Ultrasound of the Pediatric Female Pelvis

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Objectives

• Review development of gonads and GU tract
• Examine role of US in diagnosis and management of gynecologic disorders of pediatric pelvis
  – Ambiguous genitalia
  – Prepubertal bleeding
  – Primary amenorrhea
  – Pelvic mass
  – Pelvic pain
Imaging

- US primary modality
- CT or MR for further evaluation of congenital malformation or neoplasm
Technique

- Transabdominal and linear array probes adequate for most indications
- Perineal approach useful in young girls with urogenital malformation, hydrometrocolpos, anal atresia, labial mass
- Transvaginal probe in sexually active adolescents
- Sterile water, saline or water-soluble contrast to outline vagina, urogenital sinus or cloaca in complex congenital anomalies
Transperineal Anatomy
Normal Anatomy

• Size and shape of ovaries and uterus age-dependent and under hormonal influence
• Maternal and placental hormones produce relatively large size of neonatal uterus and ovaries compared with infancy
• Uterus and ovaries stable in size and appearance from infancy until growth spurt at 7-8 years of age
• Ovary contains mature follicles at all ages due to secretion of FSH
Normal Neonatal Uterus and Ovaries

- Neonatal cervix larger than fundus
- Uterine length approximately 3.5 cm, maximal thickness 1.4 cm; ovaries slightly > 1 cm³
- Endometrial lining often echogenic
- Fluid may be seen in endometrial cavity
Prepubertal Uterus and Ovaries

- Cervical thickness greater or equal to that of fundus
- Endometrial lining relatively inconspicuous
- Uterine length 2.5-4 cm, thickness ≤ 1 cm
- Ovarian volume slightly < 1 cm³
Pubertal/Postpubertal Uterus and Ovaries

- Uterine fundus elongates and thickens
- Larger than cervix
- Endometrial lining undergoes cyclical changes associated with menstrual cycle
- Ovarian volume averages 8-10 cm³
Development of Female Reproductive Tract

- Cellular differentiation, migration, fusion, canalization and apoptosis (programmed cell death)
- Complex and integrated sequence of events associated with many opportunities for abnormal development and structural anomalies
Development of Reproductive Tract

- Primordia of female and male reproductive tracts develop together in first 3 months of life
- Early embryonic gonad bipotential
- Gonad becomes ovary or testis depending on differentially expressed genes
- Gonadal differentiation leads to testicular and ovarian hormone production and induction of anatomical differences
Development of Reproductive Tract

- Urorectal septum forms by the seventh week, separating the urogenital sinus from the rectum
- Cell layers involved in formation of the female reproductive tract
  - mesoderm, endoderm, ectoderm
- Müllerian ducts arise from mesoderm and develop into uterus, upper vagina and fallopian tubes
- Urogenital sinus arises from endoderm and develops into lower vagina, bladder, urethra, and vestibule
Development of Reproductive Tract

- Fusion of endoderm and ectoderm required for canalization
- Defects lead to duplication anomalies or obstructive lesions
Uterine Duplication Anomalies

- Septate, bicornuate, didelphys, unicornuate
Anomalies of Female Reproductive Tract

• May manifest at different stages of life
• Most abnormalities of external genitalia obvious at birth
• Obstructive and nonobstructive lesions may be apparent at birth or only later (childhood, puberty, adolescence, or adulthood)
Pelvic Pain and Severe Menstrual Cramping: Septate Uterus
Patient with VATER Association and RLQ Pain

Right

Left
Bicornuate, Bicollis Uterus with Complete Vertical Vaginal Septum
17-Year-Old Girl with MRKH Syndrome: Uterus Didelphys
Ambiguous Genitalia

- Any deviation from normal of the appearance of the genitalia should prompt immediate investigation
- Bilateral cryptorchidism
- Unilateral cryptorchidism with incomplete scrotal fusion
- Hypospadias
- Labial fusion
- Clitororomegaly
Disorders of Sexual Development (DSD)

- Congenital conditions with atypical development of chromosomal, gonadal, or anatomic sex
- Sex chromosome DSD (karyotype not the usual 46, XX or 46, XY)
- 46, XX DSD (chromosomally female)
- 46, XY DSD (chromosomally male)
- Karyotyping, hormonal analysis and imaging evaluation required for proper characterization
Imaging Evaluation

- US used to determine location of gonads and presence of a uterus
- Retrograde genitogram and/or VCUG to delineate anatomy of urethra, vagina, urogenital sinus, and cervix
- In setting of cloacal anomaly, delineation of the connections of the urinary, genital and GI tracts to the cloaca can be ascertained
Sex Chromosome DSD

- Turner syndrome most common
- Partial or complete absence of one X-chromosome
  - 45,X most common
- Associated abnormalities
  - hypothyroidism, congenital heart disease, renal anomalies, autoimmune disorders
- Characteristic physical features
  - short stature, broad chest, low-set ears, webbed neck
- Gonadal dysfunction
  - amenorrhea, sterility

www.turnerssyndrome.net
18-Year-Old Woman with Karyotype 45,X

Rt ovary volume 2.1 cc

Lt ovary volume 1.3 cc

Rt

Lt
46,XX DSD

- Most cases due to congenital adrenal hyperplasia
  - normal uterus and ovaries
  - enlarged adrenal glands
  - masculinization of lower urogenital tract
    - urogenital sinus most common
- Rare causes of masculinized, chromosomally normal females
  - androgen ingestion in early pregnancy
  - masculinizing ovarian tumor
Congenital Adrenal Hyperplasia

Courtesy David Diamond, M.D., Boston
Prepubertal Bleeding

- Vulvovaginitis
  - most common
- Precocious puberty
- Vaginal foreign body
- Genital trauma
  - accidental, self-inflicted, physical or sexual abuse
- Vaginal mass
Precocious Puberty

• Onset of pubertal changes prior to 8 years of age
• True isosexual precocious puberty
  – increased serum levels of gonadotrophins in response to GRF stimulation test
  – >80% due to idiopathic activation of hypothalamic-pituitary-gonadal axis
  – remainder due to hypothalamic or pituitary lesions
• Pseudosexual precocious puberty
  – increased serum estrogen with low gonadotrophin
  – ovarian tumors, adrenal adenoma and carcinoma, neurofibromatosis, McCune-Albright syndrome, estrogen ingestion
Precocious Puberty

• Premature thelarche
  – isolated breast development

• Premature adrenarche
  – isolated pubic hair development

• US used to determine ovarian and uterine size and morphology to confirm or exclude a postpubertal appearance of internal genitalia

• US can exclude an ovarian or adrenal tumor

• US can monitor the effect of medical treatment
Three-Year-Old Girl with Isosexual Precocious Puberty
Six-Year-Old Girl with Pseudosexual Precocious Puberty
Adrenocortical Carcinoma

2.5 year later....
8-Year-Old Girl with Abdominal Pain
Vaginal Foreign Body

- Presents with vaginal bleeding, discharge, urinary symptoms, abdominal or pelvic pain
- Toilet paper, fibrous material from clothing and carpets most common
- Self-exploration
- Sexual abuse
- Both radiopaque and nonradiopaque objects appear echogenic
  - distal acoustic shadowing may be present
  - may be slight indentation on posterior bladder wall
Vaginal Masses

• Benign
  – common
    • cyst
    • polyp

• Malignant
  – rare
    • rhabdomyosarcoma
    • endodermal sinus tumor
    • clear cell sarcoma
16-Year-Old Girl with Gartner’s Duct Cyst
Adolescent with Exophytic Vaginal Mass: Fibroepithelial Polyp
17-Year-Old Girl with Menorrhagia and Pelvic Pain: Vaginal Rhabdomyosarcoma

Courtesy Marilyn Siegel, M.D., St. Louis
Vaginal Rhabdomyosarcoma

- Usually arises in anterior wall adjacent to cervix
- Uterine involvement secondary to direct extension from vaginal tumor
- Presents with vaginal bleeding and vaginal, perineal, or vulvar mass
- Metastases to liver, lymph nodes, lung, and bone
- Bimodal distribution:
  - first peak between 2-6 years of age
  - second peak between 14-18 years of age
- Embryonal and botryoid subtypes most common
Primary Amenorrhea

- Absence of menarche by age 16 years in presence of normal growth and secondary sexual characteristics

- Causes:
  - DSD (e.g. Turner syndrome)-50%
  - hypothalamic hypogonadism-20%
  - absence of uterus, cervix, and/or vagina-15%
  - transverse vaginal septum or imperforate hymen-5%
  - pituitary disease-5%
  - other disorders (androgen insensitivity, congenital adrenal hyperplasia, polycystic ovarian syndrome)-5%
14-Year-Old Girl with Amenorrhea

Rt ovary

Lt ovary
Müllerian Agenesis
Müllerian Agenesis

- (a.k.a. Mayer-Rokitansky-Küster-Hauser Syndrome)
- Normal karyotype (46,XX)
- Vaginal atresia
- Absent or rudimentary uterus (unicornuate or bicornuate)
- Functional endometrium in 6%-10% may lead to unilateral hematometra
Müllerian Agenesis

• Hymen usually present along with a small distal vaginal pouch or vaginal dimple
  – hymen and distal vagina both derived embryologically from urogenital sinus
• Normal ovaries
• Renal anomalies in 50%
  – agenesis, ectopia
• Skeletal or spinal anomalies in 12%
Obstructive Müllerian Anomalies

• Most neonates with hydrometrocolpos have a urogenital sinus or cloacal malformation
• Adolescents with obstructive uterovaginal anomalies present with amenorrhea and cyclic abdominal pain
• US useful in differentiating hemato(metro)colpos due to imperforate hymen or transverse vaginal septum (common) from hematometra due to cervical dysgenesis (rare)
Neonate with a Single Perineal Opening
Cloacal Malformation

Courtesy Jeanne Chow, M.D., Boston
Cloacal Malformation

- Occurs exclusively in females
- Incidence of 1 in 40,000 to 50,000 newborns
- Failure of urorectal septum to form or fuse properly with incomplete separation of urinary, genital and GI tracts
- Cloaca is the common confluence
- Single perineal orifice
- Associated failure of fusion of müllerian ducts leads to duplication of uterus and proximal vagina
Cloacal Malformation

- Urogenital sinus persists
- Hymen and lower vagina do not form appropriately
- Drainage of pooled urine through vagina necessary to prevent bladder outlet obstruction
- GI tract decompressed with colostomy
- Many affected children have additional anomalies
- Detailed diagnosis usually requires genitography and contrast examination of distal limb of colostomy
Transverse Vaginal Septum

- Failure of fusion and/or canalization of müllerian ducts and urogenital sinus
- External genitalia appear normal
- Upper (46%), mid (40%) or lower (14%) vagina (14%)
- Vagina short or a blind-ending pouch
- Septa generally < 1cm thick
- Extend completely or incompletely across vagina
Transverse Vaginal Septum
Distal Vaginal Atresia

- Lower portion of the vagina fails to develop from urogenital sinus
- Normal uterus, cervix, and upper vagina
- Fibrous tissue replaces absent distal vagina
- Normal secondary sexual characteristics
Distal Vaginal Atresia
Treatment

• Hematometrocolpos cured by relieving obstruction
• Complete vaginal obstruction may lead to endometriosis
Pelvic Pain

• Adnexal torsion
• Hemorrhagic ovarian cyst
• Pelvic inflammatory disease
• Ectopic pregnancy
Adnexal Torsion

• Involves ovary and fallopian tube
• Partial or complete rotation of ovary on its vascular pedicle
• Compromise of lymphatic, venous and arterial flow
• Pathology ranges from massive edema to necrosis
• Nearly always unilateral
• Can occur in normal adnexa or in association with underlying mass
• Most common in adolescents and young women
Adnexal Torsion

• Lesions predisposing to torsion
  – cyst in neonate
  – cyst or teratoma in adolescent
• Hypermobility due to lax supporting ligaments thought to account for torsion of normal adnexum
• Affected ovary at least 5-6 times larger than normal contralateral ovary
• Multiple peripherally located cysts only relatively specific sign of torsion
Prenatal Diagnosis of Ovarian Cyst: In-Utero Torsion of Cyst with Hemorrhage
Hemorrhagic Ovarian Cyst

• Complication of cyst in neonate or adolescent girl

• Neonatal cyst
  – maternal hormonal stimulation *in utero* results in exaggerated follicular development
  – hemorrhage often indicative of associated torsion

• Adolescent cyst
  – graafian follicle grows after failed ovulation or corpus luteum persists after ovulation

• US features reflect age of blood
  – fresh blood hyperechoic
  – older blood heterogeneous
  – blood becomes anechoic as clot lyses
Hemorrhagic Ovarian Cyst

- Most are complex masses with interdigitating septations
  - fishnet/cobweb/spiderweb appearance
- Fluid-debris level
- Clotted blood
- Complicated by torsion and rupture
Pelvic Inflammatory Disease

• Affects girls of reproductive age
• Usually due to ascending sexually transmitted infection
• Common causative organisms *Neisseria gonorrhoeae* and *Chlamydia trachomatis*
• Diagnosis usually established clinically
  – fever, pelvic pain, tenderness, vaginal discharge
• Sonography useful in identifying complications
  – pyosalpinx, tubo-ovarian abscess
Pelvic Inflammatory Disease

- Sonographic findings depend on stage of inflammatory process
- Minimal if any US abnormalities early in course of infection
- Pyosalpinx appears as thick-walled, fluid-filled tubular structure, oval or round mass with low-level echoes due to purulent debris
- Spectrum of ovarian involvement
  - enlarged and echogenic
  - complex adnexal mass due to abnormal ovary and fallopian tube matted together (tubo-ovarian complex)
  - Complex intra-ovarian mass (tubo-ovarian abscess)
Tuboovarian Complex
Tuboovarian Abscess and Pyosalpinx
Ectopic Pregnancy

- Rare in young adolescents
- Highest reported death rate
- Consider diagnosis in presence of pelvic pain, abnormal beta human chorionic gonadotropin (β-hCG) level, irregular vaginal bleeding, and missed menstrual period
Gynecological Pelvic Masses

• Ovarian cyst
• Peritoneal inclusion cyst
• Hematocolpos
• Primary ovarian neoplasm
  – 2/3 benign
  – 1/3 malignant
• Metastatic ovarian disease
  – extremely rare in children
Gynecological Pelvic Masses

- Primary vaginal and uterine neoplasms
  - rare in childhood
  - usually malignant

- Pregnancy
  - consider in differential diagnosis of a pelvic mass in girls 9 years of age and older
  - increased risk of complications in pediatric pregnancy
    - toxemia, preeclampsia, placental abruption, laceration, cesarean section
Peritoneal Inclusion Cyst

- Ovaries are main source of production of peritoneal fluid in young women
- Fluid usually transported throughout peritoneal cavity and absorbed by mesentery
- In setting of extensive peritoneal adhesions fluid entrapped in pelvis
- US shows cystic, septated masses
Ovarian Tumors

• May arise from germ cells, stroma, or surface epithelium
• Most occur in second decade of life
• Teratoma most common
  – composed of elements of all 3 germ cell layers
  – 90% classified as mature (cystic)
  – 10% classified as immature (contain embryonic neural elements) or malignant
• Ectodermal components predominate in cystic teratomas (dermoid cysts)
Cystic Teratoma

- Accounts for >90% of all benign ovarian tumors
- Usually unilateral
- 10% to 20% bilateral
- Most 5-10 cm in greatest diameter
- US appearance depends on relative amounts of sebum, serous fluid, calcium, hair and fat
- Spectrum ranges from purely cystic to completely echogenic
- Most appear complex
- Usually contain <50% soft tissue elements
Malignant Ovarian Neoplasms

• 1%-2% of all malignant neoplasms in children less than 17 years of age
• Germ cell tumors- 60% to 90%
  – teratoma, dysgerminoma, endodermal sinus tumor, mixed malignant germ cell tumor, embryonal carcinoma
• Rare germ cell tumors (<1%)
  – choriocarcinoma, polyembryoma, leiomyosarcoma
• Stromal tumors-10% to 13%
  – Granulosa-theca cell, Sertoli-Leydig cell, undifferentiated
• Epithelial carcinomas-5% to 11%
Malignant Germ Cell Tumors

- Usually occur in postmenarcheal girls
- Asymptomatic pelvic or abdominal mass
- Typically larger than 10 cm diameter at diagnosis
- Intra-abdominal spread to lymph nodes and liver
- Dysgerminoma most common in childhood
  - 10% bilateral
- Malignant teratoma contains >50% soft tissue elements
- Other germ cell tumors may be homogeneous and echogenic or complex with cystic areas due to hemorrhage
Immature Teratoma
Dysgerminoma
Sex Cord-Stromal Tumors

• Low-grade malignancies arising from granulosa theca cells and Sertoli cells of embryonic gonad
• More common in premenarcheal girls
• Usually symptomatic
• Granulosa theca cell tumor causes isosexual precocity due to estrogen production
• Sertoli-Leydig cell tumor causes virilization due to androgen production
• Generally heterogeneous masses with cystic areas
• Metastases rare
  – peritoneal surfaces and liver
9-Year-Old Girl with Vaginal Bleeding: Juvenile Granulosa Cell Tumor
Other Ovarian Malignancies

• Epithelial tumors rare before puberty
  – serous and mucinous cystadenocarcinoma
• Difficult to differentiate benign from malignant and serous from cystic lesions on basis of US features
• Ovaries may be sanctuary for leukemia
• Site for metastatic spread from neuroblastoma, lymphoma and colon cancer
  – involvement usually asymptomatic
  – diagnosis made at autopsy
Summary

- US main imaging modality of pediatric female pelvis
- Useful in assessing anatomy and hormonal status of children with ambiguous genitalia, precocious puberty and primary amenorrhea
- Valuable in determining cause of pelvic pain and pelvic mass