IS-009
A case of triple gastrointestinal anomalies of imperforate anus, malrotation, and Hirschsprung’s disease associated with holoprosencephaly

National Center for Child Health and Development
Nobuyuki Morikawa, Toshiro Honna, Tatsuo Kuroda, Miwako Nakano, Kiyoshi Tanaka, Yoshihiro Kitano, Hiromitsu Matsuda, Keiichiro Tanaka, Noriko Kawashima

A one-month-old girl presented with anocutaneous fistula and mild facial phenotypes of holoprosencephaly such as median cleft lip and ocular hypotelorism. Plain radiographs demonstrated a dilated sigmoid colon and right-sided small bowel location, suggesting the association of Hirschsprung’s disease and malrotation. Contrast studies of the gastrointestinal tract and pathology of the rectal biopsy confirmed the diagnosis of malrotation and Hirschsprung’s disease. One-stage operation including cut back, Ladd, and endorectal pull-through procedures was performed at 2 months of age. Although she developed severe hypoglycemia due to hypopituitarism requiring hormonal replacement, the postoperative course was otherwise uneventful. It has been reported that Hedgehog signals are essential for organogenesis of the mammalian gastrointestinal tract. Since Hedgehog mutations have been shown to cause multiple gastrointestinal anomalies including imperforate anus, malrotation, and Hirschsprung’s disease in mice as well as holoprosencephaly in humans, mutations in Hedgehog gene may account for the pathogenesis of the complicated anomalies in this patient. Although this is the first report of the triple association of imperforate anus, malrotation, and Hirschsprung’s disease diagnosed preoperatively followed by one-stage operation, one should consider such unusual coexistent pathologies for correct diagnosis and appropriate management.

IS-010
Proper treatment of megacolon after various anoplasties for anorectal malformation

Division of Pediatric Surgery, Yonsei University College of Medicine, Seoul, Korea
Seung Hoon Choi, Airi Han, Yong Tak Koh, Jung Tak Oh, Seok Joo Han, Eui Ho Hwang

Purpose: The megacolon after repairing an anorectal malformation is not a rare complication, and there is much controversy on the causes, the treatment choice and the results after a longterm follow-up. We present 5 cases of a megacolon after the repair of an anorectal malformation, which were controlled with either a surgical resection or conservative treatment.

Methods: Five patients with a megacolon after the repair of an anorectal malformation were studied. A retrospective chart review was done and fecal continence was evaluated with an individual interview.

Results: All five patients initially underwent conservative treatment with laxatives and/or enemas. One patient responded well to conservative treatment and the diameter of the bowel reduced to normal size. Another patient responded to conservative treatment after correcting the location of the anus. Three patients needed a surgical resection and one of those needed a further procedure to correct the anal location. After the surgical resection of the megacolon and/or correction of the anus, they soon reported an almost normal bowel habit.

Conclusion: The first step in treating a megacolon after repairing an anorectal malformation was conservative treatment. However patients without an adequate response to conservative treatment are best managed with a surgical resection. The cause of the megacolon is now under investigation and the lack of adequate management after repair is one of the subjects.