



Case Report

Wilson's disease: An autopsy case report

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ABSTRACT

Wilson's disease is an autosomal recessive disease which result in defective copper metabolism, usually seen in young adults, predominantly affecting liver and brain. Although it is common in India, variation in epidemiology, clinical presentation and course are reported. However, community-based incidence and prevalence rates are not available in India and incidences are limited to hospital based reports. Most often, the diagnosis is delayed. We present a clinical autopsy case in a 12 year-old male who had presented with clinical symptoms at 10 years of age. Patient's parent had non-consanguineous marriage and patient is 3rd by birth order.

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1. Introduction

Wilson's disease (WD) is an autosomal recessive disease involving brain and liver secondary to altered copper metabolism. About 47% and 55% of cases reported have positive family history and consanguinity, respectively.¹ The liver disease may be asymptomatic, with only biochemical abnormalities of cirrhosis.^{1,2} A patient (5-40 years old) presenting with liver disease, with a decrease in serum ceruloplasmin and detectable Kayser – Fleisher (KF) rings are generally regarded as having classic WD.³ Delay in diagnosis of WD is observed across all the health care levels.⁴

2. Case Report

12 year old male boy born by Non consanguineous marriage 3rd by birth order came with complaints of fever, jaundice and altered sensorium since 01 day.

On General examination pallor, icterus, anasarca, ascites and depressed reflexes (Glasgow coma scale 3/15 (normal

i.e. Good chances of survival >8). No investigations were performed during this admission. Patient died within 35 minutes of admission.

Past contribution history is admission in hospital 2 times, during that time following investigations were done. Patient was never asymptomatic since then and continued similar complaints i.e. anasarca.

2.1. Investigations

1. Ultrasonography performed which was suggestive of liver cirrhosis with portal hypertension with moderate ascites
2. CT abdomen performed which showed similar features as in USG
3. Upper gastrointestinal scopy done which showed fundic varices

So, with all clinical and radiological details further investigations advised

On the basis of following biochemical investigations

1. Serum Copper-27 21mg/dl(10-15mg/dl.)
2. 24 hour urine copper-427 92mg(53 mg.)

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So with the clinical, radiological and biochemical investigations; diagnosed him as Wilson's disease

Patient put on D- penicillamine and patient was being followed up

Table 1:

Test	Findings	Normal range
Haemoglobin	9.4gm%	12-15gm%
WBC count	19600/mm ³	6000-11000/mm ³
Red blood cell morphology	Hypochromic microcytic	NN
Platelet count	80000/mm ³	1.5-4.0 lac/mm ³
Total bilirubin	2.2mg%	<01-0mg%
Direct bilirubin	1.4mg%	Up to 0.5 mg%
BUN	14.0mg%	10-15mg%
Sr. Creatinine	0.9mg%	01-02mg%

Table 2:

Test	Findings	Normal range
Serum copper	27.2mg/dl	10-15mg/dl
24 hr. urinary copper	312.38mg	<53.9mg
Serum ceruloplasmin	2.71mg/dl	30-65mg/dl
ALP	514IU/L	44-140IU/L

Consent for full body autopsy was taken from the family members. The salient features of autopsy were as follows. On external examination: Ascites, Icterus, Pallor, Brown ring identified at the junction of cornea and conjunctiva which is suggestive of Kayser Fleischer ring.



Fig. 1: Kayser Fleischer ring, pallor and icterus on external examination

2.2. Microscopy

1. Liver showed mixed nodular cirrhosis with hepatitis
2. Spleen showed congestion in red pulp
3. Lungs showed features suggestive of bronchopneumonia, pulmonary oedema and intrapulmonary haemorrhage
4. In brain, multiple petechial haemorrhages seen



Fig. 2: Liver (450gms) showed features of mixed cirrhosis



Fig. 3: Spleen weighed 150 grams, External and cut section showed congested parenchyma



Fig. 4: Bilateral lungs were rubbery to feel. External and cut section was congested

5. Mesentery showed abundant haemorrhage
6. Colon showed features of mild inflammation suggestive of Serositis
7. Pancreas, gastrooesophageal junction, adrenal, stomach, kidneys and Heart were unremarkable

1. Special stains were performed with Masson Trichrome, PAS, Retic, Orcein, Prussian blue and PAS diastase
2. PAS, Diastase positive

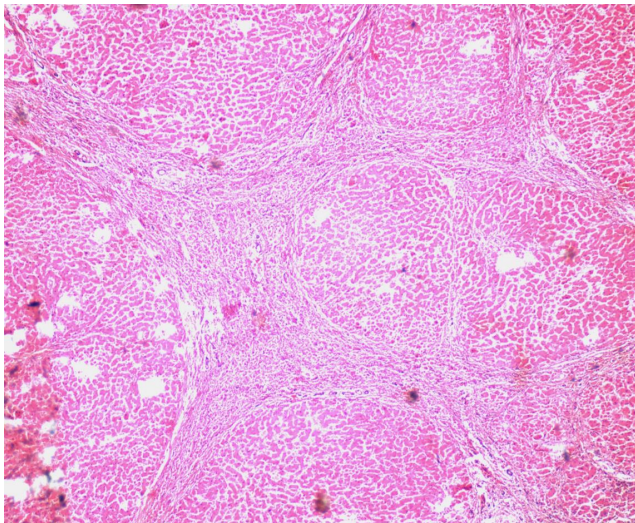


Fig. 5: Liver showed mixed nodular cirrhosis with steatosis

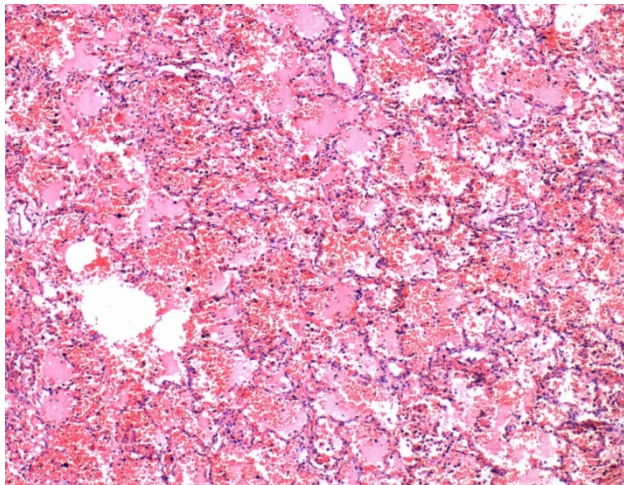


Fig. 6: Lungs showed features suggestive of bronchopneumonia, pulmonary oedema and intrapulmonary haemorrhage

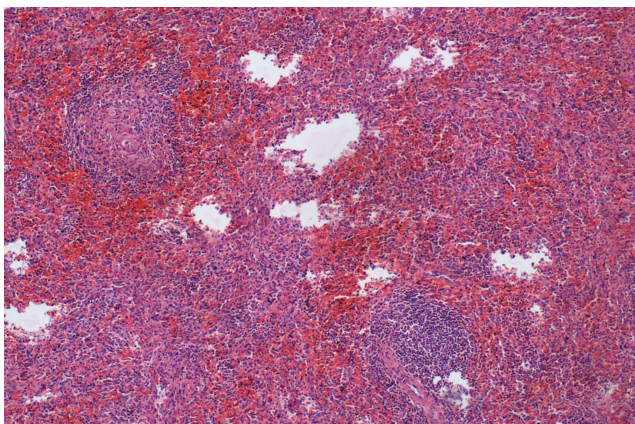


Fig. 7: Spleen showed congestion in red pulp

3. Retic stain showed enhanced fibrosis in portal region
4. Orcein shows focal positivity
5. Masson Trichrome showed weak positivity in bands
6. PB negative

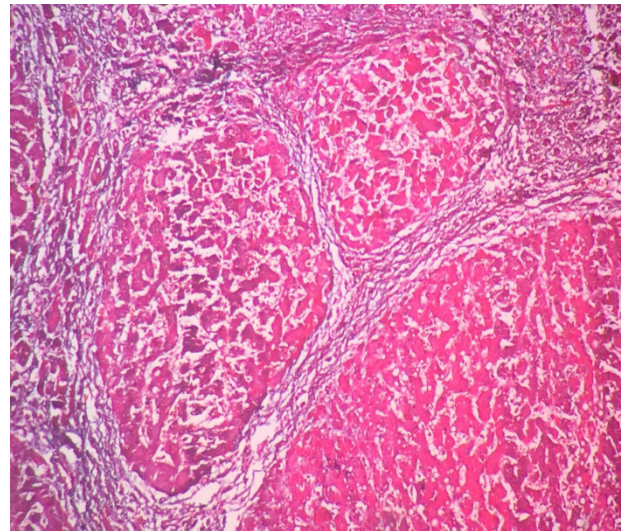


Fig. 8: Masson trichrome stain positive in the bands

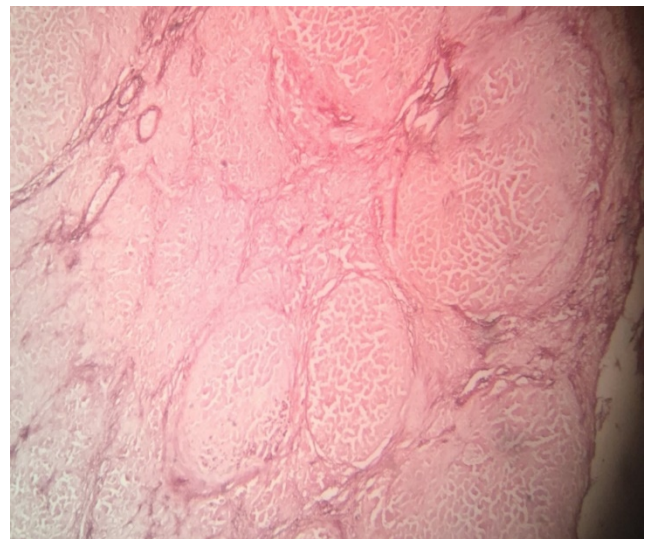


Fig. 9: Orcein stain shows focal positivity

Final cause of death was intrapulmonary haemorrhage and bronchopneumonia in a case of Wilson's disease.

3. Discussion

In 1968, first case of WD was reported in India.^{2,3} WD is an autosomal recessive disease having inborn error of copper metabolism and potentially curable disease if recognized and treated early.² Worldwide the average prevalence is

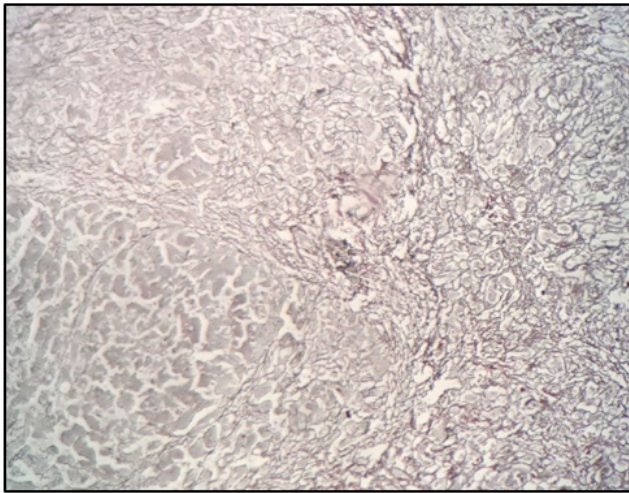


Fig. 10: Retic stain showed enhanced fibrosis in portal region

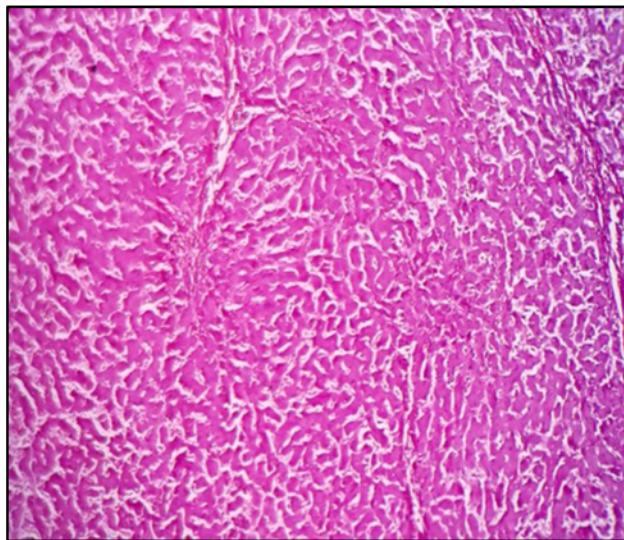


Fig. 11: PAS stain positive

about 30 individuals per million populations.^{3–5} Recorded community based incidence and prevalence are not available in India (many are hospital based reports), although variation in epidemiology, clinical presentation and course are reported.^{1,2,6} In WD, the defect is in copper transport by hepatic lysosomes caused by impaired function of metal transporting P-type adenosine triphosphatase (ATPase) expressed in hepatocyte, encoded by ATP7B gene, located on chromosome 13q14. Decreased or nonfunction of ATP7B protein causes decreased hepatocellular excretion of copper into bile giving rise to excess hepatic copper accumulation and cell injury. There is also failure of copper binding to ceruloplasmin. The early manifestation are generally hepatic or neurological (40% each), while the remainders present with psychiatric, hematological, renal or osteochondrotic manifestation. The hepatic form of the

disease is more commonly seen in children and young adults than older adults.^{2,4} The incorrect diagnoses of WD are diverse and form the differential diagnosis. They are flat feet, myxedema, myasthenia gravis, encephalitis, multiple sclerosis, Parkinson's disease, schizophrenia, depression, anxiety state, acute/chronic hepatitis/cirrhosis of any cause, etc.^{4,5} WD is potentially treatable disease, however fatal if left untreated. The screening tests in suspected cases are examination for KF rings, ultrasound examination of liver, serum copper/ceruloplasmin and 24 h urine copper especially in asymptomatic sibs of index cases.⁴ Early molecular genetic and biochemical studies can be done to confirm diagnosis. Natural course of the neurological form of WD with limited liver disease may progress rather slowly extending over a period of 20 years and has better life expectancy.⁷ Untreated, the disease progress inexorably resulting in extrapyramidal disease and patient in bed ridden status.

4. Source of Support

Nil.

5. Conflict of Interest

None declared.

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