Prevalence and Genetic Variability of Inborn Errors of Metabolism in Bahrain

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Objective: To evaluate the prevalence and the pattern of inborn errors of metabolism (IEMs), mutation spectrum and outcome.

Design: A Retrospective Analysis.

Setting: Salmaniya Medical Complex, Bahrain.

Methods: A seventeen-year retrospective study of patients diagnosed with IEMs was performed. The following were documented: IEMs categories, age, sex, origin, and consanguinity. In addition, molecular genetic result and outcome were documented.

Result: One hundred eighty-eight patients with IEM were included in the study. One hundred seventy-seven (94.1%) were consanguineous. Ninety-three (49.5%) patients were identified to have small molecules disorders, while 95 (50.5%) were large molecules disorders. Mutation analysis was done on 124 (66%) patients, and novel mutations were detected in 72 (38.3%). The overall death rate was 41.5%.

Conclusion: The high rate of IEMs in Bahrain warrants the need for implementing a national neonatal screening program to evaluate the exact burden of these disorders to reduce mortality and morbidity by early management. The detection of molecular genetic mutations in our population will help a prevention program through preimplantation genetic diagnosis; in addition, the presence of a large number of novel mutations will invariably help identifying the genetic variability in this region.

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