

## Case Report

# Congenital Absence of Cervix with Atrial Septal Defect In An Adolescent Girl

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## ABSTRACT

**Background**-Cervical agenesis is a rare congenital Mullerian anomaly. Its association with vaginal agenesis is further rare, reported in 39% of cases of cervical agenesis.

**Case Presentation**-Case report of a 15-year-old adolescent girl congenital absence of cervix with atrial septal defect is reported here. Case presented with a history of Primary amenorrhea with cyclical pain in the abdomen for 3-4 months. Investigations were done. The diagnosis was made & was advised for surgical correction.

**KEYWORDS:** agenesis, Mullerian anomaly, Absence of cervix

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## INTRODUCTION

Cervical dysgenesis is a congenital anomaly which is extremely rare. According to the American Society of Reproductive Medicine (formerly American Fertility Society), cervical agenesis is classified as type Ib Mullerian anomaly [1]. In 2013, European Society of Human Reproduction and Embryology (ESHRE) has proposed a newer classification system for Mullerian anomalies on the basis of uterine anatomy. In addition, anomalies of the cervix and vagina are classified separately. In the ESHRE classification, cervical and vaginal agenesis are classified as C4 and V4, respectively [2].

This anomaly has a mean prevalence of 4.3% of the general population. The incidence of cervical agenesis is about 1 in 80,000 to 100,000 births. The review of literature lists less than 200 reported cases since 1900 [3].

Cervical agenesis manifests itself in the absence of a cervix, the connecting structure between the uterus and vagina. Uterus, fallopian tubes, the cervix, and vagina gets differentiated from the Müllerian ducts. When normal development of the Mullerian ducts is not happening properly, many malformations can occur.

Affected patients of cervical agenesis have a functional uterus. Because of lack of cervix the

patients may develop primary amenorrhoea with cyclic pelvic pain & haematometra. Usually growth and pubertal development is found normal. Cervical or cervicovaginal agenesis usually presents around the age of menarche with complaints of primary amenorrhea and/or cyclical abdominal pain. In these cases, endometrium functions normally, but menstrual blood has no path to exit from vagina, leading to hematocolpos followed by hematometra formation. The condition can further aggravated with hematosalpinx and endometriosis if left undiagnosed [4].

Diagnosis can be made in early adolescent period (12–17 years) due to cyclic abdominal pain, but others cases may be diagnosed later with amenorrhoea. management should be done early in these cases to avoid progression of the disease.

Appropriate & early diagnosis of cervical agenesis, noting for any other associated congenital anomalies, psychosocial counseling for patients are considered as the most vital steps in the effective management of müllerian agenesis.

The first line of therapy after diagnosis typically involves the administration of the combined oral contraceptive pill, medroxy progesterone acetate or a gonadotropin-releasing hormone agonist to suppress menstruation and thereby relieve pain.

A number of surgical approaches are in practice to correct the deformity including vaginal elongation, creation of a neovagina etc. The goals of reconstructive surgery for cervical malformations are to provide a conduit for menstruation, to relieve pain and preserve reproductive potential.

### **CASE REPORT**

A 15-year-old girl, presented to Gynecology OPD in January with complaints of cyclical lower abdominal pain for last 3-4 months. The pain is described as tearing type, radiating to the flanks not relieved with rest or analgesics, and was associated with nausea and vomiting. Each monthly episode lasts for 3-4 days. The patient's history evidenced primary amenorrhea. Medical and surgical history was normal,

except for the presence of left-eye microphthalmia & generalized folliculitis. Microphthalmia has been present since birth and is not associated with vision loss. Pustular lesions of mixed type-(active and healing) with hyperpigmented spots were noticed on both arms and legs, suggestive of folliculitis. Vital parameters and routine blood investigations were normal except urine microscopy findings with many pus cells indicating urinary tract infection. BMI was only 16.66.

Physical examination revealed normal breast development (Tanner III) and secondary sexual characteristics. Axillary hair and pubic hair also were well developed (Tanner III). External genitalia appeared normal with well-differentiated labia majora and minora. Examination revealed a vaginal length of approximately 3cm with non tender bilateral fornices. Cervix was not felt. Per abdominal assessment suggested an enlarged bulky uterus that corresponds to a fundal height of 6-8 weeks. Guarding and rigidity were also observed.

An ultrasonography & MRI was done for diagnostic purposes. MRI plain of the pelvis was advised and was performed. The MRI findings correlated with the sonographic findings. MRI findings suggested haematometra with non-visualization of normal cervical stroma with presence of cystic lesions in regions of cervix, suggestive of congenital absence of cervix.

Diagnosis of the congenital absence of cervix with hematometra was confirmed, and the parents of the girl were counseled about various management options, including the possibilities of reconstruction, risks of failure, etc. Cervical and vaginal reconstruction surgery was recommended. McIntoe's procedure for vaginal repair was the surgical intervention advised. An anesthetic and cardiac opinion was sought before undertaking the surgical intervention. An ejection systolic murmur of grade III was heard in pulmonic areas on auscultation. She was not having any dyspnea, orthopnea, or other associated difficulties. 2D ECHO was advised. ECHO revealed a large Ostium Secundum Atrial Septal Defect with left to right shunt. Mild PAH



**Fig. 1:** Trans abdominal pelvic ultrasound revealed a mildly bulky uterus that measured 10.7 cm × 3.8cm × 2 cm with a volume of 113.2cc. Moderate to gross amount of fluid collection was noted within the endometrial cavity and endometrial canal with diffuse homogeneous internal echoses, suggestive of hemato metro colpos, likely due to a imperforate hymen.

was also observed. Right atrium & Right ventricle appeared dilated with normal left ventricular function. LVEF was 77%.Surgical closure of atrial septal defect was advised for her and was started on with T.Metoprolol 12.5 mg daily once. Parents were informed about the cardiac defect ruled out during investigations. They were advised to get the Atrial Septal Defect closure first and then followed by Mcintoe procedure for vaginal repair. Symptomatic treatment were given to the girl during the time of hospitalization, which included pain relief with Inj.Tramadol , management of nausea and vomiting with Inj. Odandesetron,treatment of urinary tract infections with T. Nitrofurantonin 100 mg BD for 10 days.

## DISCUSSION

Patients with cervical agenesis typically present in early adolescence, around the time of menarche, with amenorrhea and cyclic pelvic pain caused by the obstruction of menstrual flow from the uterus. Obstructive uterine anomalies occlude the normal menstrual flow, resulting in primary amenorrhea.

Any abdominal or pelvic pain in a pubescent girl must evoke the suspicion of possible obstructive genital anomaly. If untreated, the accumulation of menstrual fluid in the uterus caused by cervical agenesis can lead to hematocolpos, hematosalpinx, endometriosis, endometrioma & pelvic adhesions.

In the current case also, the presenting features were the same - amenorrhea & cyclical abdominal pain. The girl presented to the health care in adolescent age period -at 15 years of age Stagnation of menstrual fluid due to imperforate hymen lead to heamatometro colpos in this scenario. The diagnosis of cervical agenesis can be made by magnetic resonance imaging, which is used to determine the presence or absence of a cervix. MRI & USG were the diagnostic modalities used in this case, too.

Evaluation for associated congenital anomalies is essential, because up to 53% of patients with müllerian agenesis have concomitant congenital malformations. Variety of uterine anomalies, including müllerian agenesis, can

be seen with VATER/VACTERL association (vertebral anomalies, anorectal malformations, cardiovascular anomalies, tracheoesophageal fistula, esophageal atresia, renal anomalies, limb defects). In this case, a cardiovascular anomaly -A large ostium secundum atrial septal defect was identified on examination that demanded surgical closure. Left eye microphthalmia -another congenital anomaly in which eyes are abnormally small and have anatomic malformations were also present in this case too.

McIndoe procedure-procedure of performing a careful dissection between the bladder and rectum, thus producing a cavity which will be formed by inserting a vaginal mold covered with a skin graft was the recommended surgery. The psychologic effect of the diagnosis of müllerian agenesis should not be underestimated. Many patients experience anxiety and depression, question their female identity, and grieve in their infertility.

## CONCLUSION

Cervicovaginal agenesis is a rare congenital anomaly of the female genital tract. Only few scattered cases and case series are reported in the literature. Cervicovaginal agenesis is to be kept in mind whenever a young girl presents with primary amenorrhea and cyclical abdominal pain. Early diagnosis and management are crucial for successful outcome. Exact delineation of pelvic anatomy on imaging is must to avoid complications. The first-line surgical option is cervicovaginoplasty, but management has to be individualized in each patient with keeping the possibility of the need for a hysterectomy. Psychological counseling and long-term follow-up are essential. Considering these aspects, the adolescent girl and family are counseled well, informed in depth about the treatment options available, and reassured.

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## Author Contributions

**Jissa Donel:** Manuscript drafting and correspondence with the Journal,

**Binu Mathew:** Concept and design of the study,

**Bagavathi R:** Literature collection and review

All Author-Co-Author Contributed sufficiently to bring out the quality of the manuscript

## Conflicts of Interests: None

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