Priya John

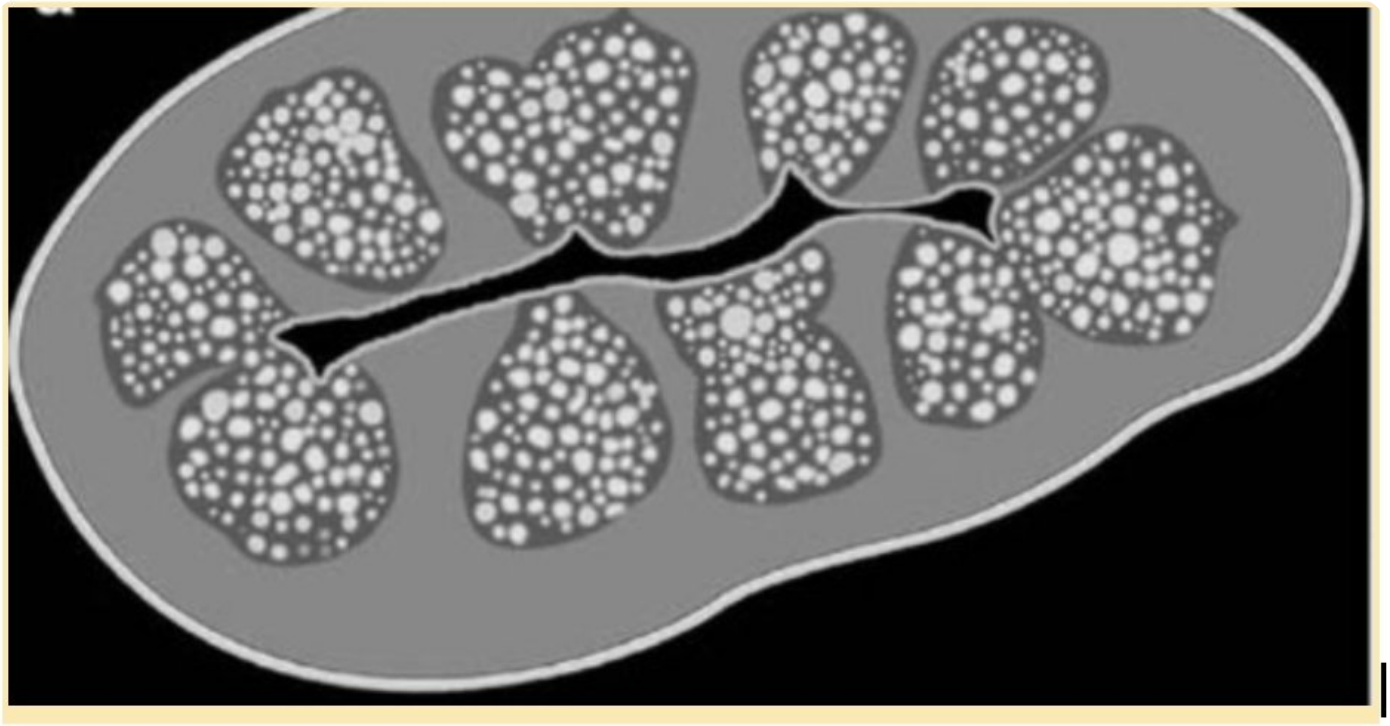
@drpriyajohn

27 Tweets • 2022-09-26 00:46:30 UTC • See on Twitter

rattibha.com

Hello #NephTwitter #Renal #MedTwitter @aspneph radiology webinar for august was Autosomal Recessive Polycystic Kidney Disease (#ARPKD). Let's get started

#PedNeph #Transplant



PMID: 19089418

1/Let’s start with a poll!
Do you know the one third rule of #ARPKD
@myadla @bhavya_dasara @dakidneydoc
@DrPSVali @priti899 @NephroSeeker
@Sandyrvsdav @dr_sourabha @nephromythri
@divyaa24 @krithicism @NamrataYParikh
@snamratarao @nephrologik @AnandhUrmila

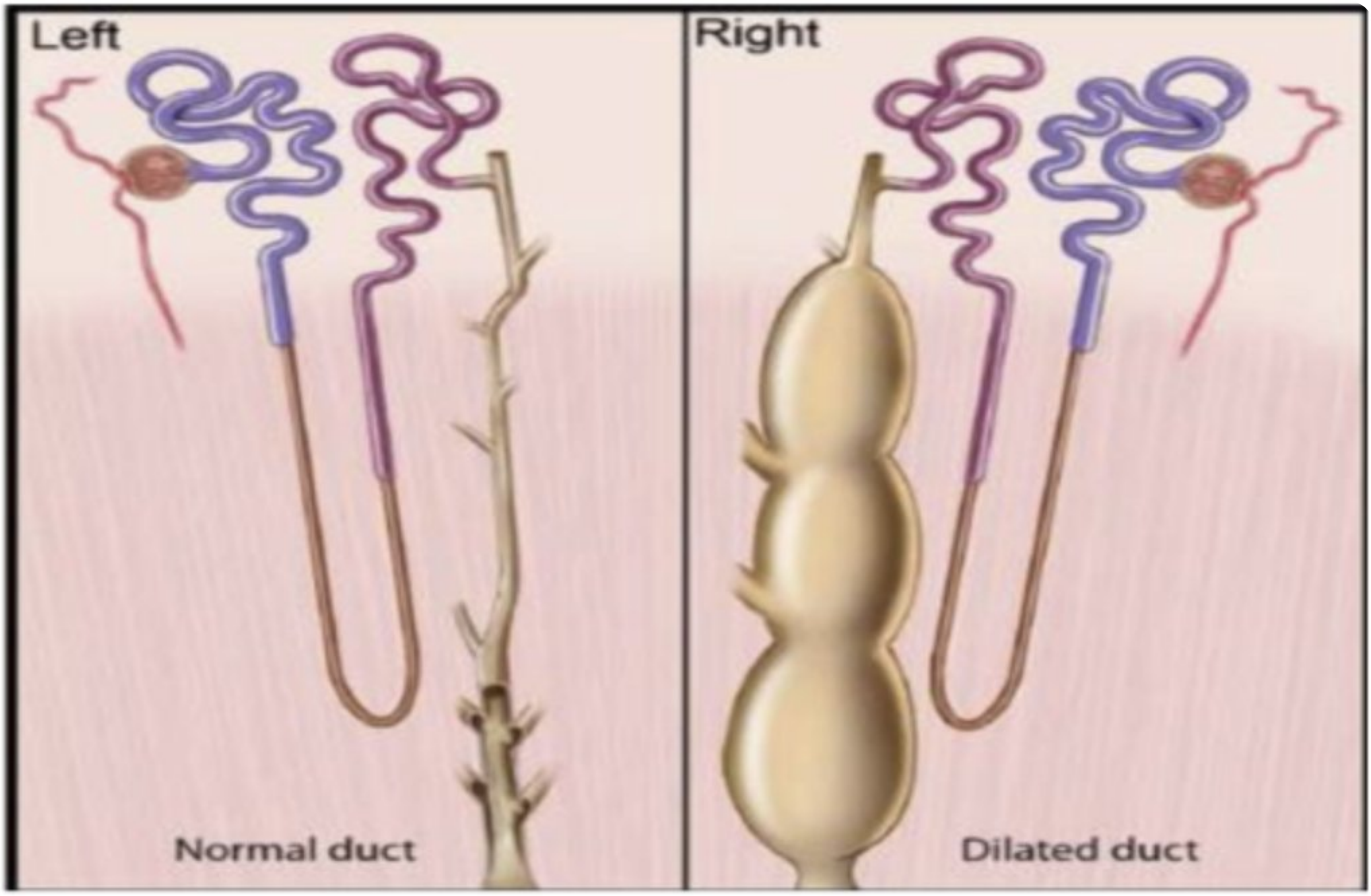
2/ Clinical Presentation of ARPKD follows ‘ONE-THIRD RULE’
📌1/3 of the patients present during the first year of life,
📌1/3 present between 1 and 20 years of age, and
📌the rest 1/3 after 20.
📌The incidence of ARPKD is 1 in 20000 live births.

https://pubmed.ncbi.nlm.nih.gov/16523049/

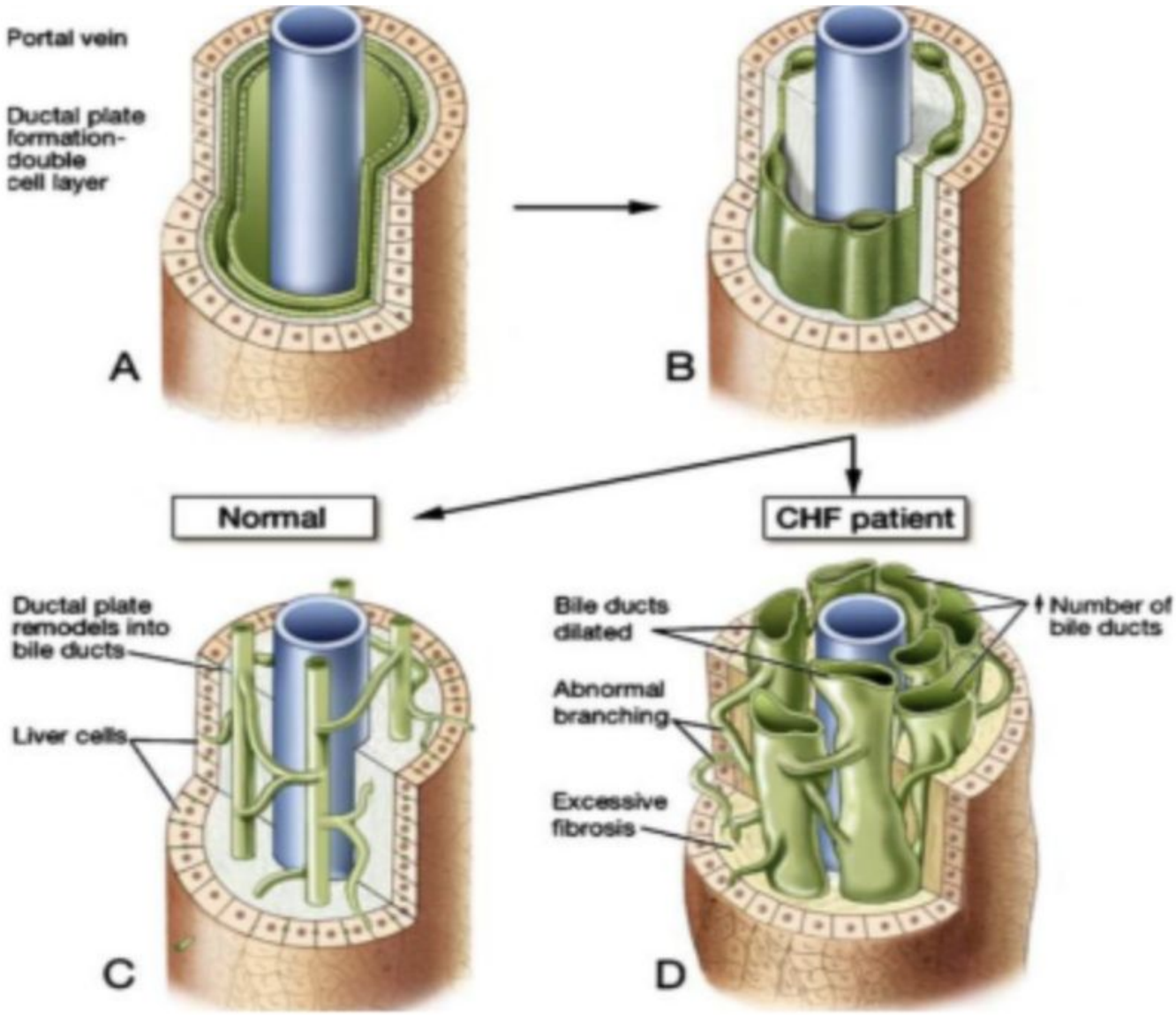
3/ ARPKD is an inherited hepatorenal disorder characterised by

♦Cystic dilations of the renal collecting ducts &

♦Congenital hepatic fibrosis due to developmental defects in ductal plates of the hepatobiliary system.



PMID: 19089418



PMID: 19089418

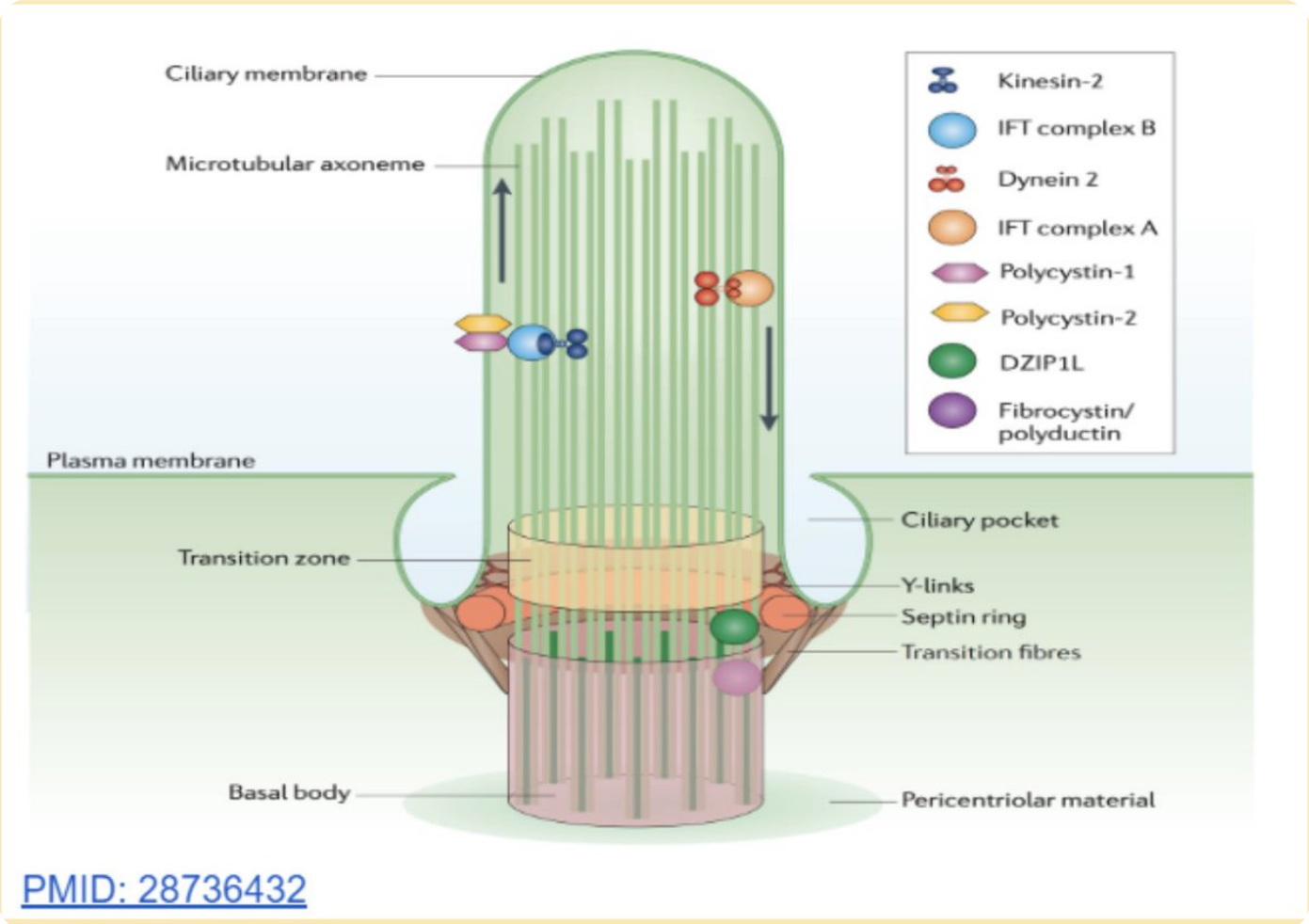
4/ Let's talk genetics!

Which of the following is true about genes in ARPKD

5/ The ans is a.

♦ARPKD is predominantly caused by mutations in PKHD1 located on chromosome 6p21 & DZIP1L genes (less common), which encodes fibrocystin and DAZ interacting protein 1 respectively.

♦PKHD1-localized to the basal body of the cilia.DZIP1L- localizes to centriole.



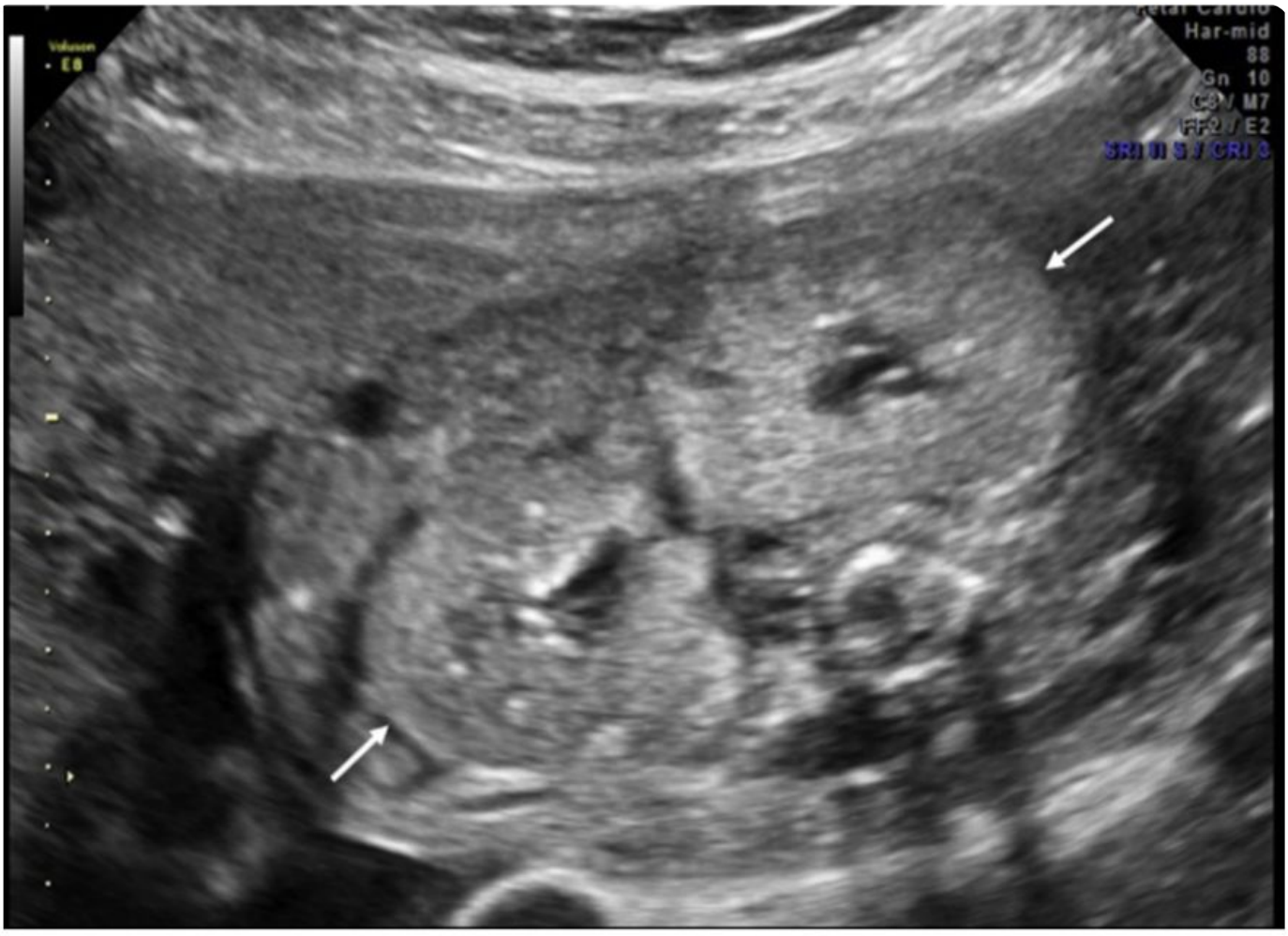
PMID: 28736432

6/ Prenatal imaging of ARPKD on ultrasound shows

🔷Bilateral enlarged kidneys with loss of corticomedullary differentiation,

🔷Subcortical hypo echoic rim with predominantly medullary cysts- “Snow Storm” appearance.

🔷Oligohydramnios and absence of urine in the bladder.



PMID 28669735

7/ In neonates, ARPKD presents with respiratory failure and kidney impairment.

🔷Respiratory failure - pulmonary hypoplasia (Potter Sequence) &/ or impaired excursion of diaphragm due to enlarged kidneys.

🔷HTN& ⬆️creatinine at birth - 🚩possibility of severe renal impairment.

Potter sequence

#ASPNFOAM Group
American Society of Pediatric Nephrology

PSEUDOEPICANTHUS

PULMONARY HYPOLASIA

FLATTENED NOSE

FLATTENED EARS

RECESSED CHIN

CLUB FEET & HIP DISLOCATION

@drpriyajohn

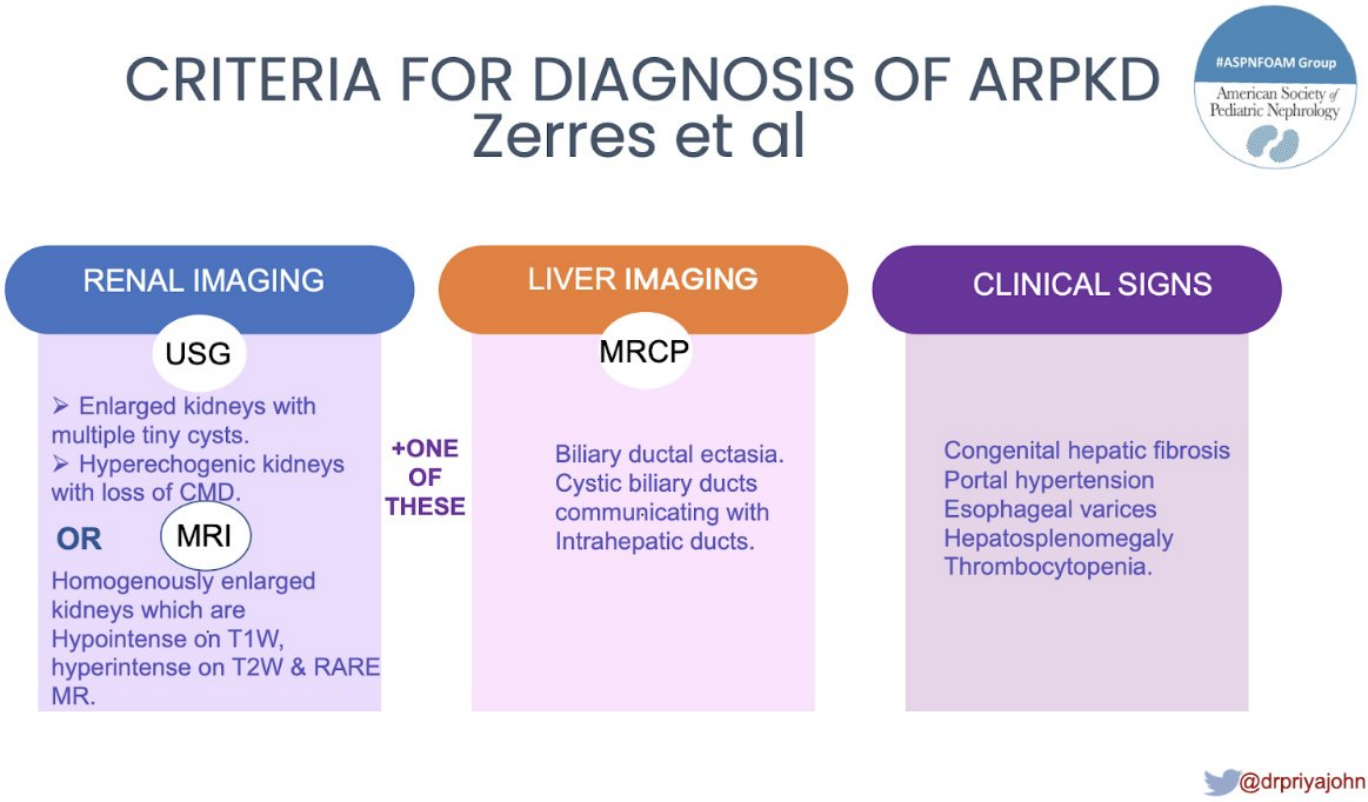
8/ Children who survive the neonatal period progress to infancy and childhood with-

📌some PLATEAU IN RENAL FUNCTION owing to the golden period of renal maturation in the first few years of life followed by

📌A steady decline in renal function later.
PMID: 25113295

9/ 📌 Biliary dysgenesis, congenital hepatic fibrosis, dilatation of the intrahepatic bile ducts (Caroli disease), portal hypertension, and cholangitis are prominent hepatobiliary presentations in children with ARPKD during adulthood.PMID: 24114580

10/ Specific diagnostic criteria of ARPKD are defined by
Zerres at al

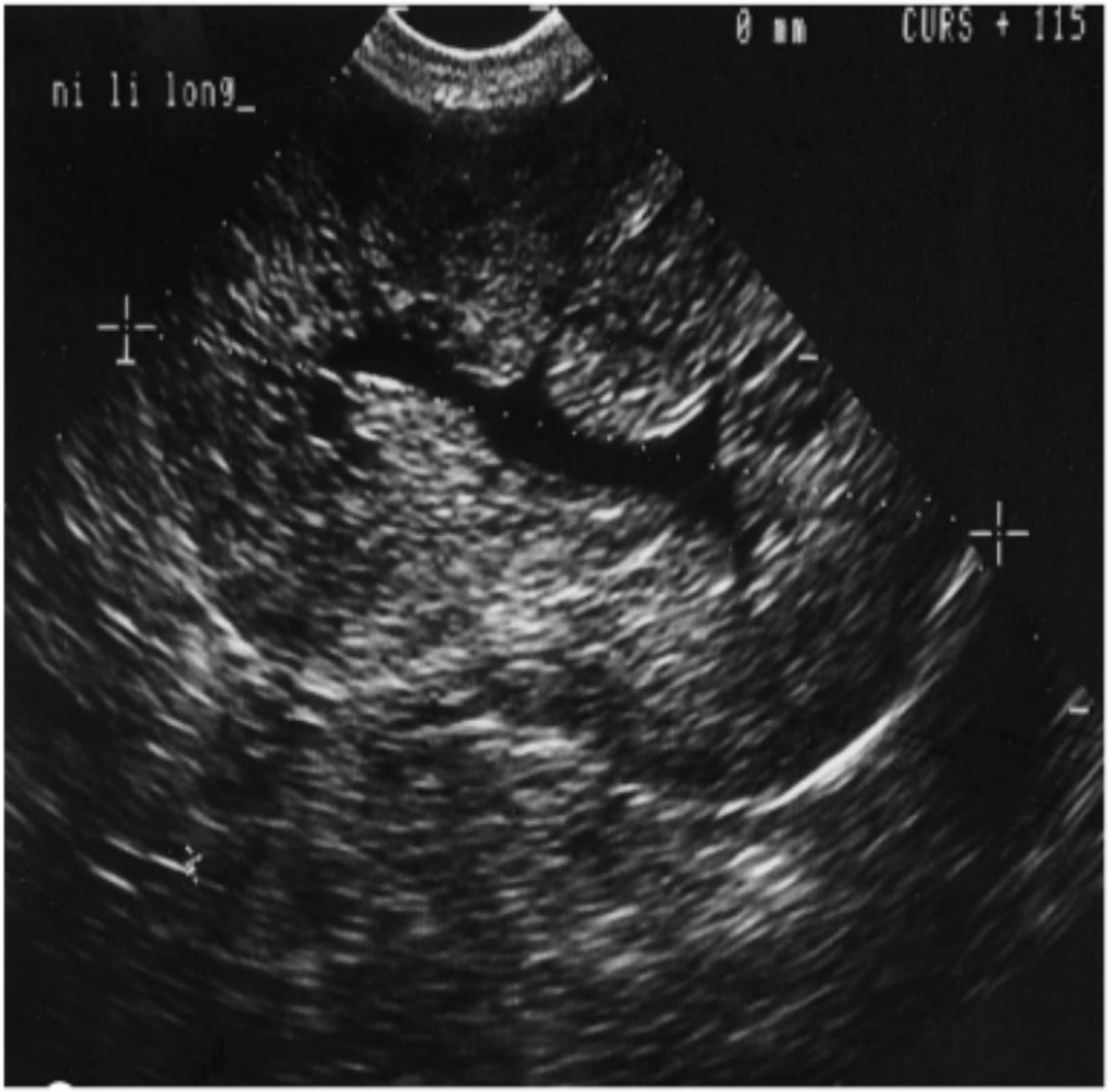


11/ Imaging by Ultrasound is usually diagnostic of ARPKD.

📌Ultrasound shows enlarged kidneys with multiple tiny cysts of diameter < 3mm.

📌High-resolution ultrasonography shows medullary ductal dilations where medullary cysts are predominantly present.

PMID 20413436



US imaging of 3-month-old child with ARPKD showing salt and pepper or snow storm appearance due to the hyperechogenic cortex and medulla

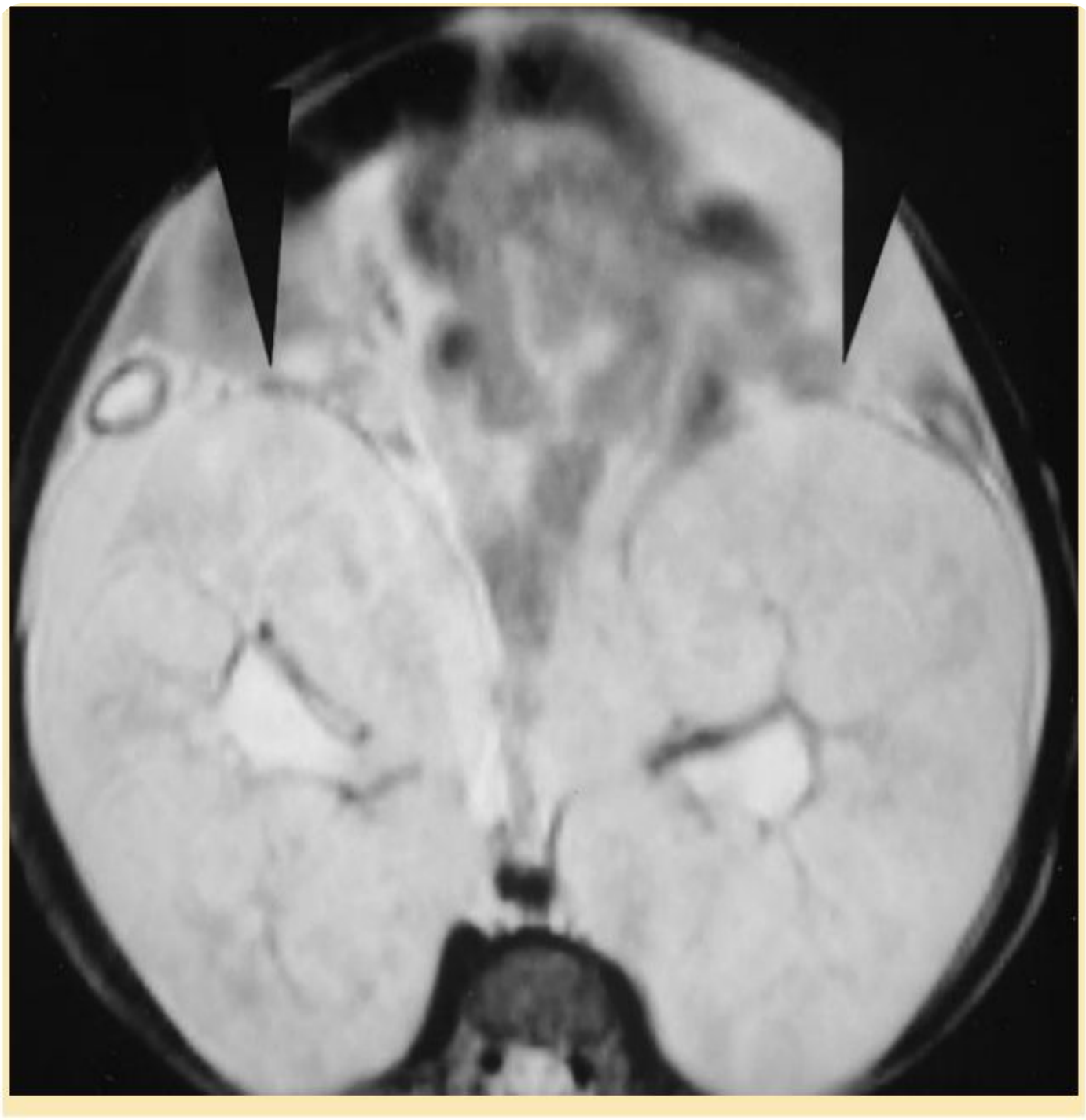
PMID: 10755752

12/ In cases with ambiguous findings on ultrasound, MRI confirms the diagnosis.

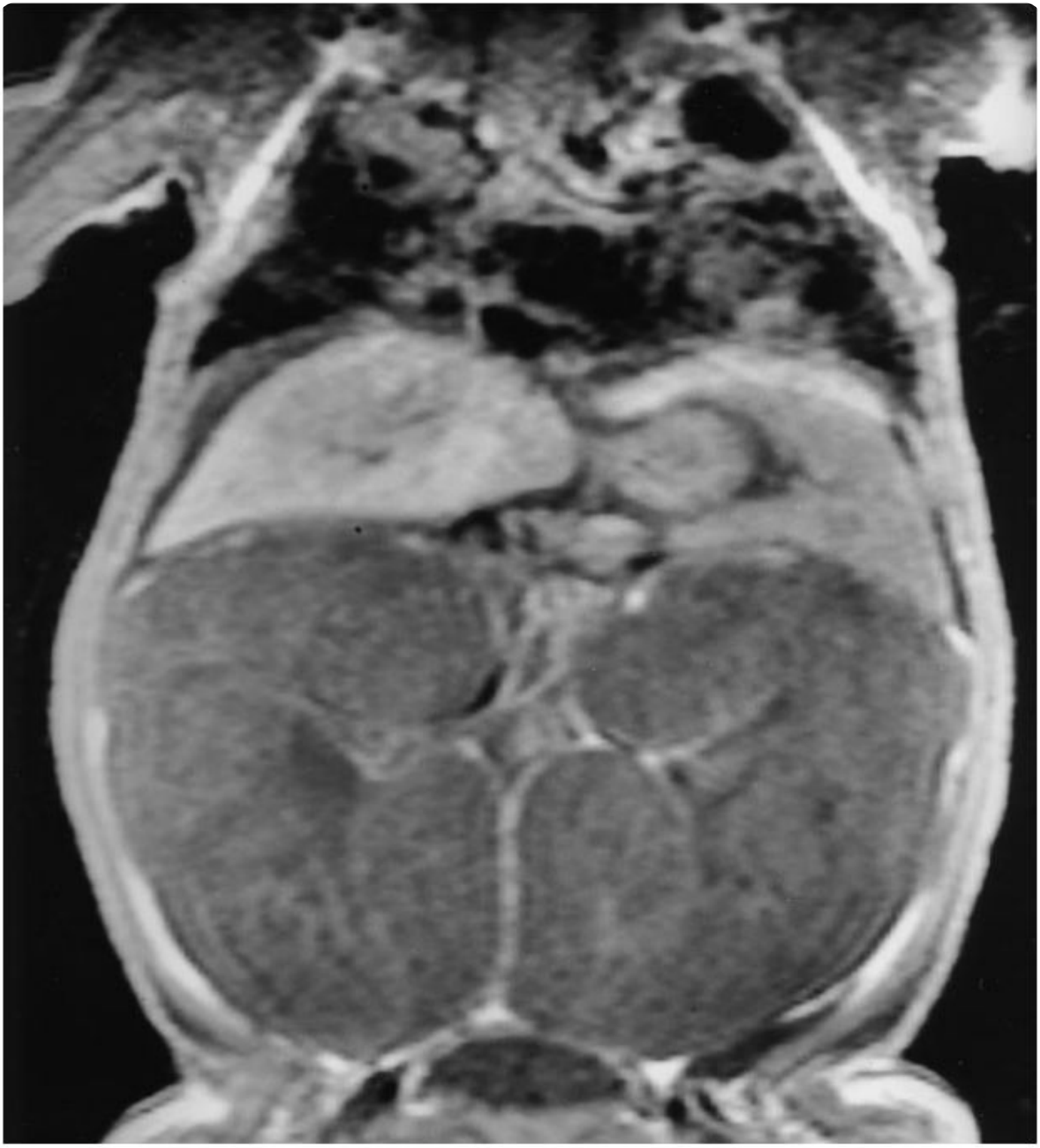
🔶Hyperintense enlarged kidneys on T2W

🔶RARE-MRI (rapid acquisition with relaxation enhancement)- microcystic dilatation & hyperintense, linear radial pattern in the cortex & medulla.

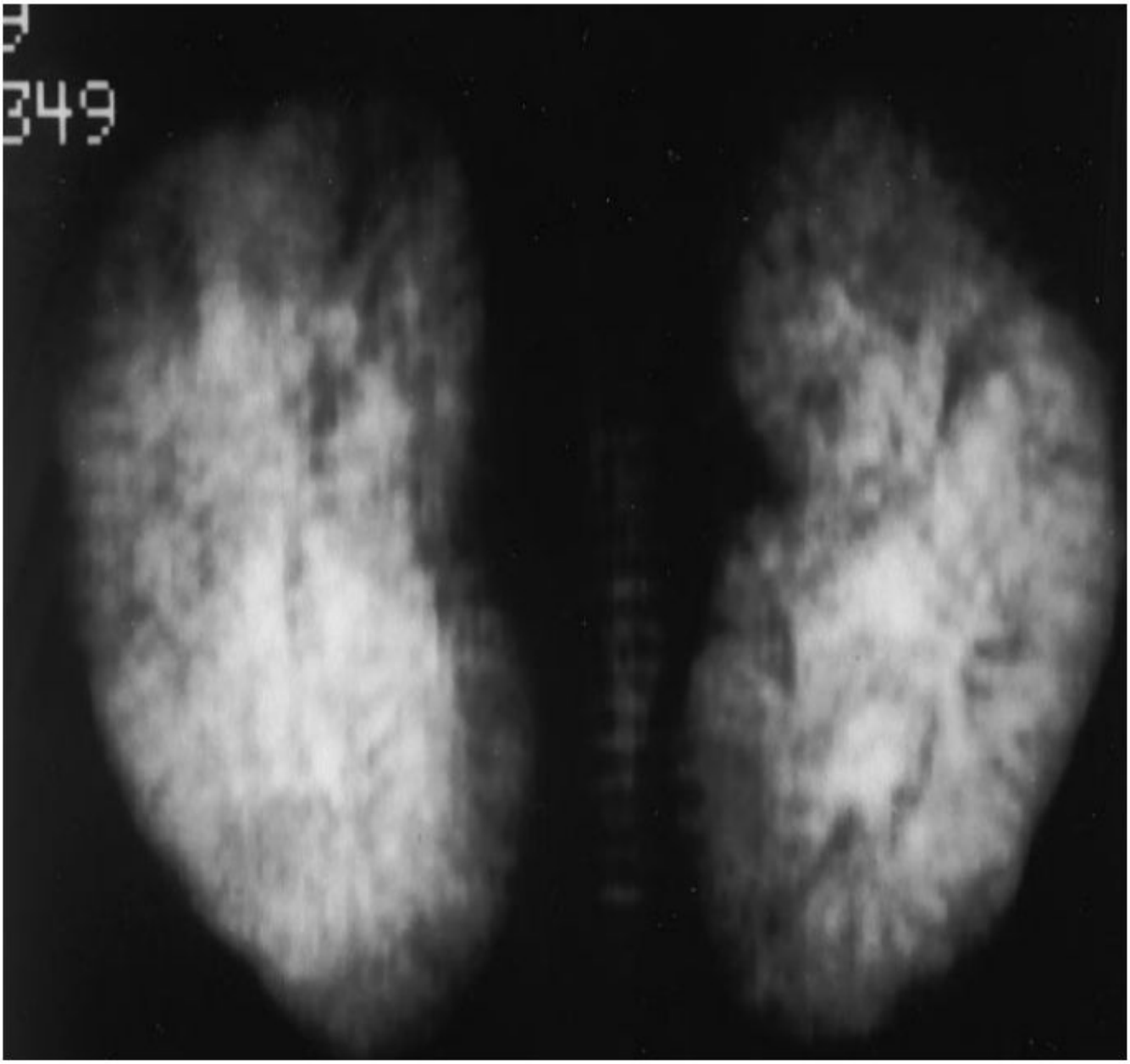
PMID 10755752



Hyperintense kidneys on T2 W

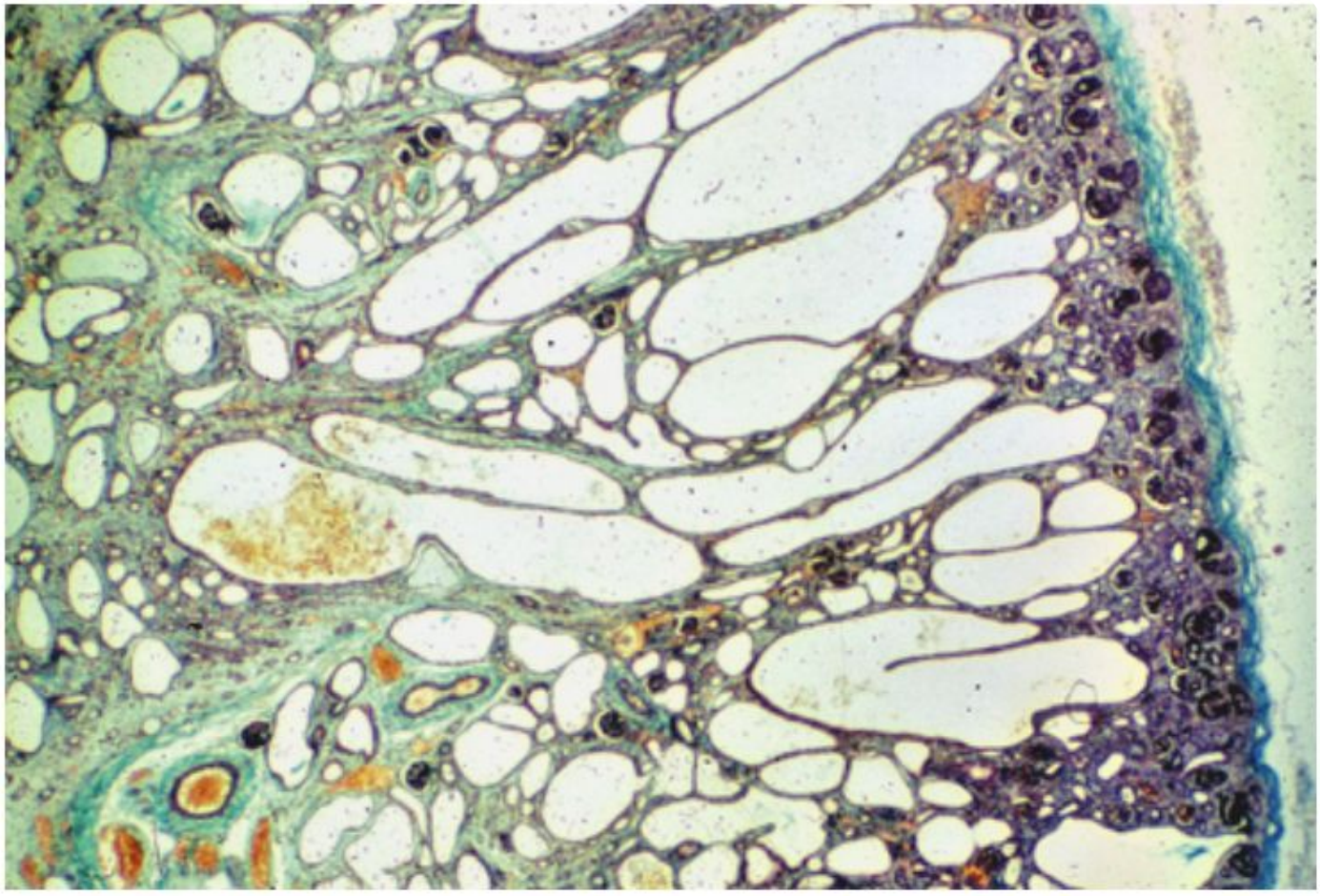


Hypointense kidneys on Coronal view T1W



RARE MR urography showing linear radial pattern in cortex and medulla.

13/ Histopathology of ARPKD shows collecting tubules dilation with cysts and flattened epithelium



2022 UpToDate

14/ Genetic testing is not routinely done in ARPKD.

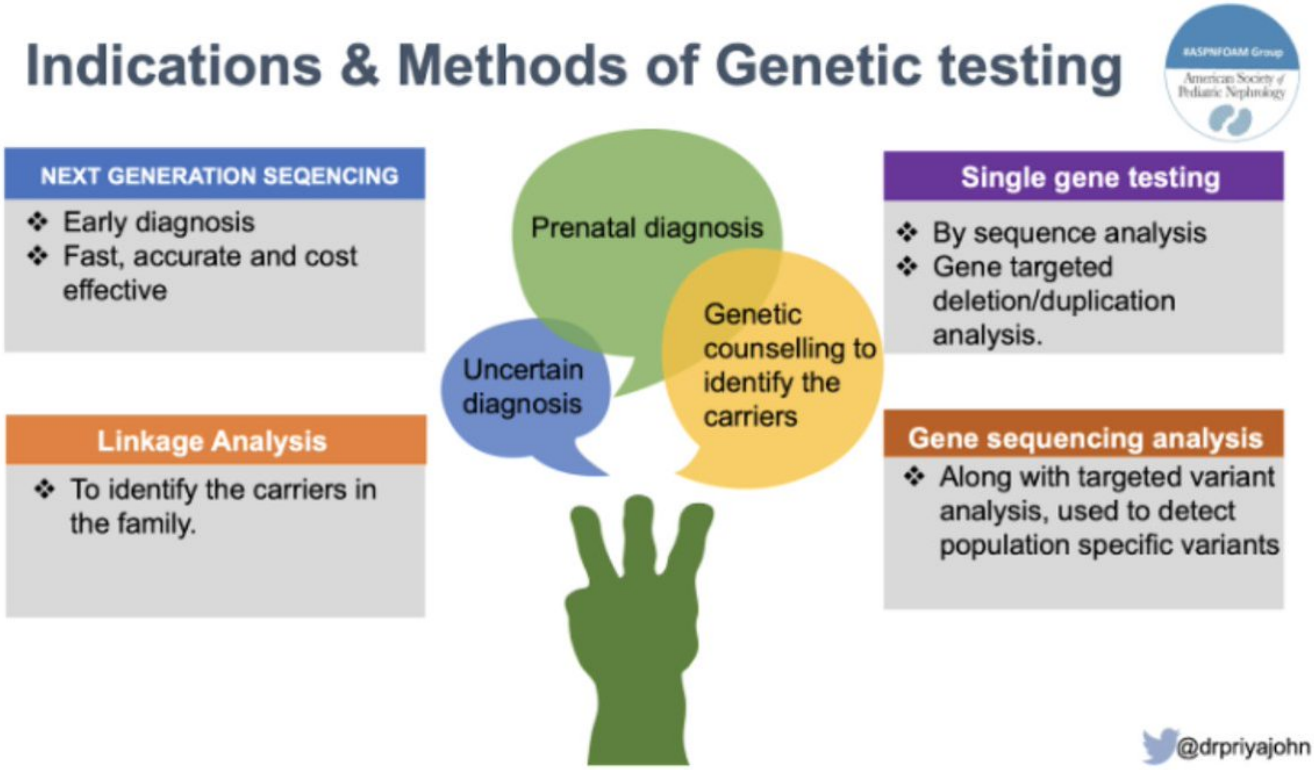
📌Indications include-

🔷Uncertain clinical diagnosis

🔷Preimplantation testing in parents who previously had a baby with ARPKD

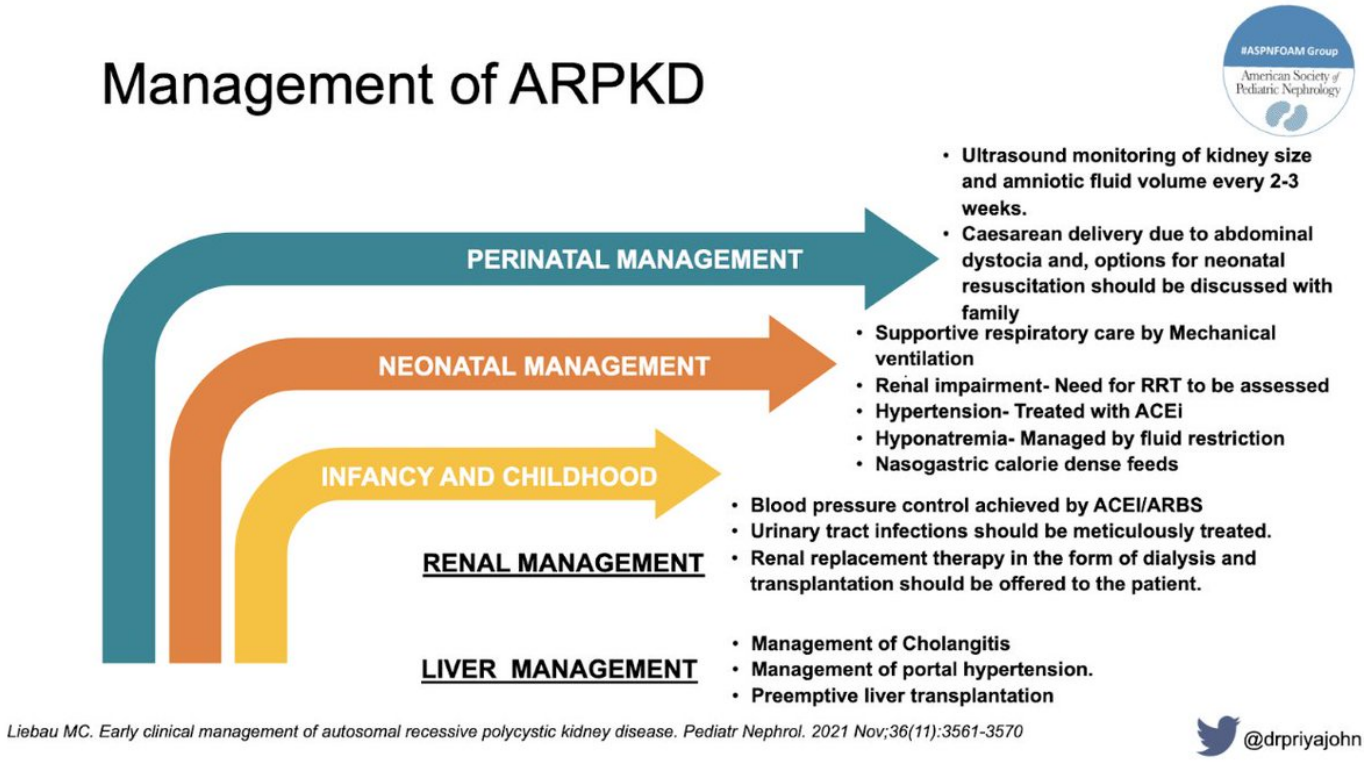
🔷To identify the carriers in the family to facilitate genetic counseling.

15/ Indications and genetic testing methods are listed in the following infographic.
If you would like to know more about genetic testing and kidney disease, here is an excellent tweetorial by @swastithinks



PMID: 21046169

16/ Management of ARPKD needs multidisciplinary approach more so during perinatal and neonatal period. The following infographic summarizes management of ARPKD according to the time of presentation



17/ What of the following is false regarding genetic counseling in ARPKD

18/ Yes, the correct ans is a.

🚩There is a 25% risk of inheriting both pathogenic variants & being affected

🚩50% risk of inheriting a pathogenic variant but a carrier

🚩25% chance of inheriting neither a pathogenic variant nor being a carrier.

Gene reviews Sweeney et al

19/ Long-term outcomes of ARPKD children who survive the first month of life have >80% chance of survival beyond 15 years.
📌 In a study of 164 children with ARPKD-renal survival was 85% at 5 yrs,70% at 10 yrs & 40% at 20 yrs.
📌HTN & ⬇️ liver function in future
PMID 15698423

20/ Which of the following is true with respect to peritoneal dialysis in ARPKD

21/ The answer is c.
📌IPPN(International Pediatric Peritoneal Dialysis Network) studied 79 children on PD with ARPKD , congenital nephrotic syndrome(CNS) and CAKUT.
📌Survival in ARPKD 78% and CNS 73% was comparable and higher in CAKUT 95%.

22/ Overall PD is well tolerated in ARPKD with minor adaptations. A higher UF with lower glucose concentration was observed, likely due to portal HTN.
PMID: 31983502

23/ Big question- When Is nephrectomy advised in ARPKD-ARegPKD study with- comparing children with very early bilateral nephrectomies (<3 mo) (VEBNE) with nephrectomies done (4-15 mo) ;controls- children with very early dialysis (<3mo) without nephrectomy & those with ⬆️ TKV.

24/ Children with VEBNE developed neurological complications and severe hypotensive episodes compared to other study groups.
PMID: 32994492

25/ For further information on expert opinion for the diagnosis and management of ARPKD refer to
PMID: 25015577

For a case-based clinical discussion on #ARPKD with an expert - login to @ASPNeph website, Aug 2022 webinar #Membereducation
Special thanks to #ASPNFOAM group @nefron1310 @SwastiThinks @drM_Sudha