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Case Report

Pregnancy in wilson's disease

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ABSTRACT

Wilson's disease is a rare autosomal recessive disorder with mutation of ATP 7B on chromosome 13q14 which leads to impaired biliary excretion and ceruloplasmin incorporation causing copper accumulation mainly in the liver and brain. This accumulation results in liver cirrhosis and nervous system manifestations such as neuropsychiatric symptoms, movement disorders and ataxia. Untreated Wilson's disease usually causes subfertility and in cases where pregnancy does occur, it often results in spontaneous miscarriage. However, therapeutic evolution in the past decades has resulted in multiple successful pregnancy outcomes in patients with Wilson's disease. We report such a case of successful pregnancy outcome in a women with Wilson's disease.

1. Introduction

Wilson's disease is an autosomal recessive genetic disorder affecting copper transport leading to hepatic and/ or neuropsychiatric manifestation. Untreated Wilson's disease in females may cause subfertility or spontaneous miscarriage. Pregnant women need close monitoring and multidisciplinary management. Anticopper therapy during pregnancy and breastfeeding are safe. Treatment should be maintained during pregnancy and pregnant women should be treated by a multidisciplinary team. With adequate medical treatment and close monitoring before and during pregnancy, a successful outcome of mother and newborn can be achieved.

2. Case Report

A 24 year old primigravida, presented to our antenatal clinic for the first time at 37.3 weeks of gestation in latent phase of labor. She was diagnosed to have Wilson's disease at the age of eighteen years in 2018 when she started having symptoms of altered behaviour, irrelevant talks and agitation for which she was on antipsychotic treatment for 1 year. But later on in 2019, she developed bilateral tremors of upper and lower limb associated with rigidity, stiffness and difficulty in walking. Serum copper and ceruloplasmin levels were then obtained which were 500 μg/dL and 0.07g/L, respectively (Normal range S. Copper: 85-180 microgram/dl, S. ceruloplasmin :0.2-0.6 g/dl. She had her ANC check up done only once.

She had one antenatal scan suggestive of single live intrauterine gestation of 33.6 weeks of pregnancy, cephalic presentation, placenta posterior, AFI adequate. In antenatal workup all ANC investigations such as hemogram, blood group, virology (HIV, HCV, HBsAg, VDRL), S.TSH, Urine-routine and microscopy, blood glucose were done. All other ANC investigations were found to normal except she was found to be Rh negative (O- negative). Her MRI brain showed abnormal T2 /FLAIR intensities on bilateral basal ganglia, thalami and brainstem along with bilateral caudate and putamen atrophy suggestive of Wilson's disease. On reviewing her records she had stopped her medications on her own 9 months back. She was on tab. penicillamine initially and later on she was prescribed tab Zinc Sulphate 50 mg twice daily along with antipsychotic

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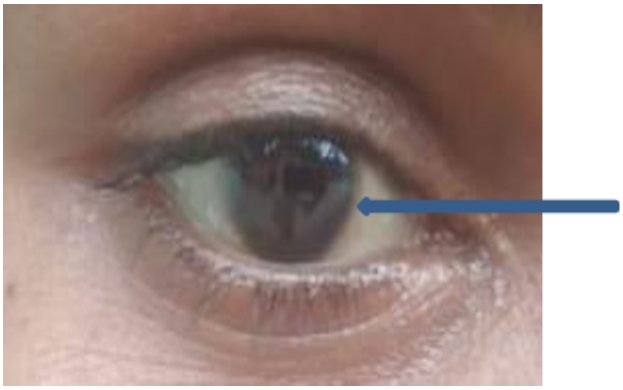

Figure 1: Opthalmology examination kayser fleischer (KF) ring present in wilson's disease

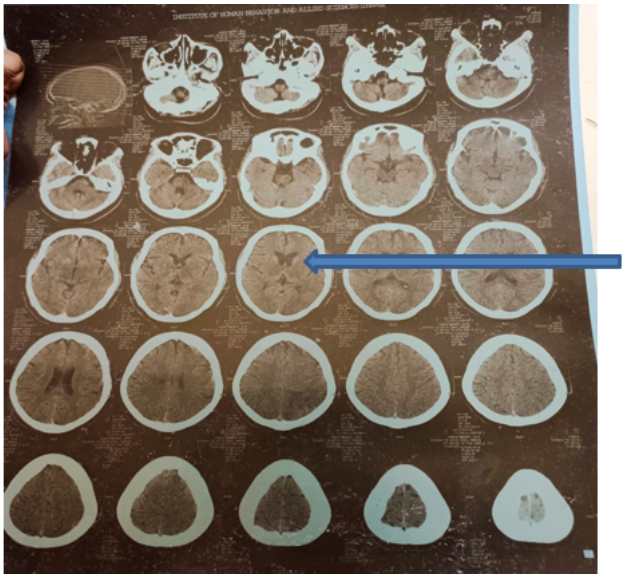

Figure 2: MRI (brain) bilateral caudate and putamen atrophy suggestive of wilson's disease

drugs according to her old medical prescriptions. She presented in latent phase of labor with irritability and altered behavior for which the neurology team was consulted. The dose of tab zinc sulphate was increased to 50 mg thrice a day along with tab Trihexyphenidyl 2mg once a day, tab pyridoxine 40 mg once a day, tab lorazepam 2 mg once a day and tab aripirazole 5 mg once a day after which the neurological symptoms subsided gradually. Ophthalmologic examination revealed Kayser-Fleischer rings. Serum copper and ceruloplasmin levels were then obtained which were normal. Maternal ultrasound of the upper abdomen revealed inflammatory changes in the liver with echogenicity of the parenchyma along with mild splenomegaly. Renal and liver function tests were within normal limits except for S. ALP which was raised upto 556.7U/L. Intra-partum course of events was unremarkable and a healthy female weighing 2.5 kg with a good Apgar score was delivered vaginally.

3. Discussion

Wilson's disease is an autosomal recessive inherited disorder of human copper metabolism. The underlying molecular defect is a dysfunction of P -type ATPase ATP 7B on chromosome 13q14 which is essential for copper transport across cellular membranes.[1] The incidence of this autosomal recessive disease is reported to be about 1 in 30000. Impaired Biliary copper excretion leads to pathological copper accumulation in liver, brain and other tissues. Clinical manifestation of patients are variable. Some patients are asymptomatic, others experience acute or chronic liver failures or neuropsychiatric deteriorations. Women with Wilson's disease may require infertility treatment but many patients conceive spontaneously as seen in our case report.[2] Wilson's disease, if not treated promptly, can lead to significant morbidity and can be potentially fatal. Nowadays Zinc is increasingly being used as a therapeutic option in managing Wilson's disease.[3] Zinc interferes with absorption of copper from gastrointestinal tract by induction of intestinal cell metallothionein which has a higher affinity for copper and prevents serosal transfer of copper into blood.[4] Also, Zinc seems to have an excellent safety profile with no congenital abnormalities being found. In maternal outcomes, these patient might be at risk for other complications including pregnancy induced hypertension or preeclampsia, placental abruption and thrombocytopenia and deranged coagulation.[5] Such complications were not found in our case.

4. Conclusion

Patients with Wilson's disease receiving regular treatment who remain asymptomatic are usually able to conceive with successful outcomes. Zinc sulphate is an effective therapeutic option and can be safely used in managing patient with Wilson's disease throughout the pregnancy. With proper medical treatment, good compliance of patients and interdisciplinary monitoring of pregnancy, successful outcome for mother and newborn can be expected.

5. Source of Funding

None.

6. Conflict of Interest

None.

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