Sonographic and Hysteroscopic Assessment of Uterine Congenital Malformations: A Retrospective Study

Orestis Tsonis¹*, Fani Gkrozou², Evangelos Dimitriou³, Iordanis Navrozoglou¹ and Minas Paschopoulos¹

¹Department of Obstetrics and Gynaecology, University Hospital of Ioannina, Greece.
²Department of Obstetrics and Gynaecology, University Hospitals of Birmingham, UK.
³MSc in Biostatistics, Department of Hygiene, Epidemiology and Medical Statistics, Medical School, National and Kapodistrian University of Athens, Greece.


Correspondence:
Dr Orestis Tsonis, MD, PhD, Senior Registrar in Obstetrics and Gynaecology, Department of Obstetrics and Gynaecology, University Hospital of Ioannina, Greece, Tel: 00306986620604.

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ABSTRACT

Aim: Congenital uterine malformations are often diagnosed via Transvaginal Sonography (TVS) and/or office hysteroscopy. Few studies address the diagnostic accuracy of both techniques in detecting these abnormalities. The aim of this study is to evaluate sonographic and hysteroscopic findings in women with uterine congenital malformations.

Methods and Materials: A nested retrospective study on 137 medical records of women with congenital malformations undergoing vaginoscopic office hysteroscopic and transvaginal sonographic assessment in the Endoscopic Unit, Department of Gynaecology, University Hospital of Ioannina, Greece was conducted. All women were examined initially via Transvaginal Sonography (TVS) and the sonographic findings were correlated with the hysteroscopic findings. Women with septate or bicornuate uteri confirmed their pathology through laparoscopy, a proposed gold standard technique.

Result: Hysteroscopy was able to detect 136 out of 137 cases of congenital malformations with a false positive bicornuate uterus. Transvaginal sonography (TVS) showed moderate diagnostic accuracy accompanied by positive predictive value (PPV) at 79.79%, negative predictive value (NPV) at 99.17%, sensitivity at 79.79%, and finally specificity at 99.17%. Endometrial thickness assessed by TVS was found to be higher in cases of complete septate and bicornuate uteri. The incidence of congenital malformation within the total population was estimated at 4.56%.

Conclusion: Diagnostic hysteroscopy is a reliable tool in detection uterine congenital malformations compared to two-dimensional sonography. Office hysteroscopy demonstrates high diagnostic accuracy and should replace traditional sonographic assessment in determining congenital uterine malformations.

Clinical significance: Hysteroscopy is superior than TVS in detecting uterine congenital malformations.

Keywords
Office hysteroscopy, Congenital malformations, Diagnosis, Transvaginal ultrasound.

Introduction
Congenital malformations of the female genital tract are a result of failure of fusion of paramesonephric structures originated during the development of Müllerian ducts. The incidence of this condition varies and reaches up to 7% in general population [1,2]. These anomalies cover a wide range from minimal defects to complete absence of vaginal and uterine formation as presented in Mayer-Rokitansky-Küster-Hauser (MRKH), often affecting reproductive and obstetric outcomes [3]. Scientific evidence demonstrates higher incidence of these congenital abnormalities in
women with recurrent miscarriages and fertility issues [2]. Septate uterus remains the most common uterine congenital malformation within this subgroup of women as well as, in general population accompanied by the poorest reproductive outcome [3,4]. Operative hysteroscopy is considered the treatment of choice in cases of septate uterus offering a simple option in an outpatient facility [5].

Scientific evidence dictates appropriate and efficient management of these patients in terms of diagnosis and treatment. The gold standard technique in the diagnosis of uterine congenital malformation is hysteroscopy combined with laparoscopic assessment of the uterus reaching 100% of diagnostic accuracy when methods combined [2]. Office hysteroscopy requires no analgesia nor cervical preparation allowing clinicians to accurately assess the endometrial cavity’s integrity at a single appointment [6-9]. Other imaging techniques such as Transvaginal Sonography, 3D sonography, Saline Infusion Sonography (SIS) and especially, Magnetic Resonance Imaging (MRI) could also provide adequate data, but hysteroscopy permits the direct view of the endometrial cavity and thus, increased diagnostic validity in setting the diagnosis [1].

In this nested retrospective study, a total of 137 cases of hysteroscopically confirmed congenital malformations were recruited. Additionally, all patients were evaluated sonographically and in cases of septate or bicornuate uteri, laparoscopy confirmed the hysteroscopic diagnosis. Diagnostic accuracy of TVS was estimated compared to hysteroscopic results.

Materials and Methods
Subjects
In this study, all records of office hysteroscopies from April 2010 till December 2019 performed at the Endoscopic Unit, Department of Gynaecology, University Hospital of Ioannina were searched. A total of 3000 cases of vaginoscopic office hysteroscopy were retrieved and 137 cases of them identified as confirmed congenital malformations. The unit performs over the years many diagnostic hysteroscopies for common hysteroscopic indications. Sonographic data for these cases was also included. In cases of septate or bicornuate uteri, patients underwent laparoscopy to confirm the diagnosis.

Technique and Equipment
Prior to vaginoscopic office hysteroscopy, all women were assessed by a two-dimensional transvaginal sonography (TVS) by different operators. The equipment used was the rigid Bettocchi hysteroscope 4 mm (Karl Storz), with forward-oblique vision of 30°. The distension medium in use was normal saline. Hysteroscopy performed with the atraumatic technique of vaginoscopic approach. All interventions performed in the proliferative phase of the cycle. The procedure was successful in all selected cases (3000 out of 2675) thus, failure rate was estimated at 0%. All hysteroscopies performed by 6 different hysteroscopists. No analgesia or cervical preparation was administered prior to the intervention. In cases of bicornuate and septate uteri, laparoscopic confirmation was performed based on the local protocol of the Department of Gynaecology, University Hospital of Ioannina, Greece.

Statistical Analysis
The diagnostic results of 2D TVS were compared to hysteroscopic results, which in cases of bicornuate/septate uteri were laparoscopically confirmed. For quantitative variables, data was expressed as mean ± standard deviation (SD). Sensitivity, specificity, positive predictive value (PPV), negative predictive value (NPV) and finally, overall diagnostic accuracy for TVS were calculated. Statistical analysis was performed with the use of SPSS Statistics software (Version 23.0). P-value of <.05 was considered significant.

Results
Main Findings
In 137 out of 3000 patients, congenital malformations were confirmed resulting to an incidence of 4.56%. The mean of the age of those patients were 32.74 years and the 95% confidence interval of the age was 31.48 to 34.00 years of age. Indications for office hysteroscopies were reported. The majority of cases (72 out of 137, 52.55%) had sonographic evidence of congenital malformation, while the rest distributed as follows: IVF screening/infertility issues in 44 cases (32.11%), abnormal uterine bleeding (AUB) in 11 cases (8.02%) and other common hysteroscopic indications in 10 cases (7.29%). In these 3000 cases, 718 patients had infertility issues, thus the estimated prevalence of congenital malformations in this subgroup was estimated at 6.12% (44 out of 718 cases of women suffering from infertility issues). The exact distribution of congenital malformations, after laparoscopic confirmation, was partial septate (n=57 | 41.7%), complete septate (n=29 | 21.2%), unicornuate (n=18 | 13.1%), bicornuate (n=16 | 11.8%), arcuate (n=12 | 8.7%), hypoplasia (n=3 | 2.1%), didelphys (n=1 | 0.7%) and DES drug related (T shaped) (n= 1| 0.7%). The test Pap showed normal finding in 97 patients, abnormal findings (parakeratosis etc.) in 6 patients and infection in 6 patients, and for the rest no data was available (Table 1).

Table 1: Number of uterine congenital malformations diagnosed with MRI, office hysteroscopy and TVS

<table>
<thead>
<tr>
<th>Malformation</th>
<th>Confirmed with Laparoscopy</th>
<th>Office Hysteroscopy</th>
<th>TVS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Unicornuate uterus</td>
<td>-</td>
<td>18</td>
<td>2</td>
</tr>
<tr>
<td>Bicornuate uterus</td>
<td>16</td>
<td>17</td>
<td>12</td>
</tr>
<tr>
<td>Complete Septum</td>
<td>29</td>
<td>28</td>
<td>23</td>
</tr>
<tr>
<td>Partial Septum</td>
<td>57</td>
<td>57</td>
<td>32</td>
</tr>
<tr>
<td>Arcuate uterus</td>
<td>-</td>
<td>12</td>
<td>3</td>
</tr>
<tr>
<td>Hypoplasia</td>
<td>-</td>
<td>3</td>
<td>1</td>
</tr>
<tr>
<td>T-shaped uterus</td>
<td>-</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Didelphys</td>
<td>-</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Success rate</td>
<td>102/102 (100%)</td>
<td>136/137 (99.27%)</td>
<td>72/137 (52.55%)</td>
</tr>
</tbody>
</table>

(TVS: Transvaginal Sonography; -: no laparoscopic confirmation needed)

Sonographic findings in women with congenital malformations
Transvaginal sonography was able to identify congenital abnormalities in 72 out of 137 cases (52.55%). The remaining 55
cases had normal sonographic results. All women sonographically suspected of congenital malformations were examined hysteroscopically. Endometrial thickness was assessed in each patient. Mean endometrial thickness was estimated. The mean of endometrial thickness of the patients with unicornuate and bicornuate uteri was 9.05 mm and 9.4 mm, respectively. Partial and complete uterine septum demonstrated a mean endometrial thickness of 6.45 mm and 10.37 mm, respectively. The mean of endometrial thickness of the patients with arcuate was 7.05 mm (Figure 1).

![Figure 1: Endometrial thickness assessed by Transvaginal Ultrasound in women with confirmed congenital malformation.](image)

**Sonographic accuracy in detecting congenital malformation compared to office hysteroscopy**

Besides congenital malformations, hysteroscopy was able to reveal concurrent pathologies. The hysteroscopic results were normal in 81 out of 137 patients (59.12%), 13 patients were diagnosed with endometrial polyps (9.48%), 24 patients were diagnosed with micropolyps (17.51%), 8 with adhesions and 6 with stromal oedema and hyperaemia attributed to endometritis (4.37%). The histology confirmed endometritis in 93.33% (28 out of 30) cases of micropolyps and hysteroscopic signs suggestive of endometritis. All cases with histologic confirmation of endometritis were diagnosed with septate uterus, partial or complete. Finally, laparoscopy revealed a false positive hysteroscopic diagnosis of bicornuate uterus, instead of complete uterine septum.

The ability of TVS to diagnose congenital malformations was assessed and the diagnostic accuracy of the technique was estimated as follows: positive predictive value (PPV) of TVS was estimated at 79.79%, negative predictive value (NPV) at 99.17%, sensitivity at 79.79%, and finally specificity reached 99.17%.

**Discussion**

Hysteroscopy is minimally invasive technique that provides reliable information about the endocervical canal and uterine cavity [7]. Main limitation of this technique regarding the detection of congenital uterine malformations is that hysteroscopy does not evaluate the external contours or the thickness of the uterine wall and does not differentiate the septate uterus from the bicornuate uteri, since no data regarding uterine contour can be extracted by this diagnostic method [10]. Our study suggests that women with congenital malformations are more likely to undergo an office hysteroscopy due to infertility issues arise by this condition [11,12]. Interestingly, histology confirmed the concurrence of endometritis in cases of partial or complete uterine septum possibly suggesting that infertility issues do not solely attributed to anatomical defects but also, to underlying mechanisms affecting implantation. No available data was able to retrieved and further studies need to be conducted in order to test this hypothesis.

Our study also suggests only moderate accuracy of TVS in detecting congenital uterine malformations. High specificity and NPV suggest that in sonographic traits not compatible for congenital malformations, TVS set the diagnosis accurately, while moderate sensitivity and PPV suggest that diagnosing congenital malformations is only limited via TVS, according to our results. Scientific evidence estimates the sensitivity of two-dimensional ultrasound at approximately 90% to 92% [13,14], although our study reveals only moderate accuracy of the technique. In cases of complete uterine septum or bicornuate uteri, increased endometrial thickness was sonographically noted, reflecting potentially myometrial tissue of which they are consisted. Sonographic diagnosis of congenital malformations based on criteria. For instance, uterine septum had sonographic evidence of fundal contour, convex or flat and depending on the proportion between myometrial or fibre tissue, its appearance fluctuated in echogenicity compared to the myometrium. Some studies suggest that endometrial thickness might diagnose accurately between septate and bicornuate uteri via endometrial thickness, since septate uteri demonstrate higher measures than bicornuate ones [15,16]. Our findings do not confirm the hypothesis and the suitability of TVS in these cases remains controversial.

Compared to MRI, office hysteroscopy offers a reliable, cost-effective and simple tool not only to detect congenital malformation but to treat in cases of partial/complete uterine septum or rarely in cases of T-shaped uterus [5,17]. In contrast to hysteroscopy, 3D ultrasound is a potential alternative approach to MRI, offering high diagnostic accuracy but postponing treatment [2,10]. In our study, MRI was not performed for these cases since the majority of the participants had hysteroscopic indications addressing infertility issues, for which office hysteroscopy is considered a first-line assessment tool. According to scientific evidence, 3-D ultrasonography might facilitate a pre-diagnosis screening test by providing essential data to the hysteroscopist prior to the intervention. Therefore, women diagnosed with uterine malformation that can be treated by means of hysteroscopic resection might be conveniently scheduled for operative hysteroscopy rather than diagnostic laparoscopy. The cost saving of 3-D ultrasonography compared with MRI or diagnostic laparoscopy can also be highlighted [10,18]. In any case, based on our study findings office hysteroscopy is a simple, safe and
effective technique in diagnosing congenital malformations [19].
In cases of septate or bicornuate uteri, authors suggest additional
assessment by 3D ultrasound, as scientific evidence proposes [18].

The main strengths of the study are (a) the large sample of patients,
upon which a valid incidence of congenital abnormalities within
general population can be proposed and (b) that all women
underwent laparoscopy in order to estimate both hysteroscopic
and sonographic diagnostic accuracy. The main limitation is the
retrospective design of this study.

Conclusions
This study has confirmed that combined laparoscopy and
hysteroscopy are valuable tools for the diagnosis and classification
of female genital system anomalies. In addition, hysteroscopy can
offer efficient treatment of some particular uterine malformations,
such as, septate uteri. In our study, only the traditional USS
was used. By comparing its results with hysteroscopy, it is
obvious that hysteroscopy is superior in diagnosis of intrauterine
malformations. According to the literature, the 3-D USS or the
MRI can add to the diagnosis of these conditions in a way very
similar to hysteroscopy. The main limitation of this technique
though, is their inability to offer treatment options in the same time,
proving hysteroscopy superior as a technique in cases of congenital
uterine malformations. It is worth mentioning that in women with
sonographically suspected uterine malformations, hysteroscopic
assessment should follow and set accurately the diagnosis.

Authors’ contribution
OT and FG share first authorship. ED was in charge of the
statistical analysis of the data. MP performed the hysteroscopies
and supervised the project. IN gathered the literature.

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