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Case Report

Severe generalized dystonia in paediatric onset wilsons disease

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ABSTRACT

Wilson disease (WD) is a rare autosomal recessive disorder with defect in copper transport mechanism with varied clinical manifestation predominantly hepatic, neurological, ophthalmological and multi-systemic involvement. WD in paediatrics age group manifest differently from the adults. In this case report, Authors have described the first case report presenting with neurological involvement in the form of severe generalized dystonia in a paediatric onset WD. This case report is of greater significance in detecting the most often undetected paediatric WD presenting with a usual hepatic manifestation occurring early in the course.

Keywords: Atypical, Dystonia, Kayser-Fleischer ring, Paediatric, Wilsons disease

INTRODUCTION

Wilson disease (WD) is a rare autosomal recessive disorder with defect in copper transport mechanism wherein, mutations in the ATP7B gene is implicated as major pathogenic factor. The final result in the pathogenesis of WD is systemic overload of copper. WD in paediatrics differs from adult, as the first presentation is usually of hepatic manifestation than of neurological mainfestations.[1] Neurological and psychiatric symptoms usually develop in the second or third decade of life.[2] Dystonia is seen in at least one third of patients with neurological manifestations of WD. The severity of dystonia varies with focal, segmental, multifocal, or generalized involvement.[3] Here, authors present a rare case of early paediatric onset WD with severe generalized dystonia in a 10 year old male patient with mild liver involvement.

CASE REPORT

A 10 year old boy presented with complaints of fever and episodes of abnormal posturing of both upper and lower limbs since 8 months, progressive speech disturbance since 4 months. Initially the patient had intermittent low grade fever, associated with chills and rigor, relieved on taking medications with colicky type of lower abdominal pain associated with nausea and non-projectile vomiting. Following this he developed posturing of upper limbs and lower limbs since 3 months, started first in the left lower limb in the form of flexion of left knee ,ankle and bending of toes over 15 days progressed to involve other lower limb and the upper limbs. This was associated with stiffness and pain in all of the four limbs. After which child developed frequent spasms over the next two months with protrusion of the tongue. Patient also developed speech disturbances in the form of slurring of speech progressing to anarthria. Patient developed difficulty in swallowing and drooling of saliva and chocking while taking food. Patient had difficulty while initiating micturition but no incontinence. The child was a first born to a non-consanguineous parents with a normal birth history. No other similar illness in the family. No history of any chronic drug use. On examination Child was conscious obeying commands with anarthria, jaw opening was present with protruded

tongue and generalized dystonia in the form of flexion of both elbow, wrist and knee with flexion of both feet were noted (Figure 1).

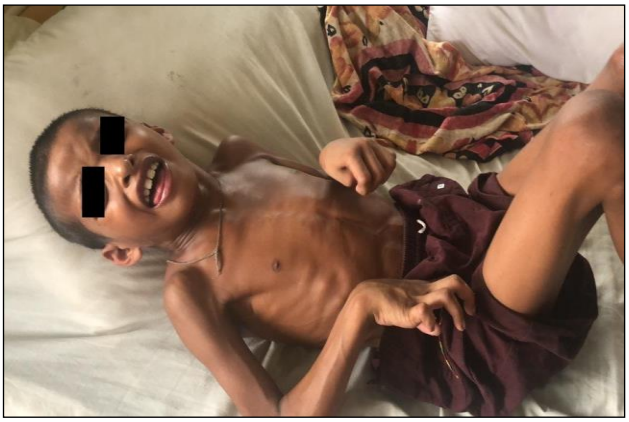

Figure 1: Generalized dystonia.

Cranial nerves examination was normal. Fundus examination was normal. Contractures noted in both elbow, wrist, hip, knee and ankle. Power was 3/5 in all four limbs, deep tendon reflexes 2 and ankle jerk was absent. Systemic examination was normal. At this point differential diagnosis of structural lesion, demyelinating and metabolic disorders were consider. MRI brain revealed bilateral caudate and lentiform nuclease hyperintensity. Considering a possibility of WD at this point, child was subjected to a slit lamp examination and found to have bilateral Kayser-Fleischer ring (Figure 2).

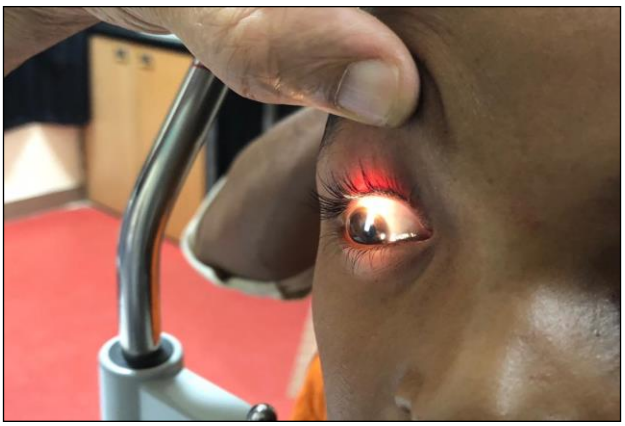

Figure 2: Bilateral Kayser-Fleischer.

Investigations revealed normocytic normochromic blood picture with mild increase in eosinophils , low serum ceruloplasmin and copper level and mildly elevated liver enzymes. Creatinine kinase was slightly raised. Levels of homocysteine and ammonia were within normal limit done to rule out in born errors of metabolism . In view of these features the child was started in the lines of treatment of WD with status dystonicus. Following which child improved with decrease in frequency of dystonic spasms and discharged with advice for further neurological rehabilitation.

DISCUSSION

Classical case of Wilson disease of paediatric onset usually present with hepatic involvement occurring early in the course but may also go unnoticed.[1] Hepatic involvement in WD may range from subclinical liver disease to acute fulminant hepatitis.[1,4] In our patient hepatic involvement was mild with only derangement of enzymes. The neurological manifestations of WD usually present with dysarthria and tremors of hands and in the juvenile age group presentation include personality changes, drooling of saliva, dysphagia, dystonia, clumsiness of hands, abnormal gait and fall at work in school.[5]

The overall reported prevalence of neurological features in WD widely varies.[6] As per studies, dystonia is seen in at least one third of patients with neurological manifestations of WD. Range of severity of dystonia can vary from focal, segmental, multifocal, or generalized. A common focal manifestation seen is dystonic facial expression known as risus sardonicus. Other types of movement disorders in WD include tremor, parkinsonism, chorea, and ataxia.[3] Neurological abnormalities in patients with WD usually present in the second or third decade as: (1) an akinetic-rigid syndrome resembling parkinsonism; (2) postural and intention tremor with ataxia, titubation and dysarthria "pseudosclerosis"; or (3) a generalized dystonic syndrome. And few patient seem to develop dystonia in later course of disease despite adequate treatment.[2] The deposition of copper in the lenticular nuclei, brainstem and cerebellum could contribute for neurological features of WD.[9] A study by International Parkinson's and Movement Disorder Society has reported dystonia in 36% of patients with WD. Many patients had mixed dystonia and other neurological manifestations. Generalized dystonia was observed only in four patients (10%).[8]

The patient showed early pediatric age onset of severe generalized dystonia with less hepatic involvement suggesting that neurological symptoms can manifests in pediatric age group with minimal liver involvement Hence, it is crucial to evaluate for WD in paediatric patients present.

CONCLUSION

In recent years, WD has been seen with more atypical and rare manifestation which further necessitates a need for high index of suspicion for detection of WD. As there are effective treatment available for Wilson's disease, early disease recognition and treatment is of paramount importance which could limit the development of long term disability. Wilson's disease should be considered in adolescents and young adults presenting with following features: (1) elevated liver enzymes found incidentally or in the context of an acute episode of hepatitis; (2) dysphagia or dysarthria not explained by another neurological disorder; (3) any type of unexplained

movement disorder; (4) psychiatric symptoms with liver disease; (5) adolescents with mood disorders and minor elevation of liver transaminase; (6) Coombs-negative hemolytic anemia; and (7) unexplained liver cirrhosis or hepatic failure.

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