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Review Article

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A RARE CASE OF HIRSCHSPRUNG'S DISEASE IN A PATIENT WITH DIFFUSE ADENOMATOUS POLYPOSIS

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ABSTRACT

The case of an 11-year-old Kazakh male who had a past history of Hirschsprung's disease is presented. He underwent transanal endorectal resection of the rectosigmoidal colon at the age of 46 months. He was also diagnosed as having diffuse adenomatous polyposis of large intestine. The aim of the publication is to analyze modern ideas about the peculiarities of diagnosis in the combination of Hirschsprung's disease with diffuse adenomatous polyposis of large intestine in children and to present a rare clinical case from our own practice. This is the first case of familial adenomatous polyposis associated with Hirschsprung's disease from Kazakhstan.

KEYWORDS: Familial adenomatous polyposis, Diffuse familial polyposis, Hirschsprung's disease, surgical treatment.

INTRODUCTION

Hirschsprung's disease is a congenital disorder characterized by absence of ganglion cells, which leads to obstruction in the distal gastrointestinal tract.[1] Little is known about the relation of Hirschsprung's disease to other congenital intestinal malformations, such us familial adenomatous polyposis (FAP). FAP is an autosomal dominant inherited disorder defined by numerous polyps of colon, which develop colorectal carcinoma if treatment is not provided.[2] A boy with FAP associated Hirschsprung's disease presented. Case presentation A fullterm (39 weeks gestation) 3950 g., a Kazakh baby boy was born to a second gradiva mother with moderately severe iron deficiency anemia. Mother lives on the territory of the former Semipalatinsk nuclear test site in the Abay region. He had failed to pass meconium spontaneously for the first 2 day and later passed after conservative treatment. A barium study was suggestive of dolichosigma. From 3 months of age, the child started having abdominal distension and constipation. The child was periodically hospitalized with the diagnosis Dolichosigma. At the age of 2 years, an irrigography was performed at Semey medical university hospital. A narrowing was revealed in the rectosigmoidal part of the large intestine for 4-5 cm and above it there was a suprastenotic expansion (Figure 1). Fibrocolonoscopy with biopsy showed the absence of ganglionic cells in the nervus plexus of the colon, supporting the diagnosis of Hirschsprung's disease, rectosigmoidal form, subcompensated stage. Unfortunately parents refused from surgical l treatment. There was a deterioration in the condition, all persistent constipation disturbed, bowel movements were carried out only with the help of an enema. The transanal endorectal resection of the rectosigmoidal part of the large intestine was performed at the age of 46 months according to De la Torre-Mondragan. A narrowed area with an enlarged part within 12 cm was resected. Histological examination revealed the diagnosis of Hirschsprung's disease. At approximately 5 years of age, the patient complained about recurrent abdominal pain, constipation, fecal incontinence, mainly at night. He had failure to thrive. Height was 100 cm, weight was 17.2 kg. Examined by an endocrinologist, somatogenic dwarfism and subclinical hypothyroidism was diagnosed. On the irrigogram a year after the operation, megacolon phenomena persist, but the width of the intestinal wall is not expanded in comparison with the picture before the operation (Figure 2). At the age of 6 years performed fibrocolonoscopy with biopsy: over 25-27 cm of the colon wall throughout the entire length of multiple polyps measuring from 0.8-1.0 to 1.0 ×1.5 cm with altered mucosa (more than 100 in total) was founded. Hypertrophic adenomatous polyps of the colon, the descending part and the sigmoid colon were resected within 35 cm. Biopsy findings revealed multiple hypertrophic and adenomatous colon polyps (figure 3). Endoscopic investigation of the upper gastrointestinal tract showed absence of polyps in the stomach and duodenum. In the postoperative period, a positive trend

was noted: appetite improved, constipation and abdominal pain resolved. We made colonoscopy annually from 2017 to 2021, no pathology was revealed. After the operation, the child has an increase in height and weight. Currently, the child is 11 years old, weight 29 kg, height 130 cm, BMI 17.4. Discussion Hirschsprung's disease occurs with additional congenital and chromosomal anomalies in 30% of cases. It also associated with cancer predisposition syndromes as familial medullary thyroid carcinoma, FAP and even tumors (neuroblastoma).[3] This emphasizes the importance of careful evaluation of all Hirschsprung's disease patients. The diagnosis of FAP is mainly made by colonoscopy and the presence of 100 or more polyps that are later confirmed adenomas is a clear indication of the diagnosis. FAP is the result of a tumor suppressor mutation of the APC (Adenomatous polyposis Coli) gene, which initial clinical presentation in patients is hematochezia.[4] The most cases are asymptomatic until the polyps are large enough to cause gastrointestinal bleeding. We found only two cases of FAP associated Hirschsprung's disease. Ikeda reported 14-years-old Japanese girl with past history of Hirschsprung's disease. She had colonoscopy because of five members of her family diagnosed as having FAP. Multiple polyps in stomach, duodenum and large intestine were determined.[5] Another case characterized with an early onset of the disease from the age of 3 with intestinal manifestations, abdominal pain, and physical retardation. FAP was diagnosed by colonoscopy and genetic screening. Relatives did not have this disease.[6] In the presented clinical case, relatives also do not have a diagnosis of polyposis. There is a late diagnosis of Hirschsprung's disease and untimely surgical treatment in our case. A timely diagnosis of FAP and dynamic observation helps to recognize the presence of a malignant tumor and provides an opportunity for treatment in the early stages of the disease. To diagnose and assess the risk of cancer and better prognosis, a multidisciplinary approach of pediatric gastroenterologist, surgeons, proctologists and genetics is required. Conclusion We report a rare case of the co-inheritance of both Hirschsprung's disease and FAP. Further research is necessary to elucidate the relationship between Hirschsprung's disease and FAP.

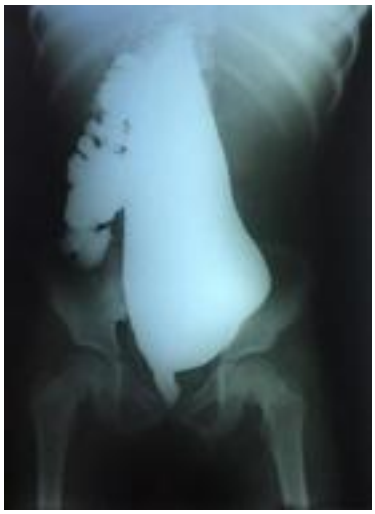

Figure 1 A narrowing with suprastenotic expansion.

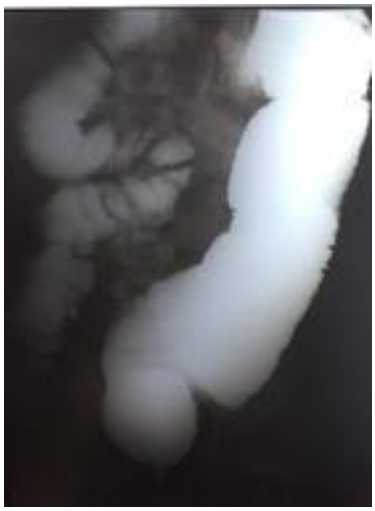

Figure 2: Irrigogram after surgical treatment of Hirschsprung′s Disease.

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