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Resumenes de las comunicaciones presentadas al 5° Congreso de la Sociedad Gallega de Nefrologia. Comunicaciones orales

Development of strategies for genetic diagnosis of hereditary glomerulopathies, tubulopathies and cystic kidney diseases by the sequencing sets of genes

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Introduction: Accurate clinical diagnosis in certain renal pathologies, especially glomerular and tubular disease, has proven to be difficult, as different pathologies may appear as clinical phenocopies. Genetic studies have the advantage of ensuring an accurate diagnosis and anticipate the disease. The implementation of NGS technology into routine genetic diagnostic practices allows the screening of large sets of genes in a single test.

Methods: Our group, based on the clinical classification, generated different panels for the sequence of renal disease genes in single tests: (1) panel for cystic kidney disease (up to 72 genes); (2) panel for glomerular disease (26 genes), and (3) panel for tubular disease (36 genes). Also, our group solved one of the limitations of conventional pre-designed NGS kits for target enrichment in regions with high homology pseudogenes (such as the *PKD1* gene) by developing particular primers to amplify specifically the replicated region of *PKD1* gene (exons 1-34).

Results: Our technology proved to be more efficient compared to other technologies used in the diagnostic routine. By analyzing a cohort of 291 families with PKD clinical diagnosis, we identified the causal mutation in 88% ($n = 255$) of the families. In 94% ($n = 240$) of these cases the clinical and genetic diagnosis were concordant. Of the 71 patients with a clinical diagnosis of glomerular disease and 31 with tubular disease subjected to genetic analysis, we identified the causal mutation in 62% ($n = 44$) and 52% ($n = 16$) of the cases, respectively. The concordance between genetic and clinical diagnosis was 66% ($n = 29$) for the glomerular cohort and 69% ($n = 11$) for tubular cohort. Most cases of misdiagnosis were associated with syndromic diseases with very similar phenotypes, such as Gitelman and Bartter syndromes.

Conclusions: The strategy of grouping genes by phenotype for genetic testing probed to be efficient in finding the causal mutation. Our results make clear the need of a genetic test to avoid misdiagnosis of certain renal pathologies.

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Enfermedad renal crónica (ERC) y estatus socioeconómico (SES). Estudio Epidemiología de la enfermedad renal crónica en España (EPIRCE)

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Introducción: Las desigualdades socioeconómicas son un problema clave en salud pública, pero los factores de riesgo cardiovascular (FRCV) convencionales solo explican parcialmente la relación entre SES y nivel de salud. En ERC es posible