**W23: Transitioning Care; The Evolving care of women with congenital genitourinary anomalies**

Workshop Chair: Margaret Mueller, United States  
29 August 2018 16:00 - 17:30

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<th>Speakers</th>
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<td>Introduction to Transitioning care for women with Congenital GU anomalies</td>
<td>Margaret Mueller MD</td>
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<td>Surgical management of complex GU anomalies</td>
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<td>Lia Bernardi MD</td>
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<td>Pelvic Floor Disorders in women with GU anomalies</td>
<td>Kimberly Kenton MD, MS</td>
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**Aims of Workshop**
1. Describe common congenital genitourinary (GU) anomalies and their effects in women during different stages of life (adolescence, reproductive years, perimenopause/menopausal years).
2. Describe the medical and surgical management of GU anomalies and the associated sequelae of these interventions.
3. Uncover the gaps in care for women with GU anomalies, and strategies to maintain adequate care in this population.

**Learning Objectives**
1. Define the need and objectives of a “transitions clinic” for women with congenital GU anomalies and identify types of GU anomalies, with which women are diagnosed, which might benefit from a transitions clinic.
2. Illustrate the fertility, pelvic floor and sexual health concerns and needs of women with congenital genitourinary anomalies.
3. Demonstrate integrated and comprehensive management for women with congenital genitourinary anomalies.

**Learning Outcomes**
After attending this course the participant will be able to identify the sequelae of common genitourinary anomalies as well as strategies for managing these chronic conditions.

**Target Audience**
Urogynaecologists; urologists; female pelvic medicine and reconstructive surgery specialists; trainees (residents/fellows).

**Advanced/Basic**
Advanced

**Conditions for Learning**
Learners will be exposed to different speakers who will convey their information through powerpoint presentations.

**Suggested Learning before Workshop Attendance**
MA Hall-Craggs et al./Journal of Pediatric Urology (3013) 9, 27-32  

**Suggested Reading**
R Chan, UROLOGY 84: 1544e1548, 2014  
H Stephany, UROLOGY 85:959e963, 2015  
V Trofimenko, Current Opinion Urology 26 (2016) 357-62

**Margaret Mueller, MD**
This workshop will focus on the evolving care of women with congenital GU anomalies. First, we will highlight the idea of "transitioning care" for these women. Specifically, the transition requires a seamless and integrated "hand-off" between pediatric providers and specialized adult providers well versed in anticipating future needs of these adolescents and young women with respect to the known sequelae- pelvic floor disorders, sexual dysfunction and fertility challenges. Urogynecologists
are well-suited to orchestrate this transition as surgeons and pelvic health specialists. Additionally, sexual health providers, reproductive endocrinologists, physical therapists are crucial to the interdisciplinary management of these young women.

Briefly, we will illustrate both common and uncommon GU anomalies and their associated specific sequelae as well as their initial surgical management. Attention will be paid to the pelvic reconstructive techniques including vaginoplasty.

**Sexual health concerns with GU anomalies**  
Maureen Sheetz APN

Sexual health is of utmost concern to practitioners caring for adolescents and young women with GU anomalies. Often times this is overlooked, and these women are not afforded the screening for and prevention of sexual health disorders. Moreover, women whom have undergone reconstructive procedures as children, face unique challenges with respect to sexual function as adolescents and adults. The timing and approach to reconstructive vaginal surgery (vaginoplasty) is extremely important and an issue of evolving complexity. Finally, issues related to decreased sexual satisfaction relating to pain, embarrassment and fear are present in this population. We will focus on the appropriate evaluation and management of sexual dysfunction in women with GU anomalies.

**Fertility and Reproductive Potential with GU anomalies**  
Lia Bernardi MD

Gentiourinary anomalies can impact the ability of a woman to conceive as well as her capacity to carry a pregnancy to term. In this workshop we will review how genitourinary anomalies can affect fertility and will discuss treatment options aimed at optimizing pregnancy outcomes. Specifically, we will examine the most common uterine malformations including septate, bicornuate, unicorneate and didelphys uterus, discuss their impact on fertility and pregnancy outcomes and explain surgical treatments that may increase likelihood of reproductive success. We will describe how congenital adrenal hyperplasia can make achieving a pregnancy more challenging and will discuss options that can optimize an individual’s ability to successfully conceive. In addition, we will discuss Mayer-Rokitansky-Kuster-Hauser syndrome and explain options for family building in the setting of müllerian agenesis. We will also review the latest outcome data on uterine transplant in this patient population.

**Pelvic Floor Disorders in women with GU anomalies**  
Kimberly Kenton MD, MS

Pelvic floor disorders including prolapse and incontinence are very common in this population. Given the presence of anomalies and the associated surgical corrections, managing PFDs in this population is complex and can be challenging. We will focus on the evaluation and management of prolapse and incontinence in women with different GU anomalies. Specifically, we will examine the challenges of surgical management of prolapse and incontinence within this population, and the anticipated outcomes.
Transitioning care; The evolving care of women with GU anomalies
Maggie Mueller, MD
Urogynecology and Reconstructive Surgery
Northwestern Medicine

Transitioning care

• 22 year old presents to establish care
  – PMH
  • Bladder extrophy
  • GU reconstruction as infant
  • Multiple surgical reconstructions

• What do you do????
  – Fertility/pregnancy
  – Sexual function
  – Urogynecologic concerns

Learning objectives

• Define the need and objectives of a “transitions clinic” for women with congenital GU anomalies and identify types of GU anomalies with which women are diagnosed which might benefit from a transitions clinic
• Illustrate the fertility, pelvic floor and sexual health concerns and needs of women with congenital genitourinary anomalies
• Demonstrate integrated and comprehensive management for women with congenital genitourinary anomalies

Embryology of genitourinary system

• Organs involved in reproduction and forming and voiding urine

Important embryologic concepts

Development of the genitourinary system

Specialists for Congenital Anomalies of the Genitourinary System in Women

- Urogynecologic specialists
- Pediatric surgeons from Ann & Robert H. Lurie Children’s Hospital
- Reproductive endocrinologists
- Plastic and reconstructive surgeons
- Physical therapists
- Psychologists

Important embryologic concepts

Development of the genitourinary system

Embryology of renal system
  – kidney
  – ureter
  – bladder
  – urethra

Embryology of uterus and vagina

Embryology of external genitalia

All derived from intermediate mesenchyme (mesoderm) derived from the dorsal body wall of the embryo
**Embryology**

**Development of the kidney**

- ~5 weeks
- ~8 weeks

**Development of urinary bladder**

- ~5 weeks
- Bladder develops from vesical part of UG sinus
- Allantois and bladder are continuous
- ~12 wks

**Development of genital system**

- Yellow - urogenital sinus
- Pink - mesonephric duct
- Blue - paramesonephric duct

**Development of uterus and upper vagina**

- Urogenital sinus
- Paramesonephric ducts
- Solid tip of PM ducts come into contact with UG Sinus
- Sinovaginal bulbs grow from sinus and produce a vaginal plate
- ~5 months, Fully cannulated vagina
- Proliferation increased the space between uterus and UG sinus

**Common genitourinary anomalies**
GU anomalies

• Anomalies requiring reconstruction of the genital system
  − Vaginal agenesis
  − Vaginal septa

• Anomalies requiring reconstruction of the urinary and genital system
  − Congenital adrenal hyperplasia
  − Bladder Exstrophy
  − Cloacal Exstrophy

Müllerian Anomalies

• Abnormal fusion/migration of Müllerian ducts
  • Absence/atresia of ducts (vaginal atresia, absence of uterus)
  • Aberrations in fusion (septate or didelphys)
  • Arrest of migration of the Müllerian ducts (persistent urogenital sinus)

• Prevalence 2-3%
• Commonly associated with renal anomalies

Non-obstructive anomalies

• Vaginal
  − Longitudinal vaginal septae
  − MRKH

• Uterine
  − Arcuate
  − Bicornuate
  − Septate

Obstructive Müllerian Anomalies

• Vaginal
  − Hymen pose problems as neonate (maternal estrogen stimulation) or at menarche
  − Transverse vaginal septum
    • Typically present as cyclical pain or primary amenorrhea
    • Low, mid-position or high
    • OHVIRA (obstructed hemivagina ipsilateral renal agenesis) can delay diagnosis

• Uterine
  − Non-communicating horns

Vaginal septums

• Failure of vertical vaginal fusion
• Failure of fused müllerian ducts to unite with UG sinus at müllerian tubercle
• Failure of fusion of endodermal vaginal primordium with mesodermal müllerian ducts
**MRI- Very helpful**

**Management of Müllerian anomalies**

- Non-obstructive
  - Indicated for sexual function or fertility
- Obstructive
  - Typically time sensitive
  - Can suppress cycle to provide more time

**Herlyn Werner Wunderlich Syndrome**

CHVRA

- Uterine didelphys
- Obstructed hemivagina
- Ipsilateral renal agenesis

**MRI coronal**

- 2 uterine cavities

**MRI coronal**

- 2 cervixes

**MRI coronal**

- Hematocolpos
Diagnostic algorithm - obstructive mullerian anomalies

Skinner and Colle, JMIGS 2018

- History
- Physical
- Imaging

Excision of vaginal septums

- 2-3 mm thick – entire length of the vagina
- TV septum vs imperforate hymen vs vaginal atresia
  - MRI

Mayer Rokitansky Küster Hauser Syndrome

- Spectrum of Müllerian anomalies in 46XX females
  - Vaginal agenesis
  - Uterovaginal agenesis
- Cessation of müllerian duct development caudal to genitoinguinal inguinal ligaments (round)
- Urinary and skeletal system defects

Creation of Neovagina

- Non Surgical
  - Simple Pressure (Frank)
  - Pressure from bicycle seat (Ingram)
  - Regular intercourse
- Surgical
  - Surgical creation of neovaginal space between rectum and bladder
  - Split thickness graft (Abbe, McIndoe)
  - Allogenic tissue (Amnion)
  - Peritoneum (Davydov)
  - Artificial grafts
  - Autologous graft (buccal mucosa)
  - Bowel vaginoplasty
  - Surgical traction (Vecchietti)
Creating a neovagina

Vecchietti

- Hailed as first line therapy
- Dilates vagina by passive traction on ovoid bead or olive attached by bilateral traction sutures to abdominal wall

Absence of uterus/vagina

Finding vesico-rectal window

Laparoscopic Vecchietti

Video 2
Laparoscopic Vecchietti

- Graft to make lining of neo vagina
  - Split thickness skin from buttocks or groin
  - Interceed, repliform, OASIS (SIS)
  - Amnion
- Soft dilator wrapped in graft and left in vagina
- Post-op dilation for 3-6 months - forever

Creating a neovagina

Abbe Mondoe
Anomalies affecting both the urinary and genital tracts

### Congenital adrenal hyperplasia (CAH)

- Group of autosomal recessive disorders of cortisol biosynthesis
  - 21 hydroxylase deficiency (90% of cases)
  - Overproduction of cortisol precursors → biosynthesis of sex hormones
  - Classic 21 hydroxylase deficiency
    - 1/16,000 births
    - Simple virilizing and salt wasting phenotypes

### CAH

**Clinical manifestations**

- Phenotypic abnormalities in 46 XX
- Genital abnormalities
  - Enlarged clitoris
  - Partly fused labia majora
  - Common urogenital sinus
- Skeletal abnormalities
  - Short stature (premature epiphyseal closure or glucocorticoid inhibition of growth axis)

### Embryology

**Development of external genitalia**

- **Congenital Adrenal Hyperplasia Grades**

<table>
<thead>
<tr>
<th>Female genitalia</th>
<th>Increasing virilization</th>
<th>Male genitalia</th>
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</table>

1. Hyperplastic clitoris with otherwise normal female genitalia
2. Hyperplastic clitoris, urogenital sinus, vaginal and perineal openings covered
3. Hyperplastic clitoris, narrow and deep urogenital sinus
4. Phallic with small urogenital sinus
5. Male genitalia
### Principles of surgical reconstruction
- Clitoroplasty
- Total urogenital sinus mobilization
- Vaginal reconstruction
- Single operation versus staged procedure

### Surgical approach

#### Total urogenital sinus mobilization (TUM)
- Entire UG sinus is dissected from perineum as a unit
- The “unit” will reach perineum, and a flap from the sinus can be attached to normal caliber vagina
- Urethra maintains orthotopic location
- Mobilized tissue can be used for vaginoplasty

### Video 4
CAH Surgical Steps

#### Exstrophy-Epispidias Complex
- Rare spectrum of multisystem birth defects involving the genitourinary system, gastrointestinal tract and musculoskeletal system
- Disorder of the development of the cloacal membrane
  - Classic Bladder Exstrophy (CBE)
  - Cloacal Exstrophy (CE)
- CBE results if the rupture occurs after the urorectal septum divides the gastrointestinal from the genitourinary tracts while CE results if the rupture occurs before this separation

### Embryology
- Failure of mesodermal lateral fold migration
- Weakness of anterior abdominal wall
- Rupture of the cloacal membrane
- Timing determines CE vs CBE

#### Anatomic manifestations
- Low-set umbilicus
- Bladder
- Ureteral orifices
- Separated pubis
- Anteriorly deviated anus

### Classic Bladder Exstrophy

#### Anatomic manifestations
Classic Bladder Exstrophy

**Initial surgical management**

- Goals
  - Adequate low pressure urine storage
  - Urinary continence
  - Preserved renal function
  - Functional and cosmetically acceptable genitalia

**Typical surgical repair**

- Closure of bladder and abdominal wall
- Ureteral reimplantation
- Bladder neck reconstruction
- +/- pelvic osteotomies

**Secondary surgeries**

- Augmentation cystoplasty
- Bladder neck closure with continent urinary diversion

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Cloacal Exstrophy

**OES Complex (omphalocele, exstrophy of bladder, imperforate anus, spinal abnormalities)**

- 1/200,000 to 1/400,000 live births
- "Classic Exstrophy"
  - Omphalocele
  - Exstrophy of bladder (bivalved) and bowel
  - Imperforate anus and blind ending colon
  - Diastasis of symphysis
  - Duplicated uterus
  - Mons, clitoris, and labia are separated
  - Shortened and anteriorly displaced vagina

**Individualized repair of genitourinary and gastrointestinal defects**

- Secure closure of bladder and anterior abdominal wall
- Preservation of renal function
- Prevention of short gut syndrome
- Attainment of functional and cosmetically acceptable genitalia
- Continence

**Timing of operative management varies with respect to strategies to narrow/secure pubic diastasis**

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Cloacal Exstrophy

**Initial Surgical approach**

- Separation of bladder halves from cecal plate
- Closure of omphalocoele
- Hindgut preservation
  - End colostomy
  - Colonic pull through
- Vaginal reconstruction
  - Native vagina or neovagina

**Options for vaginal reconstruction**

- Pull-through vaginoplasty
- Bowel vaginoplasty
- Vaginoplasty with native bladder

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Classic Bladder Exstrophy

**Surgical repair**

- Typical surgical repair
  - Closure of bladder and abdominal wall
  - Ureteral reimplantation
  - Bladder neck reconstruction
  - +/- pelvic osteotomies

**Secondary surgeries**

- Augmentation cystoplasty
- Bladder neck closure with continent urinary diversion
Cloacal Exstrophy

Factors affecting success

Initial bladder closure of the cloacal exstrophy complex: Outcome related risk factors and keys to success

Mohsin R. Shah, M.D., Bethany DiCicco, M.D., Seth D. Goldstein, M.D., Philip M. Pierpont, M.D., Brian M. Iniguez, M.D., Eric Z. Maxey, M.D., Abbe Her, M.D., Jane Kinsky, M.D., Paul Oppenheimer, M.D., and John P. Canavan, M.D.

100 patients with CE and primary closure

• 26 patients with failed closure compared to 34 with successful closure

Table 2

Univariate logistic analysis of variables affecting primary closure failure.

<table>
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<th>Variable</th>
<th>Odds ratio</th>
<th>P-value</th>
<th>95% CI</th>
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<td>46XY Genotype</td>
<td>0.130</td>
<td>0.056</td>
<td>0.013–0.007</td>
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<tr>
<td>Premature closure (congenital)</td>
<td>2.626</td>
<td>0.004</td>
<td>1.303–5.299</td>
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<tr>
<td>Dysplasia/ampliﬁcations</td>
<td>0.034</td>
<td>0.822</td>
<td>0.003–13.00</td>
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<tr>
<td>Age at closure (months)</td>
<td>0.086</td>
<td>0.002</td>
<td>0.023–0.003</td>
</tr>
<tr>
<td>Obesity</td>
<td>0.001</td>
<td>0.005</td>
<td>0.001–0.003</td>
</tr>
<tr>
<td>Delay between menarche and closure</td>
<td>0.025</td>
<td>0.001</td>
<td>0.003–0.003</td>
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* Only calculated using patients who underwent surgery.

How do you transition care?

What to do in your office

Transitioning care

• Detailed description of surgical and medical history
• Fertility and pregnancy potential/ capability
• Sexual health/function
• Risks for other quality of life disorders (prolapse)
• Coordination and management of chronic conditions (incontinence, voiding dysfunction)

Are they ready

To identify those who are ready to transition to adult gynecological care:

• I can explain MRKH (and any other medical conditions I have) to my GYN and medical providers. (If not, it’s very helpful to provide your GYN and other medical providers with a copy of the MRKH guide for health care providers.)
• I know the names and phone numbers of all of my health care providers.
• I look questions during my GYN and other medical appointments...
• I know what medications I take and I have a list that I bring to my GYN and medical appointments. I tell my providers when I no longer take certain medication(s) and when I start a new medication(s)
• I feel comfortable responding to questions my gynecologist and other medical providers ask.
• I am able to schedule my GYN and other medical appointments by myself and I have a way of keeping track of them so I don’t miss appointments.
• I know about my health insurance coverage and how much my co-pay is for different medical services.
• I know where and how to obtain a copy of my medical records.
• I am able to get to my GYN and other medical appointments by myself.
• I know where to get my prescriptions ﬁlled, and have called the pharmacy when I’ve had questions on needed refills.

Transitioning care

Transitioning from pediatric to adult care surgery for patients with disorders of sexual development

• I can explain MRKH (and any other medical conditions I have) to my GYN and medical providers. (If not, it’s very helpful to provide your GYN and other medical providers with a copy of the MRKH guide for health care providers.)
• I know the names and phone numbers of all of my health care providers.
• I ask questions during my GYN and other medical appointments.
• I know what medications I take and I have a list that I bring to my GYN and medical appointments. I tell my providers when I no longer take certain medication(s) and when I start a new medication(s)
• I feel comfortable responding to questions my gynecologist and other medical providers ask.
• I am able to schedule my GYN and other medical appointments by myself and I have a way of keeping track of them so I don’t miss appointments.
• I know about my health insurance coverage and how much my co-pay is for different medical services.
• I know where and how to obtain a copy of my medical records.
• I am able to get to my GYN and other medical appointments by myself.
• I know where to get my prescriptions ﬁlled, and have called the pharmacy when I’ve had questions on needed refills.
Take home points

- GU anomalies are common and affect sexual health, fertility and the pelvic floor
- Care involves a multidisciplinary approach focusing on transition from pediatric to adult provider
- Anticipate the obstacles that these women face

Thank you!
Surgical management of Pelvic Floor Disorders in women with congenital GU anomalies

- Transitioning Care: ICS 2018
- Wednesday, August 29, 2018
- Sarah A. Collins, MD

Overview
Surgical management of PFDs in women with GU anomalies

- Bladder Exstrophy- Epispadias complex
  - Bladder Exstrophy: predisposed to POP
    - Skeletal abnormalities affecting muscular and connective tissue support:
      - Absent symphysis
      - Incomplete pubic rami
      - Incompletely formed sacrum
      - Absent coccyx
      - 30-50% develop POP by middle age
  - Bladder exstrophy: common reconstructive surgeries
    - Modern Staged Repair of Bladder Exstrophy: MSRBE
      - Newborn: Closure of bladder, urethra, abdominal wall
      - 4-5 years: Bladder neck reconstruction with bilateral ureteral reimplantation
    - Complete Primary Repair of Bladder Exstrophy: CPRBE
      - All repairs at once in first days of life
      - Many patients still require multiple operations
    - Osteotomy:
      - Mean pubic diastasis:
        - Normal: 0.6 cm
        - Classic BD:
          - 4 cm at birth
          - 8 cm at age 10

- MRKH: POP after neovagina
  - Case report evidence: presentation and management
- POP after surgery for Herlyn-Werner-Wunderlich Syndrome
  - Northwestern’s experience
- CAH: Continence after repair of persistent urogenital sinus

Exstrophy- Epispadias Complex

- Classic Bladder Exstrophy
- Cloacal exstrophy

Exstrophy- Epispadias Complex

Life long considerations
Urogynaccological and obstetric issues in women with the exstrophy-epispadias complex

- Series of 34 women who responded to mailed surveys
  - Female Epispadias: N=4
    - Classical Bladder Exstrophy: N=24
      - 10 with urinary incontinence
      - 10 with POP: mean age 16
      - 2 with rectal prolapse
    - Cloacal Exstrophy: N=6
      - 1 with incontinence
      - 1 with POP at 21 and 24 years
      - 1 with rectal prolapse

Exstrophy- Epispadias Complex

- Bladder Exstrophy: Epispadias complex
- Predisposing factors for PFDs
- Management of POP and UI
- MRKH: POP after neovagina
  - Case report evidence: presentation and management
  - POP after surgery for Herlyn-Werner-Wunderlich Syndrome
  - Northwestern’s experience
  - CAH: Continence after repair of persistent urogenital sinus

Exstrophy- Epispadias Complex

- https://www CENTRALKESCLINIC.BIZ/urinary-tract-2/exstrophy.html, last updated 20 April 2018

Exstrophy- Epispadias Complex

Bladder Exstrophy: common reconstructive surgeries

- Pelvic osteotomy
  - Decreases tension on bladder and abdominal closures
  - Does not appear to decrease risk of pelvic organ prolapse


Classic Bladder Exstrophy

Genital prolapse in adult women with classical bladder exstrophy

Raia S. Nakhal1, Rebecca Deans1, Sarah M. Creighton1, Dea Wood2, Christopher R. J. Woodhouse1
1st Urogynecol 2012;3:191-195

52 women after primary repair bladder exstrophy identified

- Mean age 33, mean number of GU surgeries 9
- 27 (52%) women had prolapse
  - 85% managed with surgery (hysterosacropexy)
  - 75% successful
  - 15% managed conservatively
- Repair of POP in BE patients also improves urinary continence and sexual function


Cloacal Exstrophy

Northwestern’s Experience

- 23 yo with cloacal exstrophy spontaneously conceives
  - Pregnancy complications
    - Prolapse
    - Hospital admission for pyelo
    - Hospital admission for SBO
    - Hospital admission for HELLP
  - Classical C/S at 34 wks
  - Symptomatic POP continued post-partum
    - Pessary not able to manage patient’s symptoms
    - Underwent abdominal sacral colpopexy

Cloacal Exstrophy

Prolapse before and after reconstructive surgery

Classic Bladder Exstrophy

Continence

Exchanging long-term outcomes of bladder exstrophy: a 20-year follow-up

Angelo D. Gupta, Sameer K. Goel, Christopher R.J. Woodhouse and Dan Wood
Department of Urogynecology, Johns Hopkins Medical Institutions, Baltimore, MD, USA, and University College London
International, London, UK
BJU International 2013;112:157–161

61 patients were identified (>20 years of age, men and women)

- All had undergone additional surgeries after their original reconstructive procedures
- 21 patients responded to this survey (15 men, 6 women)
  - 20 performed CSC
  - 1 was continent and self voiding
  - 13 patients completely continent, no leakage episodes on ICIQ

Cloacal Exstrophy

Continence

Clinical Outcome of Cloacal Exstrophy, Current Status, and a Change in Surgical Management

Rob van Vliet1, Lut A. J. Rokhlo2, Rianne Kossoul-Kirchmeier2, Robert P. E. de Car2
Hard L. Claeken-van der Gronden3, Chris Verhaak4, Alan J. Homan5, Catharina C. M. Beersbroek6
Ernst J. van Linden7, Michel A. A. P. Willemen3, Marc H. M. Wijnen3, Wood F. J. Jelks1, Hox de Blaauw8

- 14 patients identified with CE (5 women)
  - Initial bladder closure attempted in 13 cases, 9 initial failures
  - 5 underwent bladder neck reconstruction, 10 underwent later augmentation cystoplasty or diversion
  - 2 remain continent
Exstrophy-Episadias Complex

- Urinary incontinence:
  - Requirements for urinary continence:
    - Adequate sphincteric resistance
    - Sufficient bladder capacity without overactivity
    - Bladder regimen
  - Procedures available:
    - Bladder neck reconstruction (BNR)
    - Urethral slings
    - Artificial urinary sphincters
  - Urethral bulking:
    - Can add a modest increase in continence when used after failed BNR
    - Sufficient
    - Staged repair:
      - Bladder regimen

Case report:
Series of 20 children (boys and girls) with CBE
Transobturator
1 year postoperatively: complete vault eversion, TVL 4 cm, scarring
Urethral slings
Artificial
Sufficient
Staged repair:
Bladder regimen
Adequate
Can add a modest increase in continence when used after failed BNR

28 year old with MRKH
Self
Case report
Procedures available:
Bladder neck reconstruction (BNR)
Case report
Even after additional surgeries, 7 remained incontinent
4 continent after BNR

40 year old with MRKH
Self
Case report
Procedures available:
Bladder neck reconstruction (BNR)
Case report
Additional 2 continent after BNI
8 years later: vaginal prolapse beyond the hymen repaired by a
Postoperative vaginal dilation and vaginal estrogen for 3 months
Augmentation

• Urethral bulking:
  - Can add a modest increase in continence when used after failed BNR
  - Series of 20 children (boys and girls) with CBE:
    - 4 continent after BNR
    - Additional 2 continent after BNI
    - Even after additional surgeries, 7 remained incontinent

Prolapse of the neovagina
Self-dilation: primary repairs

Case report: 21 year old with MRKH who attained a 10 cm vagina with self-
dilation and maintained it with intercoarse developed prolapse beyond the
hymen. This was repaired with a mesh augmented bilateral sacrospinous
ligament suspension complicated by a mesh erosion.3

Case report: 23 year old with MRKH who attained a 7.5 cm vagina with self-
dilation (from a 3 cm dimple at presentation) developed complete vaginal
eversion. This was repaired with a laparoscopic sacral colpopexy, and vagina
lengthened to 9 cm postoperatively.4

Case report: 42 year old with MRKH who attained a 7 cm vagina with self-
dilation at age 17 developed stage 3 apical prolapse. This was repaired with a
laparoscopic "nerve-sparing" suspension of the vagina with polypropylene
mesh and polyester suture to the anterior longitudinal ligament at L5.5

Prolapse of the Neovagina

Self-dilation: primary repairs

Case report: A 41 year old with MRKH who underwent sigmoid neovagina
creation at age 17 presented with stage 3 apical prolapse and a "bearing
down" sensation. She underwent laparoscopic "nerve-sparing" suspension of
the vagina with a hybrid monofilament polypropylene mesh and polyester
with polyglactin suture to the anterior longitudinal ligament at L5.6

Case report: A 72 year old with MRKH who underwent sigmoid neovagina
creation at age 21 presented with stage 4 apical prolapse. She underwent
laparoscopic sacral colpopexy with polypropylene mesh and polyester
suture.7

Prolapse of the Neovagina

Case report: A 41 year old with MRKH who underwent sigmoid neovagina
creation at age 17 presented with stage 3 apical prolapse and a "bearing
down" sensation. She underwent laparoscopic "nerve-sparing" suspension of
the vagina with a hybrid monofilament polypropylene mesh and polyester
with polyglactin suture to the anterior longitudinal ligament at L5.6

- Recurrences and their repairs
  - 28 year old with MRKH:
    - Self-dilated at age 19 and achieved intercourse
    - 8 years later: vaginal prolapse beyond the hymen repaired by a
      community urologist:
      - Anterior and posterior repair with AMS Elevate mesh
      - Transobturator midurethral sling
    - 1 year postoperatively: complete vault evasion, TVL 4 cm, scarring
      - Staged repair:
        - Mesh removal followed by postoperative dilation
        - 6 months later:
          - Skin grafted vaginal eversion using cadaveric dermis
          - Postoperative vaginal dilation and vaginal estrogen for 3 months
          - 6 months after surgery: excision / advancement of posterior continuation
    - 1 year follow up:
      - Aa, −3; Ba, −3; C, −5; Gh, 3.5; Pb, 3; TVL, 7.5; Ap, −2; and Bp−2
**Prolapse of the Neovagina**

Recurrences and their repairs

- A 45-year-old with MRKH presented after multiple surgeries for POP of her sigmoid neovagina with a feeling of "bearing down" and increased vaginal discharge
  - Original sigmoid vaginoplasty, age 34
  - At age 40, she developed symptomatic prolapse
  - Laparotomy with mesh-augmented repair, Recurred on POD 15
  - 2 years later, mesh-augmented vaginal repair, Recurred after 3 years
  - Colposuspension to the round ligaments, Recurred, POD 40
  - On presentation: protrusion of neovagina to 5 cm beyond the hymen
  - Repair:
    - Laparoscopic sacral colpopyexy
    - Polypropylene mesh
    - Polyester sutures
  - Follow up: Good symptom control with no anatomic recurrence at 40 days, 3 months, 6 months

**Herlyn-Werner-Wunderlich Syndrome**

- 17 year old with müllerian anomaly presents with pain and abnormal bulge in vagina when urinating
  - PMH:
    - Uterus didelphys – Right vaginal septum and associated hematocolpos, L solitary kidney
  - PSH:
    - 2007: resection of right transverse vaginal septum
    - 2011: repair of "anterior vaginal wall hernia"
  - Ultimately required repair of vaginal wall defect with graft

**CAH: Persistent urogenital sinus**

- TUM procedure: Total urogenital mobilization
- Retrospective chart review of children with PUGS
  - Intermediate and high defects
  - Follow up: subjective report of continence

**Complications of excision transverse vaginal septum**
Key Facts

In summary...

- GU anomalies predispose women to POP and UI
- Knowledge of common reconstructive surgeries
  - Operative reports
- In patients with Exstrophy-Episadias Complex:
  - POP may be best managed with sacral colpopexy after childbearing
  - Bulking may improve urinary incontinence after BNR
- MRKS: TV mesh may lead to failure and future revision surgeries
- Herlyn-Werner-Wunderlich Syndrome:
  - Complications of transverse septum resection: POP

Questions?

Thank You
Let's talk about sex

- Sexual activity is attainable in this population
- Many women have barriers to satisfactory sexual function
- Must address this proactively
  - Goal: Address before they have pain

Elements of female sexual function

- Sexual desire
- Arousal
- Orgasm
- Pain

- Sexual symptoms are not considered a dysfunction unless they cause personal distress for the individual
- Sexuality is always a couple issue—not just the one who appears to be presenting with the problem

Components of healthy sexuality

- Share pleasure
- Deepen and reinforce intimacy
- Achieve desired pregnancy
- Decrease tension and to alleviate the stresses inherent in life and relationships
- Energize and reinforce a feeling of desire and desirability

Evaluation: Bio-Psycho-Social approach

- Biological: health, nutrition, medications, endocrine function, neurobiology
- Psychological: depression, anxiety, performance anxiety, PTSD, ADD/ADHD
- Socio-cultural: family upbringing, societal/cultural attitudes, religious influence
- Interpersonal: quality of current and past relationships, intimacy, sense of self-worth

Adapted from Kinsberg and Reszaee (2013)

Sexual Dysfunction

- DSM V vs Sexual Medicine
  - Female sexual interest/arousal disorder
  - Female orgasmic disorder
  - Genito-pelvic pain/penetration disorder
  - Hypoactive sexual desire disorder
  - Female sexual arousal disorder
  - Female orgasmic disorder
  - Persistent genital arousal disorder
  - Vulvar pain caused by a specific disorder
  - Vulvodynia
    - Generalized vulvodynia
    - Provoked localized vestibulodynia
Pain and intimacy

- Unwelcome emotional and physical challenge
- Physical intimacy
  - Fear that sexual activity will follow all contact
  - Intercourse, sensual contact ceases
- Women avoid sexual contact
  - Sexual comfort/skills atrophy
  - Couples lose intimacy

Couple avoids touching

Pain also negatively affects:
- desire
- arousal
- orgasm

Partner effects of dyspareunia

- Also affected by dyspareunia
- Helpless, angry and depressed
- Do not understand what is going on
  - Think partner is avoiding sex for other reasons
  - Take it personally
  - Think partner doesn’t find them attractive
  - Feel rejected
- Even those who understand what’s going on → can inadvertently contribute to relationship problems by avoiding any type of physical intimacy

Sexual dysfunction in women with complex GU anomalies

- Reconstructive vaginoplasties
  - Childhood vs later in life
  - Scarring/Stricture
  - Adequate length for intercourse
  - Pelvic floor dysfunction
  - Concomitant pelvic floor disorders
  - Emotional considerations (body image)

Mo’s goals in the transitions clinic

- Prevent sexual dysfunction as sequelae of vaginal reconstructive surgery
- Prepare women for a healthy sexual relationship
- Educate women on their anatomy
- Address sexual function and concerns if and when they arise

Mo’s recipe

- Pre-operative consultation
- Post-operative close follow up
- Age appropriate history
  - Any sexual concerns
  - Any sexual activity
  - Any pelvic floor concerns
- Physical exam
  - Length and caliber of vagina
  - Assessment of vagina epithelium
  - Assessment of vaginal scarring
  - Assessment of pelvic floor musculature

Maintaining or restoring vaginal length/caliber

- Dilation therapy is critical
- Self dilation 2-3 times daily immediately post-op
- Dilating schedule/duration based on goals
Dilating 101

The right equipment

- Set expectations pre-op
- Introduce dilating ASAP postop
- Close follow-up
- Weekly visits
- Allow patient to self demonstrate dilating and progress
- Premedicate with lidocaine gel

Vaginal estrogen

- Great adjunct to dilating
  - Post-vaginoplasty vaginal epithelium retains estrogen receptors
  - Helps prevent scarring in septum resections
  - Insert 0.5 mg daily x 2wks, then maintenance twice weekly
  - Not well studied in this population
  - Can be helpful in CAH

Pelvic Floor Physical Therapy

- Assess for myofascial pelvic floor dysfunction
  - Resultant from surgical procedures
  - Resultant from dyspareunia
- Additional musculoskeletal abnormalities
- PT can be an important adjunct to dilator therapy

Vaginal Septa

Original Studies

Presenting and Long-Term Clinical Implications and Fecundity in Females with Obstructing Vaginal Malformations

- Women can develop scar tissue, most often in area of resection concentric scar at the level of the previous septum
- 2 of the 3 patients with resection of TVS required re-op for stricture
- Little known on long term sexual dysfunction
Vaginal septae

Sexual and functional results after creation of a new vagina in women with Mayer-Rokitansky-Küster-Hauser syndrome: a comparison of nonsurgical and surgical procedures

Table 2: Female Sexual Function Index scores for the two groups.

<table>
<thead>
<tr>
<th>Domain</th>
<th>Nonsurgical group (n = 20)</th>
<th>Surgical group (n = 20)</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Desire</td>
<td>4.5 ± 1.0</td>
<td>4.6 ± 0.8</td>
<td>0.5</td>
</tr>
<tr>
<td>Anxual</td>
<td>4.6 ± 1.6</td>
<td>4.3 ± 1.7</td>
<td>0.7</td>
</tr>
<tr>
<td>Lubrication</td>
<td>4.2 ± 2.0</td>
<td>4.3 ± 1.8</td>
<td>0.8</td>
</tr>
<tr>
<td>Orgasm</td>
<td>4.0 ± 1.8</td>
<td>3.8 ± 1.9</td>
<td>0.6</td>
</tr>
<tr>
<td>Satisfaction</td>
<td>4.7 ± 1.6</td>
<td>4.7 ± 1.7</td>
<td>1.0</td>
</tr>
<tr>
<td>Confort</td>
<td>3.4 ± 1.9</td>
<td>3.4 ± 1.1</td>
<td>0.6</td>
</tr>
<tr>
<td>Total PFI score</td>
<td>25.3 ± 7.3</td>
<td>25.3 ± 6.8</td>
<td>1.0</td>
</tr>
</tbody>
</table>

Values are given as mean ± standard deviation (95% confidence interval).

CAH

Total urogenital sinus mobilization (TUM)

Table 3: Female Sexual Function Index scores according to type of management

<table>
<thead>
<tr>
<th>Type of management</th>
<th>Global PFI score</th>
<th>Desire</th>
<th>Anxual</th>
<th>Lubrication</th>
<th>Orgasm</th>
<th>Satisfaction</th>
<th>Confort</th>
<th>Pain</th>
</tr>
</thead>
<tbody>
<tr>
<td>Surgery</td>
<td>30.4 ± 20.5</td>
<td>4.5 ± 2.0</td>
<td>4.6 ± 1.7</td>
<td>4.3 ± 1.8</td>
<td>3.8 ± 1.9</td>
<td>4.7 ± 1.7</td>
<td>3.4 ± 1.1</td>
<td>25.3 ± 7.3</td>
</tr>
<tr>
<td>Endocrine</td>
<td>22.3 ± 15.0</td>
<td>4.3 ± 1.5</td>
<td>4.2 ± 1.6</td>
<td>4.6 ± 1.8</td>
<td>3.8 ± 1.9</td>
<td>4.7 ± 1.7</td>
<td>3.4 ± 1.1</td>
<td>25.3 ± 7.3</td>
</tr>
<tr>
<td>Total TUM</td>
<td>25.3 ± 6.8</td>
<td>4.7 ± 1.7</td>
<td>4.7 ± 1.7</td>
<td>4.7 ± 1.7</td>
<td>4.7 ± 1.7</td>
<td>4.7 ± 1.7</td>
<td>4.7 ± 1.7</td>
<td>4.7 ± 1.7</td>
</tr>
</tbody>
</table>

Values are given as mean ± standard deviation (95% confidence interval).

• Unique challenges
  - Body image
  - Coitarche
  - Non-surgical versus various surgical procedures
  - Many have had several procedures: dilating followed by surgical management

Surgical care is not superior to dilation for the management of vaginal agenesis in MRKH syndrome: a multicenter comparative observational study in 131 patients

Mayer-Rokitansky-Küster-Hauser Syndrome

• Vaginoplasty typically undertaken at birth
• Girls at risk for stricture and vaginal stenosis
• Mobilized tissue from UG sinus can be used for vaginoplasty
• Clitoroplasty
CAH

Sexual Function and Attitudes Toward Surgery After Feminizing Genitalplasty
Riitta Fagerholm, Pekka Santtia, Päivi J. Miettinen, Aino Mattila, Riitta Rintala and Seppo Taskinen*

• Assessment of sexual function using FSFI
• 24 women (16 with CAH)
  - Clitoroplasty (mean age 3.8 years)
  - Vaginal reconstruction (mean age 4.5 years)
• 43% required surgical revision for narrowed vagina
• 54% required vaginal dilation under anesthesia

Classic Bladder Exstrophy

Anatomic manifestations

Low set umbilicus
Bladder
Ureteral orifices
Separated pubis
Anteriorly deviated anus

Pelvic Floor:
- Diastasis of pubic bone
- Posterior deviation of levators

Genitalia:
- Bifid clitoris
- Short or stenotic vagina

Cloacal Exstrophy

OES Complex (omphalocele, exstrophy of bladder, imperforate anus, spinal abnormalities)

<table>
<thead>
<tr>
<th>Genital (XX)</th>
<th>(n = 7)</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Vagina aplasia</td>
<td>1</td>
<td>14</td>
</tr>
<tr>
<td>Vagina duplex</td>
<td>5</td>
<td>71</td>
</tr>
<tr>
<td>Clitoris aplasia</td>
<td>5</td>
<td>14</td>
</tr>
<tr>
<td>Uterus didelphys</td>
<td>7</td>
<td>100</td>
</tr>
</tbody>
</table>

Life long considerations

Urogynaecological and obstetric issues in women with the exstrophy–epispadias complex
R.J. Mathews, M. Gan and J.P. Bearhart
Division of Pediatric Urology, The James Buchanan Brady Urological Institute, Baltimore, MD USA
Accepted for publication 5 February 2003

14 women included in analysis with cloacal exstrophy
6 patients underwent genital reconstructive surgery as part of the closure surgeries
Cloacal Exstrophy

- Sexual function
  - 4/6 women had further genital surgery
  - 3/5 women (>18 years) had intercourse
    - 2 reported sexual satisfaction
    - 1 discontinued sexual activity 2/2 uterine prolapse
  - 2 patients reported that they were not sexually active due to the appearance of their genitalia

Take home points

- Women in this population are sexual and can have a healthy functional sexual relationship
- Many women have barriers to satisfactory sexual function
- Must address this proactively
- Remember birth control or STI prevention if indicated!

Thank you!
Genitourinary Anomalies and Fertility

- Mullerian Anomalies
  - Unicornuate
  - Didelphys
  - Bicornuate
  - Uterine Septum
- Congential Adrenal Hyperplasia
  - Fertility implications and treatment
- Mayer-Rokitansky-Kuster-Hauser Syndrome
  - Uterine Transplantation

Prevalence of Mullerian Anomalies

<table>
<thead>
<tr>
<th>Category</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Women with Late Miscarriages &amp; Preterm Deliveries</td>
<td>&gt;25%</td>
</tr>
<tr>
<td>Women with Recurrent Miscarriage</td>
<td>5-16%</td>
</tr>
<tr>
<td>Women with Infertility</td>
<td>5-7%</td>
</tr>
<tr>
<td>Fertile Women</td>
<td>3-7%</td>
</tr>
</tbody>
</table>

*60% of women with unilateral renal agenesis have genital anomalies

Mullerian Anomalies: Reproductive outcomes

- Prevalence of Mullerian defects in fertile and infertile women. Hum Reprod 1997;12:1372

Mullerian Anomalies: Reproductive outcomes


Values shown are risk ratios (95% CI). *P < 0.05, **P < 0.01, ***P < 0.001.
Mullerian Anomalies: Reproductive outcomes

<table>
<thead>
<tr>
<th>Anomaly</th>
<th>Conception rate</th>
<th>First-trimester miscarriage</th>
<th>Second-trimester miscarriage</th>
<th>Preterm labor</th>
<th>Malpresentation at delivery</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anomalistic uterus</td>
<td>0.85 (1-1.22)</td>
<td>0.35</td>
<td>0.29</td>
<td>0.29</td>
<td>1.25</td>
</tr>
<tr>
<td>Cervical defects</td>
<td>0.80 (0.4-1.5)</td>
<td>0.36</td>
<td>0.29</td>
<td>0.29</td>
<td>1.29</td>
</tr>
<tr>
<td>Subseptum</td>
<td>0.93 (0.3-1.7)</td>
<td>0.36</td>
<td>0.29</td>
<td>0.29</td>
<td>1.29</td>
</tr>
<tr>
<td>Seprum</td>
<td>0.73 (0.1-1.7)</td>
<td>0.36</td>
<td>0.29</td>
<td>0.29</td>
<td>1.29</td>
</tr>
<tr>
<td>AS</td>
<td>0.12 (0.01-1.1)</td>
<td>0.39</td>
<td>0.29</td>
<td>0.29</td>
<td>1.29</td>
</tr>
<tr>
<td>Distortion defects</td>
<td>0.86</td>
<td>1.82</td>
<td>0.32</td>
<td>0.32</td>
<td>1.15</td>
</tr>
<tr>
<td>Didelphys</td>
<td>0.80 (0.4-1.3)</td>
<td>0.36</td>
<td>0.29</td>
<td>0.29</td>
<td>1.29</td>
</tr>
<tr>
<td>Unicornate</td>
<td>0.85 (0.3-1.7)</td>
<td>0.36</td>
<td>0.29</td>
<td>0.29</td>
<td>1.29</td>
</tr>
<tr>
<td>3D Ultrasound</td>
<td>0.80 (0.3-1.3)</td>
<td>0.36</td>
<td>0.29</td>
<td>0.29</td>
<td>1.29</td>
</tr>
</tbody>
</table>

Value denotes the risk ratio (95% CI). *P < 0.05, **P < 0.01, ***P < 0.001.

Mullerian Anomalies: Diagnosis

- **Gold Standard**
  - Hysteroscopy and Laparoscopy
- HSG or Hysteroscopy alone
  - Insufficient to evaluate the contour of the uterine fundus
- 3D Ultrasound + SIS
  - 3D Ultrasound alone: 88% Accuracy
  - 3D Ultrasound + SIS: 100% Accuracy
- MRI

Unicornuate

- Pathophysiology: Failure of development of one mullerian duct
- Reproductive Outcomes:
  - Miscarriage (25-30%)
  - Ectopic Pregnancy (3-4%)
  - Rudimentary horn
  - IUGR (10%)
  - Malpresentation

Rudimentary horn with endometrium

Unicornuate Uterus

Uterus Didelphys

• Gold Standard
  - Hysteroscopy and Laparoscopy
• HSG or Hysteroscopy alone
  - Insufficient to evaluate the contour of the uterine fundus
• 3D Ultrasound + SIS
  - 3D Ultrasound alone: 88% Accuracy
  - 3D Ultrasound + SIS: 100% Accuracy
• MRI
Uterine Didelphys

- Pathophysiology: Complete failure of fusion of the mullerian ducts and normal differentiation of each to form a cervix and hemi-uterus
- Reproductive outcomes:
  - No impact on fertility
  - Increased risk of preterm labor and malpresentation
- No evidence to support surgical treatment, except for excision of vaginal septum

Bicornuate Uterus

- Pathophysiology: Incomplete fusion of the mullerian ducts at the level of the fundus, creating two separate uterine cavities.
  - One lower uterus and single cervix
  - External midline cleft
- Reproductive Outcomes:
  - 30-40% pregnancies end in miscarriage
  - Increased risk of preterm delivery
  - Increased risk of malpresentation
- Management:
  - Metroplasty is no longer considered

Septum

- ESHRE
  - Internal indentation extending > 50% of myometrial wall thickness
- ASRM
  - Normal/Arcuate < 1.5cm
  - Septate > 1.5cm
  - Bicornuate: External fundal indentation > 1cm

Uterine Septum

Definitions: Normal/Arcuate, Partial Septum, Complete Septum, Complete Septum with Longitudinal Vaginal Septum
Uterine Septum: Fertility

- Fertility
  - Venetis et al, 2014
    - Septum with decreased probability of natural conception
    - RR 0.86, 95% CI 0.77 to 0.96, three studies
  - Tomazevic et al, 2010
    - Lower pregnancy and live birth rates in those with a septum vs. controls
    - 12.4% vs. 22.2%, P = 0.001 (Pregnancy Rate)
    - Pregnancy rates higher after incision
    - OR 2.5 (95% CI 1.5 to 4.1, P < 0.001)

- Miscarriage
  - Kupesic et al, 2002
    - 689 women with septum vs. 15,060 women in general population
    - Early miscarriage: 41.1% vs. 12.1%
    - Late miscarriage and preterm delivery: 12.6% vs. 6.3%
  - Saygili et al 2003
    - Retrospective case series
    - 361 patients with septum and primary infertility or RPL
    - Miscarriage rate decreased from 91.8% to 10.4% following septum incision

Congenital Adrenal Hyperplasia

- Congenital Adrenal Hyperplasia
  - Most frequently encountered genetic steroid disorder affecting fertility
  - Classic Disease: 1/16,000
    - Cortisol deficiency
    - Virilization
  - Non-classic CAH: 1/600
    - Often asymptomatic
    - Elevated androgen precursors from the adrenals lead to impaired fertility
    - Often presenting symptom leading to diagnosis
    - Can present similarly to PCOS

- Elevated Androgens
  - Alter GnRH pulse generator leading to elevated LH levels
  - Treatment with glucocorticoids leads to decreased androgens and normalization of LH levels
  - Impact: Ovarian Function
    - Adrenal androgens directly inhibit folliculogenesis by negative feedback on granulosa cell aromatase activity

- Elevated Progesterone
  - Alter GnRH pulse frequency
  - Impact endometrial development
  - Thicken cervical mucus

- Altered psychosocial development
  - Exposure to adrenal androgens in utero
  - Females with classic CAH can exhibit masculinized behaviors including childhood play
  - Virilization of external genitalia associated with impaired sexual functioning
  - Sexual intercourse can be prohibitively uncomfortable
Congenital Adrenal Hyperplasia: Preconception Counseling

- Classic CAH
  - Carrier frequency 1:62
  - Likelihood of having an affected child 1:120
- Non-classic CAH
  - Carrier frequency 1:5-1:16
  - Likelihood of having an affected child 1:32
- Recommend screening partner for carrier status and can offer Preimplantation genetic testing of embryos if desired

Congenital Adrenal Hyperplasia: Fertility Treatments

- Classic CAH
  - Nearly all require treatment to ovulate
    - Ovulation using only steroid maintenance therapy
    - May need additional corticosteroids to suppress adrenal progesterone production
- Non-Classic CAH
  - Androgen excess frequently correctable with glucocorticoid treatment alone
  - Ovulation induction with letrozole or clomiphene

Mayer-Rokitansky-Kuster-Hauser Syndrome

- Mullerian Agenesis
  - Absence of the vagina
  - Absent or hypoplastic uterus
  - Normal or hypoplastic fallopian tubes
  - Normal ovaries
  - Normal breast and pubic hair development
  - Primary Amenorrhea
- Pregnancy achieved through IVF and surrogacy
  - IVF challenging secondary to ovaries high in pelvis and lack of vagina for transvaginal ultrasound guided oocyte aspiration
  - May require abdominal oocyte retrieval

Earliest Attempts

- 1931
  - Lili Elbe, a transgender woman, died from organ rejection 3 months after receiving a uterine transplant
- 2000
  - Saudi Arabia. Transplantation of a uterus from a 46 yo hysterectomy patient to a 26 yo recipient
  - Uterus functioned for 99 days and then was removed for thrombosis and uterine necrosis
- 2011
  - First uterus transplant from a deceased donor was performed on Derya Sert in Turkey
  - First pregnancy, however pregnancy ended in miscarriage
Summary

- Congenital GU anomalies can have significant impacts on fertility and pregnancy outcomes
- Appropriate diagnosis can be achieved with 3D ultrasound +/- saline
- Correction of a uterine septum can improve fertility and decrease miscarriage rates
- Pregnancy is possible in women with CAH with steroid hormone replacement and/or ovulation induction
- Women with MRKH can have success with the use of IVF and a gestational surrogate. Investigation into experimental treatment with uterine transplant is ongoing.

References

- Fritz M, Speroff L. Clinical Gynecologic Endocrinology. 2010
- Yen & Jaffe. Reproductive Endocrinology. 2019
- Uterine Septum, A guideline: Practice Committee of the American Society of Reproductive Medicine

Thank You
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Northwestern Medicine