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Original Article

Evaluation of Kidney Transplantation Outcomes of Pediatric Patients with Ciliopathy: A Single Center Experience

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ABSTRACT

Aim: Ciliopathies are rare genetic diseases referring to a group of syndromic diseases characterized by the deterioration of the structure of the cilia, which may cause kidney failure in childhood. Follow-up of the patients with ciliopathy after kidney transplantation is important for graft survival.

Materials and Methods: This study was designed as a retrospective cohort trial. One hundred and fifty-one renal transplanted children (111 were non-ciliopathy and 31 were ciliopathy) were evaluated. Sociodemographic characteristics and clinical information regarding transplantation stage were recorded.

Results: The mean age of the 31 patients (16 female/15 male) with the diagnosis of ciliopathy was 11.1±3.5 years and their mean follow-up duration was 7.7±4.8 years. Four of the patients (12.9%) experienced acute rejection and two patients had graft loss. Eleven patients had polycystic kidney disease, ten patients had cystic dysplasia and ten patients had nephronophthisis as their primary diagnosis. Graft survival rates were similar for transplants from living and cadaveric donors in those patients with ciliopathy. The data of the 31 patients who underwent kidney transplantation with the diagnosis of ciliopathy were compared with the 111 patients with the diagnosis of non-ciliopathy. The rates of hypertension, acute rejection and graft loss were similar in both groups. According to a Kaplan-Meier analysis, the graft and patient survival rates for those patients with ciliopathy and for those with non-ciliopathy were similar (p=0.123, p=0.370).

Conclusion: Kidney transplant outcomes of patients with ciliopathy from well-selected living donors in terms of graft and patient survival are favorable.

Keywords: Ciliopathies, pediatric kidney transplantation, polycystic kidney disease, nephronophthisis

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Introduction

Ciliopathy is a term which covers a wide group of diseases which occur as a result of many different genetic mutations. It may affect many organ systems which have ciliary cells, making it one of the most common causes of end stage kidney disease (ESKD), especially in childhood. In this group of diseases, known as hepatorenal fibrocystic diseases, fibrocystic diseases of the liver, diabetes and skeletal dysplasia can be observed, as well as kidney, eye, and central nervous system findings (1). The three leading causes of ESKD in children during 2012-2016 were reported to be primary glomerular diseases (22.3%), congenital anomalies of the kidney and urinary tract (CAKUT; 21.9%), and cystic/hereditary/congenital disorders (11.7%), respectively (2). This ranking was the same as that in the annual report published in 2023 (3). In those patients with ciliopathy, the risk of complications after kidney transplantation may increase due to extrarenal involvement due to cilia disorder. Therefore, a careful selection of suitable candidates for kidney transplantation and a careful follow-up after transplantation are required. Common post-transplant complications may include infections, transplant rejection, and problems with cilia. Therefore, the early recognition and management of post-transplant complications and a multidisciplinary team approach with the involvement of all stakeholders are critical. Ciliopathies are rare genetic diseases characterized by a deterioration of the cilia structure, leading to kidney failure in childhood. While their multisystemic nature is well-known, pediatric data regarding post-transplant outcomes remain limited. The aim of this study was to evaluate the clinical outcomes, graft survival, and patient survival of pediatric kidney transplant recipients with ciliopathy and to compare these results with a non-ciliopathy control group in order to provide insights into the management of these complex patients.

Materials and Methods

Design and Setting

This study was designed as a retrospective cohort trial. The study protocol was approved by Ege University's Medical Research Ethics Committee (approval number: 22-12.1T/4, date: 15.12.2022). This study was conducted at our University's Renal Transplantation Center. Following the early postoperative period, all pediatric patients are monitored in the Pediatric Nephrology ward and followed up in the outpatient clinic. Kidney biopsies were performed in those patients showing clinical signs of rejection. The diagnosis of rejection was made via the examination of the kidney biopsies by two experienced pathologists.

Study Population and Sampling

We retrospectively evaluated 151 children who underwent renal transplantation in our Renal Transplantation Center. The vast majority of kidney-transplanted patients (111 patients) were diagnosed with non-ciliopathic disorders. Thirty-one children were diagnosed with ciliopathy and 9 children had unknown etiology. Those with unknown etiologies were excluded from this study. Thirty-one of ciliopathy patients who met the requirement of at least six months of follow-up agreed to participate in this study. Although complete molecular genetic data were not available for all patients included in this study, the diagnoses of those patients in the polycystic kidney disease (PKD) subgroup were genetically confirmed. The follow-up results of those patients with ciliopathy were compared with the non-ciliopathy group.

Data Collection

The date of birth of the patients, their gender, the cause of ESRD, dialysis information, donor information, mismatch numbers, warm and cold ischemia times, post-transplant treatment protocols, rejection status, graft loss, and survival data were recorded. At their last visit, the urea and creatinine levels of the patients were recorded. The updated Schwartz formula was used to calculate estimated glomerular filtration rate (eGFR) levels (4).

Statistical Analysis

We used the IBM SPSS statistical software package version 20.0. Continuous data are presented as mean±standard deviation under parametric conditions and median (minimum-maximum) under non-parametric conditions. Categorical variables are presented as numbers and percentages. Chi-square analysis was used for categorical variables. When analyzing independent continuous variables, the Mann-Whitney U test was used under nonparametric conditions. The One-Way Analysis of Variance test was used for parametric conditions and the Kruskal-Wallis test was used for non-parametric conditions in order to compare the three groups of ciliopathies. Bonferroni correction was used to distinguish any differences between groups. Survival analysis was performed with the Kaplan-Meier method using the log-rank test. The results obtained were evaluated and interpreted by all of the researchers. Statistical significance was accepted as $p<0.05$.

Results

Of the 31 patients (16 female/15 male) with the diagnosis of ciliopathy, the mean age at the time of transplantation was 11.1±3.5 years and the mean follow-up duration was

7.7±4.8 years. Two patients who were siblings, diagnosed with PKD, had graft loss. The causes of the graft loss were chronic rejection. Two patients, one with a functioning graft, died. The causes of death of the patients were due to central nervous system malignancy and fungal pneumonia. The mean creatinine and estimated GFR levels of the patients at their last visit were 1.5±1.4 mg/dL and 60±26 mL/min/1.73 m^2, respectively.

As the primary diagnosis of the patients with ciliopathy, eleven patients had PKD, ten patients had cystic dysplasia and ten patients had nephronophthisis (NPHP). Of those patients diagnosed with PKD, 8 were autosomal recessive PKD (ARPKD), and 3 were autosomal dominant PKD (ADPKD). The patients with ciliopathy were similar in terms of age, gender, follow-up time, rejection, graft loss, creatinine and eGFR at their last visit when they were grouped according to their primary diagnosis. The only statistically significant difference between the 3 groups was that all of the patients with a primary diagnosis of polycystic kidney used Anti-thymocyte globulin (ATG) in their induction regimen. The demographic and clinical characteristics of the patients in terms of their primary diagnosis of ciliopathy are shown in Table I.

Regarding the comparison with the control group, Table II shows the comparative demographic, clinical, and laboratory data between the 31 ciliopathy patients and the 111 non-ciliopathy patients. Regarding the treatment protocols, the induction regimen and the preferred calcineurin inhibitor were similar in both groups. However, azathioprine was not used as a nucleoside agent in those patients with ciliopathy. The rates of hypertension, acute rejection and graft loss were similar in both groups, and there was no significant difference between creatinine and eGFR levels measured at the last visit. The use of induction regimens and calcineurin inhibitors were similar in both groups. The graft survival rate was 93.5% in those patients with ciliopathy and 81.1% in those with non-ciliopathy. Patient survival rates were 93.5% in those patients with ciliopathy and 97.3% in those with non-ciliopathy. According to Kaplan-Meier analysis evaluating the difference in graft survival of those patients with ciliopathy and those with non-ciliopathy using a log-rank test, graft survivals of both groups were similar (p=0.123) (Figure 1). Kaplan-Meier analysis also revealed that there was no statistically significant difference in terms of patient survival between those patients with a diagnosis of ciliopathy and those with non-ciliopathy (p=0.370).

Table I. Demographic and clinical characteristics of patients in terms of primary diagnosis of ciliopathy

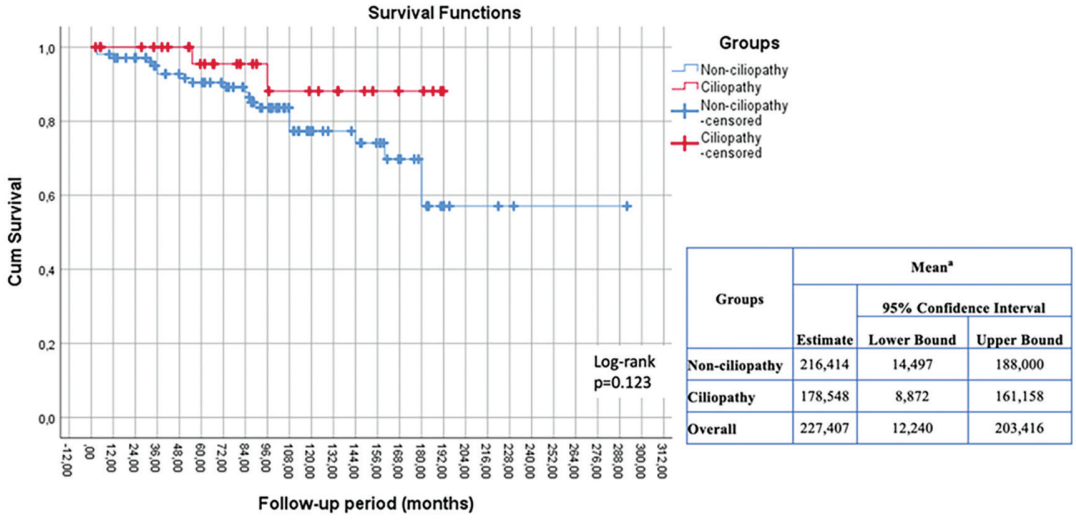
	Polycystic kidney n=11	**Cystic dysplasia n=10**	**Nephronophthisis n=10**	**Total n=31**	**p value**
Gender (male/female)	6/5	5/5	4/6	15/16	0.794
RRT (PD/HD/preemptive)	7/1/3	7/2/1	6/3/1	20/6/5	0.633
Donor type (living/cadaveric)	5/6	5/5	8/2	18/13	0.208
Induction (ATG/basiliximab)	11/0	7/3	7/3	25/6	0.049*
Calcineurin inhibitor (CsA/Tac)	5/6	5/5	5/5	15/16	0.971
Acute rejection, n (%)	3 (27%)	1 (10%)	0 (0%)	4 (12.9%)	0.108
Hypertension, n (%)	8 (72.7%)	7 (70%)	4 (40%)	19 (61.3%)	0.140
Graft loss, n (%)	2 (18.2%)	0 (0%)	0 (0%)	2 (6.5%)	0.111
Status (living/dead)	10/1	9/1	10/0	29/2	0.424
Age at transplant (years)¶	11.7±3.2	8.8±3.5	11.6±2.7	11.1±3.5	0.558
Time on RRT (months)Ω	31 (24-102)	43 (4-96)	16 (2-48)	30 (2-102)	0.191
Donor age (years)¶	21.4±21.3	27.0±16.8	34.7±10.4	29.8±17.7	0.841
Follow-up time (years)¶	7.8±2.6	11.7±2.8	6.8+5.7	7.7±4.8	0.489
Warm ischemia time (min)Ω	36 (4-90)	2.5 (1-10)	3 (1-32)	5 (1-90)	0.136
Cold ischemia time (min)Ω	450 (36-1,320)	208 (27-2,000)	55 (30-565)	121 (27-2,000)	0.259
Creatinine at last visit (mg/dL)¶	2.4±2.1	0.9±0.3	1.0±0.2	1.5±1.4	0.126
eGFR at last visit (ml/min/1.73 m^2)¶	48±33	72±22	58±12	60±26	0.107

¶Mean±SD, ΩMedian (min-max)
RRT: Renal replacement therapy, PD: Peritoneal dialysis, HD: Hemodialysis, ATG: Anti-Thymocyte Globulin, CsA: Cyclosporine, Tac: Tacrolimus

Table II. Comparison of the demographic, clinical and laboratory data of renal transplanted patients with a diagnoses of either ciliopathy or non-ciliopathy

Categorical variables	**Ciliopathy n=31**	**Non-ciliopathy n=111**	**p value¶**
Gender (male/female)	15/16	54/57	0.979
Donor type (living/cadaveric)	18/13	55/56	0.402a
Induction (ATG/basiliximab)	25/6	89/22	0.954
Calcineurin inhibitor (CsA/Tac)	15/16	50/61	0.991
Nucleoside inhibitor (MMF/AZA)	31/0	83/28	0.002*
Delayed graft function (yes), n (%)	4 (12.9 %)	9 (8.1%)	0.477
Hypertension after transplantation, n (%)	19 (61.2%)	65 (58.5%)	0.664
Acute rejection, n (%)	4 (12.9%)	20 (18%)	0.453
Graft loss, n (%)	2 (6.5%)	21 (18.9%)	0.096
Status (dead), n (%)	2 (6.5)	3 (2.7%)	0.090
Continuous variables			**p valueΩ**
Age at Tx (years)	10.9 (3.8-17.6)	12.9 (1.7-17.9)	0.183
Follow-up time (years)	7.4 (2.8-15.9)	8.5 (0.8-16.1)	0.449
Warm ischemia time (min)	5 (1-90)	5 (1-60)	0.850
Cold ischemia time (min)	121 (27-2,000)	67 (15-2,800)	0.683
Creatinine at 1st year of Tx	0.7 (0.4-1.7)	0.8 (0.3-1.8)	0.146
Creatinine at 5th year of Tx	0.9 (0.5-9.9)	1.0 (0.4-9.6)	0.282
Urea at last visit (mg/dL)	40 (17-207)	37 (14-155)	0.669
Creatinine at last visit (mg/dL)	1.24 (0.57-6.0)	1.33 (0.49-10.2)	0.099
eGFR at last visit (mL/min/1.73 m^2)	56 (9-123)	54 (5-125)	0.444

¶Chi-square test, ΩMann-Whitney U
RRT: Renal replacement therapy, PD: Peritoneal dialysis, HD: Hemodialysis, ATG: Anti-thymocyte globulin, CsA: Cyclosporine, Tac: Tacrolimus, AZA: Azathioprine, MMF: Mycophenolic acid, NTx: Transplantation



Groups	Mean[a]		
		95% Confidence Interval	
	Estimate	Lower Bound	Upper Bound
Non-ciliopathy	216,414	14,497	188,000
Ciliopathy	178,548	8,872	161,158
Overall	227,407	12,240	203,416

Figure 1. Graft survival of those patients with ciliopathy or non-ciliopathy

Discussion

Ciliopathic dysfunctions occur in any organ and the ones most predominately affected include the kidney, eye, liver and brain, with the kidneys being the most commonly affected (1). Kidney manifestations display pathologies ranging from a urinary concentration defect in normal appearing kidneys to cystic dysplastic kidneys. ADPKD and ARPKD represent the most common ones, followed by NPHP, cystic dysplastic kidneys, medullary sponge kidney, and several overlapping phenotypes. PKD is a group of monogenic disorders characterized by the presence of multiple cysts, primarily in the kidney and liver (5,6).

ADPKD is one of the most commonly encountered genetic origins of chronic kidney disease (CKD). Its incidence is estimated to be between 1 in 400 to 1 in 1,000 individuals. Conversely, ARPKD is a rare condition, with an estimated incidence of 1 in 20,000 individuals (1).

NPHP stands out as the most prevalent genetic cause of CKD disease occurring within the first three decades of life. Its prevalence among the population experiencing end-stage renal failure in childhood is estimated to be approximately 5%. Patients typically present with symptoms which include polyuria, polydipsia, enuresis, and anemia (1,7,8).

Renal dysplasia occurs as a result of defective differentiation of the renal parenchyma during kidney development (9). Histologically, dysplastic features may include incompletely branched collecting ducts surrounded by undifferentiated mesenchymal stroma. It is important to note that renal dysplasia has been observed in several ciliopathic disorders, such as Bardet-Biedl syndrome and Meckel-Gruber syndrome (1,9,10). Given that renal dysplasia is fundamentally a developmental phenotype, its presence within the context of a ciliopathy likely indicates a more severe genetic makeup.

Almost 3% of the population reach ESRD in the United States and as one of the leading causes of CKD, PKDs are common indications for dialysis or kidney transplant. While many studies have evaluated the results of kidney transplant in patients with PKD, most of these have only published the results after kidney transplant in patients with ADPKD (11-16).

In our study, we wanted to present the kidney transplantation results of all ciliopathies, including both PKDs (ADPKD and ARPKD), NPHP and cystic dysplasia. Our single center study comprises our 31-year experience and it is one of the largest single center studies covering children with different types of PKD who underwent kidney transplant. As expected, most of our study patients had PKD, similar to recent studies (17). In children, ADPKD is typically characterized by preserved renal function despite progressive structural changes (18). Reaching end-stage kidney disease (ESKD) is uncommon during childhood, with approximately 50% of patients reaching this stage by age 60. However, research has identified specific high-risk subgroups and systemic causes which can accelerate this progression. The progression to ESKD in the pediatric population varies greatly and depends largely on the age of onset and genetic factors (19). Cadnapaphornchai (20) reported a subset of ADPKD diagnosed before 18 months of age (very early-onset ADPKD) which is at the highest risk for early loss of kidney function and progression to ESKD during childhood or adolescence. While ADPKD typically progresses to ESKD in adulthood, the presence of three ADPKD patients in our pediatric cohort suggests a more aggressive clinical phenotype. In our study, the mean age at diagnosis for the three patients with ADPKD was 26 months. Although this exceeds the 18-month threshold established in the literature for very early-onset ADPKD, we believe this discrepancy is due to the small sample size (n=3), which prevents alignment with the standard definition. Despite the small cohort, these cases demonstrate clinical and structural characteristics consistent with early-stage disease progression. Therefore, we argue that this patient group should be considered within the very early-onset ADPKD spectrum for early loss of kidney function and progression toward ESKD as described in the literature.

In our study, two siblings diagnosed with PKD experienced graft loss due to chronic rejection. This finding is noteworthy and unlikely to be coincidental; it raises the possibility of shared genetic factors or familial disease-related triggers which may influence the immune response post-transplantation. The lack of molecular genetic data for all of the study group was a limitation in our retrospective study; however, the clinical presentation of the two siblings was consistent with severe polycystic disease.

Hypertension is an early and common symptom throughout the progression of both ADPKD and ARPKD and it also contributes to increased cardiovascular morbidity and mortality. Hypertension often develops without a decrease in kidney function. This indicates the existence of extrarenal causes in addition to renal causes in the etiology of hypertension. In some publications, the prevalence of hypertension in this patient group has been reported as being 50-70% (17,21-24). Conversely, in NPHP, blood pressure usually remains within the normal range without

progression to ESRD (25). In our study, we observed a high rate of hypertension in those patients diagnosed with ciliopathy, however, the frequency of hypertension was not different from the transplant recipients in the non-ciliopathy group. Regarding the frequency of HT in patients with PKD, NPHP and dysplasia, although post-transplant HT was more common in PKD and dysplasia, this difference was not statistically significant.

The necessity of native kidney nephrectomy before, after, or during kidney transplantation remains a matter of debate (26). None of the patients in this study required nephrectomy.

Most studies have reported more satisfactory results of kidney transplants from living donors than from cadavers (27). It has also been emphasized that living kidney donation deserves special attention and that genetic testing should be performed in transplants from living kidney candidates under the age of 30 (28). Due to the genetically inherited nature of the disease, adult studies have reported higher rates of cadaveric transplantation in those patients with ciliopathy (17). In our study, there was no difference in the frequency of cadaveric and living donors between the patient groups with and without ciliopathy. This can be explained by the fact that the median donor age (parents) in our patients was 40 years and above. The older average parental age can be considered as a reason for the increased frequency of transplants with living-related donors.

In the past years, it has been reported that the frequency of rejection in those patients who underwent kidney transplantation with a diagnosis of PKD was higher than in those with a diagnosis of non-ciliopathy (29). In our study, although the rejection rate was higher in patients with ciliopathy, it was not statistically significant.

Kanaan et al. (28) published a study showing excellent patient and graft survival rates after kidney transplantation in ADPKD patients. Barbouch et al. (29), in their study comparing kidney transplant patients due to ADPKD and other nephropathies in terms of graft and patient survival, reported that they did not observe any significant difference between the two groups. In our study, patient and graft survival were found to be similar in the ciliopathy and non-ciliopathy groups. This result shows that promising results can be obtained with close monitoring, even in genetic diseases with multisystem involvement, such as ciliopathy.

Mehrabi et al. (17) reported that the 1-year, 3-year, 5-year, and 10-year graft survival rates for cadaveric donor transplant recipients in 250 cases of PKD were 97%, 96%, 95%, and 85%. In the same study, cumulative 1-year, 3-year, 5-year and 10-year graft survival rates for living donor transplant recipients were reported as being 100%, 100%, 100% and 75%. Graft survival rates were found to be similar for kidney transplants from living and cadaveric donors (17). In our study, eighteen of the patients received transplants from a living donor and 13 from a cadaveric donor. The 1-year, 5-year and 10-year graft survival rates were 92%, 92%, and 85% for the cadaver donors and 100%, 100%, and 100% for the living donors, respectively. Our study supports that the idea that transplantation from a carefully selected living donor may be a good option even in genetically inherited diseases such as ciliopathy.

Our study highlights the complexity of pediatric ciliopathies in the context of transplantation. However, it is essential to acknowledge that "ciliopathy" functions as an umbrella term for a highly heterogeneous group of diseases, including ADPKD, ARPKD, and NPHP, which differ significantly in their pathophysiology and systemic manifestations. While combining these into a single analytical category was necessary for statistical viability in this rare pediatric cohort, it inherently limits our ability to derive disease-specific conclusions. Specifically, the inclusion of ADPKD, a condition more frequently studied in adults, underscores the aggressive nature of early-onset phenotypes within the ciliopathy spectrum.

Regarding prognosis, our findings suggest that kidney transplants from well-selected living donors yield more favorable outcomes in terms of graft and patient survival for those patients with ciliopathy.

Study Limitations

Several limitations of this small, single-center retrospective cohort study warrant consideration. First, the retrospective design prevented us from obtaining complete molecular genetic data for all patients, a significant drawback when studying genetically determined disorders. Second, the comparative analysis between the ciliopathy and non-ciliopathy groups was not fully adjusted for potential confounders, such as the duration of ESKD or the specific severity of extrarenal involvements. Additionally, unadjusted factors such as varying immunosuppressive regimens, driven by the primary disease's systemic manifestations, may have influenced the interpretability of our results.

Conclusion

Finally, our study was constrained by its small sample size, particularly after subdividing into specific disease groups. This reduced the statistical power to detect minor differences; therefore, the lack of significant differences in survival rates might reflect a Type II error (lack of power) rather than true clinical equivalence. Consequently, these results should be interpreted with caution and they should be validated by larger, multicenter studies.

Ethics

Ethics Committee Approval: The study protocol was approved by Ege University's Medical Research Ethics Committee (approval number: 22-12.1T/4, date: 15.12.2022).

Informed Consent: This study was designed as a retrospective cohort trial.

Footnotes

Authorship Contributions

Surgical and Medical Practices: S.T., B.S.K., S.Ş., G.K., T.Ö.S., S.C.T., A.K., İ.K.B., Concept: S.T., N.B.Ö., İ.K.B., Design: S.T., N.B.Ö., İ.K.B., Data Collection or Processing: S.T., N.B.Ö., S.Ö., Analysis or Interpretation: S.T., N.B.Ö., S.Ö., Literature Search: S.T., N.B.Ö., B.S.K., S.Ş., İ.K.B., Writing: S.T., N.B.Ö., G.K., İ.K.B.

Conflict of Interest: Four authors of this article, Sevgin Taner, Su Özgür, Ahmet Keskinoğlu and İpek Kaplan Bulut are members of the Editorial Board of the Journal of Pediatric Research. However, they did not involved in any stage of the editorial decision of the manuscript. The editors who evaluated this manuscript are from different institutions. The other authors declared no conflict of interest.

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