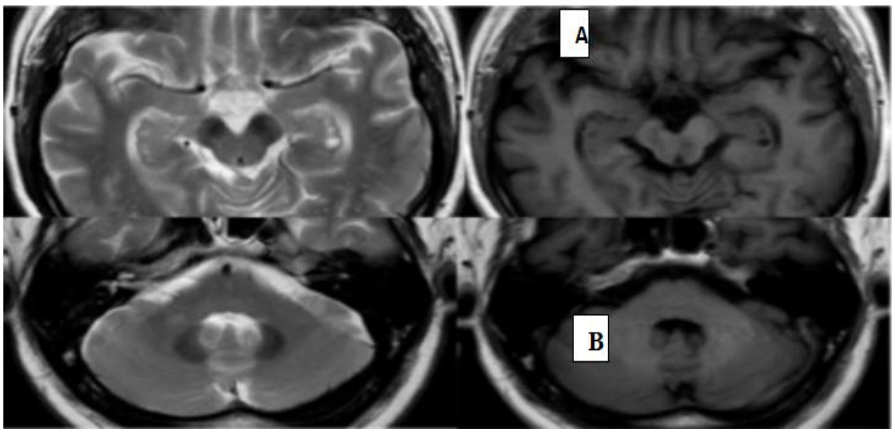
Routine blood investigations were normal. Kidney Function, Liver Function and Serum iron studies did not revealed any abnormality.NCCT head was done, which showed multiple hyperdense foci in bilateral basal ganglia. MRI brain was planned on day 3 of admission. MRI brain (plain+contrast) was done with multiplanar images taken in T1W axial+ sagittal , T2W axial, FLAIR axial, DWI, SWI and T1 W+C sequences. Significant hypointensity noted in the region of bilateral globus pallidus and substantia niagra (Image). Features on imaging were suggestive of Neurodegeneration with brain iron accumulation. Hence after excluding all the other possiblities & differential diagnosis, diagnosis of neurodegeneration with brain iron accumulation was made.

Course of Treatment

Patient was started in Tab trihexyphenidyl 2 mg once a day dosing and Tab Baclofen 10mg in Thrice a day dosing . Patient was asymptomatic since starting of baclofen with no further episode of stiffness.Patient was lost to follow up after 3 months.

Discussion

Over the past decade, the genetic based treatments have been considered for NBIA Disorders. Based on their overlapping phenotypes and pathologies, there was reason to expect their disease genes to reveal new pathways critical for neuronal health. The pathways implicate mitochondrial bioenergetics, lipid metabolism, and autophagy/mitophagy as common processes are of the causes of NBIA disorders. Specific defects in these pathways damage selectively vulnerable cells in the basal ganglia and lead to iron dyshomeostasis. Despite these rapid genetic advances, there are still no curative or disease-modifying therapies for any of the NBIA disorders.



T2 weighted T1 weighted

The above image shows comparison between the normal Brain (A) in T2 & T1 weigted images while the same scan done in Iron accumulated brain (B) is shown.

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