CONGRESUL NAȚIONAL DE NEFROLOGIE
EDIȚIA A XII-A

Outcomes of kidney transplantation in patients with Fabry disease: real world experience

SOCIETATEA ROMÂNĂ DE NEFROLOGIE

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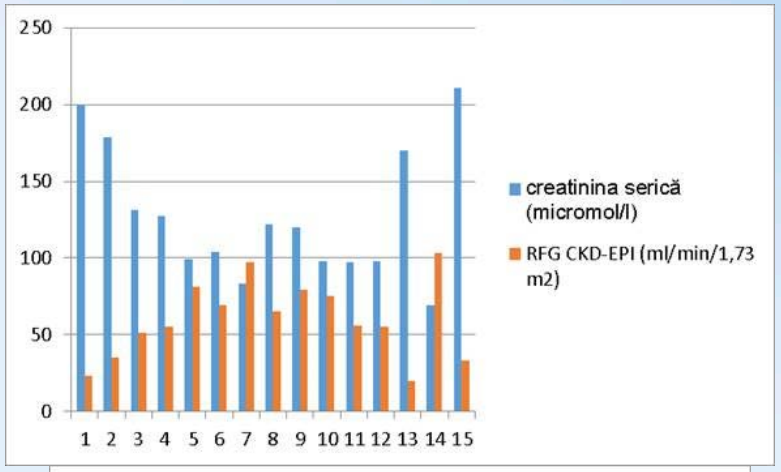
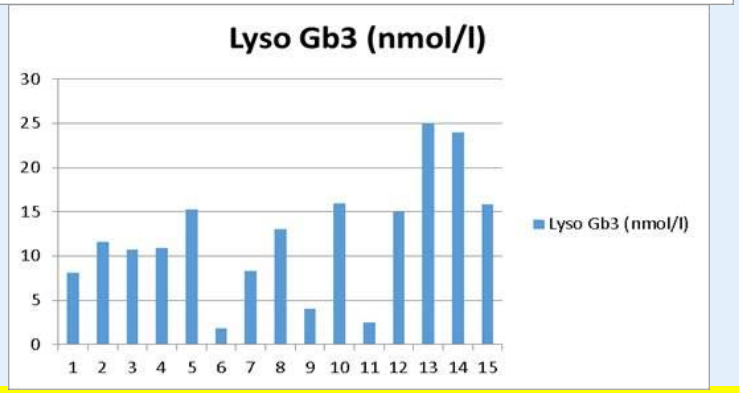
Introduction:
Fabry disease (FD) (OMIM #301500) is an X-linked genetic disorder of glycosphingolipid metabolism. Renal involvement is one of the major features of FD. Data on patients with FD and kidney transplant are scarce in the literature.

Methods:
Fifteen patients with classic FD (80% males) were treated in the French Referal Center for Fabry disease (www.centre-geneo.com) for a mean post-transplant follow-up of 12.6 years (SD ± 8.2). Mean age was 54.7 years (± 8.9). All patients were treated by enzyme replacement therapy (ERT).

Results:
Patient survival was 91.7% at 5 years and 82% at 10 years. Two patients died of cardiovascular causes and one from cancer. Graft survival was 93.4% at 5 years and 82% at 10 years. The mean glomerular filtration rate estimated by the CKD-EPI formula was 63.2 ml/min/1,73 m^2 (± 24.3), mean urinary protein to creatinine ratio was 0.28 g/g (± 0.16).
Immunosuppressive treatment comprised tacrolimus (70%), cyclosporine A (20%), mycophenolate mofetil (64%), azathioprine (18%) and corticosteroids (50%).
Mean duration of ERT post-transplantation was of 14.4 years (SD± 6,3).
All females who needed kidney transplant had unfavorable skewed X chromosome inactivation.
Amenable mutations were present in 33% of patients.
One patient switched to migalastat after 13 years of ERT.





Conclusion: Kidney transplant in association with continued ERT appears generally favorable in patients with FD with good patient and graft survival, stabilized renal function and absence of proteinuria.