

Accuracy and Predictivity of Cystoscopy and Genitography to Operative Findings in Persistent Urogenital Sinus due to Virilized Female Congenital Adrenal Hyperplasia

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ABSTRACT

Congenital adrenal hyperplasia in females leads to virilization of the external genitalia and an anomalous genitourinary tract, with a termination of the urethra and vagina into the persistent urogenital sinus. Surgical repair during infancy is indicated to raise a female child with external genitalia matching her gender and to avoid psychosocial and genitourinary complications. Earlier surgery is believed to be easier and of better results. Twenty five female patients with virilized external genitalia and persistent urogenital sinus due to congenital adrenal hyperplasia were studied. Interpretation of the genitography and cystoscopy was analyzed and compared to the operative findings to assess their accuracy and predictability. Sensitivity of cystoscopy and genitography is comparable (78.5% and 73% respectively), but cystoscopy has a higher specificity than genitography (91% and 83% respectively). Both investigations had a similar predictivity and were able to reveal the internal anatomy and operative difficulty in 56% of cases. In 12% both investigations had a similar predictivity, but failed to expect the operative difficulty. Cystoscopy was superior to genitography in 24%, but surprisingly genitography was more predictive in 8%. Neither genitography nor cystoscopy were independently predictive of the internal anatomy and intra-operative findings or difficulty of the studied cases. We therefore recommend doing both investigations as a routine preoperative workup. The benefit is to assess these cases properly and support decision making and choice of surgery.

Key words: DSD, Feminizing Genitoplasty, Cystoscopy, Genitography.

INTRODUCTION

In urogenital sinus anomalies, there is a persistent communication of the vagina with the urinary tract. In the majority of cases this connection occurs within the middle portion of the urethra. The two structures terminate into a common channel that exits on the perineum as a single opening^[1]. The vagina is shortened and fails to descend to the perineum and enters the urethra distal to the bladder neck. The older descriptions classify this as either a “high” confluence if the connection is proximal or a “low” confluence for more distal connections^[2].

Regarding the external genitalia; the virilization manifests as hypertrophy of the clitoris. Clitoromegaly ranges from minimal to a male-appearing phallus, with a urethra extending to the tip. The labia may be masculinized to form labioscrotal folds or, in severe cases, fused together as a scrotum^[3].

Persistent urogenital sinus (UGS) due to congenital adrenal hyperplasia in females is more recently regarded to occur in a spectrum, rather than simply “high” or “low”, and therefore evaluation and management must be tailored to each case individually^[1].

The recent surgical techniques imply mobilization of the urogenital sinus, either partially or totally. For these techniques to be successful as regards the urinary continence and voiding function, the urethra should have an adequate length above the confluence. This is a subjective judgment and an arbitrarily cutoff point of 15 to 20 mm has been used by previous reports^[4,5].

When the urethra is too short the risk of incontinence is high and, therefore a technique that preserves the urogenital sinus as a urethra should be used. The other point that has an impact on the surgical difficulty and outcome is

the depth (level) of urethral-vaginal confluence.^[1]

All patients with a urogenital sinus anomaly used to undergo a contrast genitography. The retrograde instillation of radiopaque dye is used to define the anatomy. Proper studies should identify the length of the common sinus, the depth (level) of the vaginal-urethral confluence, and the length of the proximal urethra. Some recent reports argue that genitography generally has limited usefulness in CAH^[6,7].

Endoscopic evaluation of the anatomy is generally done under the same anesthetic as definitive surgical correction, particularly for lower or intermediate lesions. If the anatomy is believed to be complex after genitography, cystoscopy may be performed initially alone in order to better decipher the anatomy, prepare the patient and consent the family for a major and challenging reconstructive surgery^[8].

The purpose of this report is description of the findings of both genitography and cystoscopy in virilized female CAH and analysis of the ability of each investigation to predict the internal anatomy and its impact on the surgical decision making.

PATIENTS & METHODS

Twenty five prepubertal females with virilized atypical genitalia due to congenital adrenal hyperplasia, presenting to the outpatient clinics of Cairo University Specialized Paediatric Hospital in the period from June 2011 to June 2013, were reviewed. The included cases were prepubertal females (46, XX karyotype). The excluded cases were male disorders of sexual development and debatable sex of rearing as; mixed gonadal dysgenesis or ovotesticular DSD.

For all cases an external genital examination was done. Serum electrolytes were measured at diagnosis and during follow-up. Karyotyping was done to confirm being 46XX. A hormonal profile was done at diagnosis and during follow up and an abdominal ultrasonography was done to visualize the uterus and ovaries.

Genitography was done under mild sedation (attended to by an anesthetist) for the delineation of the internal anatomy. It starts by positioning the patient in the exact lateral position on the X-ray table. The tip of the plastic syringe containing contrast is inserted into the external

meatus. Contrast material was injected under fluoroscopic control. The injection continued under a moderate pressure until the confluence appears, followed by the bifurcation of the urethra anteriorly and the vagina posteriorly, at this point serial spots were taken. The depth (level) of the confluence from the perineal floor was noted as well as the direction of the common channel, whether it was in a vertical, oblique or horizontal lie. The distance between the confluence and the bladder neck was determined to assess the urethral length proximal to the confluence. The caliber and length of the vagina proximal to the confluence were determined, if it was filled by contrast. The perineum as a level was determined by an experienced paediatric radiologist as the level of the distal end (meatus) of the common channel, which is further confirmed by the junction between the horizontal (superficial) and vertical (deep) portions of the urogenital sinus, delineated by the contrast.

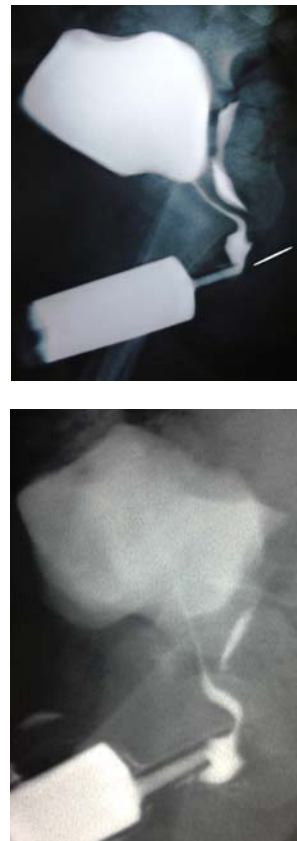


Fig. [1]: Genitography showing intermediate to high confluence



Fig. [2]: Genitography showing low confluence

Cystoscopy was done immediately prior to surgery. The procedure used Karl-Storz Endoskope size 9Fr. or 11Fr. with a working port. Saline flow was used to distend the urethra; this allowed appreciation of the vaginal orifice in the back wall of the urethra. The vaginal orifice was looked for around the pseudo-verumontanum. Balloon catheters were introduced into both the vagina and the bladder over guide-wires.

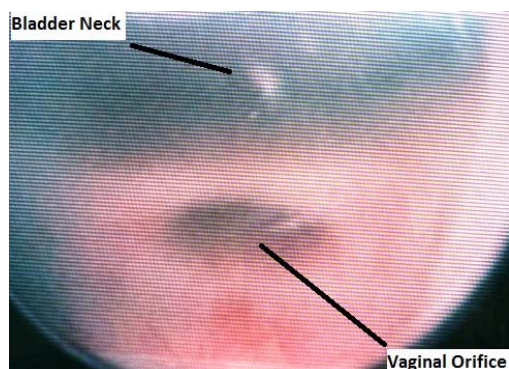


Fig. [3]: Cystoscopy showing high vagina in close proximity to the bladder neck

Our observations were analyzed after all cases of the study were operated upon. The operative technique was a feminizing genitoplasty, correcting both the external genitalia and internal anatomy at one stage. For the internal anatomy partial urogenital mobilization (PUM) was used whenever possible as our procedure of choice. Flap vaginoplasty, Passerini-Glazel flap and total urogenital mobilization (TUM) were done in few cases. For the external appearance; reduction clitoroplasty, preputial reconstruction of the labia minora and V-Y labio-scroto-plasty were done for all cases.

RESULTS

Twenty five cases of virilized females due to congenital adrenal hyperplasia were admitted to the care of the DSD team at Cairo University Specialized Paediatric Hospital (Aboulreish Hospital). All were adjusted by glucocorticoid (with or without mineralocorticoid) replacement therapy and operated upon. In this series the average age at which surgery was performed was 22 months; the median age was 18.5 months, range (6 months to 78 months).

The genitography showed a sensitivity of 72% and a specificity of 83%. The cystoscopy showed a slightly higher sensitivity of 78.5% and a significantly higher specificity of 91%.

The genitography and cystoscopy findings were equivalent and provided similar information regarding the site of vaginal entry, the length of the proximal urethra and the operative difficulty in 17 cases (68%). Among these 68%, neither could accurately predict the operative difficulty and operative findings in 3 cases (12%). These cases were among our most difficult cases, and required an operative time of 4 hours or more. In the remaining 14 cases (56%) they were both able to predict the surgical anatomy [Table 1].

Table [1]: Comparison of Genitography and Cystoscopy

<i>Result</i>	<i>Number of Cases (Percentage)</i>
Genitography & Cystoscopy both similar and predictive	14 cases (56%)
Genitography and Cystoscopy both failed to predict surgical anatomy	3 Cases (12%)
Cystoscopy more accurate than Genitography	6 Cases (24%)
Genitography more accurate than Cystoscopy	2 Cases (8%)

Surprisingly, the cystoscopy interpretation was inaccurate and less capable of anticipating the operative difficulty in 2 cases (8%). In these cases the genitography was more predictive. These two cases were not of a specific anatomy and represented average operative difficulty. The cystoscopy excelled the genitography in anticipating the operative difficulty in 6 cases (24%). In all these cases the cystoscopy expected a more proximal vaginal entry and was found to meet the operative findings better than the genitography.

The distal urogenital sinus was in a vertical (directly downwards) direction in 8 cases (32%), horizontally transverse in 4 cases (16%), and oblique in 13 cases (52%). The mean length of the proximal urethra was 21.5mm (range 10mm to 35mm). The vaginal depth was estimated by dividing the distance from the perineum to the confluence by the distance from the perineum to the bladder neck, this ratio has a Mean of 0.4441 (Median = 0.4667, Mode = 0.5), and follows a normal distribution curve.

The operative technique for correcting the internal anatomy was partial urogenital mobilization (PUM) in 19 cases (76%). Flap vaginoplasty was done in 3 cases (12%). Three cases (12%) needed more invasive procedures; two cases needed a Passerini-Glazel flap to be added to the PUM, and one case needed a TUM (dissection anterior to the urethra).

DISCUSSION

Current practice usually entails an early ultrasonography, which should identify a uterus if present. Although a gonad may be seen, ultrasonography is not useful in differentiating a testis, ovotestis, or ovary^[9]. In this series abdomino-pelvic ultrasound was capable of detecting a prepubertal uterus in 23 out 25 cases, however, it was not helpful to visualize the vagina or provide information to guide the choice of surgical approach.

Successful reconstruction depends upon an accurate preoperative definition of the anatomy. The genitourinary tract is traditionally evaluated by genitography and endoscopy. These investigations are used to identify the level of the vaginal-urethral confluence and the length of the proximal urethra^[10,11]. Some reports undermine the value of genitography as a high confluence

may not be identified radiographically and may be noted only on careful endoscopy by a few small punctuate openings at or near an apparently flattened verumontanum^[7,12].

VanderBrink et al. retrospectively evaluated the role of preoperative genitogram in surgical planning. Their genitograms revealed the complete anatomy of the urogenital sinus in 72%, and did not reveal the anatomy completely in 25% of the patients. The investigators could not catheterize the UGS in 2%^[7]. Compared to our series, the sample sizes are similar. The present study has the advantage of being prospective, and it was decided to contribute to this debatable point from the beginning of the study, hence genitographies were done with more precision. A member of the paediatric surgical DSD team attended all the genitographies with an experienced paediatric radiologist, and sedation was used in all cases. On the contrary to that study, all our cases could be catheterized with careful examination under sedation.

Our motive to defend the value of genitography is the consensus that one of the main determinants of operative difficulty is the depth of the confluence^[13]. Lateral view genitographies help to measure two distances, on which surgical decisions are made; first; the depth (level) of the vaginal-urethral confluence from the perineum, as all our surgical corrections were from a perineal approach and second; the length of the proximal urethra.

This lateral view radiographic evaluation is complementary to the cystoscopic measurement of the proximal urethra. It is important to note that the urogenital sinus distal to the confluence could be either superficially transverse or deeply vertical and could be either long or short without having an effect on surgical difficulty and outcome. The cystoscope cannot evaluate the direction and inclination (obliquity) of the urogenital sinus.

Podesta and Urcullo expressed a similar opinion to ours, that combining both investigations gives the best understanding of the confluence depth and proximal urethra. Proximal urethra is an indisputable determinant of the operative difficulty and outcome. The deeper the confluence, the higher the vagina, the shorter the proximal urethra and the more difficult the operation would be^[14].

CONCLUSIONS

Cystoscopy is more specific and predictive than genitography. The cystoscopic assessment immediately preoperatively is the mainstay of anticipating the operative difficulty and surgical approach. Genitography is still very important and completes the preoperative workup. In the lateral view, it adds very valuable information about the vaginal depth. We recommend routinely doing both genitography and cystoscopy to study these complex cases.

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