

Key Takeaways for Clinicians from the KDIGO 2025 Clinical Practice Guideline for the Evaluation, Management, and Treatment of ADPKD: Nomenclature, diagnosis, prognosis, and prevalence



1

Definition of ADPKD

ADPKD is defined as a group of dominantly inherited disorders associated with kidney cysts and extrarenal manifestations, caused by a pathogenic variant in a gene associated with this disease. The disease is progressive, usually manifesting clinically in adulthood, often resulting in kidney failure.

2

Prevalence

ADPKD is the most prevalent monogenic kidney disease associated with kidney failure, accounting for 5%–10% of individuals with kidney failure worldwide. ADPKD affects all populations, with generally no common pathogenic variant enriching the disease in a geographic area or racial and/or ethnic group.

3

Diagnosing ADPKD

Abdominal ultrasound should initially be employed to screen adults at risk of ADPKD (usually ones with affected parent). Age-specific cyst number criteria have been employed to diagnose or exclude ADPKD. Kidney MRI or CT can also be useful, if available, and genetic testing can be of value (Figure 1).

4

Genetic testing

Although often not required to diagnose ADPKD in a person with a typical presentation, genetic testing can be particularly informative for people with an uncertain diagnosis based on kidney imaging and can aid in obtaining a definitive diagnosis in those with a negative or unknown family history. Due to the heterogeneity in the genetic causes of ADPKD, genetic testing should screen a panel of known PKD genes, not just *PKD1* and *PKD2*.

5

Prognostic markers

Methods to assess risk of rapid progression vary in different regions of the world. Where imaging is available to assess the height-adjusted total kidney volume (htTKV) for age in people with typical disease, the Mayo Image Classification (MIC) is currently the most straightforward way to identify patients with rapidly progressive disease. The slope of eGFR decline and the Predicting Renal Outcome in Polycystic Kidney Disease (PROPCKD) score, employing genetics, sex, and clinical factors, are useful if quantitative imaging is not available.

6

Nomenclature

Use a common nomenclature including the disease name followed by the gene name (e.g., ADPKD-*PKD1*) when a genetic diagnosis is available and pathogenicity is well supported. This nomenclature uses the familiar disease name but, by adding the gene name, provides more specific details of the disease in the affected individual. A similar nomenclature is recommended for autosomal dominant polycystic liver disease or ADPLD (e.g., ADPLD-*SEC63*).

7

Incidentally detected kidney and/or liver cysts: Obtaining a diagnosis

In a person with incidentally detected kidney and/or liver cysts (without a family history), detailed clinical and imaging assessment and genetic testing can help obtain a firm diagnosis (Figure 2).

8

Factors associated with disease progression

A range of clinical, genetic, lifestyle, environmental, and chance factors are thought to influence the rate of disease progression in ADPKD (Figure 3).

9

Considerations when testing for ADPKD

Testing for ADPKD in an at-risk adult either by imaging or genetics should employ a patient-centered approach. This should prioritize patients' values and preferences with an understanding of possible negative, as well as positive, outcomes from the testing.

Figure 1

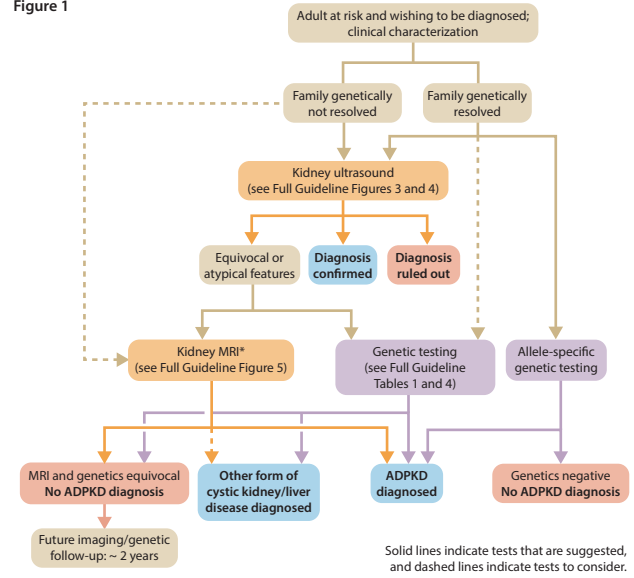


Figure 2

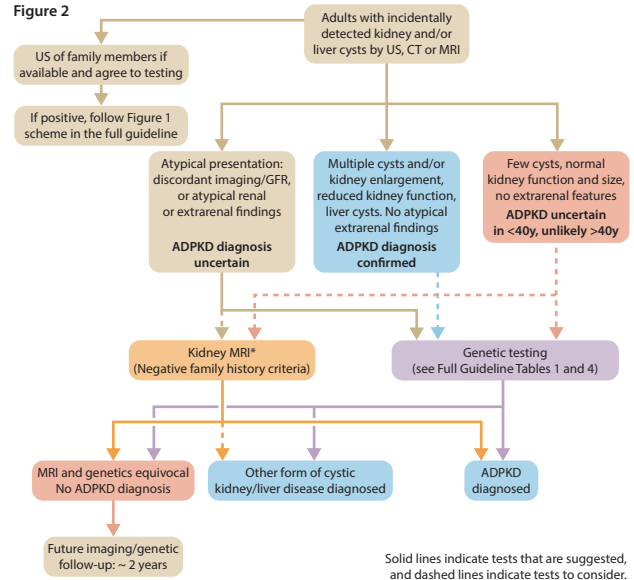


Figure 3

