

1. J Gastroenterol Hepatol. 2008 Aug;23(8 Pt 2):e428-31. Epub 2008 May 7.

## **Cholestatic liver disease in long-term infantile nephropathic cystinosis.**

Cornelis T, Claes K, Gillard P, Nijs E, Roskams T, Lombaerts R, Nevens F, Cassiman D.

Department of Nephrology, University Hospital Gasthuisberg, University of Leuven, Leuven, Belgium.

### **Abstract**

**BACKGROUND:** Cystinosis is a metabolic disease characterized by accumulation of cystine in different organs and tissues, leading to potentially life-threatening organ dysfunction. Infantile cystinosis typically leads to end-stage renal disease, necessitating renal replacement therapy. Liver disease in cystinosis is rare and is mostly reported as nodular regenerative hyperplasia leading to portal hypertension.

**METHODS:** Two patients with infantile cystinosis developed cholestatic liver disease (increasing alkaline phosphatases, gamma-glutamyltransferase and mild increase in transaminases). Severe accumulation of cystine was demonstrated on liver biopsy, predominantly localized in Kupffer cells, together with morphological signs of sclerosing cholangitis on liver biopsy. One patient showed changes compatible with sclerosing cholangitis on magnetic resonance imaging. Therapy with ursodeoxycholic acid led to biochemical improvement in one and stabilization in the other patient.

**CONCLUSION:** Long-term infantile nephropathic cystinosis can be associated with a form of sclerosing cholangitis, which can respond to therapy with ursodeoxycholic acid.

PMID: 18466290 [PubMed - indexed for MEDLINE]