# Article information:

The genetic landscape of autosomal dominant polycystic kidney disease in Kuwait - PMC
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9900584/>

# Article summary:

1. This study provides a comprehensive description of the pathogenic variants linked to autosomal dominant polycystic kidney disease (ADPKD) in the Kuwaiti population.

2. A total of 29 ADPKD pathogenic mutations were identified from 36 families, with 28/36 (77.8%) families having pathogenic mutations in PKD1 and 1/36 (2.8%) having a pathogenic variant in the gene,IFT140.

3. Clinical analysis indicated that genetically unresolved ADPKD cases had no apparent association between kidney volume and age.

# Article rating:

May be slightly imbalanced: The article presents the information in a generally reliable way, but there are minor points of consideration that could be explored further or claims that are not fully backed by appropriate evidence. Some perspectives may also be omitted, and you are encouraged to use the research topics section to explore the topic further.

# Article analysis:

The article is generally reliable and trustworthy, as it is based on a study conducted by a team of researchers from multiple institutions in Kuwait, including Mubarak Al-Kabeer Hospital, Kuwait University and the Kuwait Institute for Medical Specialization. The study was also reviewed and approved by the Ministry of Health Research Ethics Committee, which adds to its credibility. Furthermore, the article provides detailed information about the methods used in the study, such as targeted next-generation sequencing (tNGS), long-range polymerase chain reaction (LR-PCR), Sanger sequencing and multiplex ligation-dependent probe amplification (MLPA). This indicates that the authors have taken great care to ensure accuracy and reliability of their results.

However, there are some potential biases that should be noted when considering this article's trustworthiness and reliability. Firstly, although 126 patients were recruited into the study, only 36 families were included in the genetic analysis due to limited resources available for genetic testing at Mubarak Al-Kabeer Hospital. This could lead to an underestimation of genetic heterogeneity underlying ADPKD in Kuwaiti population as well as an underestimation of intrafamilial phenotypic variation associated with ADPKD mutations. Secondly, although clinical evaluation was conducted through renal function testing and ultrasonographic kidney volume analysis, other factors such as extrarenal manifestations such as polycystic liver disease (PLD), cerebral aneurysms and cardiovascular abnormalities were not considered or discussed in detail in this article. Finally, while 28/36 (77.8%) families had pathogenic mutations in PKD1 identified by tNGS panel analysis, 7% of ADPKD cases remain genetically unresolved or are due to pathogenic variants in other genes such as GANAB, DNAJB11 and ALG9 which were not explored or discussed further in this article due to limited

# Topics for further research:

* Polycystic Liver Disease (PLD)
* Cerebral Aneurysms
* Cardiovascular Abnormalities
* GANAB Gene Mutations
* DNAJB11 Gene Mutations
* ALG9 Gene Mutations

# Report location:

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