Does whole genome sequencing clarify the cause of unexplained chronic kidney disease in Hong Kong adults?

ISN INTERNATIONAL SOCIETY OF NEPHROLOGY

Prospective cohort study
October 2022 to June 2024

131 adults with chronic kidney disease of unexplained cause

Whole genome sequencing focused on 637 kidney disease genes

Variant classification based on American College of Medical Genetics criteria

92% Chinese

Mean age 35 years

21% presented with kidney failure

36% with family history

Pathogenic or likely pathogenic variants in 13 patients

Diagnostic yield: 10%

One third of variants were novel

Most frequent genes: *COL4A3, COL4A4, COL4A5*

Alport spectrum disorders represented **77%** of positive cases

Positive results strongly associated with family history

p < 0.001

Conclusion: Monogenic etiology was identified in 10% of adults with chronic kidney disease of unexplained cause by whole genome sequencing. Alport spectrum disorders were the leading diagnoses.

Becky Mingyao Ma, Shirley Pik Ying Hue, et al

Visual abstract by **Cristina Popa**
𝕏 @nephroseeker

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