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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

This review aimed to describe the prevalence and outcomes of PKD in the African population.

METHODS

A literature search of PubMed, AJOL, and Google Scholar databases between 2000 and 2023 was performed. The Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) 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.

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 1,000 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 (AJOL) 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 Polycystic kidney disease (PKD) cases; (2) studies that offer a description of clinical manifestations at presentation; and (3) studies published in English or French. (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.

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].

A total of 943 patients with polycystic kidney disease (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 (KFI), abdominal mass and hypertension (HTA) 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 (SNP) detected. The most frequently reported mew 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 autosomic recessive PKD (ARPKD) were reported in Egypt with a mutation (c.3367G>A, p.G1123S) in PKHD1 [15].

DISCUSSION

Polycystic kidney disease (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. Autosomal dominant polycystic kidney disease (ADPKD) and autosomal recessive polycystic kidney disease (ARPKD) are its two main types. ADPKD is commonly

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France and Canada where 22% and 25% respectively presented with ESKD at the time of diagnosis [25,26].

In the USA, 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 20,000 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

Polycystic kidney disease 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.

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