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Chapter 1

Nomenclature, Diagnosis, Prognosis, and Prevalence

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ADPKD Is the Most Prevalent Monogenic Kidney Disease Associated With Kidney Failure

Prevalence estimates vary across population and genetic studies

Studies have estimated ADPKD prevalence in 2012 as 2-4 per 10,000 in the US and ~4 per 10,000 in the EU

One study conducted in the US in 2020 found that 2.6% of dialysis patients had cystic kidneys and were starting KRT

ADPKD, autosomal dominant polycystic kidney disease; EU, European Union; KRT, kidney replacement therapy; US, United States.

KDIGO. *Kidney Int*. 2025;107(2S):S1–S239.

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Mutations to the *PKD1* and *PKD2* Genes Are the Most Common Pathogenic Variants That Cause ADPKD

- ADPKD is an inherited disorder caused by single pathogenic variants in one ADPKD–causing gene
- While mutations in the *PKD1* and *PKD2* genes account for >90% of affected families, mutations in other minor genes can also lead to the development of ADPKD

Gene	Screened families, %	No. of families	Disease designation	Kidney phenotype	Extrarenal phenotype	Comments
The major ADPKD genes and nomenclature for unknown, not screened, and unresolved typical cases						
Unknown/not screened/ unresolved			ADPKD	Bilateral PKD, kidney enlargement, age-related CKD, may result in KF	Liver cysts including severe PLD, increased risk of ICA	Wide phenotypic rise in terms of TKV and KF risk and timing
PKD1	~48	>3250	Truncating pathogenic variant: ADKPD-*PKD1*	Bilateral PKD, early kidney enlargement, CKD stage G3, age ~40 y, KF in 50s	Liver cysts including severe PLD, increased risk of ICA	Some disease variability, including a more benign course, sometimes associated with mosaicism
	~19	>1750	Nontruncating pathogenic variant: ADPKD-*PKD1*	Bilateral PKD, kidney enlargement, age-related CKD, may result in KF	Liver cysts including severe PLD, increased risk of ICA	Phenotype ranges from as severe as *PKD1* truncating to mild PKD in old age, in part depending on the degree of residual protein function
PKD2	~15	>1000	ADPKD-*PKD2*	Bilateral PKD, milder and later kidney enlargement, CKD stage G3, age ~55 y, KF in 70s	Liver cysts including severe PLD, increased risk of ICA	Some disease variability, including a more severe or more benign course

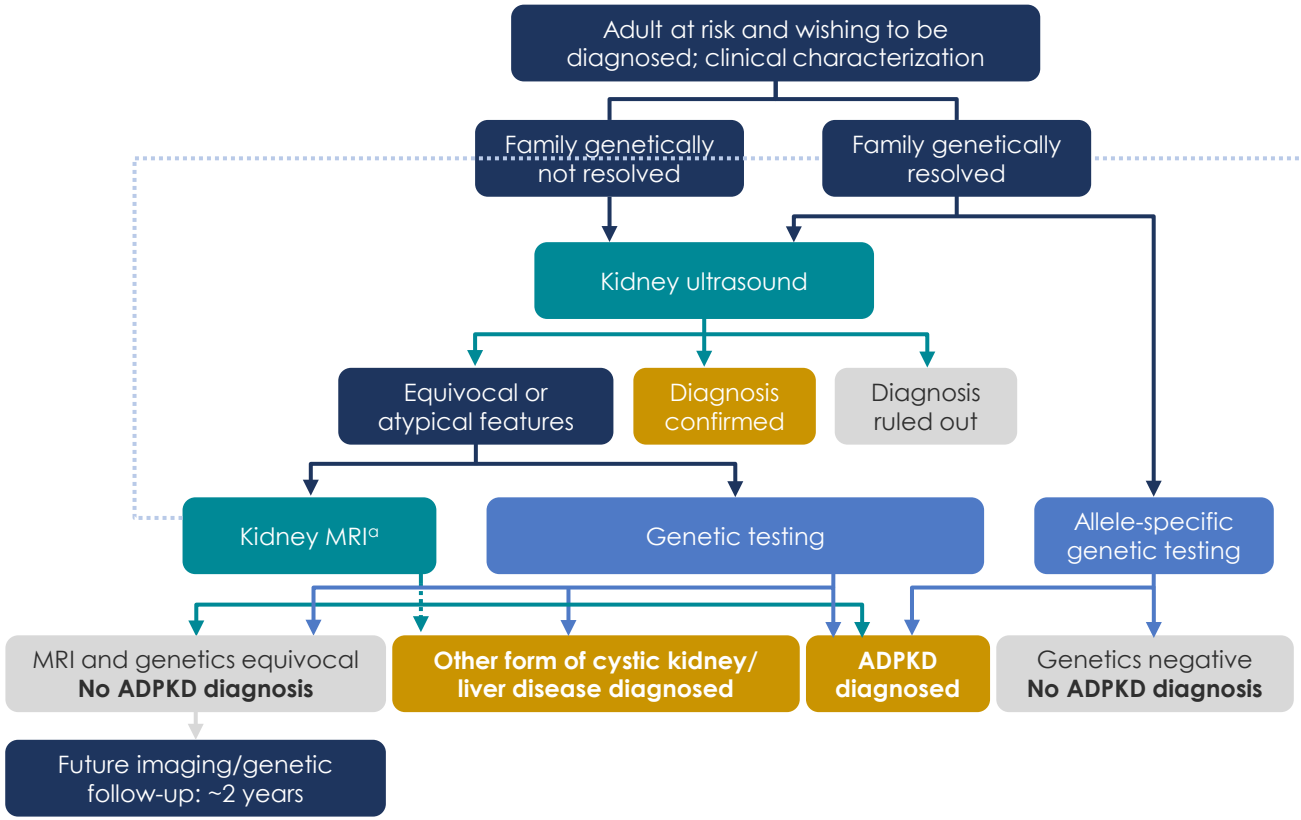
ADPKD, autosomal dominant polycystic kidney disease; CKD, chronic kidney disease; ICA, intracranial aneurysms; KF, kidney failure; PKD1, polycystic kidney disease 1; PKD2, polycystic kidney disease 2; PLD, polycystic liver disease; TKV, total kidney volume.

KDIGO. *Kidney Int*. 2025;107(2S):S1–S239.

A Diagnosis of ADPKD Can Be Determined Based on a Combination of Imaging and Clinical Features

- For screening adults at risk of ADPKD, we recommend first using abdominal imaging by ultrasound, in the context of the family history, kidney function, and comorbidities **(Recommendation 1.3.1 [1B])**
- Follow-up MRI, CT imaging, and/or genetic testing may clarify the diagnosis and further characterize the disease (Practice Point 1.3.4)

Diagnostic Algorithm for ADPKD



[a]Computed tomography, either with or without contrast, can also be used. Abdominal ultrasound is suggested as the first imaging analysis, with follow-up MRI analysis and/or genetic testing recommended in people with equivocal imaging or atypical extrarenal features.

ADPKD, autosomal dominant polycystic kidney disease; CT, computed tomography; MRI, magnetic resonance imaging.

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Genetic Testing Can Help Provide a Definite Diagnosis and Determine Prognosis of ADPKD

Genetic testing is not required to make an initial diagnosis; however, it is recommended if there is:

No family history of PKD

A presence of atypical imaging

Limited number of cysts

In patients with ADPKD, the genetic variant can impact disease severity

Understanding the genotype and associated variant can help elucidate the rate of disease progression and predict patient outcomes, which can be used to inform treatment decisions

Scan to learn more about genetic testing

ADPKD, autosomal dominant polycystic kidney disease; PKD, polycystic kidney disease.

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Identifying Patients With Rapidly Progressing Disease Can Help Optimize Treatment Strategies

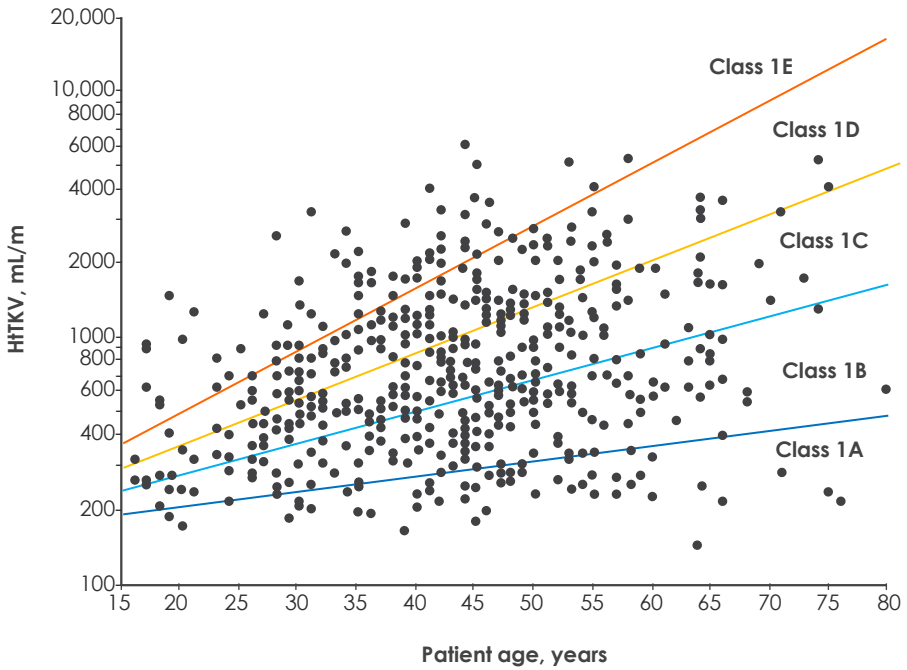
- Due to the heterogeneous nature of ADPKD, there is remarkable phenotypic variability in the progression of disease
- Various predictors can help identify patients at risk of rapid progression, specifically decrease in eGFR, increase in htTKV, and high PROPKD score

ADPKD, autosomal dominant polycystic kidney disease; AKI, acute kidney injury; BMI, body mass index; BP, blood pressure; eGFR, estimated glomerular filtration rate; GFR, glomerular filtration rate; htTKV, height-adjusted total kidney volume; PKD, polycystic kidney disease; PROPKD, Predicting Renal Outcomes in PKD; TKV, total kidney volume; UTI, urinary tract infection.

KDIGO. *Kidney Int*. 2025;107(2S):S1–S239.

Mayo Imaging Classification (MIC) Is Currently the Most Validated Model for Identifying Individuals at Risk of Rapid Progression

The MIC should be used to predict future decline in kidney function and time to kidney failure in patients with typical ADPKD (Recommendation 1.4.2.1 [1B])



Scan to access an online ADPKD simulator!

FPO

Scan to learn more about rapid progression!

FPO

ADPKD, autosomal dominant polycystic kidney disease; htTKV, height-adjusted total kidney volume.

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The Predicting Renal Outcome in Polycystic Kidney Disease (PROPKD) Score Can Help Identify Individuals at Risk of Rapid Progression

- The PROPKD score provides an additional clinical framework to identify risk for progression where either historical rate of decline in eGFR and/or MIC is equivocal or unavailable
- The PROPKD was developed for individuals 35 years and older

Risk Assessment Based on PROPKD Score

Variable	Points for PROPKD Score
Sex	
Female	0
Male	1
Hypertension before age 35 years	
No	0
Yes	2
≥1 urologic event before age 35 years	
No	0
Yes	2
Mutation	
PKD2	0
PKD1 nontruncating	2
PKD1 truncating	4

PROPKD score >6 points forecasts onset of kidney failure before age 60 years

eGFR, estimated glomerular filtration rate; MIC, Mayo Imaging Classification; PKD, polycystic kidney disease; PROPKD, Predicting Renal Outcomes in PKD.

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