A ‘never before’ presentation of a common Mullerian abnormality

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INTRODUCTION

Congenital anomalies of the Müllerian duct reflect some of the most intriguing challenges that infertility consultants encounter. They span from agenesis/duplication of uterus and/or vagina to minor uterine cavity abnormalities at the other end of spectrum. Although incidence of Mullerian anomalies in general population is not uncommon, a high transvaginal septum - a vertical fusion defect (different from imperforate hymen) is rarely seen. One such case being investigated for infertility is reported here, the diagnosis of which was unnoticed all along, but once made required simple surgical intervention with excellent outcome.

CASE REPORT

Presented here is an interesting case of a 30 year old lady married for 6 years, cohabiting for the same period and not able to conceive.

On examination she was of average built for her age.

Her menstrual cycles were normal 3/30 days with average flow. She had achieved menarche at the age of 13 years.

There was no history of tuberculosis /thyroid disorder in the past. Her husband was in a white collar job and there was no coital difficulty. There was no history of any genital infection, or spinal trauma in the past. The patient had undergone thorough investigations in a premier tertiary level hospital earlier. Her HSG was done two months prior, which showed uterus to be of normal shape and size. Fallopian tubes could not be visualised, and there was no peritoneal spill present. Diagnosis of B/L tubal block was made. General physical examination and systemic examination were unremarkable. On local exam her vagina was 2” long and vault had 2 small holes at the top, only a needle tip could be negotiated through the left hole, while a uterine sound followed by EB curette could be inserted through the right hole. There was no feeling of resistance of internal os of cervix. Uterine cervical length was 5.5 cm.
Hormone assay was normal, (LH - 1.54 IU/L, FSH - 2.5 IU/L, prolactin - 19.48 µg/L, and TSH - 3.38 mIU/L).

X ray chest was normal.

EB was inadequate; TB PCR on menstrual blood was negative. Ultrasound showed normal sized uterus with ET 7.2 mm and normal ovaries. After visualising uterus on ultrasound on her own the author made another attempt for EB without success.

Patient was taken up for diagnostic laparo-hysteroscopy under general anaesthesia.

At laparoscopy, normal size uterus with normal looking fallopian tubes was visualised. As hysterocope was put through the right hole and we were struggling to find the cervical canal, lo and behold!! A normal cervix was visualised at the farther end. Diagnosis of transverse vaginal septum was made. The vagina was incised from site of hysteroscopy hole and the process of chrome pertubation done. Bilateral tubes were patent.

Retrospectively it was realised that the earlier HSG done, was nothing but vagina ballooned with dye!!! The cervix had never been reached.

HSG, which was wrongly interpreted as normal uterus, in fact only demonstrated distended vagina that had a shape resembling multipara uterine cavity. The point missed out was that a nulliparous uterine cavity is much smaller and narrower (Figure 1).

MRI was also reported to be normal initially but a careful review later revealed presence of septum in upper 1/3 of vagina just caudal to external os (Figure 2).

Once the problem was identified and corrective surgery performed patient was able to conceive in the very first cycle following surgery-end of a six year long wait (Figure 3 & 4).
DISCUSSION

A transvaginal septum (TVS) is a type of rare congenital utero-vaginal anomaly (class II under the Rock and Adam classification). This is a type of vertical fusion defect. A transverse vaginal septum may either be perforate (incomplete) - more common or imperforate (complete) and result from varying degrees of failure in reabsorption of the tissue between the vaginal plate and the caudal aspect of the fused Mullerian ducts. The recanalisation or vacuolation, as this is called is usually complete by 20th week of embryogenesis.

The TVS may occur in isolation or may be associated with other Mullerian abnormalities such as bicornuate uterus (Figure 5).

The condition needs to be differentiated from imperforate hymen. In the adult, a transverse vaginal septum adjacent to the cervix is quite thick. In addition adjacent vaginal segment may be underdeveloped. In contrast Hymen is a membrane separating the vaginal caudal end of the fused paramesonephric ducts from the evaginated urogenital sinus. The hymen may persist if centrally placed epithelial cells do not degenerate. Accumulated menstrual blood may cause the bluish bulge of membranous hymen seen on speculum examination. Imperforate Hymen does not constitute Mullerian anomaly.

The septum may be of complete/incomplete/perforate variety. Usually the septae are about 1 cm thick and located at junction of upper 1/3rd and lower 2/3rd of vagina (46%), Some are located in mid vagina (40%) and few (14%) reported in lower vagina. He more cranially located vaginal septa are relatively thicker and are fibromuscular in nature.

An autosomal recessive gene has been found responsible in some cases of Amish descent. This gene as yet unidentified is reported to occur on chromosome band 20p12.6,7

HSG has no role in diagnosis of transverse vaginal septum. MRI is useful in demonstrating obstructed vagina, exact localisation of the septum and thickness of the septum to plan surgical approach in these cases. Collapse vagina is difficult to visualise and may need to be distended with ultrasound gel or tampon.

The successful pregnancy in surgically corrected height transverse vaginal septum has been reported to be 25% in John Hopkin study. This compares poorly with reported 86% pregnancy success in surgically corrected hymen patients.8

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