To the Editors:

Rare presentation of Wilson disease

Wilson disease is an autosomal recessively inherited disorder (1) characterised by degenerative changes in the brain, liver disease and Kayser-Fleischer rings in the cornea due to deranged copper homeostasis. The incidence is 1:30 000-100 000 births (2). It is at present a significant cause of chronic liver disease in children in Sri Lanka (personal observation).

A 9-year old boy was admitted to the Professorial Paediatric Unit at Lady Ridgeway Hospital for further investigation of a painless and increasing limp, and generalised pigmentation of 2 months' duration. Pigmentation was generalised and gradually increased in severity. He was otherwise well. He was the only child of non-consanguineous parents. He had no significant illness in the past.

On examination he was found to have generalised pigmentation, bilateral Kayser-Fleischer rings, hepatomegaly and a limp. He was not icteric and had no neurological abnormalities. Slit lamp examination of his eyes confirmed bilateral Kayser-Fleischer rings and bilateral early sunflower cataracts. Serum caeruloplasmin was 2.8 mg/dl (normal 16.8 - 34.2 mg/dl) and the 24 h urinary copper excretion was 304.8 mg/dl. (normal <70 mg/dl). The diagnosis of Wilson disease was confirmed by repeating the 24 h urinary copper excretion (1394 mg/dl) after a loading dose of penicillamine. There was no family history of Wilson disease. Following the diagnosis, oral penicillamine was started and necessary dietary advice was given. After initial monitoring he was discharged on penicillamine.

Wilson disease is a rare disorder of hepatobiliary copper excretion and the clinical sequelae result from excessive deposition of copper in various body tissues.

It is predominantly a disease of children, adolescents and young adults. In children hepatic manifestations predominate, whereas in adults neuro-psychological manifestations are more frequent (3). Children could also present with non-spherocytic, Coomb's negative haemolytic anaemia or renal tubular acidosis due to toxicity on red blood cells and renal tubules. Rarely the disease can manifest with features related to rheumatologic, dermatologic, cardiac and endocrine systems (1). Several bone changes have also been described (1) in Wilson disease. This patient presented with two rare manifestations of the disease, namely the limp and generalised pigmentation. Rapid diagnostic investigations facilitated early treatment with oral penicillamine, which is the first-line drug. With this the patient improved clinically as well as biochemically, as assessed by copper excretion studies. This rare presentation of Wilson disease highlights the need to maintain a high index of suspicion to diagnose this readily treatable condition.

References

1. Yanze JC, Martin P, Munoz SJ, Friedmon LS. Wilson's disease: current status. *American Journal of Medicine* 1992; 92: 643-32.
2. Balistreri WF. Metabolic diseases of the liver: Wilson disease. Nelson textbook of paediatrics, IS'" ed. Philadelphia: WB Saunders Company, 1996; 1139-40.
3. Brist NRM, Keenaway WG, Powell BR. Disorders of metal metabolism: Wilson's disease. Forfar and Arneil's Textbook of Paediatrics, 4* ed. London; Churchill Livingstone, 1996; 1213-14.

G N N de Silva, *Registrar, and* **Sanath P Lamabadusuriya,** *Senior Professor in Paediatrics, Department of Paediatrics, Faculty of Medicine, University of Colombo. (Corresponding author SPL*

Accepted 25 August 2001)