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Hassab's operation for Joubert syndrome with congenital hepatic fibrosis: A case report

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ABSTRACT

INTRODUCTION: Joubert syndrome is characterized by psychomotor developmental delay, hypotonia, oculomotor abnormalities, occasional retinal dystrophy and cystic kidneys, and frequent and often, striking breathing abnormalities, especially in the neonatal period, with panting tachypnea followed by apnea. We report a case of Joubert syndrome with hepatic fibrosis, portal hypertension, and pancytopenia treated by Hassab's operation.

PRESENTATION OF CASE: Our patient was a 27-year-old woman with a history of tachypnea, muscle hypotonia, and psychomotor retardation shortly after birth and a diagnosis of Joubert syndrome at 2 years of age. At 19 years of age, she was diagnosed with progressive pancytopenia. At 27 years of age, she visited her local doctor for sudden-onset hematemesis. Endoscopy revealed esophageal varices exhibiting the red color sign and no evidence of recent bleeding. Splenomegaly and development of portal collateral circulation were observed on computed tomography scans.

The patient was referred to our hospital, where she was diagnosed with Joubert syndrome and hepatic fibrosis, portal hypertension, and hypersplenism. After performing Hassab's operation, the pancytopenia improved, but anticoagulant therapy was required for splenic vein thrombosis. The patient was discharged on postoperative day 25. Two years following surgery, the gastroesophageal varices were controlled, and no progression of the splenic vein thrombosis or hepatic failure was evident.

CONCLUSION: This is the first case report of Hassab's operation for congenital hepatic fibrosis in a patient with Joubert syndrome, a rare congenital condition. We achieved a favorable clinical outcome.

1. Introduction

Joubert syndrome is an autosomal recessive disorder that first reported in 1969 [1]. It is characterized by psychomotor developmental delay, hypotonia, oculomotor abnormalities, occasional retinal dystrophy and cystic kidneys, and frequent and often, striking breathing abnormalities, especially in the neonatal period, with panting tachypnea followed by apnea. Joubert syndrome and related disorders (JSRD) are used to describe individuals with Joubert syndrome accompanied by additional findings including retinal dystrophy, renal disease, ocular colobomas, occipital encephalocele, polydactyly, oral hamartomas, endocrine abnormalities and hepatic fibrosis [2]. We report a case of Joubert syndrome with hepatic fibrosis, portal hypertension (PH), and pancytopenia treated by Hassab's operation. To the best of our knowledge, our patient is the first such case to be reported.

2. Presentation of case

Patient: 27-year-old woman

Family history: Older sister with Dandy–Walker syndrome

History of present illness: On postnatal day 2, the patient was treated by her local doctor for tachypnea. At 1 year of age, she was noted to have muscle hypotonia and psychomotor retardation, and she was diagnosed with Joubert syndrome. At 19 years of age, she suffered from increasingly severe anemia, with pancytopenia progressing despite the prescription of iron formulations. At 25 years of age, a bone marrow biopsy ruled out hematological disease. At 27 years of age, following the sudden onset of hematemesis, an upper gastrointestinal endoscopy revealed esophageal varices that were positive for the red color (RC) sign without any evidence of active

Abbreviations: JSRD, Joubert syndrome and related disorders; PH, portal hypertension; RC sing, red color sign; CT, computed tomography; RV, reference value; CHF, congenital hepatic fibrosis; EVL, endoscopic variceal ligation; EIS, endoscopic injection sclerotherapy; PSE, partial splenic artery embolization; DOAC, Direct oral anticoagulant; VKA, vitamin K antagonist; PSVT, : portal vein or splenic vein thrombosis.

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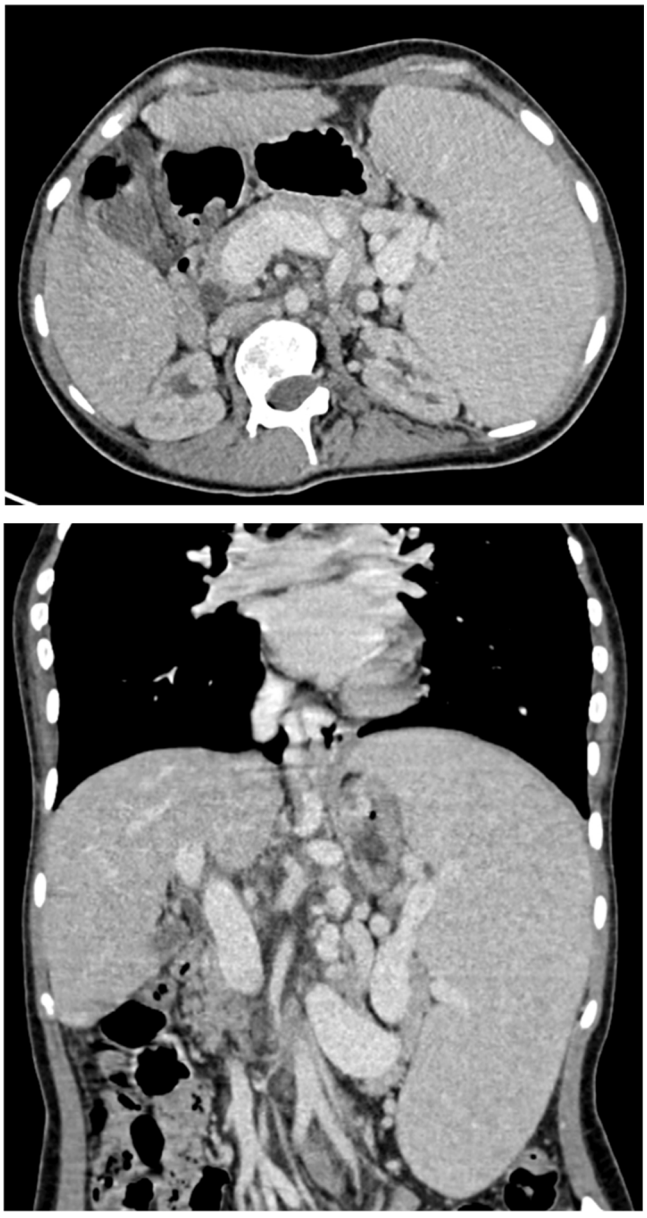

Fig. 1. Contrast-enhanced CT scan before operation. Spleen enlargement was observed, and development of portal collateral circulation and gastroesophageal varices were noted. A small volume of ascites was present, and bilateral renal atrophy was observed.

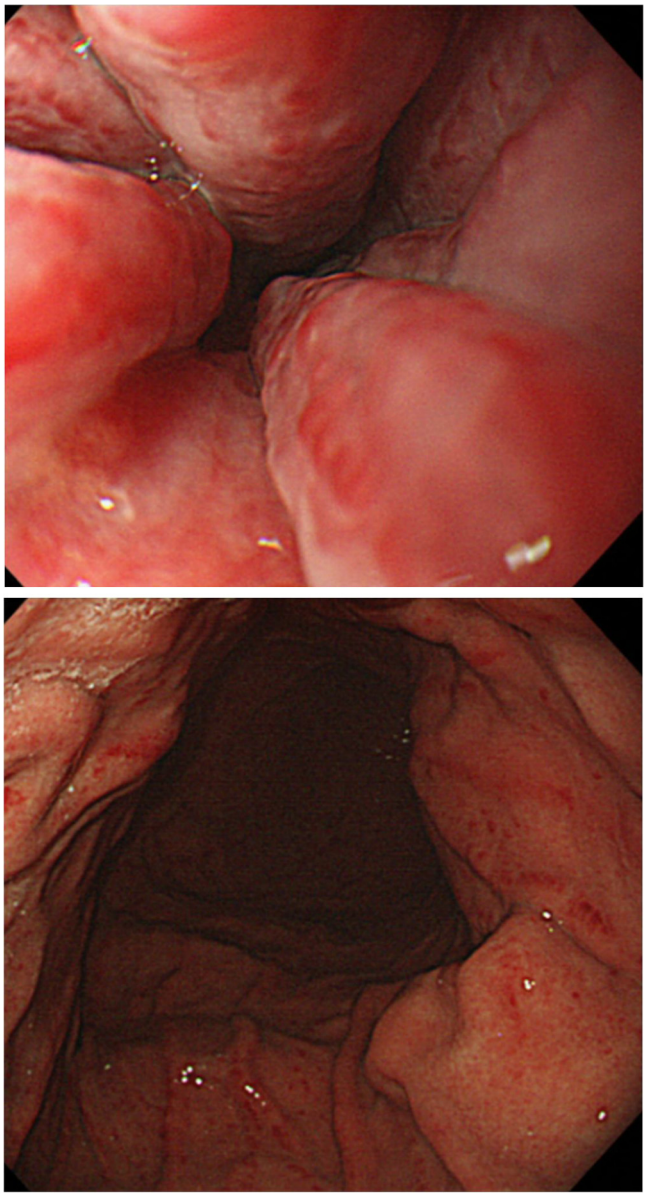

Fig. 2. Upper gastrointestinal endoscopy before operation. Esophageal (in the upper third of the esophagus, F2-3, Cb, and RC2) and gastric (F1, RC0) varices near the cardiac orifice were confirmed.

bleeding. She was kept nil per mouth and started on proton pump inhibitors with no recurrence of hematemesis. Because the computed tomography (CT) scans and abdominal ultrasound revealed splenomegaly, the patient was referred to our department for further treatment of the splenomegaly, pancytopenia, and esophageal varices.

On initial examination, body height was 156.3 cm, body weight was 33.8 kg, body mass index was 13.8 kg/m^2, body temperature was 37.2 °C, blood pressure was 99/65 mmHg, and pulse was 85 beats/min. An enlarged spleen was palpable as a mass on the left abdominal region. Laboratory findings were a white blood cell count of 1000/μl [reference value(RV): 3300–9000/μL], her hemoglobin was 9.5 g/dL (RV: 11.5–15 g/dL), and her platelet count was 20×10^3/mm^3 (RV:150–400 × 103/mm3). She had a Child–Pugh score of A (6 points). The indocyanine green retention rate at 15 min was 13%. The blood urea nitrogen level was 33 mg/dL (RV: 8–20 mg/dL), serum creatinine level was 2.13 mg/dL (RV: 0.4–0.8 mg/dL).

An abdominal CT scan found that the hepatic margin appeared blunted. Spleen enlargement was observed, and development of portal collateral circulation and gastroesophageal varices were noted. A small volume of ascites was present, and bilateral renal atrophy and renal cysts were observed (Fig. 1). Upper gastrointestinal endoscopy found esophageal (in the upper third of the esophagus, F2-3, Cb, and RC2) and gastric (F1, RC0) varices near the cardiac orifice (Fig. 2). Gastroesophageal varices were classified on the basis of the criteria used to describe endoscopic findings in Japan. In brief, the severity of gastroesophageal varices was classified as follows: F1, straight and small-caliber varices; F2, beady varices; F3, tumor-shaped varices; Cw, white varices; Cb, blue varices; RC0, absence of red color (RC) sign; RC1, a few RC signs; RC2, several RC signs; and RC2, many RC signs. The patient was diagnosed with Joubert syndrome with congenital hepatic fibrosis (CHF), which was deemed to be consistent with the observation of