Colovesical fistula in a 11-year-old girl with ambiguous genitalia and recurrent urinary tract infection; a brief report

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**ABSTRACT**

VACTERL (V-Vertebral, A-Anorectal, C-Cardiac, TE-Tracheoesophageal, R-Renal, L-Limb) is an acronym for multiple organ dysfunction, which often occurs together. These prospects may affect patients with other abnormalities, such as urogenital abnormalities. Here we present a case of VACTERL syndrome with ambiguous genitalia who was referred and hospitalized due to recurrent urinary tract infection (UTI) and urosepsis. A colovesical fistula was discovered in the following examinations, which could explain this complication.

**Keywords:** VACTERL association, fistula, recurrent urinary tract infection, ambiguous genitalia

**Implication for health policy/practice/research/medical education:**
It is essential to be aware of the co-existing comorbidities associated with VACTERL (V-Vertebral, A-Anorectal, C-Cardiac, TE-Tracheoesophageal, R-Renal, L-Limb) syndrome, as these may have a crucial impact on prognosis and outcome.


**Introduction**

Ambiguous genitalia is a birth anomaly that embodies an unrecognizable gender due to the atypical appearance of the external genitalia. A disruption in the sex chromosome caused by environmental and genetic components results in ambiguous genitalia (1). Sex chromosome differences of sex development as; ovotesticular, common in karyotype 46xx in VACTERL (V-Vertebral, A-Anorectal, C-Cardiac, TE-Tracheoesophageal, R-Renal, L-Limb) patients (2).

VACTERL is a rare congenital multi-organ malfunction with an incidence rate 1/10,000-1/40,000 in the newborn (3). The diagnosis of VACTERL is in the presence of three or more of these involvements (4). abnormalities of the renal and urinary were commonly reported in 60-90% of VACTERL patients, and several studies showed that they were diagnosed in patients with VACTERL syndrome (5). A study conducted by Solomon. et al. showed that recto-vestibular or recto-urethral fistula, often associated with anorectal malformations, could be related to VACTERL syndrome (6). In addition, statistical data provided evidence of urogenital abnormalities, which are common in VAVTERL patients with renal abnormalities and anorectal atresia (7,8).

Due to the complicity of the seven core characteristics of VACTERL syndrome, as well as the other anomalies that accompany them (e.g., genitourinary anomalies), identifying all abnormalities and initiating clinical treatment could optimize medical care. Here we presented a brief report of a known VACTERL syndrome involving genitourinary system anomalies with recurrent urinary tract infection (UTI), who was finally diagnosed with colovesical fistula.

**Case Report**

A 11-year-old girl was referred to our center because of an increase in serum creatinine, colostomy dysfunction and abdominal pain. She was the last child of unrelated parents who came from Afghanistan. Her older siblings were...
healthy and showed no abnormalities. She had a history of an imperforate anus, a single kidney, and recurrent UTIs. In addition, she was diagnosed with VACTERL due to multiple organ abnormalities and her karyotype analysis at another center revealed a female chromosome (XX) genotype (Figure 1).

In addition, the colostomy bag had been used for an extended period of time due to an unsuccessful operation to open the imperforate anus. It was noticeable that her UTI recurred and persisted on various antibiotics. During her examinations, we found sacral agenesis and vertebral abnormalities related to her history and fever. At our center we performed a VCUG (voiding cystourethrogram) which revealed a high grade (grade V) vesicoureteral reflux (VUR) in the left kidney. In addition, due to the fever and history of recurrent UTIs, and colostomy malfunctions, a barium enema was performed, which revealed a colovesical fistula (Figure 2).

Due to her poor kidney function and electrolyte imbalance, as well as intermittent volume overload, she was eventually diagnosed with end-stage renal disease (ESRD). Three-weekly hemodialysis was considered. She is also a candidate for surgical procedures for colovesical fixation, genital reconstruction, and colostomy repair.

**Discussion**

Here we presented a rare case of colovesical fistula in a known diagnosed VACTERL syndrome with barium enema. Colovesical fistula is a rare malformation in children that is an abnormal connection between the bladder and the large intestine (9). Colovesical fistulas can be fatal from urosepsis (10), with an incidence rate of approximately 1 in 3000 patients hospitalized for surgery. The rate of all anorectal malformations is approximately 1 in 5000 live births and this colovesical fistula is an anorectal malformation (11). Therefore, early diagnosis and treatment of colovesical fistulas are essential.

Another essential manifestation in our patients was ambiguous genitalia. In a study of 105 patients with VACTERL syndrome, 56% of women had urogenital abnormalities, 33% had urogenital fistula, and 20% had structural abnormalities of the genitals such as micropenis (6). In addition, one of the most complex congenital anomalies of the pelvic organ in women, with an incidence rate of 1 in 50 000 births, is cloacal malformation. Up to 10% of these patients present with syndromic abnormalities associated with chromosomal or genetic abnormalities (12).

In addition, due to the close development of the urogenital and renal systems from the hindgut, these malformations mostly occur together. Several studies have demonstrated that renal and urogenital anomalies are common (69-93%), particularly when anorectal malformations, urogenital defects or lower vertebral defects are present (13). These results are consistent with our case report. In our case study, there was a predisposition to recurrent UTIs. A study of 220 VACTERL patients showed that the most common renal manifestation in patients was VUR causing UTIs and renal scarring (14). However, the cause of recurrent UTIs in our patients was a colovesical fistula. We need to consider other comorbidities like VUR. A long-term follow-up of 12 VACTERL patients showed that the development of ESRD was more common in VACTERL patients compared to the placebo-matched group, and had more deficient dialysis and transplant outcomes (15).

**Conclusion**

In summary, abnormalities that could cause complications such as UTIs may go unnoticed. The study of relative anomalies should be conducted with extreme care. The diagnosis would be beneficial in deciding on the best treatment, particularly in symptomatic patients (such as fever and abdominal pain as characterized in our VACTERL case) and/or those suffering from organ failure.

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**Authors’ contribution**

**Conceptualization:** Paniz Pourpashang, Seyed Mohammad Taghi Hosseini Tabatabaei.
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Conflicts of interest
The authors declare that they have no competing interests.

Ethical issues
This case report was conducted in accord with the World Medical Association Declaration of Helsinki. Informed consent was obtained from the parents of the patient prior to their child's inclusion in the study. Ethical issues (including plagiarism, data fabrication, double publication) have been completely observed by the authors.

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