

A case of neurologic Wilson's disease presenting without Kayser-Fleischer Rings

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Madam, Wilson's disease (WD) is an underdiagnosed disorder. Although the prevalence of clinically diagnosed WD has traditionally been around 1 in 30,000, more recent genetic testing has shown that the actual prevalence may be closer to 1 in 7021.¹ One of the causes of this discrepancy may be frequent failure to diagnose this disorder. It is essential for physicians to familiarize themselves with different presentations of WD in order to make sure they do not misdiagnose this manageable disorder. A study by Verma R, et al. concluded that the correct diagnosis of Wilson's disease on first consultation is only made in one-third of the cases.² This is because Wilson's disease presents with extremely varied symptoms, making it hard to pinpoint the disorder.³ Amongst the wide range of non-distinguishing features seen in Wilson's disease, one symptom is highly specific for the disease - the Kayser Fleischer (KF) ring. Presence of KF rings, which are best detected by a slit-lamp examination, almost certainly means the patient is afflicted with WD.⁴

In her research article, Suvarna JC et al states that KF rings may be one of the earliest manifestations of WD, therefore helping physicians to promptly halt the progression of the disease.⁵ This means that the absence of KF rings can make the diagnosis of WD delayed and difficult. This was the case with a 13-year-old male who was brought to the Emergency Room of Civil Hospital on 20th November 2016 with multiple complaints suggestive of neurological WD, but absence of KF rings on slit-lamp examination. The patient had a history of haematemesis, two episodes of focal fits in the last 24 hours and had been in and out of the hospital for the last two years on account of recurrent jaundice and mild symptoms such as fatigue and diarrhoea. After carrying out several investigations that helped rule out possibilities of other diseases, copper studies helped us finalize our diagnosis of decompensated chronic liver disease with neurological symptoms secondary to

Wilson's disease. A brain Magnetic Resonance Imaging (MRI) scan revealed hyperintense regions, suggesting copper deposits. The patient obtained a score of 6 on the international scoring system for diagnosis of Wilson's disease (> 4 being required for confirmed diagnosis).⁶ Due to the extent of liver damage and occurrence of neurological signs the patient was classified in stage C,⁷ which has a poor prognosis. In conclusion, if hepatic and/or neurologic WD is part of a differential diagnosis, WD should not be eliminated just because KF rings are absent, or because of an atypical presenting age. Children may present without KF rings more often than adults, which may lead to a higher risk of missed or delayed diagnosis.⁸ A further work up should be done including ceruloplasmin and urinary copper levels. Genetic testing, although still expensive and unreliable, could be a useful means of diagnosis in the future. The international scoring system for diagnosis of WD is a reliable tool, and should be used by all physicians considering a diagnosis of WD.⁶

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