Original Article

Sensitivity of Flush Genitogram in the Diagnosis of Disorder of Sex Development (DSD)

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Abstract

Background: Disorder of sex development (DSD), previously known as ambiguous genitalia are group of Individuals who have a genital appearance that does not permit gender declaration at birth. Gender assignment in children with DSD must be done as early as possible and requires close correlation between the history and physical examination, appropriate genetic and laboratory studies and imaging studies. **Objective:** The objective of this study is to assess the sensitivity of the flush genitogram in the evaluation of children with DSD. **Materials and Methods:** A prospective study was done over 5 years; the flush genitogram findings of the children were compared with the laparotomy findings at mini lap. **Results:** Over 5 years, 39 children were evaluated using both a flush genitogram and mini laparotomy. The correct sex was identified by a flush genitogram in 18 children, with a sensitivity of 56% and a false positive in 6 children. **Conclusion:** The sensitivity of the flush genitogram is found to be low, other methods of assessment are necessary before gender assignment in children with DSD.

Keywords: Diagnosis, disorder, flush genitogram, sex development

Introduction

Disorder of sex development (DSD), previously known as ambiguous genitalia are group of Individuals who have a genital appearance that does not permit gender declaration at birth.[1,2]

Gender assignment in children with DSD must be done as early as possible and requires close correlation between the history and physical examination, appropriate genetic and laboratory studies, as well as imaging studies.[3,4]

They are considered social and medical emergencies, especially for salt-losing group of patients and early assignment of gender has a profound effect on the successful outcome for the family and the child.[5]

Ambiguous genitalia have been reported worldwide to occur in about 1 in 1000 births as an isolated abnormality, although it can be associated with other physical abnormalities as well.[6,7]

There are several classifications of DSD, one of the most common is as follows:

1. Female pseudohermaphroditism (46 XX, with two ovaries)
2. Male pseudohermaphroditism (46 XY, with two testes)
3. Ovotesticular DSD, i.e. both ovarian and testicular tissues present
4. Gonadal dysgenesis, either mixed (a testis and a streak of gonads) or pure (bilateral streaks of gonads).[8-11]

In our environment, most of the patients with DSD still come to the hospital late and the diagnosis may be difficult. Various methods of investigation are used in our environment; the best investigation for DSD is yet to be determined.[12]

Radiological investigations have a significant role in making an accurate diagnosis; a flush genitogram is one of these common investigations in the diagnosis of DSD.[13,14]

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Some studies tried to report the accuracy of genitogram in the diagnosis of DSD, but few studies compared it with actual diagnosis during surgery in order to identify its accuracy in the diagnosis.[15]

This study aimed to determine the sensitivity of the flush genitogram in the diagnosis of DSD, by comparing it with the findings of mini laparotomy.

Materials and Methods

This is a prospective study over 5 years after obtaining ethical approval from the ethical committee of our hospital. All children presenting to the paediatric surgery unit with ambiguous genitalia, single perineal opening and undescended testis and having undergone a flush genitogram during the study period were included in the study.

The results of the flush genitogram were compared with intraoperative findings during the mini laparotomy and the presence of well-developed uterus at mini laparotomy is considered to be the standard to be compared with the findings of the flush genitogram. The positive result was considered as the ability of the flush genitogram to detect the presence of the uterus in children with a well-developed uterus, which was used to calculate the overall sensitivity of the procedure.

Flush genitogram procedure

During the procedure of a flush genitogram, a preliminary plain X-ray film of the pelvis was obtained in the supine and lateral positions. The perineal orifices were canulated using a feeding tube and a contrast medium was injected under fluoroscopy. When the pelvic chamber was filled with the contrast, a supine and lateral view was taken. The procedure was carried out by a senior registrar in the radiology department of the hospital. The findings of the genitogram was recorded and documented.

Results

Within the 5-year period of the study, 39 patients were seen with DSD in paediatric surgery units of our hospital. The initial genders assigned by the parents were 22 males and 17 females, respectively [Figure 1].

The age at presentation ranged between 3 months and 14 years, but most of the patients presented before the age of 2 years [Figure 2].

Eight patients have palpable gonads on examination (5 left and 3 right palpable gonads).

At mini laparotomy, four patients amongst the six ovotesticular DSD have a complete uterus, while two patients have a hemi-uterus. Three among those with unilateral palpable gonad were found to be males, two have female internal genitalia and three have ovo-testicular DSD[Figures 3,4,5].

The only ovo-testicular patient, in whom Mullerian structure was identified with a genitogram, has a complete uterus.

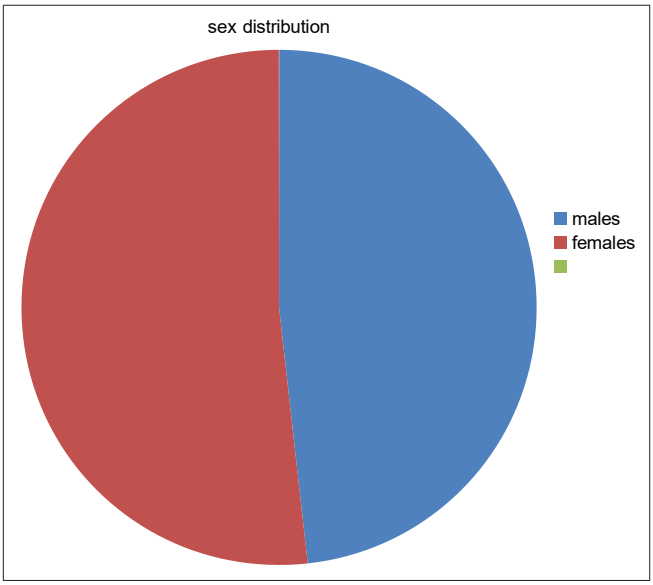


Figure 1: Sex distribution at presentation (Initial gender assigned by parents)

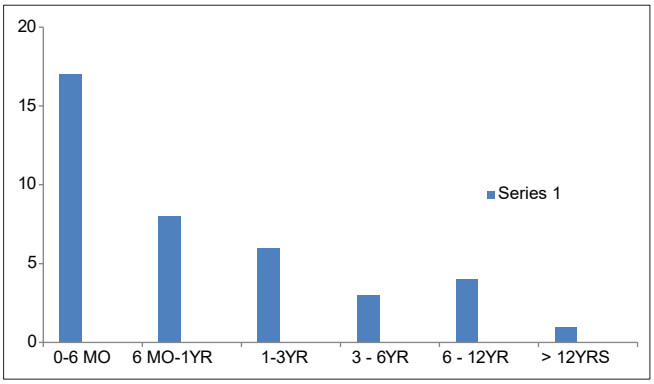


Figure 2: Age at presentation



Figure 3: Pre-operative picture

Perhaps, the patients with hemiuterus had some obstruction which prevented the Mullerian structure from being visualised by genitogram.



Figure 4: Flush genitogram (lateral view)

Table 1: Flush genitogram and mini laparotomy results

	Minilap (uterus present)	Minilap (uterus absent)	Total
Genitogram (uterus present)	13	4	17
Genitogram (uterus absent)	10	12	22
Total	23	16	39

After evaluation using mini laparotomy, 19 were found to have uterus (females), amongst which 5 were initially assigned male by the parents, 14 were found to have no uterus (male) and 6 have ovotesticular DSD, with 4 of them having well developed uterus and 2 have hemi uterus.

Among the 39 children who had mini laparotomy 6 have ovotesticular DSD, with 4 of them having well developed uterus and both ovary and testicular tissue and 2 have hemi uterus. Nineteen have well developed uterus with no evidence of testicular tissue. The remaining 14 have no evidence of the uterus at laparotomy.

The flush genitogram was able to identify 13 children with uterus (1 of them has ovotesticular DSD) correctly and reported 4 children without uterus as positive for the presence of uterus, and the remaining 22 children were reported to have no uterus, out of them ten have well developed uterus at minilaparotomy. Four out of those reported to have uterus by genitogram were found to have no uterus at mini laparotomy [Table 1].

1. Sensitivity = True positive/true positive + false negative × 100 = 13/13 + 10 × 100 = 56.5%
2. Specificity = True negative/true negative + false positive × 100 = 12/10 + 4 = 10/14 × 100 = 85.7%
3. Positive predictive value = True positive/true positive + false positive × 100 = 13/13 + 4 × 100 = 13/17 = 76.5%

4 Negative predictive value = True negative/true negative + false negative × 100 = 12/12 + 10 × 100 = 12/22 = 54.5%



Figure 5: Mini laparotomy (intraoperative picture)

Discussion

The age distribution in this study ranges between 3 months and 14 years, but majority presented before 2 years of age, which is similar to what was observed in other studies.[16,17]

A flush genitogram is often the radiological investigation done to delineate the internal genitalia when evaluating patients with genital ambiguity. It is readily available in most centres.[18,19]

During the period of our study, gender was found correctly in 23 children using a flush genitogram (10 males and 13 females). Four males were reported to have the uterus (females) while eight females were reported to have no uterus. Four out of six children with ovo-testicular DSD were reported by the flush genitogram to have no uterus (males).

The sensitivity (56.5%) and specificity (85.7%) of the flush genitogram in this study are similar to the sensitivity 55.6% reported by Elhalaby, in Egypt, but lower than the sensitivity of 60% reported by Chartin *et al*.[20,21]

In another study by AlJurayyan reported the sensitivity of genitogram was reported to be 84.2%, which is said to be lower than the sensitivity of pelvic ultrasound.[22] However, Vanderbrink *et al*. reported a lower sensitivity of 72% using genitogram in the evaluation of children with disorders of sex development.[18]

The specificity of the flush genitogram was found to be 85.7%, while the positive and negative predictive values were found in our study to be 76.5% and 54.5%, respectively.

The low sensitivity in this study could be improved if more attention is paid to technical details when taking the images

Furthermore, when combined with trans-abdominal USS, the yield of a flush genitogram can be improved

Conclusion

The sensitivity of the flush genitogram is found to be low, hence other investigation tools are required for proper evaluation before definitive treatment.

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Conflicts of interest

There are no conflicts of interest.

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