Gastroschisis and VACTERL Association In a 28-Year-Old Male

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ABSTRACT

In gastroschisis the intestine protrude through a paramedian defect in anterior abdominal wall. VACTERL is an acronym for vertebral, anorectal, cardiovascular anomalies, tracheoesophageal fistula, renal and limb anomalies. Although literature has discussed association of gastroschisis with cardiovascular anomalies, its association with VACTERL has not been described. This report describes a case of a 28-year-old male diagnosed as having gastroschisis together with VACTERL association.

Key words *Gastroschisis, VACTERL association, Adult.*

INTRODUCTION:

Gastroschisis is a condition in which the intestines protrude through a defect in the anterior abdominal wall. Its incidence range from 0.46 to 4.18 per 10,000 live births.[1,2] The defect is usually in paraumbilical region with herniation of small bowel loops. It has a high mortality rate in low income countries.[3] VATER/VACTERL association was first described in the 1970s. Its reported incidence is approximately 1 in 10,000 to 40,000 live births. For the diagnosis of VACTERL, at least three of the features should be present.[4]

Associated congenital anomalies are common with omphalocele and rare with gastroschisis.[1] Gastroschisis is related to bowel abnormalities such as malrotation or intestinal atresia. Sometimes, the fetus may suffer from intrauterine growth restriction. However, association of gastroschisis and VACTERL association is rare. We report a case of gastroschisis and VACTERL association and are of view that it will be an interesting addition to the literature.

CASE REPORT:

A 28-years-old male resident of rural area was referred from urology outpatient with active complaint of suprapubic pain for 6 months and burning micturition for 2 weeks. He had a history of gut coming out from an anterior abdominal wall defect and imperforate anus since birth. He had normal vaginal delivery at home conducted by traditional birth attendant. Later on he was checked by a local physician of his village who advised surgery, but they did not seek medical attention due to lack of awareness, resources and fear of death.On examination, the gut herniated through upper abdominal wall defect and abdomen was tender at suprapubic region (Fig I). He underwent CT scan abdomen that demonstrated a large anterior abdominal wall defect with herniation of small bowel loops, mesentry and mesentric vessels without covering suggesting gastroschisis (Fig IIA). Rectum showed abrupt cut off at upper sacral level representing high anal atresia. There was absence of coccyx, partial agenesis of lower sacrum and bony ankylosis of lower

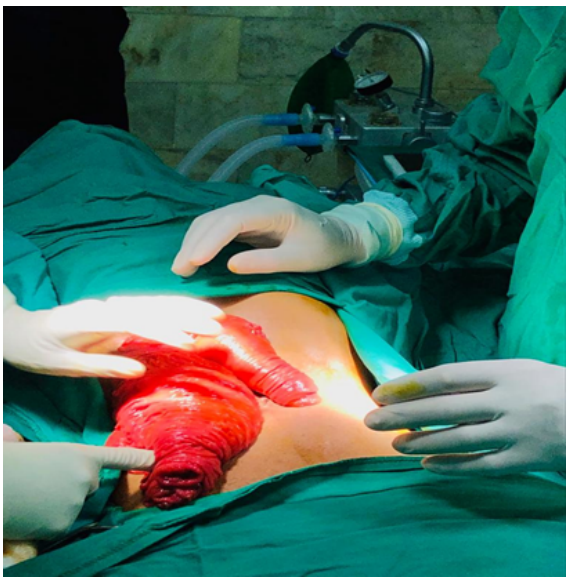

Fig I: Prolapse bowel loops from anterior abdominal wall defect.

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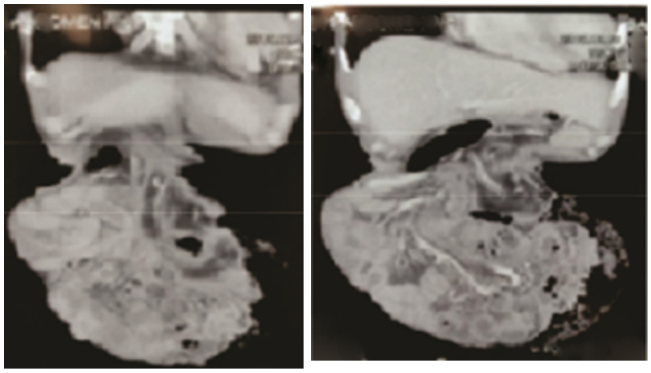

Fig-I A CT scan showing the herniation of the gut.

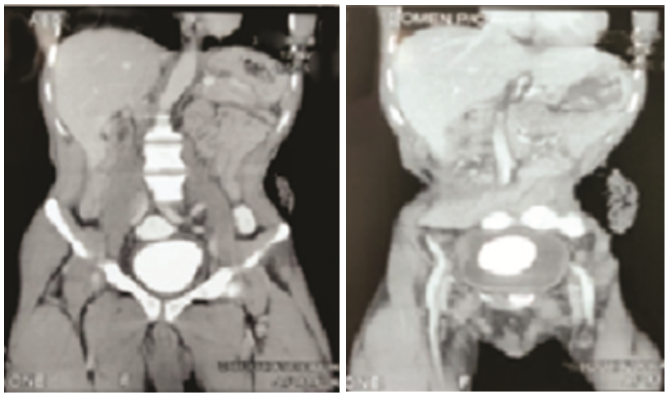

Fig-I B: CT scan showing the urinary calculus

lumbar and upper sacral vertebrae with spina bifida at lower lumbar levels. Left kidney was not visualised, likely congenitally absent. All these findings were consistent with VACTERL association. A urinary bladder calculus was also seen (Fig II B).

MRI pelvis confirmed the findings of high anal atresia and also demonstrated absent prostate and seminal vesicles. Rudimentary anal sphincter and anal canal were seen with distorted normal anatomy. His cardiac workup demonstrated mitral valve regurgitation.

The surgery and urology teams made the decision for exploration under anesthesia. Preoperatively, transverse colon found protruding out from upper abdominal wall defect. There was presence of intussusception with cecum, ascending colon, and ileal loops in transverse colon as intussusceptum. Descending colon had stenosed lumen and therefore, distal transverse colon and descending colon were resected. The calculus was removed followed by bladder repair. (Fig III)

Left kidney was absent. Liver, gall bladder, spleen, stomach, jujenum, duodenum and ileum were normal. Double barrel colostomy was made at left iliac fossa with transverse colon and sigmoid colon (Fig IV). The cavity was washed, pelvic drain was placed and skin closed. Patient recovered well and was discharged.

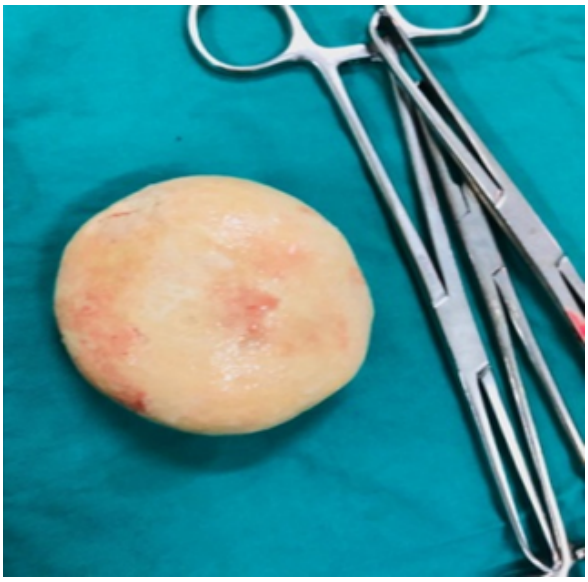

Fig-III Calculus removed from the urinary bladder.

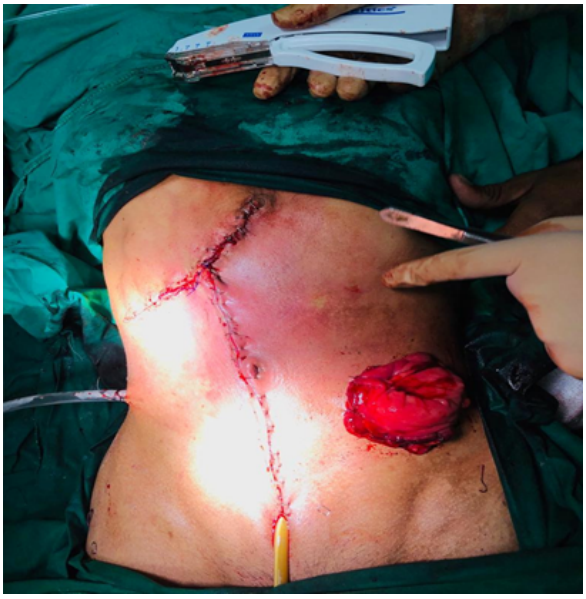

Fig IV: Final appearance following surgery.

DISCUSSION:

Abdominal wall defects (AWDs) are a part of a wide range of congenital abnormalities. The prevalence of abdominal wall defect is 6 per 10,000 live births.[5] Gastroschisis is a common type of AWD. Its etiology is thought to be abnormality associated with lateral body wall folding having deficient mesenchyme.[6]

VACTERL association is usually sporadic in nature.[7,8] Vertebral abnormalities are present in almost 60% to 80% of the cases with another visceral anomalies.[4]

Our case was unique for several features. First, the age of presentation was quite late. Gastroschisis is usually diagnosed antenatally and operated at birth. Second, the presenting complaint was urological rather than related to gut related symptoms. Although cardiac malformations have been reported with gastroschisis,[9] its association with VACTREL is quite rare.

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Received for publication: 22-07-2019

Accepted after revision: 20-12-2019

Author's Contributions:
Halar Habibullah: Manuscript writing, literature search and bibliography.
Sughra Parveen: Initial concept development and discussion with literature search.
Imran Khan: Literature search and bibliography.
Tanveer Ahmed: Literature search and bibliography.

Conflict of Interest:
The authors declare that they have no conflict of interest.

Source of Funding: None

How to cite this article:
Habibullah H, Perveen S, Khan I, Ahmed T. Gastroschisis and VACTERL association in a 28-year-old male. J Surg Pakistan. 2019;25(1):44-46. Doi:10.21699/jsp.25.1.11.