Pseudoxanthoma elasticum: a rare cause of recurrent gastrointestinal bleeding in a child

Pseudoxanthoma elasticum (PXE) is an inherited connective tissue disorder characterized histologically by elastorrhexis affecting the elastic tissues in the dermis, blood vessels and Bruch’s membrane of the eye. The diagnosis is based on the presence of classical skin lesions, angiod streaks, and demonstration of characteristic findings on skin biopsy.1 Patients with PXE are known to present with recurrent upper gastrointestinal (UGI) bleeding.2,5

An 11-year-old boy presented with recurrent painless UGI bleeding (hematemesis and melena) since the age of 4 years. None of the bleeding episodes was associated with ingestion of drugs, jaundice or encephalopathy. Repeated UGI endoscopies, abdominal ultrasonography, barium meal follow-through and liver biopsy had not revealed any diagnosis. During the present admission, he had severe pallor and splenomegaly. On UGI endoscopy, there were no esophageal varices but there was a pool of blood in the gastric fundus. In addition, the gastric folds appeared prominent in the gastric fundus and an active ooze was seen. Ultrasonography showed normal liver and portal vein; the splenic vein could not be visualized.

Despite blood transfusion and gastric tamponade with a Sengstaken-Blakemore tube, GI bleeding continued. With a provisional diagnosis of portal hypertension due to splenic vein thrombosis, gastric devascularization and splenectomy were done.

One month after surgery, the patient was noted to have multiple papules of 1 mm to 4 mm diameter; a few of those coalesced to form plaques, all round the neck, axilla and groin. The skin was lax and redundant in the neck and its surface was rough and plebby. Ocular fundus examination revealed angiod streaks and multiple peripapillary mottling in both eyes. Skin biopsy from the involved area in the neck showed focally thickened epidermis, fragmented elastic fibers in the reticuldermis along with collection of histiocytes, and giant cells in the dermis and calcification. Repeat endoscopy showed normal gastric mucosa. Histology of the splenectomy specimen revealed fragmented elastic fibers in the large-sized muscular arteries, with calcification. Ultrasonography and CT scan of the abdomen showed diffusely scattered calcific specks in the cortex and medulla of both the kidneys, suggesting nephrocalcinosis. Biochemical investigations including serum calcium, serum phosphate, serum proteins, renal function tests, and 24-hour urinary excretion of sodium, potassium, calcium, inorganic phosphate, creatine and proteins were within normal limits. Urinary ammonium chloride load test for acid excretion and arterial pH were also normal.

In a review of 200 patients with PXE collected from the literature, GI bleeding was reported in 13% of patients.2 It is usually gastric in origin, and recurrent.2-5 Our patient had the first episode of UGI bleeding at the age of 4 years, which is the youngest age at onset of bleeding reported in patients with PXE. GI bleeding is thought to result from degeneration of the elastic fibers in the arterial wall, which leads to aneurysmal dilatation and subsequent rupture of the vessels.2 The inability of arterioles to retract also increases chances of hemorrhage from unrelated causes such as peptic ulcer disease or other mucosal injuries.2,3,4 The characteristic endoscopic findings include distinctive yellow cobblestone appearance or nodular raised submucosal lesions similar to xanthoma-like lesions of the skin as seen in this condition.5

Our patient also had evidence of nephrocalcinosis. The renal calcification in PXE is generally limited to the cortico-medullary junction. To the best of our knowledge, diffuse renal calcification has not been described earlier in patients with PXE. It is possible that these calcific specks represent areas of vascular degeneration with calcification in the renal parenchyma.

There is no specific treatment for PXE. Anti-secretory drugs and vasoconstricting agents are frequently unsuccessful in controlling bleeding.2,5 Angiographic embolization of involved vessels has been used with variable results.2,3,4 Partial gastrectomy, total gastrectomy, oversewing of the bleeding site and gastric devascularization are the usual surgical options in patients who have recurrent UGI bleeding.2,5 Gastric devascularization done in our patient with a presumptive diagnosis of portal hypertension incidentally is also a mode of treatment for gastric bleeding in patients with PXE.

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Second primary malignancy of oropharynx with hepatocellular carcinoma

A 60-year-old man, consuming alcohol for over 20 years, was on regular follow up for ulcer-type dyspepsia of 2 years’ duration.
At endoscopy he had had a benign, gastric ulcer in the juxta-pyloric region, which was treated. He presented recently
with dysphagia and hoarseness of voice for 15 days, in addition to anorexia, epigastric pain and discomfort.

On examination, the patient was sick; tenderness was present in the epigastrum. Liver was enlarged; it felt nodular and hard. Ultrasonography showed a heterogenous mass, 8 cm
× 5 cm, involving the 5th and 8th segments of the liver, and two 2 cm × 2 cm satellite bull’s eye lesions adjacent to the large
tumor. Serum alpha fetoprotein level was 60,000 ng/mL. Fine needle aspiration cytology of the lesion confirmed hepatocellular carcinoma (HCC). Upper GI endoscopy revealed a poly-poidal fleshy growth involving the oropharynx posterior to the epiglottis. Biopsy confirmed squamous cell carcinoma.

Oropharyngeal squamous cell carcinoma has high predilection for development of a second primary lesion, the incidence varying from 10%-27%.1,2 A majority are in the upper aerodigestive system. The ‘field cancerization theory’ suggests that multiple neoplastic lesions of independent origin occur within an epithelial field in response to chronic tobacco and alcohol exposure in combination with endogenous processes.3

There is no reported increase in risk of cancer in viscosa outside the respiratory and upper digestive tract. In one series4 of 440 cases of HCC, 13 had second primary and in 4 the site was colorectum. One case of esophageal carcinoma was identified. None had oropharyngeal carcinoma. This case is being reported for the rare combination of two primary sites of malignancy of the GI tract of two different cell origin.

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