Vol. 5 No. 2 (2026)

Pakistan Journal of Medical & Cardiological Review

https://pakjmcr.com/index.php/1/about

Online ISSN 3007-2387

Print ISSN 3007-2379

Khan et al., 2026

FREQUENCY AND ULTRASONOGRAPHIC FINDINGS OF POLYCYSTIC KIDNEY DISEASE AMONG PATIENTS UNDERGOING ABDOMINAL ULTRASONOGRAPHY: A DESCRIPTIVE CROSS-SECTIONAL STUDY

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Online ISSN 3007-2387

Print ISSN 3007-2379

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Received on 24 May, 2026
Accepted on 29 June, 2026
Published on 30 June, 2026

Abstract

Background: Polycystic kidney disease (PKD) is a common inherited renal disorder causing progressive cystic enlargement and renal dysfunction. Ultrasonography is a practical, non-invasive tool for its early detection, monitoring, and differentiation from other cystic kidney diseases.To determine the frequency of PKD and evaluate its ultrasonographic findings among patients undergoing abdominal ultrasound examinations. **Methods:** This descriptive cross-sectional study was conducted over four months at Lady Reading Hospital, Peshawar. Ninety-three patients undergoing abdominal ultrasonography were enrolled through convenient sampling. Data were collected using a structured questionnaire together with ultrasonographic findings. Patients with indeterminate PKD, kidney transplant, or prior kidney surgery were excluded. Data were analyzed using SPSS version 22, and results were expressed as frequencies and percentages. **Results:** Of 93 participants, 72 (77.4%) were diagnosed with PKD on ultrasonography, while 21 (22.6%) had normal findings. Bilateral renal involvement was the most common pattern (24.7%), followed by right-sided (40.9%) and left-sided (14.0%) involvement. A positive family history of kidney disease was reported more frequently among hypertensive participants than non-hypertensive participants. **Conclusion:** PKD was identified in a substantial proportion of patients undergoing abdominal ultrasonography, with bilateral renal involvement predominating. Family history and hypertension were important associated factors. Ultrasonography remains an effective, accessible tool for the early detection of PKD.

Keywords: Polycystic Kidney Disease; Autosomal Dominant Polycystic Kidney Disease; Ultrasonography; Abdominal Ultrasound; Renal Cysts; Renal Imaging

INTRODUCTION

Polycystic kidney disease (PKD) is one of the most common inherited renal disorders, characterized by the progressive development and enlargement of multiple fluid-filled cysts within the kidneys.[1] PKD distorts normal renal architecture and leads to a gradual decline in kidney function, culminating in end-stage renal disease (ESRD) in a significant proportion of affected individuals. Among the various forms of PKD, Autosomal

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Online ISSN 3007-2387
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Dominant Polycystic Kidney Disease (ADPKD) is the most prevalent hereditary kidney disorder, accounting for approximately 5–10% of patients requiring renal replacement therapy worldwide. [2], [3]

ADPKD results primarily from mutations in the PKD1 and PKD2 genes, which encode polycystin-1 and polycystin-2—proteins essential for normal tubular epithelial architecture. These mutations cause abnormal cellular proliferation and fluid secretion, leading to progressive bilateral cyst formation and eventual impairment of renal function. [4], [5] Although the genetic basis of ADPKD is well characterized, its clinical expression varies considerably even among members of the same family. The estimated prevalence of ADPKD ranges from 1 in 400 to 1 in 1,000 live births, making it one of the most common monogenic disorders globally; many affected individuals remain undiagnosed in early, asymptomatic stages. [6]

Clinical presentation is diverse and age-dependent. Patients may remain asymptomatic for decades before developing hypertension, flank pain, hematuria, urinary tract infections, nephrolithiasis, or progressive renal insufficiency, with hypertension often the earliest manifestation. Extra-renal features—hepatic cysts, intracranial aneurysms, cardiac valvular abnormalities, and colonic diverticulosis—further contribute to disease morbidity. [7]

Among available imaging modalities—ultrasonography, computed tomography, and magnetic resonance imaging—ultrasonography remains the first-line technique for evaluating cystic kidney disease because it is non-invasive, free of ionizing radiation, widely available, and diagnostically accurate. [8] Validated ultrasound criteria allow reliable identification of renal cysts, assessment of kidney size, and disease monitoring over time, and the modality is highly sensitive for cysts larger than 1 cm. Because abdominal ultrasound is frequently performed for unrelated indications, renal cysts and polycystic kidneys are often detected incidentally, offering valuable opportunities for early diagnosis.[9] Ultrasonography is equally important for distinguishing PKD from other cystic renal disorders—such as simple renal cysts, acquired cystic kidney disease, medullary sponge kidney, and tuberous sclerosis-associated cystic disease—through comprehensive assessment of cyst number, size, distribution, bilaterality, and associated extra-renal findings.[7]

The genetic basis of ADPKD has been extensively investigated; approximately 85% of cases arise from PKD1 mutations, with the remainder linked to PKD2.[10] These mutations disrupt intracellular signaling, producing abnormal proliferation and progressive cyst growth.[11] Epidemiological data indicate that ADPKD may affect more than 12 million people worldwide and imposes a considerable healthcare burden through hospitalization, dialysis, and associated costs.[12] Approximately half of affected individuals progress to ESRD by the sixth decade of life, with severity influenced by genetic factors, hypertension, total kidney volume, and age at diagnosis.[13,16] Hypertension itself is frequent and often precedes measurable renal decline, occurring in roughly 60% of young adults with ADPKD and contributing independently to disease progression.[14]

Unified ultrasonographic diagnostic criteria for individuals at risk of ADPKD have demonstrated high sensitivity and specificity, particularly among adults with a positive family history, and are now widely used in clinical practice.[19] Advances in ultrasound technology have further improved assessment of cyst number, size, and renal dimensions, supporting longitudinal disease monitoring without radiation exposure.[20,21] Total kidney volume, in particular, correlates strongly with the rate of renal function decline, and although magnetic resonance imaging offers the most precise volumetric assessment, ultrasonography remains a practical and cost-effective alternative in routine practice.[17,22] Early identification through imaging-based screening enables timely blood pressure control, patient counseling, and consideration of disease-modifying therapies such as tolvaptan, which has been shown to slow kidney volume progression.[2,9]

Despite the recognized clinical and public health importance of PKD, most epidemiological data originate from North America and Europe, where structured screening programs are well established; comparable local data from developing countries, including Pakistan, remain scarce.[3] Variations in genetic background, healthcare access, environmental factors, and referral patterns may influence disease prevalence and presentation, and regional studies are therefore needed to characterize the burden of PKD within specific populations and clinical settings.[26] The increasing use of abdominal ultrasonography in routine practice offers a practical opportunity to assess

PKD frequency among patients undergoing imaging for diverse indications, potentially improving referral pathways, patient counseling, and screening strategies.

Given this gap in local evidence, the present study was designed to determine the frequency of PKD and to characterize its ultrasonographic features among patients undergoing abdominal ultrasonography at a tertiary care hospital in Peshawar, Pakistan, thereby contributing population-specific epidemiological and diagnostic data to the existing literature.

Objectives

The objectives of this study were: (i) to determine the frequency of polycystic kidney disease among patients undergoing abdominal ultrasonography, and (ii) to evaluate the ultrasonographic findings of polycystic kidney disease in affected patients.

MATERIAL AND METHODS

Study Design, Setting, and Duration

This descriptive cross-sectional study was conducted over a period of four months in the Radiology Department of Lady Reading Hospital, Peshawar, Pakistan.

Study Population and Sampling

The study population comprised patients undergoing abdominal ultrasonography at Lady Reading Hospital, Peshawar. A total of 93 patients were enrolled using a non-probability convenient sampling technique. The sample size was calculated using the Cochrane formula, $n = Z^2p(1-p)/E^2$, with a 95% confidence interval ($Z = 1.96$), an expected prevalence of 6.4% based on previously reported data,[1] and a margin of error of 5% ($E = 0.05$), yielding a minimum required sample size of 93.

Selection Criteria

Inclusion criteria: Patients of either gender undergoing renal or abdominal ultrasonography during the study period who were willing to participate and provide informed consent.

Exclusion criteria: Patients with indeterminate findings of polycystic kidney disease, those with a prior kidney transplant, and those with a history of previous kidney surgery were excluded.

Data Collection Procedure

Following approval from the Institutional Ethical Review Committee, a structured, pre-tested questionnaire was administered to eligible participants by the investigator. The questionnaire captured demographic characteristics, clinical history, and ultrasonographic findings. Items were phrased in simple, comprehensible language. Each participant was briefed on the purpose, benefits, and potential risks of the study prior to data collection, and participation was entirely voluntary.

Ethical Considerations

Ethical approval was obtained from the Institutional Ethical Review Committee prior to the commencement of the study. Written informed consent was obtained from all participants after explaining the study objectives; participation was voluntary, and withdrawal at any stage did not affect routine clinical care. Confidentiality was maintained by coding the data and omitting personal identifiers. The study did not interfere with participants' standard diagnostic or clinical management.

Data Analysis

Data were analyzed using IBM SPSS Statistics, version 22 (IBM Corp., Armonk, NY, USA). Descriptive statistics were used to summarize demographic and clinical variables; categorical variables were expressed as frequencies and percentages and presented using tables and bar/pie charts. Cross-tabulation was used to explore the relationship between family history of kidney disease and ultrasonographic findings of the affected kidney.

RESULTS

A total of 93 patients undergoing abdominal ultrasonography were included in the final analysis. The demographic and clinical characteristics of the study participants are summarized in Tables I and II.

Table I: *Demographic Characteristics of Study Participants (N = 93)*

Variable	Category	Frequency (n)	Percentage (%)
Age (years)	12–18	43	46.2
	19–45	45	48.4
	>45	5	5.4

Variable	Category	Frequency (n)	Percentage (%)
Gender	Male	44	47.3
	Female	49	52.7
Residence	Urban	49	52.7
	Rural	44	47.3

The majority of participants belonged to the 19–45-year age group (48.4%), followed by the 12–18-year group (46.2%); only 5.4% were older than 45 years. Females constituted a slightly higher proportion of the sample (52.7%) than males (47.3%), and urban residents (52.7%) slightly outnumbered rural residents (47.3%).

Figure 1: Age Distribution of Study Participants

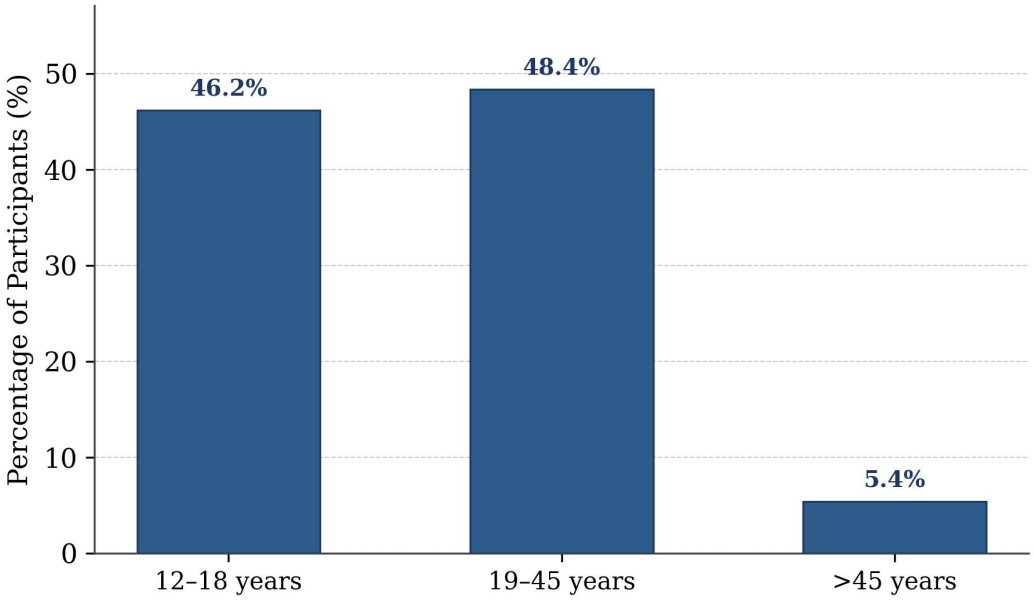

Figure 1: Age distribution of study participants (N = 93).

Figure 2: Gender and Residential Distribution of Participants

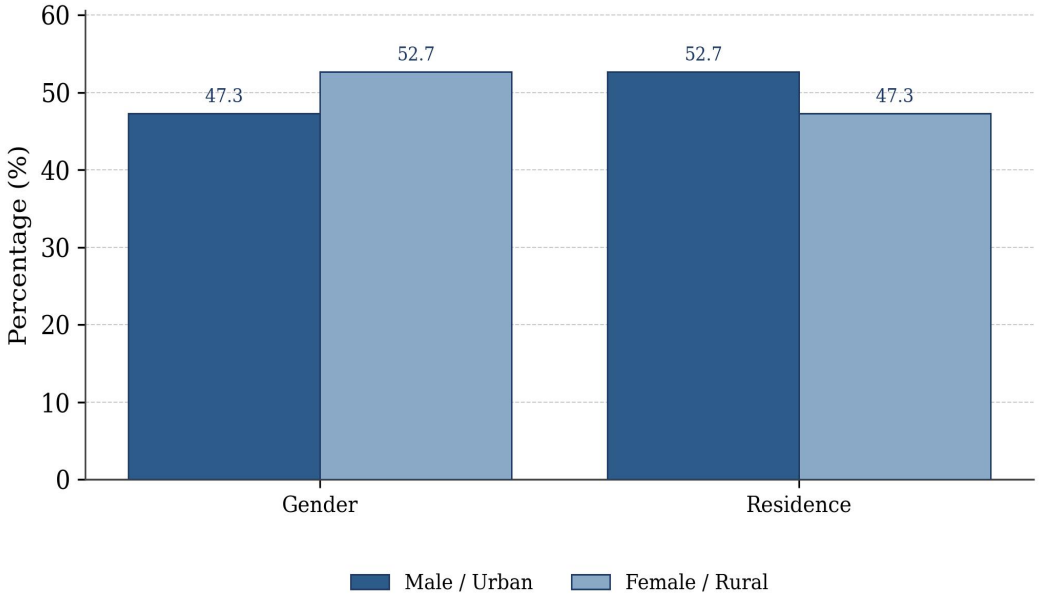

Figure 2: Gender and residential distribution of study participants (N = 93).

Table II: *Ultrasonographic Findings Among Study Participants (N = 93)*

Ultrasonographic Finding	Category	Frequency (n)	Percentage (%)
Overall impression	PKD positive	72	77.4
	Normal	21	22.6
Kidney involvement	Right kidney only	38	40.9
(among all participants)	Both kidneys (bilateral)	23	24.7
	Left kidney only	13	14.0
	Normal	19	20.4

Of the 93 participants, 72 (77.4%) were diagnosed with PKD on ultrasonography, while 21 (22.6%) had normal findings (Table II). Among the patterns of renal involvement, right kidney involvement was the most frequent (40.9%), followed by bilateral involvement of both kidneys (24.7%); isolated left kidney involvement was least common (14.0%), and 20.4% of participants had normal kidneys on ultrasound.

Figure 3: Overall Ultrasonographic Impression

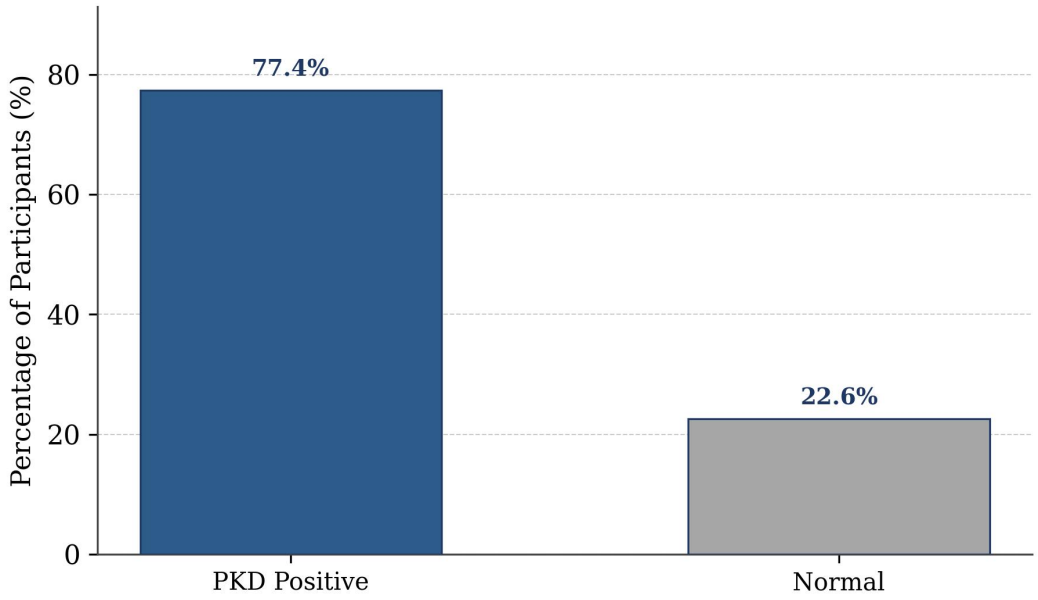

Figure 3: Overall ultrasonographic impression among study participants (N = 93).

Figure 4: Pattern of Renal Involvement on Ultrasonography

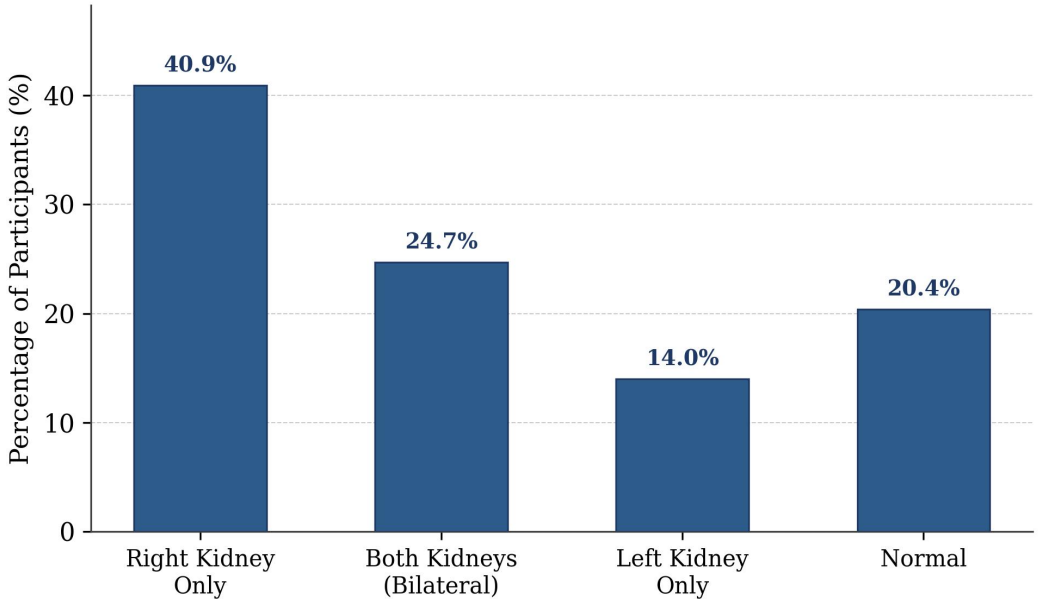

Figure 4: Pattern of renal involvement on ultrasonography (N = 93).

Table III: *Clinical Symptoms and Comorbid Conditions Among Study Participants (N = 93)*

Clinical Variable	Present, n (%)	Absent, n (%)
Abdominal or flank pain	40 (43.0)	53 (57.0)
Hematuria (blood in urine)	49 (52.7)	44 (47.3)
Recurrent urinary tract infection	40 (43.0)	53 (57.0)
Prior diagnosis of chronic kidney disease	51 (54.8)	42 (45.2)
High cholesterol	47 (50.5)	46 (49.5)
History of hypertension	34 (36.6)	59 (63.4)

Abdominal or flank pain was reported by 43.0% of participants and hematuria by 52.7%. Recurrent urinary tract infection was present in 43.0%, and 54.8% of participants had a prior diagnosis of chronic kidney disease. High cholesterol was reported by 50.5% of participants, and a history of hypertension was present in 36.6% (Table III).

Figure 5: Clinical Symptoms and Comorbid Conditions

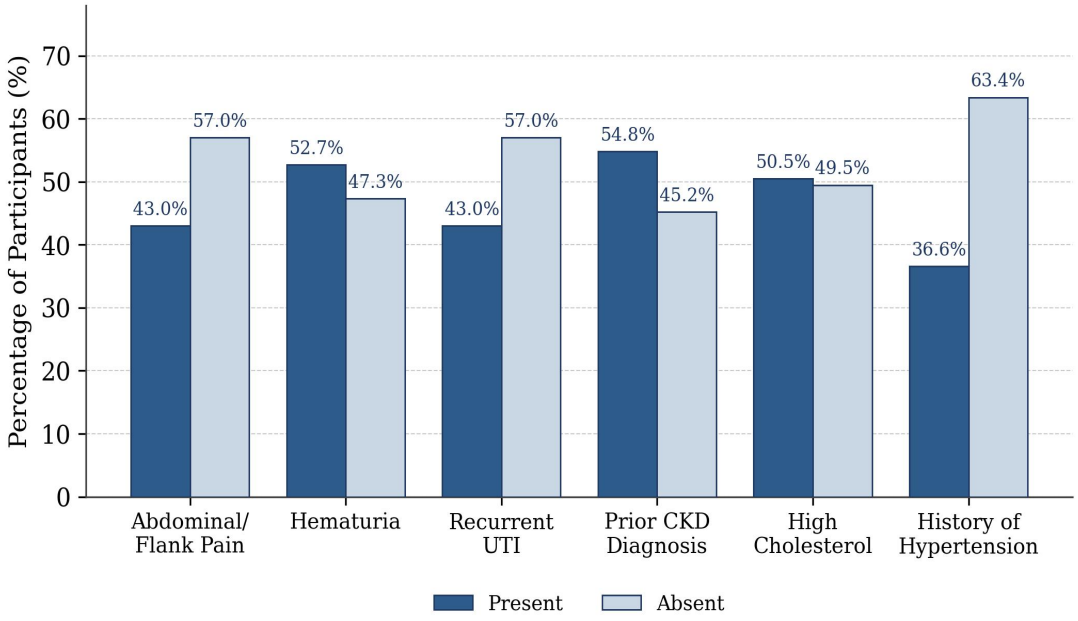

Figure 5: Clinical symptoms and comorbid conditions among study participants (N = 93).

Table IV: *Family History of Kidney Disease Among Study Participants (N = 93)*

Family History (First-Degree Relative Affected)	Frequency (n)	Percentage (%)
Yes	34	36.6
No	34	36.6
Uncertain / Don't know	25	26.9

A positive family history of kidney disease in a first-degree relative was reported by 36.6% of participants, an equal proportion (36.6%) reported no family history, and 26.9% were uncertain (Table IV).

Figure 6: Family History of Kidney Disease Among Participants

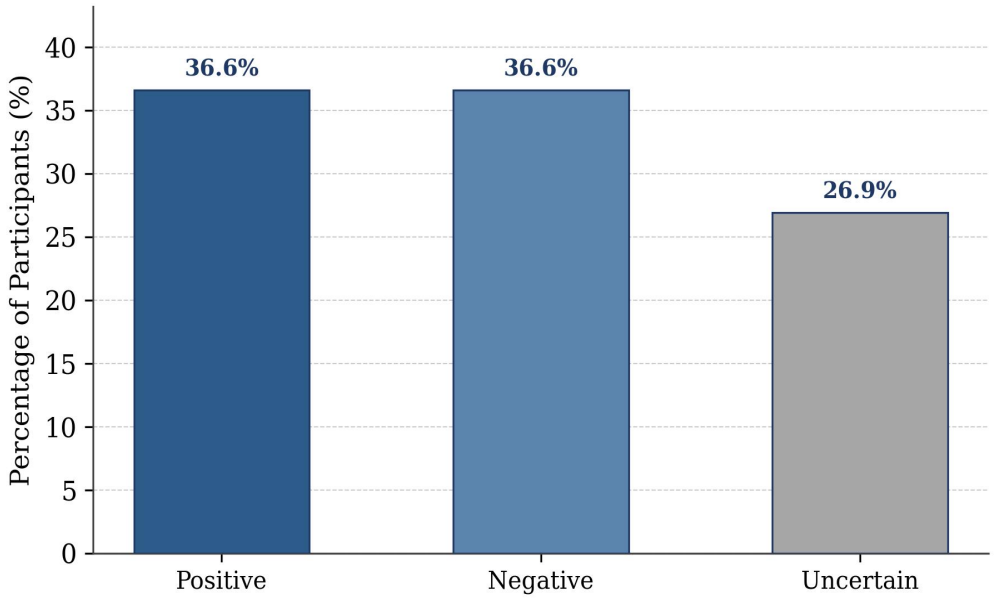

Figure 6: Family history of kidney disease among study participants (N = 93).

Table V: *Relationship Between Family History of Kidney Disease and Ultrasonographic Pattern of Renal Involvement*

Ultrasonographic Finding	Positive Family History, n (%)	No Family History, n (%)	Uncertain, n (%)	Total
Left kidney involvement (n=13)	9 (69.2)	4 (30.8)	0 (0.0)	13

Ultrasonographic Finding	Positive Family History, n (%)	No Family History, n (%)	Uncertain, n (%)	Total
Right kidney involvement (n=38)	8 (21.1)	18 (47.4)	12 (31.6)	38
Bilateral involvement (n=23)	9 (39.1)	8 (34.8)	6 (26.1)	23
Normal (n=19)	8 (42.1)	4 (21.1)	7 (36.8)	19

Table V presents the relationship between family history of kidney disease and the pattern of renal involvement identified on ultrasound. Among patients with isolated left kidney involvement, a positive family history was most common (69.2%). Among those with right kidney involvement, the largest proportion reported no family history (47.4%), while 31.6% were uncertain. Among patients with bilateral involvement, family history was fairly evenly distributed across the three categories, and a positive family history was also reported by 42.1% of participants with normal ultrasound findings.

Figure 7: Family History by Pattern of Renal Involvement

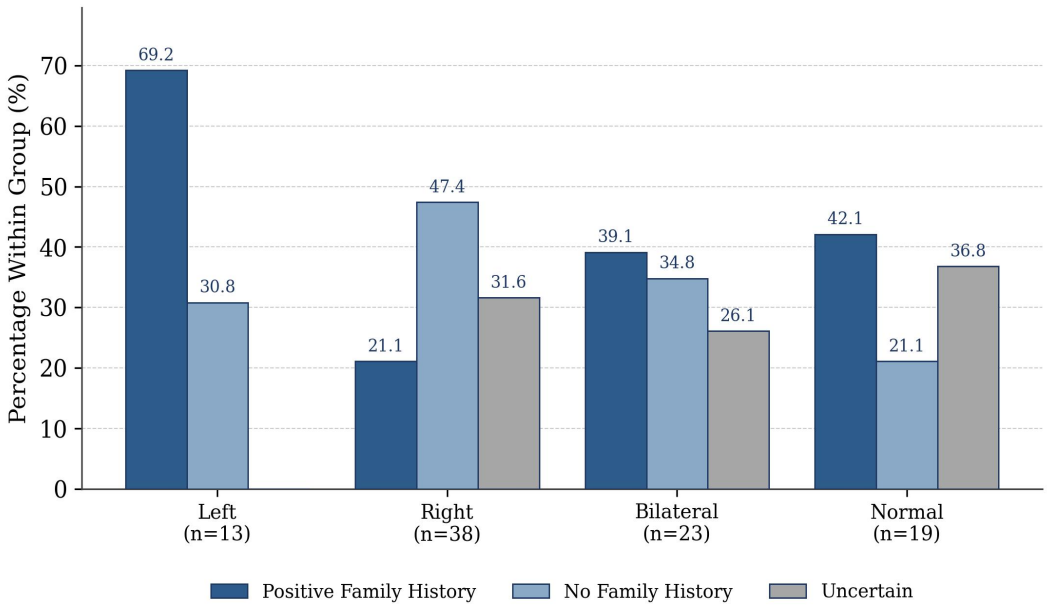

Figure 7: Family history of kidney disease by pattern of renal involvement on ultrasonography.

Table VI: *Family History of Kidney Disease by Hypertension Status (N = 93)*

Hypertension Status	Positive Family History, n (%)	Total, n
Hypertensive (n=34)	22 (64.7)	34
Non-hypertensive (n=59)	12 (35.3)*	59

A positive family history of kidney disease was reported by 64.7% of hypertensive participants, compared with 35.3% of non-hypertensive participants, indicating a higher burden of familial kidney disease among those with hypertension (Table VI).

**Percentage reflects the proportion of all participants with a positive family history (n = 34) who were non-hypertensive; figures are derived from the original cross-tabulated dataset.*

Figure 8: Positive Family History of Kidney Disease by Hypertension Status

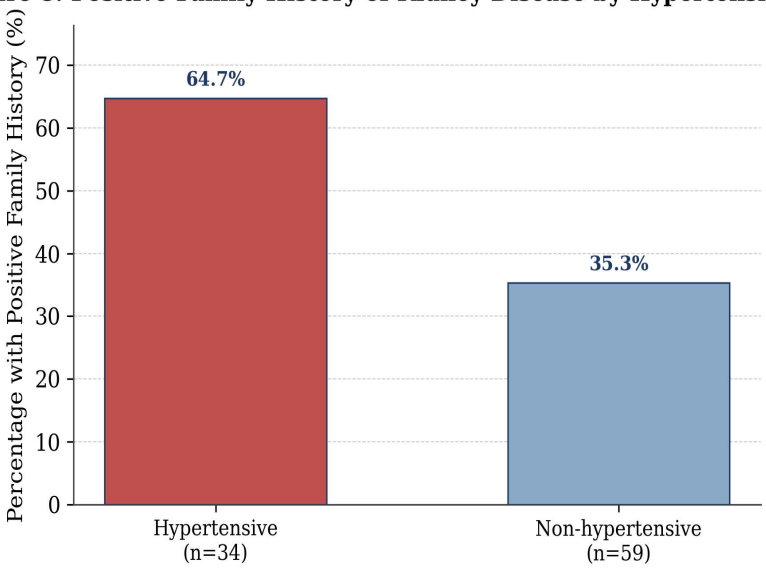

Figure 8: Positive family history of kidney disease by hypertension status.

DISCUSSION

The present study evaluated the frequency and ultrasonographic findings of polycystic kidney disease (PKD) among patients undergoing abdominal ultrasonography at a tertiary care hospital in Peshawar. PKD was identified in 77.4% of participants, while 22.6% had normal findings. This relatively high frequency likely reflects the hospital-based, referral-driven nature of the sample, since most patients were referred specifically for suspected renal abnormalities rather than drawn from the general

population. Ultrasonography has consistently been highlighted as the most widely used imaging modality for the diagnosis and screening of PKD because it is non-invasive, readily available, cost-effective, and highly sensitive for renal cysts.[7] Ali et al. similarly emphasized the central role of sonography in the evaluation and differential diagnosis of renal cystic disease.[1]

Bilateral renal involvement was the most common ultrasonographic pattern overall, consistent with the natural course of Autosomal Dominant Polycystic Kidney Disease (ADPKD), in which cyst formation typically progresses to affect both kidneys over time. This finding aligns with previous reports describing bilateral renal cysts and kidney enlargement as characteristic and diagnostically important features of ADPKD.[5,11] The proportion of isolated unilateral involvement observed in this study, particularly right-sided predominance, may in part reflect disease at an earlier stage or detection bias related to referral indications, and warrants further longitudinal evaluation.

Family history plays an important role in the diagnosis of ADPKD given its hereditary basis. In this study, 36.6% of participants reported a positive family history of kidney disease, an equal proportion reported no family history, and 26.9% were uncertain. Notably, a positive family history was most frequent among patients with isolated left kidney involvement. These findings are broadly consistent with the literature describing ADPKD as one of the most common inherited kidney disorders with marked familial clustering.[2,5] Cornec-Le Gall et al. reported that while most ADPKD cases demonstrate a positive family history, a subset of patients lack a known family history because of de novo mutations, incomplete family medical records, or undiagnosed disease among relatives—a pattern that may also explain the substantial proportion of "uncertain" responses observed in the present cohort.[3]

The relationship between family history and hypertension was also examined. A markedly higher proportion of hypertensive participants reported a positive family history of kidney disease (64.7%) compared with non-hypertensive participants (35.3%). This observation is consistent with the well-established role of hypertension as one of the earliest and most common clinical manifestations of ADPKD, frequently preceding measurable decline in renal function.[14] Ecder similarly emphasized the high prevalence of hypertension and its associated cardiovascular risk among ADPKD patients,[18] while

Schrier et al. demonstrated that elevated blood pressure is linked to accelerated renal disease progression and poorer long-term outcomes.[16] Taken together, these associations support early ultrasonographic screening of individuals with a family history of kidney disease or hypertension, enabling timely blood pressure control and closer clinical monitoring.

Overall, the findings of this study are consistent with the existing literature regarding the ultrasonographic characteristics of PKD, the contribution of family history, and the association between PKD and hypertension. The predominance of bilateral renal involvement together with the familial and hypertensive associations observed here reinforce the established understanding of ADPKD and support the continued use of ultrasonography as an effective, accessible, and non-invasive diagnostic tool for the early detection and ongoing evaluation of PKD in routine clinical practice.

Limitations

This study has several limitations. It was conducted at a single tertiary care hospital with a relatively modest sample size, which may limit the generalizability of the findings to the broader population. The cross-sectional design did not permit assessment of disease progression over time, and the use of convenient (non-probability) sampling may have introduced selection bias. Genetic testing and long-term clinical follow-up were not performed, which would have allowed more definitive confirmation of ADPKD and a more comprehensive evaluation of disease severity and progression.

CONCLUSION

Polycystic kidney disease was identified in a substantial proportion of patients undergoing abdominal ultrasonography at this tertiary care hospital, with bilateral renal involvement representing the most common ultrasonographic pattern. A considerable proportion of affected individuals reported a positive family history of kidney disease, supporting the hereditary basis of the disease, and hypertension was more prevalent among participants with a positive family history. These findings reinforce ultrasonography as an effective, accessible, and reliable tool for the diagnosis and evaluation of PKD, and highlight the value of targeted screening among individuals with a family history of kidney disease or hypertension to support early detection, timely intervention, and improved long-term renal outcomes.

Recommendations

Based on these findings, routine abdominal ultrasonography should be encouraged for the early detection and evaluation of PKD, particularly among individuals with a positive family history of kidney disease or hypertension. Patient education and awareness programs addressing the hereditary nature of PKD and the value of regular screening are recommended. Multicenter studies with larger, randomly selected samples and longitudinal follow-up—ideally incorporating genetic testing—are recommended to validate these findings and improve their generalizability.

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