CASE REPORT OPEN ACCESS

Complex Congenital Anomalies of Colorectal and Genito-urinary Systems: A Case Report

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ABSTRACT

The caudal duplication syndrome is a rare but well-recognized congenital anomaly. We describe a female who presented in neonatal period with imperforate anus with fistula in the vestibule. On further investigations anomalies related to upper urinary tract as well as genital systems were identified. Later stool was noted coming out of an additional opening behind the vagina. This turned out to be the duplication of distal part of the colorectal gut. Patient also suffered from the recurrent urinary tract infections and failure to thrive was noted. CT urogram showed duplication of the upper urinary system with massive hydronephrosis and hydroureter on right side and significant dilatation of the urinary system on left side. Patient underwent left sigmoid colostomy and bilateral ureterostomies. She is currently in follow up.

Key words *Caudal duplication syndrome, Duplication of urinary tract, Septate vagina, Uterus didelphys, Colonic duplication, Ureterostomy, Colostomy, Hydronephrosis, Renal dysplasia.*

INTRODUCTION:

Complex congenital anomalies of anorectal and genito-urinary systems pose number of challenges for the treating physicians including their recognition, diagnosis and treatment.[1] Many of these anomalies are unique in their presentation. The anatomical features as well the clinical course may vary. It is therefore important to report such patients so that others learn from these experiences. In this report, we describe a patient with complex anomalies of genito-urinary systems and anorectal canal. The clinical course of the patient was marred by infrequent follow up visits that added to the morbidity. The anomalies had few similarities with caudal duplication syndrome.[2]

CASE REPORT:

A 12-month-old female at present, resident of a rural district from low socio-economical background, had her first visit to our hospital when she was 20-days of age with 2.4-kg weight. Her mother noticed passage of stool through an opening in the vulva in an adequate amount. On examination a fistulous opening was found in the vestibule at posterior fourchette and imperforate anus. A separate urethral and vaginal openings were noted with multiple pits in the introitus in same anatomical location. The pits could not be probed and appeared blind. On ultrasound abdomen mild bilateral hydronephrosis was reported. Other VACTERL screening was normal. The plan was made for primary anorectoplasty by the age of two months. Mother was taught dilatation of fistulous opening with Hegar's dilator. However, infant was brought at the age of 10-weeks with high grade fever and passage of turbid urine. She was evaluated and managed in nephrology unit for documented urinary tract infection (UTI) as on urine culture Burkholderia grew. The passage of stool was adequate and perineal hygiene was acceptable. Ultrasound was repeated which showed moderate bilateral hydronephrosis and hydorureter. Urinary bladder appeared unremarkable. It was planned to go for voiding cystourethrogram (VCUG) after eight weeks following UTI treatment. At 16-weeks of age VCUG was done. It showed normal looking bladder with no reflux on both side and bladder emptying adequately in post-voiding films. A non-refluxing, non-obstructing vesico-ureteral junction was suspected. It was decided to request for radio-isotope DMSA scan for assessment of renal status. Renal biochemistry was reported as within normal limits.

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The girl presented again at 24-weeks of age, weighing 3.5-kg, with recurrent fever and not gaining weight. Mother also noticed baby straining during defecation. She also found stool staining from another opening just posterior to the vagina and in front of vestibular fistula (Fig. I). This might have resulted secondary to the inflammation. On follow-up ultrasound at this visit a right sided duplex renal system with massive hydronephrosis and left sided mild hydronephrosis with bilateral hydroureter were reported. DMSA scan showed well outlined left kidney with good tracer uptake, no scarring and left hydroureter with relative function of 64%. The right kidney was faintly outlined due to the reduced tracer uptake, no scarring was noted. The relative function of the kidney was 36%. A CT urogram was also performed that revealed right sided massive hydronephrosis with two separate renal moieties suggestive of a duplex system with ureters joining at distal level proximal to the urinary bladder. The cortex of left appeared thinned out with hydronephrosis. The common right ureteric appeared opening at the urethral level and left ureter at the level of bladder neck (Fig. II).

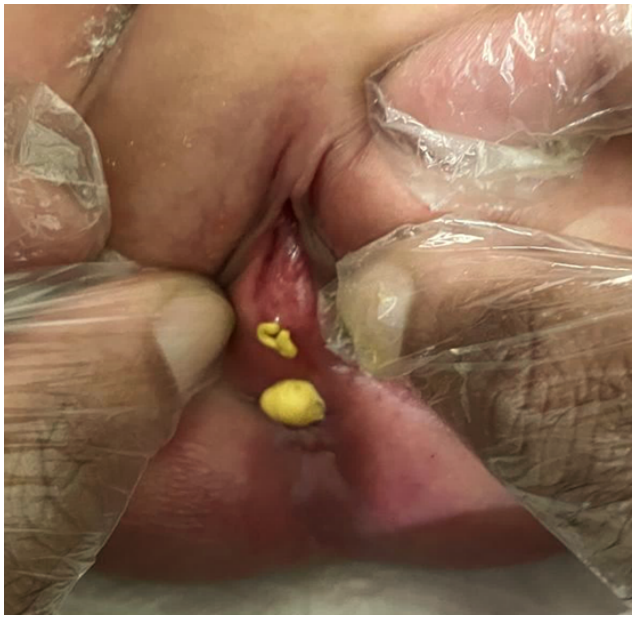


Fig. I: Two fistulous openings from which stool is noted.

During this visit cystoscopy as well as colostomy were planned to divert the stool. Examination under anesthesia revealed a septate vagina that was missed previously with multiple blind pits in the vestibule. Two fistulous openings with stool staining were also identified. On table contrast study was done through both the openings that showed duplicated colon with no communication between them proximally extending into the abdomen. Genitoscopy showed two separate hemi-vaginal openings leading to two cervices. At cystoscopy urethra was found normal looking and urinary bladder was full of debris. It was difficult to identify ureteral openings.

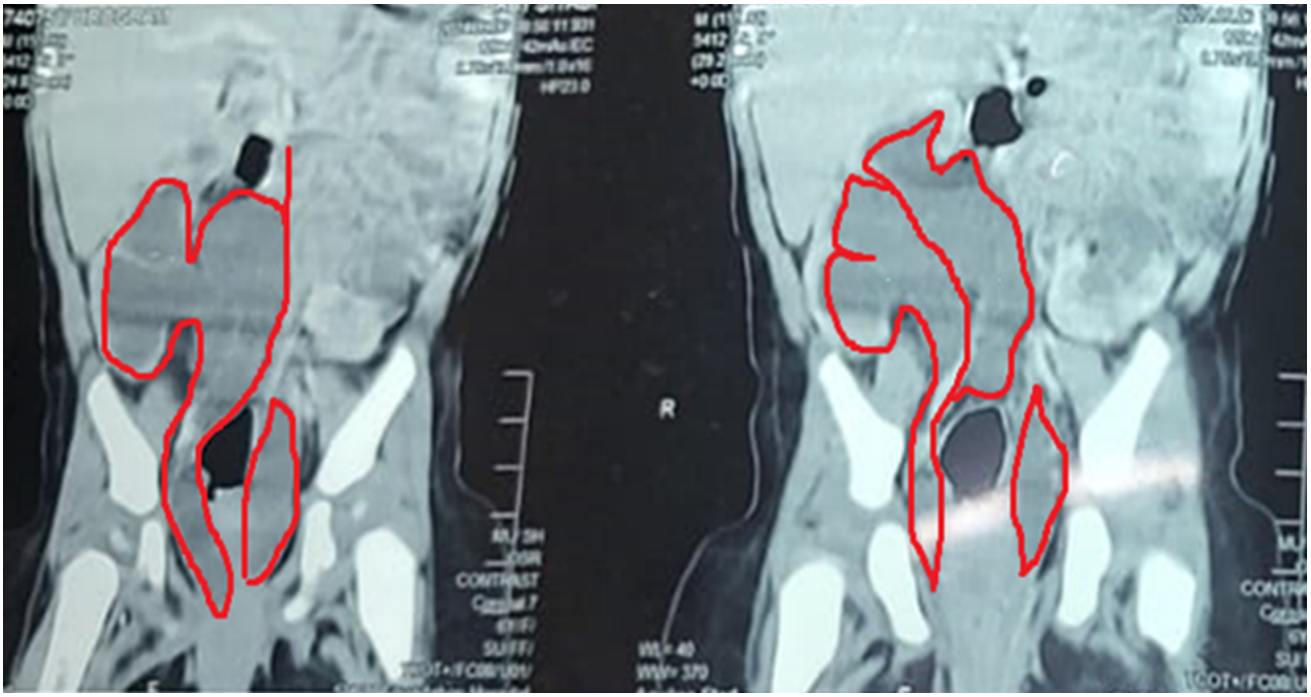


Fig. II: CT Urogram showing enlarged and hydronephrotic kidneys with partial duplex system on right side. Lower end of the ureters can be appreciated at the level bladder neck and urethra. Both ureters are dilated and tortuous; left placed more medially.

Sigmoid colostomy was done in the same sitting through left sided lower transverse abdominal incision. The proximal extent of the colonic duplication could not be delineated as incision was too low. A high divided sigmoid colostomy was made with four openings at stoma site (Fig. III). Postoperative recovery was uneventful and stomas stared functioning. A contrast study for delineating the duplicated colon was done later which showed the proximal extent up to the transverse colon (Fig. IV). Patient was discharged after taking consultation from pediatric urology team with expertise in renal transplantation. Considering recurrent UTI, urinary diversion was suggested. However, possibility of renal transplantation in future was not ruled out.

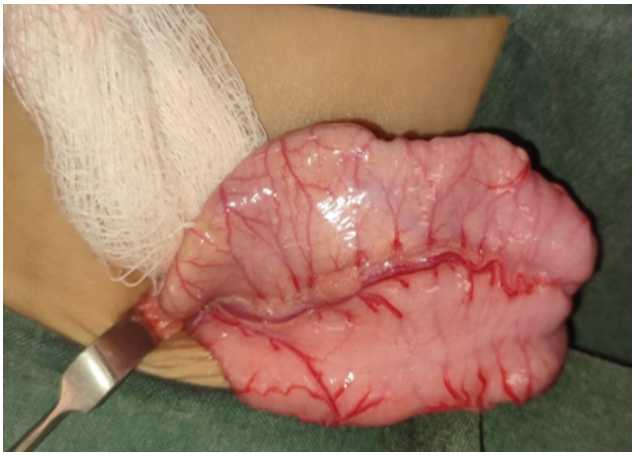


Fig. III: Duplication of the sigmoid colon.

At one year of age when patient finally presented to us with 4.5-kg weight, ultrasound showed right sided massive and left mild to moderate hydronephrosis. Repeat DMSA scan showed no functional tissue on right side and faintly outlined left kidney with reduced tracer uptake. MRI of the spine was also done which was reported as normal. It was decided to go for urinary diversion to save the upper tract. Under general anesthesia cystoscopy was done that showed wide single ureteric opening at bladder neck. Other ureteric opening could not be identified. Contrast dye was instilled through the ureteral opening that was noted. It delineated left sided hydroureter and hydronephrosis. After cystoscopy bilateral loop ureterostomies were performed; right sided in distal ureter in the common channel and on left side in proximal ureter due to the presence of colostomy in lower abdomen. Postoperatively both ureterostomies started unctioning. More urine output was noted from left side as compared to the right. No urine was reported per urethra. Renal biochemistry showed values in normal range.

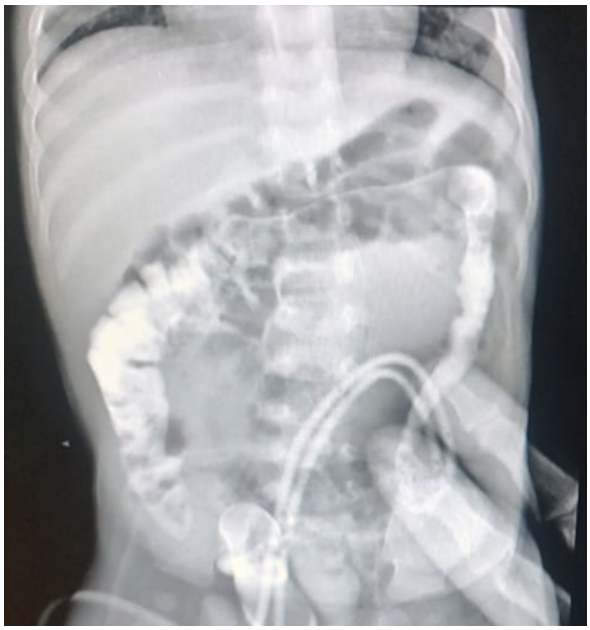


Fig. IV: Contrast fistulogram showing proximal extent of colonic duplication up to mid-transverse colon.

Nutritional advise was given and monetary support also provided to the family to ensure regular follow up and adherence to the treatment. It was planned to further investigate the upper genital system and divide the vaginal septum during the subsequent follow up. Removal of the duplicated part of the colon as well as decision about renal system shall be planned according to the status of the kidneys in follow up. The summary of the events is given in table I. The anomalies are shown in a sketch form in Fig. V.

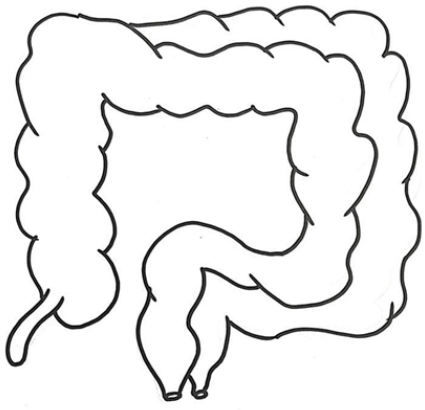
DISCUSSION:

This report adds a new set of anomalies to the literature. In particular, the complex nature of associated urinary tract malformation. This case is different from classical caudal duplication syndrome which is infrequently reported in literature. The classical caudal duplication syndrome is a rare anomaly. The reported prevalence is less than 1 per 100,000 births. The anomalies in the syndrome consists of the duplication of the variable extent of gastrointestinal system, mostly of hindgut, genitourinary system, as well as spinal cord, vertebral column. Lower limbs are also affected.[3]

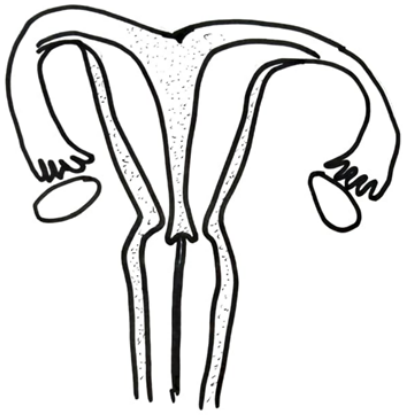
Duplication of colorectal canal is a known anomaly and can occur as an isolated malformation. Piplani et al have reported their experience of management of these patients and also reviewed the literature.[4] In our patient there was a vestibular fistula noted at birth but later passage of stool was noted from

Fig. V: Graphic representation of the anomalies,

A: Urinary tract anatomy



B: Colorectal anatomy.



C: Genital anomalies.

Table I: Summary of the Case

Age	Clinical Examination & Anomalies Identified	Investigations / Results	Surgical Procedure / Intervention
Three Weeks	Weight 2.4 kg Imperforate anus with vestibular fistula, adequate passage of stool	Ultrasound KUB: Mild bilateral hydronephrosis	Hegar's dilatation
Ten Weeks	High grade fever and passage of turbid urine	Urine culture: Positive (UTI) Ultrasound KUB: Moderate bilateral hydronephrosis	Size of dilator increased Nutritional advise
Sixteen Weeks	Failure to thrive	VCUG: Showed normal looking bladder with no reflux on both side and bladder emptying adequately in post-voiding films. A non-refluxing, non-obstructing VUJ was suspected. RFTs: Within normal range	Nutritional advise
Twenty-four Weeks	Weight: 3.5 kg Straining during the defecation. Stool staining from another opening just posterior to the vagina and in front of vestibular fistula.	Ultrasound KUB: Right sided duplex renal system with massive hydronephrosis. Left sided mild hydronephrosis. Bilateral hydroureter. DMSA Scan: Well outlined left kidney, relative function of 64%. Faintly outlined right kidney, relative function of 36%. CT Urogram: Bilateral hydonephrosis and hydroureter, with ectopic position of ureteral openings	EUA: Septate vagina with multiple pits that were not negotiable. Two fistulous opening with staining of stool. Fistulogram: Duplication of colon. Genitoscopy: two hemi-vaginal openings leading to two cervices. Cystoscopy; Normal urethra and urinary bladder. Ureters could not be identified. Sigmoid colostomy
Post-procedure		Fistulogram: Proximal extent of the duplication up to transverse colon.	
Fifty-two weeks	Weight: 4.5 kg		Bilateral Ureterostomies

a duplicated fistulous opening located between vestibular fistula and vagina. This is an unusual variant. Pampal et al have reported three patients with number of anomalies falling broadly under the spectrum of caudal duplication. Our patient also had similarity with these cases.[5]

In our patient the gradual deterioration of anatomical features of the upper urinary system is more likely due to inherent renal dysplasia rather than vesico-ureteral obstruction on right side. There was associated partial upper tract duplication on same side. The right ureter could not be identified during the cystoscopy. However, on CT urogram it appeared to be the level of urethra. As patient remained continent of urine it is assumed that pelvic diaphragm and bladder-urethral synergy is intact. The status of left sided urinary system is better. The renal functions tests are also reported as normal that indicate preserved renal function to some extent. The current status of the patient is therefore satisfactory. Ureterostomies are expected to prevent recurrent UTI which may be a cause of failure to thrive. Nephrectomy on right side in case of nonfunctioning kidney and left ureteral re-implantation is also under consideration. However, counseling for renal transplant in future is also done.

We missed septate vagina in this baby at initial presentation. The reason could be a small vaginal opening and prominent hymen lips. Further assessment of the genital system is differed as it is not an issue at present. Colostomy in our patient is expected to improve the passage of stool but stoma care may be a challenge for the family. Application of stoma bag can facilitate this. Removal of distal part of the duplicated colon will require meticulous planning. We planned to remove the smaller fistulous tract and attempt transposition of main vestibular opening to its normal position by the perineal approach taking care of the blood supply of anorectal canal as well as sphincter muscles. This will be combined with the abdominal approach to deal with the proximal part of the colonic duplication. The anastomosis between the two limbs of the duplicated part can be facilitated with the use of stapler. These anomalies are rare and have to be addressed on their merit. The plan can be modified depending upon the nutritional status of the patient as well renal function.

CONCLUSION:

Few of the anomalies were missed at initial presentation like septate vagina and duplicated fistulous rectal opening. Timely intervention was also not possible because of irregular follow-up, recurrent UTI and patient's nutritional status. At ultrasound duplication of renal system was diagnosed late. On cystoscopy ureteral openings could not be identified. However, with a continued management strategy involving nephrology team it was possible to provide best possible care to the girl.

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Authors' contribution:
Kainat Ejaz: Concept, data collection, literature search, drafting and revision of the manuscript.
Rabbia: Manuscript writing and revising.
Misbah: Manuscript writing and revising.

All authors are responsible for the content of the article.

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