Diagnosis and Management of Urological Anomalies associated with Anorectal Malformations

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in general surgery

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بِسْمِ اللهِ الرَّحْمَنِ الرَّحِيمِ

قَالُوا سُبْحَانَكَ لَا عِلْمَ لَنَا إِلَّا مَا عَلَّمْتَنَا إِنَّكَ
أَنتَ الْعَلِيمُ الْحَكِيمُ

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List of Abbreviations

ARM   :   Anorectal malformations
CT scan :   Computerized tomography scan
MRU   :   Magnetic resonance urography
RNC   :   Renal nuclear cystography
UDS   :   Urodynamic studies
URA   :   Unilateral renal agenisis
VCUG   :   Voiding cystourethrogram
VUR   :   Vesicoureteric reflux

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Introduction

Anorectal malformations are congenital anomalies that occur in approximately 1 in 5000 live births .Anorectal malformations comprise a wide spectrum of diseases, which can affect boys and girls, and involve the distal anus and rectum as well as the urinary and genital tracts. Defects range from the very minor and easily treated with an excellent functional prognosis, to those that are complex, difficult to manage, are often associated with other anomalies, and have a poor functional prognosis *(Marc and Peña,2009)*.

Several classification systems of this defect have been proposed in the surgical literature.Anorectal malformation (ARM) may be classified as high and low anomalies according to their relation to the levator muscle complex. Associated anomalies are more common in high than in low anomalies. The true incidence of urological anomalies found in patient with ARM may only be detected by complete urological evaluation *(Endo, et al, 1999)*.

The incidence of urinary tract anomalies increased with a higher level of anorectal malformation *(Cho, et al, 2001)*. VUR and renal agenesis are the most common associated urinary tract anomalies with imperforate anus. In many studies, VUR was the most common anomalies in 25.7% patients with high lesion and 52.9% patients with urinary tract anomalies. VUR was reported the most common associated urinary tract anomaly with imperforate anus *(Boemers, et al, 1996)*.

The incidence of VUR in patients with anorectal malformations was different in various studies form 19% to 47.2% *(Metts, et al, 1997)*.

The current practice is to perform ultrasound which can be used to image the kidneys, bladder and to evaluate the

spinal cord for tethering *(Beals, et al, 1993)*. *Metts and Oemers in 1997,* found VUR in 32% of their cases. *Misra, et al, in 1996,* reported that 7.5% of patients with low deformity had VUR, but *Tohda and Moore, in 1995,* reported the incidence of VUR only in 0.7% and 5.4% of their patients. This wide variation in incidence of VUR is related to the different methods of study. In some studies VCUG was performed only when sonographic findings were abnormal *(Vasishali, et al, 2005)*.

In other study, hydronephrosis and renal agenesis were the most common abnormalities of the upper urinary tract, and neurovesical dysfunction is a frequent finding in children with anorectal malformations *(Bernard, et al, 1985)*.

It was found hydronephrosis and renal agenesis as two common of the upper urinary tract abnormalities. Neurovesical dysfunction commonly is associated with sacrospinal deformities. Some authors recommend evaluation of all patients with MRI, because spinal cord anomalies may occur without obvious sacrospinal anomalies *(Beals, et al, 1993)*.

Urodynamic studies (UDS) are reserved for those children with either a deformity of the spine or a spinal cord defect *(Bernard, et al, 1985)*. urological malformation may cause renal damage and chronic renal failure if not detected in time.Immediate care of the newborn with ARM centers around accurate classification of the anorectal anomaly and restoration of normal intestinal anatomy and function.However,not enough emphasis has been placed on the urgency of urological evaluation.Secondary importance is often placed on the evaluation and management of significant anomalies of the urinary tract *(Mark, et al, 1983)*.

Aim of the work

The aim of this work is to highlight through literature review urological anomalies associated with anorectal malformation regarding their incidence, types, presentation, work up and management.

Embryology of Urogenital Tract

The study of embryology provides a useful foundation for the understanding of definitive human anatomy and various congenital disease processes. During the past few decades, a torrent of molecular information and novel experimental techniques has revolutionized the field of embryology, and the knowledge base continues to expand at an exponential rate. From the urologic surgeon's perspective, however, the classical, descriptive aspects of anatomic embryology continue to serve as an important reference point from which various congenital problems are solved clinically *(Achermann et al., 2001)*

Kidney Development
Early Events

Mammals develop three kidneys in the course of intrauterine life. The embryonic kidneys are, in order of their appearance, the pronephros, the mesonephros, and the metanephros. The first two kidneys regress in utero, and the third becomes the permanent kidney. Embryologically, all three kidneys develop from the intermediate mesoderm. As the notochord and neural tube develop, the mesoderm located on either side of the midline differentiates into three subdivisions: paraxial (somite), intermediate, and lateral mesoderm. As the embryo undergoes transverse folding, the intermediate mesoderm separates away from the paraxial mesoderm and migrates toward the intraembryonic coelom (the future peritoneum). At this time, there is a progressive craniocaudal development of the bilateral longitudinal mesodermal masses, called nephrogenic cords. Each cord is seen bulging from the posterior wall of the coelomic cavity, producing the urogenital ridge *(Bartlett et al., 2002).*

Pronephros and Mesonephros

The mammalian pronephros is a transitory, nonfunctional kidney, analogous to that of primitive fish. In humans, the first evidence of pronephros is seen late in the 3rd week, and it completely degenerates by the start of the 5th week. The pronephros develops as five to seven paired segments in the region of the future neck and thorax. Development of the pronephric tubules starts at the cranial end of the nephrogenic cord and progresses caudally. As each tubule matures, it immediately begins to degenerate along with the segment of the nephric duct to which the tubules are attached. *(Batourina et al., 2001)*

The second kidney, the mesonephros, is also transient, but in mammals it serves as an excretory organ for the embryo while the definitive kidney, the metanephros, begins its development. There is a gradual transition from the pronephros to the mesonephros at about the 9th and 10th somite levels. Development of the nephric ducts (also called the Wolffian ducts) precedes the development of the mesonephric tubules. The nephric ducts can be seen as a pair of solid longitudinal tissue condensations at about the 24th day, developing parallel to the nephrogenic cords in the dorsolateral aspect of the embryo. Its blind distal ends grow toward the primitive cloaca and soon fuse with it at about the 28th day. This fused region later becomes a part of the trigone and posterior wall of the bladder. As the ducts fuse with the cloaca, they begin to form a lumen at the caudal end. This process of canalization then progresses cranially in a reverse direction, transforming the solid tissue condensations into the definitive nephric ducts with excretory capability. Soon after the appearance of the nephric ducts during the 4th week, mesonephric vesicles begin to form. Initially, several spherical masses of cells are found along the medial side of the nephrogenic cords at the cranial end. This differentiation progresses caudally and

results in the formation of 40 to 42 pairs of mesonephric tubules, but only about 30 pairs are seen at any one time because the cranially located tubules start to degenerate starting at about the 5th week. By the 4th month, the human mesonephros has almost completely disappeared, except for a few elements that persist into maturity as part of the reproductive tract. In males, some of the cranially located mesonephric tubules become the efferent ductules of the testes. The epididymis and vas deferens are also formed from the nephric (Wolffian) ducts. In females, remnants of cranial and caudal mesonephric tubules form small, nonfunctional mesosalpingeal structures called epoöphoron and paroöphoron *(Achermann et al., 2001).*

The mesonephric tubules differentiate into excretory units that resemble an abbreviated version of an adult nephron. Shortly after the cell clusters are formed, they develop lumens and take the shape of vesicles. As the vesicle elongates, each end curves in an opposite direction to form an S-shaped tubule. The lateral end forms a bud that connects with the nephric duct. The medial end lengthens and enlarges to form a cup-shaped sac, which eventually wraps around a knot of glomerular capillaries to form a renal corpuscle. The tuft of glomerular capillaries originating from a branch of the dorsal aorta invades the developing glomerulus, while an efferent arteriole empties into a subcardinal sinus *(Brophy et al., 2001).*

Metanephros

The definitive kidney, or the metanephros, forms in the sacral region as a pair of new structures, called the ureteric bud, sprouts from the distal portion of the nephric duct, and comes in contact with the condensing blastema of metanephric mesenchyme at about the 28th day. The ureteric bud penetrates the metanephric mesenchyme and begins to divide dichotomously. The tip of the dividing ureteric bud, called

ampulla, interacts with the metanephric mesenchyme to induce formation of future nephrons through mesenchymal-epithelial interaction. As the ureteric bud divides and branches, each new ampulla acquires a caplike condensation of metanephric mesenchyme, thereby giving the metanephros a lobulated appearance *(Brennan et al., 2003).*

The ureteric bud and metanephric mesenchyme exert reciprocal inductive effects toward each other, and the proper differentiation of these primordial structures depends on these inductive signals. The metanephric mesenchyme induces the ureteric bud to branch and divide, and, in turn, the ureteric bud induces the metanephric mesenchyme to condense and undergo mesenchymal-epithelial conversion. The nephron, which consists of the glomerulus, proximal tubule, loop of Henle, and distal tubule, is thought to derive from the metanephric mesenchyme; and the collecting system, consisting of collecting ducts, calyces, pelvis, and ureter, is formed from the ureteric bud.

In principle, all nephrons are formed in the same way and can be classified into fairly well-defined developmental stages **(Larsson et al., 1983).** The first identifiable precursors of the nephron are cells of metanephric mesenchyme that have formed a vesicle completely separate from the ureteric bud ampulla (stage I). Cells of the stage I renal vesicle are tall and columnar in shape and are stabilized by their attachments to the newly formed basement membrane. It has not yet established a contact with the ampulla of the ureteric bud. The stage I renal vesicle then differentiates into an S-shaped stage II nephron that connects to the ureteric bud. At this stage, the cup-shaped glomerular capsule is recognized in the lowest limb of the S-shaped tubule. The rest of the S-shaped tubule develops into the proximal tubule, the loop of Henle, and the distal tubule. When the cup-shaped glomerular capsule matures into an oval structure, the nephron has now passed

into stage III of development. Now the nephron can be divided into identifiable proximal and distal tubules. The stage IV nephron is characterized by a round glomerulus that closely resembles the mature renal corpuscle. The morphology of the proximal tubule resembles that of a mature nephron, whereas the distal segments are still primitive. In some species (e.g., the rodents) all stages of nephron development are present at birth, whereas in others (e.g., the humans) all nephrons at birth are in varying steps of stage IV. Initially, vessels are seen in the cleft between the lower and middle portion of the S-shaped tubule, and they quickly branch into a portal system. Mesenchymal cells that do not become tubular epithelium either give rise to interstitial mesenchyme or undergo programmed cell death. Overall, these events are reiterated throughout the growing kidney, so that older, more differentiated nephrons are located in the inner part of the kidney near the juxtamedullary region and newer, less differentiated nephrons are found at the periphery. In humans, although renal maturation continues to take place postnatally, nephrogenesis is completed before birth *(Cancilla et al., 2001)*
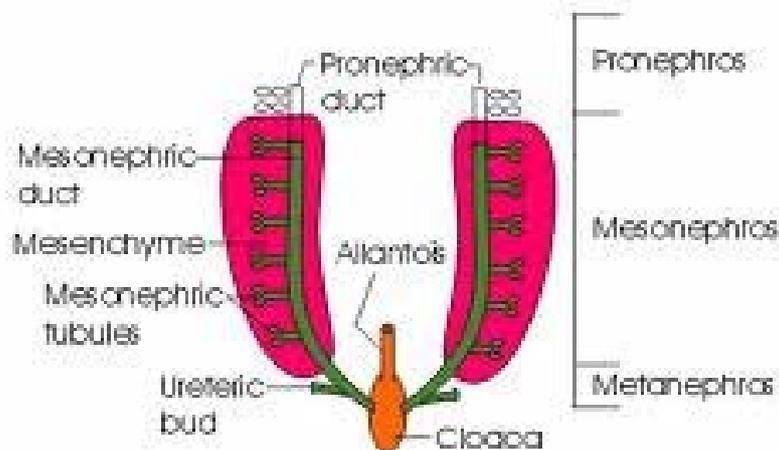

Fig. (1): The developing kidneys *(Cancilla et al., 2001)*