A CASE REPORT ON WILSON’S DISEASE-INDUCED LIVER CIRRHOSIS

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INTRODUCTION

Wilson’s disease (WD) is an inherited autosomal genetic abnormality which results in impairment in cellular copper transport. Overtime, this may lead to liver cirrhosis. The main focus of this case is to show the importance of taking a medical history. Here, we discuss a case of a 35-year-old male diagnosed with WD-induced liver cirrhosis and portal hypertension. He was physically very weak. Since the same genetic abnormality was the reason for the death of his sibling which was not considered while taking the medical history of this patient, this led to a late diagnosis of 4 years while the patient’s condition became worst. Herein, we report a case that provides an insight to medical professionals about taking proper medical history of patients

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 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 Ascazin (50 mg elemental zinc), tablet bidiliv 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.

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 is 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 spilt 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 sulphydryl group which acts as a copper chelating moiety. It helps to remove copper from less tightly bound sites on proteins, peptidases, 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