P19.001.A Genotype of autosomal dominant polycystic kidney disease using a custom gene panel in Malta

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Introduction: Polycystic Kidney Disease (PKD) is the commonest form of inherited kidney disorder. The disease can be inherited in an autosomal dominant (AD) or autosomal recessive (AR) manner. Autosomal dominant polycystic kidney disease (ADPKD) is characterized by the development of multiple renal cysts causing renal enlargement and end-stage renal disease (ESRD) in 50% of patients by 60 years of age.

Methodology: A total of 49 unrelated patients with clinical features of ADPKD were studied using a customized gene panel for genes associated with polycystic kidney disease (PKD) using next generation sequencing (NGS). The genes studied were PKD1, PKD2, GANAB, DNAJB11, PKHD1 and DZIP1L.

Results: Bioinformatic analysis has identified five different pathogenic variants in fifteen subjects. Two different novel frameshift pathogenic variants and three other previously reported frameshift, nonsense and splicing pathogenic variants were identified. The novel pathogenic variants, c.4651delC (p.Leu1551SerfsTer12) and c.1645dupG (p.Glu549GlyfsTer24) were identified in PKD1 and PKD2 respectively. Other variants of unknown clinical significance have been identified through sequencing.

Conclusion: This study helps to show that a customized gene panel is the method of choice for studying patients with ADPKD and further emphasizes the genetic variability of this condition. Further functional analysis of these novel variants is necessary to understand the mechanism underlying the development of ADPKD in the Maltese population. This research is being funded by the LifeCycle Malta Foundation through the University of Malta Research, Innovation & Development Trust (RIDT) and by the Tertiary Education Scholarship Scheme.

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