Abstract

Cholestyramine resin for erythropoietic protoporphyria with severe hepatic disease: a case report

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Erythropoietic protoporphyria (EPP) is a rare disorder of heme biosynthesis caused by mutations in the gene encoding the enzyme ferrochelatase. In EPP, deficient ferrochelatase activity leads to the excessive production and biliary excretion of protoporphyrin (PP). The major clinical features of EPP are photosensitivity and hepatobiliary disease that may progress to severe liver disease, that are caused by the toxicity of PP. EPP–related liver disease has been treated medically or surgically including liver transplantation. We described a 20-year-old male with severe liver disease who was diagnosed with EPP based on clinical and laboratory findings. He was treated with cholestyramine resin. Six months after the treatment, he was doing well without any abdominal pain or photosensitivity. (Korean J Hepatol 2010;16:83-88)

Key words: Erythropoietic protoporphyria; Cholestyramine; Liver
photosensitivity and hepatobiliary disease, are related to the biochemical abnormalities. Most symptomatic patients are photosensitive due to the photoactive injury produced by PP in skin tissue.\(^4\) Liver disease, a less common clinical manifestation of EPP, is occurring in about 10% of patients, and can vary from mild biochemical abnormalities to hepatic failure. Liver disease is caused by the toxic effects of PP on hepatobiliary structure and function.\(^5\)

Several treatment modalities for liver disease in EPP patients have been described, including suppression of erythropoiesis, the molecular adsorbents recirculating system and fractionated plasma separation and adsorption.\(^6\),\(^7\) Liver transplantation is another possibility, however, the survival rate in a study of 20 EPP patients who underwent transplantation was fairly low.\(^8\) None of these treatments are effective in all patients, each has potential problems, and none has been applied to sufficient numbers of patients to allow rigorous evaluation of efficacy.

We described an EPP patient with severe liver disease in whom administration of cholestyramine resin improved clinical symptoms and liver function.

**Case report**

A 20-year-old man visited our institution with abdominal pain, accompanied by jaundice and erythema on light-exposed areas, mainly on the forearms (Fig. 1). Erythematous skin lesions were noted soon after birth. Since his late teens, the patient was aware of a skin disorder of poorly demarcated erythema and edema mainly on both hands and the wrists under the lights, for which he was treated intermittently for many years. In August 2008, he developed jaundice and severe abdominal pain, especially at right upper quadrant. Abdominopelvic computed tomography (CT) scans showed hepatosplenomegaly without focal lesions on intraabdominal solid organs including liver, bile duct, and pancreas. During hospitalization, his liver function was disturbed. He was transferred to our center for additional evaluation and management. He had no family history of hepatitis, pancreatitis, and skin disorders. He did not display history of hepatitis and consumed alcohol socially. Physical examination showed an enlarged spleen and liver, which were tender upon palpation. Biological evaluation showed bicytopenia (white blood cells 6,700/mm\(^3\), hemoglobin 11.2 g/dL, platelets 105,000/mm\(^3\)), normal blood protein (6.6 g/dL) and albumin levels (3.6 g/dL), abnormal liver enzyme levels (aspartate aminotransferase 118 IU/L, alanine aminotransferase 26 IU/L) with jaundice (direct bilirubin/indirect bilirubin 12.3/8.2 mg/dL), elevated alkaline phosphatase (201 IU/L), and \(\gamma\)-glutamyltransferase (1,063 IU/L). Prothrombin time and activated partial thrombin time were 88.5% and 28.3 seconds, respectively. He was negative for hepatitis B virus (HBV) surface (HBs) antigen, antibody to hepatitis C virus (HCV), and IgM antibodies to hepatitis A virus, but positive for antibodies to HBs. He was positive for uroporphyrin and coproporphyrin (>250 ug/L).