ISSN: 0975-8585

Research Journal of Pharmaceutical, Biological and Chemical Sciences

Endometriosis Stage: IV Associated with a Bicornuate Uterus: A Rare Case Report.

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ABSTRACT

Endometriosis stage – IV involving a bicornuate uterus is a very exceptional condition to occur. It is an estrogen dependent chronic condition which is mostly affecting the females those who are in the reproductive phase of their life. This disease can be having both symptomatic as well as asymptomatic effects. Thus the pathogenesis of this disease is very convoluted to recognize. Mostly many cases are been reported with symptoms itself. But here in our case we are representing a unique case of a 21 year old female with stage – IV endometriosis along with a bicornuate uterus with asymptomatic effects. Endometriosis has a well-established genetic predisposition. Thus in our case we are using some of the classic cytogenetic tools to find out if there are any kind of chromosomal anomalies are present in the patient. According to the cytogenetic analysis the result of the chromosomal anomalies we got is a somatic chromosomal mutation at chromosome 17q which was the main causative agent in inculcating this disease to the patient. Hence the study of genetic as a distinct dimensional quantitative assay is likely to prove an important strategy for future researches in the field of endometriosis disease.

Keywords: endometriosis, bicornuate uterus,

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ISSN: 0975-8585

INTRODUCTION

Endometriosis is a gynaecological conditions were the endometrium (which makes the inside surface of the uterus) grows outside the uterus cavity or the ovary which is also called as ectopic or extra-gonadal endometriosis [1, 3]. It was first acknowledged by Baron Carl von Rokitansky in 1860. The most suited site for the occurrence of this disease is in the pelvis but there are reports that it can occur in some of the other places such as on the ovaries, uterus, fallopian tubes, uterosacral ligaments, broad ligaments, round ligaments, cul-de-sac or ovarian fossa, as well as on the appendix, large bowel, ureters, bladder, or rectovaginal septum [1]. Endometriosis is affecting roughly around 5-10% of the women who are in the reproductive phase as it is an estrogen-dependent chronic inflammatory disease and may also reach up to 40 to 60% women among with dysmenorrhoea [1, 2, 5]. The incidence of endometriosis is also been observed in infertile women almost among 20 to 25% [5]. The episode of this disease is mainly due to a process known as retrograde menstruation [2]. Retrograde menstruation happens when menstrual blood flows back through the fallopian tubes into the pelvic cavity instead of the body. The symptoms linked with this disease are dysmenorrhea – painful, sometimes disabling cramps during the menstrual period pain may get worse over time (progressive pain), also lower back pains linked to the pelvis; chronic pelvic pain – typically accompanied by lower back pain or abdominal pain; dyspareunia – painful sex; dysuria – urinary urgency, frequency, and sometimes painful voiding; infertility and fatigue [1,2]. But many a times it could also be asymptomatic or could be recognized during a laparoscopy or exploratory surgery [1]. Endometriosis is been differentiated into four stages by the American Society of Reproductive Medicine such as stage I – minimal; stage II – mild; stage III – moderate and stage IV – severe. The susceptibility of the endometriosis disease is usually directed by genetic factors. Only one or two cases with this disease are an act of isolation or interact either with ubiquitous non-genetic factors or an exogenous factor. Thus in our case study we are going to report on an out of the order type of stage 4 endometriosis female along with a bicornuate uterus and for furthermore analysis we went ahead with the genetic studies which finally revealed that the female had an 17q somatic chromosomal mutations which was an evidence for causing of the disease.

Case Report

A 21 year old woman married with nonconsanguineous for 6 months visited our department with chief complaints of abdominal pain for past 15 days of duration. The pain was dull and of mild in nature. Her past medical as well as family histories were normal. She got menarche at 13 years old with regular menstrual periods with few episodes of menstrual pain and occasional constipation. She did not give a history of cyclical pelvic uneasiness or major dysmenorrhoea. She did not go for any remedial for infertility in the past. There were no histories of her for a hysterotomy or any other pelvic surgery, including laparoscopy, in the past.

Her CBC and other blood parameters were normal. Even her test for TB that is the mantoux and PCR also showed the negative results. Henceforth with our suggestions the woman was been asked to do all sort of local examinations such as ultrasound scan, MRI scan, hysteroscopy and laproscopy. The results of the ultrasound scan showed a cystic tubular mass of about 3*5 cms in the right fornix with non-tenderness and hydrosalpinx in the right side (Image 1,2,3). MRI scan also confirmed the same and also showed a bicornuate uterus with a rudimentary right horn, right hydrosalpinx and a chocolate cyst. Further the diagnostic hysterolaparoscopy and chromopertubation showed the following results:-

- Bicornuate uterus,
- Left ovary and tube were normal,
- Right tube was buried in the adhesions and only fimbrial end was seen,
- Right ovary was enlarged enormously with huge endometriotic cyst,
- Another cyst was also present in the pod about 4*5 cms which was completely obliterated,
- All over the right horn and adnexa an endometriotic vesicles was observed,
- Right tubal ostia was not been visualised.

Hence according to the above mentioned scanning reports it was been proved that the women was suffering from Stage – IV type of endometriosis.

ISSN: 0975-8585

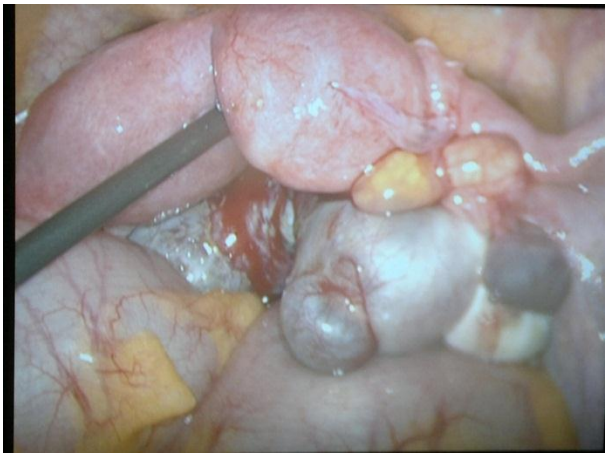

Image 1

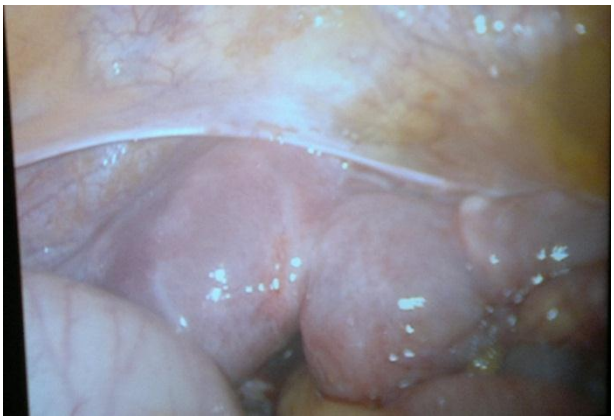

Image 2

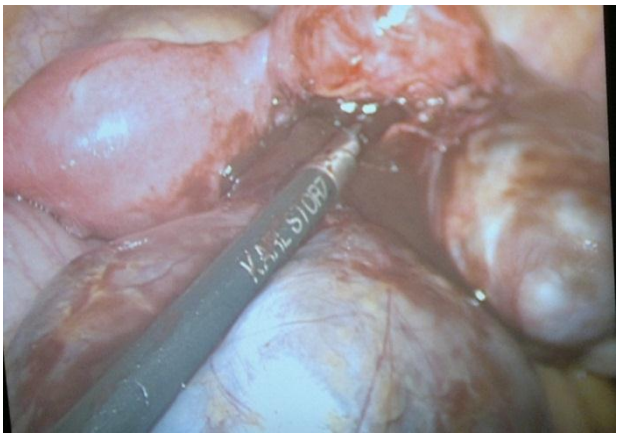

Image 3

After all these scanning report we collected 5ml of peripheral blood from the woman to check for any kind of chromosomal anomalies were present or not. After which the karyotyping and fluorescences in situ hybridisation (FISH) was been done. The result of the following karyotyping and FISH showed that there was a somatic chromosomal mutation at the chromosome 17q. Thus according to these findings it shows that there might be some kind of hereditary influence in inculcating this disease to the patient.

Finally the contents of the chocolate cyst were drained out by using a small gentle insertion using the dissection method. After the dissecting surgery the patient was been monitored for 3-4 hours and was been asked for a regular monthly checkups. After a month she was been prescribed with three cycles of Gnrh

analogues was given to relieve pain, slow down the growth of endometriosis tissue, improve fertility and to prevent the disease returning. Following this after 4months she had a successful spontaneous conception which was been confirmed by UPT examination.

CONCLUSION

Endometriosis is a sporadic disease which has been caused in the reproductive organs of the female. The pathogenesis of this disease is very impulsive and complicated to discover. But there are two most accepted theories for this diseases pathogenesis and they are the first theory is “hypothesis of migration – or the retrograde menstruation” while the second theory is the “induction theory – coelomic metaplasia” [3]. The second theory is based on the interpretation that the coelomic pluripotent cells can get differentiated into both endometrial as well as the peritoneal cells [3].

In the finale of this case report we are representing a case about a female with stage – IV endometriosis with a bicornuate uterus without any symptoms. As there were not even a single medical or symptomatic history with the patient for the causing of this disease we went for a cytogenetic genetic study to investigate further for the pathogenesis of the disease. Thus as the result of our cytogenetic studies we finally got into the conclusion that the female was having a somatic chromosomal mutations of the chromosome 17q. Hence the causative agent for the disease with a stage – IV endometriosis including a bicornuate uterus was this somatic chromosomal mutation.

Endometriosis is a very rare case to be identified in the earlier stage itself. And the consequence of the late stage identification could lead to either colorectal cancer or ovarian cancer. Thus the clinicians as well as the patients must have regular checkups as soon as their menstruations cycle starts to overcome with the verse situations in the future as the clear pathogenesis or the causative agent of this disease is not yet clearly identified.

ACKNOWLEDGMENT

Author Divya James Fenn would like to extend my gratitude to my institution, Sree Balaji Medical College and Hospital and sincere thanks to my Head of the Department Dr.K.Saraswathi, Dr.Aravind, Department of paediatrics, Sreebalaji Medical college and hospital for the guidance given**.** Author D.J.F thank Dr.V.Balachandar for assisting the manuscript preparation.

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