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Persistent Urogenital Sinus (PUGS) Anomaly in Children: A Single Center Experience

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KEYWORDS

Urogenital Sinus, Persistent Urogenital Sinus Children, DSD.

ABSTRACT:

Background: Urogenital sinus (UGS) or Persistent urogenital sinus (PUGS) is an uncommon congenital malformation characterized by failure of normal separation of the urinary and genital tract during embryonic development.It may present as an isolated defect or in association with syndrome or Disorders of Sex development (DSD), most frequently congenital adrenal hyperplasia (CAH). Evidence regarding clinical presentation and surgical outcomes from developing countries remain limited.

This study aimed to analysed the demographic profiles, clinical presentation, operation approaches and short –term outcomes of children diagnosed with PUGS.

Methods: This retrospective study was conducted in the Department of Paediatric Surgery at Bangladesh Medical University (BMU) between January 2016 and December 2020.

Results: Eleven cases were managed during the five years period. Age ranged from (1.5- 16) years with a median of 8.75 years. DSD, prodominantly CAH was identified 727% cases. Low-confluence PUGS was observed in 63.7 patients. Management of low-confluence PUGS included, flap vaginoplasty through perineal route or TUM (Total urogenital mobilization) method, while high- confluence cases required a 'Pull Through' procedure. Postoperative complications included vaginal stenosis in 27.3% and flap disruption in 18.2% patients.

Conclusion: PUGS was frequently associated with DSD and was more common of the low-confluence type. Selectionof surgical technique should be individualized based on anatomocal configuration.

Introduction:

Persistent Urogenital Sinus is a common channel formed by the urethra and vagina. During embryogenesis, the cloaca divides into anteriorly urogenital sinus and posteriorly anorectal canal. Due to failure of complete separation of the vagina and urethra from the urogenital sinus causing varying degrees of PUGS[1.] This affects the genetic females (46, XX) and frequently associated with congenital adrenal hyperplasia [2.] PUGS is two types, low -confluence and high -confluence which significantly influences surgical complexity and outcomes[3.]

Despite improvement in reconstructive surgery, outcome data are still variable. This study presented our institutional experience with urogenital sinus in children, focusing on the demographics, clinical presentation, surgical techniques and short-term outcomes.

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Methods and Materials:

This was a retrospective study conducted in the Pediatric Surgery Department of BMU from January 01,2016 to December 31,2020. The age of the child ranged from (1.5-16) years with a median age 8.75years.Upon admission, all patients were clinically investigated. The degree of virilization of the patient's external genitalia was recorded using the Prader Scale.

Hormonal assays, including TSH, FSH, LH, Testosterone, Oestrogen, Progesterone and Cortisol were performed. Karyotyping, USG of whole abdomen, Genitogram, Cystoscopy and MRI for assessing the vaginal anomalies and length were also conducted for all patients.

These datas were collected from medical records and operative theatre registry. Age at diagnosis, surgical techniques and outcomes were also recorded from these sources.

Statistical Analysis:

Data were analyzed using SPSS (Statistical Package for Social Science). Categorical variables were expressed in frequencies and percentage and continuous variables in mean $\pm$ standard deviation.

Results:

A total of 11 (eleven) patients were included in the study. The median age was 8.75 year (age range: 1.5 year to 16 year). The most (72.7%) common associated diagnosis was DSD, particularly CAH.

Demography and Clinical Profile of the patients

	Value	**Percentage (%)**
Total patient	11	
Age(range)	(1.5-16) years	
Median age	8.75year	
Karyotype 46XX	11	100
CAH	08	72.7
Ambiguous genitalia	02	18.2
Hydrometrocolpos	03	27.3
Amenorrhoea	02	18.2
Others: Urinary tract infection/ dys pareunia	04	36.4

Low -confluence PUGS was more common than High -confluence PUGS.

Anatomical Classification of PUGS(n=11)

Type	Number of Patient	Percentage (%)
Low confluence	07	63.6
High confluence	04	36.4

In case of low -confluence PUGS, total urogenital mobilization (TUM) was performed. Treatment of choice for high -confluence PUGS was 'Pull Through' the most (36.4%) common postoperative complication was vaginal stenosis.

Surgical techniques and outcome (n=11)

	Number of Patient	Percentage (%)
Techniques		
TUM	05	45.5
Flap Vaginoplasty	03	27.3
Pull Through	02	18.2
Intestinal Vaginoplasty	01	9.1
Outcomes		
Vaginal Stenosis	04	36.4
Disruption of flap	02	18.2

Discussion:

Persistent urogenital sinus (PUGS) is a congenital defect in females where the urethra snd vagina fail to separate resulting in a single perineal opening[4]. It occurs in approximately 6 per 100,000 female birth[5]. In children,this anomaly is commonly associated with Disorders of Sex Development (DSD), particularly Congenital Adrenal Hyperplasia (CAH), where the

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incidence is 1: 500[2]. Other anomalies and syndromes such as VACTERL, Townes - Brocks syndrome, MKS and Bardet-Biedl syndrome may also be associated with PUGS[6]. Diagnosis becomes difficult when patients present with overlapping phenotypes[7].

Study Demographic and Association:

In our study, the presenting age of patients ranged from 1.5-16 years. In contrast, Aboalazayem A and his colleagues[2] reported a patient age at surgery of 9-109 month. This delayed presentation in our study may be attributed to a lack of awareness, insufficient knowledge among local health care providers and low literacy rates.

In this study, eight patients were diagnosed with DSD, specifically CAU as an associated condition. This aligns with finding by Podesta and his colleague[8], who identified 12 patients with urogenital sinus anomalies associated with DSD.

We found that low -confluence PUGS more (63.7%) common than high -confluence PUGS, a result consistent with the finding of Oshiba A and his colleagues[9].

Clinical findings of PUGS:

Pelvic mass (hydrometrocolpos/dilated bladder) and ambiguous genitalia[10] are the common presentation of PUGS. However, the PUGS may be present with recurrent urinary tract infection, amenorrhea and dyspareunia.PUGS most commonly associated with DSD,particularly CAU[11]. In our study, patients presented with similar clinical features. On the other hand, Fiorentino R and his colleagues[12], reported that their patients presented only with a pelvic mass.

Diagnostic and Therapeutic Approach:

The diagnosis of PUGS is challenging due to its rarity and complexity. This anomaly is often overlooked when the anus is normally positioned and external genitalia appear female. A high index of suspicion for PUGS (as in our case), should be maintain when a single perineal opening is present within the vestible, particularly when associated with hydrometrocolpos or pelvic cystic mass. This finding is considered pathognomonic[13,14].

A combined strategy involving clinical examination, karyotyping, hormonal profiling and imaging tools are required for the diagnosis and therapeutic management of PUGS, especially since it is often association with multiple clinical entities.

Surgical Challenges and Techniques:

The most challenging aspect is the treatment of PUGS. In cases of low- confluence PUGS, the separation of the urinary and genital tract is usually straightforward. However, when PUGS involves high- confluence, these two sysytem can be difficult to separate[15]. If separation is unsuccessful, the PUGS may persist or clinical features of urethral injury develop[16].

During the IVth World Congress of the International Society of Hypospadias and Disorders of the Sex Development (ISHID), a voting system of global deligates (78%) favour an optimal age for female genital reconstruction before two year. They also recommended one-stage procedure[17]. This timing for surgical correction is preferred as it causes fewer psychological effects than delayed surgery[1].

Surgical Standards by Common Channel Length:

Skilled surgical techniques and the selection an accurate surgical approach are crucial for favourable outcomes.

According to Prader Scale:

Prade Class 1: The preferred treatment is urogenital sinus incision.

Prader Class I and II: Perineal flap vaginoplasty is planned.

Prader Class II or above:

'Pull Through' procedure should be planned based on the location of the vaginal confluence point and length of the common channel.

According to the length of common channel Marei MM etal[18] and TugtepeH etal[19] were performed surgical treatment as below:

Common Channel Length	Recommended Procedure
<20mm (or <2cm)	PUM (Partial Urogenital Sinus Mobilization)
25 to 35 mm (2.5-3.5cm)	TUM (Total Urogenital Sinus Mobilization)

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>40mm (>4cm)	‘Pull Through' procedure

In low -confluence PUGS, PUM or TUM is considered the gold standard treatment, this procedure can also be performed in high- confluence PUGS. However, for most high -confluence PUGS cases the ‘Pull Through' procedure remain the preferable option. Our study findings were consistent with the aforementioned literature in the management of PUGS.

Currently, there are no standardized follow-up protocols after vaginal reconstruction. Most outcomes rely on clinical exam and patient's report rather than validated assessment instrument.

Vaginal stenosis was common after surgery[3]. This observation parallels our study results.

Conclusion: Persistent UGS represents an uncommon and anatomically challenging congenital malformation in the pediatric population. Optimal management often requires coordinated input from pediatric surgeon, endocrinologist and other specialists.

Successful reconstruction deoends detailed preoperative anatomical evaluation and individualized surgical planning.

In our series, the anomaly was predominantly associated with DSD especially congenital adrenal hyperplasia, which aligns with previously reported observations.

Limitations:

1.Sample size in this study was very small.

2. The study only examined the short-term surgical outcome of these patients.

Recommendation:

1. Life long follow-up is essential to assess the functional, psychological and reproductive issues for these patients.
2. Continued research is essential to establish universally standardized clinical guidelines for the management of patients with PUGS

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