
International Foundation for Pediatric Imaging Aid

Genetic and non-genetic cystic kidney

An overview, morphological classification and diagnostic value of imaging procedures

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Aid

Introduction

- Renal cystic disease comprises a mixed group of heritable developmental and acquired diseases.
- The ultimate diagnosis of many of the “cystic kidneys” requires clinical, genetic, radiological and pathological information.
- The diagnosis is important for prognosis, treatment and genetic counseling.

Introduction

- Because of the diversity in etiology, histology and clinical presentation of “cystic kidney” a single scheme classification is not accepted.
- From practical point of view the “cystic kidney diseases” should be divided in genetic and non-genetic groups.

Congenital cystic renal diseases

Genetic cystic kidney

1. Autosomal dominant polycystic kidney disease (ADPKD)
2. Autosomal recessive polycystic kidney disease (ARPKD)
3. Tuberous sclerosis complex
4. Medullary cystic kidney and juvenile nephronophitisis

Non-genetic cystic kidney

1. Multicystic dysplastic kidney (MCDK)
2. Malformation syndromes with renal cysts
3. Multicystic nephroma
4. Medullary sponge kidney

Congenital cystic renal diseases

- This presentation includes the clinical and morphological findings of 132 infants and children suffering from cystic kidney disease, following a retrospective study.
- In a large number of neonates a cystic kidney was suspected by antenatal sonography.
- Postnatal ultrasound was performed routinely as the first diagnostic procedure and repeated frequently to follow the morphological changes.
- Additional imaging modalities were used depending on clinical or ultrasound findings and to image other affected organs.

Autosomal Dominant Polycystic Kidney Disease (ADPKD)

- Frequency 1 : 400/1000 life birth.
- Mutation 85% PKD1 gene, Ch.L: 16p.3.3. 15% PKD2 gene, Ch.L.: 4q21.2.
- 25 patients were reviewed (M=14, F=11; age range 1 d- 13 yrs)

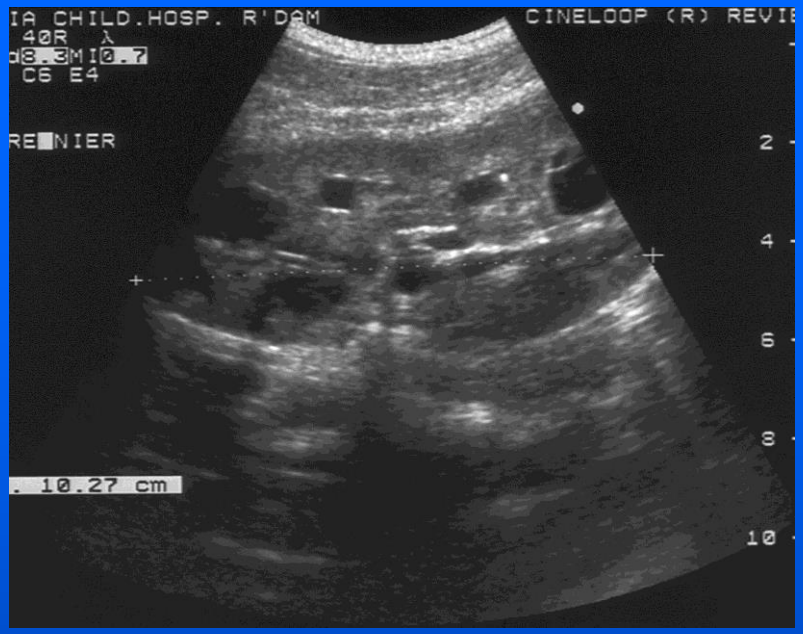
Clinical manifestation:

Renal:

Decrease in renal concentrating ability, microalbuminuria, cyst infection, renal failure, chronic abdominal pain (1), hypertension (1), hematuria (2),.

Extrarenal:

Hepatic cysts (2), intracranial aneurysms, other vascular abnormalities, cardial disease, diverticular disease, hernias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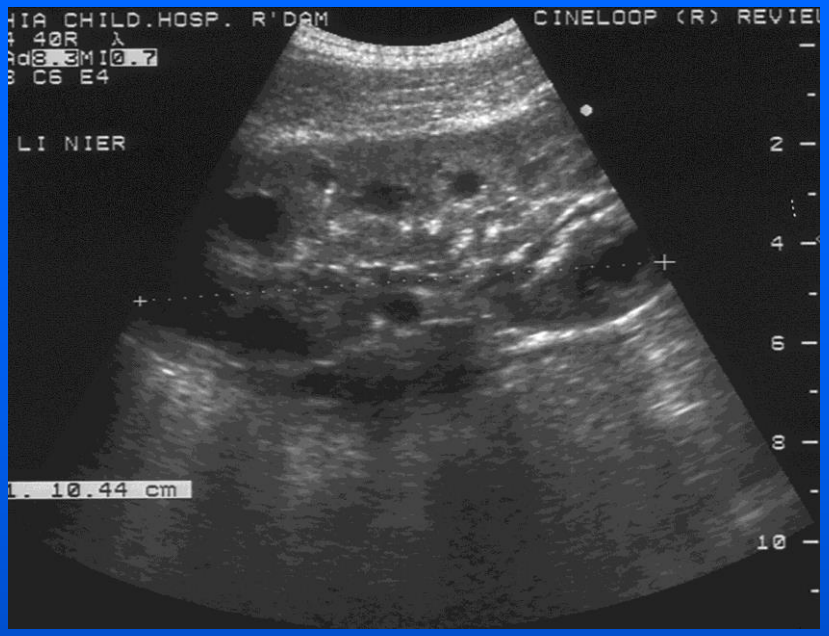
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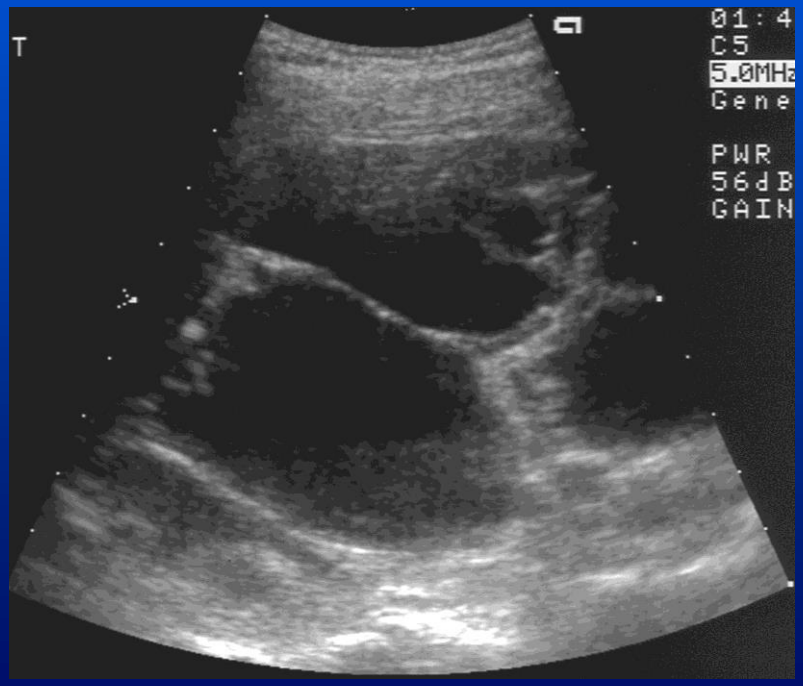
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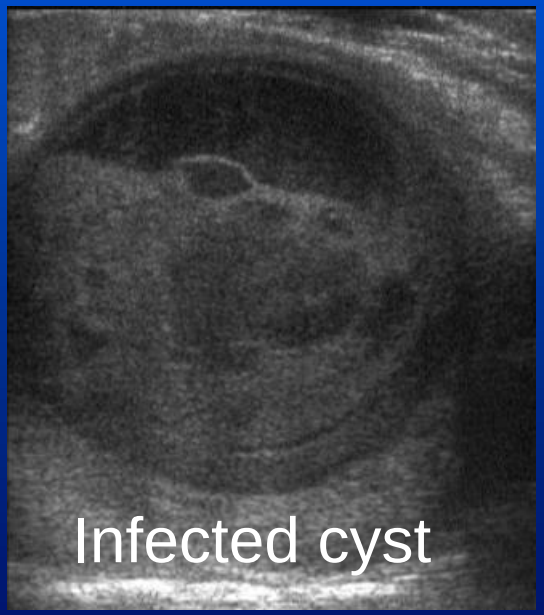
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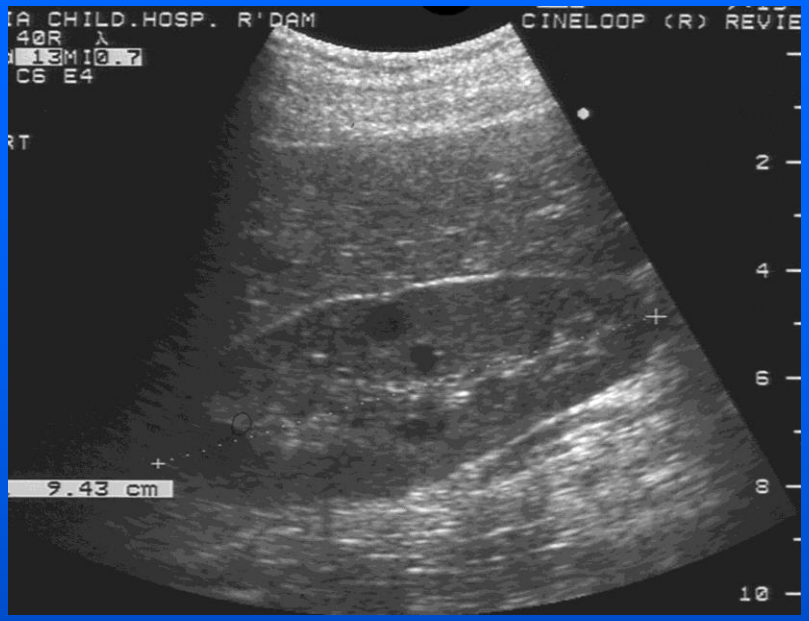
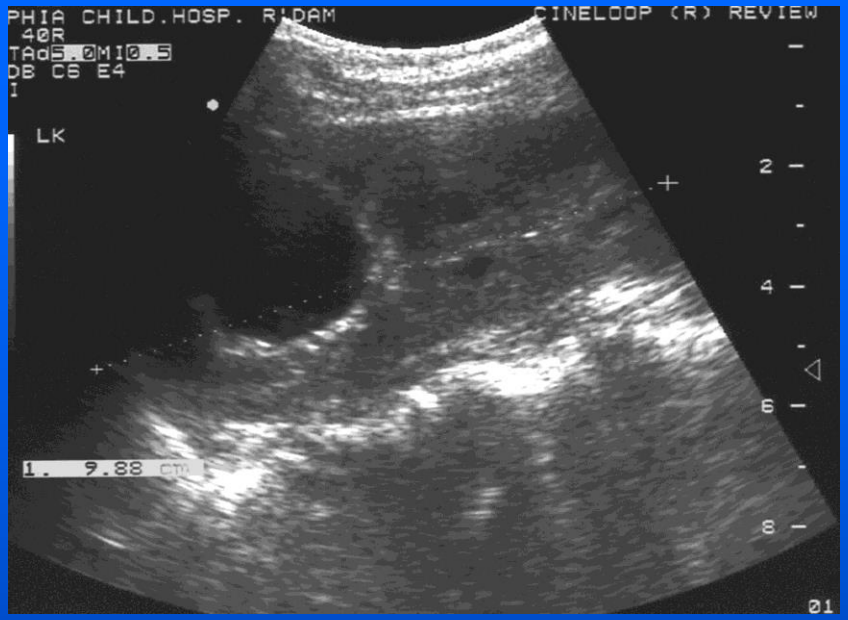
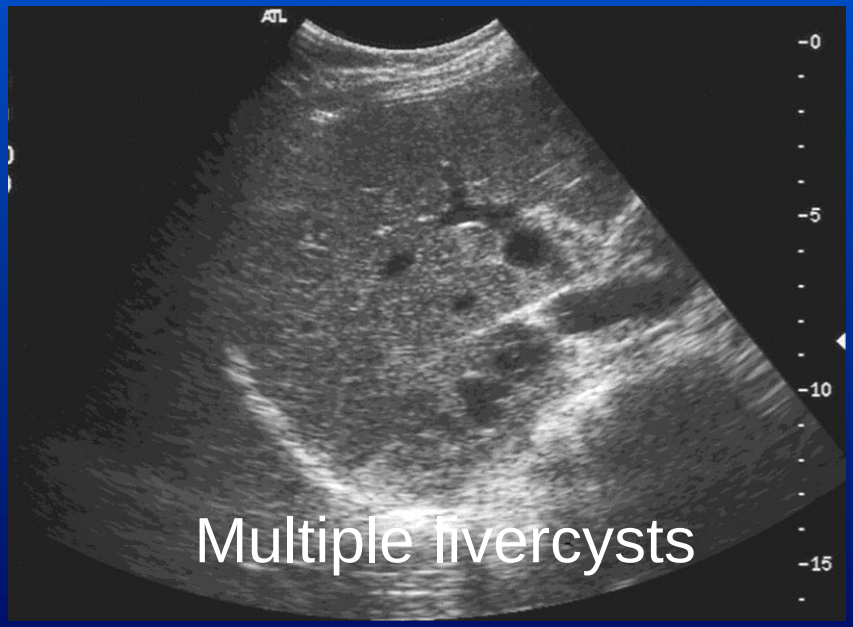






Multiple livercysts

Autosomal Recessive Polycystic Kidney Disease (ARPKD)

- Affects 1 : 10.000-40.000 individuals.
- Mutation gene, Ch.L.: 6p21.1.
- 11 patients were reviewed (M=2, F=9; age range 2 d-3 yrs)

Clinical manifestation:

Prenatally:

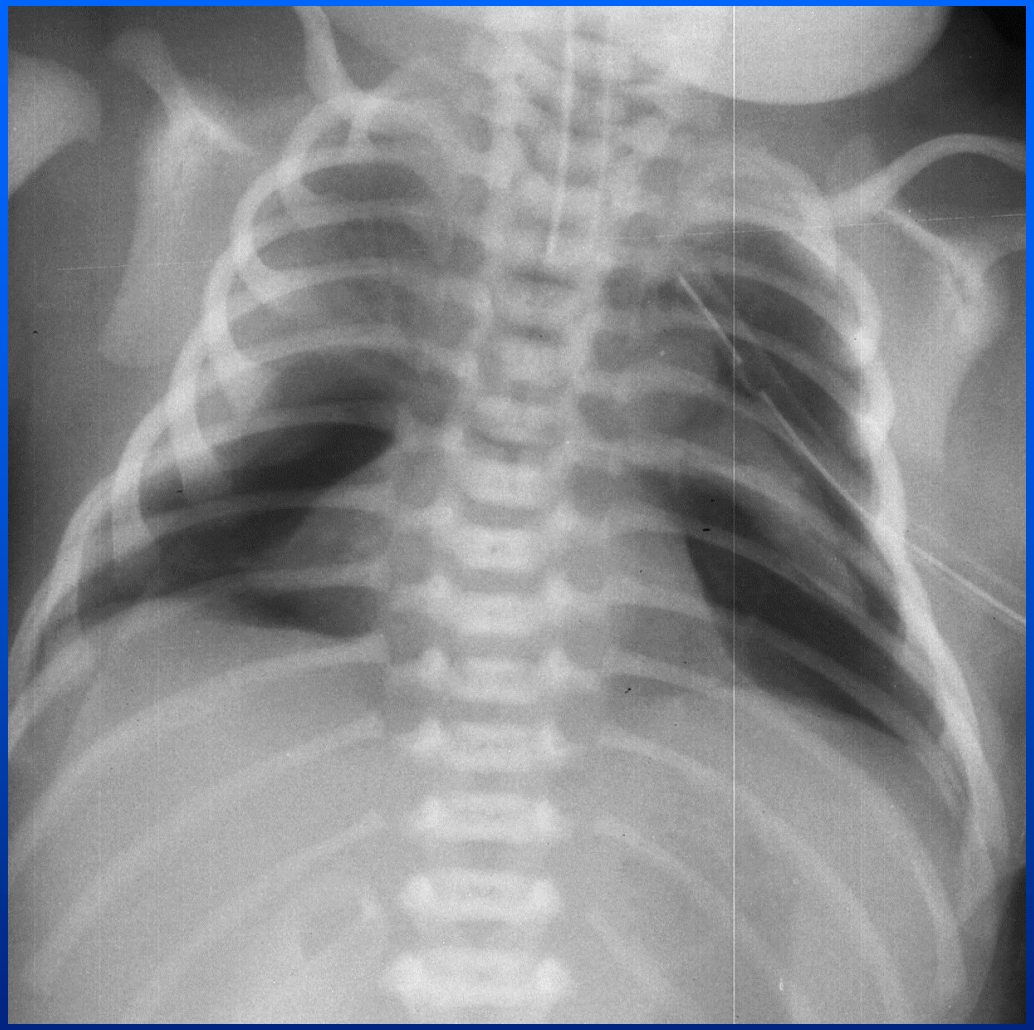
Oligohydramnion, enlarged kidneys, lung hypoplasia.

Infancy:

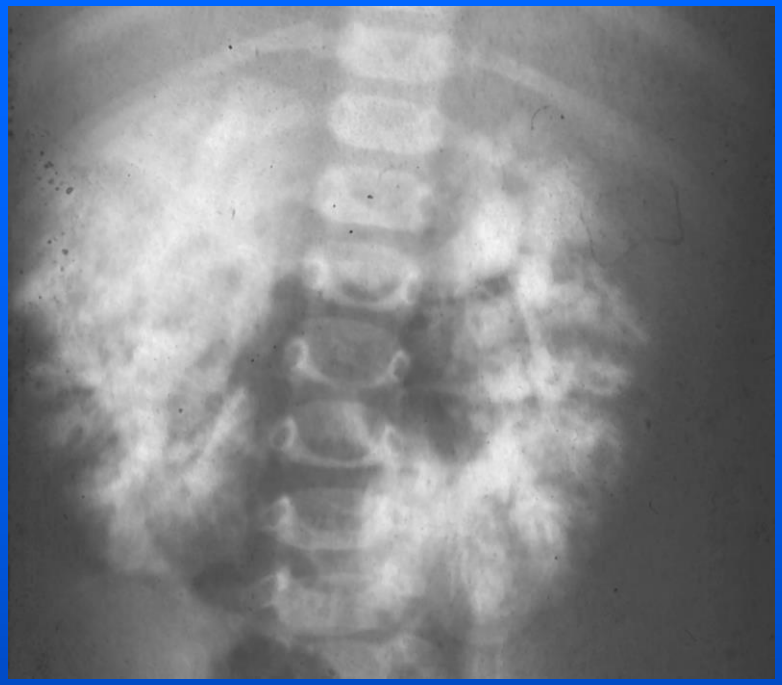
Pneumothorax (1), hypertension, renal failure (5)

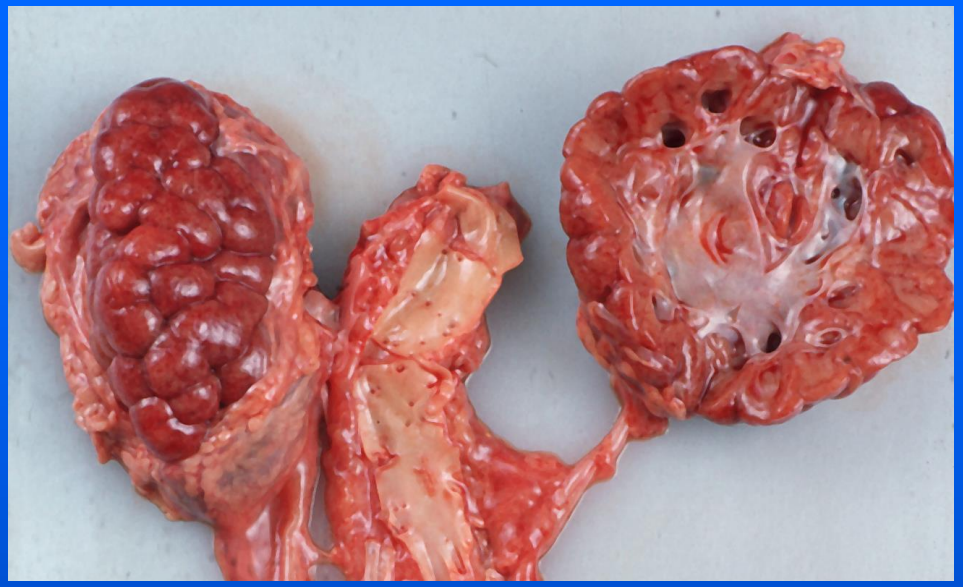
Older children:

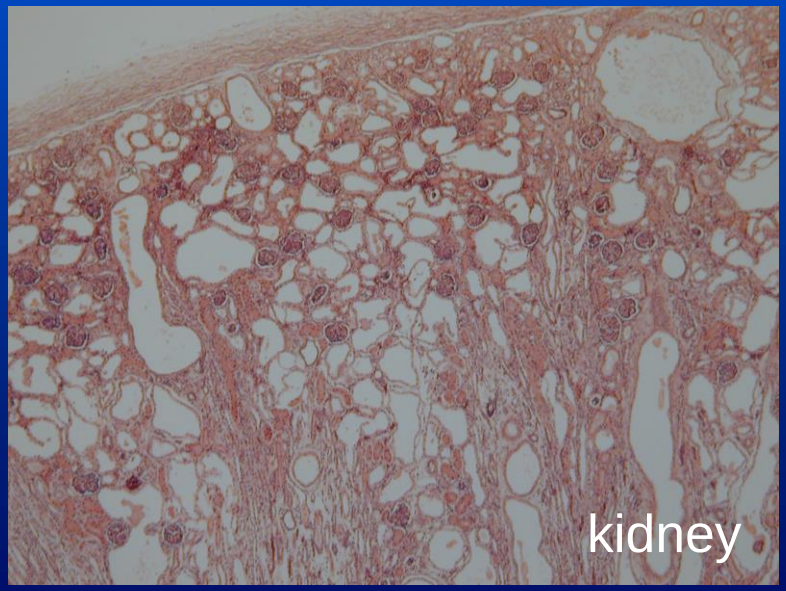
Hepatic fibrosis (6), portal hypertension, Caroli's disease (3)



Pneumothorax



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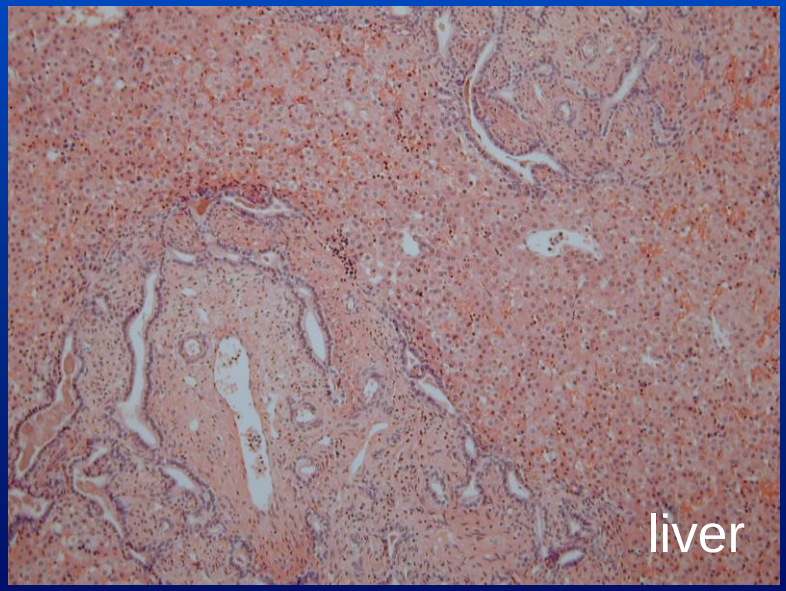
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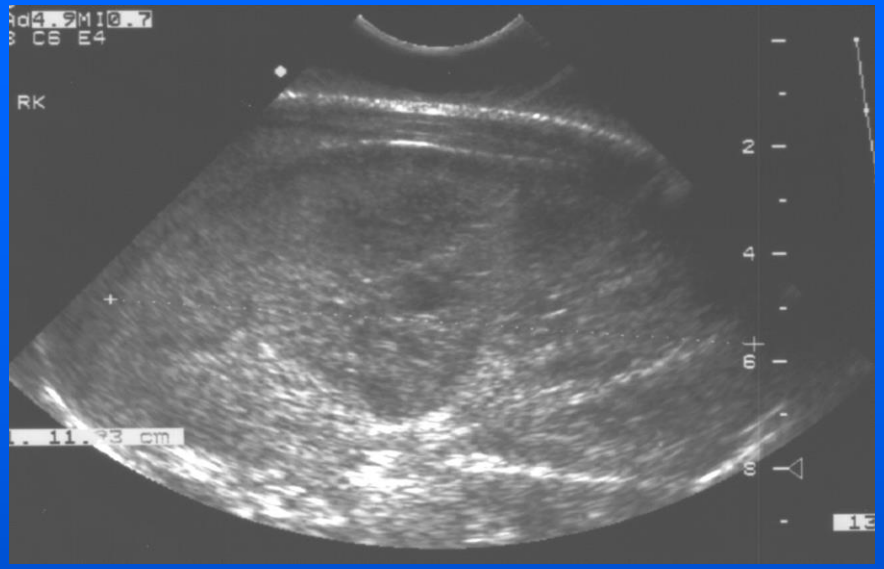
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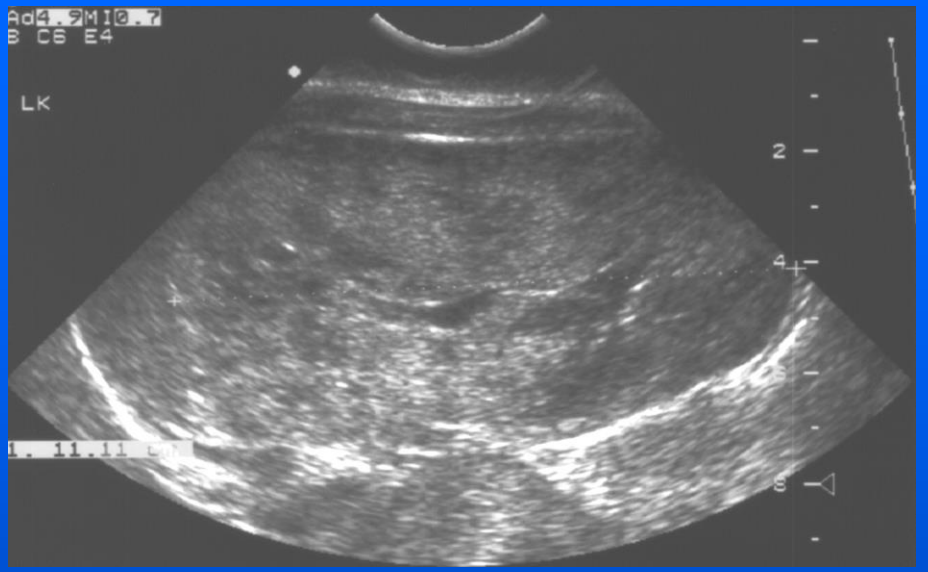
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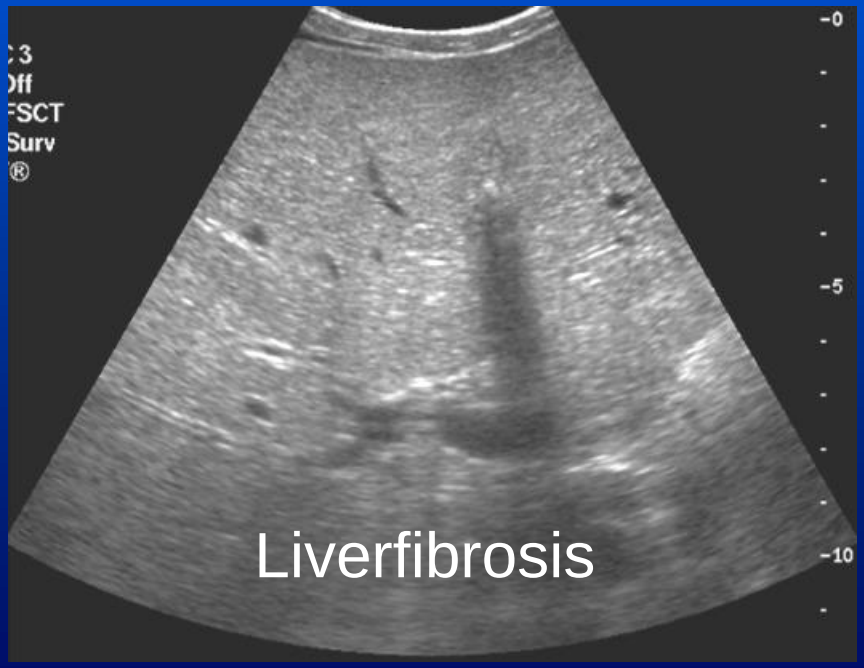
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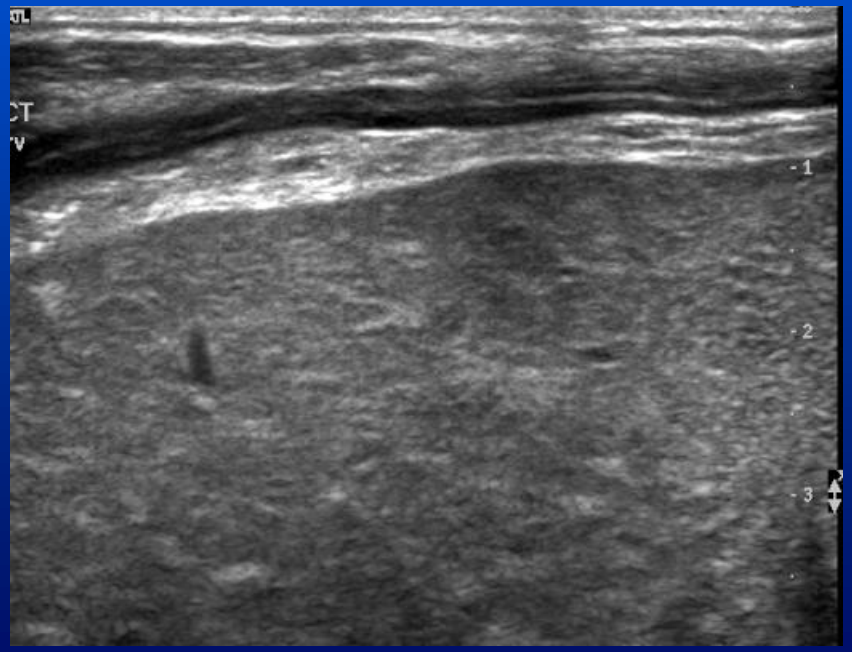
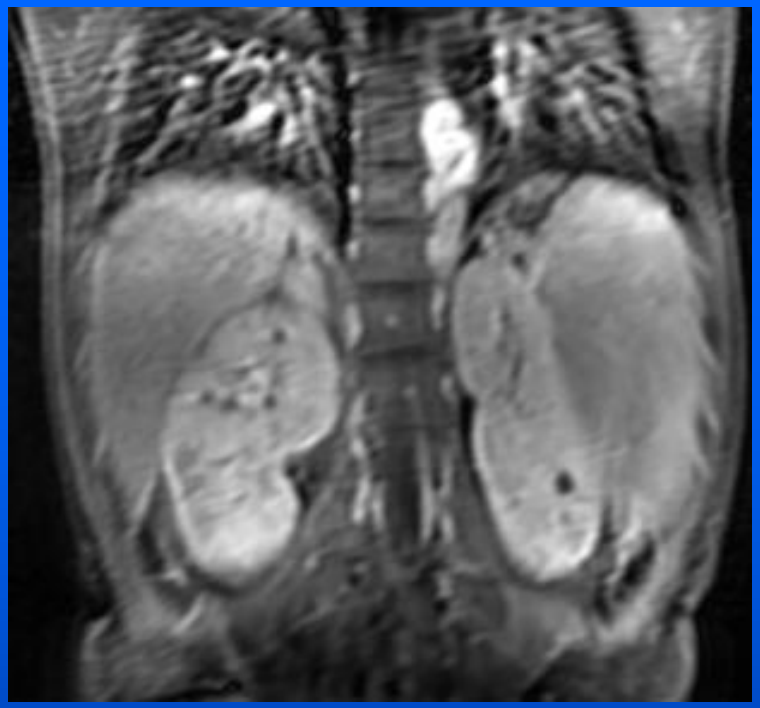
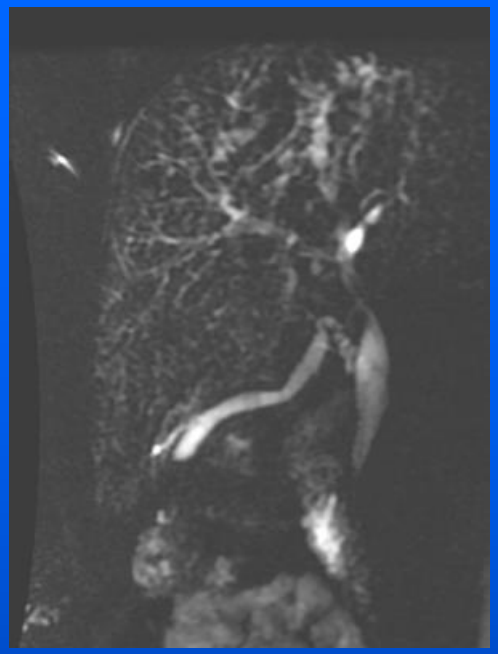
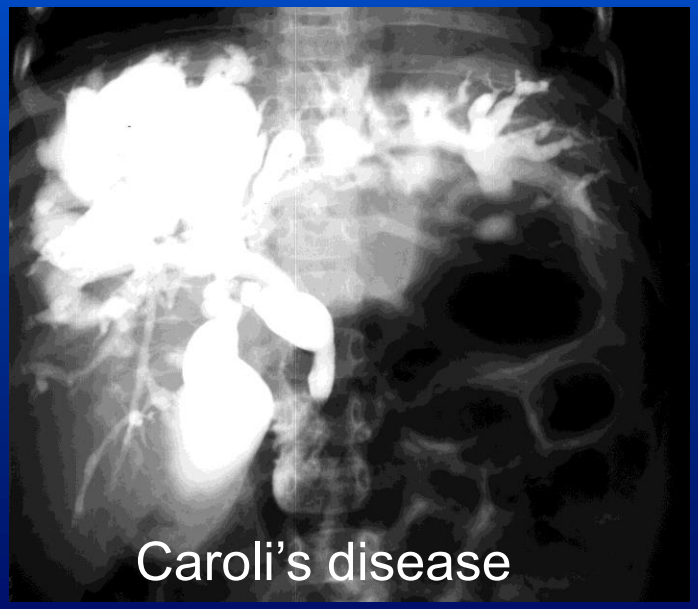






Portal hypertension with varicosis



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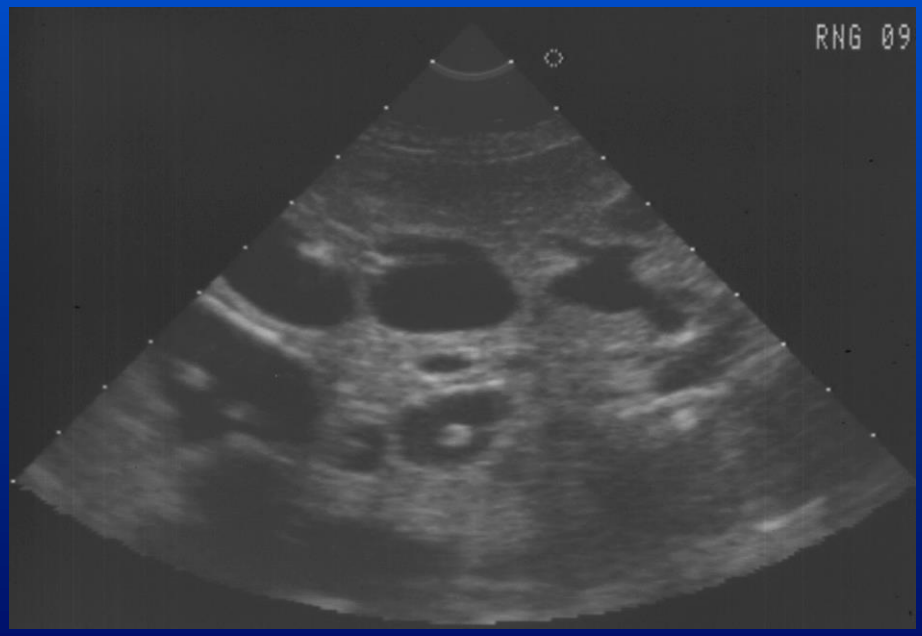


Tuberous sclerosis complex (TSC)

- Affects approximately 2 million people worldwide
- MI: AR
- Mutation:
  -TSC1 gene located on chromosome 9q34
  -TSC2 gene located on chromosome 16p13.3
- 2 patients were reviewed

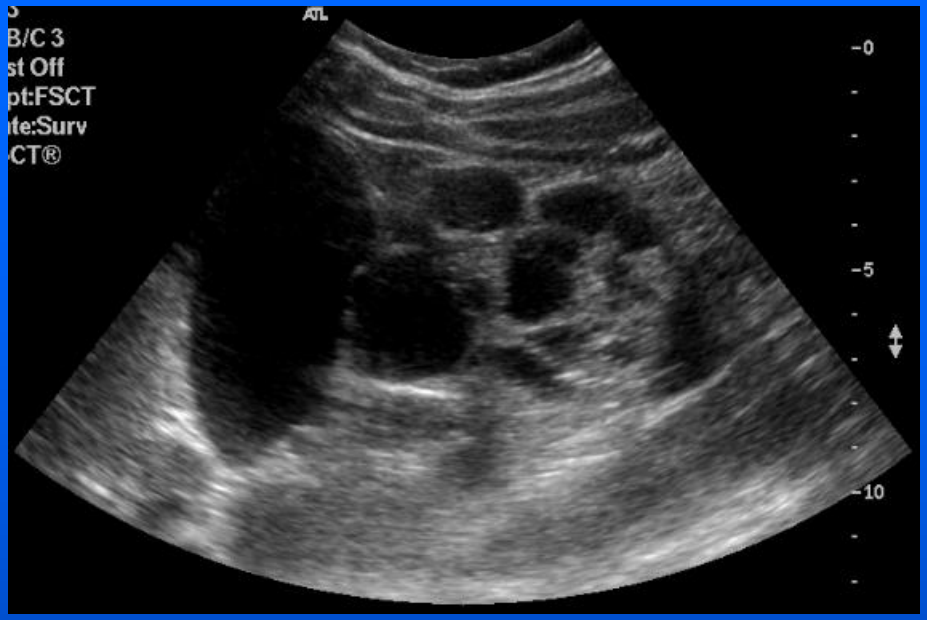
Clinical manifestation:

Renal:

Angiomyolipomas (70-80%), multiple renal cysts (20%)

Extrarenal:

Skin, central nervous system, cardiac pulmonary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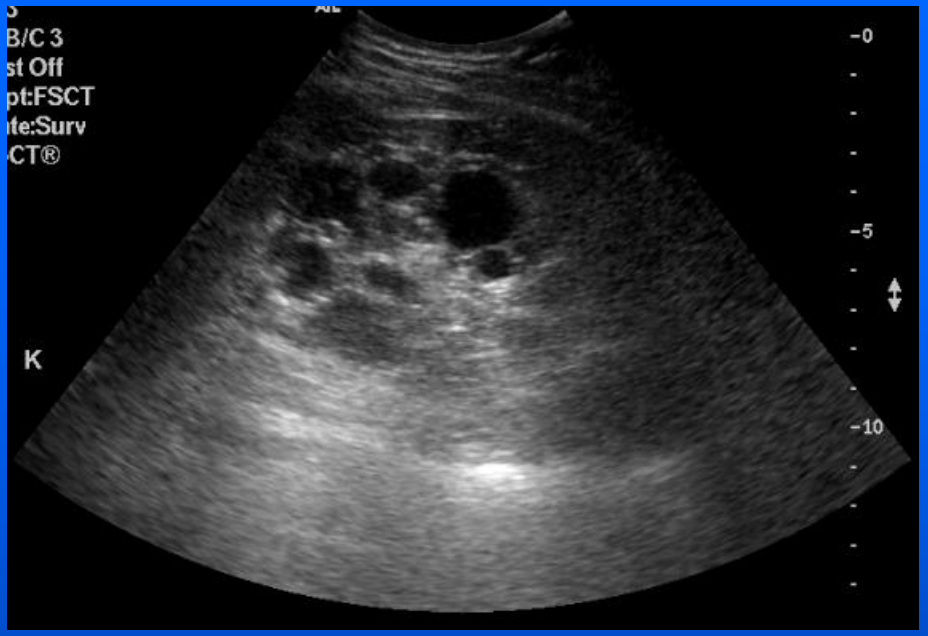
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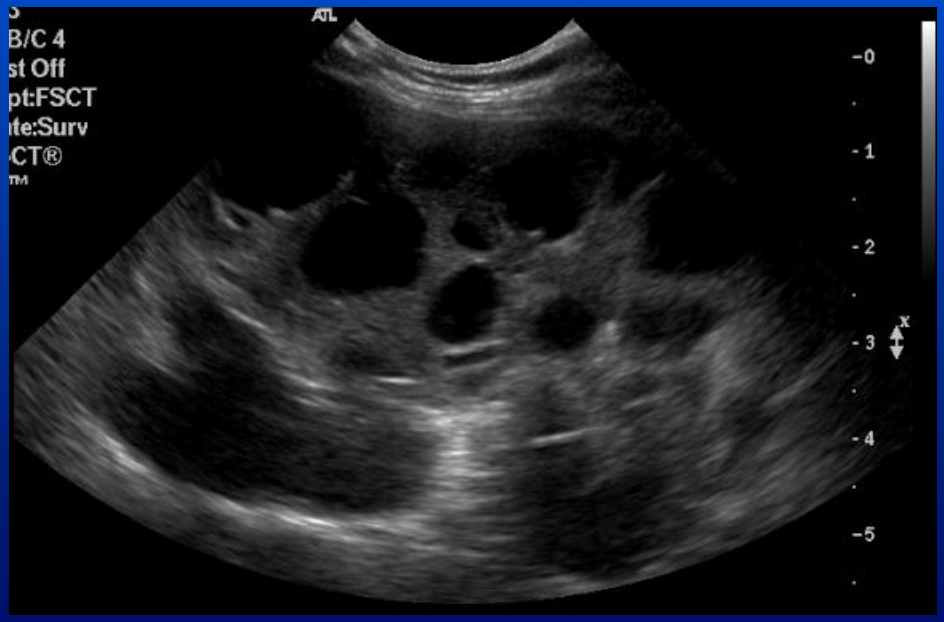
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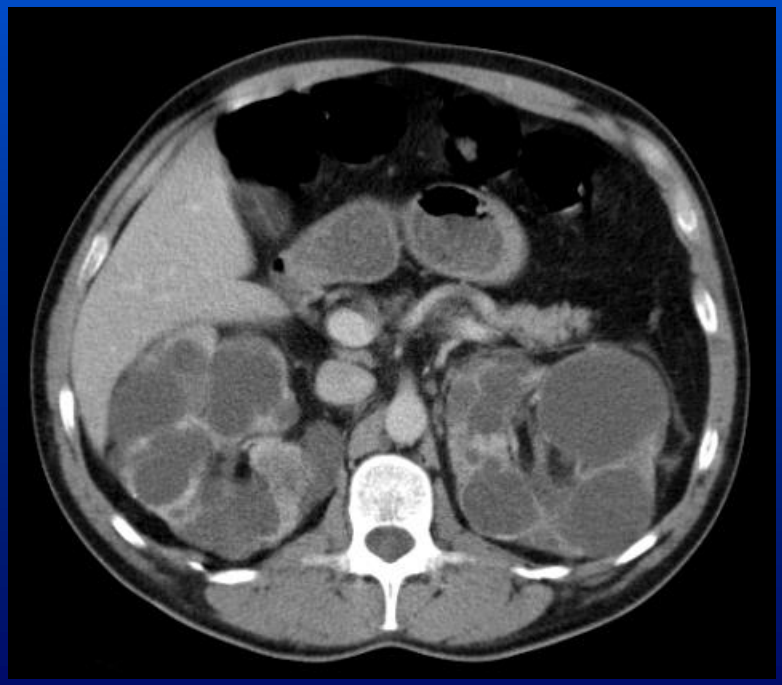


Juvenile nephronophitisis

- Frequency: unknown
- MI: AR. Mutation gene Ch.L.: 2p13.
- Medullary cystic kidney disease is the adult type (AD)
- 4 patients were reviewed (M=3, F=1; age range 5 m-16 yrs)

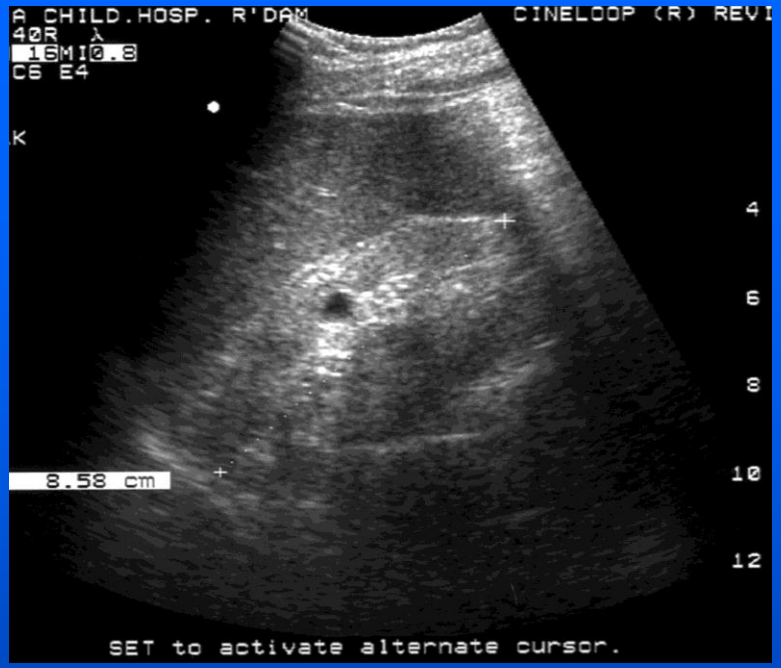
Clinical manifestation:

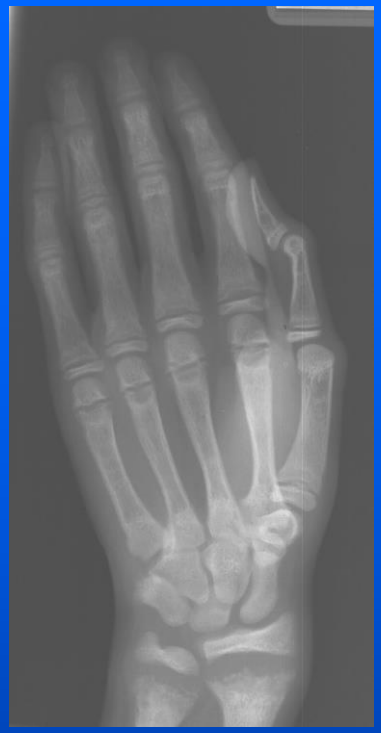
Renal:

Hyposthenuria, anemia, polyuria, polydipsia, renal failure

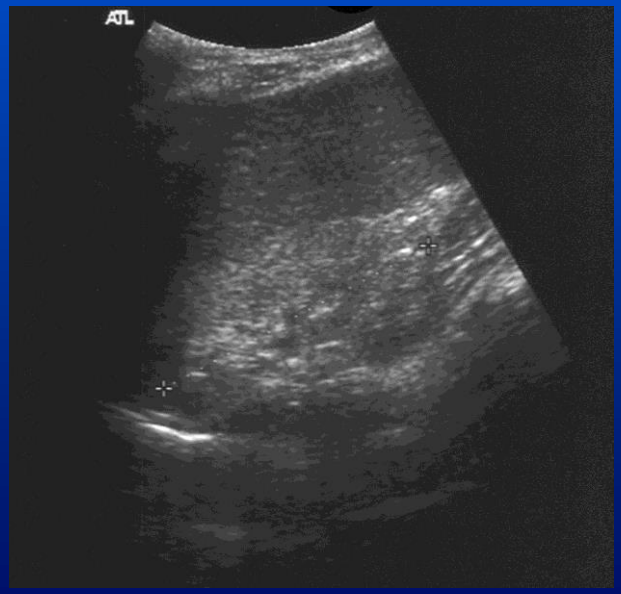
Extrarenal:

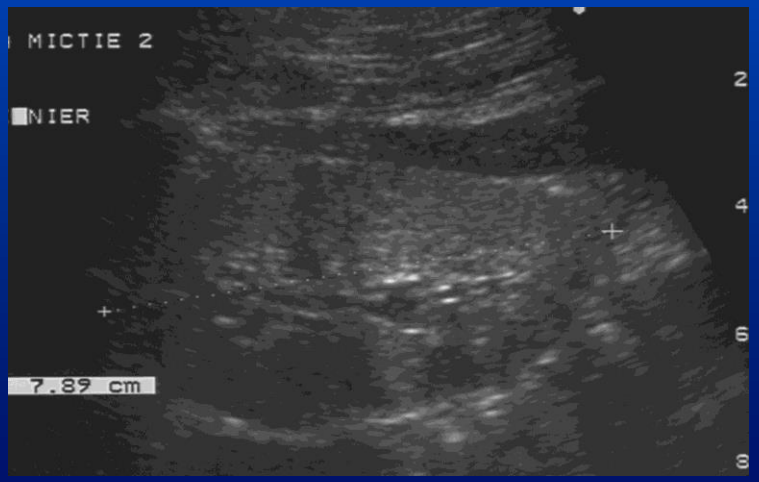
Brown bone tumor mandible (1), renal osteodystrophia (3), psychomotoric retardation, vermis atrophia





Renal osteodystrophia





Multicystic dysplastic kidney (MCDK)

- Non-hereditary
- Developmental anomaly of kidney with multiple smooth nonfunctioning, no communicating cysts, variable in size and number
- Little or no normal parenchyma
- Relatively high incidence of severe contralateral anomalies of kidney (hydronephrosis)
- Incidentally complicated with gastrointestinal and cardiac anomalies
- Detected frequently during antenatal ultrasonography
- In past managed by nephrectomy, but currently a more conservative approach is utilized

MCDK

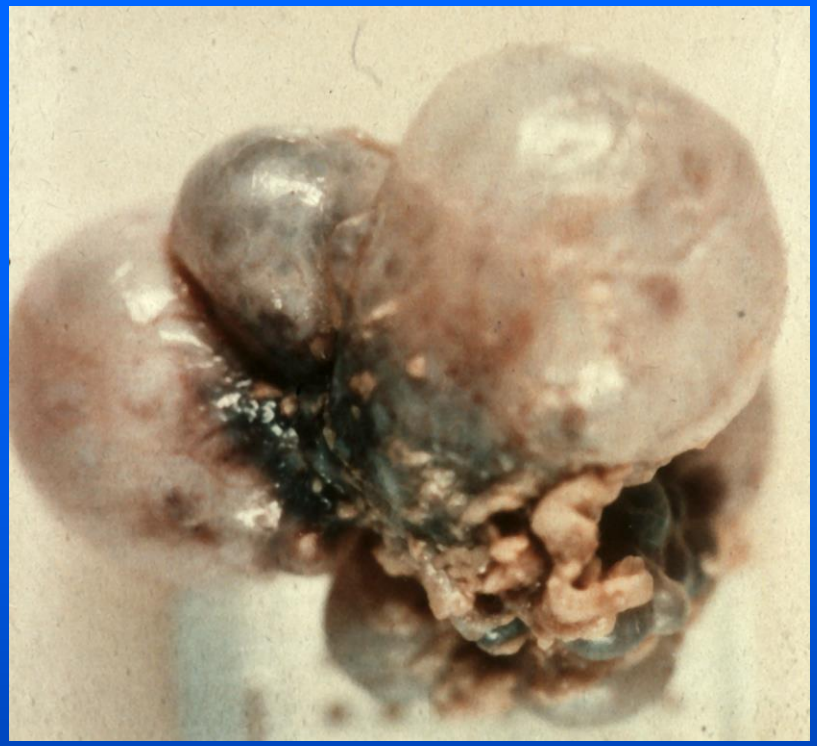
- 80 patients were reviewed (M=52, F=28; age range 1 d-10 yrs)
- 45 Antenatal ultrasound reported 28 as MCDK, 14 as hydronephrosis and 3 as renal agenesis.

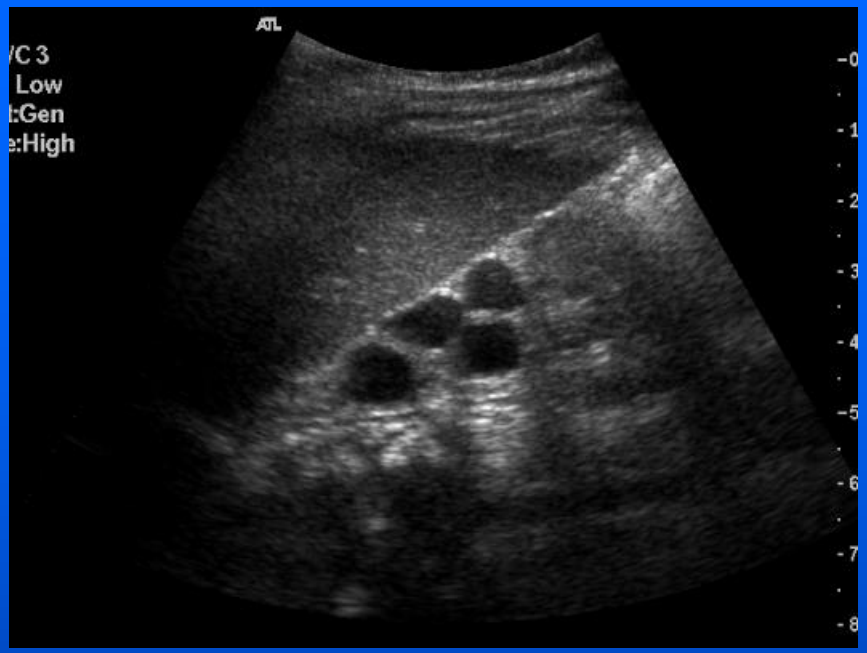
Used imaging modalities:

- Ultrasound in all 80 patients (1-3 times)
- Micturation cystography in 43 patients (5 times reflux found)
- Radionuclide study in 8 patients

Additional findings in 80 cases of MCDK:

Anorectal malformation (2), congenital heartfailure (2), contralateral hydronephrosis (6), duplicated urinary system (3), ectopic ureter (11), atretic ureter (2), ectopic located MCDK (4)

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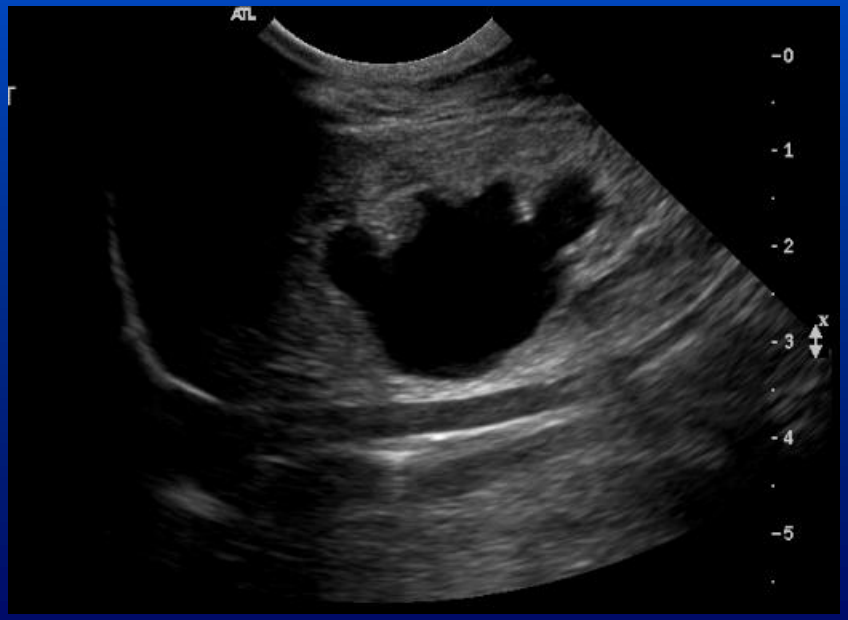
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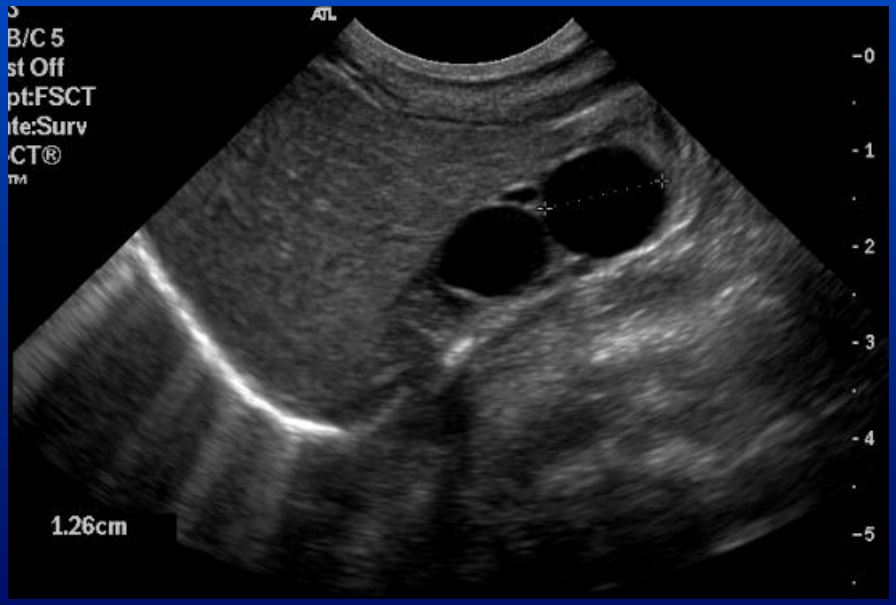
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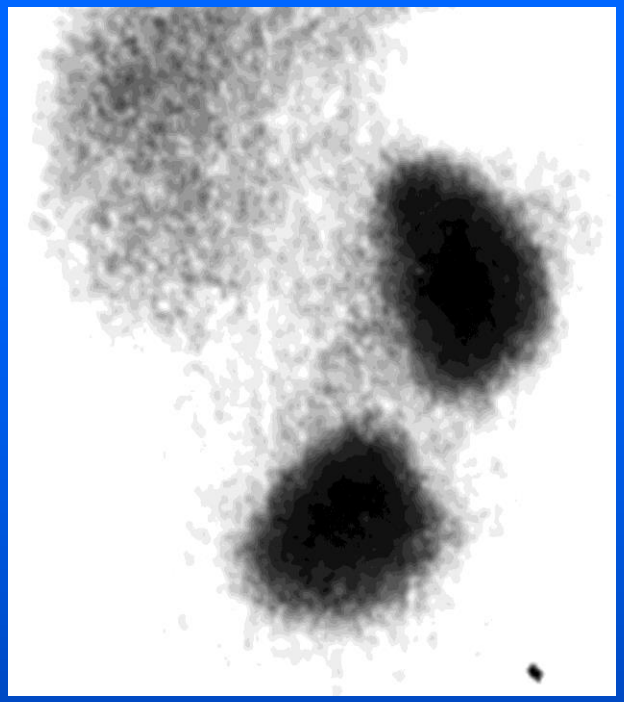

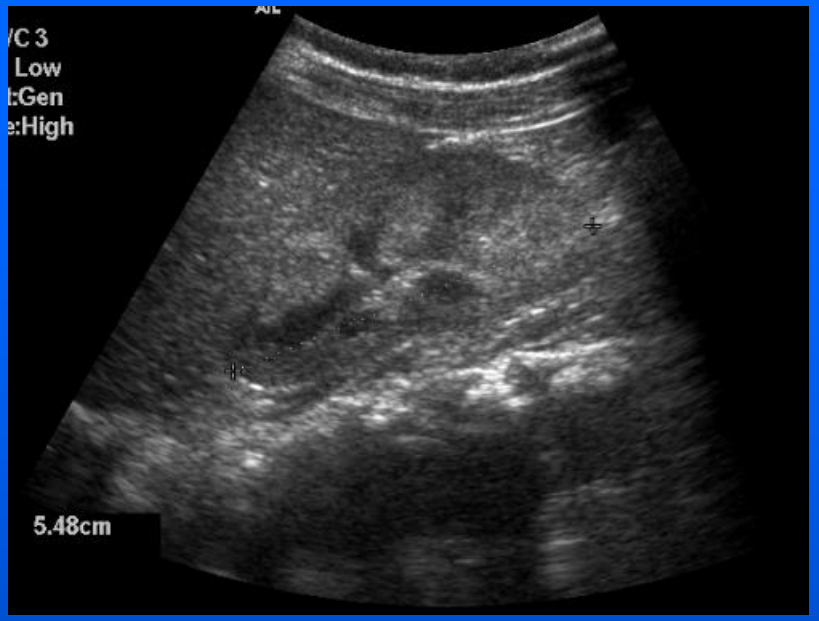
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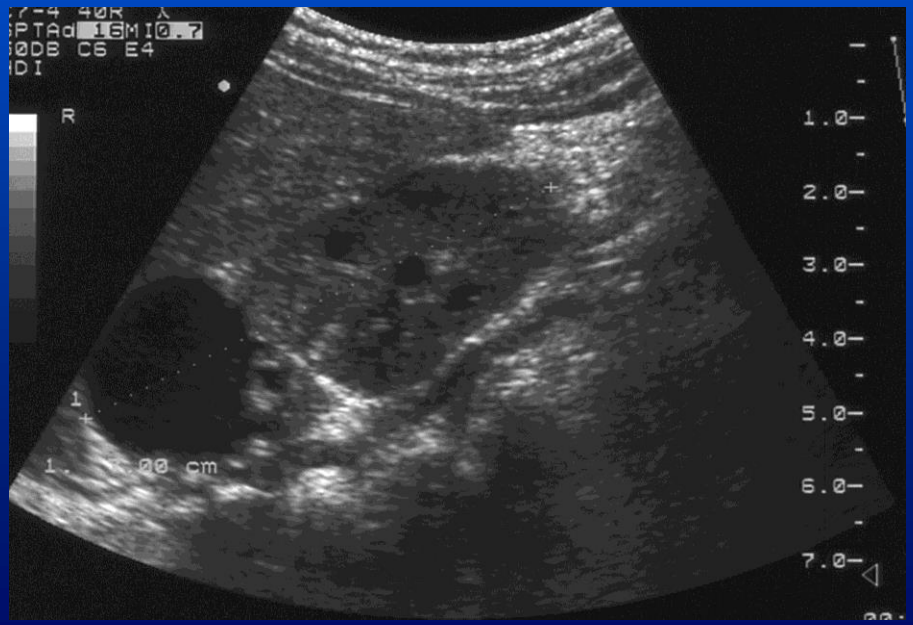
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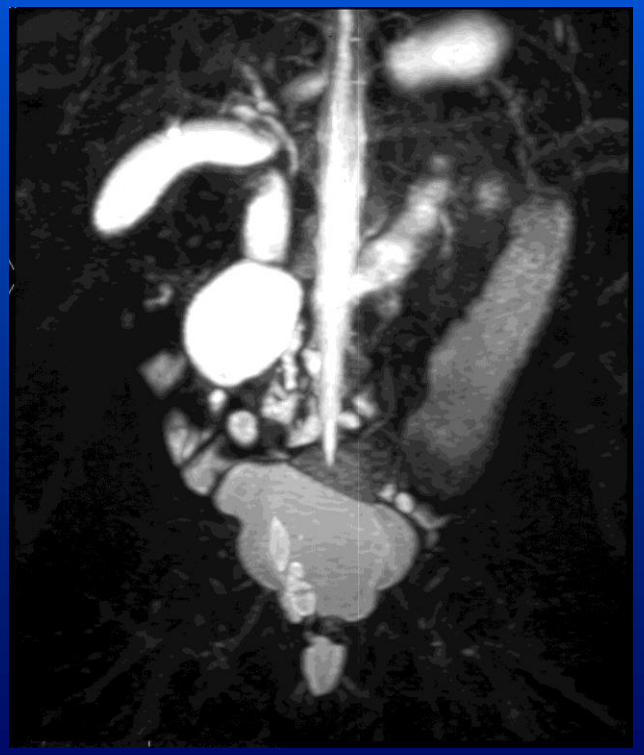
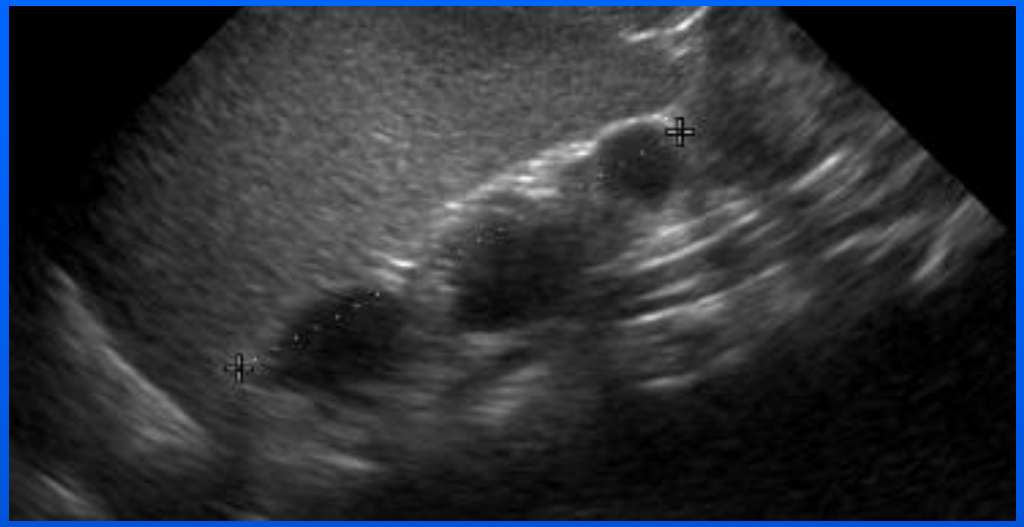
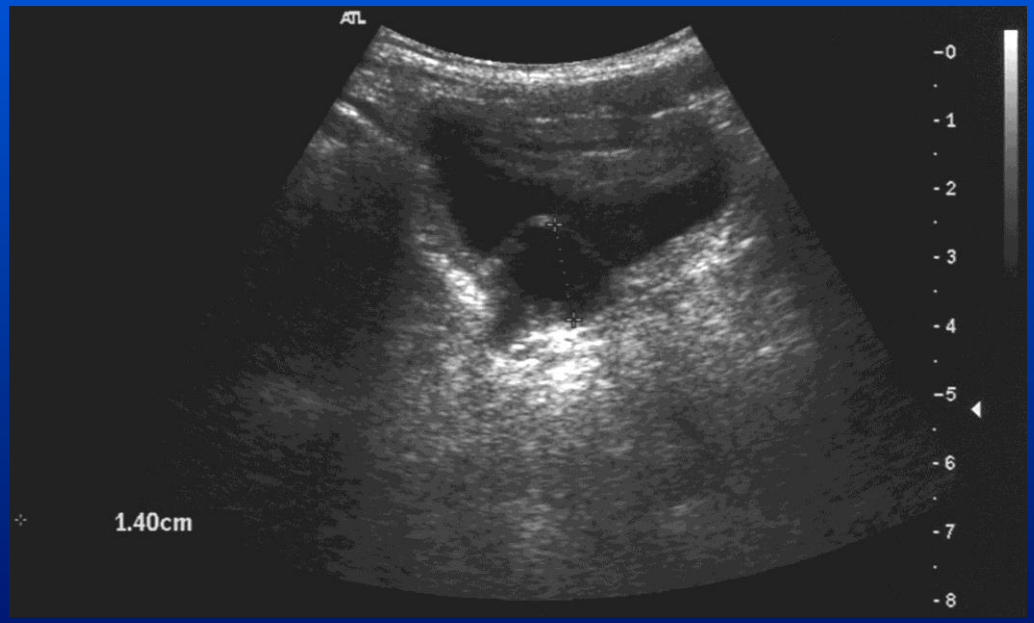
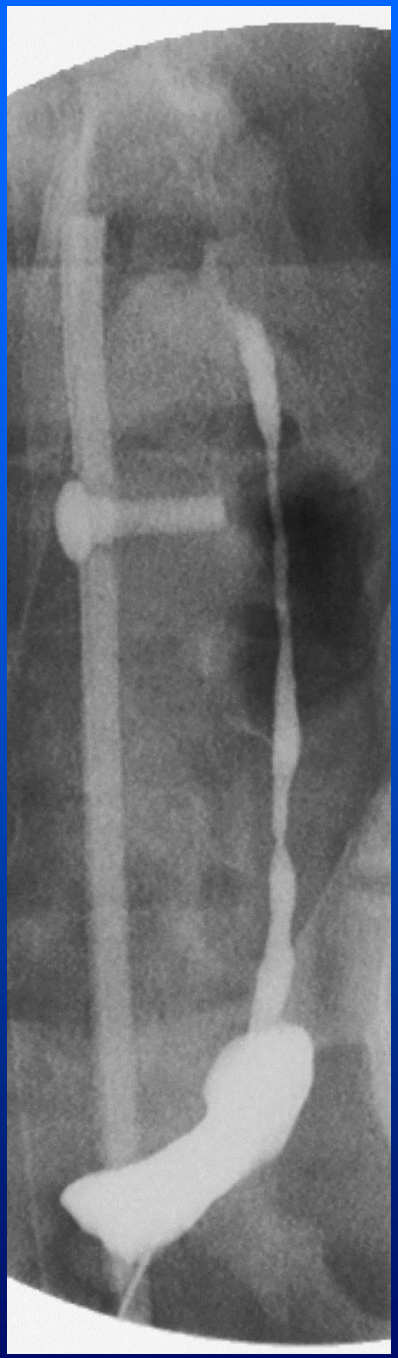
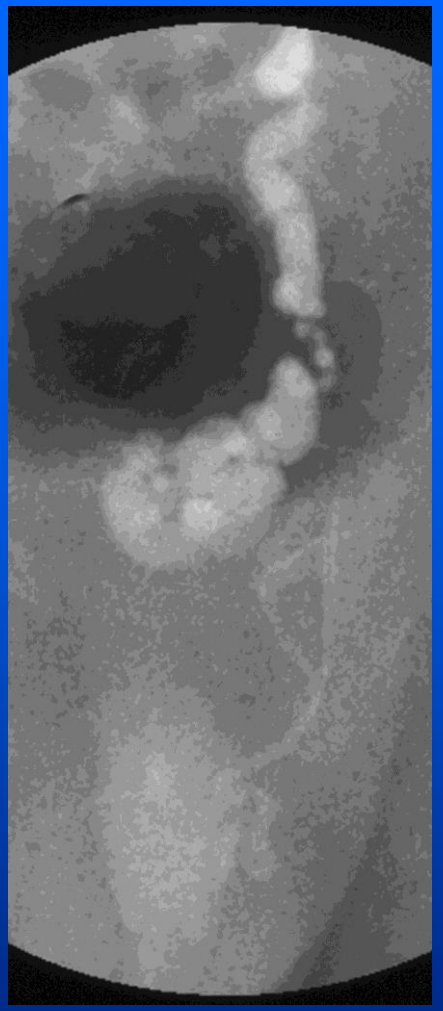






Ureterocele

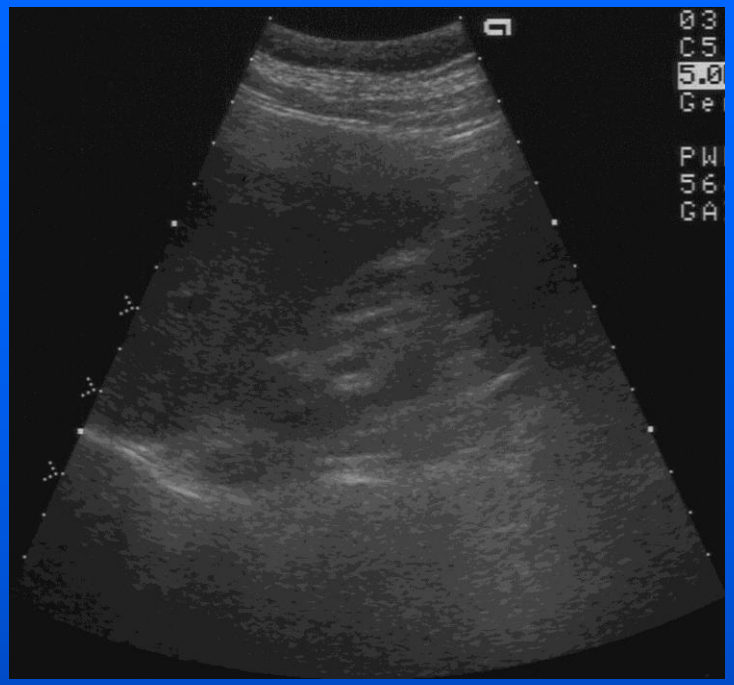


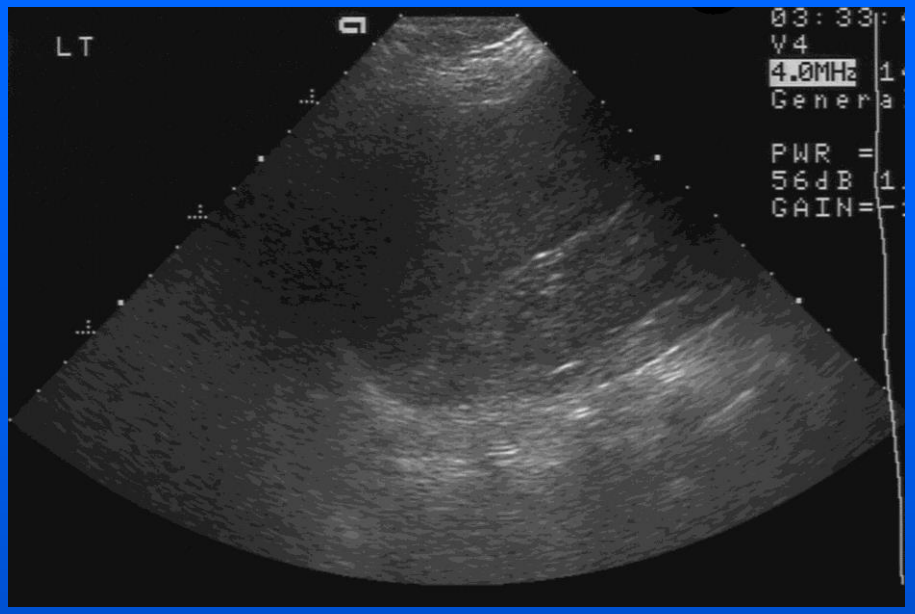


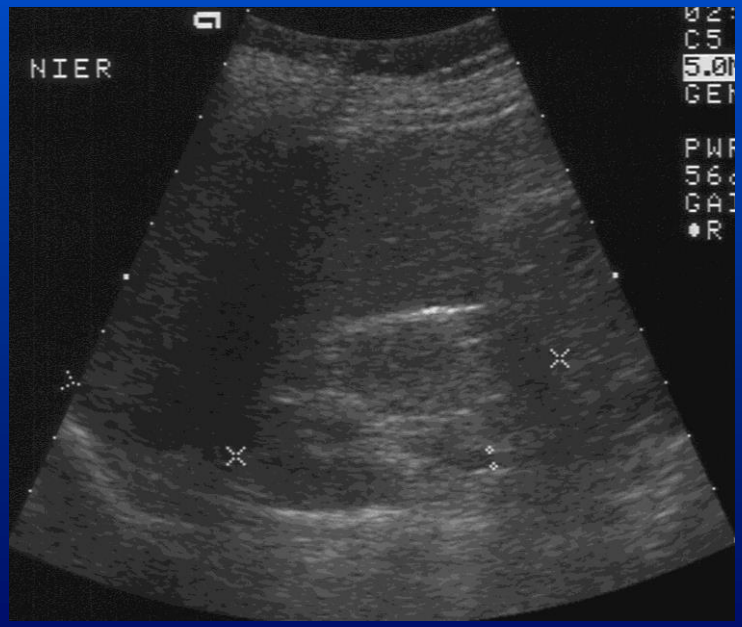
Ectopic ureter in vesiculae seminalis

Malformation syndromes with renal cysts

- Over 80 syndromes are associated with some form of cystic renal disease.
- The presence of cysts in certain syndromes is an integral part of the definition of diseases as Zellweger and Meckel's syndrome.
- In other syndromes renal cysts are a frequent finding as in Bardet-Biedl or asphyxiating thoracic dysplasia.
- In this patient material only one case of Bardet-Biedl was reviewed in a 8-year-old boy.



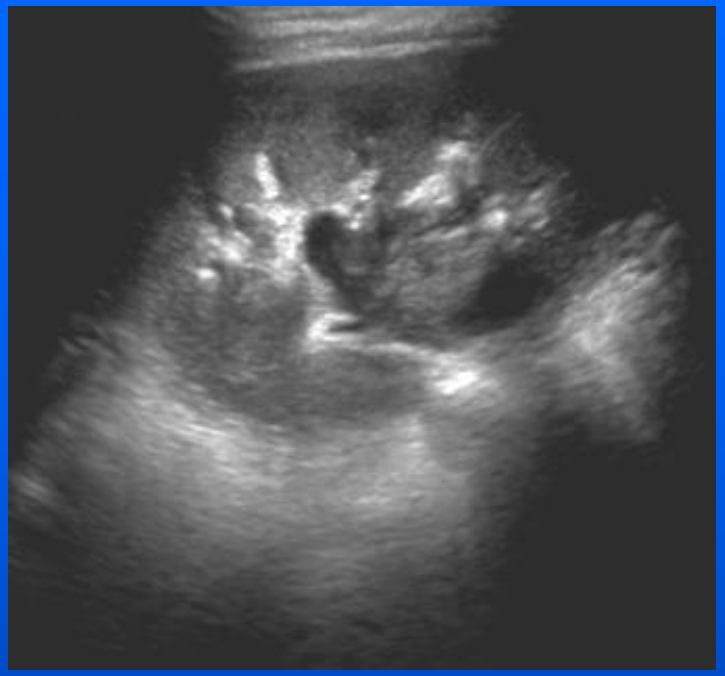


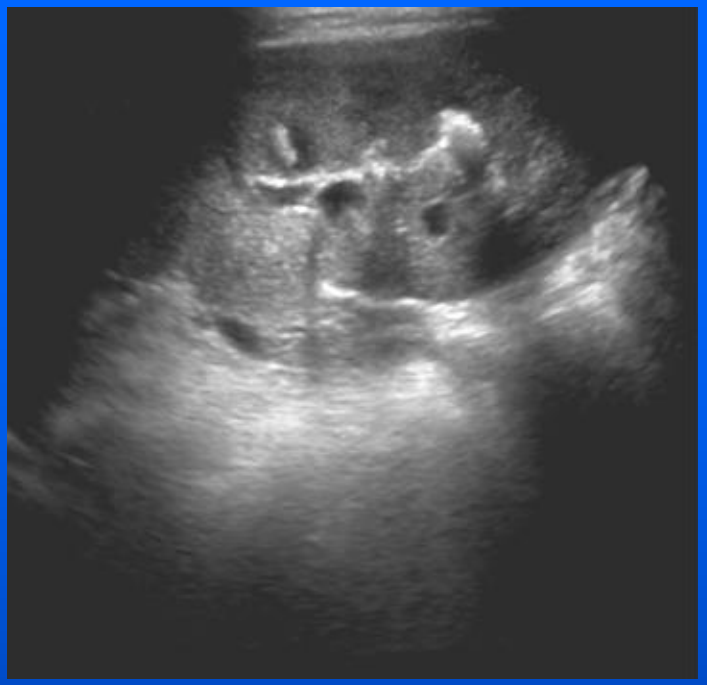


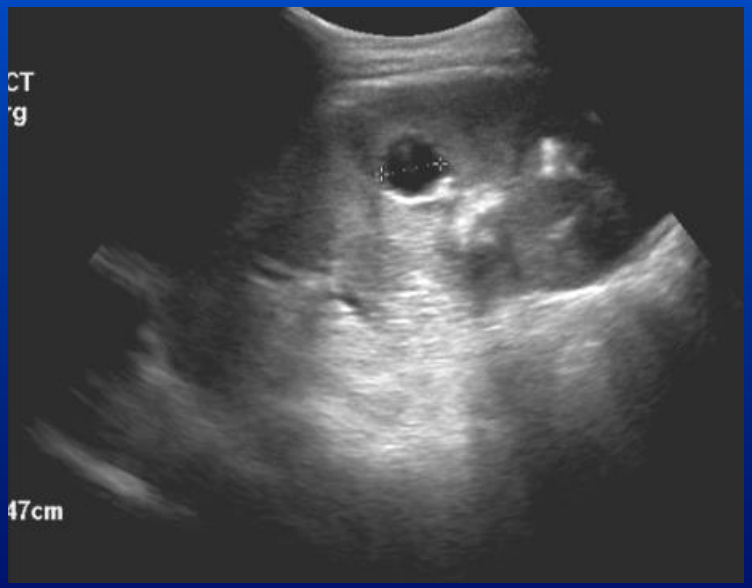
Microcystic kidneys in Bardet-Biedl syndrome

Medullary sponge kidney

- Nonhereditary disorder
- More frequently observed in adults than in children.
- Is incidentally associated with stone formation or urinary infection.
- The radiological changes are due to the focal dilatation of the collecting tubules.



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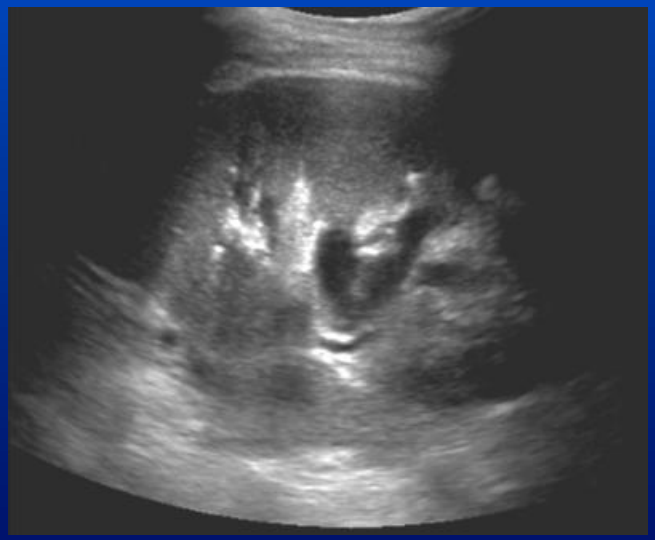
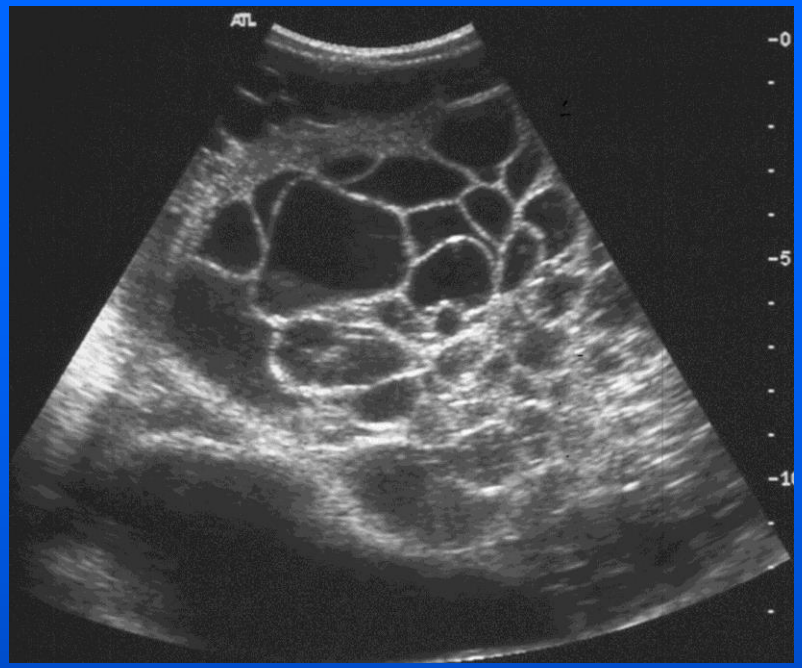
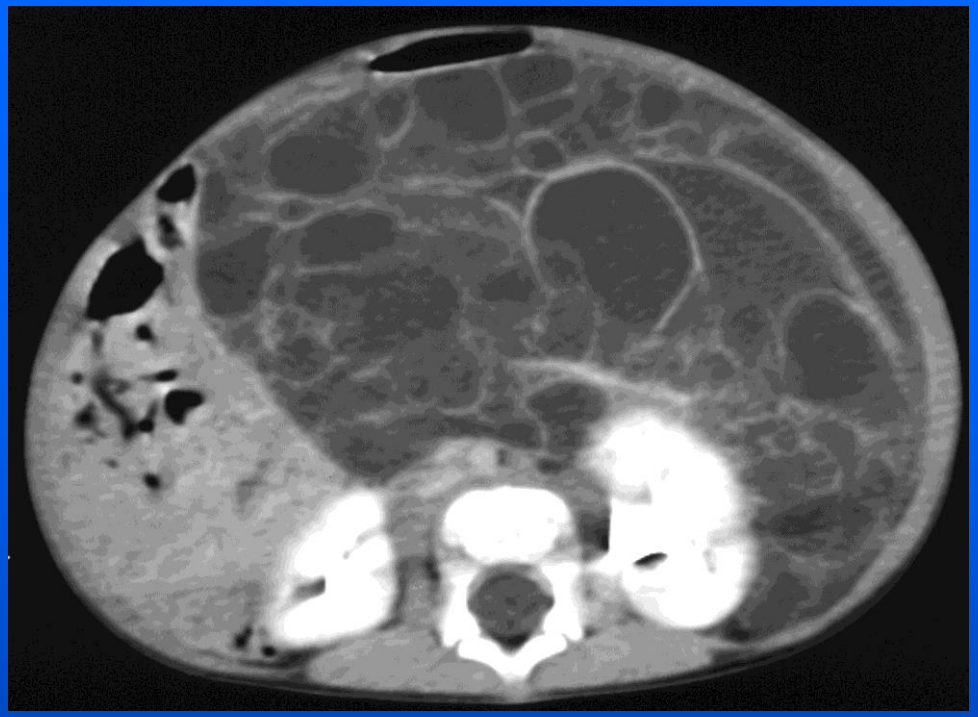
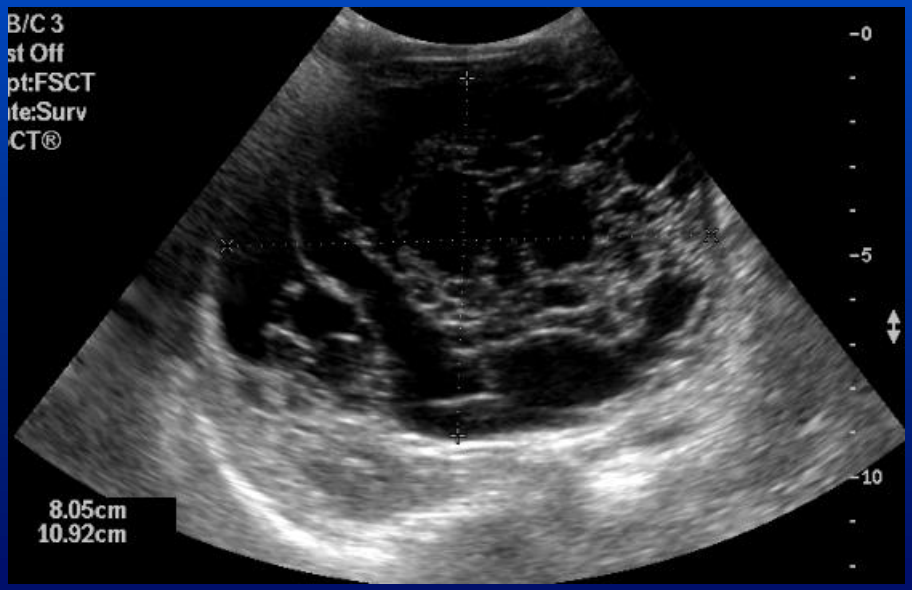


Multicystic nephroma

- Multilocular cystic nephroma is a renal mass derived from metanephric blastoma.
- Histology varies from complete benign to malignant Wilms tumor.
- An abdominal mass is mostly the clinical presentation.
- This patient material includes 2 cases of benign multicystic nephroma.



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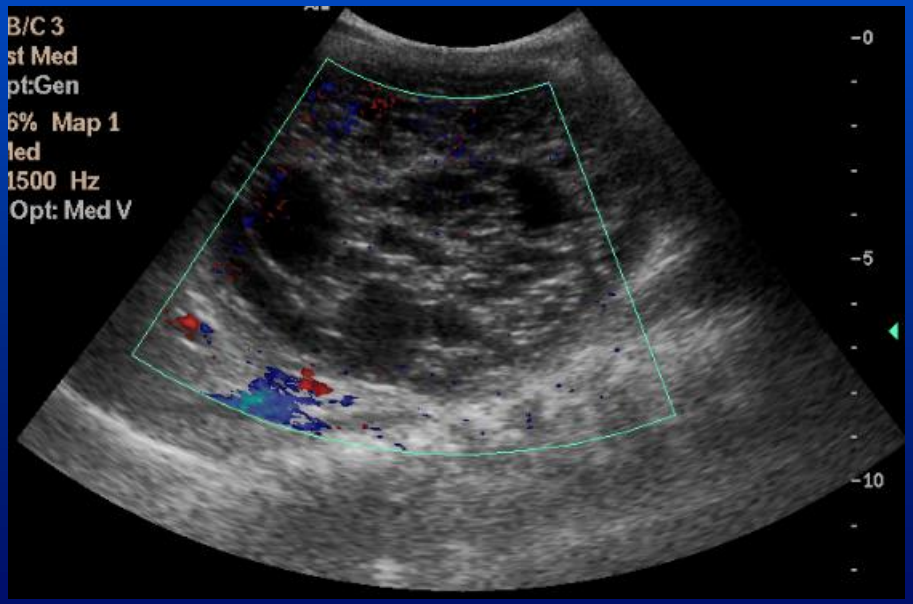
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Conclusion I

- Nearly all double sided cystic kidneys are inherited with exception of sponge kidneys and renal cysts in some syndromes.
- Genetic renal cystic diseases manifest themselves in different age periods, depending on the type of the disease.

Conclusion II

- Multicystic dysplastic kidney should be differentiated from hydronephrosis especially in prenatal and postnatal period.
- Hepatic cysts are a frequent finding in ADPKD, but is presented in later life.
- Caroli's disease and liver fibrosis with portal hypertension is the most frequent finding in ARPKD.
- Pneumothorax and pneumomediastinum are important complications of hypoplastic lungs in infants with ARPKD.

Conclusion III

- Contralateral hydronephrosis in patients with MCKD is an incidental finding.
- Vesico-ureteral reflux, ectopic or atretic ureter as well as ectopic location of dysplastic kidney are frequent findings in MCKD.

- Ultrasound is the cornerstone in the diagnosis of renal cystic diseases.
- CT, MRI and radionuclide studies should be performed in complicated cases.