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CASE REPORT

Megacolon Caused by Hirschsprung's Disease in Pregnant Women from a Rural Area in Indonesia: A Case Report

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ABSTRACT

This case report describes a rare occurrence of Hirschsprung's disease presenting in a 26-year-old pregnant woman from a rural area in Indonesia. Initially asymptomatic for pregnancy-related issues, the patient had experienced lifelong constipation, frequently managed with laxatives, and had significant abdominal distension. At approximately 34-35 weeks of gestation, an ultrasound during a routine obstetric check-up revealed intestinal dilation potentially compressing the uterus. Subsequent referral to a regional hospital confirmed the diagnosis of Hirschsprung's disease, differentiated from teratoma through imaging and postoperative findings. A classical caesarean section was performed at 38 weeks, during which a significantly dilated sigmoid megacolon and fecaloma were discovered and managed with a distal colostomy. This case highlights the challenges of diagnosing and managing rare gastrointestinal disorders in pregnant patients, emphasizing the importance of considering congenital anomalies in differential diagnoses to ensure appropriate prenatal care and surgical intervention.

Keywords: Congenital, Hirschsprung, Pregnancy, Rural community.

INTRODUCTION

Hirschsprung's disease, referred to as congenital aganglionic megacolon, is defined by the proximal dilatation of the afflicted intestine segment due to the absence of submucosal and myenteric plexus ganglion cells in the intestinal segment.[1] The incidence of Hirschsprung's disease is higher in Indonesia, i.e., 31 instances per 100,000 live births while it is lower in Europeans, African Americans, and Asians i.e., 15, 21, and 28 cases per 100,000 live births, respectively.[2] Typically, clinicians diagnosed about 80% of Hirschsprung cases during the first year of life, making it infrequent in adolescents and adults. Common symptoms include a delay in passing meconium beyond 48 hours postnatally, abdominal distension, and vomiting. Cases in adolescents or adults often present in short and ultrashort segment forms.[3] In rare cases, adult Hirschsprung's disease can be found in pregnant women.

This case report discusses adult Hirschsprung's disease in pregnancy. The patient lives in a rural area, 40 km away from the center of the Jember. Jember is located in the province of East Java, 950 km from the capital city of Jakarta, Indonesia. Most of the people in the patient's neighborhood work as farmers. The nearest healthcare facility provides general practitioner services, while surgeons and obstetricians can be found in the center of cities.

CASE REPORT

A 26-year-old pregnant woman (G1P0000) at 34-35 weeks of gestation came to polyclinic obstetric Soebandi Regional Hospital Jember. The patient had no pregnancy-related complaints. She had noticed her stomach growing since childhood, particularly when she experienced difficulty defecating. The patient had been dealing with bowel issues since infancy, with meconium coming out after seven days postnatally, as reported by her mother. Throughout her life, the patient had relied on laxatives to facilitate defecation. She typically defecated twice daily in limited amounts, with small, hard, brown stools daily, without any signs of blood, abdominal pain, diarrhea, nausea, or vomiting.

The patient reported attending eight antenatal care appointments at the midwife and health center near her home. During her ninth follow-up for pregnancy check-up with the obstetrician, an ultrasound revealed a black shadow filled with gas, which raised suspicions of intestinal dilation pressing against the uterus and fetus. Subsequently, she was referred to the regional hospital for further evaluation and management.

Examination

The patient's general condition looked good. She had normal vital signs. Her height was 150 cm, weight was 61 kg, with a BMI of 27.1 kg/m^2. On obstetric examination, the abdomen looked distended, bowel sounds decreased, and fetal heart rate was 140 times/minute. Top of fundus height was three fingers below the Xyphoid processus (42 cm), and Head in Spine was not found. Leopold's examination showed the fetus presented with the head, back on the right, and had not yet entered the pelvic door. Genital and other physical examinations did not reveal any abnormalities. A complete blood laboratory examination revealed hemoglobin 10.1 g/dL and serum albumin 2.8 g/dL, while other parameters were within normal limits.

The patient was diagnosed with G1P0000 34-35 gestation weeks with Hirschsprung's disease, and the differential diagnosis was teratoma based on imaging findings. The patient received 60 mg of ferrous sulfate daily and planned an elective section caesarian (SC) at 38 weeks gestation. The SC surgery performed was a classical SC because the obstetrician had difficulty accessing the lower uterine segment due to a pushed uterus.

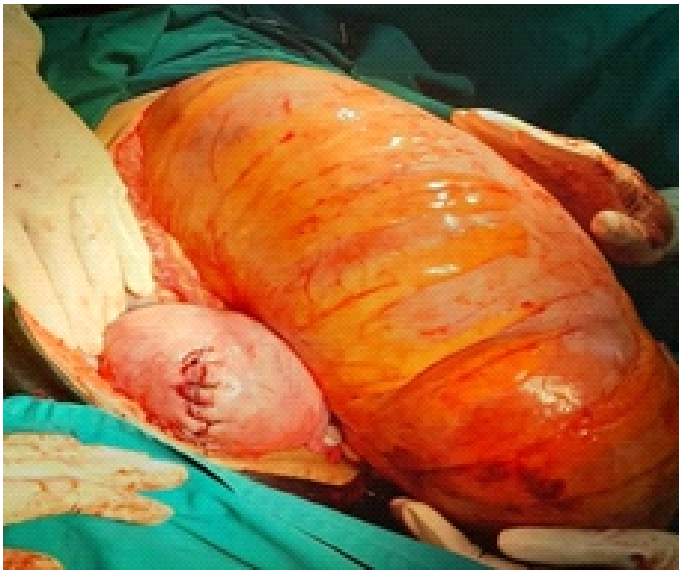

Figure 1: Uterus with dilated megacolon sigmoid during surgery

A gravid uterus with a dilated sigmoid megacolon was found (Figure 1). The baby girl was born crying immediately, weighing 2800 grams, with a body length of 48 cm and an appearance, pulse, grimace, activity, and respiration (APGAR) score of 7-8. A Surgeon then continued the surgery. The megacolon was found in the distal 1/3 of the sigmoid to the ascending colon, with a sigmoid diameter of 20 cm. In addition, fecaloma was found in the sigmoid up to the transverse colon. The evacuation of fecaloma through an incision in the sigmoid colon. The volume of feces was ±10 liters, followed by a true distal colostomy using the transverse colon segment.

Imaging

X-ray examination of the thorax showed no abnormalities. Abdominal ultrasound examination revealed meteorism, indicating distended bowel accumulating on the anterior abdominal side (left lateral) and pushing the uterus to the right. A single fetus lives intrauterine with an estimated gestational age of 34 weeks one day, with the placenta in the fundus (Figure 2). Plain abdominal radiographs in an erect position showed a ground-glass appearance with calcification, raising suspicions of a teratoma mass (Figure 3). Two weeks postoperatively, a Colon in-loop examination was performed, and the results showed a short segment description of Hirschsprung's disease (Figure 4).

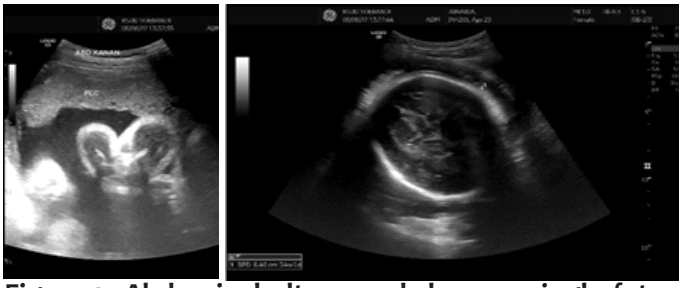

Figure 2: Abdominal ultrasound shows a single fetus living intrauterine with a gestational age of 34 weeks, and the placenta is located in the fundus

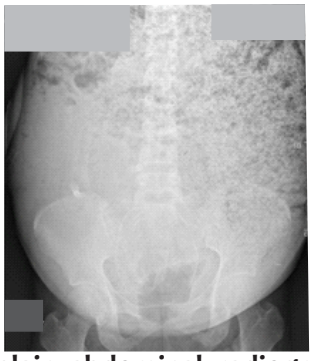

Figure 3: The plain abdominal radiograph shows a ground-glass appearance with calcification

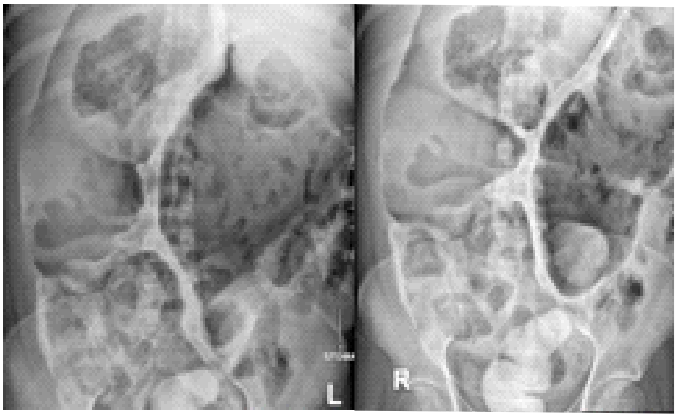

Figure 4: Contrast Imaging of the large intestine examination shows a short segment of Hirschsprung

Histopathology

A full-thickness rectal biopsy was held after the surgical wound had healed. The histopathological examination of the biopsy tissue showed the absence of ganglion cells in the middle to the distal rectum.

DISCUSSION

Megacolon is characterized by a caecum diameter greater than 12 cm, ascending colon more than 8 cm, or rectosigmoid more than 6.5 cm.[4] Though uncommon during pregnancy, its complications can markedly affect the mother's capacity to sustain the pregnancy due to pressure exerted on the uterus by the dilated colon.[5] In other cases, the mother may experience nausea, vomiting, lower abdominal pain, or severe colic pain, indicating obstruction. Those conditions can lead to delayed fetal growth and distress.[5]

Megacolons can be divided into three types: toxic megacolon, idiopathic megacolon, and acute megacolon. In a previous study, the diagnosis of Hirschsprung's disease was confirmed when the patient's mother disclosed that the patient had just passed meconium one week after birth, strongly indicating the presence of the condition.[6] In another previously published case report the patient denies fever and bloody diarrhea, excluding the toxic megacolon diagnosis due to inflammation or infection.[7] The chronic nature of the patient's megacolon rules out the diagnosis of toxic megacolon, which typically involves acute colonic dilation within 12 hours. In this case, the patient presents with adult Hirschsprung's disease, a term applied to individuals older than ten years, with a median age of 24 years. This diagnosis often stems from an undetected congenital cause (5%) or a misdiagnosis. Adult Hirschsprung's disease is frequently undiagnosed, particularly in individuals from low socioeconomic backgrounds, families with limited education, and regions with restricted access to healthcare facilities.[8] In the current study the patient's house was 40 km away from the district referral hospital, with few transportation options. The patient was a high school graduate and a housewife. Her parents worked as farmers, and her husband was a salesman with a limited salary. The patient often visited primary health facilities because of her constipation. However, she was only given the laxatives and was never advised to a reference health facility for further examination.

The first-line therapy for adult Hirschsprung's disease includes increasing dietary fiber, water intake, and daily exercise. If these are ineffective, using enemas, laxatives, and manual disimpaction is second-line therapy.[5] In this case patient admitted having never arranged her diet but regularly used a laxative enema. The patient lives in a rural area with abundant vegetables. She did not need to buy vegetables, but the patient was not consuming them.

Farmers' knowledge must be expanded, especially about potential health issues in rural areas. Only 10.8% of Indonesian farmers use the internet, which is a low usage rate. The majority of internet users are men, living on Java Island, who are young and highly educated. Regretfully, only a few Indonesian farmers exhibit these traits.[9] Farmers aware of a health issue are more likely to seek medical attention quickly.

The challenges of managing Hirschsprung cases in rural areas of Indonesia stem from patients, as mentioned previously, the characteristics of the disease, and health facilities. In Indonesia, Hirschsprung's disease is linked to particular genetic variations at RET (rs2435357) and NRG1 (rs7835688, rs16879552).[2] It is recommended to give a careful attention to genetic variation as it may also lead to additional disparities in clinical characteristics.

CONCLUSION

Hirschsprung's disease in pregnant women from rural areas is sporadic because the patient can maintain her pregnancy for up to 38 weeks without any fetal complications. Socio-economic factors, habits, and reach of health services need to be considered in the treatment. Early diagnosis is an essential step in preventing complications. History taking, physical examination, additional tests, and proper treatment were carried out to avoid complications from Hirschsprung's disease experienced by the patient,

especially during pregnancy.

PATIENT'S CONSENT: The authors certify that they have obtained all appropriate patient consent forms. In the form patient has given her consent for her clinical information to be reported in the journal. The patient understands that her name and initials will not be published.

CONFLICT OF INTEREST: All authors do not have any conflict of interest.

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