Congenital malformations of the female genital tract: the need for a new classification system

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Current proposals for classifying female genital anomalies seem to be associated with limitations in effective categorization, creating the need for a new classification system that is as simple as possible, clear and accurate in its definitions, comprehensive, and correlated with patients’ clinical presentation, prognosis, and treatment on an evidence-based foundation. Although creating a new classification system is not an easy task, it is feasible when taking into account the experience gained from applying the existing classification systems, mainly that of the American Fertility Society. (Fertil Steril® 2010; ∞; ∞; ∞. ©2010 by American Society for Reproductive Medicine.)

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Congenital malformations of the female genital tract consist of a group of miscellaneous deviations from normal anatomy. They result from embryologic maldevelopment of the Mullerian or paramesonephric ducts during fetal life: failure of one or more Mullerian ducts to either develop (e.g., aplasia in parts of the system, unicorneate uterus without rudimentary horn) or canalize (e.g., unicorneate uterus with rudimentary horn but without proper cavity), abnormal fusion of the ducts or a lack of duct fusion (e.g., didelphys uterus, bicornuate uterus), and failure to absorb the septum, either totally or partially (e.g., septate uterus, arcuate uterus, obstructing or non-obstructing cervical or vaginal septum) (1–7).

Although certain types of congenital malformations are the result of a clear disturbance in one stage of embryologic development, others are the result of disturbances in more than one stage of normal formation (3). This multistage involvement seems to be the reason for the extremely wide anatomical variations and the large number of combinations observed in congenital malformations of the female genital tract. Importantly, uterine malformations are only a part, although the greater part, of female genital malformations.

Congenital anomalies are very common, but their true prevalence in the general population is not absolutely known mainly owing to methodological bias (6–8). However, the current availability of a wide range of noninvasive diagnostic procedures provides the opportunity to detect anatomical variations in an accurate way. In an older pooled analysis of all studies, the mean prevalence of female congenital malformation in the general population was ~4% (6); in a more recent systematic review that included studies using more accurate diagnostic methods, the mean prevalence in the general population was up to ~7% (7).

Therefore, congenital anomalies represent a more frequent clinical entity than previously expected. The effective appraisal of the anomalies requires a representative classification system.

THE CHARACTERISTICS OF A CLASSIFICATION SYSTEM

Classification systems are extremely useful and necessary for organizing any kind of knowledge, especially medical knowledge. In medicine, categorization enables a better understanding of disease processes as well as more effective diagnosis and treatment. Classification systems are based on the systematic categorization of the patients into groups with similar characteristics. The basic characteristics selected for patient grouping and how they are used in a system are important and create the differences between the systems proposed for a group of diseases. The acceptance of a system indicates its ability to effectively correspond to the needs of the clinicians in understanding, diagnosing, and treating patients.

The ideal classification system should have the following characteristics: [1] clear enough and accurate for diagnosis and differential diagnosis; [2] comprehensive, incorporating all possible anatomical variations; [3] correlated with the clinical presentation and prognosis of the patients; [4] correlated with the treatment of the patients; and [5] as simple as possible.

CURRENT PROPOSALS FOR THE CLASSIFICATION OF CONGENITAL MALFORMATIONS

In 1979, Buttram and Gibbons (9) did the first serious work on the classification of congenital malformations, which was the basis for the American Fertility Society (AFS) classification system, which was published in 1988 (4). The AFS classification system has been widely accepted and is currently the most widely used system. Two other proposals have also been published; in 2004, Acien et al. (2) proposed a new system based on the embryological origin of the anomalies, and 1 year later, Oppelt et al. (10) proposed the vagina, cervix, uterus, adnexae, and associated malformations (VCUAM)
system. Although it is too early for permanent conclusions, these systems have not been widely accepted.

**AFS Classification**

The basis for the AFS classification system is the anatomy of the female genital tract, especially the uterine anatomy (Table 1). This system is simple, user-friendly, and clear enough. It has been successfully adopted as the main classification system for almost two decades. The fact that the vast majority of the female congenital malformations are uterine, which is the basic characteristic for patient grouping, explains the wide acceptability. Also, the classification of congenital malformations according to the degree of uterine deformity seems to correlate well with patient prognoses, mainly the impact on pregnancy outcome, which is another notable parameter for explaining the system’s acceptance.

However, as time passes, many problems seem to be associated with the use of the AFS system:

1. Many clinicians are faced with congenital anomalies that are not included in the main categories or subcategories of the AFS classification system, such as bicervical septate uterus with or without vaginal septum, didelphys with obstructing vaginal septum, and bicornuate with cervical/vaginal aplasia (2, 3, 11–35).

2. There has been extensive discussion as to whether arcuate uterus needs to be a separate class in the classification system or should be included with septate uterus (6, 36).

3. Class I includes cases with hypoplasia and/or dysgenesis of the vagina, cervix, uterus, and/or adnexae; the grouping of these patients into one category is very general and not functional, and it seems to be an effort to avoid a problem rather than to solve it. Although these types of anomalies represent an extremely low percentage of the total number of malformations (37), they incorporate severe anomalies (in terms of deviation from the normal) and complex (more than one organ) types of congenital anomalies; they are associated with very serious clinical manifestations, and they usually need complex surgical treatments (18, 38–40). A clear and accurate classification is a prerequisite for their treatment. Furthermore, the two proposals for a different classification system (2, 10), as well as some subdivisions proposed for certain categories of genital malformations (40, 32), are derived from the inability of the AFS classification system to effectively classify “complex” anomalies.

4. “Obstructive” anomalies, which are the result of 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 (40, 32). These anomalies are either placed together in the first class or are not clearly distinguished in the other classes (2, 10).

It seems, therefore, that the AFS classification system “could function as a framework for the description of anomalies rather than an exhaustive list of all possible anomaly types.”

**Clinical and Embryological Classification**

The basis for the clinical and embryological classification system (2) is the embryological origin of the different elements of the genital tract (Table 1). This system could lead to a better understanding of the pathogenesis of female genital tract anomalies, which seems to be the main purpose of its inventors. Also, the system may be more effective at classifying the complex anomalies because it is based on their pathogenesis, a hypothesis that needs to be tested. However, this system has an inherent limitation that dramatically reduces its chances of acceptance; there is a radical change on the basis of the classification system from anatomy, which is the basis of the widely accepted AFS classification system, to embryogenesis. Because congenital malformations of the female genital tract are defined as miscellaneous deviations from normal anatomy, it seems more logical to choose anatomy per se as the basis for their classification. It should also be noted that current theory about female genital tract embryogenesis, which seems to be reliable, could undergo many changes in the future as new evidence comes to light. This shift could cause changes in the basis of the classification system and the selected and proposed categories.

Furthermore, patients’ clinical manifestations, prognoses, and treatments seem to be the most important parameters in choosing the basis for a classification system. Because most therapeutic interventions tend to restore normal anatomy, and the clinical problems seem to be associated with anatomical deviations from the norm, anatomy per se seems to be the more functional basis for the classification system. Of course, it is useful to explain and understand the pathogenesis of any genital malformation before undertaking the best corrective or therapeutic approach. Finally, this system is not very simple or friendly to use.

**VCUAM Classification**

The basis of the VCUAM system is the anatomy of the female genital tract (Table 3). Each organ is classified separately, similar to the tumor, node, and metastasis classification system for breast tumors (10). This approach provides the opportunity to have a precise, detailed, and extremely representative manner of classification; each type of anomaly could be described using this system, and the clinician could have an accurate idea of each individual’s genital tract anatomy.

The main disadvantage of this system is that it is not simple or user-friendly. Patients are classified only with the help of the classification system’s tables. “Translating” what a “V5b, C2b, U4b, A0, MR” patient has (a woman with Mayer-Rokitansky-Kuster-Hauser syndrome) is not easily done without the use of the appropriate tables. This system is not practical for everyday use and explains the low acceptance of this idea.

From the design of the VCUAM classification system, the anomalies of each separate organ of the female genital tract have the same independent importance in the classification of each patient, which does not allow their frequency to be taken into account in the patient’s classification. Thus, extremely frequent congenital anomalies, such as uterine septum, have the same importance in the classification as the extremely rare anomalies, such as cervical aplasia. This overestimation of the anatomy seems to not be very functional because frequency is another important parameter in the design of the classification systems.

Another misleading problem of the VCUAM system is that there is a need to discuss the groups for each separate organ from the beginning; according to this system, uterine anomalies are classified into 0, normal; 1a, arcuate; 1b, septate <50%; 1c, septate >50%; 2, bicornuate; 3, hypoplastic; 4a, unilaterally rudimentary or aplastic; 4b, bilaterally rudimentary or aplastic; +, other; and #, unknown (Table 3). As easily, one could understand there is a need to discuss from the beginning the adequacy of the proposed classification to cover all kinds of uterine anomalies (e.g., there is a need for a distinct class of uterus didelphys and unicorneate uterus), the necessity of each separate class, and the definitions of each distinct category.
Thus, although the VCUAM classification system may serve as an exhaustive list of all possible anomalies, it could not easily serve as a functional framework for describing the anomalies.

**THE NEED FOR A NEW CLASSIFICATION SYSTEM**

The need for a new classification system for congenital anomalies of the female genital tract seems obvious. The new system should seriously and critically take into account all of the experience gained from the application of the current classification systems, incorporating their advantages and avoiding their disadvantages and fulfilling, as much as possible, all the criteria for an ideal classification system. This is not an easy task, but it is a necessary task for patient management.

**Clear and Accurate Definitions**

Hysterosalpingography does not really help diagnose a large number of congenital malformations (e.g., all cases of aplasia or cases with obstructing vaginal septum that do not allow approximation of the cervix) or offer an effective differential diagnosis between others (e.g., between septate and bicornuate uterus), but it was the main noninvasive diagnostic procedure during the 1980s (7, 8, 41–47). However, two-dimensional ultrasound using the transabdominal and/or transvaginal route, as well as sonohysterography later, were major noninvasive steps forward in the diagnosis of congenital malformations, and a real improvement was made with the introduction of three-dimensional ultrasound, enabling the physician to estimate both the endometrial cavity and the outer shape of the uterus in a more accurate and patient-friendly way (7, 42, 43–58). More recent advances in the field of computer science and ultrasound offer the opportunity for the three-dimensional computerized ultrasound reconstruction of the uterine cavity, a diagnostic approach that could be undertaken as virtual hysteroscopy (59).

Magnetic resonance imaging began a new era in the noninvasive diagnosis of the uterus. females of reproductive age show three distinct layers in the myometrium on T2-weighted images: hyperintense inner endometrium, surrounded by the hypointense junctional zone (JZ), and the intermediate hypointense outer myometrium (60–69). The JZ is functionally important in reproduction and ontogenetically related to the endometrium (61, 63, 65). Although its diagnostic accuracy in the management of congenital anomalies is not yet fully elucidated, it seems to be an extremely useful tool, especially in cases of obstructive or nonobstructive vaginal and/or cervical aplasia or dysplasia (8, 11, 24, 25, 70–77). All these improvements, together with advances in the field of endoscopy (e.g., office hysteroscopy, hydrohysteroscopy, better imaging in laparoscopy), greatly increase our efficacy for an accurate, clear, and detailed estimation of the anatomy of the female genital system (78–81).

Ideally, the classification system should be very clear in the description of its classes, as well as its possible subclasses, to enable the clinician to efficiently recognize anatomical variations and classify them easily and exactly. Clarity will enable the clinician to develop a diagnostic strategy. The anatomy of the female genital tract should be defined as objectively as possible, and for this reason the examination protocol should be supplemented as needed by any imaging modality that contributes to this goal. Clear definitions of each type may help avoid subjectivity in the criteria used to recognize and classify each anomaly and avoid “transitional” cases (3, 6, 7, 82).

A clear and accurate classification of the congenital malformations allows for the correct assessment of their prevalence and the prevalence of the different types. In addition, the need for standardization is exemplified by the lack of an internationally accepted orientation system for the display of uterine images. In view of the worldwide popularity of ultrasound, an internationally accepted
standard orientation would assist clinicians in their interpretation of the literature.

**Comprehensive Incorporation of All Potential Variations**

An ongoing number of publications have reported a number of new variations of undescribed anomalies with unclear classification: septate bicornuate uterus with or without vaginal septum, didelphys uterus with obstructing vaginal septum, cervical aplasia/dysplasia with functioning uterine corpus, and so on (2, 3, 11–35, 83). These described new variations are probably the result of our increasing ability to more efficiently detect anomalies and identify anatomy in detail using the new diagnostic modalities. The classification system must be able to incorporate the new entities and be open to new possible combinations.

**Correlation with Clinical Presentation and Prognosis**

Ongoing evidence supports the initial perception that uterine anomalies are associated with poor pregnancy outcome (6, 84–90). The pregnancy-related consequences of each type of anomaly may be different (6, 87, 89–92) or not between different groups (e.g., arcuate and septate uterus) or subgroups of the same group (e.g., partial, complete, and bicervical septate uterus) (36, 93, 94). Some of the consequences are related to obstetrical complications that are dangerous to a woman’s life, such as pregnancy in a noncommunicating uterine horn of a unicornuate uterus (91, 95–99).

Women with obstructive congenital anomalies (normal functioning uterus with cervical and/or vaginal aplasia or dysplasia) are usually associated with severe health problems, such as cyclic pain resulting from “trapped menses” and coexisting endometriosis, an inability to establish a sexual life, and reproductive problems achieving a pregnancy, even with assisted reproductive techniques (18, 32, 39, 40, 82, 100–105). The treatment of these patients is urgent in nature, and an inability to restore the continuity of the female genital tract is substituted only by total hysterectomy (18, 82, 83, 100–102, 104, 106–108). This group of women has a completely different clinical presentation and therapeutic requirements than the group of patients with vaginal aplasia/dysplasia and an absent functioning uterus. This anomaly is associated with a lasting negative psychological impact on women. However, the main problems are the inability to establish a sexual life and reproductive problems, which could wait until beginning sexual activity (38, 109–113).

Thus, it is extremely important for the evidence-based prognosis of female genital anomalies to have a new classification system with clear definitions of each group and subgroup. Ideally, the system should incorporate the different categories of congenital malformations in a very distinct way; uterine malformations are usually associated with a different prognosis than that of the cervix and vagina.

**Correlation with Treatment**

Laparoscopy and hysteroscopy markedly increase our therapeutic options (6, 77, 78, 98, 114–121). Furthermore, many new therapeutic options have been proposed for the restoration of vaginal and/or cervical aplasia/dysplasia in the presence or not of a functional uterus (18, 26, 32, 82, 100–104, 106, 113, 122–131). More importantly, the results of their treatment could be evaluated in clinical series or in reviews of case reports or case series (18, 32, 82, 100–103, 106, 113, 122, 125, 128, 129, 132–135).

When planning a clear therapeutic strategy for female genital malformations the following questions should be asked for every category: [1] Is treatment needed? For example, restoring the continuity of the female genital tract is a necessity in cases of obstructing anomalies. [2] Is there a treatment? For example, hysteroscopic septum resection can restore normal uterine anatomy in cases of septate uterus, but restoration of the uterine anatomy is not feasible for unicornuate uterus. And [3] will treatment restore the functional problems related to the anomaly?

In planning our therapeutic strategy, there is an obvious need for an evidence-based patient classification according to the patient’s...
therapeutic needs. The classification system should, therefore, incorporate treatment-related options in its design. This approach will make it feasible to evaluate the safety, efficacy, and effectiveness of the procedure to restore normal anatomy and function.

**Simple and User-friendly**

The system should, ideally, be simple, user-friendly, and functional. To be user-friendly and functional, there should be a direct and obvious association with the anatomy of the female genital system without using complicated tables. As the AFS committee for the classification of congenital anomalies pointed out, the scheme of the classification system should be given in one page. The system should have both complete and simple; it is not necessary to be analytical and extremely detailed. The frequency of the congenital anomalies should also be taken into account.

**CONCLUSION**

Congenital malformations of the female genital system seem to represent a common clinical entity, especially in patients with infertility problems. The most commonly used classification system is that of the AFS, but its appropriateness today has been challenged. There is an urgent need to have a more exact, clear, and accurate definition of the different malformations to facilitate researchers and clinicians in providing correct answers for the prognosis and treatment of the different malformations.

The European Academy of Gynecological Surgery, in collaboration with the European Society of Gynecological Endoscopy, recognizes the scientific and clinical significance of an evidence-based updated classification of female congenital malformations and has established a working group specifically for this issue. To achieve a most broad consensus, all scientific societies working in a field related to this subject will be invited to participate in this project. Using the Delphi procedure, a cost-efficient oversight communication is guaranteed.

The task definition is obvious but not easy; a new classification system should have the practicality of the AFS classification system, but definitions should avoid subjectivity in the criteria used for patient grouping and take into account the modern diagnostic and therapeutic modalities. Anatomically, it should preferably be the basis of an updated classification system taking into account, if possible, embryogenesis. The different types of anomalies should be categorized in a strict manner. Scientific interest has been focused on uterine anomalies rather than on the genital tract in general, and, therefore, this might be taken into account in an updated classification.Completeness, flexibility, simplicity, and friendliness of use should be discussed.

It is planned to use the Delphi procedure (136) to find a consensus among the experts in the field. The Delphi procedure is an affordable and user-friendly communication model, characterized by anonymity, to avoid dominance; and where processes occur in rounds, it allows individuals to change their opinion. Through controlled feedback and statistical group response, the expert group comes to a form of consensus. This review article provides a necessary overview of the existing problem and will be used as the starting point for the working group containing a minimum of 20 practicing clinicians, surgeons, radiologists, ultrasound specialists, geneticists, and researchers interested in uterine embryology. This process is only an intermediate step in the problem solution because the existence of a consensus does not mean that the answer has been found. Each answer needs to be matched to observable events and needs its clinical approval in the field.

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