

# Atypical Presentation of Wilson's Disease

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## Abstract

Wilson's disease, an inborn error of copper metabolism, is a relatively rare familial disorder inherited as an autosomal recessive trait but treatable condition that often presents diagnostic dilemmas. We report below a case with some uncommon but well documented features of the disease with review of literature

**Key words:** *Wilson's disease (WD); KF ring; Hyperpigmentation; Vitamin A deficiency*

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## Short Report

A nine year aged male child, hailing from Kawasoti, Nawalparasi, was admitted to College of Medical Sciences, Bharatpur, Nepal with complaints of generalized hyperpigmentation of skin; more seen in lower limbs for six months, which was progressive in nature. He also had fever, jaundice and abdominal distension three months back which had lasted for 15 days. According to mother, child was having diminished vision in the evening for the last two months. However, child recovered from the jaundice but hyperpigmentation persisted. The patient was born at home; birth weight was not known but was of average size according to mother. His immunization was complete according to the EPI schedule. There was no history of consanguineous marriages. His sister died two years ago with a similar illness. There was no history of blood transfusion, convulsions, loss of consciousness, tremors, hematemesis, melaena, drug intake, bleeding from any other sites and joint pain. They eat foods in the steel plate and use aluminum utensils for cooking purpose. They have never used brass vessels. All developmental milestones were appropriate for age. Child was of average built. His height, weight and head circumference were 133cm, 28kg and 49cm respectively, which were within normal limits. His pulse was 90/min, respiratory rate 18/min, temperature 98°F and BP110/60 mmHg. He was anicteric but had mild anaemia. Rest of the general examination was normal. The eye examination was normal. On examination of the abdomen, the spleen was palpable 3cm below left subcostal margin along the splenic axis, which was firm, smooth and non-tender. Rest of the abdomen was normal. Other systems were also normal.

Keeping the possibility of Wilson's disease an ophthalmological consultation was done. Kayser-Fleischer (KF) ring (Bilaterally) was confirmed by slit-lamp examination.

His hemoglobin was 9.5 gm/dl. Total and differential count, ESR, peripheral blood film, Platelet count, reticulocyte, Prothrombin time, APTT, bilirubin, electrolytes, urea, creatinine, calcium, phosphate, sugar, urine routine, stool routine were all within normal limits. No hemoparasites were seen in the peripheral blood film. Direct coomb's test was negative. Serum protein and albumin were 6.6gm/dl and 3.0gm/dl respectively. SGOT, SGPT and Alkaline phosphatase were 121.1 U/L, 60.3 U/L and 192.0 U/L. His HBsAg and Anti HCV status were negative. Ultrasonogram of the whole abdomen was unremarkable except for mild splenomegaly measuring 120 mm. Serological test for Kala-azar was negative. Chest X-ray showed no abnormality.

The serum ceruloplasmin was 13mg/dl (Normal Value=22-61mg/dl). 24 hour urinary copper was 100 microgram/L (Normal Value=40microgram/L). On the basis of strong family history, clinical findings, presence of KF ring and laboratory findings of low ceruloplasmin level and high 24hour urinary copper level, the diagnosis of Wilson's disease was established presenting with hyperpigmentation and associated Vitamin A deficiency.

The child was managed with D-penicillamine and Vitamin A supplementation for night blindness. However, child showed improvement in vision but hyperpigmentation persisted even when child was seen after two months. Child was kept on regular follow-up.

## Discussion

Wilson's disease has an autosomal recessive inheritance; therefore it is rare in Nepal, because of very low rate of parental consanguinity. The presentation of Wilson's disease is sufficiently protean, the disease is quiet rare and also majority of patients are heralded by hepatic insufficiency as copper first accumulates in the liver<sup>1,2,3</sup>. The other presentations include neuropsychiatric disorders, hemolytic anemia and renal tubular lesions and osseous muscular defects with bony deformities (knock-knees) suggestive of resistant rickets. Osteomalacia, spontaneous fractures, and arthropathy are also seen. Renal calculi are also known. A well known but tricky presentation of Wilson's disease is acute or recurrent Coomb's negative hemolytic anemia with or without associated liver dysfunction. Gallstones too are common in Wilson's disease. Other reported manifestations are cardiac involvement, skin hyperpigmentation, ovarian dysfunction, and hypoparathyroidism<sup>2,3,4</sup>. KF rings are present in 50-60% of children without neurological symptoms. This ring does not affect vision<sup>5</sup>.

The uncommon features of Wilson's disease in our case include the child presenting with hyperpigmentation and associated night blindness due to Vitamin A deficiency. The hyperpigmentation has been reported occasionally and is the development of unusually dark skin patches, a hypermelanotic pigmentation<sup>6,7,8</sup>. The night blindness; nyctalopia due to Vitamin A deficiency encountered in this child was of liver involvement. The mechanism implicated in the pathogenesis of Vitamin A deficiency is that, in the body Vitamin A is synthesized from carotene, a provitamin. One molecule of beta-carotene gives 2 molecule of Vitamin A. In man the liver is believed to be the only organ, which performs this conversion<sup>9</sup>.

The importance is stressed of hyperpigmentation of the body, an atypical presentation, as a pointer in the diagnosis of Wilson's disease. Therefore, it is concluded that hyperpigmentation may be the presenting manifestation of Wilson's disease even in the absence of clinical evidence of hepatic involvement. It is well known and documented that patients with hyperpigmentation in Wilson disease do not respond well in cure of the pigmentation

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## References

1. Schoen RE, Sternlieb I. Clinical Aspects of Wilson's Disease. *Am J Gastroenterol*. 1990; 85:1453-1457.
2. Yuce A, Kocak N, Gurakan F, Ozen H. Wilson's Disease with Hepatic Presentation in Childhood. *Indian Pediatr* 2000; 37: 31-36.
3. Gow PJ, Smallwood RA, Angus PW, Smith AL, Wall AJ, Sewell RB. Diagnosis of Wilson's Disease: An Experience over Three Decades. *Gut* 2000; 46: 415-419.
4. Pandit A, Bavdekar A, Bhave S. Wilson's Disease. *Indian J Pediatr* 2002; 69(9): 785-91.
5. Gaffney D, Fell GS, O'Reilly D St J. Wilson's Disease: Acute and Presymptomatic Laboratory Diagnosis and Monitoring. *J Clin Pathol* 2000; 53:807-812.
6. Thaipisuttikul Y. Acanthosis Nigricans associated with Hepatolenticular Degeneration. *J Dermatol*. 1997Jun; 24(6): 305-400.
7. Steiner P, Frey P, Lupi EA, Kister HJ. Familial Wilson's Disease: Copper Induced Hemolysis, Hypersplenism and Hyperpigmentation as the Main Symptoms. *Schweiz Med Wochenschr*. 1975 Jul 5; 105(27): 872-9.
8. Proctor P. Psychosis, Dyskinesia and Hyperpigmentation. *Lancet*. 1971 May 22; 1(7708): 1069.
9. Segal P, Ruszkowski M, Masiak M. Abortive Wilson's Syndrome with Dark Adaptation Disorder. *Klin Oczna*. 1956; 26(4):379-87.