A First Case Report Of Mcardle Disease And Alive Kidney Transplantation

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Abstract

McArdle disease (Glycogen storage disease type V, GSDV) is an inherited disorder of glycogen metabolism affecting only skeletal muscles. A 56-year-old male patient with McArdle disease had the symptoms such as fatigue, muscle weakness since he was 8 years old in his history. A Pre-emptive kidney transplant from his wife was performed on the patient who has gone through rhabdomyolysis attacks during his life period. As far as known, this is the first case in the literature. We aim to share this process before and after the transplantation.

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Abstract

McArdle disease (Glycogen storage disease type V, GSDV) is an inherited disorder of glycogen metabolism affecting only skeletal muscles. A 56-year-old male patient with McArdle disease had the symptoms such as fatigue, muscle weakness since he was 8 years old in his history. A Pre-emptive kidney transplant from his wife was performed on the patient who has gone through rhabdomyolysis attacks during his life period. As far as known, this is the first case in the literature. We aim to share this process before and after the transplantation.

What's already known about this topic?

McArdle disease can be cause of acute kidney injury. But progression to end stage renal disease (ESRD) has never been reported.

What does this article add?

The repetitive CK elevation in Mcardle disease might cause ESRD or accelarate this process. In this case, we share our experience of renal transplantation in a patient with ESRD caused by probably Mcardle Disease.

Key words: Mcardle disease, Glycogen storage disease, kidney transplant, CK elevation

Introduction

McArdle disease (Glycogen storage disease type V, GSDV) is an inherited disorder of glycogen metabolism affecting only skeletal muscles. The disease was first reported by Brian Mcardle. Brian McArdle described an exercise intolerant patient who had muscle ache, cramps, and was unable to produce lactate during ischemic

exercise. (ischemic exercise?) Afterward, glycogen phosphorylase deficiency was detected in the patients. There was no detectable glycogen phosphorylase activity in the most of the patients. (1-4) The deficiency of myophosphorylase disrupts in muscle cells function and breakdown of the muscle cells. Myophosphosrilase deficiency is an autosomal recessive disease caused by mutations in the muscle isoform of phosphorylase (muscle glycogen phosphorylase [PYGM]) found in 11q13. (5) Although the symptoms are present in the first decade, the patients typically present in adolescence or early adulthood with exercise intolerance, fatigue, muscle pain, cramps, muscle swelling, and weakness. Creatinine kinase (CK) elevation and rhabdomylosis attacks can be seen in the patients (6). The aim of this study is to examine the results of a live donor kidney transplant (not previously shared in the literature to the best of our knowledge) before and after transplantation and to share our observations and experiences.

Case Report

The fifty-six-year-old male patient had previously symptoms such as fatigue, weakness, and muscle cramp since he was 8 years old. When he was eighteen years old, he was investigated with a pre-diagnosis of glomerulonephritis when he observed dark urine color as a result of tonsillitis. No biopsy was performed to the patient at that time.

And additionally, there was no report found which was associated with the diagnosis of glomerulonephritis.

In the followsup, the patient had symptoms such as fatigue, muscle cramps following the exercise and there were elevated CK levels and dark urine caused by myoglobinuria in the laboratory analysis.

The patient, who was evaluated in the neurology department with the increase of his current complaints at the age of thirty-five, was diagnosed with myophosphorylase deficiency (McArdle's disease) after muscle biopsy and genetic analysis (mutation was detected in the PYGM gene encoding the myophosphorylase enzyme). Afterward, myophosphorylase deficiency was detected in the 6 cousins of him following the screening of the family members in terms of Mcardle disease.

The elevated creatinine level was detected in the follow-ups at the age of 50. A kidney biopsy was recommended to the patient and the patient did not accept the kidney biopsy. After the increase in the creatinine levels, the patient was regularly followed up in our clinic. He was diagnosed with hypertension during the 6-year follow-up. Besides, recurrent rhabdomyolysis attacks and CK elevation were observed (Lifetime more than 50 times). Preparations for preemptive kidney transplantation were made as the creatinine level of the patient increased over time.

Preoperative, Intraoperative, and Postoperative Management:

In addition to the routine transplantation preparations for the donor and recipient, a carbohydrate-based diet was initiated for the patient. The appropriate intravenous (IV) anesthetics and neuromuscular blockers and IV hydration were administered to prevent the possible malignant hyperthermia and rhabdomyolysis. And Dantralone sodium was also kept available to prevent malignant hyperthermia. To reduce the risk of rhabdomyolysis, the rocuronium bromide was administered for the purpose of neuromuscular blockage. IV anesthetics (propofol and remiphentanil hydrochloride) were preferred instead of inhaler anesthetics. A dramatic decrease in CK and creatinine levels were observed in the follow-up postoperatively. The patient was discharged without complication on the 5th day. No rhabdomyolysis attacks or CK elevation was observed within the first month of postoperative (Table).

Discussion

In this case report, we shared the pretransplantation and posttransplantation process of McArdle patient who had kidney transplantation from a living donor (husband). To the best of our knowledge, this case was a first in the literature.Mcardle is a rare disease that is generally lately diagnosed. The long-term and intensive exercise cause rhabdomyolysis in this patient. The most common laboratory results are myoglobinuria and elevated CK levels. The recurrent myoglobinuria attacks are seen in more than %50 of patients. Myoglobin is excreted by the kidney and can cause kidney injury. Myoglobinuria has a toxic effect on renal tubular

cells and causes damage through the tubular obstruction. Acute kidney disease is extremely rare in McArdle patients.

In the Spanish national registry, acute kidney injury has been reported at a rate of 4 percent. In a study in the United Kingdom, myoglobinuria and acute kidney injury are reported at the rate of 62% and 11% percent respectively. In our patient, there was history of hypertension which started with elevated creatinin levels. He had no high blood pressure levels which can cause end stage renal disease. In addition, there were no history of habits such as cigarette, and alchohol or comorbid diseases, which could explain the end stage renal disease. The history of more than 50 rabdomylosis attacks and absense of any other possible etiology, which can explain the pathology, suggested the rabdomylosis attacks as a most possible cause of end stage renal disease.

The rabdomyolysis following the kidney transplantation, delayed graft rejection, and even graft loss were among the possible post-transplantation risks. Rhabdomyolysis has been reported in kidney transplantation cases due to the immunosuppressions used (tacrolimus and CD 25 monoclonal antibody). (10-11) We did not observe rabdomylosis in the post operative period in our case in which tacrolimus and anti-thymocyte were used as induction or immunsuppressant agents. Kidney transplantation can be performed in McArdle patients with end-stage renal disease in the appropriate preoperative and postoperative conditions.

Data Availability Statement

All datasets are available from the corresponding author upon reasonable request.

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Table 1 : Laboratory results of the patient

	Unit	**Before kidney Tx**										
			1st day	**2nd day**	**2nd day**	**3rd day**	**4th day**	**5th day**	**10th day**	**15th day**	**1st month**	**3 r**
Urea	mg/dl	182	167	61	61	32	38	35	46	34	36	4
Creatinine	mg/dl	7.28	5.26	2.01	2.01	1.0	0.87	0.82	1.23	1.12	1.04	0
CK	IU/L	4267	3539	3829	3829	4267	1177	374	221	156	141	3
Calcium	mg/dl	9.3	8.8	7.2	7.2	7.7	8.2	8.5	8.3	8.8	8.98	9
Magnesium	mg/dl	2.63				2.93						
Phosphorus	mg/dl	3.8	2.4				2.3	3.4	3.6		1.2	2
AST	U/L	14	23	28	28	57	45	26	14	18	16	2
ALT	U/L	13	15	15	15	20	23	25	11	20	17	2
Albumin	g/L	41	31.3	30	30	31	32	34	34	38	39	4
Potassium	mmol/L	4.89	3.37	4	4	4.18	3.69	4.26	4.77	5.07	4.76	4
WBC	10^3/uL	7.46	10.20	8.9	8.9	8.55	9.01	11.8	12.64	13.2	11.12	7
Hemoglobin	g/dl	11.4	10.9	9.1	9.1	9.8	10.4	10.5	9.7	10.2	11	1
Platelets	10^3/uL	275	242	195	195	188	193	207	316	293	296	3
Urine analysis	ph	7	6.5	6	6	6.5	7	5.5	5.5	5	5	
	Gravity	1007	1007	1008	1008	1007	1007	1007	1011	1019	1017	1
	Erythrocyte		+	-	-	-	-	-	-	-	-	-
	Protein	+	-	-	-	-	-	-	-	-	-	-
	Leukocyte	-	+	+	+	-	-	-	-	-	-	-
	ketone	-	-	-	-	-	-	-	-	–	-	-

CK: Creatine kinase