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Congenital Absence of the Cervix - Its Place in the Present Classification of Female Genital Tract Malformations.

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ABSTRACT

Background: Aims and Objective: To outline and group cases of cervical agenesis / dysgenesis according to the associated uterine and vaginal abnormalities, as per the proposed ESHRE/ESGE CONUTA classification. **Methods:** The study was conducted in a tertiary care hospital and involved a total of 93 young girls diagnosed as having an absent cervix. Patients with utero-vaginal anomalies alone, with a patent cervix - such as uterine septae, uterus didelphys, bicornuate uterus; or transverse vaginal septae, imperforate hymen and complete or partial vaginal atresia, were excluded. All cases were subjected to specific clinical and local examination. Detailed 2 D Ultrasound was done to study the uterus. Identification of the cervix, its presence or absence, length of the cervical canal, hemato-cervix or hematocolpos was done by trans-abdominal route. All the cases were operated by the same team of surgeons, with the first author as the chief surgeon, to confirm and make a final diagnosis. They were then tabulated as per the CONUTA classification. **Result:** There was absence of the cervix (aplasia / dysplasia) in 93 cases as per the clinical examination, ultrasound, MRI and operative findings. There was complete or partial vaginal atresia (aplasia) in 69 (74.19%) cases and a normal vagina in 22(23.65%) cases. There were 33 cases (35.4%) with a normal uterus, with no cases of T-shaped uterus. There were 29/33 cases (87.89%) without vagina. There was one case with septate uterus and complete cervical and vaginal aplasia. Cases with two Bicornuate uteri with a common aplastic/dysgenetic cervix was seen in 4 cases. In 3 cases the cervix was partially patent in the upper side and obstruction was present only in the lower 4-5mm, causing 'hemato-cervix'. There were 14 cases (15.1%) with unilaterally formed uterine horn, with the non-communicating rudimentary horn. There were 13 cases (13.9%) of cervical aplasia of the unilateral formed uterine horn. In 11 out of 93 cases (11.8%) of cervical aplasia, uterine aplasia / dysplasia and rudimentary horns with cavity were present. **Conclusion:** Our findings underscore the fact the patients with congenital absence of the cervix present a diagnostic challenge and that this entity should be thoroughly evaluated. The new ESHRE/ESGE classification system has the potential to overcome the limitations of the previous classification systems as it provides an effective and comprehensive categorization of almost all the currently known anomalies of the female genital tract.

Keywords: Cervix, Female Genital Tract Malformations.

INTRODUCTION

Congenital malformations of the uterus and vagina are not-uncommon. However, absence of the cervix in the presence of a functioning uterus is rare.[1,2] Menstrual blood collects in the uterus leading to hematometra and hematosalpinx, causing tremendous cyclical pain abdomen incapacitating the young girls. More often than not, there is associated atresia of the vagina – complete or partial, compounding the problem in attempts at construction of a utero-cervico-vaginal canal.[3,4] Due to difficulty in reconstruction and maintainence of patency of the cervix, these unfortunate girls often end up in hysterectomy.

Absence of the Cervix has probably the worst reproductive outcome amongst all uterine malformations; and therefore has great clinical significance for these young teenagers. However, the status of the anomaly of the absence of the cervix, has not been appropriately appreciated in relation to the anomalies of the uterus and vagina as per the American Fertility Society (AFS) 5 and the need for a more inclusive classification was sought.[5,6] The classification proposed recently by the European Society of Human Reproduction and Embryology (ESHRE) and the European Society for Gynaecological Endoscopy (ESGE) common Working Group for the classification of genital tract anomalies - CONgenital UTerine Anomalies (CONUTA) - is relatively more comprehensive.[7-9] However, the main class is Uterine abnormality, and even the sub-classes are further variations of uterine developmental anomalies. It gives only a 'co-

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existent sub-class' status to the cervical or vaginal components.

Anomalies involving cervical absence are not appropriately understood and management is not streamlined. Knowledge of the embryogenesis and its deviation is the foundation of analysis of the clinical presentation, of avoiding pitfalls in making a correct diagnosis, and of institution of appropriate surgical therapy.[10,11] It is necessary to classify the cases from the Gynaecological point of view, where the main pathology is cervical agenesis or dysgenesis, and its association with a normal or anomalous uterus and vagina; so as to be able to counsel and manage the young girls suitably.

Our study of cases with severe dysmenorrhoea and usually with primary ammenorrhoea, due to absence of the cervix as the primary condition, in the presence of a functioning uterus is presented, as per the CONUTA group classification; and perhaps to set the stage for a new classification for 'absence of the uterine cervix'.

Aims and objectives

To outline and group cases of cervical agenesis / dysgenesis according to the associated uterine and vaginal abnormalities, as per the proposed ESHRE/ESGE CONUTA classification.[7-9]

MATERIALS AND METHODS

Study subjects:

A total of 93 young girls diagnosed as having an absent cervix, seen over the last 20 years, since January 1998.

Setting:

Hospital based Tertiary care referral Gynaecology center.

Exclusion criteria:

Patients with utero-vaginal anomalies alone, with a patent cervix - such as uterine septae, uterus didelphys, bicornuate uterus; or transverse vaginal septae, imperforate hymen and complete or partial vaginal atresia, were not included.

All cases were subjected to specific clinical and local examination, if necessary under anesthesia. Rectal examination for hematocolpos which ruled out a cervical problem was done. Detailed 2 D Ultrasound to define the uterus – size and site of the uterine hematometra – whether smaller than 3-4 cms in length and whether away from the midline, presence of hematosalpinx, endometrioma was done. Identification of the cervix, its presence or absence, length of the cervical canal, hemato-cervix or hematocolpos was done usually by trans-abdominal route if the vagina was blind, or the girl was uncooperative; rarely by the vaginal route. Ultrasound of kidneys, echocardiography, skeletal X-ray were done for any associated abnormalities. Karyotype was not routinely done unless there were physical signs / facies suggestive of chromosomal anomalies.

All the cases were operated by the same team of surgeons, with the first author as the chief surgeon, to confirm and make a final diagnosis. They were then tabulated as per the CONUTA classification.

RESULTS

The large number of cases referred to our Institute from all over the populous country and neighbouring countries have helped us in accumulating a large series and grouping them as CONUTA Main Class / Main sub-class 0 – V a/b (Table 1). All the cases were referred due to severe cyclical pain abdomen, often after several futile surgeries tried outside locally.

There was absence of the cervix (aplasia / dysplasia) in 93 cases as per the clinical examination, ultrasound, MRI and operative findings - Co-existent sub-class CONUTA C4 [Figure 1, Table 1]. There was complete or partial vaginal atresia (aplasia) in 69(74.19%) cases, obstructing vaginal septum in 2 cases, and a normal vagina in 22(23.65%) cases - Co-existent sub-class V0/4.

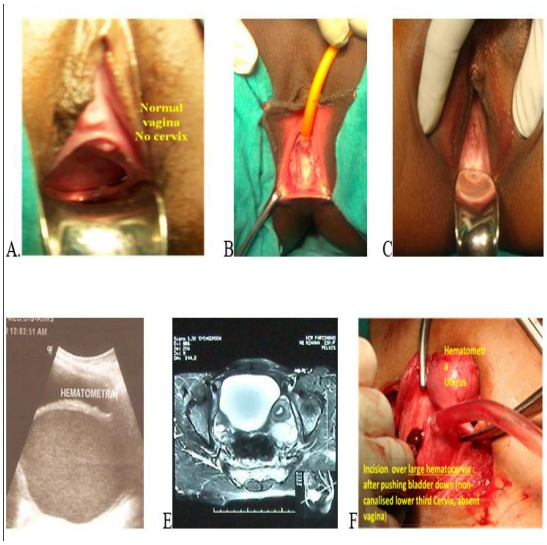

Figure 1: A. Showing a normal vagina but no cervix. B- Blind vagina in complete vaginal aplasia. C-Blind vagina in partial upper vaginal aplasia. D-Ultrasound showing large hematometra. E-MRI showing large hematometra. F- Hematometra, and hematocervix at laparotomy.

Table 1: Showing the 93 cases of Cervical absence, placed in the various Classes / sub-classes / co-existant subclasses of the CONUTA classification.

	Main class	Main sub-class	Co-existent sub-class	No of cases
	Uterine anomaly		**Cervical/vaginal anomaly**	
Class 0	Normal uterus		Cervix C0:Normal C1:Septate C2:Double 'normal' C3:Unilateral aplasia/dysplasia C4: Aplasia/dysplasia Vagina V0: Normal vagina V1: Longitudinal non-obstructing vaginal septum V2: Longitudinal obstructing vaginal septum V3: Transverse vaginal septum/imperforate hymen V4: Vaginal aplasia	Class 0 - 33 cases C4,V0- 4, V4: 29
Class I	Dysmorphic uterus	a. T-shaped b. Infantilis		Class I a – Nil Class Ib-10 cases C4,V0 - 0, V 4- 10
Class II	Septate uterus	a. Partial b. Complete		Class II a – Nil Class IIb – 1 case C4: V0 – 0, V4 – 1
Class III	Dysfused uterus (including dysfused 'septate')	a. Partial (bi-cornuate) b. Complete (didelphys)		Class III a - 4 cases C4:V0 - 1, V4- 3 Class III b – 7 cases C 4, V0 – 5 V4-Longitudinalobs- tructing septum-1 case, Oblique obs-tructing septum -1 Aplasia - 0
Class IV	Unilaterally formed uterus	a. Rudimentary horn with cavity(communicating/not) b. Rudimentary horn without cavity/aplasia (no horn)		Class IV a - 14 cases C4:V0- 7, V4 – 7 Class IV b -13 cases C4: V0 – 5, V4 – 8
Class V	Aplastic/ dysplastic	a. Rudimentary horn with cavity (bi- or unilateral) b. Rudimentary horn without cavity (bi- or unilateral)/aplasia		Class V- 11 cases C4: V0 – 0 V4 - 11
Class VI	Unclassified malformations			Nil

CONUTA Class U0 C4 V0 / V4 cases with a normal uterus (33 cases) were the ones which gave maximum independent status to the absence of the Cervix. There were 29/33 cases (87.89%) without vagina, while only 4 cases had a patent vagina [Figure 2, Table 2].

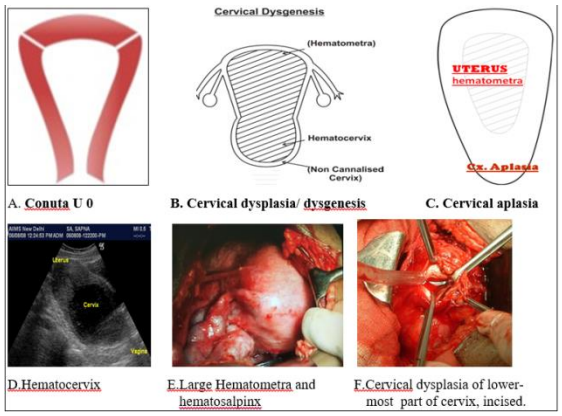

Figure 2: Absent Cervix Aplasia / Dysplasia, with Normal Uterus, with/ without vaginal aplasia, classified as per ESHRE/ ESGE classification consensus - CONUTA Class U0 C4 V0 / V4. A,B,C: Diagramatic pictures of cervical dysplasia, hematocervix, hematometra in a normal sized uterus. Showing large hematometra, and hematocervix at laparotomy.

Table 2: Absent Cervix with Normal Uterus, with/without patent vagina, Cases classified as - CONUTA Class U0 C4 V0 / V4

Class U0 – Normal uterus -33 cases, C4: Cervix – Aplasia / dysplasia

V0: Vagina: Normal - 4 cases, V4: Vagina: Aplasia – 29 cases

Patients with Minimal hematometra due to small size of the uterus - Dymorphic Infantile CONUTA U 1b, C4, V 0/4 - who had severe incapacitating dysmenorrhoea, were seen in 10 girls [Figure 3, Table 3]. There were no cases of T-shaped uterus

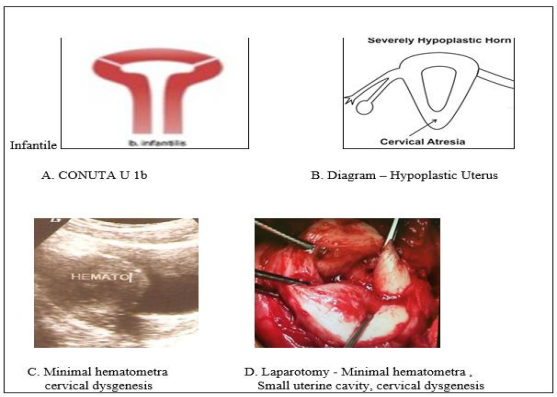

Figure 3: Absent Cervix with/without vaginal aplasia and Dymorphic Infantile Uterus.

Table 3: Absent Cervix with/ without patent vagina, with Infantile Hypoplastic Uterus, with minimal hematometra, Cases classified as - CONUTA Class U1b, C4 V0 / V4
Class U – I b, Dysmorphic uterus, Infantile - 10, C4: Aplasia/d
V0: Vagina: Normal - 0 cases, V4: Vagina: Aplasia - 10 cases

There was one case with septate uterus and complete cervical and vaginal aplasia –CONUTA IIb V0/4 [Figure 4, Table 4]

Figure 4: Absent Cervix with Septate Uterus, with/ without vaginal aplasia - CONUTA U II b , C4 V0 / V4

Table 4: Absent Cervix with Septate Uterus, with vaginal aplasia - Case classified as - CONUTA U IIb, C4 V0 / V4
Class U2 – Septate Uterus - 1, C4: Cervix - Aplasia/dysplasia
V0: Vagina: Normal – 0 cases, V4: Vagina : Aplasia - 1 case.

Cases with two Bicornuate uterii – two formed uterine horns with a common cervix which was aplastic or dysgenetic CONUTA U III a V0/4, was seen in 4 cases. In 3 cases the cervix was partially patent in the upper side and obstruction was present only in the lower 4-5mm, causing 'hemato-cervix' [Figure 5, Table 5].

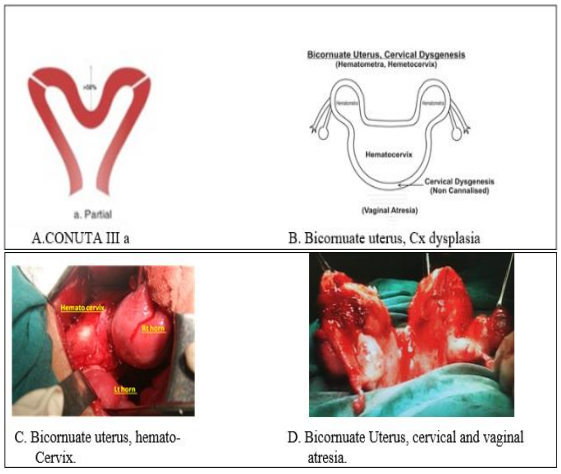

Figure 5: Absent Cervix with/ without vaginal aplasia and Dysfused uterus - Partial (Bicornuate), CONUTA Class U III a, C4, V0 / V4

Table 5: Absent Cervix with Bicornuate Uterus – Cases classified as - CONUTA Class U IIIa, C4 V0 / V4 – 4 cases
Class U III a - 4 cases, C4: Aplasia/dysplasia
V0: Vagina: Normal - 1 cases, V4: Vagina: Aplasia – 3 cases

There were 7 cases of Dysfused uterus complete (didelphys) where the girls had severe dysmenorrhoea - CONUTA IIIb C4, V0 / 4. The cervix on one side was absent leading to one sided hematometra of a fully developed uterine horn : normal vagina, normal menses - 5 cases, amenorrhoea with longitudinal obstructing septum - 1 case, obstructing oblique septum -1 case [Figure 6, Table 6].

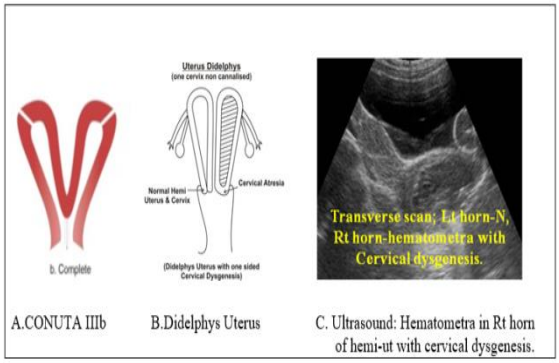

Figure 6: Absent Cervix with Dysfused uterus- Complete (Didelphys) : CONUTA UIII b, C4 V0/4

Table 6: Absent Cervix with Didelphys Uterus- CONUTA UIII b, C4 V0 / V4 -7 cases
C3: Unilateral Aplasia/dysplasia
V0: Vagina: Normal - 5 cases
V4: Vagina: Longitudinal obstructing septum – 1 case, obstructing oblique septum -1 case
Complete Aplasia - 0 cases

There were 14 cases with unilaterally formed uterine horn, with the non-communicating rudimentary horn with cavity having hematometra – CONNUTA U IVa, V0/V. Of these - 7 cases had obstruction to menstrual flow due to absent cervix of the fully formed horn also. The other 7 cases had patent cervix with normal menstrual flow but with severe dysmenorrhoea due to the associated rudimentary horn hematoma [Figure 7, Table 7].

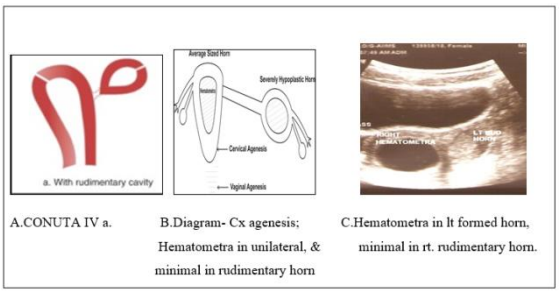

Figure 7: Absent Cervix with Hemi Uterus, Rudimentary horn with cavity- CONUTA U IV a, C4 V0 / V4.

Table 7: Absent Cervix with Unicornuate Unilaterally formed Hemi-Uterus, other Rudimentary horn with cavity : CONUTA Class U IVa, C4 V0 / V4 – 14 cases C4: Aplasia/dysplasia - in rudimentary horn -14, also in formed horn – 7
V0: Vagina: Normal - 7 cases
V4: Vagina: Aplasia – 7 cases

Similar to these cases, there were 13 cases of cervical aplasia of the unilateral formed uterine horn, with no cavity in the other rudimentary uterine horn CONUTA U IVb V0/4. There was associated vaginal aplasia in 8 of these cases [Figure 8, Table 8].

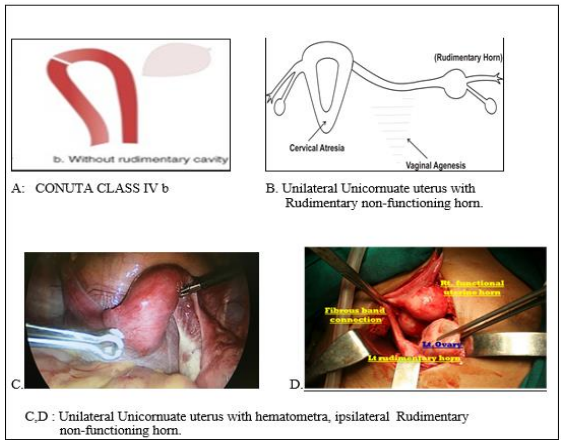

Figure 8: Absent Cervix with Hemi Uterus-Rudimentary horn without cavity -CONUTA U IVb, C4 V0 / V4

Table 8: Absent Cervix with formed Hemi-Uterus, Rudimentary horn without cavity - CONUTA U IV b, C4 V0 / V4 - 13 cases
C4: Aplasia/dysplasia
V0: Vagina: Normal - 5 cases, V4: Vagina : Aplasia – 8 cases

Finally, 11 / 93 cases of cervical aplasia had Uterine Aplasia / dysplasia and Rudimentary horns with cavity – CONNUTA U Va, V0/V. All the cases had hematometra, causing severe pain abdomen [Figure 9, Table 9].

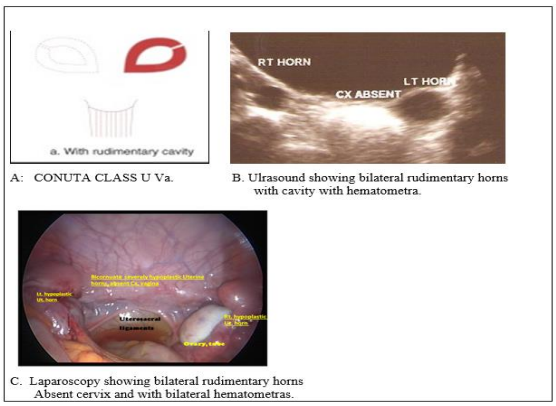

Figure 9: Absent Cervix with Rudimentary horn with cavity (bi-or unilateral) - CONUTA U Va, C4 V0 / V4.

Table 9: Absent Cervix with Rudimentary horn with cavity (bi-or unilateral) – Cases classified as CONUTA U V, C4 V0 / V4
Class U 5/ Aplastic Uterus – 11
C4: Aplasia V0: Vagina : Normal – 0 cases, V4: Vagina : Aplasia - 11 cases

DISCUSSION

Absence of the Cervix has probably the worst reproductive outcome amongst uterine malformations with most ending in hysterectomies; and hence has great clinical relevance for these young teenagers.[12] As our cases described had the main pathology as absence of the cervix, causing hematometra and its consequences, it is necessary to classify cases in a new manner, so as to be able to categorize and manage them appropriately.

The Cervix could be aplastic / agenesis or dysplastic/ dysgenetic. A dysgenetic cervix has some degree of cervical tissue, a band or a cord, or even an intact body which is non-canalised.[13] Patients with cervical aplasia and a functioning midline uterine corpus may have aplasia of the lower two thirds of the vagina with an upper vaginal pouch. On the other hand, some patients may have a considerable atretic segment of vagina and an upper vaginal pouch with a poorly developed uterine cervix and corpus above.[14] It is important to differentiate between these two mullerian anomalies. With cervical dysgenesis, there is no vaginal dilatation from the accumulation of blood, as seen with a high transverse vaginal septum. Imaging modalities like ultrasound and MRI are most helpful when they are correlated with the findings of a careful pelvic examination.[15]

Rock JA et al,[16] conducted a hospital-based study at tertiary-care reproductive endocrine infertility units with the intent to document the anatomical variations of cervical agenesis. A total of thirty women with congenital uterine-cervical anomalies were studied through exploratory laparotomy. All surgical findings were carefully reviewed to understand and determine the anatomic characteristics of the malformed cervix. The authors noted that patients lacked a cervix (cervical agenesis) or had one of three variants of cervical dysgenesis. Patients with cervical dysgenesis were distinguishably characterized as having: a) an intact cervical body with obstruction of the cervical os; b) a cervical body consisting of a fibrous band or cord; or c) cervical fragmentation.

In recent years, there has been a significant upsurge in the understanding and clinically managing congenital cervical agenesis and dysgenesis. One of the most interesting publications on this aspects comes from Grimbizis et al.[17] who published a complete review of the literature wherein reporting of 116 cases of transverse cervical defects since 1900 has been appropriately presented. While

categorizing cervical agenesis and dysgenesis is one aspect, there is not enough clarity of the best possible way to manage this condition. There is a stark lack of carefully designed cohort or randomized trials to provide evidence on a suitable surgical practice. Consequently, the management of uterine cervical anomalies remains murky. Absence of the Cervix has not been clear or much discussed in academic classifications. A clear and efficient classification is essential for planning management. Cases of absence of Cervix- aplasia or dysplasia could not be categorized as per the American Fertility Society (AFS) classification. According to the American Society of Reproductive Medicine (formerly American Fertility Society), cervical agenesis is classified as type Ib Mullerian anomaly. Cervical atresia has been further classified into different types as follows (a) The cervical body is intact with obstruction of the cervical os (b) The cervical body consists of a fibrous band (c) Fragmented portions of the cervix are noted (d) The mid-portion of the cervix is hypoplastic with a bulbous tip.[4,18-20] It is quite interesting to note that the anomalies resulting due to cervical and/or vaginal aplasias and/or dysplasias in the presence of either a normal or deformed but functional uterus, are not represented in the AFS classification system,[5,21] It is important to note that malformations with anatomical characteristics included in more than one category cannot be classified individually and precisely. However, AFS class I, including cases with hypoplasia and/or dysgenesis of the vagina, cervix, uterus and/or adnexae, incorporates severe and complex types of congenital anomalies with serious clinical manifestations usually needing complex surgical treatments.[5,6,22] Such clubbing of different anatomical abnormalities under one category does not help much when it comes to deciding the management. A clear and accurate classification is therefore a prerequisite for treatment of anatomical anomalies, which is not the case with the AFS system.[22]

The new ESHRE / ESGE classification system CONUTA proposed for female genital tract malformations appears to be partially useful in addressing the issue 7-9. Individual anomalies of the Uterus, Cervix and Vagina, and the associated anomalies of more than one have been grouped. The cases of primary Cervical absence have been described as a co-existant sub-class as a secondary add-on to the main class/ subclass of 'Uterine anomaly'.[7,8] Even this has led to a comparatively better understanding of the status of such cases for us. India being a very populous country – cases of the 'rare' cervical agenesis/ dysgenesis are in plenty and referred to our tertiary canter in large numbers. We could not group them earlier, however with the ESHRE/ESGE classification, we have made a decent cataloguing. Very often our patients had been non-appropriately and unsuccessfully operated upon before referral, probably as the understanding of the distorted anatomy was not understood by the referring surgeons.

A. Di Spiezio Sardo,[22] conducted a study to understand how comprehensive is the recently published European Society of Human Reproduction and Embryology (ESHRE)/ European Society for Gynaecological Endoscopy (ESGE) classification system of female genital anomalies. The authors reported single organ anomaly in 12 (30.8%) out of the 39 types of anomalies, while multiple organs and/or segments of Müllerian ducts (complex anomaly) were involved in 27 (69.2%) types. Uterus was the organ most frequently involved (30/39: 76.9%), followed by cervix (26/39: 66.7%) and vagina (23/39: 59%). In our cases, there was complete or partial vaginal atresia (aplasia) in 69(74.19%) cases, obstructing vaginal septum in 2 cases, and a normal vagina in 22(23.65%) cases. Further, normal uterus was present in 33 cases (35.4%) and out of these, in 29 cases (29/33; 87.89%) there was no vagina, while only 4 cases had a patent vagina. An important and noteworthy characteristic of the ESHRE/ESGE classification system is the independent categorization of uterine, cervical and vaginal anomalies.[7-9] All these malformations would have otherwise been labelled as "nondescript" or "inappropriately grouped together" into the same class I of the AFS classification system. However in the ESHRE/ESGE system, these could be properly and correctly classified into subclasses, expressing each one as a specific anatomical deviation.[7-9] Complex anomalies could be easily classified, due to the possibility to describe independently the anomalies of different areas of the genital tract (uterus, cervix and vagina) and combine them case by case. This makes this system simple and functional.

CONCLUSION

Our findings underscore the fact the patients with congenital absence of the cervix present a diagnostic challenge and that this entity should be thoroughly evaluated. The diagnosis can be reliably made by routine ultrasound evaluation and it can provide appropriate information on the cervical anatomy. It is pivotal that an 'ideal' classification system should be comprehensive and able to incorporate all possible variations and offers a clear and distinct description and categorization for them. This will, undoubtedly, facilitate appropriate diagnosis, provide useful cues to the prognosis and would help in efficient planning of the management. The new ESHRE/ESGE classification system has the potential to overcome the limitations of the previous classification systems as it provides an effective and comprehensive categorization of almost all the currently known anomalies of the female genital tract.

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