

CASE REPORT

ASYMMETRICAL GONADAL DYSGENESIS IN A MALE PATIENT: A RARE CASE REPORT

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HOW TO CITE THIS ARTICLE:

Anilkumar Sirasagi, Mirza Asif Baig. "Asymmetrical Gonadal Dysgenesis in a Male Patient: A Rare Case Report". Journal of Evolution of Medical and Dental Sciences 2015; Vol. 4, Issue 26, March 30; Page: 4531-4534, DOI: 10.14260/jemds/2015/655

ABSTRACT: BACKGROUND: Asymmetric Gonadal dysgenesis (AGD) very rare intersex disorder seen in less than 5% of all intersex disorders. Asymmetric gonadal dysgenesis refers to individuals who have abnormal and symmetrical gonadal development, usually a differentiated gonad on one side and a streak gonad or streak testis on other side. The importance of identifying these lesions are important because risk of development of malignancy as high as 30%, which always influenced by y-chromosome. **CASE REPORT:** A 21 year old male patient presented with left sided obstructed inguinal hernia with cryptorchidism. On exploratory laparotomy, uterus with bilateral adnexa and testis are seen. Microscopy revealed features of asymmetric gonadal dysgenesis. True hermaphroditism was the closest differential diagnosis and was excluded by the strict histological criteria necessary for diagnosis. **CONCLUSION:** Asymmetric gonadal dysgenesis is a rare disorder. The rare nature of this entity and its clinical implications merits its reporting.

KEYWORDS: Asymmetric gonadal dysgenesis, cryptorchidism, gonadoblastomas.

KEYMESSAGES: Asymmetric gonadal dysgenesis is a rare disorder. It is important to diagnosis this entity because 30% of cases progress to gonadoblastomas. The rare nature of this entity and its clinical implications merits its reporting.

INTRODUCTION: Disorders of sex development are very complex diseases. Most of these disorders are congenital conditions in which development of anatomical, chromosomal or gonadal sex are atypical. True incidence in India is not known. In western countries it is as high as 1 in 2000 live birth. Asymmetric gonadal dysgenesis [AGD] is one of rare entities and part of intersex disorders accounts for 7 % of all intersex disorders.^{1,2}

AGD refers to individuals who usually have a differentiated gonad on one side and a streak gonad or streak testis on other side. Few authors apply the term to patients who show testicular differentiation on either side, bilateral streak testis or bilateral dysgenetic testis or gonadoblastomas on other side. They usually present as symptomatic male phenotype. In asymptomatic patients the diagnosis may delayed till their adult life. Rarely they it is incidental finding in the male or female phenotypes.^{1,2,3}

CASE HISTORY: A 21 Years old married male patient and father of 2 children, presented with acute lower abdomen pain to emergency unit. On examination, bilateral cryptorchidism and mass in left inguinal canal noted. He had a male phenotype with well develop secondary sexual characters. External genitalia were normal. Ultra sonogram of abdomen and pelvis revealed bilateral cryptorchidism and obstructed inguinal hernia. Semen analysis, karyotyping and other investigations were not done since patient presented with surgical emergency and clinically it was not suspected. On exploratory laparotomy, uterus with bilateral fallopian tubes and ovaries were noted and was sent for histopathological examination.

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Macroscopically Uterus measures 8x5x4 cm, both tubes measures 4cms and? ovaries 2.5x2x1cm (Pic. 1). Microscopic examination of uterus and both side fallopian tubes showed normal histology. Section studied through globular tissue attached to fallopian tubes show vas-deference and epidydemal and seminiferous tubular structures (Pic. 2). No ovarian stroma or primordial follicles are seen. Section studied through separately sent testicular biopsy show features of incomplete spermatocytic maturation arrest (Pic. 3). A Diagnosis of Asymmetrical gonadal dysgenesis was made and advised for bilateral gonadectomy since chances for development of malignancy in gonads are very high. However we lost the follow up the case.

Fig. 1: Uterus with both side fallopian tubes and ovary like nodular structure.



Fig. 1

Fig. 2: Microscopic picture shows adjacent to fallopian tube, vas deference and epidydemis structures seen. (10X).

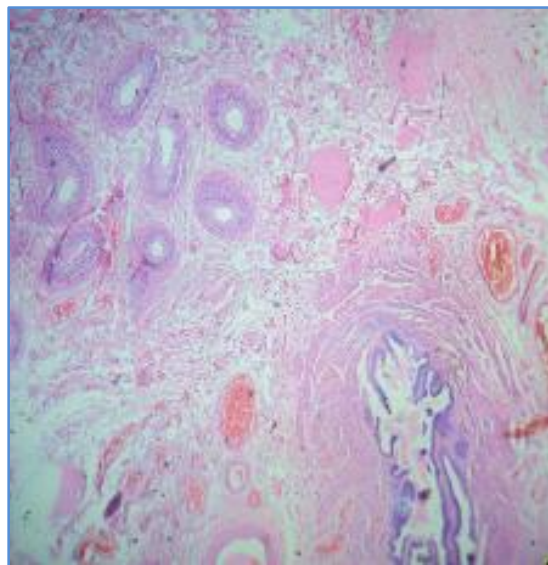


Fig. 2

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Fig. 3: Testicular biopsy shows atrophied seminiferous tubules with Incomplete Spermatocytic Maturation Arrest. (40X).

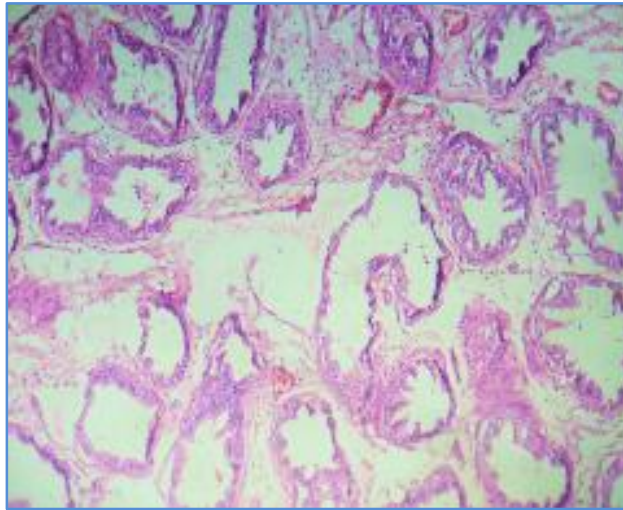


Fig. 3

DISCUSSION: Children born with intersex disorder comprise about 1.7% of all live birth in western literature. Asymmetrical Gonadal Dysgenesis is one of the rare and important intersex disorder constitutes around 5 - 7% of all intersex disorders. The exact incidence in India is not known. This disorder diagnosed if one side gonad shows more complete development and can be identified as an ovary or testis (usually testis) and other side gonad is streak or malignancy.^{1,2,4}

Majority of patients with Asymmetrical gonadal dysgenesis have testicular tissue on one side and a streak gonad or streak testis on other side. Some authors reserve the term MGD for patients who have 45X/46XY karyotype with testis on one side and streak ovary on the other side.^{5,6,7} Other apply the term to all patients with varying degree of AGD with testicular differentiation on either side, bilateral streak testis or bilateral dysgenetic testis and karyotyping will be done in female phenotype patient or for research purpose. In our case karyotyping is not done due, due to loss of follow up. However the presence of testis which in-turn indicates presence of y chromosome. Latter identification is must since it influences the development of gonadoblastomas in later age.

Persistent Mullerian duct syndrome is excluded base upon macroscopic and microscopic findings. True hermaphroditism was the closest differential diagnosis and was excluded by the strict histological criteria necessary for diagnosis i, e absence of seminiferous tubules with immature sertoli cells and few primitive germs cell and ovarian stroma comprised of numerous primordial or mature follicles containing primary oocytes adjacent to the fallopian tube. It is important to differentiate the two condition because it is necessary for gender assignment and in AGD 30% patients develop germ cell tumors like dysgerminoma, yolksac tumor, Embryonal carcinoma and hence gonadectomy is necessary in such cases.^{7,8} The diagnosis of pseudohermaphroditism due to endocrine disorders and true hermaphroditism was excluded on clinical and histological grounds.

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CONCLUSION: Asymmetric gonadal dysgenesis is rare phenomenon and it is important to diagnose this entity in early age because of proper gender assignment and 30% of these cases progress to Gonadoblastomas. The very rare nature of this entity and medical significance merits its reporting.

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FINANCIAL OR OTHER

COMPETING INTERESTS: None

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Date of Submission: 02/03/2015.
Date of Peer Review: 04/03/2015.
Date of Acceptance: 18/03/2015.
Date of Publishing: 30/03/2015.