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GENETIC DISEASES AND MOLECULAR GENETICS

SP031 SCREENING FABRY DISEASE IN PATIENTS WITH CHRONIC KIDNEY DISEASE WITHOUT RENAL REPLACEMENT THERAPY: PRELIMINARY RESULTS OF A MULTICENTER STUDY

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Introduction and Aims: Fabry's disease (FD) is an X-linked inherited,rare,progressive, multisystem disorder of glycosphingolipid metabolism affecting multiple organs. Deficient activity of α-galactosidase A enzyme gives rise to accumulation of glycosphingolipids particularly globotriaosylceramide.The data concerning the prevalance of the disesase in CKD other than ESRD is lacking.We aimed to determine the prevalence of FD in CKD.
Methods: CKD patients other than ESRD were screened for α-galactosidase A deficiency.Dried blood samples were used to analyse enzyme activity and genetic testing when required. All tests were performed by ARCHIMED Life Science GmbH Laboratories Vienna,Austria.Study protocol was approved by ethical committee.
Results: A total of 736 patients were screened in 5 centers. Male ratio was 52,45%. Mean age, serum creatinine level, proteinuria level and α-galactosidase A activity were 61.7±14.3 years, 2.0±1.0 mg/dL, 1.4±2.6 g/day, 2.31±2.42 μmol/L/h repectively.80 patients had low levels of alfa galactosidase enzyme (<1.2 μmol/L/h). 3 of them had mutations in the α-galactosidase A gene specific for FD.The prevalance of FD mutation in CKD other than ESRD was 0.4%. Mutation detected patients were clinically evaluated(Table).The 3rd patient rejected further evaluation.
Conclusions: The reported FD prevalence changes between 1/40,000-117,000 in normal population and 2/1000 in hemodialysis patients. The prevalence of FD was 3/736.FD is speculated to cause end stage renal disease requiring renal replacement therapy at 5th decades. However;our patients had a better renal function considering their ages. It is not clear whether this difference should be attributed to the racial, ethnic or regional differences,or genetic variations.

SP031 Index cases

	Case I	Case II
Age/Gender	70 years/M	83 years/M
Clinical manifestations	Cornea verticillata Cataract Type 2 DM Coronary artery disease LVH Sensorineural hearing loss Carpal tunnel syndrome Osteoporosis Chronic lacunar infarcts Renal cortical exophytic cysts	Cornea verticillata Hypertension Coronary artery disease LVH Sympathetic autonomic dysfunction
Previous Diagnosis	Diabetic nephropathy	Hypertensive nephropathy
Symptoms	Hearing loss	Intolerance to heat Sweating disorder
Family history	Premature coronary heart disease	-
Serum creatinine	1.95 mg/dL	1.81 mg/dL
Proteinuria	50 mg/dL	58.75 mg/dL
α-galactosidase A	0.5 μmol/l/h	0.3 μmol/l/h
Mutations	c.[937G>T] p.[D313Y])	c.[427G>A] p.[A143T]
Kidney Biopsy	Focal Segmental Glomerulosclerosis	Not accepted