



Prevalence and outcomes of polycystic kidney disease in African populations: A systematic review

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Abstract

BACKGROUND

Polycystic kidney disease (PKD) is the most common genetic cause of kidney disease. It is a progressive and irreversible condition that can lead to end-stage renal disease and many other visceral complications. Current comprehensive data on PKD patterns in Africa is lacking.

AIM

To describe the prevalence and outcomes of PKD in the African population.

METHODS

A literature search of PubMed, African journal online, and Google Scholar databases between 2000 and 2023 was performed. The Preferred Reporting Items for Systematic Reviews and Meta-Analyses were followed to design the study. Clinical presentations and outcomes of patients were extracted from the included studies.

RESULTS

Out of 106 articles, we included 13 studies from 7 African countries. Ten of them were retrospective descriptive studies concerning 943 PKD patients with a mean age of 47.9 years. The accurate prevalence and incidence of PKD were not known but it represented the third causal nephropathy among dialysis patients. In

majority of patients, the diagnosis of the disease was often delayed. Kidney function impairment, abdominal mass, and hypertension were the leading symptoms at presentation with a pooled prevalence of 72.1% (69.1–75.1), 65.8% (62.2–69.4), and 57.4% (54.2–60.6) respectively. Hematuria and infections were the most frequent complications. Genotyping was performed in few studies that revealed a high proportion of new mutations mainly in the *PKD1* gene.

CONCLUSION

The prevalence of PKD in African populations is not clearly defined. Clinical symptoms were almost present with most patients who had kidney function impairment and abdominal mass at the diagnostic. Larger studies including genetic testing are needed to determine the burden of PKD in African populations.

Key Words: Polycystic kidney disease; Africa; Genetic disorder; Systematic review

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Core Tip: Polycystic kidney disease is the most common genetic disorder affecting the kidney. The two main forms are autosomal dominant polycystic disease and autosomal recessive polycystic disease. It can lead to numerous complications with a natural progression leading to End stage kidney disease. Though the disease is well known and described in developed countries, its characteristics are still poorly understood in Africa. Indeed, as it appear in the present review, few studies regarding this disease were performed in the continent but reveal that advanced symptoms are already present in most of patients at the time of the diagnostic and the few studies with genetic testing revealed many new mutations.

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INTRODUCTION

Polycystic kidney disease (PKD) is the leading hereditary cause of chronic kidney disease. Autosomal dominant PKD (ADPKD) is its most frequent type with a reported prevalence to be between 1 in 400 and 1 in 1000 Live births in the world and is typically diagnosed later in life than autosomal recessive PKD (ARPKD)[1]. The prevalence, clinical and prognosis patterns of the disease are now well-documented in high-income countries. These advances have led to new therapeutic approaches that help slowing disease progression[2]. However, in low-resource settings such as in African countries, the lack of robust data on epidemiology, clinical presentation and prognosis of PKD are scarce. Also, a later diagnosis, fewer access to healthcare and new treatments are all factors that can explain a different epidemiology. We performed this systematic review to clarify the prevalence and outcomes of PKD in the African population.

MATERIALS AND METHODS

This systematic review was conducted in October 2023 to assess the prevalence and outcomes of PKD in the African populations. The Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) were followed to shape the study design[3].

A literature search of relevant articles published from January 2000 to September 2023 were performed on the online database PubMed, African journal online and google scholars. We also screened references of included articles to identify other potential studies. The keywords used for searching included: “Polycystic kidney disease” and “each of the 54 African countries name”. The search was realized by using different combination of these terms.

We included: (1) Observational studies with a description of the number of PKD cases; (2) Studies that offer a description of clinical manifestations at presentation; (3) Studies published in English or French; and (4) Case reports and case series with descriptions of genetic anomalies were also included.

Studies were excluded if they presented any one or more of the following criteria: case report, case series, abstracts, commenters or letter to the editor, systematic review and meta-analysis; language other than French or English, and study with age restriction of the participant.

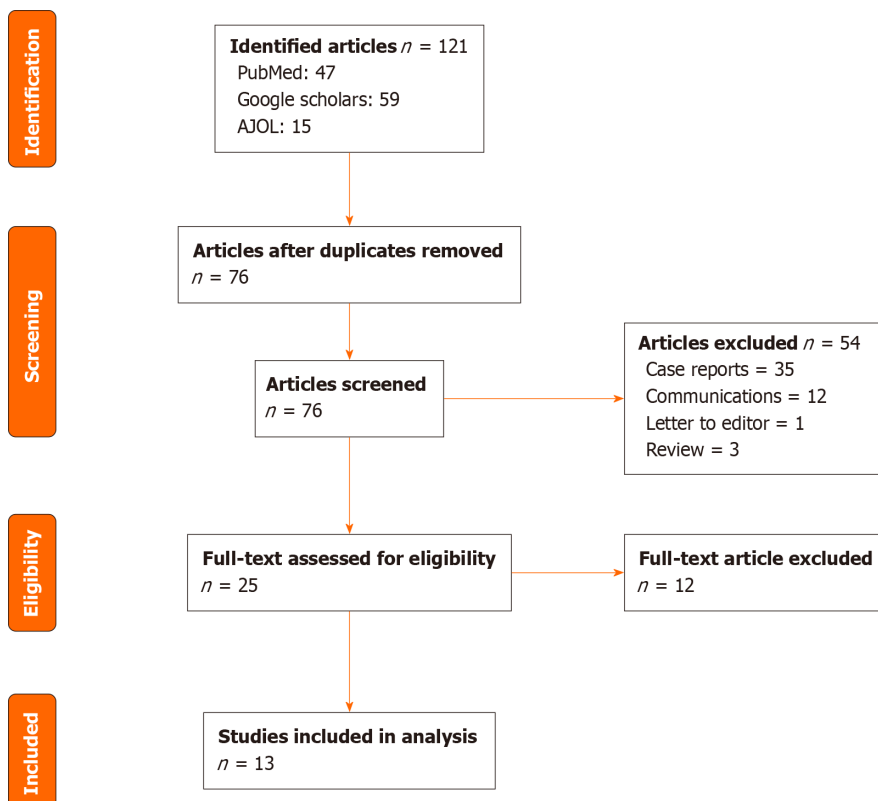
After eliminating duplicates, the titles and abstracts of all articles were reviewed and full texts of all articles designated for inclusion was obtained to ensure that they met the criteria for inclusion in this analysis.

For each study, we extracted the following data: study design, country, number of subjects included, demographic characteristics of patients, symptoms (hypertension, flank pain, hematuria, kidney function impairment), genetic mutation, complications, and prognosis.

Table 1 Characteristics of the different included studies

Ref.	Country	Patients	Design	Study period	Number of cases	Mean age (yr)	Gender M/F
Hajji <i>et al</i> [4], 2019	Tunisia	ADPKD	Observational descriptive	1969-2016	569	48.5	297/272
Arogundade <i>et al</i> [5], 2018	Nigeria	ADPKD	Observational descriptive	1996-2010	41	48.6 4.6	23/18
Chijioke <i>et al</i> [6], 2010	Nigeria	ADPKD	Observational descriptive	1994-2009	78	49.8 3.6	52/26
Ogiator <i>et al</i> [7], 2021	Nigeria	ADPKD	Observational descriptive	2013-2020	19	42.8 16.9	10/9
Mawufemo <i>et al</i> [8], 2018	Togo	ADPKD	Observational descriptive	2010-2017	27	51.6 16.4	10/17
Fary Ka <i>et al</i> [9], 2010	Senegal	ADPKD	Observational descriptive	1995-2005	55	47.0 5.0	31/24
Okyere <i>et al</i> [10], 2021	Ghana	ADPKD	Observational descriptive	2007-2018	82	43.8 15.7	43/39
Laleye <i>et al</i> [11], 2012	Benin	ADPKD	Observational descriptive	2000-2010	32	47.2	17/15
Abdelwahed <i>et al</i> [12], 2022	Tunisia	ADPKD	Observational descriptive	NA	19	47 18	10/9
Abdelwahed <i>et al</i> [13], 2018	Tunisia	ADPKD	Observational descriptive	NA	18	45	8/10
Seck <i>et al</i> [14], 2013	Senegal	ADPKD	Case report	NA	1	41	0/1
Sahnoun <i>et al</i> [16], 2015	Tunisia	ADPKD	Case report	NA	1	52	1/0
Nabhan <i>et al</i> [15], 2015	Egypt	ARPKD	Case report	NA	1	2.5	0/1
Total					943	47.9	502/441

ADPKD: Autosomic dominant polycystic kidney disease; ARPKD: Autosomic recessive polycystic kidney disease; NA: Not available.

**Figure 1 PRISMA flowchart of the review shape.**

RESULTS

Figure 1 present the PRISMA flow diagram detailing the review shape and studies selection process. We included 13 studies from different countries as detailed in Table 1. Ten of them were retrospective observational descriptive studies[4-13] and 3 were cases reports with genetic testing performed[14-16].

Table 2 Main clinical manifestations at presentation

Ref.	HTN (%)	Pain (%)	Hematuria (%)	Abdominal mass (%)	KFI (%)	ESKD (%)
Hajji <i>et al</i> [4], 2019	58.8	51.9	24.6	66.0	74.7	23.0
Arogundade <i>et al</i> [5], 2018	87.8	68.3	36.6	82.9	100.0	19.5
Chijioke <i>et al</i> [6], 2010	26.9	14.1	NA	NA	32.0	NA
Ogiator <i>et al</i> [7], 2021	42.1	68.4	31.6	NA	63.2	15.8
Mawufemo <i>et al</i> [8], 2018	77.8	63.0	22.2	63.0	63.0	25.9
Fary Ka <i>et al</i> [9], 2010	65.4	52.7	25.4	NA	NA	NA
Okyere <i>et al</i> [10], 2021	50.0	39.0	2.4	NA	81.7	15.9
Laleye <i>et al</i> [11], 2012	59.0	62.0	46.0	43.0	72.0	NA
Abdelwahed <i>et al</i> [12], 2022	57.9	NA	26.3	NA	89.5	NA
Abdelwahed <i>et al</i> [13], 2018	72.2	NA	NA	NA	NA	NA
Pooled prevalence [95%IC]	57.4 [54.2–60.6]	49.1 [45.8–52.3]	24.0 [21.1–26.9]	65.8 [62.2–69.4]	72.1 [69.1–75.1]	21.9 [18.9–24.9]

NA: Not available; HTN: Hypertension; KFI: Kidney function impairment; ESKD: End stage kidney disease.

A total of 943 patients with PKD were collected. The mean age were 47.9 years with a sex-ratio M/F of 1.14.

Clinical symptoms were described in all the descriptive studies. Overall, kidney function impairment, abdominal mass and hypertension were the most frequent finding at presentation, present in 72.1%, 65.8% and 57.4% of patients respectively (Table 2).

Genetics testing were performed in 5 study with a cumulated total of 40 patients[12–16]. All these patients had genetic disorders with 13 novels mutation/single nucleotide polymorphism detected. The most frequently reported new mutations were c.496 C>T, p.L166 among exon4; c.696 T>G, p.C232W among exon5; c.7290_7291delinsCTGCA among exon18 and c.12276 A>G, p.A4092 among exon45 in the *PKD1* gene (Table 3). The mutations concerned in 92.5% of cases the *PKD1* gene and in 7.5% the *PKD2* gene. In sub-Saharan Africa, seven new mutations were reported from Benin and one from a Senegal[11,14].

One case of ARPKD were reported in Egypt with a mutation (c.3367G>A, p.G1123S) in *PKHD1*[15].

DISCUSSION

PKD is a major public health problem that concerns all continents and ethnic groups. It is an incurable condition with a natural evolution leading to end-stage renal disease and can cause many other visceral complications. ADPKD and ARPKD are its two main types. ADPKD is commonly described in adults, whereas ARPKD is less frequent and usually presents during early childhood. ADPKD is the most frequent genetic cause of renal failure in adults, accounting for 6%–10% of end-stage renal disease cases. Its reported prevalence is similar around the globe. In the United States the reported diagnostic prevalence of ADPKD was 4.3 per 10000[17]. A large review study including 19 European countries revealed a prevalence of 3.96 per 10000[18]. However, as it appears in this review, the prevalence of PKD in Africa remains difficult to establish. Indeed, only one study from the Seychelles has reported a nationwide prevalence of 5.7 per 10000[19]. A broad range of mutations in *PKD* genes can lead to ADPKD. These disorders are widely distributed and can occur across the entire sequence of these genes, named *PKD1* and *PKD2*. The *PKD1* gene region is larger and counts 46 exons and its mutations are responsible for around 85% of cases[20]. The *PKD2* region is shorter and comprise only 15 exons with mutations causing 15% of ADPKD cases[20]. More than 1500 mutations of these two genes are indexed in ADPKD mutation databases[21]. Genotyping is usually necessary in persons with suspected PKD who do not meet the echographic criteria and/or compatible familial history. In the United Kingdom, new *PKD1* mutations represented 5% of ADPKD patients[22]. Results of a few genetic tests performed in African patients found that 90% of mutations were located in the *PKD1* gene and 48.1% of them were new mutations not previously described in non-African populations. Such findings expose the need for broader genetic testing for a better PKD description in the continent. Despite similar clinical manifestations, mutations in the *PKD1* gene are associated with an earlier onset of symptoms and ESKD compared to *PKD2* mutation[23]. In the literature, the reported ages at ADPKD diagnosis were 42 and 56 years, respectively for *PKD1* and *PKD2* patients[23]. In the present review, the mean age of patients at diagnosis was 47.9 years. In India, this age was 45.8 years[24].

Furthermore, in the majority of patients, clinical symptoms are already present at diagnosis. Hypertension, abdominal mass, flank pain, hematuria, urolithiasis, infection, and kidney function impairment were the main symptoms reported in African patients with ADPKD. The pooled proportion of ESKD was 21.9% and comparable to data from France and Canada where 22% and 25% respectively presented with ESKD at the time of diagnosis[25,26].

Table 3 Genetic analysis finding

Gene	Exon/Intron	Nucleotide change	Aminoacid change	Number of patients	Ref.
PKD 1	4	c.496 C>T	p.L166	2	[12]
	5	c.696 T>G	p.C232W	2	[12,13]
	5	c.688 T>G	p.C230G	1	[12]
	5	c.690 C>G	p.C230W	1	[12]
	7	c.1522 T>C	p.C508R	1	[12]
	8	c.1702 G>A	p.A568T	1	[12]
	9	c.1745_1761dup	NA	1	[11]
	15	c.4264 G>A	p.A1422T	1	[12]
	15	c.5577 T>C	p.A1859	1	[12]
	15	c.4495 C>T	p.L1499	2	[12]
	15	NA	p.Q1651X	1	[11]
	15	NA	p.W1666X	1	[11]
	15	NA	c.6575_6581del	1	[11]
	18	c.7290_7291delinsCTGCA	NA	2	[11,14]
	IVS22	c.8161-1 G>A	Likely silent	4	[12]
	IVS42	c.11712+28 C>T	Likely silent	1	[12]
	23	c.8679 C>G	p.S893	1	[12]
	23	c.8715 C>T	p.V2905	1	[12]
	23	c.8748 T>C	p.P2916	2	[12]
	23	c.8522 A>G	p.N2841S	1	[12]
	23	NA	p.Q2824X	1	[11]
	26	c.9397+1_9397+8del	NA	1	[11]
	28	c.9669 G>A	p.T3223	2	[12]
	30	c.3367G>A	p.G1123S	1	[15]
	31	c.10165 G>C	p.E3389Gln	1	[12]
	44	c.12133 A>G	p.I4045V	1	[12]
	45	c.12276 A>G	p.A4092	2	[12,16]
PKD 2	1	c.568 G>A	p.A190T	2	[12]
	1	c.83 G>C	p.A28P	2	[12]
	IVS1	c.596-16 C>T	Likely silent	1	[12]
PKHD1		c.3367G>A	p.G1123S	1	[15]

In bold: New mutation found. NA: Not available.

In the United States, ADPKD is the fourth leading cause of ESKD requiring dialysis and transplantation[27].

Less common than ADPKD, ARPKD is a childhood-onset disease with symptoms that can appear in perinatal. It is linked to the mutations *PKHD1* gene with an estimated prevalence of 1 in 20000 live births in Caucasians[28]. In Africa, its prevalence is still not known, one case was reported in an Egyptian child. A mean age at diagnosis of 4 years was reported with around 60% of patients with ESKD before adulthood[29].

CONCLUSION

PKD represents the most frequent genetic disorder. ADPKD is by far more frequent than ARPKD. In Africa, little data on the prevalence, clinical presentation, and evolution of this disease are available, and genetic testing is even more lacking.

Clinical symptoms were almost present with most patients who had kidney function impairment and abdominal mass at the diagnosis. As shown in this review, many new mutations were found in the *PKD1* gene. More large-scale studies are needed to describe the patterns of these diseases.

ARTICLE HIGHLIGHTS

Research background

Polycystic kidney disease is known as the most common genetic cause of chronic kidney disease. Its natural evolution lead to end-stage kidney disease. However, unlike developed countries, clinical and prognosis outcomes data of the disease are lacking in African population.

Research motivation

Mapping the data of polycystosis in African population and emphasize the gap between data from international literature and those available in our specific population and outline points for further studies.

Research objectives

Describe the prevalence, clinical, and genetic aspects of polycystic kidney disease in an African population.

Research methods

A literature review and meta-analysis of available data were performed from January 2000 to September 2023 to identify reported data of prevalence, clinical manifestation, and genetics anomalies of patients with polycystic kidney disease in the continent.

Research results

A total of 943 patients with polycystic kidney disease were reported in the period of research but the real prevalence of the disease is not known in the continent. Most patients present with symptoms at diagnosis mainly kidney function impairment and abdominal mass. Nevertheless, the mean age at diagnosis is similar to the literature data. Genetic testing was not frequent, however, they showed a high proportion of new mutations.

Research conclusions

Most African patients with polycystic kidney disease present with severe symptoms and complications at diagnosis. A high proportion of new mutations were reported in this population particularly in the *PKD1* gene.

Research perspectives

Further researches are needed to better assess the real prevalence of PKD and the spectrum of mutations in the continent.

FOOTNOTES

Author contributions: Seck SM and Ndongo M designed the study and performed the research; Seck SM and Ndongo M analyzed the data and wrote the manuscript; Ndongo M, Nehemie LM, Coundoul B, Diouara AAM and Seck SM edited and reviewed the manuscript; the manuscript has been read and approved by all authors.

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