KIDNEY TRANSPLANTATION IN PATIENT WITH CYSTINURIA – CASE REPORT

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Introduction

Cystinuria is rare autosomal recessive genetic disorder affecting tubular reabsorbtion of cystin (and three other dibasic aminoacids), leading to cystin lithiasis, and expressed also in intestinal epithelium without known clinical manifestations. It rarely leads to end stage renal disease (ESRD) and need for kidney transplantation (TX). It is different from cystinosis, a lysosomal storage disease, in which the cystin accumulates in cells of different organs, leading to renal tubular disorder and ESRD. There are only few cases of kidney TX in patients with cystinuria described in literature to date.

Aim

To present a rare case of patient with cystinuria and kidney TX, the first and the only in Croatia according to our knowledge.

Case report

A 53 old man underwent kidney TX from deceased donor. He was previously treated by hemodialysis for two years. His basic renal illness was cystinuria with recurrent nephrolithiasis. He was diagnosed with nephrolithiasis at the age of 12 years. Infrared spectroscopy showed that stones consisted of cystin. His history included many nonconservative procedures for kidney calculi. Four years preceding the TX he presented with clinically malignant hypertension and nephrotic proteinuria and he underwent kidney biopsy that showed heavy nephroangiosclerosis and perihilar focal segmental glomerulosclerosis. His sister suffers from cystinuria, too, however, her kidney function is preserved and episodes of lithiasis rare. He was not immunized and received standard immunosuppressive therapy. The kidney TX was followed by delayed graft function. Now he is four months after the TX with stable renal function (creatininemia 113 μmol/L) and without further complications.

Conclusion

Cystinuria rarely leads to ESRD, therefore kidney TX for that diagnosis is not frequent. The first and only Croatian patient with cystinuria and kidney TX had successful TX and good kidney transplant function so far. Cystinuria is limited to native kidneys and does not recur in kidney transplant.

Removed renal cystin stones