

Cases from the Armed Forces Institute of Pathology

Peutz-Jeghers Polyp of the Gallbladder

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Introduction:

Peutz-Jeghers syndrome (PJS) is an autosomal-dominantly inherited syndrome characterized by hamartomatous polyps and pigmented macules of mucous membranes and skin. The polyps occur most commonly in the gastrointestinal tract but extra-intestinal polyps may occur rarely. Possible sites include the respiratory tract, urogenital tract, and gallbladder. Herein we report a case of a Peutz-Jeghers polyp of the gallbladder.

Key Words: Peutz-Jeghers syndrome, hamartomatous polyps, gallbladder, and STK11

History:

A 16 year old boy with questionable history of hepatitis (presumed Epstein Barr virus hepatitis) presented to the emergency room with severe abdominal pain with vomiting.

Physical Examination: The physical examination revealed a tall, thin boy wearing glasses and braces. The review of symptoms was unremarkable except for a vague, non-localized abdominal pain

Radiographic Studies: Abdominal ultrasound demonstrated thickening of the gallbladder with a vascular mass and mildly dilated common bile duct. There was no evidence of cholelithiasis and cholecystitis. These findings were confirmed by CT scan, the mass enhanced in both the arterial and venous phases. A fecalith in the appendix was also noted. Fluoroscopic images of the endoscopic retrograde cholangiopancreatography showed a fusiform dilatation of the common bile duct and common hepatic duct, consistent with a choledochal cyst.

Procedure: The patient underwent cholecystectomy with removal of choledochal cyst and reconstruction by Roux-en-Y hepaticojejunostomy, appendectomy, and needle biopsy of the liver.

Operative Findings: An extended Chevron incision was made. The liver appeared grossly normal. The gallbladder was full and had a “rubbery” feel on gentle pressure and the bile duct was enlarged.

Pathology: On gross examination the gallbladder contained a tan-red exophytic, papilliferous mass attached to the mucosal surface at the fundus. It measured 4.6 X 3.7 X 2.4 cms. Histologically, the mass was composed of
hyperplastic and disorganized mixture of smooth muscle and mucosa. Intestinal metaplasia was readily evident. Focally, the lamina propria was expanded by edema. Although the epithelium had reactive and regenerative atypia, there was no evidence of dysplasia.

Discussion:

Peutz-Jeghers syndrome (PJS) is an autosomal-dominantly inherited syndrome characterized by hamartomatous polyps in the gastrointestinal tract, pigmented macules of mucous membranes and skin, and a predisposition to gastrointestinal and other tumors. There is no sex predilection. The usual age of presentation is in the second to third decade of life. The most common germline mutation is in the STK11 tumor suppressor gene on chromosome 19p13.3. Due to the variable and incomplete penetrance of the inherited allele, PJS patients show significant phenotypic variability. Microscopically, the hamartomatous polyps are characterized by an arborizing pattern of smooth muscle which is derived from the muscularis mucosae and surrounds lobules of hyperplastic glandular epithelium, typical of the involved segment of the gastrointestinal tract. The small bowel is the most common affected site followed by colon, stomach, duodenum, and appendix. Extra intestinal polyps may occur rarely and the possible sites include the respiratory tract, urogenital tract, and gallbladder, as in the current discussion. The morbidity and mortality of Peutz-Jeghers syndrome are related to the development of tumors including gastrointestinal, breast, lung, pancreas, and ovary, uterus, cervix, and Sertoli cell tumors of the testes. Cancer risks are similar in PJS patients with identified STK11 mutations and those with no detectable mutation. It has been proposed that the finding of solitary Peutz-Jeghers Polyp (PJP) may not be syndromic and that the lifetime risk for malignancy in these patients may not be as high as in the syndromic patients. However, recent studies have shown that thorough clinical examination and extensive biopsy sampling in patients who present with solitary JPs reveals evidence supportive of the syndrome. It can be speculated that these patients show extreme phenotypic variability due to incomplete penetrance of the inherited allele as discussed above. Furthermore, patients with only one PJP may have a cumulative lifetime risk of cancer similar to those with the syndrome, warranting similar follow up. Since about 30-40% of the patients do not have the STK11 gene mutation, the available genetic tests can not be used to rule out the syndrome.

Figure 1: The gross specimen showing a large polypoid mass at the fundus of the gallbladder.

Figure 2: Histopathology of the mass reveals a disorganized mixture of muscle and mucosa, characteristic of hamartomatous Peutz-Jeghers polyp.

Figure 3: Despite the large size of the lesion, the glandular epithelium shows only reactive atypia and there is no evidence of dysplasia.

Figure 4a: Longitudinal ultrasound image through the gallbladder shows a large mass, which is hyperechoic compared to the adjacent liver, filling the body and fundus of the gallbladder (arrowheads). There is no posterior acoustic shadowing. Note the normal fluid-filled gallbladder neck (arrow).

Figure 4b: Color-Doppler image shows blood flow in the mass, excluding the diagnoses of sludge or blood clot.

Figure 5a: Selected axial image from an intravenous contrast-enhanced CT demonstrate a dense mass filling the gallbladder (arrow).
Radiographic Discussion: At sonography, gallbladder polyps are isoechoic to soft tissue and, unlike gallstones, they are attached to the gallbladder wall and lack posterior acoustic shadowing (Figure 4a). Hamartomatous gallbladder polyps have rarely been described in patients with Peutz-Jeghers syndrome. They may be solitary or multiple and may be quite large. A vascular pedicle may be demonstrated with color Doppler imaging (Figure 4b). On CT images, they project into the gallbladder lumen and may be shown to enhance with intravenous iodinated contrast administration (Figures 5a & 5b).

References:


