Ahmed H. Al-Salem

An Illustrated Guide to Pediatric Urology

Springer

An Illustrated Guide to Pediatric Urology

Ahmed H. Al-Salem

An Illustrated Guide to Pediatric Urology

Springer

Ahmed H. Al-Salem
Pediatric Surgery
AlSadik Hospital
Qatif
Saudi Arabia

ISBN 978-3-319-44181-8 ISBN 978-3-319-44182-5 (eBook)
DOI 10.1007/978-3-319-44182-5

Library of Congress Control Number: 2016957149

© Springer International Publishing Switzerland 2017
This work is subject to copyright. All rights are reserved by the Publisher, whether the whole or part of the material is concerned, specifically the rights of translation, reprinting, reuse of illustrations, recitation, broadcasting, reproduction on microfilms or in any other physical way, and transmission or information storage and retrieval, electronic adaptation, computer software, or by similar or dissimilar methodology now known or hereafter developed.
The use of general descriptive names, registered names, trademarks, service marks, etc. in this publication does not imply, even in the absence of a specific statement, that such names are exempt from the relevant protective laws and regulations and therefore free for general use.
The publisher, the authors and the editors are safe to assume that the advice and information in this book are believed to be true and accurate at the date of publication. Neither the publisher nor the authors or the editors give a warranty, express or implied, with respect to the material contained herein or for any errors or omissions that may have been made.

Printed on acid-free paper

Springer Cham Heidelberg New York Dordrecht London
This Springer imprint is published by Springer Nature

Preface

The field of pediatric urology is rapidly growing and currently it is considered as a subspecialty. Pediatric urologists and pediatric surgeons care for newborns, infants, and children with congenital and acquired urological conditions. I have written this book with more than 20 years' experience in the care of infants and children with urological conditions and hope it will help all those involved in the surgical care of infants and children with urological problems. This book is written in a simple way and easy to read. It covers most areas in the field of pediatric urology with emphasis on the most important points relevant to the patient's presentation, diagnosis, and management. This book is well illustrated and includes clinical, operative, radiological, and hand-drawn illustrations. I hope it will be useful to consultant pediatric urologists, pediatric surgeons, specialists, fellows, and residents. This book should be useful also to general practitioners, general surgeons, accident and emergency doctors, pediatricians, neonatologists, trainees, medical students, and nurses.

Qatif, Saudi Arabia — Ahmed H. Al-Salem

Acknowledgments

I would like to express special thanks of gratitude to my family who supported me and made this project possible. I would also like to thank all my patients and their families.

Contents

Congenital Urological Malformations

1

1.1 Introduction

- The urinary system is comprised of two kidneys, two ureters, a bladder and a urethra.
- The kidneys contains the nephrons which are responsible to filter the blood as it passes through the kidney.
- It is estimated that the kidneys will filter around 190 l of water every day from the blood. Most of the water from the blood that is filtered is reabsorbed into the body and the remaining water is excreted as urine.
- This water travels down the ureters to the bladder which acts as a storage area for the urine.
- When the bladder reaches a certain volume, nerves in the walls of the bladder are stimulated and urination happens.
- The kidneys also play an important role in red blood cell production. Erythropoietin, which is produced in the kidneys stimulates the production of the red blood cells.
- Congenital anomalies of the kidney and urinary tract (CAKUT) are common in children.
- They represent approximately 30 % of all prenatally diagnosed malformations.
- Congenital anomalies of the kidney and urinary tract occur in 3–6 per 1,000 live births.
- They account for the most cases of pediatric end-stage kidney disease (ESKD), and predispose an individual to hypertension and cardiovascular disease throughout life.
- They are responsible for 34–59 % of chronic kidney disease (CKD) and for 31 % of all cases of end-stage kidney disease (ESKD) in children.
- Congenital anomalies of the kidney and urinary tract comprise a wide range of structural and functional malformations that occur at the level of:
  - The kidney
  - Collecting system
  - Bladder
  - Urethra
- With improved prenatal screening, many cases of CAKUT are diagnosed by antenatal ultrasonography performed on 18–20 weeks of gestation.
- Most common antenatal manifestations of CAKUT include oligohydramnios or variations in gross morphology of the kidney, ureter, or bladder.
- CAKUTs anomalies can be:
  - Sporadic
  - Familial
  - Syndromic
  - Nonsyndromic
- Syndromic CAKUTs:
  - They develop in association with other additional congenital abnormalities outside of the kidney and urinary tract.
  - They manifest clinically recognizable features of a known syndrome.

© Springer International Publishing Switzerland 2017

A.H. Al-Salem, *An Illustrated Guide to Pediatric Urology*, DOI 10.1007/978-3-319-44182-5_1

- Nonsyndromic CAKUT:
  - Congenital structural anomalies confined only to the kidney and urinary tract.
- Genetic causes have been identified in association with the syndromic forms of congenital malformations.
- The spectrum of congenital anomalies includes more common anomalies such as vesicoureteral reflux and, rarely, more severe malformations such as bilateral renal agenesis.
- These congenital anomalies can be unilateral or bilateral, and sometimes different defects coexist in the same child.
- It is important to recognize and diagnose these anomalies early as early treatment will minimize renal damage, prevent or delay the onset of ESRD, and provide supportive care to avoid complications of ESRD.

1.2 Normal Embryology

- The two most common congenital bladder abnormalities are:
  - Bladder exstrophy
  - Congenital diverticula
- An exstrophic bladder is one that is open to the outside and turned inside out, so that its inside is visible at birth, protruding from the lower abdomen.
- A diverticulum is an extension of a hollow organ, usually shaped like a pouch with a narrow opening.
- The terminal portion of the hindgut is called the cloaca.
- The cloaca forms the future urinary and gastrointestinal tracts.
- The cloaca is formed early in fetal life by incorporation of the allantois to form a common distal channel for the primitive urinary and gastrointestinal systems.
- The most caudal portion of the cloaca is the cloacal membrane.
- This separates the cloaca from the amniotic cavity.
- During the fourth to seventh weeks of development, the cloaca is divided by the urorectal septum into anterior and posterior portions to form the urogenital sinus and anal canal, respectively.
- The urogenital sinus further differentiates into three anatomic components:
  - The vesical (cranial) portion
  - The pelvic (middle) portion
  - The phallic (caudal) portion
- The pelvic and phallic portions will form the urethra and genitals, respectively.
- The vesical portion forms most of the bladder and is continuous with the allantois.
- The allantois normally constricts to a thick fibrous cord, the median umbilical ligament, and extends from the apex of the bladder to the umbilicus.
- The trigone portion of the bladder is formed from the caudal ends of the mesonephric ducts, which are incorporated into the developing bladder wall.
- The ureters are formed from the ureteral buds.
- The ureteral bud is an outgrowth of the mesonephric duct near its entrance into the cloaca. This will elongate and develop into the ureter.
- The more distal ureteral bud undergoes a complex interaction with the primitive kidney to induce differentiation of the renal parenchyma and formation of the renal pelvis, calyces, and collecting tubules.
- As the kidneys develop and ascend, traction on the ureters causes the ureteral orifices to move superolaterally, resulting in an oblique course through the muscular wall at the base of the bladder.
- The kidneys are paired organs located retroperitoneally. Their vascular supply comes from the renal arteries, and they drain into the renal veins. Each kidney excretes into a ureter, which will in turn empty into the urinary bladder. Its functional unit is the nephron.
- The urogenital system arises from intermediate mesoderm which forms a urogenital ridge on either side of the aorta.
- The urogenital ridge develops into three sets of tubular nephric structures (from head to tail): the pronephros, the mesonephros, and the metanephros.
- During the development of the kidney, there are three main structures initially, which

derive from intermediate mesoderm. These structures are pronephros, mesonephros and metanephros.

- The development of the kidney proceeds through a series of three successive phases, each marked by the development of a more advanced kidney:
  - The pronephros
  - The mesonephros
  - The metanephros
- The pronephros:
  - This is the most immature form of the kidney.
  - During approximately day 22 (fourth embryonic week) of human gestation, the paired pronephros appear towards the cranial end of the intermediate mesoderm.
  - It develop as a condensation of intermediate mesoderm in the lower cervical and upper thoracic regions extending to the cloaca, and almost entirely regresses in gestational week 4.
  - It appears as seven to ten cell groups and arrange themselves in a series of tubules called nephrotomes and join laterally with the pronephric duct.
  - The pronephric duct, which arises from dorsal and caudal evaginations of the pronephros, is preserved and ultimately will give rise to the mesonephric duct.
- The mesonephros:
  - The mesonephros begins to develop as the pronephros is regressing (fourth week).
  - The development of the pronephric duct proceeds in a cranial-to-caudal direction.
  - As it elongates caudally, the pronephric duct induces nearby intermediate mesoderm in the thoracolumbar area to become epithelial tubules called mesonephric tubules.
  - It starts as a series of S-shaped tubules.
  - Each mesonephric tubule receives a blood supply from a branch of the aorta, ending in a capillary tuft analogous to the glomerulus of the definitive nephron.
  - The tubules around the glomerulus will form a Bowmann's capsule around the capillary tuft.
  - Together this will lead to the formation of a renal corpuscle allowing for filtration of blood.
  - Laterally, the tubule enters the mesonephric collecting duct (wolffian duct).
  - This filtrate flows through the mesonephric tubule and is drained into the continuation of the pronephric duct, now called the mesonephric duct or Wolffian duct.
  - The nephrotomes of the pronephros degenerate while the mesonephric duct extends towards the most caudal end of the embryo, ultimately attaching to the cloaca.
  - These mesonephric tubules carry out some kidney function at first, but then many of the tubules regress. However, the mesonephric duct persists and opens to the cloaca at the tail of the embryo.
  - In both sexes, the ureters, renal pelvis, and bladder trigone are derived from the mesonephric duct.
  - In males, the mesonephric duct also gives rise to the vasa deferentia, epididymides, and seminal vesicles; the former is part of the duct itself, while the latter two structures arise as a result of ductal dilatation or outpouching.
  - Once the mesonephric duct comes in contact with the cloaca at the caudal aspect of the embryo, it then grows cranially as the ureteric bud until it comes in contact with the metanephric mesenchyme, forming the metanephros.
  - The ureteric bud and metanephric mesenchyme reciprocally induce growth, forming the kidney.
- The metanephros:
  - This gives rise to the definitive kidney.
  - The metanephros develops from several components:
    - An outgrowth of the caudal mesonephric duct
    - The ureteric bud
    - A condensation of nearby renogenic intermediate mesoderm, the metanephric blastema
  - The metanephros appears during the fifth week of intrauterine life.

  - The kidney has two parts:
    - The collecting system
    - The excretory system
  - The collecting system develops from the ureteric bud which is an outgrowth of the mesonephric duct.
    - The ureteric bud penetrates the metanephric tissue.
    - The bud then dilates, forming a renal pelvis.
    - The renal pelvis will differentiate into the major calyces.
    - The major calyces will further differentiate and subdivide for 12 or more generations to form the minor calyces.
    - By the fifth generation, the renal pyramids are formed.
  - The excretory system is formed because a metanephric tissue cap is induced by the collecting tubules to form renal vesicles.
  - The vesicles form an s-shaped tubules which is covered in capillaries, giving rise to glomeruli.
  - The tubules and the glomeruli form the nephron.
  - Continues expansion of the tubules will form the convoluted tubules of the kidney and the loop of Henle.
  - At birth, approximately 750,000–1 million nephrons are present in each kidney; postnatally, renal size may increase, owing to elongation of the proximal convoluted tubules.
  - With differential longitudinal growth of the embryo, the kidney ascends from its initial location in the pelvis to its final location in the upper retroperitoneum.
  - During ascent, transient blood vessels serially arise and degenerate; these arteries persist in ectopic kidneys as well as in some orthotopic renal units.
  - Concurrently, the kidneys rotate around their vertical and horizontal axes so that their final orientation is one in which the upper poles are slightly more medial and anterior than the lower poles.
- The urogenital sinus can be further subdivided into cranial (future bladder) and caudal (future prostate, urethra, and external genitalia) portions.
- The vesical epithelium is entirely derived from the endodermal layer of the urogenital sinus.
- The mesonephric duct gives rise to the ureter.
- With continued caudal growth of the embryo, the proximal end of the mesonephric duct is progressively absorbed caudally and the common portion of the mesonephric duct is absorbed into the bladder trigone and urogenital sinus.
- The discrete branches of the mesonephric duct which will become the male genital ducts and ureters becomes distinct entities attached to the urogenital sinus.
- The nonepithelial layers of the detrusor (nontrigone) portion of the bladder arise from condensations of splanchnic mesenchyme.
- The lumen of the allantois, which connects the bladder and the anterior abdominal wall, closes over time, yielding the urachus. Over time, the urachus becomes more fibrotic and becomes the median umbilical ligament.
- The prostate gland develops around 9–11 gestational weeks from the urogenital sinus, as endoderm invaginates into surrounding mesenchyme.
- Prostate development is an androgen-dependent process.
- It appears that the mesenchyme, rather than the endoderm, must be androgen-sensitive in order for normal prostatic development to occur.
- The urethra develops from the urogenital sinus, with endoderm giving rise to the epithelium and splanchnic mesenchyme giving rise to the surrounding soft tissue.
- In males, the most distal part of the urethra (the glanular portion) appears to arise from an ectodermal invagination which then joins with the endodermal epithelium of the proximal urethra to create a continuous channel.

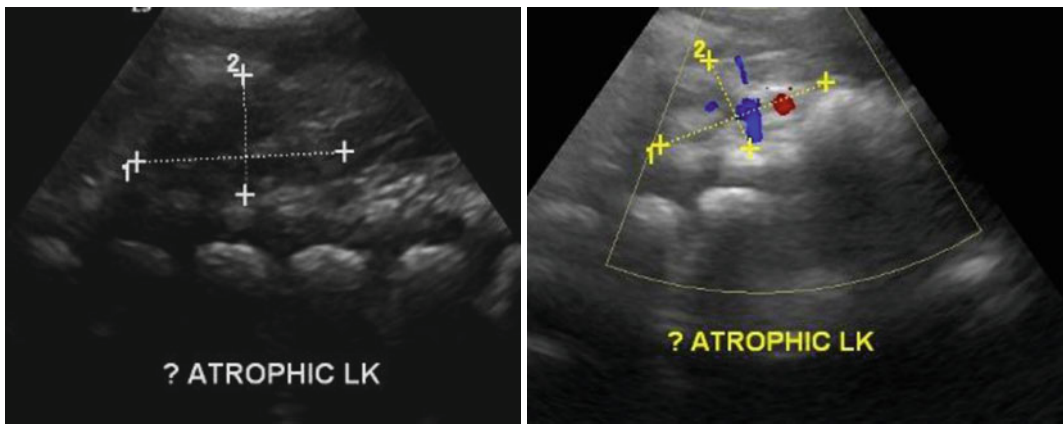
1.3 Abnormalities of the Kidney

- Normal renal development depends upon the interaction between the ureteric bud and metanephric mesenchyme, which induces organogenesis resulting in the formation of the 600,000–2 million nephrons and the collecting system of each kidney.

- The kidney is the most common site of congenital abnormalities.
- Renal malformations are often associated with other congenital defects such as a grossly deformed pinna with ipsilateral abnormalities of the facial bones.

1.3.1 Renal Agenesis

- Renal agenesis is a congenital malformation in which one (unilateral) or both (bilateral) fetal kidneys fail to develop.
- Renal agenesis can be unilateral or bilateral but almost always unilateral.
- Unilateral renal agenesis is a relatively common congenital urinary malformation.
- It is usually diagnosed during fetal ultrasonography or incidentally on ultrasound done for other reasons.
- Some cases of unilateral renal agenesis may represent involution of a previous multicystic disease of the kidney (Figs. 1.1, 1.2, and 1.3).
- Up to 40 % of women with a urogenital tract anomaly also have an associated renal tract anomaly.
- Adults with unilateral renal agenesis have considerably higher chances of hypertension.
- The annual incidence of unilateral renal agenesis is estimated at around 1 in 2,000 live newborns.
- The male to female ratio is around 1.2:1.
- Approximately 56 % of unilateral renal agenesis occurs on the left side.
- Most patients with unilateral renal agenesis are asymptomatic if the other kidney is fully functional.
- However, hypertension, proteinuria and renal failure may develop in the long term follow-ups (20–50 % of cases at the age of 30), which may be based on glomerular hyperfiltration.
- Due to this increased risk of hypertension and/or proteinuria, long-term follow-up of these patients is important.
- Associated malformations:
  - Unilateral renal agenesis may be an isolated congenital malformation or may be associated with chromosomal abnormalities or a variety of nonchromosomal syndromes including the VACTERL and MURCS associations.
  - Congenital cardiac malformations are the most common malformations associated with unilateral renal agenesis.
  - Girls with unilateral renal agenesis should have a pelvic ultrasound to look for abnormalities in the müllerian structures.
  - Vesicoureteral reflux is the most common abnormality noted in the contralateral kidney.
  - It is associated with an increased incidence of Müllerian duct abnormalities, and can



Figs. 1.1 and 1.2 Abdominal ultrasound showing left atrophic kidney. This is most likely following involution of a previous multicystic kidney

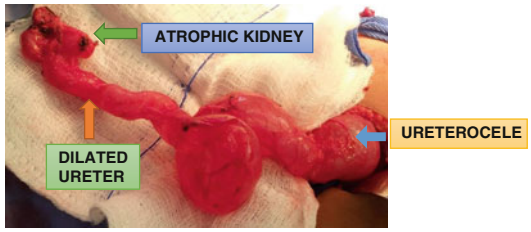


Fig. 1.3 Intraoperative photograph showing a very small atrophic kidney. Note also the massively dilated ureter secondary to an obstructive ureterocele

be a cause of infertility, hematocolpos, increased need for Caesarean sections, or other problems.
  - Herlyn-Werner-Wunderlich syndrome is one such syndrome in which unilateral renal agenesis is combined with a blind hemivagina and uterus didelphys.
  - Renal agenesis is occasionally associated with genital tract anomalies on the same side such as seminal vesicle hypoplasia and absence of the vas deferens.
  - Other anomalies occur in up to 40 % of patients, mainly cardiac (such as atrial or ventricular septal defects) and gastrointestinal (such as anorectal agenesis).
- Renal agenesis results from a development failure of the ureteric bud and the metanephric mesenchyme.
- Unilateral renal agenesis can be caused by mutations in many genes, such as *RET* (10q11.2), *BMP4* (14q22-23), FRAS1 (4q21.21), FREM1 (9p22.3 or UPK3A (22q13.31), PAX2 (10q24.31), HNF1B (17q12), DSTYK (1q32).
- Unilateral renal agenesis can occur as part of multi-organ syndromes, several of which have defined genetic bases, including Kallmann syndrome, branchio-oto-renal syndrome, diGeorge syndrome, Fraser syndrome, MURCS association, Poland syndrome, renal cysts and diabetes syndrome, and Williams-Beuren syndrome.
- Maternal diabetes mellitus or use of specific drugs during pregnancy can also result in renal agenesis.
- Prenatal suspicion of unilateral renal agenesis is confirmed by postnatal ultrasound showing an empty renal fossa, followed by renography to confirm the presence of a solitary functioning kidney.
- The size of the solitary functioning kidney is increased in the majority of patients.
- A voiding cystourethrogram should be considered in order to detect vesicoureteral reflux (VUR).
- The differential diagnoses include:
  - Extreme unilateral renal dysplasia
  - Involuted multicystic dysplastic kidney
  - Renal ectopia
- In most familial cases, unilateral renal agenesis is inherited in an autosomal dominant manner with incomplete penetrance.
- Unilateral renal agenesis can occur with dysplasia or hypoplasia of the solitary functioning kidney which makes the prognosis more serious.

1.3.2 Renal Hypoplasia

- Renal hypoplasia is a common congenital malformations.
- It is poorly understood and commonly used to describe a congenitally small kidneys with a reduced number of nephrons but normal architecture.
- There are however two distinct conditions:
  - Oligomeganephronia:
    - This is a type of renal hypoplasia that results from a quantitative defect of the renal parenchyma with a reduced number of nephrons.
  - Simple renal hypoplasia:
    - This is characterized by reduction in the renal mass but the number of nephrons is normal.
- It was estimated that renal hypoplasia affect about 2.2 % of the population.
- The exact incidence of renal hypoplasia is not known but it is estimated to occur 1 in 400 live births.
- This however is not a true incidence of pure renal hypoplasia because the majority of congenitally small kidneys also exhibit evidence of renal dysplasia.

- Severe bilateral reductions in nephron numbers that are characteristic of renal hypoplasia/dysplasia are the leading cause of childhood end stage renal disease.
- A much less reduction in nephron number caused by mild bilateral renal hypoplasia, have been associated with the development of adult-onset hypertension and chronic renal failure.
- The diagnosis of hypertension in patients with unilateral hypoplasia/dysplasia is an indication for nephrectomy.
- Oligomeganephronia:
  - This results from arrested development of the metanephric blastema at 14–20 weeks' gestation, with subsequent hypertrophy of glomeruli and tubules in the kidney.
  - This hypertrophy and hyperfiltration results in further nephron injury and sclerosis. Eventually, this progressive loss of nephrons leads to end-stage renal disease (ESRD).
  - Oligomeganephronia is usually found in infants in their first year of life and presents with anorexia, vomiting, and failure to thrive.
  - After the first year of life, individuals with oligomeganephronia most often present with short stature, polyuria and polydipsia, or proteinuria.

1.3.3 Supernumerary Kidneys

- Supernumerary kidneys are a rare congenital anomaly of the urogenital system, where there are one or two accessory kidneys.
- A third kidney may be confused with the relatively common unilateral duplication of the renal pelvis.
- Supernumerary kidney results from the aberrant division and splitting of the nephrogenic blastema into two metanephric blastemas or from separate metanephric blastemas into which partially or completely reduplicated ureteral stalks enter to form separate capsulated kidneys.
- The end result is two kidneys in association with a partially or completely duplicated ureteral bud.
- In some cases the separation of the reduplicated organ is incomplete forming fused supernumerary kidney.
- Associated anomalies:
  - Urogenital anomalies such as fusion anomalies, ectopic ureteric opening, vaginal and uretral atresia, urethral or penile duplication.
  - Non-urogenital anomalies such as coarctation of aorta, imperforate anus, ventricular septal defect and meningomyelocele.
- Supernumerary kidneys are most commonly located on the left side of the abdomen.
- A supernumerary kidney may be of same size as, larger than, or more commonly smaller than the usual kidney.
- It functions normally, possess a normal shape and capsule, and is either not attached to or loosely attached to the normal kidney but in an abnormal location.
- A supernumerary kidney may be located in front, below, above, or behind the normal kidney. They can also be found in the iliac region or anterior to the sacral promontory.
- The supernumerary kidney is thought to be an accessory organ with a separate arterial supply, venous drainage, collecting system, and distinct encapsulated tissue.
- It may have either a separate ureter or more commonly bifid ureters (50%). Rarely the ureter of the supernumerary kidney may have an ectopic opening.
- Symptoms have been noted in approximately two-thirds of the reported cases of supernumerary kidney. When symptomatic they may cause fever, pain, or palpable abdominal mass.
- The diagnosis of supernumerary kidney can be made by:
  - IVU
  - Ultrasonography
  - Nuclear scintigraphy
  - CT
  - MRI
- Bilateral supernumerary kidney is extremely rare.
- Surgery is indicated when supernumerary kidneys are affected by pathologic conditions and become symptomatic

1.3.4 Renal Dysplasia and Multicystic Kidney (Figs. 1.4, 1.5, 1.6, 1.7, 1.8, 1.9, 1.10, 1.11, and 1.12)

- Renal dysplasia is characterized by the presence of malformed renal tissue elements, including primitive tubules, interstitial fibrosis, and/or the presence of cartilage in the renal parenchyma.
- Multicystic dysplastic kidney (MCDK), a variant of renal dysplasia, is one of the most frequently identified congenital anomalies of the urinary tract.
- Other terms used to describe this condition include multicystic kidney and multicystic renal dysplasia.
- Multicystic kidney of the newborn is normally seen in only one kidney as an irregularly lobulated mass of cysts and usually absent or atretic ureter.
- Multicystic dysplastic kidney is the most common cause of an abdominal mass in the newborn and is the most common cystic malformation of the kidney in infancy.
- Renal dysplasia is considered the leading cause of end-stage renal disease in children.
- Multicystic dysplastic kidney is characterized by:
  - The presence of multiple, noncommunicating cysts of varying size separated by dysplastic parenchyma and the absence of a normal pelvocaliceal system.
  - It is associated with ureteral or ureteropelvic atresia
  - The affected kidney is nonfunctional
  - Frequently, it is associated with contralateral abnormalities, especially ureteropelvic junction obstruction.
- Dysplasia of the renal parenchyma is seen with urethral obstruction or reflux present early in pregnancy, or obstructed ureter.

1.3.5 Polycystic Kidney Disease

- Polycystic kidney disease, also known as polycystic kidney syndrome is a genetic disorder in which abnormal cysts develop and grow in the kidneys.
- It is characterized by multiple cysts typically involving both kidneys.
- About 15–17 % of cases initially present with multiple cysts in one kidney, progressing to bilateral disease in the majority.
- Polycystic kidney disease is one of the most common hereditary diseases.
- It is the cause of nearly 10 % of end-stage renal disease and affects males, and females equally.
- Signs and symptoms of polycystic disease include:
  - High blood pressure
  - Headaches
  - Abdominal pain
  - Hematuria
  - Polyuria
  - Pain in the back
- There are two types of polycystic kidney disease:
  - Autosomal dominant polycystic kidney disease (ADPKD)
  - Autosomal recessive polycystic kidney disease (ARPKD)
- Autosomal dominant polycystic kidney disease (ADPKD):
  - This is the most common of all the inherited cystic kidney diseases
  - The incidence is 1:500 live births
  - It is estimated that about 10 % of end-stage kidney disease (ESKD) patients being treated with dialysis were initially diagnosed and treated for ADPKD.
- There are three genetic mutations with similar phenotypical presentation:
  - PKD-1
  - PKD-2
  - PKD3
- Gene PKD1 is located on chromosome 16 and codes for a protein involved in regulation of cell cycle and intracellular calcium transport in epithelial cells.
- Gene PKD1 is responsible for 85 % of the cases of ADPKD.
- Gene PKD2 is located on chromosome 4 and codes for a group of voltage-linked calcium channels.