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# A CASE OF NEUROGENIC FORM OF WILSON'S DISEASE – A RARE DISORDER IN COPPER TRANSPORT.

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## ABSTRACT

Among the various disorders of metabolism existing in nature, there are certain disorders whose prognosis is good and which are treatable to some extent. The early diagnosis and treatment helps in modifying the changes caused by the diseases. The failure for early diagnosis and intervention is quiet common among people in the developing countries. This case report is an example for such negligence. The patient here presents with advanced symptoms at an elderly age and was diagnosed as Wilson's disease.

**KEY WORDS:** Ceruloplasmin, Kayser-Fleisher ring, ATP7B, Penicillamine

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# INTRODUCTION

Among a wide range of inborn errors of metabolism, Wilson's disease (WD)is noted curiously by the clinicians with special interest being a potentially curable disease if recognized and treated early.It is of 2 types according to the age of onset. Hepatic form is more common in early childhood period and neuropsychiatric form in adults. Although being an autosomal recessively inherited genetic disorder it is quite not rare in prevalence. The prevalence of WD in Asian countries varied between 33 and 68 per 10,000 in the early 2000s which is a bit more than in European population varying between 12 and 29. This difference in variation is also due to various socio-economic reasons faced by the developing countries like India. I would thus like to discuss about a case of Neurogenic Wilson's disease which was diagnosed quite late from the onset of symptoms.

# CASE REPORT

A 35 year old man of Indian origin presented with complaints of coarse tremors of head and whole body, blurred vision, anorexia gradually increasing since 2003. He is on symptomatic treatment since then and his clinical condition wasn't improving. The patient also had a history of jaundice during his childhood at the age of 8. He also had a positive family history of his aunt suffering from similar symptoms.He was examined by the neuro-physician and was found to have flapping tremors of both hands (right>left), head and whole body, hot potato voice speech and blurred vision. He had no dysphagia. On examination he was found to have Gait – stepping forward, mild cog-wheel rigidity of all limbs with a power of 5/5. Local examination of his eyes showed KF ring at the sclerocorneal junction. Hence, it was proposed to rule out Wilson's disease and certain specific laboratory and imaging studies were done. Ophthalmologist's, Nephrologist's and Gastro-enterologist's opinions were also sought. Certain other investigations were also ordered by them.

***Lab investigations and imaging studies***

1. Serum copper : 54.25 mcg/dl (70-140)
2. Serum ceruloplasmin: 8.5 mg/dl (20-60)
3. ALP : 217 IU/l (<115)
4. A:G : 1.82
5. MRI Brain: FLAIR hyperintense foci in bilateral lentiform nucleus, thalamus, mid-brain, pons, tegmentum, middle cerebellar peduncle. Few non-specific FLAIR hyperintense foci seen in bilateral corona radiate. Mild atrophic changes in the mid-brain and cerebellar hemisphere.
6. Urine routine examination : albumin +. Other investigations namely the Chest Roentgenogram, Ultrasonography of abdomen, Thyroid function tests, Serum electrolytes, Arterial blood gas, Portal Doppler, Complete hemogram, Renal function tests were normal.

***Diagnosis***

As per the above clinical features, laboratory and imaging study reports, he was diagnosed to be suffering from Neurogenic Wilson's disease.

# DISCUSSION

WD is an autosomal recessively inherited disease due to the mutations in a Gene encoding a Copper-binding P-type ATPase, encoded by ATP7B gene located on chromosome 13q14 and consists of 21 exons. Copper fails to excrete in the bile and accumulates in liver, brain, kidney and the descemet's membrane in the eye resulting in copper toxicosis. This increase in liver copper levels leads to low levels of ceruloplasmin in plasma by inhibiting the coupling of copper to apoceruloplasmin. The accumulation of copper in tissues and organs lead to various clinical features accordingly. In the study conducted by *Rahman S et al*[1], the subject had certain neurological symptoms like change in voice, difficulty in writing, blurring of vision and a sign of bilateral Kayser-Fleisher ring under the slit

lamp examination. But unlike the same study and certain other studies conducted by *Bhave Sa et al*[2], *Harry Freedberg*[3] and*Jha Sk et al*[4], our subject had no change in Hepatobiliary system and a normal hepatic echostructure. The MRI of brain shows certain features specific for WD which is almost correlating with the study conducted by Jha Sk et al[4].Our subject is definitely suffering from certain central neurological signs and symptoms, but he is devoid of any psychiatric illness unlike the study done by*Akil M et al*[5],*Hemang Shah et al*[6], *Larry V. Mcdonald et al*[7], *N. Aljukić et al*[8], *Dening Tr et al*[9]. The limitation in the study is that there wasn't a follow-up of the case after the initialization of treatment with Penicillamine, a copper chelating agent. Thus we couldn't assess the progress in the patient's neurologic symptoms or the side effects caused by the treatment as in the study conducted by *Brewer G et al*[10].

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