Case Report

Wilson disease in a Bangladeshi child: A case report

Akhter S[1], Kabir MS[2], Majumder S[3], Ferdousy SA[4],Enamullah CAHM[5], Chowdhury N[6], Gaffar F[7]

Abstract

Wilson disease, also known as hepatolenticular degeneration results from improper metabolism of copper due to deficiency or low level of caeruloplasmin which causes excessive accumulation of copper in liver, brain, eyes and different organs. This is not a curable disease. The patient needs a lifelong treatment but early diagnosis can prevent significant damage to the critical organs. This article presents a case study on Wilson disease and the role of radiology in helping to diagnose the disease and monitor patients with this condition.

Key words : Wilson disease, children, copper metabolism

Introduction

Wilson disease is an autosomal recessive genetic disorder resulting in a systemic overload of copper. This manifests as neurological or psychiatric symptoms and liver disease. It is seen in individual ranging from 3 to over 50 years of age and symptoms vary among individual and within families. The reported prevalence is 30 per million people.[1]

Case Report

A 15 years old boy was admitted into Shaheed Suhrawardy medical college hospital with the complaints of difficulty in speech for 2 years, abnormal movements in upper limbs for 6 months and enlargements of both breasts for 2 years. According to the statement of the patient's mother the boy was reasonably well two years back then he slowly developed difficulty in speech which is persisting and gradually deteriorating. His mother also noticed abnormal repeatative dancing purposeless movements in upper limbs for last 2 years and inability to grasp any object and wear his cloths without the help of others. Gradually the patient developed an abnormal rigid posture with both arms directed backwards and outwards and lack of balance. During walking he failed to maintain a straight line. His mother also complaints of behavioral changes manifested as occasional sudden emotional outburst and poor performance in his school; gradual painless enlargement of both breasts without any secretion for 2 years.

With the above complaints, he was admitted in Shaheed Suhrawardy medical college hospital on 22/06/2015 for better evaluation and management. His past illness revealed repeated attacks of jaundice 3-4 times which was persistent for 10-15 days in last five years and subside spontaneously. The boy was born by normal vaginal delivery at home with no perinatal complication. His milestones of development were normal.

Family history revealed that he is the second issue of non consanguineous parents. There was no history of similar illness among his siblings. He was immunized as per EPI schedule. He came from low socioeconomic condition. On general examination, the patient was mildly anemic, normotensiveand and nonecteric. His pulse, respiratory rate and body temperature were normal. No signs of cyanosis, clubbing was seen. Patient had bilateral gynaecomastia. On palpation, his breasts were mildly tender and no secretion was present. Neurological examination revealed dysarthria and mild emotional lability. Patient was well oriented with cooperative behaviors and unusually laughing face. There were choreoathetosis, dystonic and hypertonic movements in upper limbs with abnormal rigid posture and slight altered gait. Cranial nerve examination showed no abnormality including normal fundus of eye and jerks of lower limbs. However jerks could not be elicited in upper limbs due to abnormal movements.

1. *Dr Shamima Akhter, Assistant Professor, Department of Radiology and Imaging, Shaheed Suhrawardy Medical College, Dhaka
2. Dr Md Showkat Kabir, Assistant Professor, Department of Community Ophthalmology, Bangabandhu Sheikh Mujib Medical University, Dhaka
3. Dr Shibendu Majumder, Professor and Head, Department of Radiology and Imaging, Shaheed Suhrawardy Medical College, Dhaka
4. Dr Sultana Amena Ferdousy, Junior Consultant, Radiology and Imaging, ICH and Shishu Hospital, Dhaka
5. Dr C A H M Enamullah, Associate Professor, Radiology and Imaging, National Medical college, Dhaka
6. Dr Nirupam Chowdhury, Medical Officer, Department of Community Ophthalmology, Bangabandhu Sheikh Mujib Medical University, Dhaka
7. Dr Faria Gaffar, Medical Officer, Department of Radiology and Imaging, Shaheed Suhrawardy Medical College,Dhaka

**For correspondence*

He was reviewed by neurologist who pointed the possible presence of Kayser-Fleischer (KF) ring on both eyes. A slit lamp examination by an ophthalmologist promptly revealed greenish brown discoloration of corneal region at sclera-corneal junction (in peripheral part of Descement membrane) of both eyes suggesting Kayser-Fleischer ring in different illumination and anterior sunflower cataract (Figure-1).

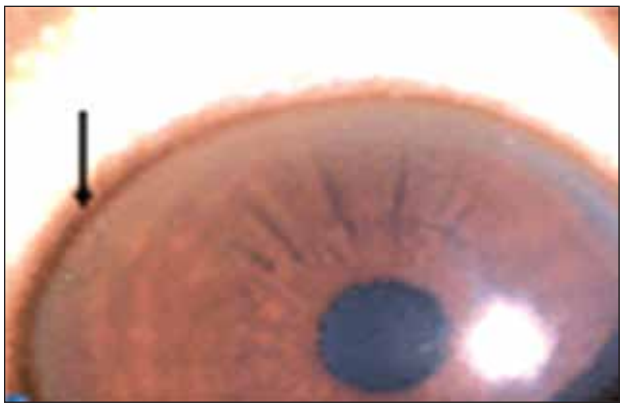

Figure- 1: *KF ring in slit lamp examination*

Urinary and serum copper levels of the patient were diagnostic of Wilson disease. (Table-I)

Table -I : Urinary and serum copper levels

Test	Result	Reference value
Urinary Copper (cu)	230μgm/ L	Concentration above 100 μgm/ L /24h (1.6 mmol/L/24h) confirm Wilson's disease
Conc. of Caeruloplasmin	14mg/dl	(15-60mg/dl)
Serum copper	70 μgm/dl	Male - 70-140 μgm/dl Female - 80-155 μgm/dl)

His Endoscopy of upper GIT and ECG examination revealed no abnormality, Echocardiography showed normal 2D/M-mode echo with good LV systolic function. Abdominal ultrasonogram scan showed coarse hepatic parenchyma. Breast sonography showed prominent fibroglandular tissue in both breasts.

Non contrast CT scan showed bilateral symmetrical hypodense areas in both thalami, basal ganglia and midbrain. Ventricles were mildly dilated (Figure-2.

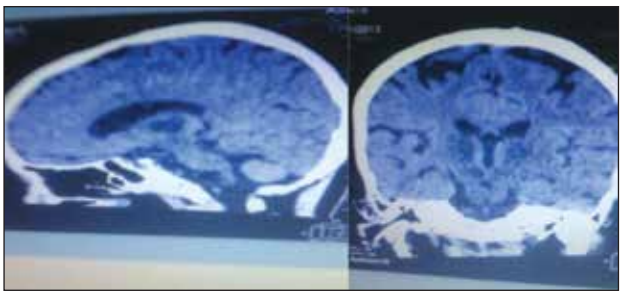

Figure-2: *Non contrast CT scan*

MRI of Brian revealed symmetrical hypointense on T_1 and hyperintense areas on T_2 & FLAIR at bilateral capsulo-ganglionic regions and thalami. Hyperintense areas are also noted at posterior aspect of mid-brain & pons (Figure-3).

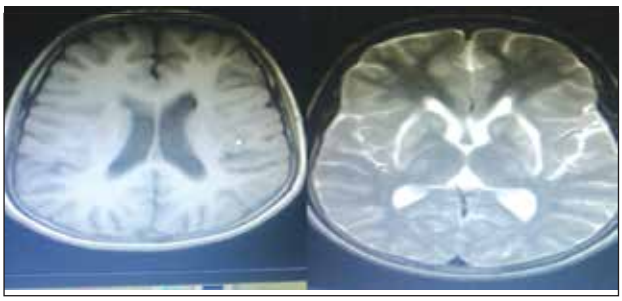

Figure-3: *MRI of Brian*

Discussion

Wilson disease is mostly reported in developed country. Children with Wilson disease are usually normal at birth and may remain healthy for variable periods of times. Most cases have been reported in developed countries.[2-4] The higher frequency of detection in North Africa compared with the rest of Africa may be explained by the higher rate of consanguinity in North Africa, which makes an autosomal recessive disease more likely to occur.[5]

The neurological features of Wilson disease are primarily due to the deposition of copper in the lenticular nuclei, although areas like the brainstem and cerebellum can be affected.[6] Most cases present in 2nd and 3rd decade of life. The disease presented with a variety of signs and symptoms like jaundice, dysarthria, clumsiness, tremor, gait distur- bances malaise and arthralgia. The mean age of onset is 12 years. Hepatic and osteo-arthral symptoms develop early and neurological symptom late.[7] A higher mortality rate was observedinhepatic, hepato-hematological and hepato-renal cases mainly due to acute hepatic failure resulting death within few weeks after onset. Cases having onlyneurological symptoms showed a more favorable prognosis with a long survival.[8]

The screening tests in suspected cases are examination for KF ring, ultrasound examination of liver, serum copper/ caeruloplasmin and 24 hour urinary copper especially in asymptomatic siblings of index cases.[9] Early molecular genetic and biochemical studies can be done to confirm diagnosis.

Natural course of neurological form of Wilson disease with limited liver disease may have better life expectancy[10]. Prognosis is fatal if remain untreated. D-Penicillamine is the drug of choice and orthotopic liver transplantation is life saving.[11,12] Reported complication of Wilson disease are hepatocellular carcinoma and cholangiocarcinoma.[13]

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