

Case Report

Wilson's Disease

Hossain MA¹, Hasan R²

Abstract

Introduction: Wilson's Disease referred to as hepatolenticular degeneration with excessive copper retention involving liver and brain. Hepatic manifestations predominate in children and neuropsychiatric manifestations predominate in adolescents, adults.

Case presentation: This is a case report of 24 years old female with long standing difficulty in walking and speech. Computerized tomography imaging showed bilateral symmetrical hypodensity in lentiform nucleus of basal ganglia, thalamus due to copper deposition and slit lamp examination showed Kayser-Fleischer (KF) ring surrounding limbus cornea.

Conclusion: Professional counselling and support in addition to drug treatment provide help to patient and her family to overcome their problems and improve the treatment outcome.

Keywords- Hepatolenticular degeneration with copper toxicosis.

Introduction

Wilson's disease is an autosomal recessive disorder caused by mutation in the ATP7B gene, with resultant impairment of biliary excretion of copper. Subsequent copper accumulation, first in the liver but ultimately in the brain and other tissues. Clinical manifestations that may include hepatic, neurological, psychiatric, ophthalmological and other derangements. Genetic testing is impractical because of the multitude of mutations that have been identified, so accurate diagnosis relies on judicious use of a battery of laboratory and other diagnostic tests. Lifelong palliative treatment with a growing stable of medications or with liver transplantation if needed, can successfully ameliorate or prevent the progressive deterioration and eventual death that would otherwise inevitably ensue.

Case presentation

Mrs. Hamida Khatun, 24 years old, housewife from Chowhali, Sirajgonj was attended in OPD, Department of Medicine, Khwaja Yunus Ali Medical College & Hospital with the complaints of difficulty in walking & speech for last 3 years which was more severe for last 1 month. She also complained emotional disturbance for 1 month & loss of weight for 6 months. On examination tremor was present, Kayser-Fleischer (KF) ring (the brown ring on the edge of the iris) was present which

was greenish brown discolouration of the corneal margin seen by slit lamp examination. Choreaathetosis, dementia & dystonia was also present. The diagnosis Wilson's disease was given to her based on clinical signs & radiological findings. Her liver function test, renal function test was normal and copper concentration in serum ceruloplasmin was elevated.



Fig:1

1. Dr. Md. Alamgir Hossain, Associate Professor, Dept. of Radiology and Imaging, KYAMCH, Enayetur, Sirajgonj, Bangladesh.
2. Dr. Rashidul Hasan, Assistant Professor, Dept. of Dermatology, KYAMCH, Enayetur, Sirajgonj, Bangladesh.



Fig:2

Fig:2 Computerized tomography (CT) has been performed and the typical features of Wilson's disease was revealed and showed bilateral symmetrical hypodensity in lentiform nucleus of basal ganglia and thalamus due to copper deposition.

Discussion

Wilson's disease is a rare autosomal-recessive disorder. A prevalence rate of 30 cases per million (one per 30,000) and a birth incidence rate of one per 30,000 to 40,000 are often quoted. A mutation in the ATP7B gene, located on chromosome 13, is responsible for Wilson's disease. The number of specific mutations that have been identified is now approaching 300. Although missense mutations are most frequent, deletions, insertions, nonsense and splice site mutations all occur. Most affected individuals are actually compound heterozygotes, having inherited different mutations from each parent. Age of onset is 7-50 years, hepatic manifestations predominate in children, neuropsychiatric manifestations predominate in adolescents and adults. Copper is an essential element for cellular function, yet free copper is extremely toxic and can produce irreversible cellular damage. To cope with this, elegant systems have evolved that bind the

copper molecule to ensure safe transport of necessary copper to intended sites and safe elimination of excess copper through the biliary system. Although the fundamental pathogenetic defect of Wilson's disease is abnormal copper metabolism but it shows some hepatic, neurological, psychiatric & ophthalmological manifestations. Patient may also presented as osteoporosis, hemolytic anemia, renal tubular dysfunction with consequent hypercalciuria & hyperphosphaturia and skin pigmentation. The diagnostic tools for Wilson's disease are serum ceruloplasmin, serum copper or free copper level, liver biopsy, neuroimaging (MRI & CT scan) and slit lamp examination of eye. With the exception of liver transplantation, treatment of Wilson's disease is only palliative and intended to restore and maintain copper balance.

Conclusion

The copper binding agent, Penicillamine is the drug of choice and the other options are trientine dihydrochloride & zinc. Abrupt discontinuation of treatment must be avoided because this may precipitate acute liver failure. Liver transplantation is indicated for fulminant liver failure or for advanced cirrhosis with liver failure. Although it is a life long disease but the prognosis is excellent, provided treatment is started before there is irreversible damage. Siblings and children of the patient with Wilson's disease must be investigated and treatment should be given to all affected individual, even if they are asymptomatic.

References

1. Thomas-Sohl KA, Vaslov DF, Maria BL: Wilson's Disease: a review. *Pediatr Neurol* 2004, 30:303-10.
2. Castillo M: Neuroradiology companion Lippincott W&W, Philadelphia; 2006:231-233.
3. Barsky SH, Rosen S, Geer DE, Noe JM: The nature and evaluation of Wilson's Disease: A computer assisted study. *J Invest Darmatol* 1980, 74: 154-157.
4. Takeoka M, Riviello JJ: Wilson's Disease [<http://emedicine.medscape.com/article/1177523-overview>].
5. Comi AM: Pathophysiology of Wilson's Disease. *J Child Neurol* 2003, 18:509-516.
6. Osborn A, Salzman K, et al.: Diagnostic imaging: Brain Salt Lake City: Amyrsis; 2004. I-1-94-97

7. Sujanski E, Conradi S: Outcome of Wilson's Disease in 52 adults. *Am J Med Gen* 1995, 57(1):35-45.
8. Rochkind S, Hoffman HJ, Hendrick EB: Wilson's Disease: natural history and prognosis. *J Epilep* 1990, 3:293.
9. Zhuo BY, Lu GJ, Ye ZZ: A case of Wilson's Disease. *Zhonghua Er Ke Zq Zhi* 2004, 42:944.
10. Khan NA, Tunbull I, MacDonald S: Wilson's Disease. [<http://emedicine.medscape.com>].