



A Rare Case of Spontaneous Conception in Untreated Wilson's Disease

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ABSTRACT

Wilson's disease is a rare, autosomal inherited disorder of abnormal ATP7B gene resulting in excessive accumulation of copper in the liver and brain. It is suspected primarily in patients with degrees of liver failure or neuropsychiatric manifestations of unknown etiology in persons below 40 years of age. Diagnosis may be strengthened by presence of Kayser-Fleischer rings, serum ceruloplasmin < 20 mg% or high 24 hours urinary copper. The gold standard is however liver biopsy. Wilson's disease usually leads to infertility or repeated miscarriages in females. It occurs in approximately 1:30,000 individuals with a carrier frequency of about 1 in 90. Here we present a rare case of spontaneous, consecutively successful pregnancies in a female with untreated Wilson's disease.

Keywords- *Wilson's disease, Pregnancy, Infertility, Miscarriage.*

INTRODUCTION

Wilson's disease is an inborn error of metabolism with autosomal recessive inheritance. It is characterized by excessive copper deposition in the liver and brain as a result of mutation in the copper transporting ATP7B gene, anywhere along the entire 21 exons, which makes identification of gene defects particularly challenging [1]. It occurs in approximately 1:30,000 individuals with a carrier frequency of about 1 in 90 [1].

CASE

A 25 year old fourthgravid with previous 3 living issues of 10, 5 and 3 years (all males) reported with complaints of acute increase in a previously distended abdomen accompanied with pain and history of abortifacient intake (for a 3 month gestation), 1 week back. On examination, her positive findings were pallor with massive abdominal as cited, rest normal.

Investigations revealed Hb 6.6gm%, platelets 59,000/ μ L, random blood sugar 101mg/dl. Liver function test; ALT/AST:52/64, Total bilirubin 0.2mg% (direct 0.1, indirect 0.1), Alkaline phosphatase 122U/L, total protein 5.6gm%, albumin 2.6gm%. Prothrombin and Activated partial thromboplastin time were mildly elevated. Renal function test was normal. Ascitic fluid microscopy showed total protein 0.5gm/dl, albumin 0.2gm/dl, sugar 93.5mg%, total count 200 cells/ μ L with 50% lymphocytes (transudate). No organisms or AFB were seen. ADA was 13.8U/L. VDRL, HIV and all other viral markers were negative. Ultrasound showed coarse echotexture, irregular surface of liver, moderate splenomegaly, dilated portal vein, gross ascites but no mesenteric lymph nodes and a single live intrauterine pregnancy of 13 weeks + 2 days. Upper GI endoscopy revealed early esophageal varices. On grounds of presence of chronic liver disease of unknown origin in a young, non-alcoholic female, she was sent for Kayser-Fleischer ring examination which were absent. However, she had low serum ceruloplasmin levels, 15mg% (18-35mg/dl, Harrison's Principles of Internal Medicine) and 24 hour urinary copper was 92.8 μ g (reference range 20-50 μ g, Harrison's Principles of Internal Medicine).

A diagnosis of Wilson's disease was made. Due to history of abortifacient intake and her abdominal distress, the pregnancy was terminated and a subsequent liver biopsy advised.



DISCUSSION

Majority of patients with Wilson's disease present with hepatic (early) or neuropsychiatric (late) manifestations [1]. Kayser-Fleischer rings provide unequivocal proof but are not mandatory for diagnosis as they can be absent in 50% of hepatic cases [1]. Untreated Wilson's disease causes subfertility [2,3], as also evidenced by use of copper intrauterine devices for contraception. In patients with cirrhosis this is probably because of associated metabolic and hormonal changes. Other manifestations may include repeated miscarriages [3,4]. Our case however was a multigravida with previous 3 live, normal children and present pregnancy, all achieved spontaneously. The current

pregnancy was terminated keeping in mind patient's well-being and future treatment. There are very few instances of spontaneous conception in untreated Wilson's disease with pregnancies carried till term without any complications. Treated Wilson's diseases, however, poses no such risk except for an element of teratogenicity when on copper chelating drugs. Treatment with Zinc, which decreases absorption of copper from the intestines, again obviates this problem^[1].

In conclusion, such cases are probably the result of evolution of these genetic mutations towards survival, which otherwise will die out on their own. However, this hypothesis needs further elucidation.

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