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A Case Report on Wilson's Disease

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Abstract **Case Report**

The accumulation of copper in the liver, brain, cornea, and kidneys is the hallmark of Wilson's disease, an uncommon autosomal recessive condition. There is no community-based research on the prevalence and incidence of Wilson's disease in India; this study is hospital-based. Overview of the Case: A 10-year-old girl with serious complaints of burning micturition, giddiness, loose stools and blood in stools, yellowish discolouration of skin and sclera, generalized body aches, fever and stomach pain with distension was brought to the paediatric department. Wilson's illness was established by abdominal USG using the increased levels of urine copper, whole blood picture, liver function tests, and serum electrolytes and decreased levels of ceruloplasmin. The prominent characteristic, which is less common in youngsters, is the Kayser Fleisher ring. It is identified by a discoloration of the corneal edge that is greenish-brown in colour and eventually goes away with treatment. Probiotics, an antibiotic, a hepatoprotective drug, and copper chelators (D-penicillamine and zinc) were used in her treatment. Gradually she showed improvement in clinical signs and LFT levels.

Keywords: Copper, Ceruloplasmin, Ferritin, Iron, Slit Lamp.

INTRODUCTION

Wilson's illness is an uncommon autosomal recessive copper metabolism disorder that is brought on by a chromosome 13 mutation in the ATP7B gene, which leads to an excess of copper in the body. This condition, where copper deposits in the brain, liver, kidney, eyes, and other organs, is also referred to as hepatolenticular degeneration (hepato-liver, lenticular-brain). Ceruloplasmin, or the P-type adenosine triphosphate family of copper transporter protein, is encoded by the ATP7B gene.

Wilson's illness is caused by a mutation in this gene that impairs ceruloplasmin synthesis. An important component of several metabolic enzymes is copper. Average daily intake of copper is 2–5 mg, with a normal estimated total body copper of 50–100 mg. The body contains a typical quantity of copper at birth. After then, it rises gradually. Usually, the symptoms start between the ages of five and forty-five years. Hepatic symptoms began as acute hepatitis and could develop into fulminant liver failure, which is characterized by digital clubbing, ascites, spider nevi, and palmar erythema. Neural injury results in dystonia, tremor, choreoathetosis, and dementia

The prominent characteristic, which is less common in youngsters, is the Kayser Fleisher ring. It is identified by a discoloration of the corneal edge that is greenish-brown in colour and eventually goes away with treatment (Sriram Shanmugam *et al.*, 2018). Hepatic copper values greater than 250 micrograms per gram of dry weight (normal 20–50) are characteristic of WD (Peter Ferenci *et al.*, 2005). The American Association for the Study of Liver Diseases recommends that first-degree relatives of patients who are newly diagnosed with WD should be screened for the disease. Screening should include genetic testing if available. Other options for screening are basic history and physical examinations, with particular attention to information regarding a history of liver disease, neurologic and psychiatric symptoms, and evaluation for Kayser-Fleischer rings (Schilsky *et al.*, 2022). Patient education regarding dietary modifications, such as avoidance of hepatotoxic medications, alcohol, and diet rich in copper including mushrooms, chocolate, nuts, dried fruits, liver, and shellfish, should be emphasized (Kannauje P K *et al.*, 2021).

CASE PRESENTATION

A 10-year-old girl was brought to the paediatric department with several serious complaints including

burning micturition, fever, generalized body pains, abdominal pain, and distension. She also complained of giddiness, loose stools, and blood in her stools [on the day of admission]. Six months ago, she was diagnosed with jaundice, recovered with the medication use. Over the previous four days, she had abdominal pain and distension. Similar family history of jaundice was noted both in her father 10 years back [recovered in one year] and grandfather. Patient weighed 33kg had an elevated body temperature of 98.6°F. On examination, she was conscious with Icterus positive. Her USG showed abdomen distension AG: 63cm.The initial diagnosis was made as viral hepatitis and started with hepatoprotectants, N-Acetyl cysteine and probiotics. On admission, hepatitis results were negative and her Serum Ceruloplasmin level was lower; on suspicion of Wilson's disease slit lamp examination of eyes was performed and revealed the presence of Kayser Fleischer rings on both eyes.

Table 1: Laboratory Investigations

S. No	PARAMETER	RESULTS	NORMAL RANGE
1.	Serum Iron	117.4	35 – 145
2.	Ferritin	>200	130 – 150
3.	Serum Ceruloplasmin	0.04	0.15 – 0.3
4.	Serum Copper	87.6 mg/dl	15 – 50
5.	TIBC	155.4	250 – 400
6.	Transferrin saturation	75.5	15 - 50
7.	Urine copper	55.8	20 – 50 mcg
8.	Urine volume	2500	-
9.	Urine copper (24 hours)	1395	480 – 1200 mcg
10.	Haemoglobin	11.9 g/dl	9.5 - 14
11.	RBC	3.9lakh/cumm	4.5 - 5
12.	WBC	12800 cells/cumm	5000 - 10000
13.	Platelets	1.68 lakhs	1,50,000 – 4,50,000
14.	Neutrophils	86%	55 – 70
15.	Eosinophils	03%	1 – 3
16.	Monocytes	10%	3 - 8
17.	Total bilirubin	45.6 mg/dl	0.1 – 1.2
18.	Direct bilirubin	22.4 mg/dl	0.2 – 0.8
19.	Indirect bilirubin	23.2 mg/dl	0.1 – 0.3
20.	SGPT (ALT)	95.2 IU/L	17 – 40
21.	SGOT (AST)	101.3 IU/L	15 – 42
22.	ALP	163.1 IU/L	30 – 130

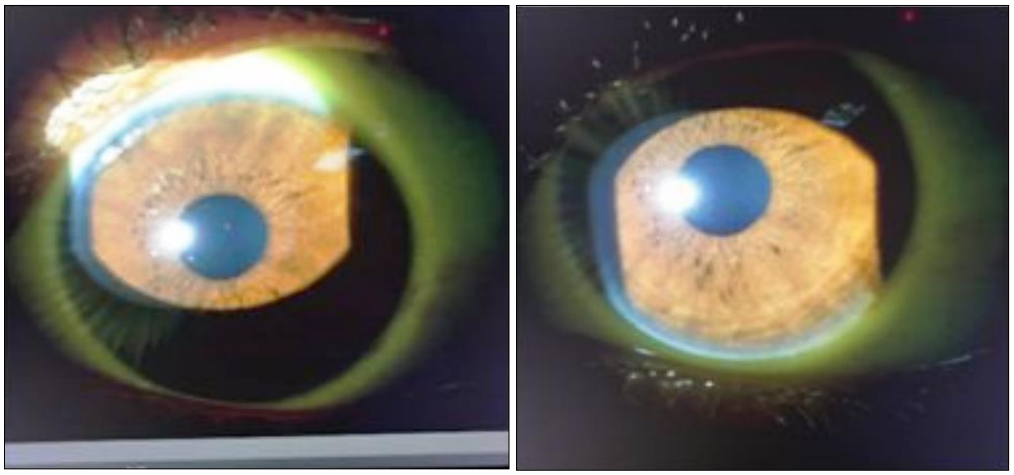

Fig. 1: Kayser Fleischer Rings (Sriram Shanmugam *et al*., 2018)

Her anti-oxidant medication containing copper was discontinued, and her treatment was restricted to hepatoprotective agent. She was told to severely avoid chocolates, animal liver, mushrooms, and nuts and was limited to a low-copper diet.

On observing all the laboratory parameters, the patient was diagnosed with Wilson's disease and was prescribed with the following medications.

Table 2: Treatment Chart

S. No.	BRAND NAME	GENERIC NAME	DOSE	FREQ	ROA
1.	TAB. URSOCOL	Ursodeoxycholic acid	300 mg	B.D.	PO
2.	CAP. CUPRIMINE	D-penicillamine	250 mg	B.D.	PO
3.	SYRUP MADZIC	Zinc gluconate	6ml	T.I.D.	PO
4.	INJ. OMNATAX	Cefotaxime	1.5g	T.I.D.	I.V
5.	SYRUP LACTAM	Lactic acid bacillus	10ml	T.I.D.	PO
6.	INJ. PAN	Pantoprazole	40 mg	O.D.	I.V
7.	INJ. VITAMIN K	Phytomenadione	5mg	O.D.	I.V
8.	ENTEROGERMINA	Bacillus clausii spores	1 to 2 mini bottles	O.D.	PO

Gastroenterologist who examined her noted that a liver transplant was necessary but patient's father denied due to economic issues and the patient was discharged with TAB. URSOCOL, CAP. CUPRIMINE, SYRUP MADZIC, SYRUP LACTAM, TAB. PAN and TAB. OMNATAX.

DISCUSSION

Wilson's disease is an uncommon autosomal recessive condition marked by copper build-up in the kidneys, liver, brain, and cornea. While it can occur at any age, most cases occur in the age range of 5 to 35. It is a progressive illness that can be lethal if neglected, yet prompt diagnosis is still difficult to come by. The primary hallmark of Wilson's illness is liver disease, indicated by our patient's yellowish discoloration of skin and sclera, burning micturition, loose and bloody faeces, fever, generalized weakness, and abdominal distension and pain. Elevated liver function tests (LFTs) were also seen. Our patient had none of the disease's neurological symptoms. It happens when too much copper builds up in the brain's lenticular region and manifests as worry and trouble walking, mood swings and depression. The patient had been found to have Kayser Fleisher rings. Zinc and penicillamine, two copper chelators, are used in the treatment of Wilson's illness, hepatoprotectants were employed as a supportive strategy. In cases of advanced Wilson's disease, liver transplantation is the life-saving treatment.

CONCLUSION

Wilson's disease is an inherited metabolic disorder. It should be suspected in young patients presented with unexplained hepatic complications. Early diagnosis and appropriate management help to prevent the systemic complications. Adherence to therapy, low copper diet and proper follow-up shows a significant reduction in morbidity and mortality. Liver transplantation is recommended in acute liver failure.

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