A Case Report on Wilson's Disease

Karisetty Basappa, G.Surender, Dr. (Mrs) M.Lahkar, Dr. Urmi Choudhury
National Institute of Pharmaceutical Education and Research, Andhrapradesh

Abstract: Wilson's disease is an autosomal-recessive disorder of copper metabolism resulting from the absence or dysfunction of a copper-transporting. The disease is mainly seen in children, adolescents and young adults, and is characterized by hepatobiliary, neurologic, psychiatric and ophthalmologic (Kayser-Fleischer rings) manifestations. The deposition of copper in tissues is the cause of virtually all the manifestations of the disease in Liver, Blood, Kidney and Brain. We present here a case study of Wilson's disease in 12 years old children. [Basappa K et al NJIRM 2013; 4(6) :143-144]

Key words: Dystonia, Kayser-Fleischer ring, Trientein hydrochloride, Wilson's disease.

Author for correspondence: Karisetty Basappa, Department of pharmacy practice, NIPER- GUWAHATI (Govt of India), Assam. E-mail: basappa.niperian12@Gmail.Com

Introduction: Wilson's disease (WD), also known as hepatolenticular degeneration was first described in 1912 by Kinnear Wilson as progressive lenticular degeneration. WD is an inherited, fatal neurological disorder accompanied by chronic liver disease leading to cirrhosis, [1] and accumulation of copper in the liver, brain because of defective biliary excretion.[2, 3] Wilson's disease is due to mutations of the ATP7B gene on chromosome 13.[4,5]

Case details: A 12 years old male patient presented with complaints of difficulty in speaking from last 3-months, dystonia from last 4 days, abnormal posture of lower limbs & tongue from last 3months.

Past history: The patient past history narroted by patient mother is as follows: He was alright approximately last 3 months back when he had an episode of fever and received medication by a private practitioner. The fever subsided after meditation, but patient had developed loose motion, difficulty in speaking and pronouncing linguals. With these compliants he was admitted in the hospital. On Radio imaging and ophthalmic examination he was diagnosed as a case of Wilson's disease and patient was started with tablet ZINCOLAC (calcium Pantothenate) and tablets DISTAMINE (D-Penicillamine) and was discharged.

After a few days patient was again admitted in the hospital with a chief complaint of abnormal behaviour and increased dystonia which progressed to involve whole left lower limb, right foot, tongue and right upper limb. At that time patient was adviced to hold DISTAMINE tablet but he was not releaved of dystonic symptoms though the behavioural symptoms decreased.

On Examination: Patient was conscious, No clubbing or cyanosis .No icterus or pallor. There was no any abnormality in cardiovascular, respiratory or gastrointestinal system. On examination his B.P was 130/80 mm Hg, Pulse rate was 90/min.

Investigations:

1.Haematological investigation revealed

- Haemoglobin % - 16%,
- Erythrocyte Sedimentation Rate: 43
- Albumin: 3.3 g/dl
- Serum Creatinine: 0.36 mg/dl
- UREA (B): 19 mg/dl
- Alanine Transaminase : 65 IU
- Aspartate Aminotranferase: 88IU
- Alkaline phosphatase : 242 IU/lit
- Creatinine Phosphokinase: 864 IU
- Serum ceruloplasmin: 374 mg/lit
- Copper: 46 µmol/dl

2. Slit Lamp Examination- KAYSER FLEISHER RING is present.

Treatment: After confirming diagnosis patient was being treated with Trientein hydrochloride 300 mg three times a day (TDS), Benzhexol 2 mg four times in a day (QDS). Follow up is being taken regularly for next six months. Patient is gradually improved and still continuing treatment.

Reference:

1. Wilson SAK. Progressive lenticular degeneration: a familial nervous disease associated with cirrhosis of the liver. Brain 1912; 34:295-507.
2. Scheinberg IH, Sternlieb I. Wilson's disease. In: Smith Jr LH, editor. Major problems in internal medicine, vol. 23. Philadelphia, PA: WB Saunders; 1984. p. 25–35.
3. Gitlin JD. Wilson disease. Gastroenterology 2003; 125:1868–1877.
4. Tao TY, Gitlin JD. Hepatic copper metabolism: insights from genetic disease. Hepatology 2003; 37:1241–1247.
5. Lutsenko S, Petris MJ. Function and regulation of the mammalian copper transporting ATPases: insights from biochemical and cell biological approaches. J Membr Biol 2003; 191:1–12.

Conflict of interest: None
Funding: None