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Clinical Genetic Testing in Adults with Kidney Disease

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Introduction

Cystic kidney disease is a common cause of end-stage renal disease in both children and adults. The two main types of monogenic cystic kidney diseases are autosomal dominant polycystic kidney disease (ADPKD) and autosomal recessive polycystic kidney disease (ARPKD). ADPKD is a common disease that primarily affects adults, whereas ARPKD is a rarer and often more severe form of polycystic kidney disease (PKD) that typically manifests during pregnancy or early childhood. Cell biological and clinical research approaches have increased our understanding of the pathogenesis of ADPKD and ARPKD, revealing some mechanistic overlap. Reduced PKD protein 'dosage' is thought to disrupt cell homeostasis and converging signalling pathways such as Ca^{2+}, cAMP, mechanistic target of rapamycin, WNT and vascular endothelial growth factor. Genetic diagnosis may benefit families and improve patient clinical management, which may be enhanced further by emerging therapeutic options. Many important questions about the pathogenesis of PKD remain unanswered. This Primer provides an overview of current knowledge about Parkinson's disease and its treatment [1].

Description

Between July 2017 and September 2018, patients were referred to one of four RGC teams based at tertiary hospitals in Melbourne, Australia and data was collected until October 2019. All reporting followed the guidelines for Strengthening the Reporting of Observational Studies in Epidemiology. Patients were discussed for study inclusion by the RGC team after being referred by their treating nephrologist. Patients were recruited if their clinical presentation fit a single cause (e.g., glomerular, tubulointerstitial, or cystic renal disease) and prioritised if they met one of the following criteria: a family history of renal disease, syndromic features, or disease onset in childhood [2].

Larger copy number variants (CNVs), some of which cause inherited kidney diseases, are not easily detected by MPS-based gene panels or ES. To detect these large CNVs from gene panel or ES data, sophisticated bioinformatic tools are required, which are not yet routinely used in all diagnostic laboratories. In many labs, the microarray-based technique [comparative genomic hybridization (CGH) or single-nucleotide polymorphism (SNP) arrays] is still the preferred methodology for routine diagnostics of large CNVs. Another method for detecting CNVs is multiplex ligation-dependent probe amplification (MLPA). Furthermore, due to the complexity of the involved genomic region, several important disease-causing variants, such as pathogenic variants in PKD1, can be missed using MPS-based panels or ES [3].

FSGS should also be distinguished from focal segmental scarring that develops as a result of postinflammatory scarring of necrotizing or proliferative lesions in immune-mediated GN (e.g., IgA nephropathy, ANCA-associated GN and lupus nephritis). A significant proportion of segmental sclerotic lesions in IgA nephropathy may represent podocyte injury with mechanisms similar to those seen in FSGS, in addition to nonspecific scarring [4].

Many families face a significant psychological burden as a result of their child's disease status15. Parents of at-risk children should be informed by appropriately trained personnel about the possibilities, limitations and consequences of genetic and clinical testing for their children. They should be aware that screening exams do not always produce definitive results and that a normal kidney ultrasonography scan has a low negative predictive value in children. Offering diagnostic screening does not imply that it is recommended, but rather gives parents the opportunity to make an informed decision. Because time for non-directional genetic counselling in general practise and adult nephrology may be limited, a referral to a geneticist or specialised ADPKD clinic may be required [5].

Conclusion

In conditions associated with hyperfiltration, glomerular capillary hypertension and glomerular hypertrophy, maladaptive FSGS results from a mismatch between glomerular load and glomerular capacity. The podocytes are subjected to significant mechanical strain as a result of hyperfiltration and glomerular capillary hypertension. Shear stress caused by increased filtrate flow through the filtration slits and over their apical surface is extremely damaging to podocytes. Glomerular hypertrophy forces podocytes to cover a larger filtration surface. However, the foot processes' ability to exhibit hypertrophic growth is limited. The podocytes may be unable to maintain a normal foot process pattern, increasing local shear stress even further. When the rheologic stress becomes unsustainable, the FPE process is initiated to redistribute the mechanical forces and reduce the stress

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Conflict of Interest

There are no conflicts of interest by author.

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