



## Caudal ‘duplication’ or ‘split’ syndrome: Is there a misnomer?



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### ABSTRACT

‘Caudal duplication syndrome’ was coined to describe the apparent duplication of organs derived from the hindgut, the neural tube and the adjacent mesoderm. Review of the anatomy suggests that the word ‘duplication’ may be a misnomer. This paper describes the management of 2 girls with caudal duplication syndrome who underwent multistage reconstructive surgery. Both had a large omphalocele and a severe diastasis of the pubic symphysis. The first patient also had an apparent duplication of the vulva, the perineum and the anus to either side of a wide midline. Each vulva contained a urethra, a hemi-clitoris with ipsilateral labium minor, and a hemi-vagina with hemi-uterus. The second child had an infrapubic sequestered appendico-cecal duplication lying between two hemi-bladders each with ipsilateral ureter and urethra. The everted duplication split the single vulva longitudinally in the midline as far as the fourchette. To each side were a hemi-clitoris, and a hemi-vagina with hemi-uterus and ipsilateral fallopian tube. Analysis of our patients’ anatomy and a literature review indicates for the most part ‘hemi’ organs on either side and suggests that the term ‘duplication’ is a misnomer such that caudal ‘split’ syndrome may be a more appropriate title.

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Caudal duplication syndrome was coined to describe the apparent duplication of the organs derived from the hindgut, neural tube and adjacent mesoderm resulting from ‘insults’ at different stages of embryogenesis [1]. The condition is rare and only few reported cases describe the combination of anomalies of the colorectum, the lower urogenital tract, the abdominal wall and spinal dysraphism.

We report our experience with two girls, born 20 years apart and in different countries. We recommend that early management should be conservative and not compromise fecal or urinary continence. Complex multistage surgery should be patient tailored toward an anatomical, functional and esthetic reconstruction [2,3], and should be delayed until after functional and dynamic evaluation of all anatomical parts. A study of our patients’ anatomy and a literature review suggest that the term ‘duplication’ in the title is, for the most part, a misnomer and caudal ‘split’ syndrome may be more appropriate.

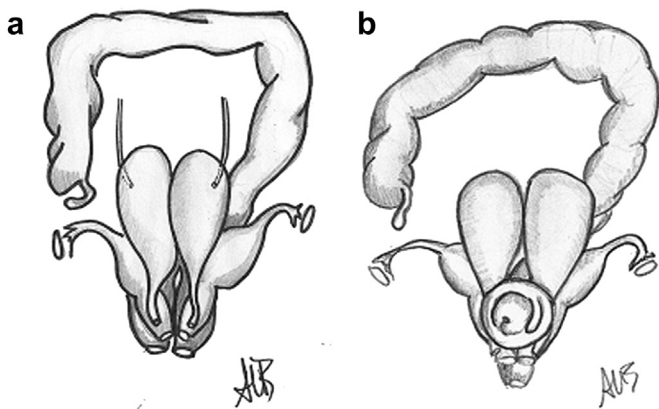
### 1. Case reports

#### 1.1. Patient 1

Born in 1980, the child was referred after a normal pregnancy and term delivery, because of a large omphalocele, severe pubic diastasis, and ano-urogenital malformations. To either side of a wide fatty midline there was a separate smaller vulva. Each contained a hemi-clitoris and ipsilateral labium minor, a complete vaginal orifice and a continent urethral orifice that passed urine. Posterior to each vulva was a perineum and an anus, with the left demonstrating an intact and continent sphincter and the right anus being open anteriorly. Both ani passed meconium. Routine pelvic radiology did not show any lumbosacral anomalies. Intravenous urography, examination under anesthesia, and cystoscopy through each urethra revealed two separate hemi-bladders lying side by side with each draining its single ipsilateral normal ureter and kidney. Bilateral vaginoscopy demonstrated a full-length hemi-vagina with cervix. At neonatal repair of the omphalocele, a laparotomy confirmed two separate hemi-bladders each with an ipsilateral single ureter, two hemi-uteri each with ipsilateral fallopian tube, and a normal ovary bilaterally (Fig. 1a). The hemi-bladders were opened longitudinally on their medial aspects and the right bladder neck was detached from its urethra and closed.

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**Fig. 1.** (a) Patient 1 had two separated hemi-bladders lying side by side, and separate Mullerian structures. The two hemi-recti pass to separate ipsilateral anal orifices. (b) Patient 2 had a sequestered cecum and appendix lying in the midline between two separate hemi-bladders and separate Mullerian structures.

The hemi-bladders were combined to form a single larger volume bladder that received both ureters and that voided through a competent left bladder neck and urethra. The right hemi-vagina with hemi-uterus and fallopian tube was detached from the right vulva and excised. The cecum, appendix and colon were single and normally sited, but the rectum was split longitudinally with each hemi-rectum passing to its ipsilateral anus. The right hemi-rectum was resected leaving the colon to evacuate through the left hemi-rectum that was controlled by a continent intact left anus and anal sphincter complex. The abnormal right anus was left in situ. No pelvic surgery was undertaken and the pubic diastasis was left open. Healing was relatively uneventful and the child was followed up as an outpatient in another institution where the residual right vulva, perineum and split anus were eventually resected. She developed normal fecal and urinary continence through the left structures. Serial radiographic and ultrasound monitoring outlined a stable large capacity bladder with no upper tract changes. Her renal function was normal and there were no urinary infections.

She returned to our institution at 25 years of age, for evaluation toward possible sexual relationships and esthetic adjustments. The pelvic diastasis was still present (Fig. 2) and had led to difficulties with appropriate clothing, but her gait was normal. She was fully continent of urine and feces through the left structures, and had

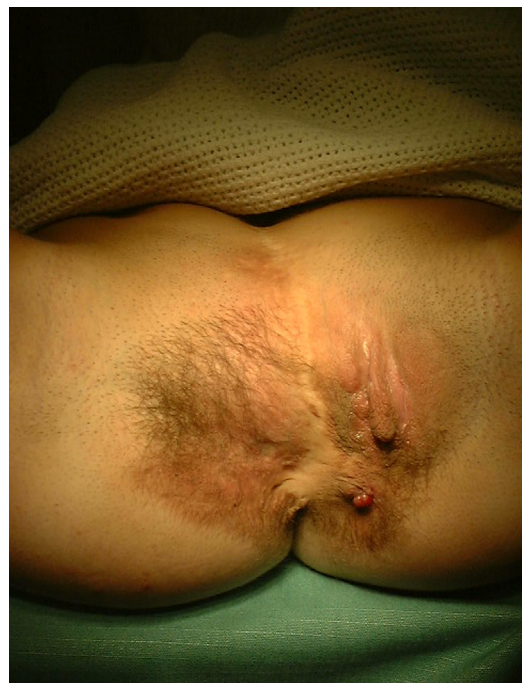


**Fig. 2.** Patient 1 – preoperative pelvic x-ray showing a severe pubic diastasis. Similar for both patients.

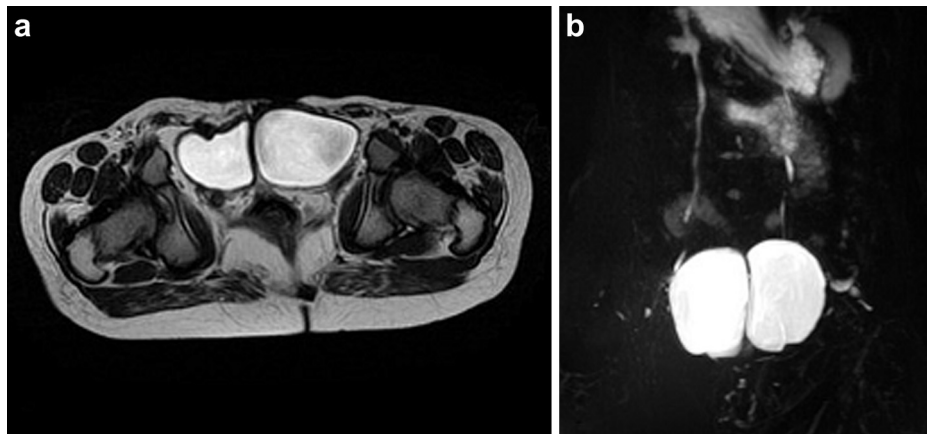
gone through normal puberty establishing regular menstrual cycles. There had not been any urinary infections and evaluation revealed a large bladder with normal upper tracts and normally functioning unscarred kidneys. Examination under anesthesia revealed a smaller vulva to the left side of a wide fatty midline. The left labium major and labium minor were normally formed but were separated from the contralateral matching right structures by a wide fatty midline. There was a left hemi-clitoris with prepuce, and a full length left vagina leading up to a left cervix. The large capacity bladder without any trabeculation was accessible through a longer angled urethra. A single ureteric orifice with hemi-trigone opened separately to either side of the previous bladder suture line. The esthetic appearance of the genitalia and perineum was poor (Fig. 3) and she was very self-conscious with strong psychological concerns relating to her lower body appearance. There had not been any sexual relationships. She was otherwise a well-adjusted, extrovert, intelligent and educated young lady.

### 1.2. Patient 2

The second patient presented as a healthy 10 year-old 46XX female with an unremarkable antenatal history and scars suggestive of surgery for a large omphalocele. The anus was normal and she was fully continent for feces, however she was continually wet because of urinary incontinence. Her serum electrolytes, renal function, and lumbosacral spine were normal. She had a severe pubic diastasis beneath which was a large midline mucosal structure that protruded through the longitudinally split vulva anterior to the intact fourchette, perineum and anus (Fig. 1b). Abdominal ultrasound scan and a barium enema revealed a cecum and appendix in the right iliac fossa in continuity with a normal single colon and rectum that did not communicate with the abnormal midline mucosal structure. Abdominal and pelvic magnetic resonance showed a single normal kidney and ureter bilaterally, each draining into a separate ipsilateral hemi-bladder (Fig. 4a and b). There was a well-formed hemi-uterus on each side. At examination



**Fig. 3.** Patient 1 – lower abdomen and perineum. Very poor esthetic result.



**Fig. 4.** (a) Sagittal magnetic resonance scan of Patient 2 showing two hemi-bladders. (b) Three-dimensional reconstruction of the two hemi-bladders lying side-by-side each with its ipsilateral ureter to each hemibladder.

under general anesthesia the infrapubic midline mucosal structure was determined to be a sequestered everted cecum with appendix, which split the single vulva longitudinally as far as the intact fourchette. To either side were an ipsilateral labium major and minor, a hemi-clitoris with partial prepuce and a urethral orifice that gave access to an ipsilateral hemi-bladder with an ipsilateral hemi-trigone and ureteric orifice. The right urethra and bladder neck appeared closed and better formed than the left that was lax and constantly dribbling urine. Posterior to each urethral orifice was a hemi-vagina with ipsilateral cervix. The sequestered everted midline cecum and appendix (Fig. 5) with its mesenteric blood supply was resected, and the hemi-vaginae were brought together and combined in the midline to form a single short but wider vagina that received both the right and left cervix. The lower abdominal wall and external genitalia were reconstructed by midline apposition of tissues within the limits allowed by the severe pubic diastasis.

At 14 years of age the patient was readmitted to our clinic. Following her operation 4 years previously, her mental attitude had improved but her quality of life was poor because of continuous urinary incontinence. Since her surgery she had not had any urinary infections, had remained fully continent for feces, and had regular monthly menstrual cycles. A cystourethrogram through each urethra again confirmed separate hemi-bladders lying side by side

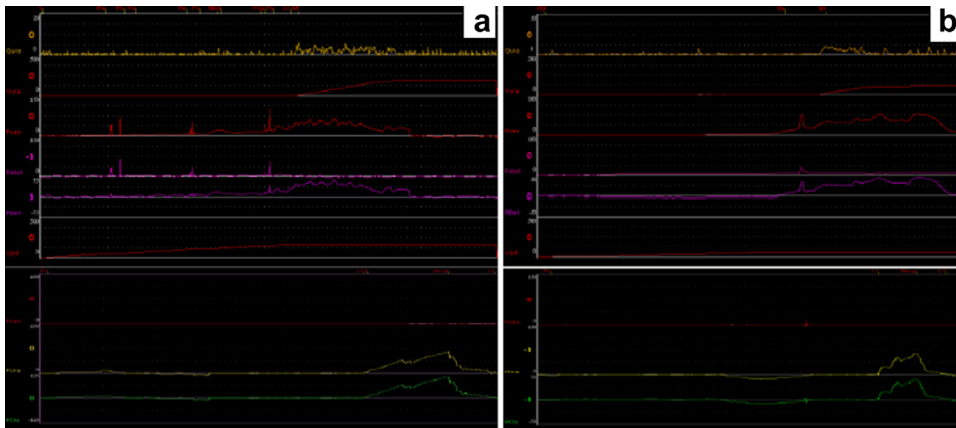
with no vesicoureteric reflux. An urodynamic study showed the right hemi-bladder to have a continent capacity of 160 cc when the patient experienced a sensation of a full bladder and a desire to pass urine (Fig. 6a). The left bladder was of smaller capacity at around 60 cc, provided no sensation and dribbled urine freely (Fig. 6b). At surgery the hemi-bladders were incised longitudinally along their medial borders, the weak left bladder neck was detached from its urethra and closed, and the hemi-bladders combined over drainage through a right urethral catheter and a suprapubic cystostomy tube. The vagina was maximally lengthened and widened by further division of the residual vaginal septum. At discharge from the hospital on the 20th post-operative day, she was dry by day and night, passing urine voluntarily every 2–3 h, and was confidently wearing normal clothing. Ultrasound scan and postoperative cystography (Fig. 7a and b) confirmed a continent bladder without vesicoureteric reflux and without post micturition residual urine. Her lower abdomen and external genitalia are acceptable within the limits of her wide pubic diastasis (Fig. 8a and b) and, together with her complete urinary continence, have led to a significant improvement in her self esteem, her quality of life and her social prospects. Six months postoperatively she remains completely continent for periods of up to 2 h by day and is dry through the night.

## 2. Discussion

Caudal duplication syndrome has an incidence at birth of 1:100,000 and there are no consistent risk factors or familial associations. The first case was reported in “Ephemerides” in 1712 from the Leopoldine Academy at Frankfurt [4], but it was only in 1993 that Dominguez et al. proposed the term ‘caudal duplication syndrome’ to describe the rare association between gastrointestinal, genitourinary, and distal neural tube malformations [1]. The pathogenesis of this anomaly that may present with a wide spectrum of anatomical variations remains unclear but it is thought to be the result of a sagittal pairing of axial structures because of notochord–hindgut interplay [5,6] during early caudal development [7]. This midline developmental division of the medial cloacal structures might also lead to a partial or complete lack of fusion of the Mullerian structures that form the internal genitalia [8,9]. In 1973, Kossow and Morales reviewed 40 cases of complete bladder duplication and found an association with duplication of the external genitalia in 90% of cases, and with lower intestinal tract duplication in 42% [10]. Our first case presented two of the rarest pediatric anomalies namely an apparent double vulva with only about 20 previously reported cases, and an apparent duplication of



**Fig. 5.** Patient 2 – everted sequestered cecum and appendix splitting the vulva longitudinally in the midline.



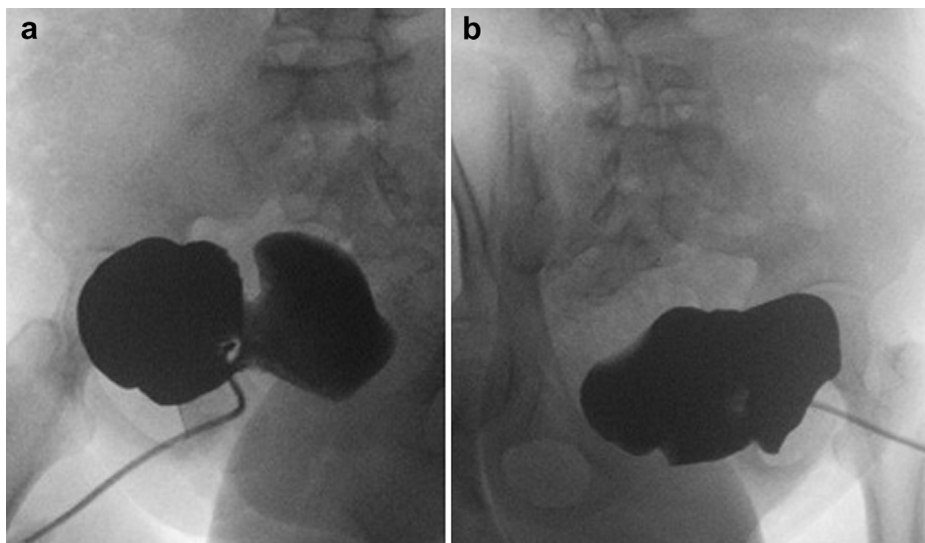
**Fig. 6.** Cystomanometry of each hemi-bladder for Patient 2: (a) The right hemi-bladder showed reduced compliance and presence of uninhibited detrusor contractions. (b) The left hemi-bladder showed a stronger uninhibited detrusor contraction.

the anal canal with no more than 35 pediatric reported cases since 1960 [11,12]. It is unfortunate that these rare conditions with unusual anatomy are often written up as case reports and find major difficulty in reaching publication such that valuable anatomical and functional detail is lost.

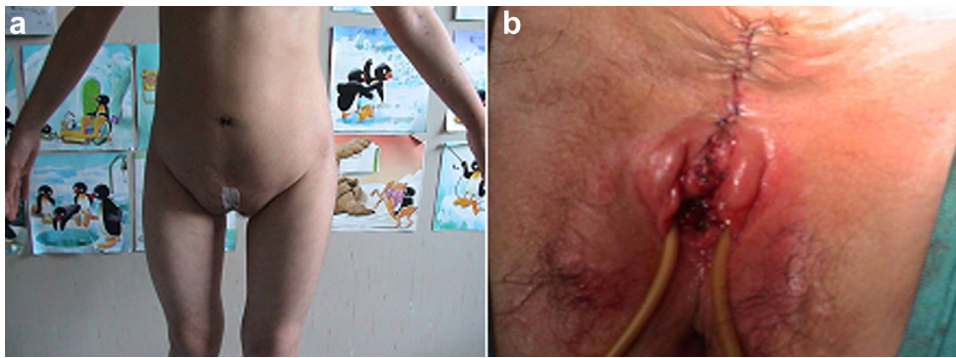
Following careful study of the actual anatomy of both our cases we are of the opinion that each structure represents the ipsilateral half of the full organ rather than a real duplication. Thus we observed that each hemi-bladder had its own hemi-trigone and drained its ipsilateral ureter and kidney. The Mullerian structures appear not to have fused and presented as bilateral hemi-vaginae each with ipsilateral hemi-uterus, cervix and fallopian tube. Each vulva had a complete labium major only on the ipsilateral side (Fig. 3). Within the vulva the clitoral glans (more likely a hemiglans) was covered over by a hemi-prepuce and with an ipsilateral complete labium minor, and was located on the end of a single corpus cavernosum that was attached to the ipsilateral pubic ramus. Analysis of the rectum in our first patient again suggested a 'split' rather than duplication, with each half passing to its ipsilateral anus (Fig. 1a). Although apparently complete only on the left side it would seem more likely that the ani also represented hemi structures. Our second child presented with a sequestered everted

cecum and appendix lying between two hemi-bladders and urethras and protruding through the longitudinally split vulva as far down as the fourchette. To each side lay a hemi-clitoris, a hemi-vagina and hemi-uterus. Contrast studies and subsequent abdominal surgery confirmed the presence in the right iliac fossa of another separate normal cecum with appendix that was in continuity with a full-length normal colon and rectum. Following resection of this sequestered midline cecal and appendicular apparent duplication with its mesenteric blood supply, it was possible to combine both hemi-bladders and to better reconstruct the external genitalia with apposition of the hemi-vaginae within the limits of the wide pubic diastasis.

Publications in the literature are unfortunately rather scant on anatomical detail. Bansal et al. reported a case of a 2-year old female with a wide pubic diastasis who had an apparent complete colonic duplication with a single cecum. The ascending colon consisted of two mucosal tubes that lay side by side within a common muscle wall that beyond the hepatic flexure separated into two complete hemi-colons on a common mesentery. Each hemi-colon ended separately, on the right side as a rectovestibular fistula and on the left at a normal continent anus at the perineum. There were two separate hemi-bladders separated by a thick muscular septum. Each



**Fig. 7.** (a, b) Postoperative cystography of the bladder for Patient 2.



**Fig. 8.** (a) Patient 2 following resection of sequestered cecum. (b) External genitalia after resection of the sequestered cecum.

bladder received its own ipsilateral ureter and urethra. The right hemi-bladder and urethra were well developed and continent, but the left bladder was thinner walled and had a stenotic urethra. The vulva had two vaginal orifices lying side by side that led into two vaginal canals of which the right was poorly developed. Each vagina received a hemi-uterus with its ipsilateral fallopian tube and ovary. The poorly developed right vagina and right hemiuterus were excised but the right ovary was preserved [13]. In another publication in 2007 Altug et al. described a 16-year old female with a pubic diastasis who at intravenous pyelography was shown to have two kidneys each with a single ureter opening into its ipsilateral separate hemi-bladder with continent urethra. Laparoscopy showed a hemi-uterus with ipsilateral fallopian tube on each side [14]. Unfortunately both sets of authors tend to use the terms 'duplication' and 'hemi-organ' rather loosely and interchangeably, which creates confusion when relating the findings to the well-accepted embryogenesis of the Mullerian ducts that describes each duct as developing separately into a hemi-vagina and hemi-uterus with ipsilateral fallopian tube, and that finally undergo fusion to form a single vagina and uterus. Similarly each hemi bladder was noted to receive its ipsilateral ureter and to drain the ipsilateral kidney. Thus we would like to suggest that valuable comparisons can only be made if careful attention is given to the use of an agreed terminology. The anatomical descriptions in both the Bansal and Altug reports are identical to our cases including the wide pubic diastasis, and the organ descriptions are suggestive of hemi organs rather than duplications. It is interesting to speculate that the two colons on a common mesentery noted by Bansal et al., may also represent two hemi-colons. It would therefore seem appropriate to suggest that the term 'duplication' within the title for this syndrome is a misnomer, since for the most part it does not reflect the actual anatomy. Indeed 'caudal split syndrome' may be more accurate.

The most plausible embryological theory was proposed by Bremer. In 6–7 week embryo, portions of the intestinal tract become obliterated by rapid proliferation of endothelial cells. As the gut grows in length, vacuoles appear in these cell masses, become longitudinally oriented and coalesce, reconstituting a single lumen. Should one or more of these vacuoles become pinched off, a second lumen is created which may become permanently separated from the main lumen by uniting the layers of opposing intestinal walls between the two channels [15]. Another theory was suggested by Campbell to explain the associated genitourinary anomalies. This theory stated that it is caused by a splitting of the vesicourethral anlage and that associated rectal anomalies will suggest whether the schism occurred before or after division of cloaca by urorectal septum [16]. A similar explanation was given by Satter and Mossman [17].

Children with caudal split syndrome require careful anatomical and functional evaluation with clinical management individually tailored toward optimal function, continence, and best esthetic

appearance. Abrahamson concludes that complete duplication is quite consistent with normal life and does not recommend treatment [18]. However we disagree with this approach since it disregards the psychological impact on the patient's self esteem and quality of life from such a severe lower abdominal and genital anomaly. Our patients had anomalies of the gastrointestinal and genitourinary systems with associated fecal (in the first case) and urinary (in the second case) incontinence. The first child was managed as a neonate with resection of the right hemi-rectum, and closure of the right bladder neck with a right-to-left bladder augmentation at the same session as the surgical closure of the omphalocele. The right hemi-vagina, hemi-uterus and fallopian tube were also resected. She underwent subsequent resection of the residual split right anus, perineum and vulva. We now consider that our approach was unnecessarily aggressive and incorrect in that it was not possible in the neonatal period to undertake urodynamic studies or to definitively determine voluntary continence. It was therefore fortuitous that the retained organs were those appropriate to urinary and fecal control. The failure to perform a pelvic osteotomy left a widely open pelvis that limited the reconstruction of the external genitalia and led to a less than satisfactory abdominal and genital appearance (Fig. 3) causing embarrassment and negating intimate relationships. The wide lower abdomen and pelvis caused considerable difficulty with clothing and contributed to a poor body image and reduced self esteem.

The late presentation of the second child at 10 years of age was perhaps fortunate in allowing us to undertake a reliable urodynamic study separately for each hemi-bladder. This determined that the better-formed right hemi-bladder had near normal detrusor and sphincter activity and was associated with a sensation of a full bladder with a wish to micturate, and the ability to retain urine and to fully micturate voluntarily. Closure of the incontinent left bladder neck and reconstruction of a single augmented bladder of increased capacity by combining both hemi-bladders, was therefore an appropriate evidence based decision. Not unexpectedly this young lady rapidly became fully continent of urine by day and by night. Six months postoperatively she remains completely dry for increasing periods presently of 2 h by day, and is dry by night. As in our first case, the failure to close the pelvis by osteotomy limited the esthetic appearance of the lower abdomen and pelvis as well as the external genital reconstruction.

### 3. Conclusion

Experience with these two patients at an interval of 20 years between them, has taught us to initially limit to cautious conservative non-destructive and potentially reversible surgery until reliable functional and dynamic studies are possible. Other experience with bladder exstrophy leads us to suggest that a neonatal pelvic

osteotomy during repair of the omphalocele would have allowed a better esthetic abdominal wall closure, and is essential to a relatively normal pelvic shape and an esthetically superior reconstruction of the external genitalia. It is the longer term esthetic as well as functional outcome that the patient is interested in, and indeed both are relevant to a good body image and to a normal quality of life. We are firmly of the view that timely multistage interventions can lead to a functionally and an esthetically superior reconstruction, and that these malformations, managed appropriately, have a potentially excellent prognosis. We recommend a *team approach* with provision of regular psychological support for these children and their families, particularly through adolescence and with long term follow-up well into adulthood.

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