Conflict of Interest: None declared.

EP20.024 Epidemiological estimates of Autosomal Dominant Polycystic Kidney Disease (ADPKD) diagnosed cases in Malta

Natalie Ciantar[1], *Emanuel Farrugia*[2], *Jean Calleja Agius*[1], *Christopher Barbara*[3], *Fiona Sammut*[4], *Graziella Zahra*[5], *Edith Said*[1,6]

[1]*University of Malta, Faculty of Medicine and Surgery, Anatomy and Cell Biology Department, Msida, Malta;* [2]*Nephrology and General Medicine Division, Department of Medicine, Mater Dei Hospital, Malta, Malta;* [3]*Mater Dei Hospital, Pathology Department, Msida, Malta;* [4]*University of Malta, Department of Statistics and Operations Research, Msida, Malta;* [5]*Mater Dei Hospital, Molecular Diagnostics Laboratory, Pathology Department, Msida, Malta;* [6]*Mater Dei Hospital, Section of Medical Genetics, Pathology Department, Msida, Malta.*

Background/Objectives: Estimating the prevalence and incidence rates of ADPKD is challenging due to the highly variable expression of the disease. Here we investigate the epidemiology among ADPKD diagnosed cases in an isolated small island of Malta.

Methods: A total of 59 unrelated cases (38 males, 21 females) over 18 years with clinical features of ADPKD were studied. A detailed three-generation pedigree was generated where possible.

Results: The estimated point prevalence of ADPKD for the Maltese adult population was 2.1 (95% CI 1.7–2.5) per 10,000 inhabitants and the estimated annual incidence rate of ADPKD was 1.6 per 100,000 person-years (95% CI 0.78–3.1). The annual incidence rate of ESRD in ADPKD patients was 0.78 (95% CI 0.21–1.98) per 100,000 person-years with a male-to-female ratio of 2.8:1. Moreover the crude percentage of clinically diagnosed ADPKD adult patients on renal replacement therapy (RRT) was 9.6%. Data on the Maltese population was retrieved from the National Statistics Office (NSO).

Conclusion: Labelled as a rare disease, ADPKD is an underestimated cause of ESRD. The high percentage of patients on RRT stimulates the need of further studies to delay ESRD. Moreover, this study highlights the need of genetic testing in order to confirm the genetic cause of renal cysts.

References: N/A

Grants: This research is being funded by the LifeCycle Malta Foundation through the University of Malta Research, Innovation