

Title: Congenital Hepatic Fibrosis Overview *GeneReview* – Autosomal Recessive Polycystic Kidney Disease / CHF

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Note: The following information is provided by the authors listed above and has not been reviewed by *GeneReviews* staff.

## Autosomal Recessive Polycystic Kidney Disease / CHF

A large group of autosomal recessive polycystic kidney disease (ARPKD) / CHF patients were evaluated at the NIH Clinical Center as a part of a prospective study ongoing since 2003 [Gunay-Aygun et al 2006]. Comprehensive cross-sectional data from this study are published [Gunay-Aygun et al 2010a, Gunay-Aygun et al 2010b, Gunay-Aygun et al 2013]; the prospective portion of the study is ongoing. Ultrasonography and magnetic resonance imaging and comprehensive biochemical studies on 73 molecularly confirmed ARPKD/CHF patients revealed that kidney and liver disease in ARPKD / CHF were independent, and variability in severity was not explainable by the type of *PKHD1* mutations. Platelet count was the best predictor of the severity of PH in ARPKD/CHF. Initial symptoms were liver related in 26% of patients, and others presented with kidney disease. Of 73 patients, 1 patient underwent liver and kidney transplantation, and ten others received kidney transplants. Four presented with cholangitis and one with variceal bleeding. Sixty-nine percent of patients had enlarged left lobes on magnetic resonance imaging, 92% had increased liver echogenicity on ultrasonography, and 65% had splenomegaly. Splenomegaly started early in life; 60% of children younger than 5 years had enlarged spleens. Spleen volume was inversely correlated with platelet count and prothrombin time but not with serum albumin level. Platelet count was the best predictor of spleen volume (area under the curve of 0.88905), and spleen length corrected for patient's height correlated inversely with platelet count ( $R(2) = 0.42$ ,  $P < 0.0001$ ). Spleen volume did not correlate with renal function or type of *PKHD1* mutation. Twenty-two of 31 patients who underwent endoscopy had varices. Five had variceal bleeding, and 2 had portosystemic shunts. Seventy percent of ARPKD/CHF patients had macroscopic biliary abnormalities. Forty percent had Caroli syndrome, and 30% had an isolated dilated common bile duct [Gunay-Aygun et al 2013].

## References

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