A Case of Progressive Familial Intrahepatic Cholestasis Type 2 diagnosed in 25-years old man

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Progressive Familial Intrahepatic Cholestasis, type 2 (PFIC II) is a rare autosomal recessive genetic disorder, which is caused by defects in the ABCB11 gene that codes for bile salt export pump (BSEP). It usually presents in childhood, and is associated with life-threatening cholestasis or liver failure. We report a very rare case which was diagnosed as PFIC II in young adult. A healthy 25-year-old male patient visited our hospital due to jaundice and pruritus for a week. He is a non-drinker, but took herbal medicine for a month. Initial laboratory tests showed elevated serum AST (132 IU/L), ALT (154 IU/L), and alkaline phosphatase (140 IU/L) levels and direct hyperbilirubinemia (total/direct bilirubin; 14.6/12.7 mg/dL), but serum rGT (24 IU/L) level was normal and prothrombin time (PT) was 86.3%. Abdominal CT scan was unremarkable. Serologic markers such as viral hepatitis A, B, and C, autoimmune disease or Wilson disease were negative. Therefore, we initially thought there was a high chance of drug induced liver injury. During the first week of hospitalization, bilirubin level and PT have worsened to 27.1 mg/dL and 67.3%, respectively. We performed liver biopsy and it showed severe cholestasis and markedly decreased BSEP expression (Figure1). This result strongly suggested PFIC II. We started high dose ursodeoxycholic acid (15mg/kg/day) and prednisolone (30mg/day). At the same time, in order to confirm PFIC II, we went through mutation test for ABCB11 gene and found out heterozygous variants (p.G78R and p.G648C) in ABCB11 gene. His parents also had heterozygote carrier of the genetic mutations. Finally he was diagnosed with PFIC II. In the first month of drug therapy, his liver function showed some improvement, but then got worse again such as PT has worsened to 42.6%. Thus, we made a decision to do liver transplantation (LT). Finally, he has undergone living donor LT, which was 4 months after onset of symptom. After LT, liver function has completely recovered.

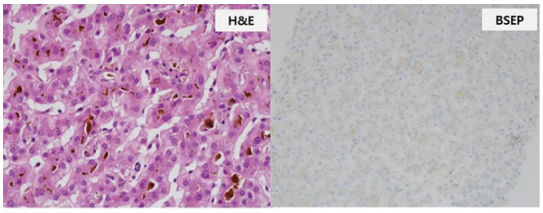

Figure 1. Histopathologic findings of liver tissues showed severe cholestasis and bile duct damage (H&E). Immunohistochemical staining for BSEP showed markedly decreased BSEP expression.