Müllerian Duct and Related Anomalies in Children and Adolescents

Monica Epelman, MD\textsuperscript{a,*}, David Dinan, MD\textsuperscript{a}, Michael S. Gee, MD, PhD\textsuperscript{b,c}, Sabah Servaes, MD\textsuperscript{d}, Edward Y. Lee, MD, MPH\textsuperscript{e}, Kassa Darge, MD, PhD\textsuperscript{f}

Objective

Mullerian Duct Anomalies:

- Identify the various types of anomalies
- Recognize associated abnormalities
Summary

Muellerian duct anomalies [MDAs]

- pre-natal MD non-development, defective fusion, incomplete septal regression
- not uncommon [6.7%]
- Hx of infertility/miscarriage: 25% MDAs
- Most common:
  1. arcuate uterus
  2. septate & subseptate defects
  3. bicornate uterus
- US primary & MRI full evaluation
- Accurate initial diagnosis for best surgical results
Introduction

EMBRYOLOGY

- Muellerian or paramesonephric ducts
  - fallopian tubes
  - uterus
  - cervix
  - vagina: upper 2/3
    [lower 1/3: urogenital sinus]
Introduction

Normal Anatomy

2-weeks-old

7-year-old

Post pubertal 15-year-old
Introduction

IMAGING

- Ultrasound
  - prenatal: maternal hormone
  - transvaginal 3D US: adult female
  - transabdominal: primary
Introduction

IMAGING

- MR imaging
  - T2 w - oblique coronal and axial [+sagittal]
  - T2 w – 3D + fat saturation
    - uterine fundal contour
    - separation endo- vs myometrium
  - T1 w + fat saturation
    - blood products
  - Gad T1 w + fat saturation
    - incidental findings

Endovaginal contrast:
- US gel
- Detection of vaginal anomalies
Introduction

IMAGING

- Ultrasound/MRI
  - associated anomalies
  - 40% renal anomalies:
    - unilateral agenesis
    - ectopia
    - renal hypoplasia
    - multicystic dysplastic kidney [MCDK]
    - urinary tract dilation
### Introduction

#### Classification

<table>
<thead>
<tr>
<th>Classification</th>
<th>Description</th>
<th>Subcategories</th>
</tr>
</thead>
<tbody>
<tr>
<td>I. Hypoplasia/agenesis</td>
<td>Early developmental failure of the Müllerian ducts</td>
<td>Vaginal (I-A) Combined (I-E)</td>
</tr>
<tr>
<td>II. Unicorneate uterus</td>
<td>Arrested development of one of the two Müllerian ducts</td>
<td>Cervical (I-B) Tubal (I-D)</td>
</tr>
<tr>
<td>III. Uterus didelphys</td>
<td>Complete failure of Müllerian duct fusion</td>
<td>Fundal (I-C)</td>
</tr>
<tr>
<td>IV. Bicorneate uterus</td>
<td>Incomplete or partial fusion of the Müllerian ducts</td>
<td>Rudimentary horn with: Communicating uterine cavity (II-A) Noncommunicating uterine cavity (II-B) No uterine cavity (II-C) No rudimentary horn (II-D)</td>
</tr>
<tr>
<td>V. Septate uterus</td>
<td>Complete or partial failure of resorption of the uterovaginal septum</td>
<td>Complete (IV-A)</td>
</tr>
<tr>
<td>VI. Arcuate uterus</td>
<td>Near complete resorption of the uterovaginal septum</td>
<td>Partial (IV-B)</td>
</tr>
<tr>
<td>VII. Diethyl-stilbestrol (DES) uterus</td>
<td>Related to the use of DES during the late 1940s to 1971</td>
<td>Complete (V-A)</td>
</tr>
</tbody>
</table>

*Endometrial cavity that communicates with the normal side

*Complete type has accompanying cervical duplication

**Table 2. Classification of Müllerian duct anomalies (MDAs) based on the American Society for Reproductive Medicine system**

**Abdom Imaging (2015) 40:192-206**

**DOI: 10.1007/s00261-014-1959-9**
Introduction

CLASSIFICATION

- uterine anomalies
- controversial
- AFS* classification
- best postpubescent females
- exact description!

*American Fetal Society

(AFNS* classification of müllerian anomalies. Redrawn from American Fertility Society, 1988.)
UTERINE Disorders
Hypoplasia/Agenesis

5%-10%

I. Hypoplasia/agenesis

A. Vaginal
B. Cervical
C. Fundal
D. Tubal
E. Combined
Hypoplasia/Agenesis

- **Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome:**
  - Complete agenesis/hypoplasia of uterus, cervix & proximal 2/3 vagina
    - MRI: rudimentary uterus (92%)
  - Normal ovaries [Ectopic]
  - Female phenotype
  - Possible association: renal, ear, or skeletal anomalies
Hypoplasia/Agenesis

MRKH syndrome
Hypoplasia/Agenesis
MRKH syndrome
Unicornuate

20%

II. Unicornuate

A. Communicating  B. Noncommunicating

C. No cavity  D. No horn
Unicornuate

- Rudimentary horn: 65%
  - 70% non-communicating
  - Obstruction – blood products
  - Endometriosis – retrograde menstruation

- Common association with renal anomalies [40%]
  - Renal agenesis ipsilateral to rudimentary/absent horn
Unicornuate non-communicating
Unicornuate non-communicating - LT: hematometrosalpinx
Unicornuate non-communicating

- RT: hemorrhage
- LT: fluid
Didelphys

5%

Normal uterus

III. Didelphys

Diagram showing the difference between a normal uterus and a uterus with Didelphys.
Didelphys

- Non-fusion of the Muellerian ducts
- 2 uterine cavities & 2 cervixes
  - non-communicating
- Diagnostic delay
  - Regular menses from non-obstructed hemiuterus
Didelphys

- Possible association
  - Obstructed hemivagina & ipsilateral renal agenesis

[Herlyn-Werner-Wunderlich syndrome]
Didelphys

- 2 uterine horns
- 2 cervixes
- Fluid filling
- 2 vaginas
Didelphys

- 2 uterine horns
- 2 cervices
- 2 vaginas
- RT vagina fluid-level
- RT obstruction
Didelphys

- 2 uterine horns
- 2 cervices
- 2 vaginas
- RT hydrocolpos
- RT hemivagina obstruction – septum
- RT ectopia
- RT involuted MCDK

RT

DEPARTMENT OF RADIOLOGY, THE CHILDREN’S HOSPITAL OF PHILADELPHIA [CHOP]
Bicornuate

10%
Bicornuate

- Incomplete fusion of the Muellerian ducts
- 2 divergent uterine fused caudally
  - Fundal cleft greater than 1 cm in depth
  - With or without a cervix duplication
Bicornuate complete

- 2 uterine horns
- Deep cleft
- Fluid filling
- Single cervix
- Single vagina
Bicornuate partial

- 2 uterine horns
- Shallow cleft
- Fluid filling
- Single cervix
- Single vagina
Bicornuate partial

- 2 uterine horns
- Shallow cleft
- Fluid filling
- Single cervix
- Single vagina
Bicornuate partial

- 2 uterine horns
- Shallow cleft
- Hematometra
- Vaginal atresia
- Pelvic kidney
Septate

50%

Normal uterus

V. Septate

A. Complete

B. Partial

Uterus septus

Uterus subseptus
Septate

- Failure to resorb of the fibromuscular septum
- Most common uterine anomalies
- MRI: T1 and T2 hypointense
- Diagnosis mostly adulthood
  - Live birth as low as 5%
  - Transvaginal resection
  - Important to differentiate from bicornuate uterus [transabdominal approach]
Septate complete
Arcuate

Class VI: Arcuate uterus
Arcuate

- Indentation at endometrial fundus
  - T2 signal isointense to myometrium
- Near complete resorption of the uterovaginal septum
- Many consider as a normal variant
VAGINAL Disorders
Obstructive Outflow Tract Disorders

- **Hydro[hemato]colpos**: fluid [blood] accumulation in vagina
- **Hydro[hemato]metrocolpos**: fluid [blood] accumulation in uterus and vagina
  - **Persistent urogenital sinus or cloacal dysgenesis**
    - Urogenital sinus: bladder + vagina – single external exit
    - Cloacal dysgenesis: bladder + vagina + rectum – single external opening
- **Causes: vaginal atresia and septa**
  - Imperforate hymen: benign, not an MDA
- **Fluoroscopic or contrast US genitography**
Obstructive Outflow Tract Disorders

- Hematometrocolpos
- Vaginal septum or lower vaginal atresia
Congenital Absence of Vagina

- Isolated or with uterine agenesis/hypoplasia
- Most common cause:
  - Mueller-Rokitansky-Kuester-Hauser syndrome
  - 1:5000 live births
  - Associated syndromes: Turner or Klippel-Feil etc.
Congenital Vaginal Septa
Congenital Vaginal Septa

- Transverse or longitudinal or combined
- Isolated or with other MDAs
- Longitudinal septa:
  - Disordered lateral fusion between lower 2 Muellerian ducts
  - 75% of uterine didelphys
- Transverse septa:
  - Variable thickness
  - Incomplete vertical fusion Muellerian duct and urogenital sinus
Congenital Vaginal Septa

Fig. 3. Transverse vaginal septum locations.

Transverse Vaginal Septum
Vaginal Cysts

- **Congenital cysts:**
  - **Mullerian cysts**
    - Various locations
  - **Gartner cysts**
    - Remnants of Wolffian [mesonephric] ducts
    - Anterolateral vaginal wall above level of inferior-most aspect of pubic symphysis
  - **Bartholin gland cyst**
    - Urogenital sinus origin
    - Caudal and inferior and posterior vaginal wall
Vaginal Cysts

- Gartner duct cyst
Muellerian duct anomalies [MDAs]

- Pre-natal MD non-development, defective fusion, incomplete septal regression
- Not uncommon [6.7%]
- Hx of infertility/miscarriage: 25% MDAs
- Most common:
  1. Arcuate uterus
  2. Septate & subseptate defects
  3. Bicornate uterus
- US primary & MRI full evaluation
- Accurate initial diagnosis for best surgical results
Thank you for your attention!