WWW.IJRPR.COM

International Journal of Research Publication and Reviews

Journal homepage: www.ijrpr.com ISSN 2582-7421

Wilson's Disease: Experience at Tertiary Care Centre

[1]Dr. Vaibhav R. Kute, [2]Dr. Shubham S Khinvasara, [3]Dr. Sumit R. Gotmare, [4]Dr. Sanket Sorate, [5]Dr. Sujit S.Kadam

[1,2,3,4,5] Department of Medicine, SMBT Medical College and Hospital, Nandi Hills, Igatpuri, Nashik-422403.

Abstract:

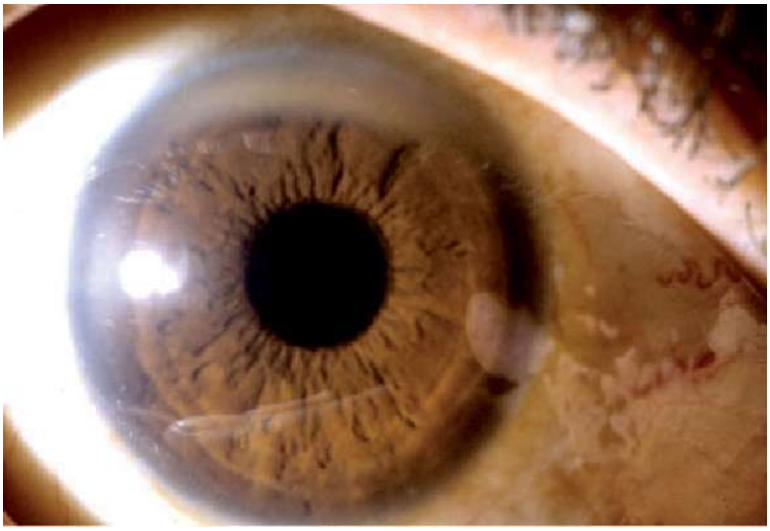
Wilson disease is a rare autosomal recessive disorder leading to abnormal copper deposition in brain,liver and cornea.If it is diagnosed early and treated promptly there is healthy outcome else it can be proven fatal

INTRODUCTION

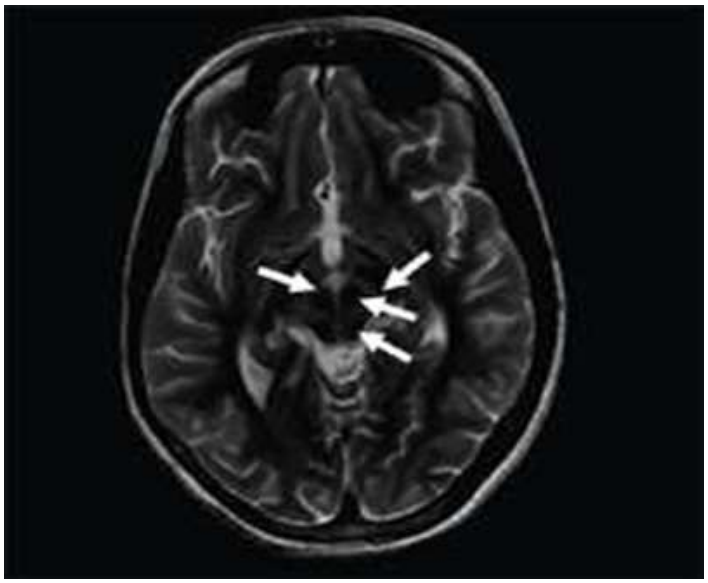
Wilson disease (hepatolenticular degeneration) is a rare, autosomal recessive disorder caused by abnormal copper accumulation in the body particularly involving the brain, liver, and cornea.

- It affects 1 in 30,000 individuals and may present as weakness, abdominal pain, jaundice, personality change, seizures, etc.
- The majority of patients with Wilson disease present within the first decade of life with liver dysfunction. T
- he neuropsychiatric features are seen in the third/fourth decade of life.
- Wilson disease is rare but if not recognized and treated, it is fatal.

IMAGE



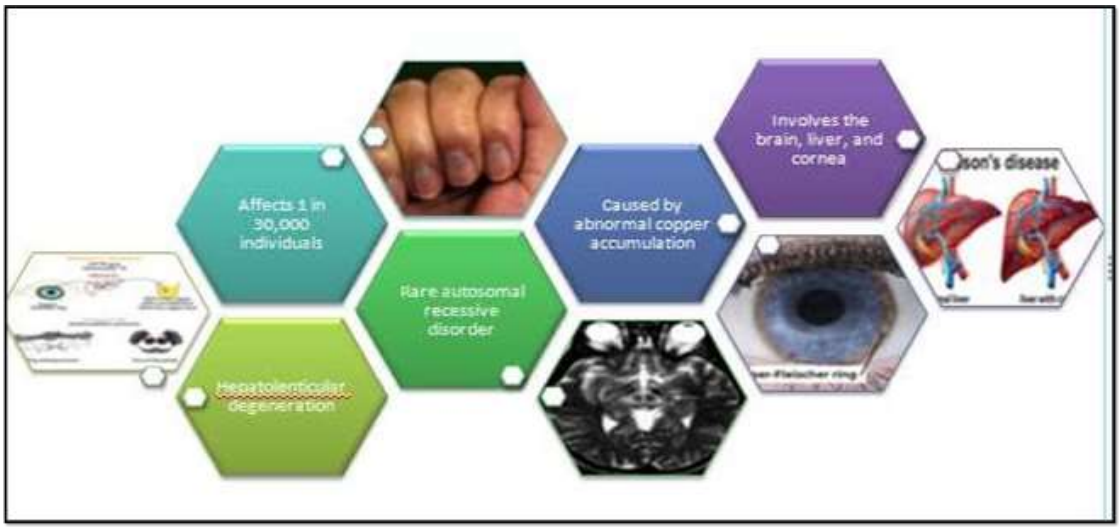
Kayser-Fleischer ring.



Face of giant panda sign.

CASE HISTORY AND DISCUSSION

- A 14 year 8 months old male patient presented to the department of Medicine, SMBT Medical College and Hospital, Nashik. with history of slurring of speech, difficulty in swallowing, imbalance while walking and right sided limb weakness.
- No family history with similar complaints.
- On examination, patient showed dysarthria, ataxic gait and tremors of the right hand.
- Ophthalmic examination revealed Kayser-Fleischer rings in both eyes.
- In laboratory investigations, we found low serum ceruloplasmin and raised 24 hour urine copper.
- Provisional diagnosis of Wilson's disease was made.
- On MRI, bilateral syammetrical well defined signal abnormalities with partial restricted diffusion in basal ganglia, internal capsule, thalami, external capsules, midbrain, pons and dentate nuclei forming "face of giant panda" sign.
- The diagnosis of Wilson's disease was confirmed and treatment with penicillamine 250 mg/day was initiated.
- Patient was discharged after 2 weeks with symptomatic improvement.



- The patients with Wilson's disease usually present with the liver disease and develop neurological manifestations later on. But in our case it was the reverse. The presenting symptom was tremor..

- There are other case reports as well where the initial presentation pertained to nervous system without involvement of liver and the presenting age was over 40 years. The presenting age in our case was 14 years.
- Paradoxically the neurological manifestations are said to become worse with penicillamine. This is attributed to mobilization of copper from the liver with elevations in unbound copper which produces worsening of neurological symptoms.
- In different studies the initial neurological deterioration was observed in 30- 75% of patients following penicillamine therapy. This was refuted by certain other reports. The tremor improved in our case after treatment with penicillamine.

CONCLUSION

- Early diagnosis of Wilson's disease with clinical, biochemical, and characteristic radiological findings is very essential to initiate the treatment early and to halt the progression of the disease.

References:

FOR MORE INFORMATION:

Ala A, Schilsky ML. Wilson disease: pathophysiology, diagnosis, treatment, and screening.

Clin Liver Dis.

2004;8:787-805.

Kaler SG. Wilson's disease. In: Cecil RL, Goldman L, Bennett JC, eds.

Cecil Textbook of Medicine.

22nd ed. Philadelphia: WB Saunders Co; 2004:1301-1305.

Kaplan MM. Clinical manifestations, diagnosis, and natural history of primary biliary cirrhosis

. *UpToDate.*

April 2005. Available at: http://patients.uptodate.com/ topic.asp?file=hep_dis/8639&title=Primary+Biliary+cirrhosis.

Roberts EA, Schilsky ML; Division of Gastroenterology and Nutrition, Hospital for Sick Children, Toronto, Ontario, Canada. A practice guideline on Wilson disease.

Hepatology.

2003;37:1475-1492.

Tung BY, Kowdley KV. Clinical manifestations and diagnosis of primary sclerosing cholangitis.

UpToDate.

March 2004. Available at: http://patients. uptodate.com/topic.asp?file=biliaryt/4959&title=Primary+Sclerosing+cholangitis