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## Screening for fabry disease among 619 hemodialysis patients in Saudi Arabia

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**Objectives:**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  $\alpha$ -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  $\alpha$ -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 48 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.

### References

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### Biography

He was experienced physician trained in the discipline of internal medicine with well-rounded experience in acute and in-patient care i have had the opportunity to provide medical care for various communities in the past during my training of Saudi board of internal medicine and as a fellow of Nephrology, I have an opportunity to teach paramedical staff, undergraduate medical students and interns Deputy of the research committee in Nephrology department at PSMC since 2019 I am keen in research and looking forward to a challenging and professionally competent environment in which I can learn and practice evidence based medicine.

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