Successful Redo Pull-through for Hirschsprung’s Disease in a Haddad Syndrome Patient

Jih-Teng Lee, Chinsu Liu*, Hsin-Lin Tsai, Taiwai Chin, Chou-Fu Wei

Division of Pediatric Surgery, Department of Surgery, Taipei Veterans General Hospital and National Yang-Ming University School of Medicine, Taipei, Taiwan, R.O.C.

Haddad syndrome is characterized by congenital central hypoventilation syndrome (Ondine’s curse) associated with segmental distal gut aganglionicosis (Hirschsprung’s disease). The prognosis of Haddad syndrome is very poor, and survival is often less than 2 years. Treatment of Hirschsprung’s disease is usually influenced by the association with Ondine’s curse. We report the case of a girl with Haddad syndrome who underwent redo pull-through with Duhamel’s method because of persistent obstruction after primary transanal pull-through surgery. After 7 years of follow-up, the patient is alive and does not suffer from recurrent partial intestinal obstruction. She performs her daily activities with the support of a portable ventilator. Thus, aggressive surgical treatment for Hirschsprung’s disease could have good outcome in terms of long-term survival in Haddad syndrome patients. [J Chin Med Assoc 2010;73(8):438–440]

Key Words: Hirschsprung’s disease, Ondine syndrome, reoperation, sleep apnea

Introduction

Haddad syndrome, described by Haddad et al in 1978, is characterized by congenital central hypoventilation syndrome (CCHS) occurring in association with Hirschsprung’s disease. Even though extensive therapy is available for the treatment of this syndrome, its prognosis is very poor. Here, we present the case of a long-term survivor of Haddad syndrome who underwent tracheotomy and redo pull-through operation.

Pull-through operation is the most favored treatment for Hirschsprung’s disease, and most patients who undergo this operation tend to do well over time. However, the surgical treatment is usually affected by the associated condition. Some investigators even question the use of aggressive care in this challenging condition. The time point required and procedures for redo pull-through operation are still being debated. The patient we describe here had Haddad syndrome and underwent redo pull-through with endogastrointestinal anastomosis (endoGIA)-assisted Duhamel operation for persistent obstruction syndrome. Although the disease course was complex, the outcome was good.

Case Report

The girl was born to a gravida 2, para 2 mother after 37 months of gestation by cesarean delivery. Polyhydramnios had been noted in the 7th month of gestation. Cyanosis was noted and the baby did not cry, so she was immediately intubated. Delayed meconium passage and persistent abdominal distension were noted after birth. Hirschsprung’s disease was diagnosed, and the transition zone was found to be over the rectosigmoid colon. She underwent transanal pull-through operation at the age of 23 days. However, weaning from ventilatory support was difficult. The patient had...
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not suffered any injuries at the time of birth and had not undergone any thoracic operation; these facts and the findings of brain magnetic resonance imaging indicated CCHS. A genetic investigation showed the presence of a point mutation in the paired-like homeobox gene 2B (PHOX2B), which was indicative of Haddad syndrome. Owing to persistent intestinal obstruction after primary surgery, another enterostomy over the transverse colon was created for fecal diversion. The patient was transferred to our hospital at the age of 9 months for transverse colostomy closure.

On admission, she weighed 6 kg (<3rd percentile). Segmental coloanal stenosis was noted after double-contrast barium enema (Figure 1). Digital examination revealed a stenosis 5 cm from the anal verge. Redo pull-through was performed using endoGIA-assisted (Tyco Auto Suture Endo GIA Straight 45-3.5; United States Surgical, A Division of Tyco Healthcare Group, Norwalk, CT, USA) Duhamel operation. Owing to previous operations, severe bowel adhesion was noted, and it was difficult to lyse the adhesive bowel. The pelvis of the little girl was shallow and small; more caution was taken during pelvic dissection for fear of pelvic organ, nerve and vessel damage. A larger retrorectal cuff space was created to prevent bowel vessels kinking and twisting, which may lead to future acquired aganglionosis. The ganglionic bowel was pulled through the posterial midline of the rectal cuff after complete resection of the stenotic segment. The patient was under full-time ventilator support before the age of 6 years. The associated Ondine’s curse did not influence the redo pull-through. A Hickman catheter was inserted immediately after the redo pull-through operation for nutritional support. The pathology report revealed the presence of ganglion cells over the proximal and distal ends of the resected colon sample. The transverse colostomy was closed 65 days after the redo pull-through operation. The Hickman catheter was removed at the same time. Unfortunately, persistent abdominal distension recurred 93 days after the redo pull-through. Mechanical bowel obstruction was suspected, so entrolysis was performed 99 days after the redo pull-through. The patient was discharged 119 days after the redo pull-through with adequate enteral feeding.

She suffered from abdominal distention and vomiting 6 and 23 months after the operation; these symptoms resolved after partial parenteral nutrition support was provided. Four years after the redo pull-through, she remained completely free of intestinal obstruction. She had pulmonary tuberculosis (TB) at the age of 3 years, which resolved after she was given a full course of anti-TB medication. She was admitted at the age of 4.5 years because of a seizure attack. Both electroencephalography and brain magnetic resonance imaging did not show any abnormal findings. When she was 6 years old, she was weaned from the full-time ventilator; since then, she has required ventilatory support only at night. She is now 8 years old, 97 cm in height (<3rd percentile), and 16 kg in weight (<3rd percentile). She attends elementary school with the support of a portable ventilator and is capable of ambulatory movement.

Discussion

Haddad syndrome is a rare congenital disorder with a high mortality rate. The prevalence of CCHS is 1 in 200,000 live births; Hirschsprung’s disease is present in 16% of these cases. However, the mortality rate is much higher in the case of Haddad syndrome than in the case of Hirschsprung’s disease alone. The mortality rate of Haddad syndrome does not differ from that of CCHS alone. The death of CCHS patients appears to be directly associated with ventilator dependence. Despite advances in home ventilator support and the surgical technique for Hirschsprung’s disease and total parenteral nutrition, the median survival for Haddad syndrome patients is still less than 2 years. Our patient suffered from recurrent pneumonia and pulmonary TB, for which she received systemic antibiotics and full-course anti-TB medication, respectively. She was in the respiratory care ward until the age of 6 years. From the age of 6, she no longer required daytime ventilation. She will require lifelong respiratory care, which will be a challenge for her.
Pull-through operation is the definitive treatment for Hirschsprung’s disease. Myectomy-myotomy has also been reported to have a good outcome for Hirschsprung’s disease in Haddad syndrome. However, the poor outcome of Haddad syndrome deters patients from undergoing aggressive treatment and leaves the colostomy open, which causes long-term suffering. Our patient was brought to our hospital because of the failure of primary pull-through with a colostomy. From their study, Schweizer et al concluded that only a second resection of the transition zone and redo pull-through procedure can provide a definite solution. EndoGIA-assisted Duhamel operation was used as the redo pull-through procedure for our patient. Langer considered this to be the “most simple type of redo pull-through”. The aganglionic zone in our patient was a short segment, which is less common in Haddad syndrome patients who usually have longer aganglionic zones. Because the segment is short, long-term administration of enteral nutrition is possible; this prevents mortality and morbidity resulting from alimentary defects. To the best of our knowledge, this is the first report on redo pull-through operation for Hirschsprung’s disease in a Haddad syndrome patient. From our experience, redo pull-through for persistent obstruction syndrome could be useful for Haddad syndrome patients.

Growth retardation was also noted in our patient (<3rd percentile); it was also reported in 2 other isolated CCHS cases. The relationship between Haddad syndrome and growth retardation is unclear, but it may involve the hypothalamic–pituitary axis.

The disease course was long and difficult in our patient. From the observations in our patient, we believe that aggressive surgical treatment and meticulous respiratory care could allow long-term survival of Haddad syndrome patients.

References