

## Commentary

Wilson's disease (WD), is an autosomal recessive inherited disorder, resulting in copper accumulation in different organs and tissues. It is due to mutations of the ATP7B gene on chromosome arm 13q.5-7, which encodes a copper-transporting P-type.<sup>[1,2]</sup> The disease is characterized by the presence of hepatic abnormalities, neurological and psychiatric symptoms, and corneal Kayser–Fleischer rings. Patients generally present with hepatic disease early decades of life, whereas neurological symptoms develop later.<sup>[2]</sup> Neurologic abnormalities are variable, most often akinetic-rigid syndrome, tremor, ataxia, and dystonia.<sup>[1]</sup> Any type of liver disease should be considered as Wilson's

disease. The symptoms of liver disease may vary widely, ranging from nonspecific hepatomegaly to severe forms of liver failure.<sup>[1]</sup> The diagnosis of WD is verified by the major laboratory features: measurements of serum ceruloplasmin, urinary copper excretion, and liver copper content, detection of Kayser – Fleischer rings.<sup>[2]</sup>

The case report by Nandi *et al.* described the development of WD with a rare, unusual finding.<sup>[3]</sup> The patient was presented with generalized hyperpigmentation of skin for last three years.<sup>[3]</sup> Neurological symptoms developed later.<sup>[3]</sup> There was no hepatic abnormalities.

Kayser – Fleischer rings was detected in Descemet's membrane of the cornea.<sup>[3]</sup> The diagnosis of WD was made based on a low serum ceruloplasmin concentration, a raised 24 hour urine copper excretion and presence of Kayser – Fleischer rings.<sup>[3]</sup> This is an unusual WD case.<sup>[3]</sup>

The skin is also involved as a result of WD itself or of penicillamine treatment.<sup>[4]</sup> The pruritus and jaundice caused by cholestasis being the most common complaints.<sup>[5,6]</sup> The skin pigmentation is more rare complaint. Although pigmentation is known to be associated with Wilson's disease,<sup>[7]</sup> the association of hyperpigmentation in WD has rarely been reported in literature.

It have been reported generalized uniform skin hyperpigmentation in this case report.<sup>[3]</sup> Conversely, previous studies have been reported that hyperpigmentation of skin predominantly lower limbs.<sup>[2,7-9]</sup> Mohizea reported a case of WD with generalized rippled hyperpigmentation.<sup>[4]</sup>

In conclusion, the diagnosis of WD in this patient was complicated by an unusual presentation and the absence of hepatic abnormalities. The significance is stressed of generalized hyperpigmentation as a pointer in the diagnosis of WD.

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## References

1. European Association for Study of Liver. EASL Clinical Practice Guidelines: Wilson's disease. J Hepatol 2012;56:671-85.
2. Bem RS, Muzzillo DA, Deguti MM, Barbosa ER, Werneck LC, Teive HA. Wilson's disease in southern Brazil: A 40-year follow-up study. Clinics 2011;66:411-6.
3. Nandi M, Sarkar S, Mondal R. Generalized hyperpigmentation in Wilson's disease: An unusual association. J Neurosci Rural Pract 2013;4:70-3.
4. Mohizea SA. Rippled hyperpigmentation in Wilson's disease. Int J Dermatol 2010;49:67-9.
5. Mak CM, Lam CW. Diagnosis of Wilson's disease: A comprehensive review. Crit Rev Clin Lab Sci 2008;45:263-90.
6. Aldersley MA, O'Grady JG. Hepatic disorders. Features and appropriate management. Drugs 1995;49:83-102.
7. Leu ML, Strickland GT, Wang CC, Chen TS. Skin pigmentation in Wilson's disease. J Am Med Assoc 1970;211:1542-3.
8. Chu NS, Hung TP. Geographic variations in Wilson's disease. J Neurol Sci 1993;117:1-7.
9. Gurubacharya SM, Gurubacharya RL. Atypical presentation of Wilson's Disease. J Nepal Paediatr Soc 2008;28:66-7.

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