Wilson’s disease (WD) is an autosomal recessive disorder caused by a mutation in the ATP7B gene involving liver and brain and which results in impairment of biliary excretion of copper. About 47% and 55% of cases reported have positive family history and consanguinity, respectively [1]. A patient (5–40 years old) presenting with liver disease, with a decrease in the level of serum ceruloplasmin, and detectable Kayser–Fleischer (KF) rings in eyes are the main symptoms of WD [2]. Delay in diagnosis of WD is observed across all the healthcare levels [3]. WD will be fatal unless diagnosed in time. The main complications of WD include brain damage and liver cirrhosis, psychiatric disturbances, i.e., depression, suicidal tendencies, and aggressive behavior motor dysfunction and corneal opacities [4]. The following case report illustrates the importance of taking the proper medical history of patients presenting with different clinical conditions.

CASE REPORT

A 35-year-old male presented with severe pain, abdominal distension, and vomiting. He had a history of intermittent hematochezia over the past 2 months and also his relatives had noticed a change in the patient’s behavior (easily irritable) over the past 1 month. Initially, patient could perform his routine daily activities without any hesitation, but for the past 1 month, he found it to be difficult. He had no history of limb weakness, headache, seizures, difficulty in swallowing, chewing, and drinking water and also no history of any major illness or any major surgery. Birth history and vaccination history are normal. He was previously admitted and evaluated at 2–3 hospitals where he was diagnosed to have liver cirrhosis. Treatment for cirrhosis was given, but no progress was observed in his condition. Finally, he was referred to our tertiary care hospital. While taking patient history, we found out that he had a consanguineous marriage and also his brother died at a young age due to WD. Hence, we send investigations for WD, serum ceruloplasmin was found to be low, 24 hrs urine copper values was 75 mcg, and he was found to have KF rings in both eyes in ophthalmology consultation. Thus, he was diagnosed to have WD-induced liver cirrhosis with portal hypertension. He had Child–Pugh score C and model of end-stage liver disease score 23. Patient was having ascites but no signs of spontaneous bacterial peritonitis. He was started with WD-specific treatment; tablet Penicillamine 250 mg, tablet Ascasin (50 mg elemental zinc), tablet Bldiv 300 mg, tablet Rifagut 250 mg, Syrup Looz 30 ml, and other hepatoprotective drugs. However, due to late diagnosis, his condition became worst, and finally, liver transplantation was the last option. Later, his liver transplantation was done where his wife was the donor. To prevent rejection, T Azoran 100 mg/day and T Everolimus 0.25 mg were started. He had a positive response to treatment and is under recovery now.

DISCUSSION

WD or hepatolenticular degeneration is an autosomal recessive genetic disorder, in which copper accumulates in tissues. It is found all over the world, with a prevalence rate of approximately 1 in 30,000 live births in most populations. Since there is a decrease in biliary copper excretion, it leads to accumulation of copper in several organs, mostly the liver, brain, and cornea. Overtime, it may lead to liver cirrhosis. A small percent of patients found to develop acute liver failure, most often in the setting of advanced fibrosis of the liver. In addition, some patients may develop neurologic complications, which can be fatal. The chance of sibling and offspring being a homozygote and developing clinical disease is 25% and 0.5%, respectively. There is a higher incidence rate in Asians attributed to consanguinity. In India, the prevalence rate is not performed because of the paucity of studies; however, it is relatively common in South India because of more consanguineous marriage (55%). In India, there are less community-based incidence and reports (many are hospital based reports) although variation in epidemiology, clinical presentation, and course is reported [4].

In this case, patient’s condition becomes worst due to late diagnosis of WD. If proper history taking was done, there would be more chance of early diagnosis because he had a consanguineous marriage and his brother died due to WD. In WD, there is a failure of copper binding to ceruloplasmin. Hence, hepatic synthesis and secretion of ceruloplasmin protein without copper give rise to apoceruloplasmin, which has decreased half-life compared with ceruloplasmin resulting in decreased serum ceruloplasmin in WD. Over accumulation of copper in hepatocytes leads to spill into circulation and thus increases the copper content in blood and get deposited in various organs such as brain, kidney, cornea, and skeletal system [5]. Patient was treated for WD with D-penicillamine; it contains a free sulfhydryl group which acts as a copper chelating moiety. It helps to remove copper from less tightly bound sites on proteins, peptides, and membranes and promotes its urinary excretion. Along with that oral zinc supplements were given...
which interfere in copper absorption, providing a rationale for its use in WD. Zinc induces metallothionein (an endogenous chelator of metals) in enterocytes, which has a higher affinity for copper than for zinc, thus binds with the luminal copper and prevent it from entering into circulation. The bound copper is excreted fecally during normal turnover of enterocytes [6].

The main focus of this case is to show the importance of taking a medical history. Since the same genetic abnormality was the reason for the death of his elder sibling which was not considered before and this led to a late diagnosis. Similar case report “WD: A clinical autopsy case report with review of literature” also points out the importance of a premedication history that may avoid the delay in getting abrupt care and which can improve the patient condition and prevent fatality.

**CONCLUSION**

The patient assessment should be entertained with a premedication history presenting with liver dysfunction and/or extrapyramidal neurological features. This case also points to the importance of taking the proper medical history of the patients for providing better treatment and to improve the quality of life of the patients.

**REFERENCES**

1. Meenakshi-Sundaram S, Mahadevan A, Taly AB, Arunodaya GR, Swamy HS, Shankar SK. Wilson’s disease: A clinico-neuropathological autopsy study. J Clin Neurosci 2008;15(4):409-17.

2. Roberts EA, Schilsky ML; American Association for Study of Liver Diseases (AASLD). Diagnosis and treatment of Wilson disease: An update. Hepatology 2008;47(6):2089-111.

3. Stefano V, Lionetti E, Rotolo N, La RM, Leonardi S. Hypercalciuria and nephrocalcinosis as early feature of Wilson disease onset: Description of a pediatric case and literature review. Hepat Mon 2012;12(8):e6233.

4. Bandyopadhyay D, Ghosh D, Chattopadhyay A, Mitea E. Curry leaves as alternative medicine in heavy metal induced occupational health hazards. Int J Pharm Pharm Sci 2016;8(8):8-20.

5. Raju K, Bangalore GN, Thuruvekere SN, Pathavanalli VN. Wilson’s disease: A Clinical autopsy case report with review of literature. J Nat Sci Biol Med 2015;6(1):248-52.

6. Egberto R, Alexandre A, Costa M, Eduardo L, Rachid C, Marta M. Wilson’s Disease: A case report and a historical review. Arq Neuropsiquiatr 2009;67(2-B):539-43.

| Lab investigations       | Result     | Normal levels       |
|--------------------------|------------|---------------------|
| Haemoglobin              | 9.66 g/dl  | 13.0-17.0 g/dl      |
| Platelets                | 115 K/uL   | 150.0-450.0 K/uL    |
| Prothrombin/partial      | 14.60 seconds | 11-14 seconds     |
| thromboplastin time      |            |                     |
| Total bilirubin          | 4.20 mg/dl | 0.2-1.2 mg/dl       |
| Serum albumin            | 1.83 g/dl  | 3.5-5.2 g/dl        |
| Total protein            | 7.64 g/dl  | 6.6-8.3 g/dl        |
| Blood urea               | 29.9 mg/dl | 17.0-43.0 mg/dl     |
| Serum creatinine         | 1.16 mg/dl | 0.84-1.4 mg/dl      |
| SGOT                     | 153.1 IU/L | 5.0-35.0 IU/L       |
| SGPT                     | 69.4 IU/L  | 5.0-45.0 IU/L       |
| Alkaline phosphatase     | 102.5 IU/L | 13.0-120.0 IU/L     |

SGOT: Serum glutamic oxaloacetic transaminase, SGPT: Serum glutamic pyruvic transaminase

Table 1: Significant laboratory investigation parameters of the present case