

Mullerian Developmental Defect Along with Multisystem Abnormalities: A Rare Case with Rare Association

Indranil Banerjee¹, Purvita Dam², Suhrita De Roy³

¹RMO cum Clinical Tutor, Malda Medical College, Malda, West Bengal, India, ²Associate Professor, Department of Obstetrics and Gynaecology, College of Medicine and Sagar Dutta Hospitals, West Bengal, India, ³Associate Professor, Department of Obstetrics and Gynaecology, Malda Medical College, Malda, West Bengal, India

Abstract

A mullerian abnormality encompasses a wide range of systemic abnormalities which throws a real challenge to the gynecologists for chalking out the appropriate strategy for their diagnosis and management. The most basic classification of mullerian duct defects consists of agenesis and hypoplasia, defects of vertical fusion, and defects of lateral fusion. Mullerian abnormalities are often associated with other systemic abnormalities. In the reported case, we encountered at least four systemic abnormalities in the same patient. The mullerian, gastrointestinal, and cardiological abnormalities were corrected on subsequent intervals. On the follow-up, the patient was seen to be heading towards a healthy life.

Keywords: Mullerian defects, Rectal agenesis, Ventricular septal defect

INTRODUCTION

Mullerian developmental defects are some of the most fascinating disorders that gynecologists encounter. The Mullerian ducts are the primordial analog of the female reproductive tract which differentiates to form the fallopian tubes, cervix, body of the uterus, and the upper part of the vagina. Frequently associated abnormalities are those of renal, cardiac, and axial skeletal system.¹

Embryology

At 6 weeks of development, the male and female genital systems are indistinguishable in appearance, constituting two sets of paired ducts: the paramesonephric (Mullerian) ducts and the mesonephric (Wolffian) ducts. In the absence of the testis-determining factor of the Y chromosome, the mesonephric ducts begin to degenerate and form a matrix for the developing paramesonephric ducts. Synchronously,

the paramesonephric ducts develop bi-directionally along the lateral aspects of the gonads. The proximal segments of the uterovaginal canal, derived from coelomic epithelium, remain unfused and open into the peritoneal cavity to form the fallopian tubes. The distal segments, induced by or derived from the adjacent mesonephric ducts, progress caudomedially and join each other before contacting the posterior aspect of the pelvic urethra at the level of the sinus tubercle. These distal segments of the uterovaginal canal give rise to the uterus and upper 4th-5th of the vagina.

Initially separated by a septum, at 9 weeks the paramesonephric ducts fuse at their inferior margin forming the single lumen of the uterovaginal canal. Regression of the uterine septum has been proposed to be a result of apoptosis, mediated by the Bcl2 gene. Absence of this gene has been implicated in the persistence of the septum. The classic theory of unidirectional regression hypothesizes that the septum regresses from the caudal to cranial aspect of the uterovaginal canal, with the uterus initially bicornuate in configuration. However, an alternative bi-directional theory has been proposed in which it is hypothesized that the process proceeds simultaneously in both the cranial and the caudal directions. This would explain anomalies such as a complete septum with a duplicated cervix or

Access this article online



www.ijss-sn.com

Month of Submission : 01-2015
Month of Peer Review : 02-2015
Month of Acceptance : 02-2015
Month of Publishing : 03-2015

Corresponding Author: Dr. Indranil Banerjee, A-92 Brahmapur More, Near Milan Tirtha Club, Garia, Kolkata - 700 096, West Bengal, India. Phone: +91 9836197019. E-mail: indranil_md@yahoo.com

isolated vertical upper vaginal septum in an otherwise unremarkable uterus.

At week 12, the uterus exhibits its normally developed configuration: A fused external uterine contour of the myometrium and a triangular-shaped endometrium. Because, the fallopian tubes are derived from a different cellular origin than are the uterus and mid- to the upper vagina, they are rarely involved in mullerian duct anomalies.

During the formation of the uterovaginal canal, the sinusal tubercle thickens and forms the sinovaginal bulbs of the primitive urogenital sinus, which gives rise to the lower 20% of the vagina. The uterovaginal canal remains separated from the sinovaginal bulbs by the horizontal vaginal plate. The vaginal plate elongates during the 3rd-5th month, and its interface with the urogenital sinus forms the hymen, which usually ruptures during the perinatal period.

The urinary and genital systems both arise from a common ridge of mesoderm arising along the dorsal body wall, and both rely on normal development of the mesonephric system. The ureters, renal calices, and collecting tubules are formed from the ureteral bud, which arises from the mesonephric ducts, which also induce formation of the kidneys. Hence, abnormal differentiation of the mesonephric and paramesonephric ducts may also be associated with anomalies of the kidneys. Renal agenesis is the most common associated anomaly, although crossed renal ectopia, cystic renal dysplasia, and duplicated collecting systems have all been described.

The ovaries arise from the mesenchyme and epithelium of the gonadal ridge and are not influenced by the formation of the mesonephric or paramesonephric ducts. The undifferentiated gonads are induced to develop by primordial germ cells that migrate from the yolk sac to the dorsal mesenchyme at 5 weeks. These germ cells induce cells of the mesonephros to form genital ridges, which in turn form primitive sex cords. If germ cells do not develop in the region of the gonads, the gonads do not form. Hence, ovarian development is a separate process from the formation of the uterovaginal canal and is not usually associated with mullerian duct anomalies.²

CASE REPORT

A 19-year-old girl came to the outpatient department on 20/1/2009 with the chief complaints of chronic lower abdominal pain for 3-4 years. Her pain was mainly located on the right hypochondrium, progressive in nature and had

exacerbations during her menstruation. Her menarche was at the age of 14 years and her menstrual history revealed that her periods were regular with mild to moderate flow and was associated with progressive dysmenorrhea.

Regarding her past history, it was revealed she was previously diagnosed with a high type of rectal atresia which was treated initially by colostomy and finally through the definite modality of posterior sagittal anorectoplasty at the age of 18 months. She also had a history of having a ventricular septal defect which was repaired between 6 and 12 months of her age.

On examination, there was a lower abdominal mass of 14-16 weeks of uterine size felt at the right hypochondrium which was later diagnosed as a chocolate cyst of right ovary. Ultrasonography of whole abdomen also gave the hint of renal agenesis on the right side with along with unicornuate uterus. Intravenous pyelography confirmed the absence of right kidney and ureter.

The patient underwent laparoscopy on 24/1/2009 which finally made the diagnosis of the case as unicornuate uterus with the left uterine horn communicating with the cervix. The right non-communicating horn had a collection inside (hematometra) with a right chocolate cyst. As a definitive management procedure, laparotomy was performed on 1/04/2009 where removal of right-sided non-communicating horn along with right ovary with chocolate cyst was performed. The left horn of the functional uterus with ipsilateral tubes and ovaries were found to be absolutely normal hence left undisturbed. Her post-operative recovery was uneventful and she was discharged on the 7th postoperative day. Her subsequent scans and follow-up were smooth and regular (Figures 1-4).

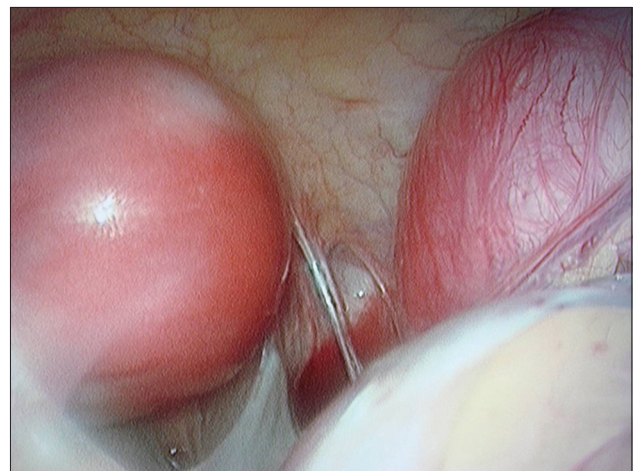


Figure 1: Right side hematometra & chocolate cyst with left side normal functional uterine horn



Figure 2: Scar of initial colostomy (before posterior sagittal anorectoplasty)



Figure 3: Scar of sternotomy (ventricular septal defect repair)

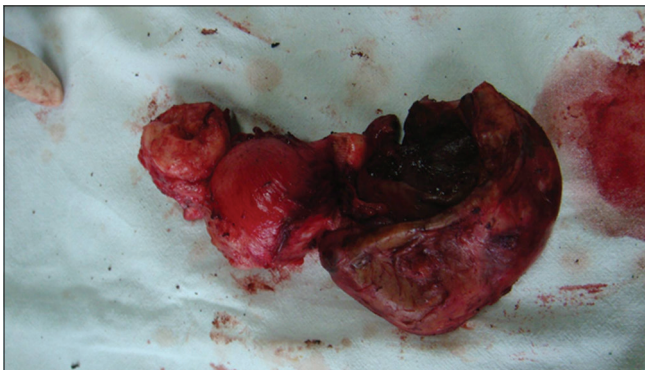


Figure 4: Uterus with cervix along with chocolate cyst

DISCUSSION

Abnormalities in the formation or fusion of the mullerian ducts can result in a variety of anomalies of the uterus and vagina. Close developmental relationship of the mullerian and the wolffian ducts explain the

frequency with which anomalies of the female genital system and urinary tract are associated. Jones and Rock have pointed out that failure of lateral fusion of the mullerian ducts with unilateral obstruction is associated consistently with absence of the kidney on the side of obstruction, which was exactly the similar scenario of the reported case.³ Reports have also described several patients with functioning endometrial tissue in one or both rudimentary uterine bulbs. These patients can develop a large hematometra because of the cyclic accumulation of trapped blood which was also one of the main features of the abovementioned case. Pittock *et al.* reported a substantial incidence of cardiac defects (16%) when reviewing a group of 25 patients with Mullerian abnormalities.⁴ The reported case also had a history of large ventricular septal defect. According to the study done by Oppelt of Germany in 2007 there is an association of rectal atresia with mullerian abnormalities which is about 2%.⁵

CONCLUSION

The uniqueness of the reported case lies in the fact that there was the presence of multiple abnormalities of the cardiac, renal, genital and gastrointestinal system in a single affected person. Most of these anomalies were treated comprehensively. Hence, it is recommended that in every case of Mullerian anomaly, associated cardiac, renal, and gastrointestinal system should be investigated in thorough so that any other associated abnormality do not escape the examiners vision.

REFERENCES

1. Li S, Qayyum A, Coakley FV, Hricak H. Association of renal agenesis and mullerian duct anomalies. *J Comput Assist Tomogr* 2000;24:829-34.
2. Larsen WJ. Development of the urogenital system. In: *Human Embryology*. New York, NY: Churchill Livingstone; 1993. p. 235-79.
3. Fedele L, Bianchi S, Agnoli B, Tozzi L, Vignali M. Urinary tract anomalies associated with unicornuate uterus. *J Urol* 1996;155:847-8.
4. Pittock ST, Babovic-Vuksanovic D, Lteif A. Mayer-Rokitansky-Küster-Hauser anomaly and its associated malformations. *Am J Med Genet A* 2005;135:314-6.
5. Oppelt P, von Have M, Paulsen M, Strissel PL, Strick R, Brucker S, *et al.* Female genital malformations and their associated abnormalities. *Fertil Steril* 2007;87:335-42.

How to cite this article: Banerjee I, Dam P, Roy SD. Mullerian Developmental Defect Along with Multisystem Abnormalities: A Rare Case with Rare Association. *Int J Sci Stud* 2015;2(12):201-203.

Source of Support: Nil, **Conflict of Interest:** None declared.