Isolated Polycystic Liver Disease (PKHI)

Abstract: PKHI isolated hepatic polycystosis, first described in 1856 by Bristowe, is most often associated with autosomal dominant polycystic kidney disease to give autosomal dominant hepato-renal polycystosis. Hepatic cysts are the most common extrarenal manifestation of autosomal dominant polycystic liver disease. We report the case of a young patient diagnosed with PKHI during a chronic abdominal pain assessment.

Keywords: Isolated polycystic liver disease - Hepatic cysts.

DEFINITION

Polycystic liver disease is defined by the presence of at least 3 cystic lesions of the liver (Debenes, B. et al., 1992; & Levy, V.G. 1989) but it is usually characterized by innumerable cysts of variable size scattered within the hepatic parenchyma (Vauthey, J. N. et al., 1991). We speak of massive polycystic liver disease beyond 15 hepatic cysts (Kaehny, W. D., & Everson, G. T. 1991, November). Polycystic liver disease, first described in 1856 by Bristowe (Bristowe, J. S. 1856), is most often associated with autosomal dominant polycystic kidney disease to give autosomal dominant hepato-renal polycystosis (Kaehny, W. D., & Everson, G. T. 1991, November; Bristowe, J. S. 1856; Kwock, M. K., & Lewin, K. J. 1988; & Chauveau, D. et al., 1995). Hepatic cysts are the most common extrarenal manifestation of autosomal dominant polycystic liver disease (Gabow, P. A. 1993; & Perrone, R.D. 1997). The familial form of isolated polycystic liver disease, without associated renal cystic disease, has also been described (Iglesias, D. M. et al., 1999; & Pirson, Y. et al., 1996).

OBSERVATION

We report the case of a 46-year-old patient, with 3 fetal deaths as antecedents, with no family history of hepato-renal polycystosis, under oral contraception, who presents for the progressive onset of HCD pain type stabbing, inhibiting breathing, radiating to the right shoulder, unrelated to food intake, without aggravating or relieving factor or other digestive or extradigestive manifestations, evolving in a context of apyrexia and CEG.

The clinical examination objectified a non-homogeneous diffuse hepatomegaly (FH at 14cm) painless, soft anterior border, without other palpable mass without jaundice without peripheral adenopathies.

The assessment objectified to abdominal ultrasound of multiple hepatic cysts, without renal cyst while the serology of hydatidosis was negative. A liver test performed did not show cytolysis or cholestasis, a 98% prothrombin level and normal tumor markers. Computed tomography confirmed the presence of multiple hepatic cysts of small and medium size of water tone with diffuse hepatic involvement without contrast enhancement after intravenous injection of the contrast agent, without renal cyst.

DISCUSSION

The prevalence of PKHI is 1 in 100,000. Women are mainly affected and have a higher number of cysts than men. Cysts are undetectable in the first years of life and usually appear after 40 years. Their number and size increase with age. Symptoms depend on mass (compression effect) and may include abdominal distension, gastroesophageal...
reflux, dyspnea, reduced mobility and back pain due to hepatomegaly. Some patients are asymptomatic. Other complications (intracystic hemorrhage or infection, torsion or rupture of the cysts) can cause acute abdominal pain. Liver function is generally normal. There is no portal hypertension. Extrahepatic manifestations are very rare and may include intracranial aneurysms (generally small and at low risk of rupture) (Chauveau, D. et al., 1997; & NAKAJIMA, F. et al., 2000; & Leão, R. N. et al., 2014) and abnormalities of the mitral valve. In rare cases, hepatomegaly can lead to sometimes lethal malnutrition.

Hepatic cysts result from excessive growth of the biliary epithelium or dilation of the peribiliary glands. Some cases occur sporadically, but most are transmitted in an autosomal dominant fashion. PKH1 is due in 30% to 50% of cases to mutations in the PRKCSH or SEC63 genes. As all cases of PKH1 do not respond to this type of mutation, there may be genes and modes of transmission that are not yet identified.

If there are no complications, liver tests are normal (Vauthey, J. N. et al., 1991; & Everson, G. T. et al., 1988). Everson et al. (1988) showed by quantitative computed tomography, in 25 patients with polycystic liver disease, a parenchymal volume of the liver preserved despite a very large cystic volume explaining the normality of liver biological parameters. A significant disturbance of the hepatic assessment must therefore lead to the search for a complication or intercurrent hepatopathy (Everson, G. T. et al., 1988; & Buffet, C., & Hagège, H. 1993).

Abdominal ultrasound is the most simple and sensitive examination for the detection of hepatic cysts and finds a more or less significant number of liquid images disseminated within a deformed parenchyma (Sanchez, H. et al., 1991; Vilgrain, V. 2001). The ultrasound appearance of these cysts is similar to that of simple biliary cysts: round or oval images, well limited with thin walls, devoid of vegetation, anechoic and transound with posterior strengthening of the echoes (Vilgrain, V. 2001; & Wan, S. K., & Cochlin, D. L. 1990). In polycystic liver disease, the very large number and the contiguity of the cysts can give images of septaendocystic or multilobulated cysts. With the disappearance of the normal architecture of the liver and a poorer visualization of the hepatic vessels, it is often difficult to find the usual anatomical landmarks. In this case, hepatic abnormalities in polycystic liver disease are best appreciated by computed tomography (Wan, S. K., & Cochlin, D. L. 1990). This review shows multiple rounded water-tone images with an attenuation coefficient between - 5 and + 20 Hounsfield units. The cyst wall does not increase after intravenous injection of contrast agent (Vilgrain, V. 2001; & Wan, S. K., & Cochlin, D. L. 1990). In these fragile patients with impaired renal function, extreme care must be taken with regard to the amount of iodinated contrast agent injected so as not to further degrade their nephrological state. Cystic parietal calcifications are sometimes the witness of an old infection or hemorrhage (Vilgrain, V. 2001; & Kutcher, R. et al., 1977) and when they are multiple, it is necessary to evoke a hyperparathyroidism secondary to the chronic renal insufficiency (Chapman, A. B. et al., 1992). Magnetic resonance imaging (MRI) is not a first-line radiological investigation in asymptomatic polycystic liver disease (Vilgrain, V. 2001) but is of great interest in complicated forms. In MRI, uncomplicated cysts appear as rounded, homogeneous images, with a perfectly hypointense signal on sequences weighted in T1 and strongly hyperintense in sequences weighted in T2 (Vilgrain, V. 2001); they do not enhance after injection of gadolinium.

All these radiological examinations also explore the kidneys of these patients: it is indeed necessary to look for a cystic renal dysmorphosis to differentiate an autosomal dominant hepato-renal polycystosis from an isolated hepatic polycystosis. Ravine et al., defined the radiological diagnostic criteria for autosomal dominant polycystic hepato-renal according to the number of renal cysts as a function of age (Ravine, D. et al., 1994); the aim of these recommendations is not to confuse dominant autosomal hepato-renal polycystosis with isolated sporadic hepatic polycystosis associated with common renal cysts, these being frequent from the age of 50 (Ravine, D. et al., 1993). Thus, before 30 years of age, the diagnosis of autosomal dominant hepato-renal polycystosis can be brought by the identification of at least 2 renal cysts, unilateral or bilateral; between 30 and 59 years, at least two cysts are needed in each kidney and after 60 years, the diagnosis of autosomal dominant polycystic liver disease requires the presence of at least 4 cysts per kidney (Ravine, D. et al., 1994).

The differential diagnosis includes multiple hepatic cysts, associated with autosomal dominant polycystic kidney disease (PRAD). Simple liver cysts are also a differential diagnosis. Caroli’s disease, characterized by cysts communicating with the bile duct, differs from imaging with the specific excretion of contrast media in the bile (Vilgrain, V. 2001).

Most patients are asymptomatic and do not need treatment. For those with symptoms, treatment depends on the size, distribution and anatomy of the cysts, and may include percutaneous aspiration of the cysts, alcoholic sclerosis,
cystic fenestration, partial hepatectomy, or even liver transplant (in rare cases excess liver growth significantly affects quality of life). Any form of estrogen therapy should be stopped as soon as possible. Recently, lanreotide and long-acting octreotides (somatostatin analogs) have been shown to be safe and effective against PKHD1 by reducing the volume of middle liver cysts and stunting their growth (Rosenfeld, L. et al., 2001). Most patients have a good life prognosis.

References