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### RESEARCH ARTICLE

#### CRYPTORCHIDISM AND 46 XY MALE GONADAL DYSGENESIS LAPAROSCOPIC DIAGNOSIS ABOUT A CASE

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#### Abstract

Disorders of Sex Development (DSD), formerly known as sexual ambiguity, are abnormalities leading to discrepancy between internal genital organs (IGO), external genitals (EGO) and secondary sexual characteristics. We report on a particularly complex case of 46 XY gonadal dysgenesis, monitored at the children's hospital of Rabat, which presented a male phenotype and whose parents consult for bilateral cryptorchidism, with well-individualized male external genital organs, a male morphotype without any other clinically detectable malformation. Our goal is to contribute to the study of the rare peculiarities of sexual ambiguities, to offer a diagnostic approach to their management.

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#### Introduction:-

Sexual dedifferentiation is the result of a series of events which follow one another in a precise chronological order, from the fertilization of the ovum by the spermatozoon, until the end of growth and puberty. The occasion for diagnosis generally arises from birth, before abnormal EGO, but can be much later, at the age of puberty, before puberty delay, amenorrhea or the appearance of discordant secondary sexual characteristics. The diagnosis must be as early as possible, sometimes prenatal, to avoid any error in the attribution of sex in which the child will be raised, an error whose psychological consequences are very serious. (1)

#### Patients and Methods:-

##### Clinical case:

Our case relates to an 8 year old child, of male morphotype, whose parents, consulted for absence of testicles in the purses. They had already consulted several times since the age of 2 years, he underwent several explorations among others abdomino-pelvic ultrasounds in search of testicles and hormonal assessments. On examination, the child present was in good general condition, with a respiratory rate of 22 beats per minute, heart rate of 102 cycles per minute, a blood pressure of 12/6, on examination of the external genital organs, the child had a normal looking penis, well individualized, normal length for age, urethral meatus in normal position on the glans, not yet circumcised, absence of testicle in the scrotum no hypospadias or vaginal opening and non-palpable testicles. Exploratory hormone checkups have found a 46XY karyotype, with normal testosterone and normal anti-Mullerian

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hormone levels. The abdomino-inguino-scrotal ultrasound found the left testicle in the left iliac fossa, the other testicle not being seen. The child was admitted to the block for laparoscopic exploration. On exploration, we found two bilateral oval-looking structures of size and shape similar to the testes, connected to the fallopian tubes and to an uterus and an internal vagina, the two "testicles" were vascularized by the uterine arteries, with channels but the prostate was absent. That confusion between ovaries and testes, an extemporaneous biopsy was realised in order to confirm if it is a testicle or ovary. The results of the pathology analysis concluded that there was testicular tissue on both sides. After a laparoscopic hysterocorproctomy was performed, the gonads and vas deferens were released and a lowering and fixation were performed.



**Figure 1:-** Laparoscopic exploration in search of ectopic testes : testicles in abdominal position, connected to fallopian tubes, uterus and internal vagina.



**Figure 2:-** Gonads after laparoscopic realise, incision in the lower and fold abdominal in order to be loweed and fixed in the scrotum

**Discussion:-**

46 XY gonadal dysgenesis is normally manifested by an abnormal development of the gonads which is manifested by the presence of external and internal female genitalia despite the male 46XY karyotype. The reason for consultation is either a sexual ambiguity, or for hypospadias, or for an absence of testicles. Our patient consulted for a cryptorchidism, he presented a well differentiated penis, of normal size and shape for the age. The absence of testicles in the purses was the element worrying the parents and therefore the reason for consultation. Mixed gonadal dysgenesis is incomplete and almost always asymmetrical. There is a strip and a dysgenetic testicle on the other. There is no follicle. The dysgenetic testis is sometimes palpable in an inguinal situation. The genitals are asymmetrical: a fallopian tube and a half-uterus are present on the side of the strip and are more or less developed on the side of the dysgenetic testicle. A vas deferens may be present, attesting to early androgenic secretion during fetal life [2]. In our case, the dysgenesis was symmetrical, and we observed two testicles with reduced size, connected to the fallopian tubes on both sides and to the uterus. The uterus was itself connected to the internal vagina. The external genitalia were of the male type: normal phallus, scrotum of good quality and good size, without trace of femininity apart from the absence of palpable testicles neither in scrotal, nor in inguinal nor abdominal. The deferent canal was present. In front of the presence of gonads having an aspect of testicles, despite the male phenotype, the XY karyotype and the hormonal tests in favor of masculinity, an extemporaneous biopsy was carried out to guide the continuation of the treatment.

**Diagnostic context:**

Most 46 XY gonadal dysgenesis is of the female phenotype at birth, and manifests itself in virization at puberty, or sometimes an appearance wrongly evoking a posterior hypospadias. In rare cases where a definitive diagnosis cannot be determined, and in infants with intra-abdominal or non-palpable testicles in whom DSD are being considered, open or laparoscopic exploration with gonad biopsy may be necessary [3]. In some cases, the differential diagnosis of DSD depends on the interpretation of the histology characteristic of the gonads [4,5]. Infants with intra-abdominal or non-palpable testicles in whom the precise diagnosis is not available with karyotyping and the serum study will require open or laparoscopic exploration with deep longitudinal gonadal biopsies for histological evaluation, which will determine the presence of ovotests, gonads, or dysgenetic testes, thus confirming the diagnosis. In our case, the phenotype is typically male, the approach being based essentially on cryptoorchidism. Ultrasound explorations had already been made in search of a possible intraabdominal localization of the testicles, objectifying the left testicle in an abdominal situation, the right testicle not being seen. After the hormonal tests which show normal levels of the male hormones, the decision of laparoscopic exploration was taken, which allowed us to discover one of the internal genital organs of female type, and doubtful gonads. As the deferred biopsy requires a waiting period and therefore another intervention, an extemporaneous biopsy is performed at the end of allowing a surgery in one time. The gonad biopsy returns in favor of the testicular tissue on both sides.

**Surgical management:**

The goal of surgery is to make the external genitalia ambiguous compatible with the assigned sex, preventing obstruction or urinary tract infections, preserving potential sexuality and reproduction, and maximizing the anatomy to improve sexual function [6]. Surgical corrections usually involve the gonads and external genitals and often the presence of a urogenital sinus. There is no indication that prophylactic removal of asymptomatic discordant structures is necessary. In general, it is recommended that the decision regarding genital surgery be made by the parents and, if possible, the patient, under the advice of the medical team. It is important to inform parents that a functional result is more important than an aesthetic result. For our case, the child already had a well individualized phallus; he was raised as a boy, although the situation of the gonads was until the unknown.

After the histology results, the parents were warned about the presence of the male internal genitalia, and, convinced that their child was a boy, a psychological interview was necessary to discuss the follow-up. charge. Thereby, in agreement with the parents and the healthcare team, the decision was a partial laparoscopic hysterocolpectomy, a release of the gonads and their pedicle, then they are lowered to fix them in the scrotum. We will not miss any doubts especially for the reproductive function following the absence of the prostate. According to some experts, early childhood genital surgery, which appears to be consistent with the sex of the child, is an important psychological support for the family. Others suggest that the appearance modification operation is not urgent and that it is more appropriate to delay surgery until a patient is old enough to be fully informed and to give consent [7]. In our case, the situation was the same, the parents were agreed to the male sex which had been apparent from birth, and the decision to eliminate female internal genital organs was taken by informed consent, written and signed by the parents of the child.

**Conclusion:-**

This is an exceptional form of gonadal dysgenesis, to which we have not found an exact classification in the literature. It would be advantageous to do an early exploration in front of any form of cryptorchidism with non-palpable testicles, despite the apparently reassuring aspect of the external genitalia. The decision to take a therapeutic approach must include the parents and the child if the latter is cared for late, according to socio-cultural standards.

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