CASE REPORT

PREGNANCY WITH WILSON'S DISEASE - A CASE REPORT

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ABSTRACT

Wilson’s disease is an autosomal recessive disorder leading to an inherited deficiency in copper binding protein (Ceruloplasmin). This leads to decreased copper excretion into the bile by the liver leading to copper accumulation and copper toxicity which can result in liver disease, and in some patients, brain damage. It is an effectively manageable disorder and early recognition and treatment can help in preventing disease progression. We are reporting a case of pregnancy in a woman already diagnosed with Wilson’s disease and discussing management of pregnant woman with this disorder.

KEY WORDS: Wilson’s disease & pregnancy, hepato lenticular degeneration, chronic liver disease.

INTRODUCTION

In 1912, Dr. Samuel Kinnier Wilson described this disorder¹. It involves deficiency of Ceruloplasmin (a copper binding protein), thus leading to increased deposition of copper in various organs of body. It is inherited as an autosomal recessive trait and has many different mutations which may present differently. Commonest is the mutation on chromosome 13 at the site 13q14.3-q21.1 & is particularly characterized by hepatic and neurological disease.

It presents as liver disease in children & adolescents while as neuropsychiatric illness in young adults. It should be considered in any individual of less than 40 years of age with unexplained chronic liver disease. Hepatic dysfunction is usually the first feature, & by the time they present with neurological features they already have liver cirrhosis. Recurrent abortions are seen to be associated in women with untreated Wilson’s disease. But properly managed women can have uneventful outcomes of their pregnancies²,³.

CASE REPORT

A 34 years old Emirati lady was referred to Obstetrics/Medicine clinic in our hospital at 13+weeks of gestation in her first pregnancy. She was a known case of Wilson’s disease diagnosed at 8 years of age as a result of family screening after a sibling died in childhood with liver failure secondary to Wilson’s disease. She was identified as having homozygous mutation in intron 12 c.2866-2A>G of the ATP7 gene.

The index pregnancy was a spontaneous conception after 06 months of marriage. She had no history of consanguinity. She was receiving penicillamine treatment at the time of conception but was changed to Zinc oxide 20 mg BID at 5+weeks by her gastroenterologist. She was monitored in a combined Obstetric medicine clinic. Her International normalized ratio (INR) and liver function tests were checked regularly and remained within normal limits throughout pregnancy. The pregnancy course remained uneventful. She had a spontaneous vaginal delivery at 38+5 weeks and gave birth to a healthy female baby of 2660 g in weight.

The infant was genetically screened and was found to be a carrier. Genetic screening detected the known familial mutation at intron 12 of the ATP7B gene (c.2866-2A>G) in the heterozygous state. Father also got himself screened & was found to have no pathological mutation for the same mutation.

DISCUSSION

Worldwide prevalence of WD is estimated to be 30 per million. In Sardinia a higher prevalence is noted and approximately 10-12 new cases per year are identified¹. It can present clinically as liver disease, a progressive neurological disorder (hepatic dysfunction being less apparent or occasionally absent), or as psychiatric illness. WD presents with liver disease more often in children and younger adult patients than in older adults. In children symptoms begin to appear by age 4. Fulminant presentation is more common in women and typically appears before 40 years age. Recurrent abortions are seen to be associated in women with untreated Wilson’s disease.²,³

It was previously considered as a fatal disorder but effective pharmacologic treatment has greatly influenced the management of WD. In 1951 the first chelating agent was introduced for the treatment of WD- British anti-lewisite (BAL or dimercaprotopropanol) ⁴,⁵. In 1956, identification and testing of D-penicillamine, an orally administered chelator by John Walsh revolutionized treatment of this disorder⁶. Other treatment modalities introduced, include zinc salts⁷,⁸ to block enteral copper absorption, tetrathiomolybdate (TM) to chelate copper and block enteral absorption, and orthotopic liver transplantation, which may be lifesaving and curative for
REFERENCES

7. Hoogenraad TU, Koevoet R, de Ruyter Korver EG. Oral low dose penicillamine and zinc sulphate are not seen to be associated with teratogenicity and are safe in pregnancy. Mother should be informed about it to ensure compliance with medicines.