Case Report

Cervico-vaginal Aplasia : A Rare Anomaly with Review of Literature

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Cervico-vaginal anomalies are thought to be a rare condition in general Obstetrics and Gynaecology practice. Cervico-vaginal aplasia is notoriously difficult to diagnose. The ability to make an accurate diagnosis prior to surgery offer significant benefits to the patient. The utmost significant of that is proper pre-operative planning and preparation. The use of Magnetic Resonance Imaging (MRI) to diagnose Cervico-vaginal atresia is a novel modality in the medical literature and it provides the advantage of diagnosis non invasively prior to surgical intervention. Here, we are reporting a rare case of Cervico-vaginal atresia who clinically presented with primary amenorrhoea and cyclical lower abdominal pain.

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Key words **: Cervico-vaginal Aplasia, MRI.**

Uterovaginal anomalies are linked to a lower rate of fertility and a variety of obstetric complications. Cervico-vaginal agenesis is a common type of such anomaly. Cervico-vaginal agenesis is an exceptionally rare congenital anomaly occurring in about 1 in 80,000-100,000 births. It can be in isolated form or combined with partial or complete vaginal agenesis[1]. Only about 200 cases have been reported since 1900, according to a review of the literature[2]. Patients with cervical agenesis mainly present with complaints of primary amenorrhea and menstrual blood retention, which initiates the symptom of cyclic low abdominal pain without menstrual flow, prompting the patient to seek gynecological evaluation and care[3] MRI can be analysed in a systematic way to allow for prompt and appropriate treatment. Associated pelvic lesions or urogenital anomalies should also be reported if noted. MRI makes it possible to diagnose obstructive causes of uterovaginal anomalies in which determining the site of obstruction is important for planning the proper surgical approach[4]. Here we present a case of Cervico-vaginal aplasia . Our case was diagnosed using transabdominal ultrasound and confirmed on MRI.

Editor's Comment :

- Cervico-Vaginal aplasia is an exceptionally rare congenital anomaly with only 200 cases reported since 1900. It is linked to a high rate of infertility and obstetric complications. Magnetic Resonance Imaging is novel, non invasive, gold standard modality for diagnosis and pre-operative planning.

Case Report

A female aged 16-year-old presented with cyclical lower abdominal pain with primary amenorrhea. Her secondary sexual characters were normally developed. She had no associated genitourinary or digestive complaints and she did not give a history of any familial or hormonal disorder. On physical examination, labia majora and minora were well developed but no separate vaginal opening was noted, only vaginal dimpling was found. On per rectal examination, uterus was palpated and appeared normal in size. An ill defined cystic mass was palpated in the left adnexa however, the right adnexa was free. Her other lab investigations were normal.

Transabdominal ultrasound showed a small conical structure in the region of cervix, however no vaginal lumen was noted. Uterus showed presence of thick dense collection with multiple moving echoes in the endometrial cavity causing thinning of the myometrium in the lower uterine segment. Both the ovaries were normal in size with the left ovary showing presence of a hemorrhagic follicle within. Adnexa showed presence of cystically dilated fallopian tubes on left side with collection containing multiple moving echoes showing layering within. Moderate loculated fluid collection was noted in the adnexa. Both the kidneys were scanned and no renal anomalies were found.

MRI was done which showed a thin fibrous cord like structure in the region of cervix and vagina with no lumen within it. A collection, which was hyperintense on T1WI and hypointense on T2WI, showing levelling within was noted in the endometrium and in the fallopian tubes on left side suggestive of hematometra and hematosalpinx. However, right fallopian tube was normal. Moderate free fluid was present in both adnexa.

Hysterectomy was performed which confirmed the diagnosis of cervicovaginal aplasia with hematometra. Hematosalpinx was demonstrated on left side, however the right Fallopian tube was normal (Figs 1&2).

Discussion

Primary amenorrhea is caused by obstructive uterine

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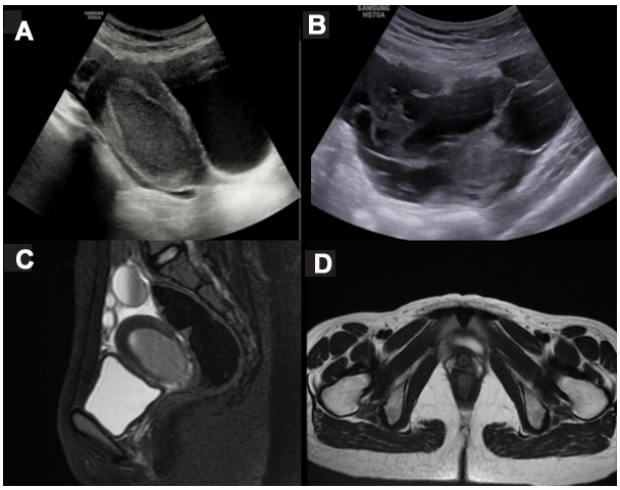

Fig 1 — Transabdominal Ultrasound (A, B) and MRI Pelvis (C, D) demonstrating Cervico-vaginal aplasia with hematocolpos

anomalies that obstruct normal menstrual flow. Any pain in the abdomen or pelvic region in a adolescent girl should raise the possibility of an obstructive cause of genital anomaly. Clinically, it can cause obstructive symptoms such as Hematometra, Hematocolpos and Cyclical lower abdominal pain[5].

The presenting symptom is usually primary amenorrhea which can be seen in a wide range of congenital uterine anomalies, including hypoplastic uterus and imperforate hymen. Ultrasonography is the preferred modality for defining internal genital anatomy and classifying the degree of obstruction or aplasia[6,7]. Despite its rarity, ultrasonography can easily detect the first signs of cervical aplasia or dysgenesis[8].

American Society of Reproductive Medicine, classified cervical aplasia as type Ib Mullerian anomaly[9]. Cervico-vaginal aplasia has been further been classified into various types which are as follows : (i) The cervical body is intact with obstruction at the level of the os of the cervix (ii) The cervical body is atrophic and replaced by a thin fibrous band (iii) Cervical Fragmentation (iv) The midportion of the cervix is hypoplastic with a bulbous tip[10]. Correlation of clinical findings with ultrasound and MRI has been helpful in proper diagnosis and management. So far, MRI has been considered as a gold standard for evaluating the vaginal and cervical anatomy[1].

There are different treatment possibilities available but patients with aplasia are typically unsuitable for canalization and total hysterectomy is the preferred management[11]. Consent of the patient was gained prior to the study and patient identity was not disclosed.

Conclusion

Congenital absence of Cervix and Vagina pose a diagnostic challenge. It is a intricate surgical obstacle that requires a thorough evaluation. Ultrasound evaluation with MRI as the gold standard modality, can diagnose this anomaly and provide pertinent details on cervical and vaginal anatomy. In primary amenorrhea, proper

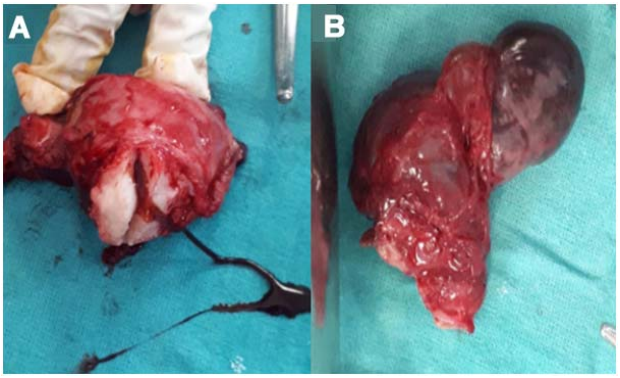

Fig 2 (A and B) — Postoperative image confirming our findings of cervicovaginal aplasia with hematosal

Cervico-vaginal examination is required and the likelihood of Cervical and Vaginal aplasia should always be thought about.

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