

Congenital hepatic fibrosis (CHF) is a rare disease that is present at birth (congenital) and affects the liver. CHF rarely occurs as an isolated problem, and is usually associated with ciliopathies that affect the kidneys, called hepatorenal fibrocystic diseases (FCD). These include polycystic kidney disease (PKD), nephronophthisis (NPHP) chronic tubulointerstitial disease, and others. Typical liver abnormalities include an enlarged liver (hepatomegaly), increased pressure in the venous system that carries blood from different organs to the liver (portal hypertension), and fiber-like connective tissue that spreads over and through the liver (hepatic fibrosis). Gastrointestinal (stomach and intestine) bleeding, splenomegaly (enlarged spleen) and hypersplenism (decreased platelet and other blood counts due to enlarged spleen) may be early signs of this condition. The frequency of CHF is not known. The prevalence has been estimated to be 1/10,000 -20,000 based on the prevalence of ciliopathies that are associated with CHF. CHF is diagnosed by ultrasound exam and magnetic resonance imaging of the liver and kidneys, and rarely, by liver biopsy. CHF and Caroli's syndrome are often associated with cystic disease of the kidneys. Family history, physical exam, and various tests including kidney ultrasound exam, kidney function tests, X-rays, eye exam, brain MRI, and molecular genetic testing can help to determine the underlying FCD syndrome.