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Case Presentation

Neuroregression as a Presentation of Wilson Disease in an Adolescent

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ABSTRACT

Background: Wilson disease (hepatolenticular degeneration) is a rare autosomal recessive disorder of copper metabolism resulting from mutations in the ATP7B gene on chromosome 13. The defective ATP7B protein leads to impaired biliary copper excretion and toxic copper accumulation in the liver, brain, cornea, and kidneys. The disease may present with hepatic, neurological, or psychiatric features, but regression of achieved milestones and seizures are relatively uncommon manifestations in children.

Case Presentation: An 11-year-old male presented with progressive drooling of saliva, nasal tone of speech, dysarthria, difficulty in walking, and intermittent aggressive behaviour for the past 1 year 5 months. He also experienced one episode of generalized tonic–clonic seizure (GTCS). On examination, cognitive decline, cerebellar signs, and hepatomegaly were noted. MRI brain revealed diffuse hyperintensity involving frontal and temporal lobes, bilateral basal ganglia, and thalami. Slit-lamp examination demonstrated Kayser–Fleischer rings bilaterally. Serum ceruloplasmin was markedly reduced (<3 mg/dl) and 24-hour urinary copper excretion after D-penicillamine challenge was elevated (3862.88 µg/day), confirming Wilson disease. The patient was managed with dietary copper restriction, D-penicillamine, zinc supplementation, and levetiracetam for seizure control.

Conclusion: This case highlights an unusual presentation of Wilson disease with developmental regression and seizures, emphasizing the need to consider this treatable metabolic disorder in children presenting with unexplained neuroregression. Early diagnosis and timely chelation therapy can prevent irreversible neurological damage.

Keywords*: Wilson disease, ATP7B mutation, neuroregression, seizure, Kayser–Fleischer ring, copper metabolism disorder.*

INTRODUCTION

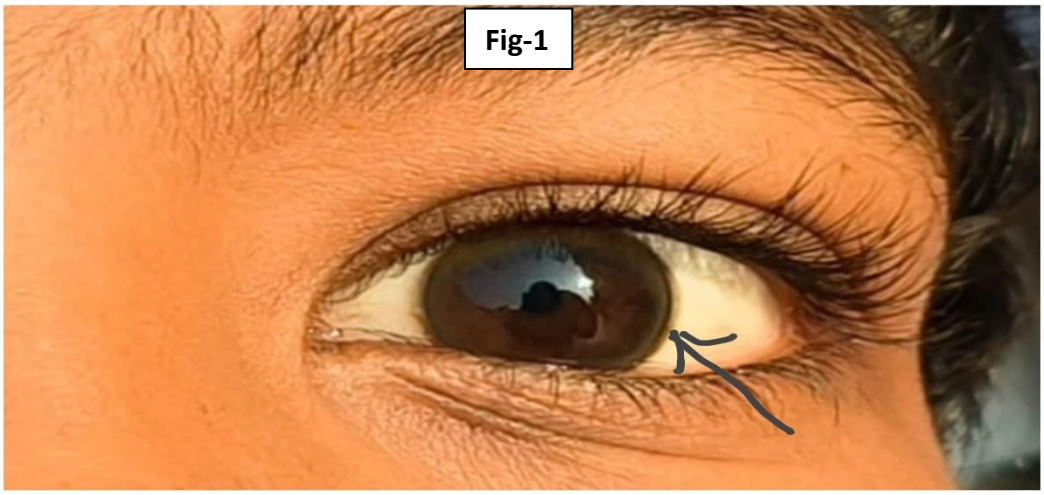
Wilson disease is a rare autosomal recessive disorder of copper metabolism resulting from pathogenic variants in the ATP7B gene on chromosome 13(13q14.3). This gene encodes a copper transporting P type ATPase essential for biliary copper excretion and incorporation of copper into ceruloplasmin. Defective or absent ATP7B function leads to impaired biliary copper excretion and toxic accumulation of copper in the liver, brain, cornea, kidneys and other organs. The worldwide incidence is approximately 1 in 30,000 live births. The clinical spectrum of wilson disease is broad and age dependent, with hepatic manifestations being more common in children and neurological or psychiatric symptoms predominating in adults. Hepatic presentations range from asymptomatic transaminitis to acute liver failure or cirrhosis, whereas neurological features include tremor,dystonia,dysarthira and parkinsonian symptoms. Psychiatric manifestations such as personality changes, depression and behavioural disturbances may also occur. A pathognomic sign is the presence of Kayser-Fleisher(KF) rings due to copper deposition in descement membrane of cornea. Diagnosis relies on clinical suspicion supported by low serum ceruloplasmin, elevated 24 hour urinary copper excretion, increased hepatic copper content and genetic testing of ATP7B mutations. Early recognition is crucial because wilson disease is potentially fatal if untreated but fully treatable with copper chelating agents such as penicillamine or trientine and zinc therapy to reduce intestinal copper absorption in advanced cases, liver transplantation remains as the only option.

CASE PRESENTATION

11 year old male child came to our hospital with total duration of illness of around 1 year 5 months. To start with he developed drooling of saliva from mouth and developed nasal tone to his speech and not able to speak clear words which worsened gradually and some difficulty in eating. After 3 months he was not able to walk properly without support and then gradually was unable to stand and sit without support. He also developed aggresive behaviour intermittently, abnormal laughter at times. In last 7 days he developed 1 episode of GTCS convulsion. There was no fever, involuntary movements, altered sensorium, sleep disturbances. There is a history of 1 sibling loss at 11 years of age, cause not known to parents. On general physical examination, patient was alert, cooperative and oriented. Neurological examination- some cognitive decline present, gag reflex was sluggish, dysarthria present. Cerebellar signs were positive like finger nose test, dysdiadokinesia, heel knee test, Ataxic gait seen. On abdominal examination, hepatomegaly present with liver span 12cms. Lab investigations including CBC, LFT, RFT were within normal limits, HbsAg, HCVAb were negative. Abdominal sonography revealed hepatomegaly.

As there was loss of achieved milestones, some cognitive decline, seizure, subtle cerebellar signs a possibility of neuroregression mixed type (gray matter+white matter+cerebellar) was thought of, MRI brain was done it showed signs of diffuse gyral swelling hyperintensity involving superior frontal and right temporal lobe, bilateral basal ganglia and thalamic diffusion restriction. We were planning to go for Whole exome sequencing. Suddenly we thought, because there was intact mentation, drooling of saliva, psychiatric features, we went for a slit lamp examination of eyes and K F rings came out to be present in both eyes. Then we did serum ceruloplasmin levels which came out to be low(<3mg/dl), total 24 hour urinary copper after D-penicillamine challenge test came out to be high(3862.88µg/day).

These helped in confirmation of diagnosis of Wilson disease, treatment regimen was planned as follows: restriction of copper rich food, penicillamine 250 mg twice daily and zinc tablet 50 mg once daily, levetiracetam 500mg twice daily was given and asked for regular follow up.



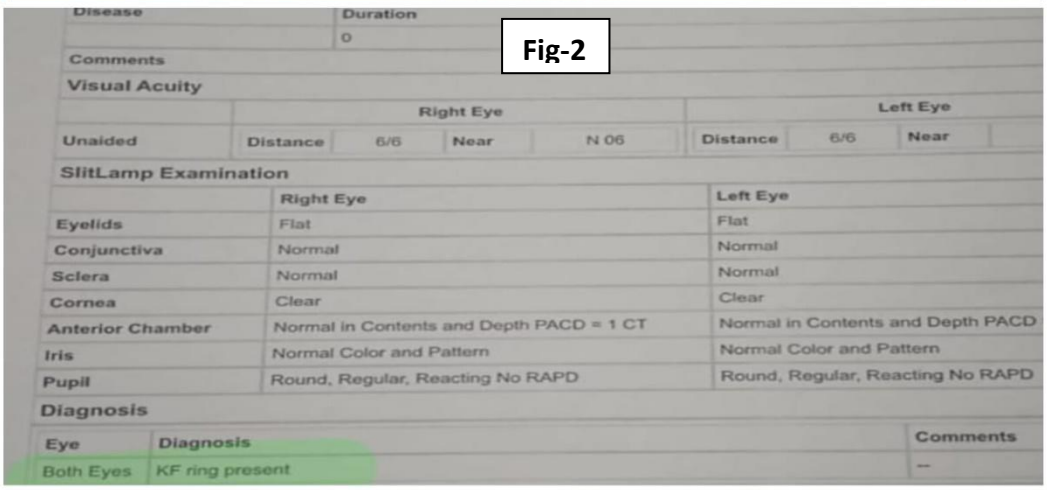


Fig-1: Kayser–Fleischer (KF) ring seen with naked eyes.
Fig-2: Kayser–Fleischer (KF) ring seen in slit-lamp examination.

Fig-3

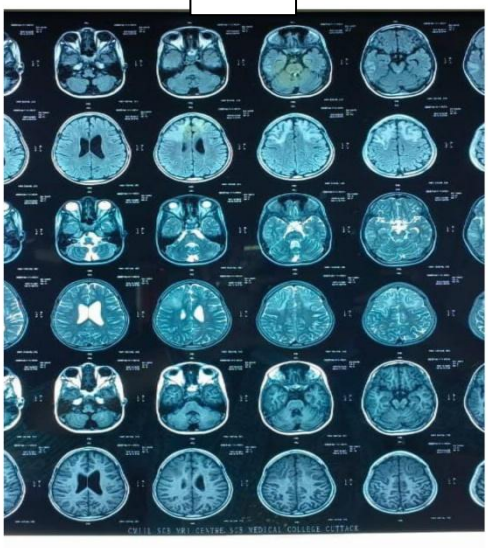


Fig-4

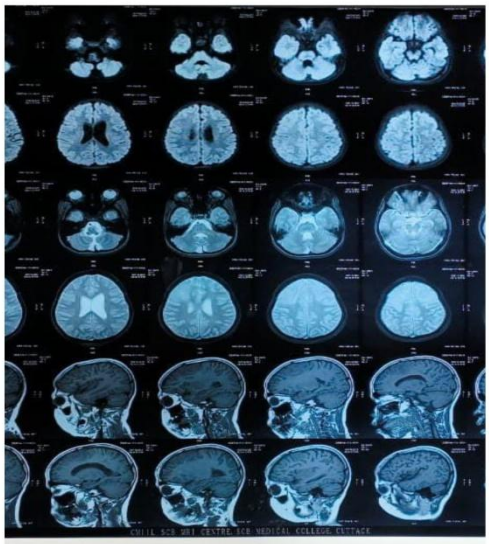


Fig-3 & Fig-4: MRI Brain Film:Diffuse Gyral Swelling WithT2wi/Flair Hyperintencity Noted Involving Bilateral Superior Frontal Lobe And Right Temporal Lobe Without Any Diffusion Restriction.

Bilateral Diffuse Symmetrical T2wi Hyperintencity Noted In Bilateral Basal Ganglia And Bilateral Thalamic Showing Central Diffusion Restriction In Thalamic Region.

Fig-5

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: Mr. DEEPAK KUMAR PRADHAN
: 494004092
: Self
: 18/10/2025 6:48:00PM
: P
: MRS. SAUDAMINI MISHRA - MINI"S CARE PLOT. NO. 2109/2402, AT RANIHAT, PO BUXI BAZAR, PS. MANGALABAG, CUTTACK CUTTACK

Age : 12 Years
Gender : Male
Reported : 20/10/2025 12:21:09PM
Report Status : Final
Processed at : LPL-KOLKATA REFERENCE LAB DR LAL PATH LABS LTD Premises No-031-0199 Plot No-CB 31/1 Str eet 199 Action Area 1C,Newtown Kolkata-700156

Test Report

Test Name	Results	Units	Bio. Ref. Interval
CERULOPLASMIN, SERUM (Immunoturbidimetric)	<3.00	mg/dL	15.00 - 30.00

Comments

Ceruloplasmin is the major copper containing protein in serum and is also the enzyme present in highest circulating concentration. Main function of ceruloplasmin is to regulate ionic state of iron and transportation of copper to tissues. It's quantification is used as a screening test for diagnosing Wilson's disease. Serum ceruloplasmin levels are reduced in Wilson's disease associated with increased urinary copper excretion and increased hepatic copper content. 15% of patients with Wilson's disease may show normal ceruloplasmin levels. Factors which increase ceruloplasmin synthesis are cytokines, pregnancy & estrogens.

WBMC 89075

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End of report

IMPORTANT INSTRUCTIONS

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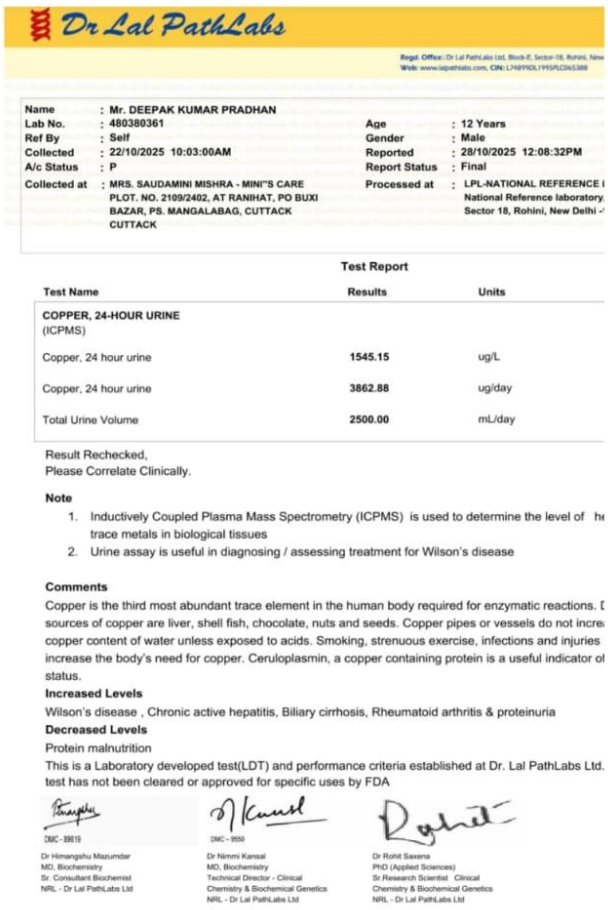
Fig-6



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Name : Mr. DEEPAK KUMAR PRADHAN
Lab No. : 480380361
Ref By : Self
Collected : 22/10/2025 10:03:00AM
A/c Status : P
Collected at : MRS. SAUDAMINI MISHRA - MINI"S CARE PLOT. NO. 2109/2402, AT RANIHAT, PO BUXI BAZAR, PS. MANGALABAG, CUTTACK CUTTACK

Age : 12 Years
Gender : Male
Reported : 28/10/2025 12:08:32PM
Report Status : Final
Processed at : LPL-NATIONAL REFERENCE National Reference laboratory, Sector 18, Rohini, New Delhi

Test Report

Test Name	Results	Units
COPPER, 24-HOUR URINE (ICPMS)		
Copper, 24 hour urine	1545.15	ug/L
Copper, 24 hour urine	3862.88	ug/day
Total Urine Volume	2500.00	mL/day

Result Rechecked,
Please Correlate Clinically.

Note

1. Inductively Coupled Plasma Mass Spectrometry (ICPMS) is used to determine the level of he trace metals in biological tissues
2. Urine assay is useful in diagnosing / assessing treatment for Wilson's disease

Comments

Copper is the third most abundant trace element in the human body required for enzymatic reactions. D sources of copper are liver, shell fish, chocolate, nuts and seeds. Copper pipes or vessels do not increa copper content of water unless exposed to acids. Smoking, strenuous exercise, infections and injuries increase the body's need for copper. Ceruloplasmin, a copper containing protein is a useful indicator of status.

Increased Levels

Wilson's disease , Chronic active hepatitis, Biliary cirrhosis, Rheumatoid arthritis & proteinuria

Decreased Levels

Protein malnutrition

This is a Laboratory developed test(LDT) and performance criteria established at Dr. Lal PathLabs Ltd. test has not been cleared or approved for specific uses by FDA

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Fig-5: Serum Ceruloplasmin report –Low (<3mg/dl)
Fig-6: 24 Hour Urine Copper Report – High (3862.88µg/day)

DISCUSSION

Wilson's disease is an autosomal recessive disorder of copper metabolism. The defect results in impaired biliary excretion of copper and its accumulation in various organs like liver, brain, cornea and kidney.

The disease usually begins in late childhood or adolescence with hepatic / neurological or psychiatric symptoms depending on prominent site of copper deposition.

In our case, the 11-year-old male presented with regression of achieved milestones and seizure, which is a relatively unusual manifestation of Wilson disease.

Neurological features such as nasopharyngeal weakness, dysarthria, cerebellar signs, and behavioural changes were noted at presentation. MRI brain showed involvement of frontal and temporal lobe involvement apart from basal ganglia and thalamus involvement. Due to these cognitive, behavioural, some psychiatric changes and dysarthria, ataxia, we were not able to correlate with hepatomegaly as LFT was normal so, we went for slit lamp examination which showed the presence of Kayser–Fleischer rings in both eyes which paved our path to drive on right path. Further detailed tests showed low serum ceruloplasmin levels, increased 24-hour urinary copper excretion after penicillamine challenge test which confirmed Wilson disease.

While movement disorders and dysarthria are common neurological features in Wilsons, seizure and developmental regression have been described in only limited subsets of patients likely due to cortical and subcortical copper deposition. Concomitant hepatic involvement evidenced by hepatomegaly, further supports multisystem nature of disease. Early recognition of such atypical presentation is essential as it is a potentially reversible cause of progressive neurological dysfunction in children. Mainstay of therapy involves copper chelators such as D-penicillamine along with zinc supplementation to reduce intestinal copper absorption. Dietary restriction of copper-rich foods plays an adjunct role. Addition of levetiracetam for seizure control was appropriate in this case. This case highlights the importance of considering Wilson's disease in any child presenting with unexplained neurological regression and new onset seizures.

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