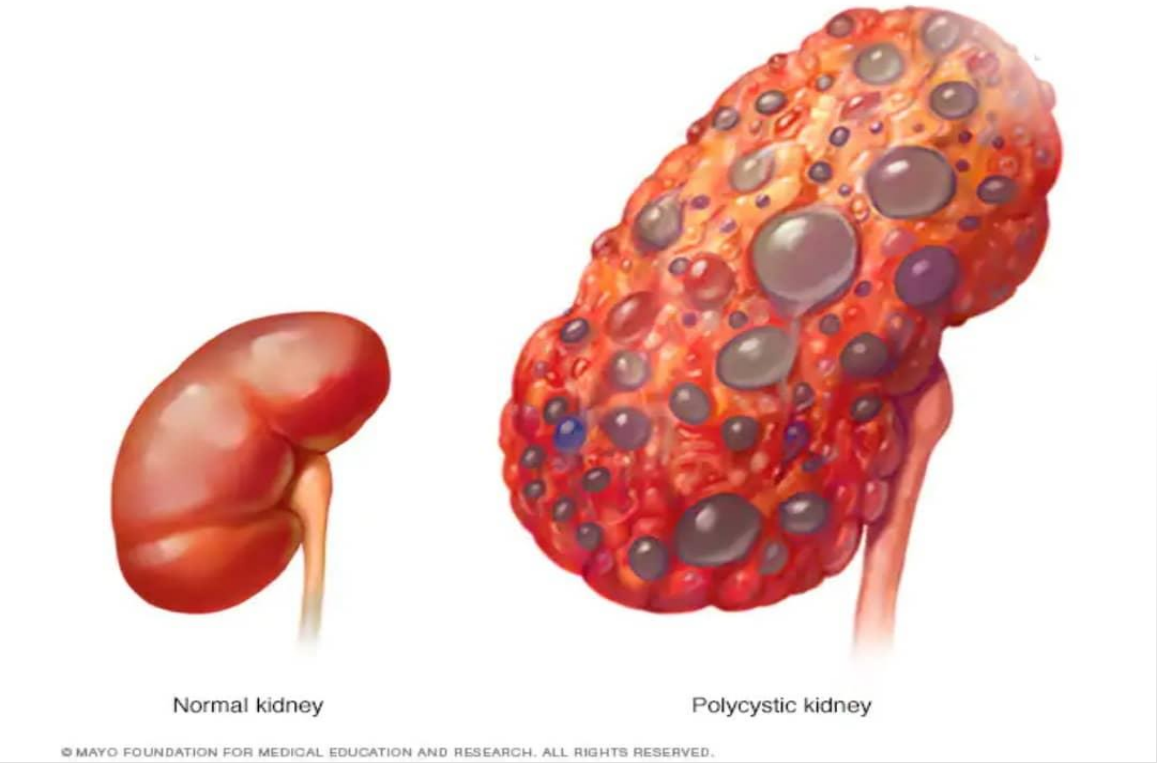

ADPKD Diagnosis

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Concerning Point

- How to diagnosis ADPKD?

  different criteria due to family history situation

  imaging modality and indications

- Predictive factors in ADPKD progression
- Novel tool to predict ADPKD outcome :

  MYO classification

  PROPKD score

- conclusion

When to suspect ADPKD

In the presence of:

- Clinical features
- Family Hx of ADPKD
- Large kidneys with extensive cysts as incidental finding in imaging

VE Torres , et al . ADPKD in adult: epidemiology, clinical presentation and diagnosis; UpToDate. Jan 2021

Diagnosis of ADPKD

- **Initial step:**
- Obtain detailed family history
- Counsel regarding risks and benefits of having an established diagnosis of ADPKD specially in asymptomatic person

What is the important data in family history?

- number and relationship of family members affected,
- their age at diagnosis,
- their age of developing ESKD (if applicable),
- any known genetic mutations in the family,

Diagnostic counseling and screening of family members

- Essential for all patients with suspected ADPKD prior to diagnostic testing:
- Advantages of testing :

  knowledge concerning the diagnosis,

  appropriate family planning,

  detect and treat associated complications

  reassurance of unaffected individuals,

  selection of unaffected relatives as possible kidney transplantation donors

Diagnostic counseling and screening of family members (continued):

- Disadvantages of testing:

  possible difficulties with employment and insurability

  psychologic impact of having a life-altering diagnosis

Diagnostic counseling and screening of family members (continued):

- **Additional genetic counseling related to family planning :**

  discussing the risk of passing the disease to the offspring,

  reproductive options (preimplantation genetic diagnosis)

  risks associated with pregnancy

VE Torres , et al . ADPKD in adult: epidemiology, clinical presentation and diagnosis; UpToDate. Jan 2021

Genetic testing:

- Reserved for atypical cases (equivocal US and MRI findings)
- To rule out ADPKD
- Need to establish an accurate diagnosis (in a young potential kidney donor or prenatal planning)

Fouad T, et al. Autosomal Dominant Polycystic Kidney Disease: Core Curriculum 2016. Am J Kidney Dis. 2016 May;67(5)

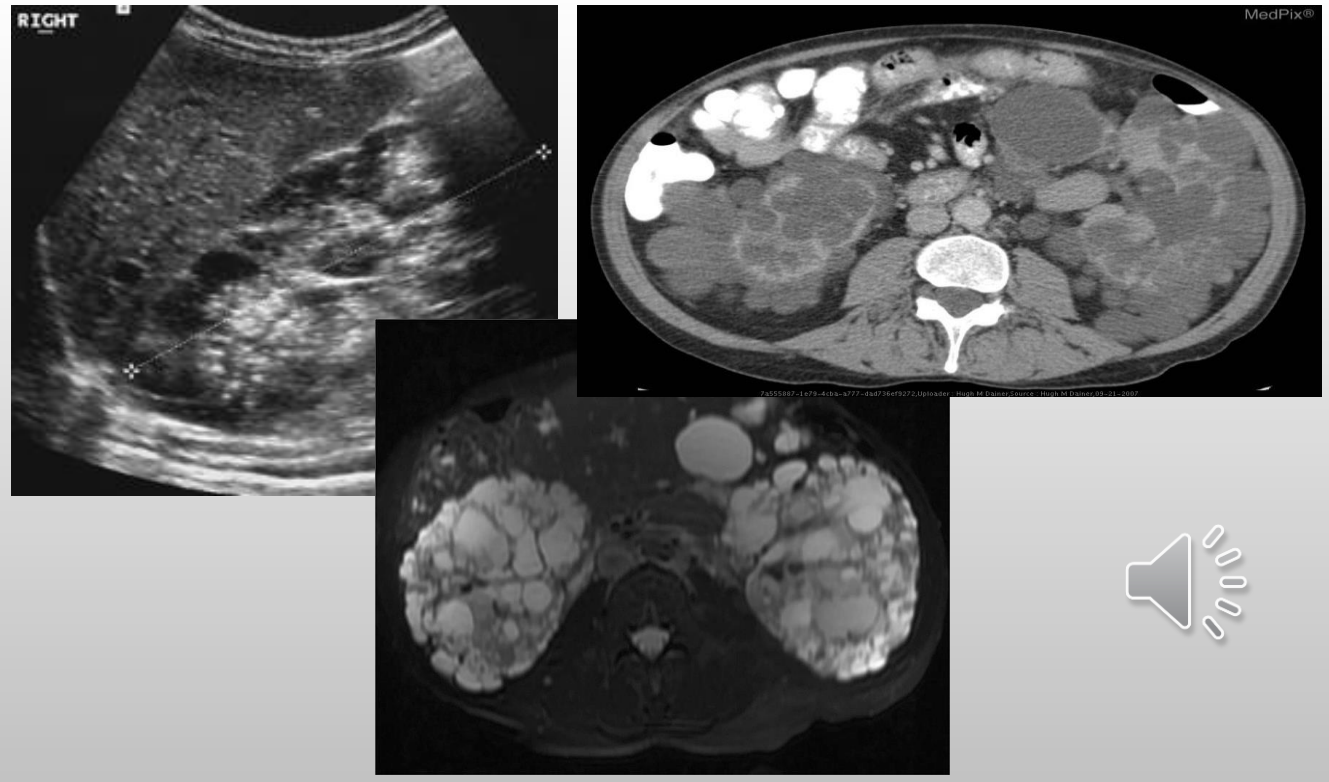
Diagnosis of ADPKD(continued)

- Primary Confirmation : imaging

  US

  CT

  MRI



- What is the choice?

VE Torres , et al. Autosomal dominant polycystic kidney disease. Lancet 2007; 369: 1287–301

Establishing the diagnosis of ADPKD

- 1.patients with a family history of ADPKD:

Criteria depend on : A. genetic type of ADPKD in their family, if known.

B. family history

C. imaging

Establishing the diagnosis of ADPKD(continued):

- In asymptomatic patients with normal kidney function who have a FH of ADPKD:

US : usually is sufficient

is inexpensive and safe,

the most commonly used imaging modality for diagnosis

Establishing the diagnosis of ADPKD(continued)

MRI indication?

US equivocal result generally in patient younger than 40 y/o

further evaluate detected complications in US (kidney mass or complex cysts).

as baseline imaging to calculate htTKV (or CT)

MRI alternative?

Some experts perform genetic testing to confirm the diagnosis, if available.

VE Torres , et al . ADPKD in adult: epidemiology, clinical presentation and diagnosis; UpToDate. Jan 2021

Establishing the diagnosis of ADPKD(continued):

- In FH+ patients with typical findings of ADPKD and/or decreased eGFR:

- CT or MRI : initial option

  benefits:

  baseline image for future comparison

  identify complications of ADPKD or disease in other organs (pancreas)

  to calculate the htTKV (prognostication and planning of treatment)

Establishing the diagnosis of ADPKD(continued):

- CT vs MRI ?
- MRI is preferred imaging modality for size determination.
- risk of contrast exposure with CT?

eGFR ≥60 mL/min/1.73 m : we perform a CT +_ contrast:

unenhanced CT: calculation of TKV

identification of any stones in the collecting system

contrast-enhanced CT: differentiation between cystic and non cystic tissue,

assessment of cyst burden

Establishing the diagnosis of ADPKD(continued):

eGFR <60 mL/min/1.73 m : we prefer MRI W/O GAD .

distinguish between cystic and noncystic tissue

is unable to reliably detect kidney stones or parenchymal calcifications.

VE Torres , et al . ADPKD in adult: epidemiology, clinical presentation and diagnosis; UpToDate. Jan 2021

Imaging Criteria for establishing the diagnosis of ADPKD

- US-based criteria of ADPKD in patients with a positive FH with machines capable to detect cysts that were1cm or more in diameter.
- most contemporary ultrasound machines detect kidney cysts of 5 mm or more thus, sensitivity of cyst detection has been increased. (from 82 to 97 % in one study).
- It is center- and operator-dependent

Imaging Criteria for establishing the diagnosis of ADPKD(continued):

- But:

  we continue to use these criteria for diagnosis in individuals with a family history of ADPKD.

  we use these same criteria for CT or MRI too.

Pei Y, et al. Imaging-based diagnosis of autosomal dominant polycystic kidney disease. J Am Soc Nephrol 2015; 26:746

Imaging Criteria for diagnosis of ADPKD

Ultrasound-based criteria for diagnosis and exclusion of ADPKD among patients with a positive family history

Diagnostic purpose	Age (years)*	Imaging findings	Family history of *PKD1*	Family history of *PKD2*	Family history with unknown gene type
Confirmation					
	15 to 29	Total of ≥3 cysts ¶	PPV, 100% Sensitivity, 94.3%	PPV, 100% Sensitivity, 69.5%	PPV, 100% Sensitivity, 81.7%
	30 to 39	Total of ≥3 cysts ¶	PPV, 100% Sensitivity, 96.6%	PPV, 100% Sensitivity, 94.9%	PPV, 100% Sensitivity, 95.5%
	40 to 59	≥2 cysts in each kidney	PPV, 100% Sensitivity, 92.6%	PPV, 100% Sensitivity, 88.8%	PPV, 100% Sensitivity, 90.0%
Exclusion					
	15 to 29	No kidney cyst	NPV, 99.1% Specificity, 97.6%	NPV, 83.5% Specificity, 96.6%	NPV, 90.8% Specificity, 97.1%
	30 to 39	No kidney cyst	NPV, 100% Specificity, 96.0%	NPV, 96.8% Specificity, 93.8%	NPV, 98.3% Specificity, 94.8%
	40 to 59	No kidney cyst	NPV, 100% Specificity, 93.9%	NPV, 100% Specificity, 93.7%	NPV, 100% Specificity, 93.9%

ADPKD: autosomal dominant polycystic kidney disease; *PKD1*: polycystic kidney disease 1 locus; *PKD2*: polycystic kidney disease 2 locus; PPV: positive predictive value; NPV: negative predictive value

Chapman AB, et al. Autosomal-dominant polycystic kidney disease (ADPKD): executive summary from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney Int 2015; 88:17

Imaging Criteria for establishing the diagnosis of ADPKD(continued):

Age	Number of cysts
15–39 years	At least 3 unilateral or bilateral kidney cysts
40–59 years	At least 2 cysts in each kidney
Above 60 years	At least 4 cysts in each kidney

Note: Data from Pei et al.[14]

Pei Y, et al. Unified criteria for ultrasonographic diagnosis of ADPKD. J Am Soc Nephrol. 2009;20(1):205–212

Imaging criteria for exclusion the diagnosis of ADPKD

- US: NO kidney cyst
- MRI : No absolute criteria up to now

- In one study including 73 affected (positive genetic testing) and 83 nonaffected (negative genetic testing) individuals has been reported that fewer than five cysts by MRI is sufficient to exclude the diagnosis of ADPKD in potential living-related kidney donors .

Pei Y, et al. Imaging-based diagnosis of autosomal dominant polycystic kidney disease. J Am Soc Nephrol 2015; 26:746

Establishing the diagnosis of ADPKD(continued):

- 2.patient without a family history of ADPKD:

  25% of ADPKD cases

- In most such cases: A. the affected parent has died without a diagnosis

  B. alive with a undetected mild form of the disease

- What will be helpful?

  Get medical information or imaging studies of parents or other family members

VE Torres , et al . ADPKD in adult: epidemiology, clinical presentation and diagnosis; UpToDate. Jan 2021

Criteria for diagnosis of ADPKD

- NO established imaging-based criteria for diagnosis:
  1. We diagnose if they have 10 or more cysts (≥5 mm) in each kidney, if the kidneys are enlarged or liver cysts are noted, with no obvious features of a different cystic disorder .
  2. Genetic testing : equivocal imaging results, to establish an accurate diagnosis

In 5% of cases there is new mutation or due to mosaicism

Tan AY, et al. Autosomal dominant polycystic kidney disease caused by somatic and germline mosaicism. Clin Genet 2015;87:373

Differential diagnosis

- Helpful clues in differential diagnosis:

  age of the patient

  family history of other genetic disorders

  presence of associated manifestations

VE Torres , et al . ADPKD in adult: epidemiology, clinical presentation and diagnosis; UpToDate. Jan 2021

Differential diagnosis(continued):

- Acquired disorders in adults in the absence of a FH of ADPKD include:

1. Medullary sponge kidney: AD inheritance in some cases

   renal cortex is spared on CT or MRI

2. Multiple benign simple cysts : difficult to differentiate from a mild

   ADPKD

3. Localized renal cystic disease : neither bilateral nor progressive

differential diagnosis(continued):

4. Acquired renal cystic disease : no FH of ADPKD and the kidneys are small to normal in size with a smooth contour , absence of the extrarenal features of ADPKD

5. bilateral parapelvic cysts : The lack of cysts in the cortex

Fouad T, et al. Autosomal Dominant Polycystic Kidney Disease: Core Curriculum 2016. Am J Kidney Dis. 2016 May;67(5)

Differential diagnosis(continued):

- Genetic disorders in adults in the absence of a family history of ADPKD :

1. Autosomal recessive polycystic kidney disease (ARPKD) :

Extrarenal (hepatic, pancreatic) cysts favor the presence of ADPKD,

while portal fibrosis or signs of portal hypertension, cholangitis, or

biliary dysgenesis favor the diagnosis of ARPKD.

US of parents of children with ARPKD will not show cysts.

Genetic testing may also be helpful in some cases

Zerres K, et al. New options for prenatal diagnosis in autosomal recessive polycystic kidney disease by mutation analysis of the PKHD1 gene. Clin Genet 2004; 66:53.

Differential diagnosis(continued):

2. Autosomal dominant tuberous sclerosis complex: presence of other features of the disease
3. von Hippel-Lindau disease : presence of other features of disease
4. Autosomal dominant tubulointerstitial kidney disease(MCKD): cysts at the corticomedullary junction, small-to-normal-size kidneys, hyperuricemia, and gout.

Differential diagnosis(continued):

5.Autosomal dominant hepatocyte nuclear factor-1beta (hnf-1b) nephropathy: presence other features of disease

6. Autosomal dominant polycystic liver disease (ADPLD): usually little or no kidney cyst burden

family history and genetic testing maybe helpful.

7. X-linked dominant orofaciodigital syndrome type I (OFD1): presence of extrarenal manifestations

VE Torres , et al . ADPKD in adult: epidemiology, clinical presentation and diagnosis; UpToDate. Jan 2021

Predictive factors of ADPKD progression

- ADPKD： substantial variability in its natural course within and between families
- Predicting factors associated with early adverse structural and/or functional outcomes:

  PKD1 mutation (particularly truncating mutation), men, early onset of HTN, early and frequent gross hematuria, and

  three or more pregnancies(in women)

  TKV (increases in TKV and decreases in GFR and renal blood flow greater than expected for a given age)

Schrier RW, et al. Predictors of autosomal dominant polycystic kidney disease progression. J Am Soc Nephrol 2014;25:2399.

Table 1. Methodologies to Determine Patients With ADPKD at Higher Risk for Progressive Disease

Method	Required Elements	Predicted Outcome	Advantages	Limitations
Genotyping[18,19,20,43,44]	• Genotype (*PKD1* vs *PKD2)* • Protein-truncating vs nontruncating mutation	• Earlier age of ESRD onset • Larger TKV and increased cyst numbers at any age • Severity: PKD1 protein-truncating > PKD1 nontruncating > PKD2	• Identification of familial mutation helpful to determine prognosis	• Expensive and not routinely obtained • Large no. of mutations, not all of which are associated with known prognostic significance • Effect of genotype as a prognostic factor is not always evident when adjusted for TKV[14-16]
CRISP2 & CRISP3[14,30]	• htTKV by MRI	• Larger htTKV predictive of greater decline in GFR	• Most accurate • Available in many facilities	• Evidence is based on a relatively small cohort of 241 pts selected for high risk for progression
PROPKD score[21]	Point score (0-9) based on: • Sex • Genotype (*PKD1* vs *PKD2),* protein-truncating vs nontruncating mutation • Clinical characteristics such as hypertension or urologic symptoms before age 35 y	• Score > 6 associated with high likelihood of ESRD by age 60 y • Score < 3 associated with low likelihood of ESRD before age 60 y	• No requirement for imaging • Validated in external populations[45]	See limitations of genotyping above
PKDOC[15,16,46]	• TKV, age, & eGFR	• Risk for 30% decline in eGFR • Risk for ESRD	• Uses TKV measured by any modality (ultrasound, CT, MRI) • Validated as a prognostic biomarker by FDA and EMA	• Modeling applied to population of participants, not an individual
Mayo Imaging Classification[17]	• Age & htTKV to calculate estimated TKV growth rate in pts with typical ADPKD (cysts scattered throughout both kidneys)	• Risk for GFR decline & progression to ESRD	• Most precise of predictive methodologies • Validated in independent populations & in secondary analyses of clinical trials[23,24]	• Based on MRI assessment of htTKV using ellipsoid formula that is not as accurate as formal assessment of TKV by stereology or boundary tracing • CT is likely sufficiently accurate but has not been specifically evaluated • Not applicable to atypical ADPKD[17]
Image texture analysis[22,41]	Age, eGFR, & htTKV with selected MRI characteristics of kidneys (entropy, correlation, & energy)	• Risk for CKD3 & 30% reduction in eGFR in 8 y	• Uses additional information from images obtained during MRI assessment of TKV	• Research methodology applied to retrospective cohort of 122 CRISP pts with GFRs > 70 mL/min at baseline • Not studied in other populations

Abbreviations and definitions: ADPKD, autosomal dominant polycystic kidney disease; CKD, chronic kidney disease; CRISP, Consortium for Radiological Imaging Studies of Polycystic Kidney Disease; CT, computed tomography (imaging); eGFR, estimated glomerular filtration rate; EMA, European Medicines Agency; ESRD, end-stage renal disease (maintenance dialysis or transplantation); FDA, US Food and Drug Administration; GFR, glomerular filtration rate; htTKV, height-adjusted total kidney volume; MRI, magnetic resonance imaging; PKDOC, Polycystic Kidney Disease Outcomes Consortium; pt, patient; TKV, total kidney volume.

Kimberly A ,et al. Addressing the Need for Clinical Trial End Points in Autosomal Dominant Polycystic Kidney Disease: A Report From the Polycystic Kidney Disease Outcomes Consortium (PKDOC). AJKD Vol XX | Iss XX | Month 2018

Preferred models to determine progressive disease

- CRISP study
- MAYO classification score
- PROPKD score

PROPKD score and MAYO imaging classification are examples of scoring systems that take into consideration the relationships between age and kidney size, along with disease progression.(clinical trial)

Clinical Trial End Points in ADPKD. Posted on June 6, 2019 by AJKD blog in Commentary // 0 Comments

Determine progressive disease :

- **The CRISP study(Consortium for Radiologic Imaging Studies of PKD1and 2):**
- A longitudinal study of 15-46 Y/O patients with creatinine clearance >= 70 ml/min, characterized the relationship between total kidney volume (TKV) and GFR.
- MRI at baseline illustrated huge phenotypic variability.
- The rate of kidney growth is quasi-exponential, unique to and variable among patients, and kidney growth precedes decline in GFR .

Determine progressive disease (continued):

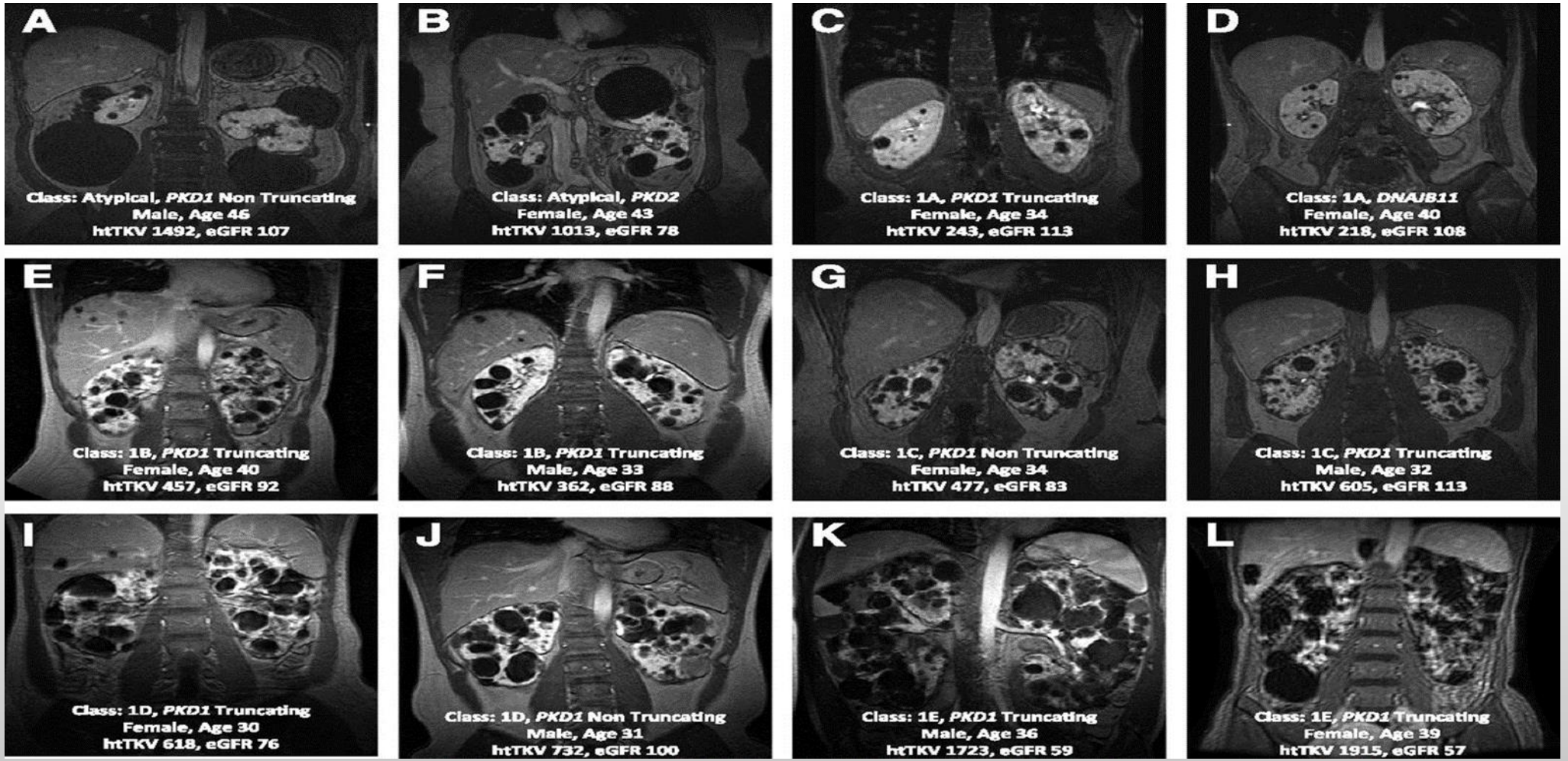
CRISP study(continued):

- TKV was the best predictor of eGFR decline.

  TKV changed significantly year after year, GFR started declining years later.

- Its value is limited in atypical cases with markedly asymmetric or coexisting ischemic disease.

Fouad T ,et al. Recent advances in the management of autosomal dominant polycystic kidney disease: clin j am soc Nephrol 13: 1765–1776, 2018



Imaging of different patients with ADPKD who enrolled in the CRISP study (creatinine clearance ≥70 ml/min), showing the large spectrum of disease severity from very mild to very severe disease at the baseline visit.

Determine progressive disease (continued):

- As in other article:
- TKV for use as a prognostic biomarker does not require high precision.
- measurements by the ellipsoid equation and various imaging modalities can be used to inform patients about their prognosis .
- Height- adjusted total kidney volume (htTKV) in concert with age predicts future GFR decline.

Fouad T, et al. Recent advances in the management of autosomal dominant polycystic kidney disease: clin j am soc Nephrol 13: 1765–1776, 2018

Determine progressive disease (continued):

- MAYO imaging classification :

IT is a simple tool that uses htTKV by MRI(or CT) and age to identify patients at the highest risk for progression independent of kidney function.

predict eGFR at any point in future using TKV at any given age(15-80).

MAYO imaging classification (continued):

- It uses criteria to exclude atypical cases and stratify typical cases(class1) into five classes (A–E) on the basis of growth rates per year estimated from patient age and a theoretical starting height-adjusted TKV (150 ml/m) with high precision modality(planimetry (the gold standard) or stereology).
- Typical cases include about 95% of ADPKD cases

María V. Imaging Classification of Autosomal Dominant Polycystic Kidney Disease: A Simple Model for Selecting Patients for Clinical Trials. J Am Soc Nephrol 26: 160–172, 2015

Table 1:

Classification of ADPKD patients by imaging

Class 1: Typical	Bilateral and diffuse distribution, with mild, moderate, or severe replacement of kidney tissue by cysts, where all cysts contribute similarly to TKV.
	Measure TKV (Could be done by nephrologist or radiologist in few minutes) http://www.mayo.edu/research/documents/pkd-center-adpkd-classification/doc-20094754

Fouad T, et al. Recent advances in the management of autosomal dominant polycystic kidney disease: clin j am soc Nephrol 13: 1765–1776, 2018

Determine progressive disease (continued) :

- 5 percent of patients : atypical cases (class 2),

  htTKV does not predict eGFR decline.

  most patients with class 2 have focal cystic disease, and a few are older

  individuals with atrophic kidneys with cysts.

- Mayo imaging classification shown to be informative in post-hoc analyses of several clinical trials.

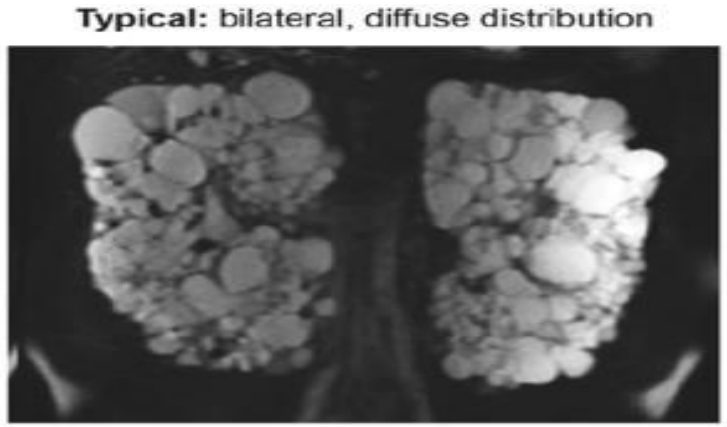
María V. Imaging Classification of Autosomal Dominant Polycystic Kidney Disease: A Simple Model for Selecting Patients for Clinical Trials. J Am Soc Nephrol 26: 160–172, 2015

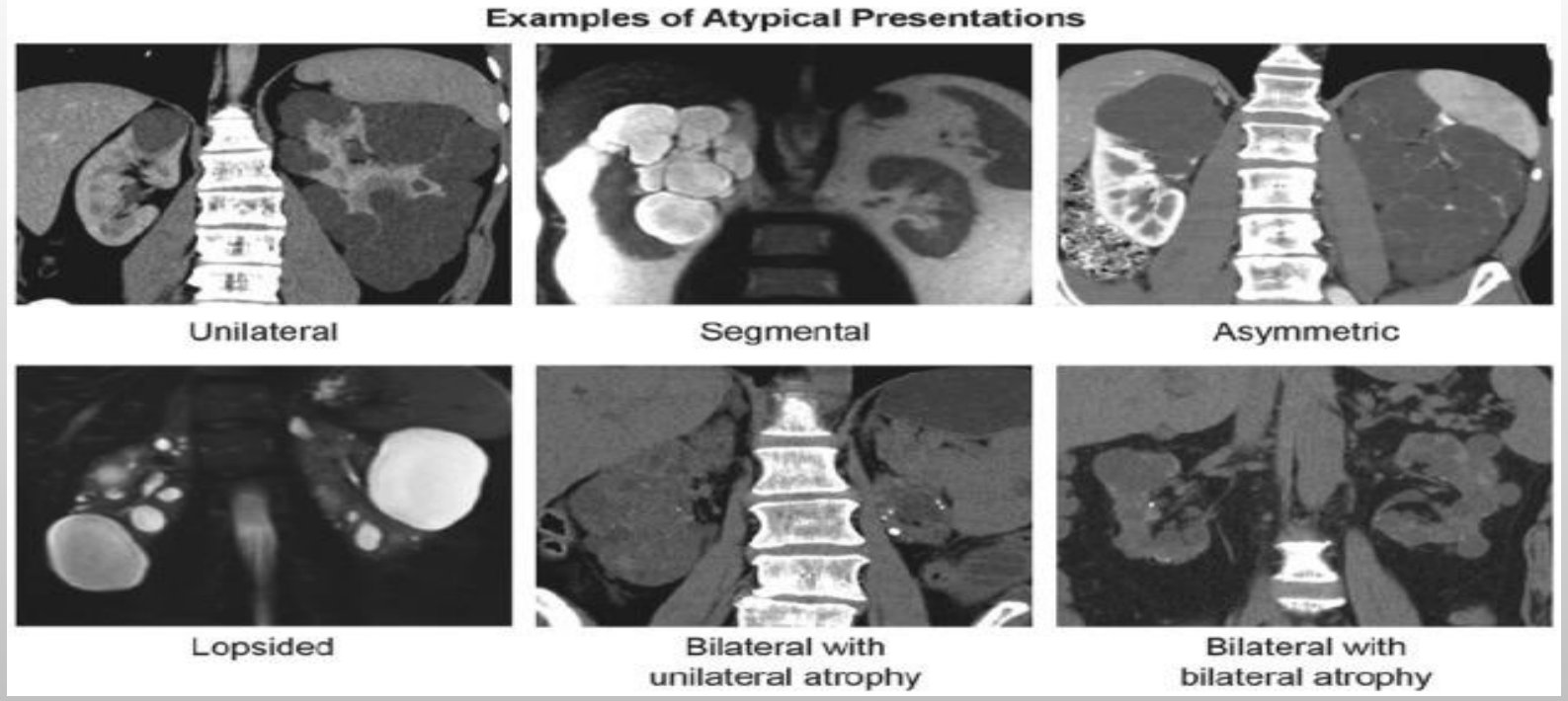
Class 2: Atypical		
Class 2 A:	Unilateral	Diffuse cystic involvement of one kidney causing marked kidney enlargement with a normal contralateral kidney defined by a normal kidney volume (<275 ml in men; <244 ml in women) and having no or only 1−2 cysts
	Segmental	Cystic disease involving only one pole of one or both kidneys and sparing the remaining kidney tissue
	Asymmetric	Diffuse cystic involvement of one kidney causing marked kidney enlargement with mild segmental or minimal diffuse involvement of the contralateral kidney defined by a small number of cysts (>2 but <10) and volume accounting for <30% of TKV
	Lopsided	Bilateral distribution of kidneycysts with mild replacement of kidney tissue with atypical cysts where ≤5 cysts account for ≥50% TKV (the largest cyst diameter is used to estimate individual cyst volume)

Class 2 B:	Bilateral presentation with acquired unilateral atrophy	Diffuse cystic involvement of one kidney causing moderate to severe kidney enlargement with contralateral acquired atrophy.
	Bilateral presentation with bilateral kidney atrophy	Impaired kidney function (serum creatinine≥1.5 mg/dl) without significant enlargement of the kidneys, defined by an average length <14.5 cm, and replacement of kidney tissue by cysts with atrophy of the parenchyma.

Fouad T , et al. Recent advances in the management of autosomal dominant polycystic kidney disease: clin j am soc Nephrol 13: 1765–1776, 2018

Mayo clinic classification:

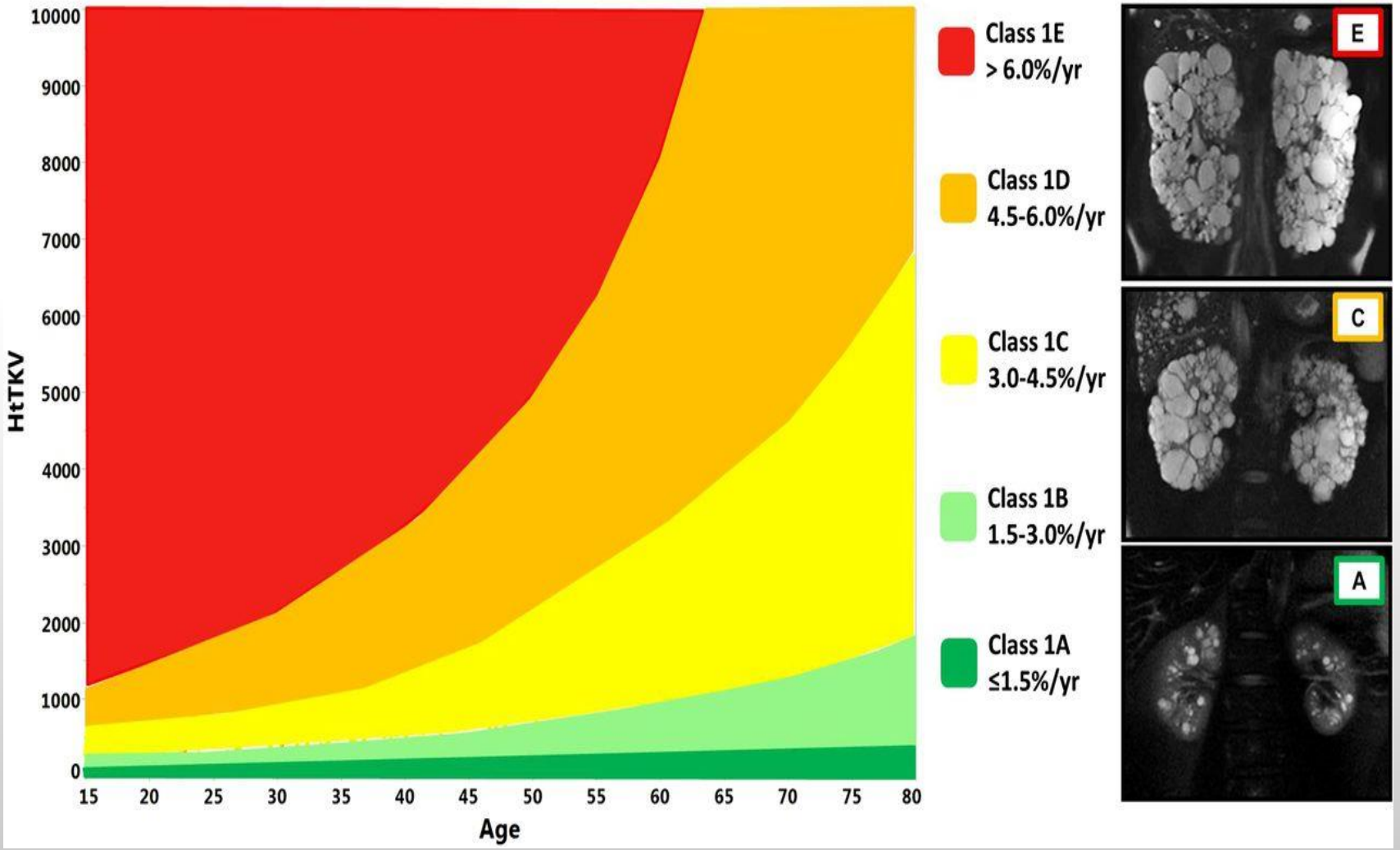




Soroka S, et al. Assessing Risk of Disease Progression and Pharmacological Management of Autosomal Dominant Polycystic Kidney Disease: A Canadian Expert Consensus. Can J Kidney Health Dis. 2017 Mar 1;4:2054358117695784.

Class 1E,1D,1C are defined as high risk for progression to ESKD:

10y ESKD rate was 2.2% in 1C and 22.3% in 1E class.



Mayo Clinic classification diagram (modified from Nephrology, Dialysis and Transplantation, in press). HtTKV, height adjusted total kidney volume (ml/m).

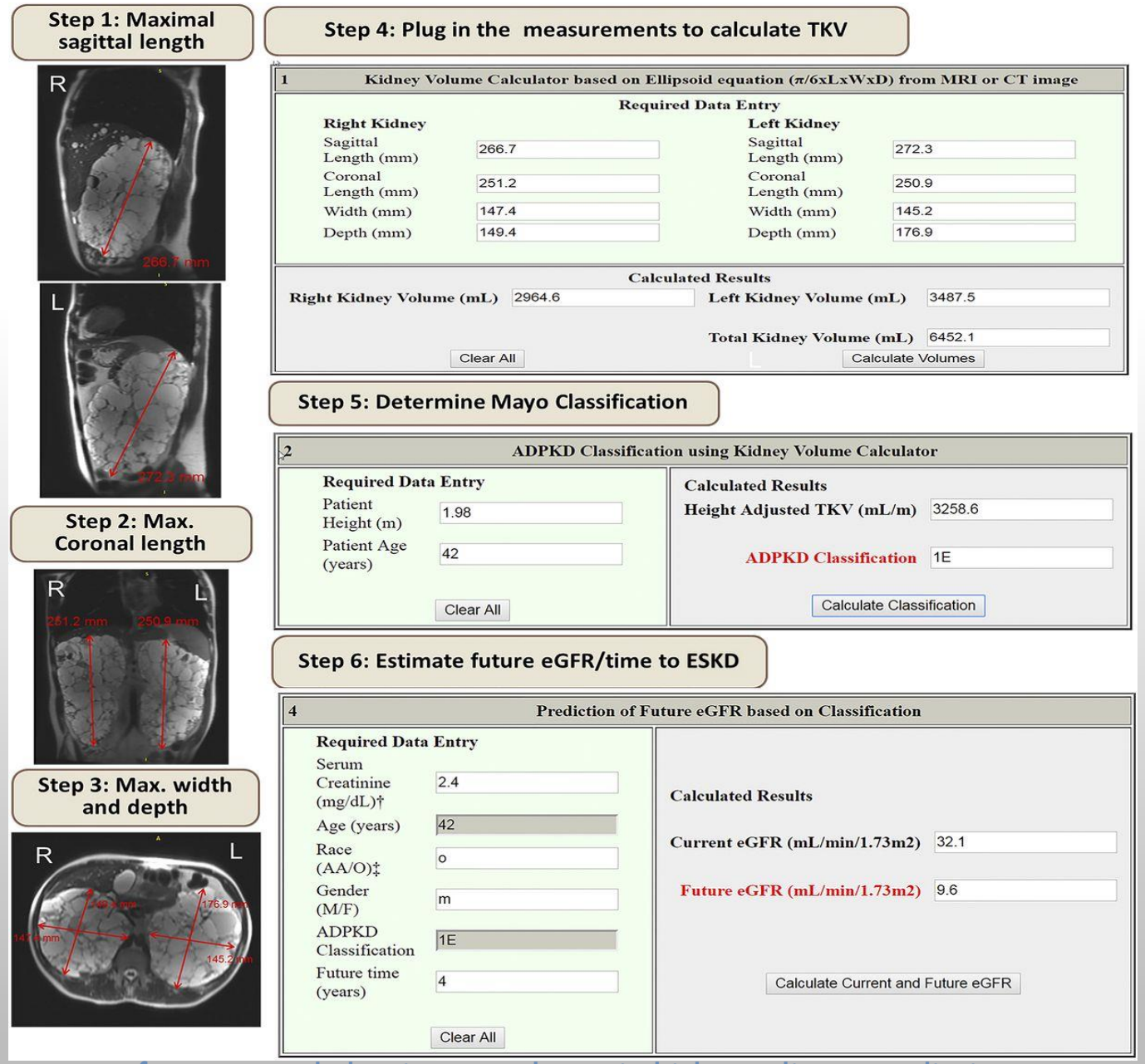
Fuad T , et al. Recent advances in the management of autosomal dominant polycystic kidney disease: clin j am soc Nephrol 13: 1765–1776, 2018

example depicting stepwise method to calculate TKV in the clinician's office using the mayo clinic ADPKD classification online tool.

This classification should be applied only to patients previously classified as Typical* ADPKD, ages 15-80.

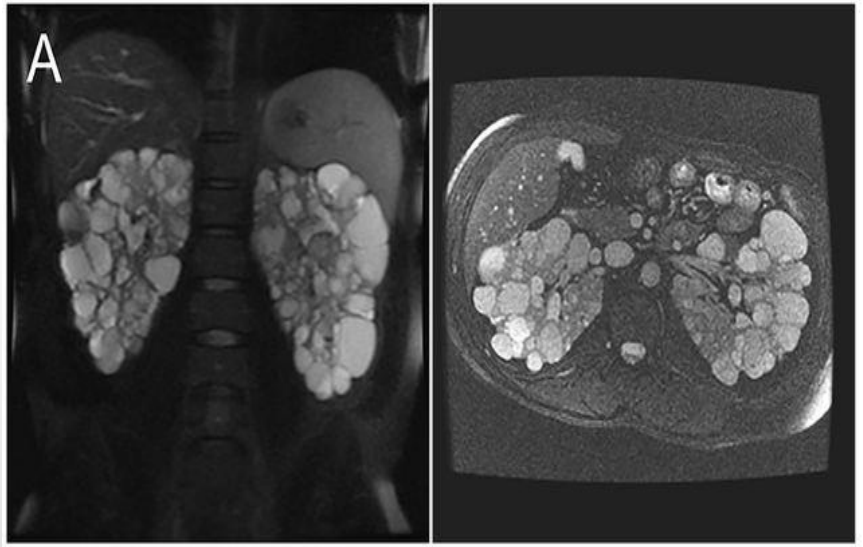
The classification is based on patient's htTKV and Age.
The Kidney Volume Calculator can be used to estimate patient's TKV using simple measurements from MRI or CT images.

If TKV has been previously calculated by Stereology technique, go straight to different calculator.

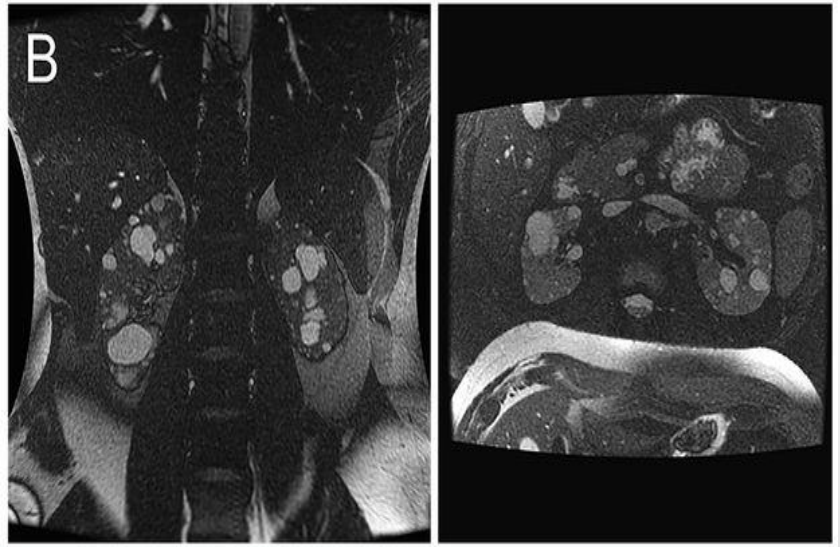


Fouad T, et al. Recent advances in the management of autosomal dominant polycystic kidney disease: clin j am soc Nephrol 13: 1765–1776, 2018

Two patients with ADPKD :
same age and GFR, substantial difference in prognosis :
on the basis of TKV and predicted rate of decline of eGFR.

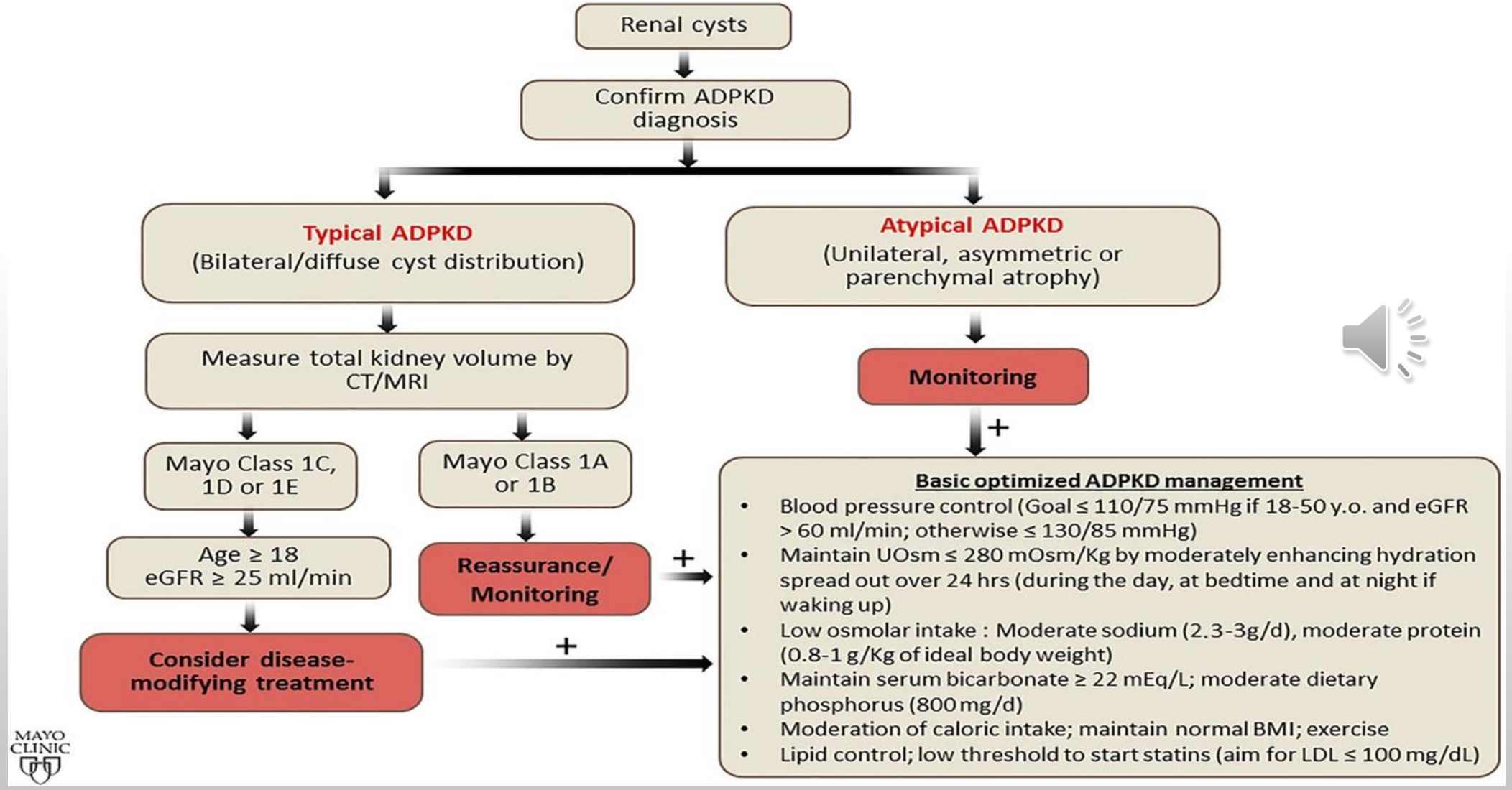


Patient A: 27 y.o male with eGFR 91 ml/min and TKV of 2064 ml
Prognosis: Mayo Class 1E, predicted ESKD date at age of 42
Recommendation: Disease-modifying treatment to slow disease progression



Patient B: 27 y.o male with eGFR 91 ml/min and TKV of 652 ml
Prognosis: Mayo Class 1B, predicted ESKD date at age of 70
Recommendation: Monitoring

Fouad T, et al. Recent advances in the management of autosomal dominant polycystic kidney disease: clin j am soc Nephrol 13: 1765–1776, 2018



Basic universal management should be recommended to all patients with ADPKD

Fuad T , et al. Recent Advances in the Management of Autosomal Dominant Polycystic Kidney Disease: Clin J Am Soc Nephrol 13: 1765–1776, 2018

Determine progressive disease (continued):

PROPKD score:

(Predicting Renal Outcome in Polycystic Kidney Disease)

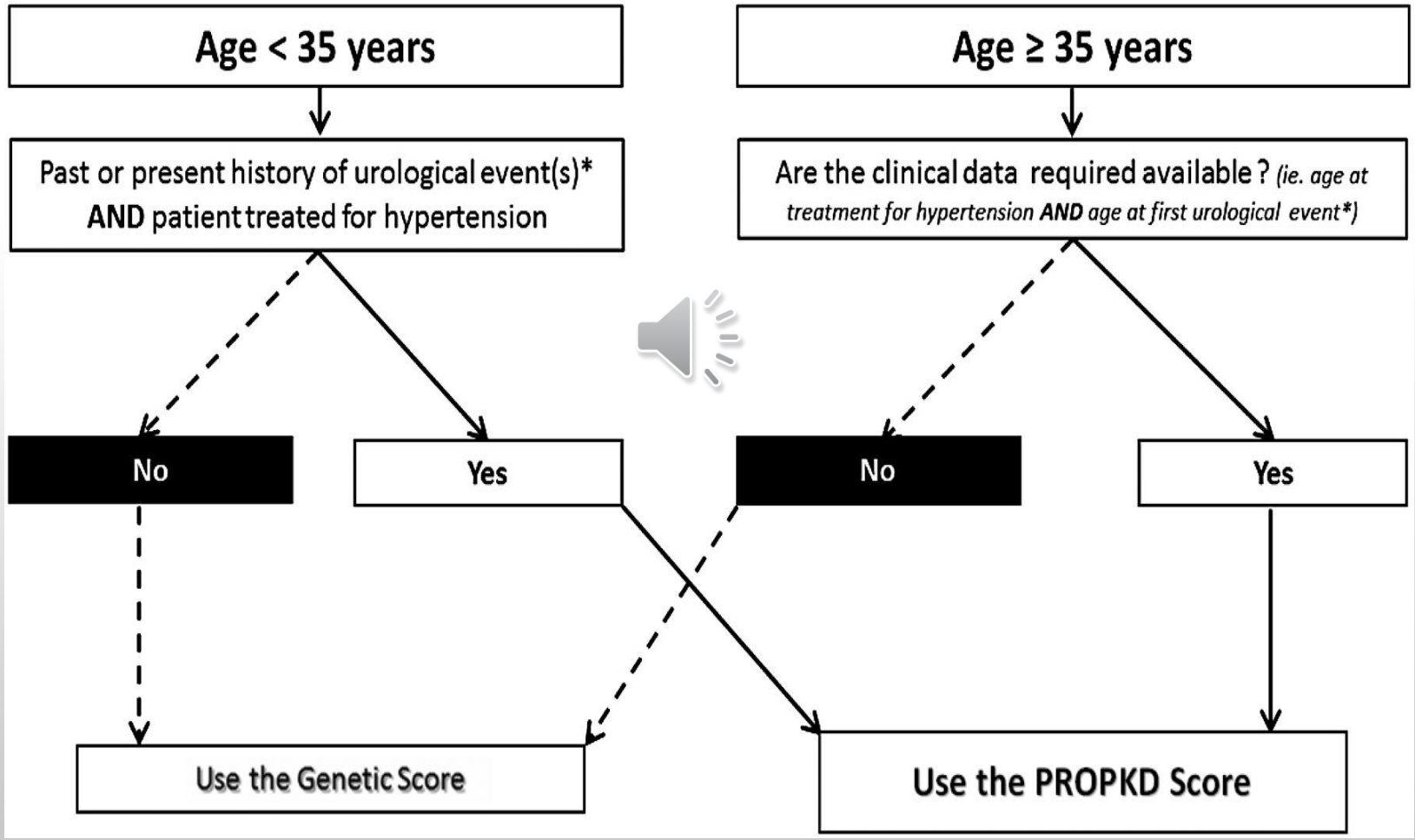
- prognostic model to predict renal outcomes in patients with ADPKD on the basis of genetic and clinical data.

  scoring system from 0 to 9:

Factor	Points
Male	1
Hypertension before age 35 y	2
First urological event before age 35 y	2
PKD2 mutation	0
Nontruncating *PKD1* mutation	2
Truncating *PKD1* mutation	4

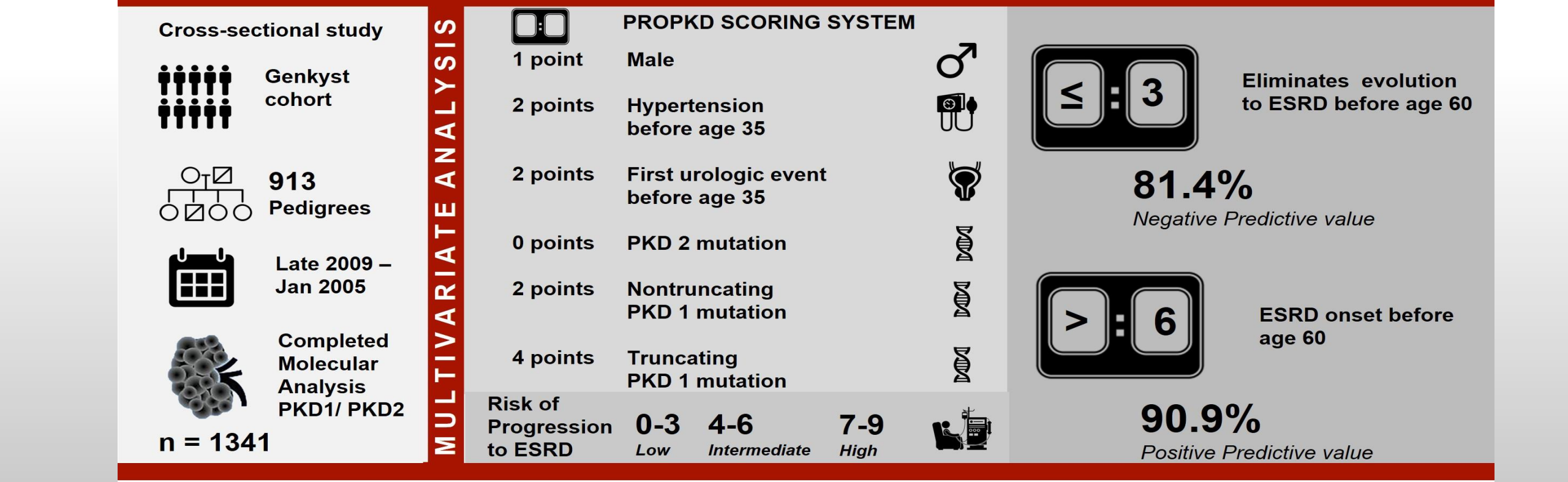
Cornec-Le Gall E ,et al. The PROPKD Score: A New Algorithm to Predict Renal Survival in Autosomal Dominant Polycystic Kidney Disease. JASN March 2016, 27 (3) 942-951

It can be used for patients younger than 35y/o who have already experienced urologic events, including gross hematuria, flank pain, or cyst infections, and are already receiving treatment for hypertension and for patients older than 35 years of age with clinical data available, the PROPKD score is applicable. for patients with ADPKD under 35 years of age and/or for whom clinical data are lacking, the genetic score, although less accurate, can be used to predict renal survival.



Cornec- Le Gall E ,et al. The PROPKD Score: A New Algorithm to Predict Renal Survival in Autosomal Dominant Polycystic Kidney Disease. JASN March 2016, 27 (3) 942-951

The PROPKD Score: A New Algorithm to Predict Renal Survival in Autosomal Dominant Polycystic Kidney Disease



Conclusion This new prognostic score accurately predicts renal outcomes in patients with ADPKD and may enable the personalization of therapeutic management of ADPKD.

Emilie Cornec-Le Gall, Marie-Pierre Audrézet, Annick Rousseau, Maryvonne Hourmant, Eric Renaudineau, Christophe Charasse, Marie-Pascale Morin, Marie-Christine Moal, Jacques Dantal, Bassem Wehbe, Régine Perrichot, Thierry Frouget, Cécile Vigneau, Jérôme Potier, Philippe Jousset, Marie-Paule Guillodo, Pascale Siohan, Nazim Terki, Théophile Sawadogo, Didier Legrand, Victorio Menoyo-Calonge, Seddik Benarbia, Dominique Besnier, Hélène Longuet, Claude Férec and Yannick Le Meur,. The PROPKD Score: A New Algorithm to Predict Renal Survival in Autosomal Dominant Polycystic Kidney Disease. JASN March 2016, 27 (3) 942-951

Visual Abstract by Edgar V. Lerma

Clinical Trial End Points in ADPKD. Posted on June 6, 2019 by AJKD blog in Commentary // 0 Comments

- **PROPKD score:**

  low risk: 0-3 point : ESKD age 70.6

  intermediate risk: 4-6 point : ESKD age: 56.9

  high risk: 7-9 point: ESKD age: 49

Cornec- Le Gall E ,et al. The PROPKD Score: A New Algorithm to Predict Renal Survival in Autosomal Dominant Polycystic Kidney Disease. JASN March 2016, 27 (3) 942-951

Progression scoring(continued):

- In other cases ?
- The genetic information used for prognosis :
- PKD1 truncating, PKD1 non truncating, and PKD2 mutations being associated to most severe, intermediate, and least severe (average age at onset of ESKD of 56, 68, and 79 years, respectively) disease, respectively.

Fuad T , et al. Recent advances in the management of autosomal dominant polycystic kidney disease: clin j am soc Nephrol 13: 1765–1776, 2018

Conclusion:

- **Criteria to identify patients high risk for progression rate:**
- MYO classification : Class 1E,1D,1C
- Adult age at 55 or younger with eGFR < 65 cc/min/1.73m2
- Average kidney length by (US,MRI,CT)>16.5 cm in patient< 50y/o (predict CKD III)
- PROPKD score >6
- PKD1 truncating mutation

Arlene B chapman, et al. ADPKD Treatment. UpToDate. Last update jul12,2021


thank
you