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The prevalence of fabry disease in hemodialysis patients of Jeju island

So mi KIM[1], Chang hyun PARK[1], Eun kyoung LEE[1], Hyun woo KIM[2], Sun hyung KIM[3], Ji HYUN[1], *Jong tae CHO[1]

[1]Division of Nephrology, Department of Internal Med, Dankook University Hospital, Dankook University, College of medicine, Korea,South, [2]Division of nephrology, Department of internal medicine, Jeju National University Hospital, Jeju National University School of Medicine, Korea,South, [3]Department of Laboratory Medicine, Jeju National University Hospital, Jeju National University School of Medicine, Korea,South

Objectives : Fabry disease (FD) is an X-linked genetic disorder, caused by mutation in the GLA gene which encodes lysosomal enzyme, a-galactosidase A (α -Gal A). The deficiency of α-Gal A could cause a renal failure, but its diagnosis is completely missed at times. Although the prevalence of FD in dialysis patients are known to between 0.16 and 1.2%, the higher prevalence was expected in island area in terms of genetic disorder. Therefore, we try to investigate the prevalence of FD in hemodialysis patients of Jeju island.

Methods : A total of 9 artificial kidney unit participated in the study. We measured plasma α-Gal A activity before starting hemodialysis. In patients with positive result, we analyzed the GLA gene, under patient's agreement.

Results : A total of 663 patients with hemodialysis were enrolled in the study. The mean age of patients was 57 year, and the male was 64 %. Among them, the 39 (5%) patients showed the low α-Gal A activity with < 0.45 nmol/min/mg protein. The gene analysis was performed in all patients with positive result. In genetic analysis, the definite GLA mutation was not found. But E66Q mutation which is debating functional variant or FD, was found in 3 female patients. Although the presumptive clinical cause of renal failure of them was DM in 1, unknown etiology in 2, the kidney biopsy was not performed.

Conclusions : Although the prevalence of FD was 0%, the E66Q mutation showed 0.04 % in hemodialysis patient of Jeju island. More accurate diagnostic tool for FD and follow-up of prognosis in patients with E66Q mutation were needed.

Keywords : fabry disease, Hemodialysis, E66Q mutation