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LETTER TO THE EDITOR

Liver transplantation due to cerebrotendinous xanthomatosis end-stage liver disease

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Cerebrotendinous xanthomatosis (CTX) is an autosomal recessive disease characterized by an increase in plasma concentrations of cholestanol and storage of sterols in multiple tissues, especially tendons and the nervous system [1]. Neonatal cholestatic jaundice has been rarely reported, but patients may progress to cirrhosis if effective treatment is not provided. Mutations in the *CYP27A1* gene leads to decreased synthesis of bile acid, excessive production of cholestanol, and consequent accumulation of cholestanol in tissues in patients with CTX [2]. The liver is rarely affected in these patients; however, neonatal cholestatic jaundice can be self-limiting. Reports on liver transplantation for CTX are lacking. Liver transplantation not only cures the liver disease, but also introduces the normal *CYP27A1* gene into the patient, which can be potentially beneficial.

An 8-month-old girl was diagnosed with CTX through the *CYP27A1* mutation analysis. This patient developed jaundice one month after birth. Chenodeoxycholic acid (CDCA) therapy was effective for controlling cholestanol, but not liver dysfunction. At the age of 8 months, the patient underwent living donor liver transplantation, in which the graft was donated by her father (left lateral lobe). The pathology report indicated cirrhosisin in the patient (Fig. 1). The liver function returned to normal one week after transplantation and was maintained evenly during the 1-year follow-up. No more CDCA therapy was needed. No newly formed xanthomas were detected in the Achilles tendons or other regions. Bone mineral density improved from $Z=-2.9$ to $Z=-2.1$. Although the patient's motor function and social behavior were all normal, a 9-month developmental delay in the patient's intellectual function was reported one year after transplantation.

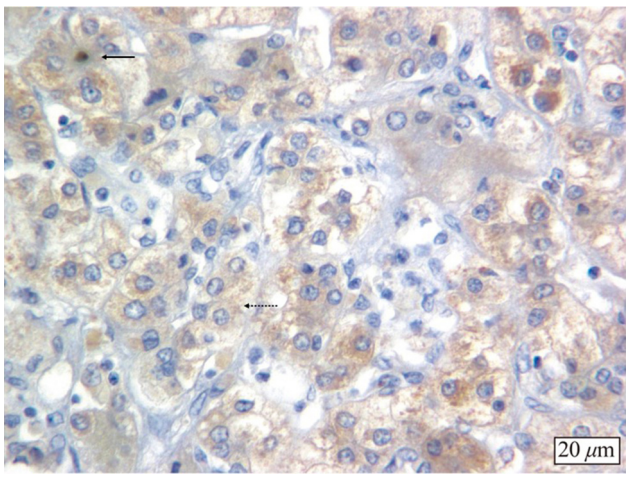

Fig. 1 Immunohistochemistry of CYP27A of the patient (original magnification × 400). Hematoxylin and eosin stain shows pale, blush-like, weak cytoplasmic marking in the hepatocytes (arrow). The dashed arrow indicates rosette of hepatocytes with bile plug and solid arrow indicates weak cytoplasmic marking in the hepatocytes

In the present case study, the patient had normal motor function and social behavior, but had impaired intelligence development before transplantation. The patient still showed retarded intelligence development 1 year after transplantation, although the MRI examination revealed no cerebral atrophy. Whether the corrected metabolism after liver transplantation could reverse the neurological deterioration in this patient needed further exploration. However, the cerebellar structure in the present case might indicate more potential for intellectual development in the future.

This study evaluated the effect of liver transplantation on CTX-associated end-stage liver disease. Liver transplantation not only restored liver function, but also corrected cholestanol metabolism in the present case. The patient did not require CDCA supplement therapy. This indicated that liver transplantation could be an effective choice for patients with CTX accompanied by liver dysfunction. However,

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whether liver transplantation can reverse the neurological damage in patients already having neurological symptoms needs to be further explored.

Author contribution ZXW proposed the study. CHS collected and analyzed the data. CHS wrote the article.

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Compliance with ethical standards

Ethical approval This study was approved by the Ethical Committee of Huashan Hosiptal, Fu Dan University, School of medicine.

Conflict of interest The authors declared no conflict of interest.

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