ORIGINAL RESEARCH PAPER

General surgery

WILKIE'S SYNDROME: A RARE CAUSE OF UPPER GASTROINTESTINAL OBSTRUCTION IN A PATIENT WITH FEMALE PSEUDOHERMAPHRODITISM

KEY WORDS: wilkie's syndrome, subacute obstruction, Pseudohermaphroditism

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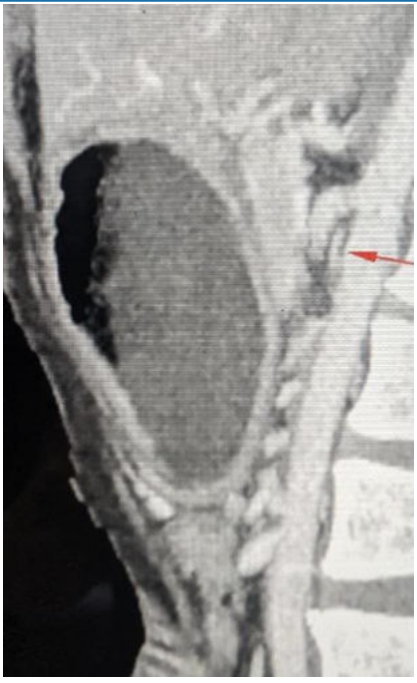
ABSTRACT

Wilkie's Syndrome, also known as Superior Mesenteric Artery Syndrome (SMAS), is a rare clinical entity that presents with symptoms of upper gastrointestinal obstruction. It occurs due to compression of the third part of the duodenum between the superior mesenteric artery and the aorta. In this article, we discuss the etiology, clinical presentation, diagnosis, and management of Wilkie's Syndrome. We also explore its association with pseudohermaphroditism and genetic abnormalities.



The Arrow In The Above Image Showing A Decreased Aorto-mesentric Angle With The Third Part Of Duodenum Compressed In Between

INTRODUCTION

Wilkie's Syndrome, first described in 1861 by Rokitansky, is a rare condition characterized by compression of the third part of the duodenum between the superior mesenteric artery and the aorta. It is also known as Superior Mesenteric Artery Syndrome (SMAS) or Cast Syndrome [1]. The prevalence of this condition is not known, but it is estimated to occur in approximately 0.1% to 3% of the population [2].The etiology of Wilkie's Syndrome is not fully understood. The condition occurs when there is a decrease in the aortomesenteric angle (AMA) from the normal range of 38-65 degrees to less than 22 degrees. This results in compression of the third part of the duodenum between the superior mesenteric artery and the aorta, leading to upper gastrointestinal obstruction [3]. Several factors have been reported to cause a decrease in the AMA, including rapid weight loss, malnutrition, scoliosis, surgery, trauma, and prolonged bed rest [4].

CASE REPORT

A 24-year-old female, presents with complaints of intermittent abdominal pain, postprandial discomfort, and unintentional weight loss over the past 2 months and the patient developed abdominal distension, vomiting and obstipation for the past 2 days. The vomitus was bilious and it occurred a few hours after good intake. Additionally, she expresses concerns about her external genitalia, specifically an enlarged clitoris resembling a male external genitalia which is giving her anxiety. The patient's assigned gender at birth is female. Upon physical examination, the patient appears underweight with evidence of malnutrition. She experiences tenderness in the epigastric region. There were no signs of peritonitis like guarding or rigidity. Additional examination reveals an enlarged clitoris resembling a penis, fused labia, and the absence of a vaginal opening. No palpable gonads are found in the inguinal regions. Laboratory tests reveal mild hypoalbuminemia, suggestive of malnutrition. Hormonal analysis indicates elevated testosterone levels, while estrogen levels remain within the normal range, consistent with female pseudohermaphroditism. Imaging with a contrast-enhanced computed tomography (CECT) of the abdomen and pelvis demonstrated a narrowed aorto-mesenteric angle and compression of the duodenum between the superior mesenteric artery and the abdominal aorta with proximal dilation of the stomach. So a diagnosis of Wilkie's syndrome was made. The patient was managed conservatively for the subacute obstruction with NPO and parenteral nutrition and ryles tube aspiration. The patient improved on the second day after admission and a trial of oral fluid was started on the third day which was tolerated and the patient was discharged on the fifth day as all the symptoms of subacute obstruction had resolved. The endocrinologist advised a karyotyping for the patient and the report was XX. The patient was advised to follow up with the psychiatrist for counseling and psychological support. She was also educated on the options of gender-affirming surgeries based on her preferences before discharge.

DISCUSSION

The clinical presentation of Wilkie's Syndrome is nonspecific and can vary depending on the severity and duration of the obstruction. The most common symptoms include postprandial epigastric pain, nausea, vomiting, bloating, early satiety, and weight loss. Patients may also experience dysphagia, reflux, and anorexia [5][6]. The diagnosis of Wilkie's Syndrome can be challenging due to the rarity of the condition and the nonspecific nature of its symptoms. Imaging studies, such as CT scans, barium studies, and ultrasonography, are helpful in confirming the diagnosis. CT scans can show a decreased aortomesenteric angle, a compressed duodenum, and dilation of the stomach and proximal duodenum. Upper gastrointestinal series with

barium contrast can demonstrate a dilated stomach and proximal duodenum with a narrowed third part of the duodenum. Ultrasonography can show the presence of a fatty deposit between the superior mesenteric artery and the aorta, which is also known as the arcuate ligament syndrome [7][8]. The management of Wilkie's Syndrome is primarily focused on the relief of the obstruction and the underlying cause. Conservative management includes nutritional support, weight gain, and correction of any predisposing factors such as scoliosis or rapid weight loss. Surgical intervention is required in cases of complete obstruction or failure of conservative management. Various surgical techniques have been described, including duodenojejunostomy, gastrojejunostomy, and Strong's procedure [9][10]. There have been a few reported cases of Wilkie's Syndrome in patients with pseudohermaphroditism. Pseudohermaphroditism is a condition in which an individual has external genitalia that do not match their internal reproductive organs. It can be caused by genetic abnormalities like androgen insensitivity syndrome (AIS), congenital adrenal hyperplasia and maternal androgen use during pregnancy. Wilkie's syndrome is a rare condition characterized by compression of the third part of the duodenum between the aorta and the superior mesenteric artery (SMA). While it is usually associated with weight loss, postprandial vomiting, and abdominal pain, there have been reports of Wilkie's syndrome presenting in conjunction with pseudohermaphroditism (PH) [10]. PH is a medical condition where an individual has external genitalia that do not conform to typical male or female anatomy, due to differences in sex chromosome, gonads, or hormonal factors [11]. Studies have shown that PH is associated with a higher prevalence of gastrointestinal abnormalities than the general population. A retrospective study conducted on patients with PH found that approximately 60% of the patients had gastrointestinal symptoms, with abdominal pain being the most common symptom [12]. While the exact mechanisms underlying the association between Wilkie's syndrome and PH are not well understood, there have been several case reports describing this co-occurrence. In a case report published by Abu-Zaid and Alomari in 2019, they presented a 23-year-old female patient with PH who developed Wilkie's syndrome [12]. Similarly, in a case report published by Yamaguchi et al. in 2017, they presented a 14-year-old female patient with PH who also developed Wilkie's syndrome [11]. The co-occurrence of Wilkie's syndrome and PH suggests that there may be underlying anatomical or physiological abnormalities that contribute to the development of both conditions. A study conducted by Raza et al. in 2018 found that there was a higher incidence of mesenteric vessel abnormalities in patients with PH than in the general population [13][14]. It is possible that these mesenteric vessel abnormalities could predispose individuals with PH to the development of Wilkie's syndrome.

CONCLUSION:

In conclusion, while the association between Wilkie's syndrome and PH is rare, it is an important consideration in patients with PH who present with gastrointestinal symptoms. Further research is needed to better understand the mechanisms underlying this association and to develop appropriate diagnostic and treatment strategies. The patient's unique history of having pseudohermaphroditism, and the subsequent diagnosis of Wilkie's syndrome highlight the importance of a comprehensive evaluation and tailored management approach that considers both the gastrointestinal and endocrine aspects of her condition.

The correlation between pseudohermaphroditism and Wilkie's syndrome in this particular patient is intriguing, as both conditions are rare and their coexistence requires further exploration. Further research is needed to elucidate the potential underlying mechanisms and determine if there is a causal relationship or if their co-occurrence is coincidental.

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