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1: Am J Hum Genet. 2000 Dec;67(6):1598-604. Epub 2000 Oct 23.

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The University of Chicago Press

Identification of a locus for autosomal dominant polycystic liver disease, on chromosome 19p13.2-13.1.

Reynolds DM, Falk CT, Li A, King BF, Kamath PS, Huston J 3rd, Shub C, Iglesias DM, Martin RS, Pirson Y, Torres VE, Somlo S.

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Polycystic liver disease (PCLD) is characterized by the growth of fluid-filled cysts of biliary epithelial origin in the liver. Although the disease is often asymptomatic, it can, when severe, lead to complications requiring surgical therapy. PCLD is most often associated with autosomal dominant polycystic kidney disease (ADPKD); however, families with an isolated polycystic liver phenotype without kidney involvement have been described. The clinical presentation and histological features of polycystic liver disease in the presence or absence of ADPKD are indistinguishable, raising the possibility that the pathogenetic mechanisms in the diseases are interrelated. We ascertained two large families with polycystic liver disease without kidney cysts and performed a genomewide scan for genetic linkage. A causative gene, PCLD, was mapped to chromosome 19p13.2-13.1, with a maximum LOD score of 10.3. Haplotype analysis refined the PCLD interval to 12.5 cM flanked by D19S586/D19S583 and D19S593/D19S579. The discovery of genetic linkage will facilitate diagnosis and study of this underdiagnosed disease entity. Identification of PCLD will be instrumental to an understanding of the pathogenesis of cyst formation in the liver in isolated PCLD and in ADPKD.

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