

Screening for Fabry disease among 619 hemodialysis patients in Saudi Arabia

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Abstract

The aim of this study was to determine the prevalence of FD among Saudi patients on hemodialysis.

Methods: This prospective study was conducted in three major hospitals in Saudi Arabia. All adult patients (> 18 years old) attending the dialysis unit who have end-stage renal disease (ESRD) and on hemodialysis were included. Known patients with FD and those who refused to participate in the study were excluded. All eligible patients were screened for FD using Dry Blood Spot (DBS) for α -Gal A. A positive DBS (enzyme activity < 40%) was followed by another confirmatory enzyme assay if the second DBS sample was also positive (enzyme activity < 40%), a Sanger sequencing of the GLA gene was performed.

Results: A total of 619 patients with ESRD and on hemodialysis were screened for FD using DBT for α -Gal A enzyme level. Enzymatic activity was below 40% in 11 samples. On retesting, three females had < 20% enzymatic activity suggesting FD. Sanger sequencing of these three females showed the variant c[1055C>G] (p.[Ala352Gly]) confirming the diagnosis of FD. Family screening of one of these three patients revealed one asymptomatic female carrying the same mutation.

Conclusion: The prevalence of FD in this cohort was 4.8 per 1000 patients. Screening of Fabry patients with

ESRD seems to be a cost-effective strategy. Furthermore, screening of relatives of patients identified by screening enhances this screening strategy.

Biography

Dr Salwa Alhemyadi graduated from the University of Taiz 2010 and went on study for the Saudi Board in Internal Medicine(KSA)

In 2013 at Prince Sultan Medical Military Centre ,Riyadh. She continued her academic career with Fellowship in Nephrology from Saudi Fellowship program in 2019. She attained MRCP(UK) in 2021. She had been organizing the resident's Academic Activities for two years in Prince Sultan Military Medical City department of medicine and was the chief organizer of the College Students' Week. Deputy of the research committee in Nephrology Department at PSMC since 2019. And she has been a member of Institutional and Academic Accreditation at PSMC 2020.