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Article: Cerebrotendinous Xanthomatosis in Three Siblings from a Chinese Family

(SMJ Vol 42 Issue 1 January 2001)

Dear editor,

I would like to comment the article of Ko and Lee on Cerebrotendinous Xanthomatosis in Three Siblings from a Chinese Family (Singapore Med J 2001; 42:30-2). The authors discuss intractable diarrhoea as a major manifestation of the disease and refer for this statement to the article of Kuriyama et al (1991), reference 7. However, the article of Kuriyama et al (1991) does not mention diarrhoea as a manifestation of cerebrotendinous xanthomatosis. The association of juvenile cataract and chronic diarrhoea was published in 1991 in another publication:

Cruysberg JRM, Wevers RA, Tolboom JJM. Juvenile cataract associated with chronic diarrhoea in pediatric cerebrotendinous xanthomatosis. Am J Ophthalmol 1991; 112:606-7.

With kind regards,

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Reply From Author

Dear Editor,

Thank you for providing me the chance to reply to the letter.

In addition to neurologic symptoms, patients with Cerebrotendinous Xanthomatosis (CTX) develop cataracts, diarrhoea, Archilles tendon xanthoma, atherosclerotic vascular disease and many other abnormalities[1]. As far as diarrhoea is concerned, there are a number of articles that describe diarrhoea as a manifestation of the disease in the literature[2-4]. Bindl et al regarded the diarrhoea as "chologenic"[5]. While diarrhoea was thought to be a dominating symptom in children[3], it has been described in adult patients. One of two female patients diagnosed with Cerebrotendinous Xanthomatosis during their forties was reported to have chronic diarrhoea[4]. In a study, diarrhoea was said to cease immediately after starting treatment with chenodeoxycholic acid[3]. Whereas these articles were related to diarrhoea in Cerebrotendinous Xanthomatosis, the article of Kuriyama et al mainly presented the clinical features, biochemical and investigational abnormalities of eight patients with Cerebrotendinous Xanthomatosis and had data on 136 patients reported throughout the world reviewed[6].

One purpose of writing the case report was to attract the attention of the clinicians to the existence of this very rare entiry among Chinese patients. Since after its publication, a few more patients with Cerebrotendinous Xanthomatosis were diagnosed in Hong Kong.

Thank you for your attention!

Yours sincerely,

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