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Ocular Manifestations of Autosomal Dominant Polycystic Kidney Diseases

Sankha Subhra Chaudhuri[1], Mousumi Bandypadhyay[2], Kali Sankar Das*[3], Soumyadeep Majumdar[4], Madhurima Kumar[5], Purban Ganguly[6]

[1,5]Senior Resident, Regional Institute of Ophthalmology, Kolkata, India
[2] Principal, Burdwan Medical College, Burdwan, India
[3]Associate Professor, Regional Institute of Ophthalmology, Kolkata, India
[4,6]Assistant Professor, Regional Institute of Ophthalmology, Kolkata, India

*Corresponding Author: Kali Sankar Das Email: docksdas97@gmail.com

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Abstract: INTRODUCTION: At about 6 weeks of gestation both eyes and kidney start to develop and hence they have a strong connection**.** As a result, a patient who suffers from any kidney disease ends up with different ocular disease manifestation. Many congenital anomalies of kidneys can also present as an ocular syndrome. Autosomal dominant polycystic kidney disease (ADPKD) is an inherited kidney disease, which also manifests numerous eye problems. The study helps to identify the ocular associations with ADPKD with creates awareness towards frequent eye checkup. **AIMs and OBJECTIVES:** To report the ocular manifestation of Inherited Renal Disease and to identify and characterize the ocular manifestation among patients diagnosed with inherited renal disease. This is a hospital based Cross sectional study, 61 patients were included. **RESULTS:** Most common ocular complaint was blurring of vision in 52 (85.24%) patients. After refractive error (55.73%), next most common ocular manifestations are, Arcus senilis (36.10%) and hypertensive retinopathy (34.2%). 39 (63.93 %) had no history of previous eye check-up. Most affected age group was patients aged > 35 – 40 years. **CONCLUSION:** Ocular manifestations can occur before systemic effects of disease starts to appear, thus identifying characteristic ocular manifestation can give a clinical clue allowing definitive diagnosis without expensive and invasive procedures in early course of disease allowing timely intervention.

Keywords: ADPKD, eye, Arcus senilis, retinopathy, kidney, ocular disease.

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INTRODUCTION

The eyes and kidney, both these develop during the same embryonic age at around 6th week of gestation. Hence, they share a strong connection and correlation in between eyes and kidneys disease process. Both these organs are a common target of systemic diseases. In many times it is evident that the renal disease or its treatment has a fatal consequence on eyes. As a result, a patient who suffers from any kidney disease end up with different ocular disease manifestation. Actually, patients with renal diseases are always at a greater risk to have any ocular disease condition in near future. Many congenital anomalies of kidneys can also present as an ocular syndrome [1].

It is evident that in various diseases pathogenesis, the ocular and renal systems are supposed to targets of end-organ damage. Many studies suggested that many renal conditions lead to cause secondary ocular involvement [2]. Likewise, chronic kidney disease (CKD), diabetic retinopathy (DR), age-related macular degeneration (ARMD), glaucoma, and cataract are associated with age, metabolic, and vascular risk factors such as diabetes, hypertension, and smoking [3].

Inherited renal/kidney diseases (IKD) is one of the major causes of end stage renal disease (ESRD), attributing to about two-third of renal failure in children and 10% to 20% in adults [4]. Prevalence can vary from 1 in 1000 for adult polycystic kidney diseases (ADPKD) to 1 in 10,000. Presence of characteristic ocular manifestation can help in giving diagnostic clue in these conditions. This study indirectly outlines the awareness of eye check-up in patients with ADPKD and outlines the delay on the part of patients in presenting to ophthalmologist for eye evaluation.

AIMS AND OBJECTIVES

- AIM
  - To report the ocular manifestation of Inherited Renal Disease.
- OBJECTIVES

- To identify and characterize the ocular manifestation among patients diagnosed with inherited renal disease.

METHODOLOGY

This is a hospital based Cross sectional study. A total of 61 patients were included in this study. All these patients were diagnosed with Autossomal dominant polycystic kidney disease (ADPKD) . They were attended Nephrology OPD or admitted under Nephrology ward. The study conducted at department of Nephrology, Medical College, Kolkata and department of Regional Institute of Ophthalmology (RIO), Kolkata during a tenure of 3 years (September 2019-July 2022). A consent was taken from all these 61 patients along with proper history. We largely selected those patients who gave history of chronic kidney disease, chronic renal disease, kidney, haemodialysis with eye, retinopathy, cataract, cornea, conjunctiva, glaucoma and ARMD, retinal vessels, and oculorenal syndromes.

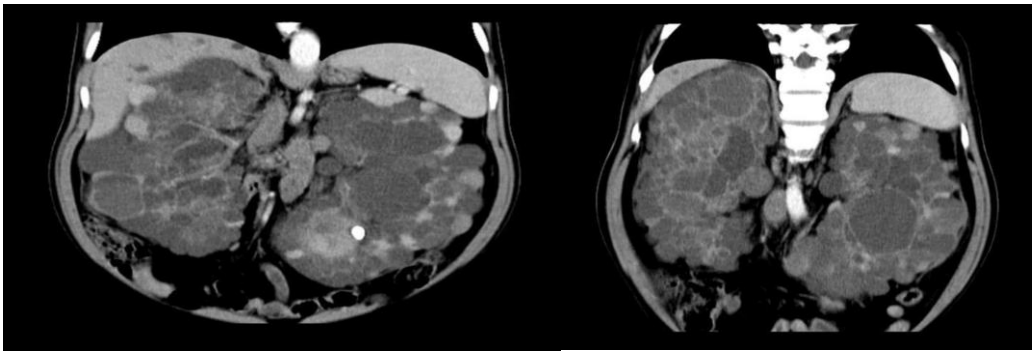
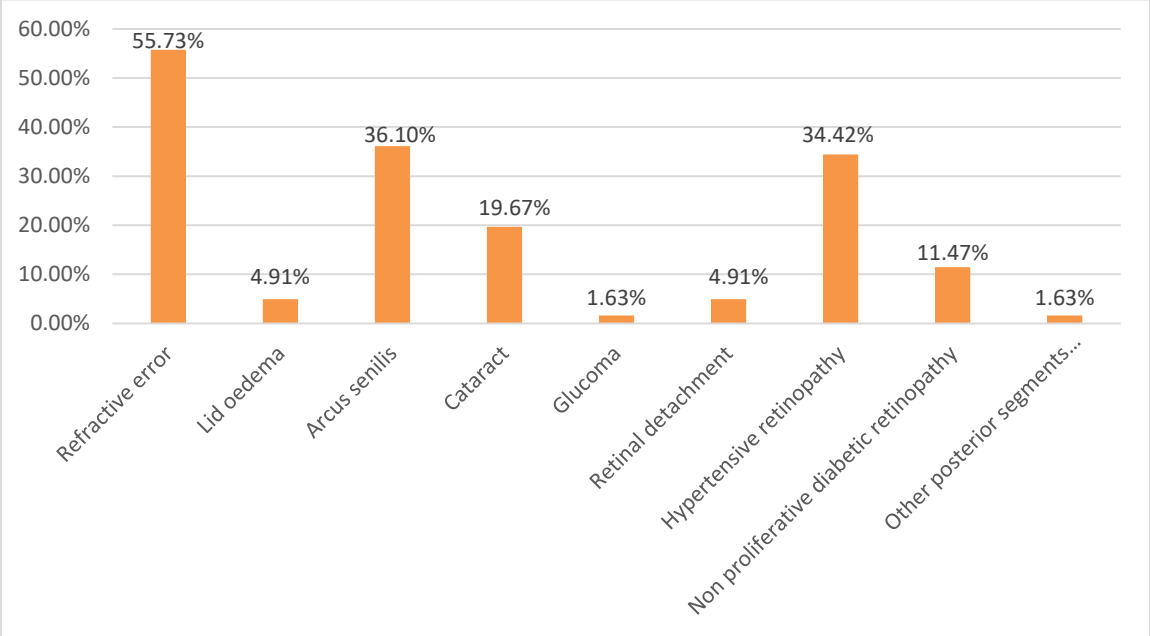


Fig-1 **Fig-2**

Figure-1 and 2: shows both kidneys are grossly enlarged with loss of normal reniform contour. Healthy renal parenchyma is seen replaced with multiple thin walled cysts (average diameter 2.5 cm to 5.0 cm)- suggestive of autosomal polycystic kidney disease (ADPKD). No calculus or hydronephrosis is seen on any side.

RESULTS

Most common ocular complaint was blurring of vision in 52 (85.24%) patients. Out of 61 patients, 39 (63.93 %) had no history of previous eye check-up following diagnosis of ADPKD. 16 (26.22%) patients were on regular follow up with an ophthalmologist. Best corrected visual acuity in 43 patients was 6/6.(Fig.1).



After refractive error (55.73%), next most common ocular manifestations are, Arcus senilis (36.10%) and hypertensive retinopathy (34.2%). Incidence of glaucoma is rare though (1.63%).

Table-1: Distribution of age and gender

Age group	N=61	Gender distribution	
		Male (n=37)	Female (n=24)
20-25 years	11	6	5
>25 – 30 years	9	6	3
>30-35 years	16	10	6
>35-40 years	21	12	9
> 45 years	4	3	1

Male were predominating one. Most affected age group was patients aged > 35 – 40 years followed by patients of >30-35 years age group.

Table-2: Distribution of comorbidities

Type of comorbidities	N=61
Diabetes mellitus	17
Hyperlipidemia	11
Hypertension	13
Obesity	9
Vit D deficiency	11

Most common co-morbidity associated was Diabetes mellitus (27.87%). Hypertension was second most common. It was found among 13 patients (21.31%). Hyperlipidemia and Vit D deficiency was found among 11 patients.

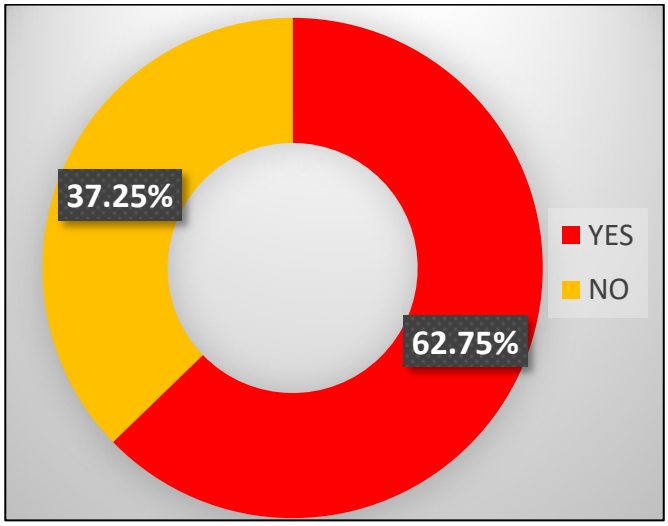


Figure-3: Distribution of smoking habits among the affected patients

DISCUSSION

The kidneys and eyes share developmental, structural, physiological, and pathological pathways, because the development of the kidneys and retina occur at the same embryonic stage (about the fourth to sixth week of gestation), hence strong correlation is seen between kidney and eye diseases [2-5]. Both glomerulus and choroid have extensive vascular networks of similar structure. Inner retina and glomerular filtration barrier share similar developmental pathways. The renin–angiotensin–aldosterone system (RAAS) is found in both the kidneys and various ocular tissues, which regulates blood volume and systemic vascular resistance [4]. Glomerular basement membrane (GBM) and Bruch's membrane–both membranes contain 3, 4, and 5 type IV collagen chains [6,7] Thus, diseases involving type IV collagen can affect both eyes and kidneys, resulting in retinopathy and nephropathy developing simultaneously, as seen in Alport syndrome [8,9]. In anti-GBM disease, IgG autoantibodies develop against 3 chains, which get deposited on GBM resulting in glomerulonephritis [10]. Similarly, IgG deposition on Bruch's membrane results in the development of choroidal ischemia.

High Myopia is also reported to be seen in APKCD, but none of the cases included in this study showed high myopia [11]. Lid oedema was noted in 14% cases

associated with generalized body swelling. All these patients were suffering from end stage renal disease. None of the cases of ADPKD had typical features of blepharochalasis as suggested by Alain Meyrier et al, which included oblique drooping of the upper lid to such an extent that eyelashes appear to be emerging beneath the eyelid [12]. Most of these findings include retinal changes secondary to diabetes mellitus and hypertension. In literature present till date association of retinal detachment in ADPKD is not mentioned except in one study done in transgenic mice where retinal degenerations were noted and these changes were correlated with presence of mutated polycystic kidney disease (Pkd) gene in transgenic rat [13]. Few reports of retinal vessel occlusion (RVO) related to systemic vasculopathy of ADPKD are available. In a case report Qi Qian reported retinal vascular occlusion in ADPKD patients with no known risk factors and controlled hypertension and he stated that RVO can occur in younger age group in patients with ADPKD [14].

CONCLUSION

Variable presentation and requirement of extensive work-up involving costly investigations makes the diagnosis of ADPKD difficult. In many instances ocular manifestations can occur before systemic effects of disease starts to appear, thus identifying characteristic ocular manifestation can give a clinical clue allowing definitive diagnosis without expensive and invasive procedures in early course of disease allowing timely intervention.

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