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Persistent mullerian duct syndrome: A 24-year experience☆,☆☆

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ABSTRACT

Background: Persistence of mullerian duct derivatives in otherwise normal male child is a very rare disorder. This may lead to diagnostic as well as management dilemma.
Materials and methods: The medical record of 27 cases of persistent mullerian duct syndrome (PMDS) operated in three teaching hospitals more than a period of 24 years is retrieved and analyzed for demography, clinical presentation, investigations, and treatment.
Results: There were a total of twenty seven male children with PMDS. The age was ranged between 3 months and 19 years. Ten patients presented with isolated bilateral UDT, six patients with bilateral UDT and unilateral inguinal hernia (4 left and 2 right sided inguinal hernia), and eight patients presented with right inguinal hernia and left sided UDT. Eight of 27 patients showed familial trends i.e. four pairs of brothers had PMDS in our series. In 21 patients, the diagnosis was made incidentally while operating for UDT and inguinal hernia. At operation 5 patients had female type of PMDS and 22 patients had male type PMDS. In 6 patients (male type), the PMDS was associated with transverse testicular ectopia. In 18 patients the initial operation was performed through inguinal incision with excision of mullerian remnants in the same settings in 12 patients. In 4 patients, straightforward laparotomy performed (familial cases) to excise mullerian remnants. In 5 patients, the PMDS was diagnosed on laparoscopy; initially biopsy of these remnants and gonads was done followed by excision of remnants by laparotomy approach. Biopsies taken from gonads in each patient revealed testicular tissue with variable degree of immaturity and dysplasia. The biopsy of mullerian remnants did not reveal any malignancy. All patients were genotypically male.
Conclusion: Isolated undescended testes, left UDT and right inguinal hernia, bilateral UDT and unilateral inguinal hernia are the main presenting features of PMDS. About 30% of the patients showed familial tendency. Inguinal exploration for UDT or inguinal hernia, and laparoscopy for UDT reveal incidental findings of mullerian remnants. PMDS can be managed as single stage procedure however two stage procedure including gonadal biopsies in first stage followed by mullerian remnants excision and orchidopexy in the second stage can be opted if there is doubt about gonads and genotype.

PMDS is characterized by the presence of mullerian duct structures as uterus, cervix, fallopian tubes, and rarely upper vagina in otherwise phenotypically normal 46-XY male. The condition is quite often associated with cryptorchidism and inguinal hernia. In majority PMDS is discovered incidentally while operating for undescended testis or inguinal hernia. The failure of formation, release, or action of anti-mullerian hormone (AMH) secreted by the testes in intra uterine life is responsible for this condition [1–4].

This case series describes our 24-year experience of managing this condition at three tertiary health facilities of Pakistan. The clinical and pathological features and management of the condition are discussed along with a review of the literature.

1. Materials and methods

It is a retrospective study of 27 patients of PMDS managed from July 1990 to May 2014; in the Departments of Pediatric Surgery of The Children's Hospital and The Institute of Child Health, Lahore and Mayo

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The record of these patients, consisting of demography, history, clinical examination, biochemical tests, operative findings, postoperative events, and follow up of most of the patients was retrieved and reviewed.

2. Results

2.1. Demography

We came across a total of 27 patients having mullerian duct remnants in otherwise healthy male children. The age was ranged between 3 months to 19 years with a mean of 5.14 years (SD ± 5.2).

2.2. Presentation

The majority of patients presented with bilateral undescended testes associated with inguinal hernia. Of 27, ten patients presented with isolated bilateral UDT, six patients with bilateral UDT and unilateral inguinal hernia (4 left and 2 right sided inguinal hernia), and eight patients presented with right inguinal hernia and left sided UDT. Table 1 describes the clinical presentation of the study population.

2.3. Familial tendency

Out of 27 patients, 8 patients showed familial trends i.e. four pairs of brothers had PMDS in our series.

2.4. Diagnosis

In 21 patients, the diagnosis was made incidentally while operating for UDT and inguinal hernia. Ultrasound suspected mullerian duct remnants in 5 cases. In 6 patients, we suspected the diagnosis of PMDS pre-operatively on the basis of clinical findings of thick structures palpable in the hernial sac, ultrasound suspicion and history of PMDS in their elder brothers (4 patients).

In all patients the external genitalia were phenotypically male with normal phallus size.

Serum testosterone was refused by most of patients because of the cost factors. In 2 patients it was normal whereas in 3 other patients it was on lower normal levels.

2.5. Operative details

At operation 5 (18.5%) patients had female type (Fig. 1) of PMDS and 22 patients had male type PMDS (Fig. 2) including transverse testicular ectopia in 6 (22.2%) patients.

In 18 patients the initial operation was performed through inguinal incision for the management of UDT and/or inguinal hernia. Out of these 18, mullerian remnants were excised in the same operation in 12 patients (7 through the same inguinal incision and in 5 through laparotomy). In the remaining 6 of 18 patients, initially only biopsies were taken from gonads and mullerian remnants and excision of mullerian remnants were performed later on through the laparotomy in 4; 2 cases are in pipeline for excision of remnants and orchidopexy.

Table 1
Clinical presentation of the study population.

No. of patients	Presentation	Proportion
10	Bilateral UDT	37%
2	Bilateral UDT and bilateral inguinal hernia	7.4%
6	Bilateral UDT and unilateral inguinal hernia	22.2%
8	Left UDT and right inguinal hernia*α	29.6%
1	Right UDT and left inguinal hernia	3.7%

α in 3 cases vas deferens were not uniting with the testes.
* One patient had irreducible inguinal hernia.

In 4 patients, straightforward laparotomy was performed (familial cases) as to high suspicion of PMDS as supported by USG and mullerian remnants excised along with orchidopexy.

In 5 patients, the PMDS was diagnosed on laparoscopy; initially biopsy of these remnants and gonads was done followed by excision of remnants by laparotomy approach.

Table 2 describes summary of operative details.

2.6. Histopathology

Biopsies taken from gonads and remnants revealed testicular tissue with variable degree of immaturity and dysplasia and presence of uterus and fallopian tubes with variable degree of maturity in all the patients, respectively. No malignancy was found in any specimen.

2.7. Genetics

A Barr body test was performed in 10 patients (−ve) during initial tenure of the study when karyotyping was not available to us; karyotyping was performed after the operation in the remaining cases that showed 46 XY genome.

2.8. Fertility

Four patients have been married but there were no offspring. Majority of patients lost to follow-up.

3. Discussion

PMDS was first recognized 80 years ago [4]. More than 200 cases have been reported in the English world literature [1,2,5–9]. Nelson presented the first complete review of early cases in 1939 and termed it as "*hernia utri inguinale*" [4].

Classically, these patients present with unilateral or bilateral undescended testes with unilateral or bilateral inguinal hernia in 80% of cases [2,5]. In our series, 37% patients presented with isolated bilateral UDT, 30% patients presented with left UDT and right inguinal hernia, and 22% patients presented with bilateral UDT and unilateral inguinal hernia. At times, they present with irreducible inguinal hernia [14] as one of our case. They usually have normal external genitalia i.e. well-developed penis and scrotum. That is why hermaphroditism is not suspected before operation. The syndrome is usually discovered incidentally while operating for inguinal hernias or undescended testes. Similarly in our series, 21 of 27 patients were not suspected for PMDS before operation. Genotypically they are all XY males as also found in our case series.

This syndrome can be classified in to three anatomical types on the basis of location of testes and mullerian duct structures [1,7,14]. The first and most common is "male form" which is found in 80–90% of cases [1,14,18,19]. In this form at least one testis is present in the hernial sac; uterus and ipsilateral tubes are either present in the sac or can be drawn, on gentle traction into the sac bringing the contra lateral testis with it. The second and least common type is "female form", seen in 10–20% of cases, characterized by the presence of testes in the pseudo-ovarian position attached to fimbrial end of fallopian tubes and uterus in the normal female pelvic position [1,14,18]. These cases usually present with bilateral non-palpable abdominal testis without hernia. In third type, both the testes are present on the same side, either in inguinal canal or scrotum, and labeled as "transverse testicular ectopia". In our series 5 (18.5%) of 27 patients had female type of gonadal arrangement. In the rest 22 patients male type arrangement was found including six (22.2%) cases of transverse testicular ectopia. More than 60 cases of transverse testicular ectopia with persistent mullerian duct syndrome are reported in the literature [7,9,11–16]. Out of these, nearly 50% of cases are reported in children [9,11,13,22,23].