Professional Ultrasound Services
244 20th Avenue San Francisco, CA 94121-2203
www.jimbaun.com (415) 751-4090

Fetal Anatomy: Genitourinary

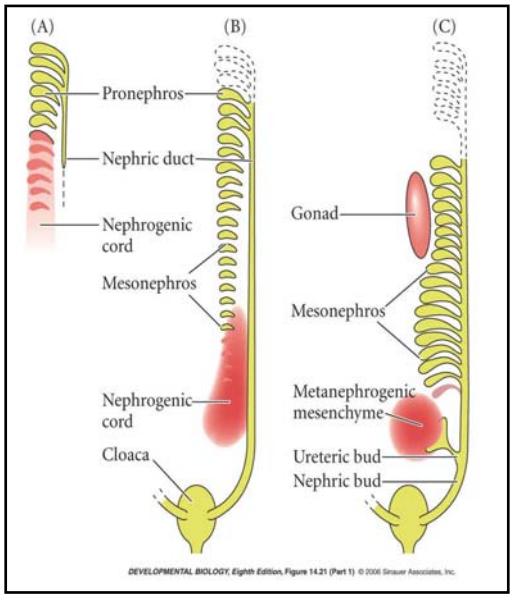
Embryology

Three sets of excretory organs or kidneys develop in human embryos:

- Pronephros
- Mesonephros
- Metanephros

The first set of kidneys - the ***pronephroi*** (plural of pronephros) - is rudimentary and nonfunctional. They are analogous to the kidneys in primitive fishes. The second set of kidneys - the ***mesonephroi*** - is well developed and functions briefly; they are analogous to the kidneys of amphibians. The third set of kidneys - the ***metanephroi*** - becomes the permanent kidneys.

Pronephros: These transitory, nonfunctional structures appear in human embryos early in the fourth week. They are represented by a few cell clusters and tortuous tubular structures in the rostral (neck) region of the embryo. The pronephric ducts run caudally and open into the ***cloaca***, the embryonic "sewer" (the word *cloaca* comes from the Latin expression describing a waste pipe that carries away sewage or surface water.) The rudimentary pronephroi soon degenerate; however, most of the pronephric ducts persist and are incorporated into next set of embryonic genitourinary structures.



Mesonephros: The next stage in renal development is the "middle kidneys". These large, elongated, excretory organs appear late in the fourth week after conception, caudal (toward the tail) to the rudimentary pronephroi. They are well developed and function as interim kidneys until the permanent kidneys develop. The mesonephric kidneys consist of glomeruli and mesonephric tubules. The tubules open into the mesonephric duct, which was originally the pronephric duct.

Metanephros: The mesonephroi, or permanent kidneys, begin to develop early in the fifth week post conception and begin functioning about 4 weeks later. They produce urine which continues throughout fetal life. Fetal urine is excreted into the amniotic cavity and mixes with the amniotic fluid. In later gestation, fetal

urine is the primary source of the amniotic fluid which is integral to normal growth and development. A mature fetus swallows several hundred milliliters of this amniotic fluid each day, which is absorbed by the intestine. The waste products are transferred through the placental membrane into the maternal blood for elimination. (*See Amniotic Fluid and Membranes).*

1. Background and Clinical Significance

a. Fetal period
   i. Most common site of embryological anomalies identified in utero are GU anomalies.
   ii. Prenatal ultrasound may help in identifying GU anomalies such as:
      1. Renal agenesis
      2. Partial renal malformation
      3. Duplications
      4. Obstructions
      5. Hydrometrocolpos

b. Neonatal period
   i. Sonographer most frequently finds anomalies in this age group.
   ii. Menstrual irregularities make many GU anomalies apparent

2. Expression of Gender

a. Gender determined at fertilization
   i. Female gamete contains X chromosome
   ii. Male contributes X chromosome = XX = girl.
   iii. Male contribute Y chromosome = XY = boy.

b. Primordial germ cells:
   i. First discernible in the embryo 21 days after fertilization.
   ii. Genital (gonadal) ridge: precursors of the ovaries in females; testes in males
   iii. Located antero-medial to the wolffian body in region where kidneys develop.
   iv. Urinary system and genital system are intimately associated in origin and development.

c. Inducer germ cells
   i. Germ cells migrate from yolk sac along dorsal membrane
   ii. Invade gonadal ridge by sixth week.
   iii. Indifferent gonad stage

3. Genital ducts

a. Formation of the Fallopian tubes
b. Formation of the broad ligament
c. Formation of the vagina

Normal Fetal GU Anatomy

Fetal kidneys and bladder are routinely studied during a prenatal sonographic screening examination at 17-21 weeks gestation. The adrenals can also be seen in most fetuses and should not be misinterpreted as a suprarenal mass unless other sonographic criteria are met.

Kidneys

Fetal kidneys can be identified as early as 12-14 weeks as two relatively sonolucent structures adjacent to spine in transverse sections. Echopenic (hypoechoic) renal pyramids are distributed evenly throughout parenchyma. Renal sinus fat is more echogenic than surrounding parenchymal tissue and can be seen in hilus of kidney. In many fetuses, the renal pelvis may contain a small amount of fluid which is a normal finding and is not indicative of obstructive uropathy unless other diagnostic criteria are met. Normal amounts of renal pelvis fluid are seen in 18% of fetuses after 24 weeks.

Normal Measurements:

- Renal length: ≈ 1mm/week of gestation
- Renal: AC = 0.3 throughout pregnancy
- Renal pelvis: measured AP in transverse section

AP Renal Pelvis Measurements	
≤ 5mm	Normal
5-10mm	Probably normal, follow-up
≥ 10mm	85% have anatomic anomaly

Age Related Renal Pelvis Measurements	
Weeks	AP Measurement (mm)
13-20	5
20-30	8
> 30	10

Bladder

The fetal bladder can be definitively identified by 16 - 20 weeks (frequently before that) and its sonographic demonstration is an important indicator of active renal function. In demonstrating the fetal bladder, the iliac wings and pelvic

vascular structures are important anatomic landmarks. Sitting in the midline, the normal fetal bladder can be seen as an anechoic structure in the anterior pelvis. The iliac vessels, which run postero-lateral to the bladder, can be reliably imaged with color Doppler imaging.

The urinary bladder is a dynamic structure that empties and fills in 30-45 minute cycles. The fetus micturates into the amniotic cavity and is responsible for virtually all of the amniotic fluid as gestation advances. The apparent absence of the bladder during the early minutes of an obstetric sonogram does not necessarily indicate abnormality. A recheck after 20 - 30 minutes frequently demonstrates the bladder as it fills with urine.

Adrenal

The adrenal glands are relatively large in fetus. About 90% of the gland is composed of cortex which quickly involutes after birth. The adrenals can be seen sonographically as an oval mass of echo-poor tissue lying superior to the kidney on sagittal plane of section. Transversely, the gland appears as a long, thin, echogenic line of medulla surrounded by the thicker sonolucent rim of cortex. The adrenal should be smaller than normal kidney. In fetuses with renal agenesis, the adrenals frequently opportunistically hypertrophy in to empty renal fossa.

Genitalia

Determination of the gender of a fetus may assist in the differential diagnosis of genitourinary anomalies and/or chromosomal syndromes.

Renal Agenesis

Unilateral Renal Agenesis: Very common 1: 1,000 births. Causes no symptoms and is not, of itself, a threat to the well-being of the fetus, neonate or adult. It usually goes undetected until adulthood as long as the single native kidney maintains adequate renal function.

Bilateral Renal Agenesis – (BRA) - Potter's Syndrome

Etiologies include:

- Chromosomal Disorders
- Autosomal Recessive Disorders
- Autosomal Dominant Disorders
- Non-mendelian Disorders.
- Sporadic Syndromes. Some teratogenic conditions, such as diabetes mellitus, have been associated with BRA.
- Non-syndromic BRA.

Embryology: BRA may result from
- Interruption of normal embryologic sequence from pronephros to metanephros.
- Failure of development of the ureteral bud.
- Lack of stimulation of nephron formation in the metanephric blastema.

Pathology: Classical Potter's syndrome consists of:
- Bilateral renal agenesis (BRA)
- Pulmonary hypoplasia
- Facial anomalies
- Patients present as small for dates

Associated anomalies: Associated with other chromosomal abnormalities including:
- Cardiovascular
- Musculoskeletal
- Central nervous system
- Gastrointestinal
- Others

Sonographic Findings:
- Absent fetal bladder (due to anuria).
- Severe oligohydramnios between 16-28 weeks should strongly suggest renal anomalies.
- Bilateral absence of fetal kidneys.

Renal Dysplasias

Potter syndrome and Potter phenotype is a complex of findings associated with a lack of amniotic fluid and with kidney failure that develops before an infant is born In ***Potter syndrome*** the primary defect is kidney failure that occurs before the baby is born, either from failure of the kidneys to develop (bilateral renal agenesis), or from other diseases that cause the kidneys to fail. The kidneys normally produce the amniotic fluid (as urine).

Potter phenotype describes a typical appearance that, in the newborn, is determined by the absence of amniotic fluid (oligohydramnios). In the absence of amniotic fluid, the infant is not cushioned from the walls of the uterus. The pressure of the uterine wall causes a typical facial appearance (Potter's facies) that includes widely separated eyes with epicanthal folds, broad nasal bridge, low set ears and receding chin.

In addition, because of limited space in the uterus, the limbs may be abnormal, or held in abnormal positions or contractures. Oligohydramnios also stops development of the lungs (hypoplastic lungs), so that at birth the lungs do not function properly.

Potter's classification:

- Type I Autosomal recessive (PKD): Infantile PKD
- Type II Multicystic dysplastic kidney disease (MDK)
- Type III Autosomal dominant polycystic kidney disease: Adult PKD
- Type IV Cystic renal dysplasia

Polycystic Kidney Disease (PKD)

PKD is an inherited kidney disorder characterized by multiple bilateral renal cysts, which cause enlargement of the total renal size while reducing, by compression, the functioning renal tissue. Clusters of fluid-filled sacs, called cysts, develop in the kidneys and, by compressing normal developing renal parenchyma, interfere with their ability to filter blood and produce urine.

The two major forms of polycystic kidney disease are distinguished by the usual age of onset and their pattern of inheritance.

Autosomal recessive type – (ARPKD) - Infantile Polycystic Kidney Disease Potter Type I

Autosomal recessive polycystic kidney disease (ARPKD) - sometimes called Infantile polycystic kidney disease (IPKD) - is an autosomal recessive disorder characterized by bilateral and symmetrical enlargement of the kidneys. Normal parenchyma is replaced by dilated collecting tubules. There is no increased amount of connective tissue. ARPKD is much rarer than the dominant genotype and is often lethal early in life. The signs and symptoms of this condition are usually apparent at birth or in early infancy. ***Autosomal recessive type diagnosed in utero.***

- Perinatal PKD: renal; failure in utero
- Neonatal: occurs within 1st month after birth. Death within 1 year.
- Infantile: occurs by 3-6 months with 20% renal involvement, hepatosplenomegaly. Progresses to renal failure, HTN, portal hypertension
- Juvenile: appears at 1-5 years. Less renal involvement. Course is similar to infantile PKD.

Autosomal dominant type – (ADPKD) - Adult Polycystic Kidney Disease Potter Type III

Autosomal dominant polycystic kidney disease (ADPKD) - sometimes called adult polycystic kidney disease (APKD) is an autosomal dominant disease characterized by replacement of renal parenchyma with multiple cysts of variable size due to dilatation of the collecting tubules and other tubular segments of the nephrons. ADPKD has signs and symptoms that typically begin in adulthood, although cysts in the kidney are often present from childhood. Autosomal dominant polycystic kidney disease can be further divided into type 1 and type 2, depending on which gene is mutated.

Individuals with ADPKD may be unaware of the presence of renal dysplastic disease well into adulthood. Cysts may also be found in the liver and pancreas. Clinical complications associated with ADPKD include:

- Hypertension
- Pain in the back or sides
- Hematuria
- Recurrent urinary tract infections
- Kidney stones
- Heart valve abnormalities
- Aortic or cerebral aneurysms

Sonographic Findings – in utero:

- Bilaterally enlarged, hyperechoic kidneys
- Increased Renal: AC ratio
- Oligohydramnios
- Absent, or small, fetal bladder.
- Loss of cortico-medullary differentiation
- The typical hyperechogenic texture is attributed to sound enhancement by the microscopic cystic structures present in the renal parenchyma.

Multicystic Dysplastic Kidneys (MKD) – Potter Type II

Multicystic kidney disease is a congenital renal disorder characterized by cystic lesions on the kidney. It is postulated that these cystic areas represent dilated collecting tubules caused by. Most cases are unilateral and asymptomatic. Bilateral MCDK is incompatible with life. Affected children are stillborn or die in the early postnatal period. Bilateral disease has been demonstrated in as many as 25% of cases of prenatally detected MCDK. MKD is present at birth (congenital), and the cysts are usually visible on prenatal sonographic examination. Two patterns of cysts have been described:

- “Classic” random distribution of cysts around the kidney.
- Discernible, dilated renal pelvis surrounded by the cystic structures.

Typically, the functional parenchyma in a multicystic kidney atrophies leaving grape-like cluster of cysts held together by loose fibrous tissue. There is no functional parenchyma. In the “classic” type, the calyceal drainage system is absent. Potential complications associated with MKD are hypertension and diminished renal function. When MKD affects both kidneys, it usually causes kidney failure shortly after birth.

Associated Anomalies:

Unilateral:

- CNS: hydrocephaly, anencephaly, spina bifida, myelomeningocele
- GI: esophageal atresia, imperforate anus, duodenal bands, tracheo-esophageal fistula

- Cardiac: ventricular septal defect
- Others: talipes equinovarus, hypospadias, vesical diverticulum, and patent urachus. Chromosomal anomalies can also occur

Bilateral:

- Cardiovascular malformations
- CNS abnormalities (anencephaly, hydrocephalus, iniencephaly, spina bifida, occipital meningocele)
- Others: Diaphragmatic hernia, cleft palate, microphthalmia, duodenal stenosis and imperforate anus, tracheo-esophageal fistula, and bilateral absence of radius and thumb.

Sonographic Findings

Unilateral

- Multiple cysts (as above) in one kidney.
- Normal appearing contralateral kidney.
- Bladder seen in presence of adequately functioning renal tissue

Bilateral:

- Multiple, peripheral, randomly located variable sized cysts
- Inability to visualize fetal bladder when renal insufficiency present
- Oligohydramnios possible

Solid Renal Masses

Several types of solid renal and adrenal masses may occur in utero such as: congenital mesoblastic nephroma, Wilm's tumor (nephroblastoma), and congenital adrenal neuroblastoma.

Sonographic Findings:

- Presence of a solid mass in the renal fossa
- Differentiation is usually not possible with ultrasound

Obstructive Uropathies

An obstructive uropathy is a condition caused by a blockage of urine flowing through the urinary tract. As anatomical structures dilated under the increased pressure of urine, renal function can be compromised. Chronic obstructions can result in permanent damage to the kidney affected.

The obstruction can occur anywhere along the genitourinary tract:

- Uretero-pelvic area
- Along the ureter
- Distal ureter’s insertion into the urinary bladder
- Urethral

Ureteropelvic Junction (UPJ) Obstruction

Obstruction of the urinary tract at the junction of the renal pelvis and the ureter causes urine to dilate the renal pelvico-calyceal system. The uretero-pelvic junction is a frequent site of obstruction of the urinary tract. Anatomic causes responsible for UPJ obstruction include:

- Fibrous adhesions, bands, kinks,
- Ureteral valves
- Aberrant lower pole vessels
- Abnormal ureteral insertion and unusual shapes of the pyelo-ureteral outlet.

Associated Anomalies:

- Vesicoureteric reflux
- Bilateral ureteral duplication
- Bilateral obstructed megaureter
- Contralateral non-functioning kidney
- Contralateral renal agenesis

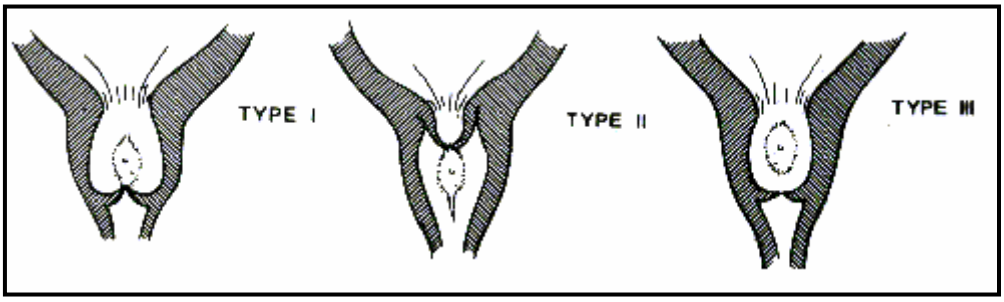
Sonographic Findings:

- Demonstration of a dilated renal pelvis
- Presence of variable caliectasis
- Thinning of renal cortex in chronic states

Posterior Urethral Valves

Lower urinary tract obstruction caused by a membrane-like structure in the posterior urethra. It affects male fetuses almost exclusively. Three types of valves have been described.

- Type I valves are folds distal to the verumontanum that insert into the lateral wall of the urethra.
- Type II valves are folds arising in the verumontanum, passing proximally to the bladder neck where they divide into fingerlike membranes.
- Type III valves consist of a diaphragm-like structure with a small perforation and are located distal to the verumontanum but not attached to it.



Associated Anomalies:

- Megacystis
- Megaureter
- Hydronephrosis
- Paraureteral diverticula
- Other anomalies of the genitourinary tract that are associated with PUV include duplication of the urethra, megalourethra, cryptorchidism, and hypospadias.

Sonographic Findings

- Marked hydronephrosis and a dilated, tortuous ureter
- Bladder wall thickening
- Male gender identified

Ectopic Ureterocele

Ectopic ureteroceles are congenital anomalies caused by abnormal embryogenesis and development of the intravesical ureter, the kidney, and the collecting system. It is a saccular out-pouching of the distal ureter into the urinary bladder producing progressive and self-obstructing cystic dilatation of the ureters and renal pelvis. It can lead to potential loss of renal function.

Sonographic Findings

- May be unilateral.
- Large, tortuous, fluid filled ureter.
- Level of obstruction may be seen occasionally.
- Ipsilateral hydronephrosis

Megaureter

Megaureter is a dilated ureter with or without dilatation of the renal pelvis and calyces. Its cause is frequently idiopathic. Ureteral enlargement may be caused by obstruction to the flow of urine, vesicoureteral reflux, or conditions in which neither obstruction nor reflux is present.

Primary megaureter: The term “primary” refers to a ureteral defect. In primary obstructive megaureter, the obstruction is at or just above the ureterovesical junction. The obstacle may be caused by stenosis of the ureteral valves, but the most common cause is the presence of a narrow juxtavesical ureteral segment that does not dilate or transmit the peristaltic wave. The pathologic basis for the obstruction may be segmental fibrosis or a localized absence of muscle.

Secondary Megaureter: “Secondary” refers to a pathologic process in another organ leading to dilatation of the ureter. In secondary obstructive megaureter, ureterectasis is due to an extrinsic pressure, such as by a vessel or tumor in the pelvis.

Associated Anomalies

- Unilateral renal agenesis,
- Complete or incomplete duplex system
- Ectopic kidney
- Contralateral cystic dysplastic kidney
- Horseshoe kidney
- Hirschsprung's disease.

Sonographic Findings

- Presence of tortuous anechoic or hypoechoic intra-abdominal structures that can be traced to the renal pelvis.
- Presence of a normal sized bladder which rules out lower urinary tract obstruction.
- Hydronephrosis may or may not be seen.

Bladder Outlet Obstruction (Megacystis)

Bladder outlet obstruction refers to any condition that blocks urine flow from the bladder. Posterior urethral valves are the most common cause. As discussed above, mucosal folds between the urethral wall and the verumontanum block outflow from the bladder causing excessive dilatation of the bladder and reflux up the ureters into the renal pelvis.

Sonographic Findings:

- Over-distended urinary bladder that does not empty within a 30 - 45 minute period.
- Bilateral dilated ureters and renal pelvi.
- Oligohydramnios.

Prune Belly Syndrome

- Triad syndrome:
- Anterior abdominal wall distention,
- Urinary tract obstruction,
- Cryptorchidism.

Often associated with severe GU anomalies particularly hydronephrosis. Theoretically, massive distention of urinary bladder and ureters causes pressure atrophy of abdominal wall. Decompression of hydronephrosis post-natally causes retraction and wrinkling of skin giving the appearance of a prune. Other causes that could lead to structural features of the syndrome such as ischemic abdominal wall muscles. 96 - 97% of cases occur in males.

Sonographic Findings:

- Abnormal compression of abdominal wall by fetal small parts.
- Movement of fetal abdominal wall when maternal abdomen is percussed.

Hydrocele

The accumulation of serous fluid around the testicle beneath the tunica vaginalis. A small amount of fluid in fetal scrotum is a normal finding. There are two types of hydrocele:

- ***Communicating hydrocele*** -- a scrotal fluid collection that communicates with the fluids of the abdominal cavity. A communicating hydrocele is caused by the failure of the processus vaginalis (the thin membrane that extends through the inguinal canal and descends into the scrotum) to close completely during prenatal development. If this membrane remains open, there is a potential for both a hernia and a hydrocele to develop.

- ***Non-communicating hydrocele*** -- a scrotal fluid collection present at birth or that develops years later for no obvious reason. A non-communicating hydrocele usually remains the same size or grows very slowly.

Sonographic Findings:

- Intra-scrotal fluid collection
- Dilated ureters
- Male genitalia