Genetic Science Spotlight

Harvard Medical School Recommends Molecular Genetic Diagnosis for all Early-Onset Chronic Kidney Diseases (CKD) Cases

It is widely known that the etiology of early-onset (below 25yr) chronic kidney diseases (CKD) is primarily different from adult-onset CKD. The recent advent of next-generation sequencing (NGS) has provided researchers and healthcare professionals with one of the most effective tools in uncovering the genetic predispositions of CKD in children. Recent studies have confirmed that a substantial proportion of early-onset CKD cases have a monogenic cause. To this date, over 200 genes have been identified as answerable to monogenic forms of CKD if mutated. Indication-driven gene panel analysis, as well as whole exome sequencing (WES) have both been effective in unravelling CKD etiology. Given the inherent nature of comprehensive coverage, WES has also provided novel gene candidates contributing to the condition that had not been previously recognized. While molecular genetic diagnostics enables prenatal testing and may have prognostic and sometimes therapeutic implications, it is advisable for all early-onset CKD subjects to undergo molecular diagnostic tests.

https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5202482/

You are welcome to contact us for more information!

臉書 Facebook
Le GENE 領先基因

香港領先基因

領先基因
www.le-gene.com

Le GENE Lifestyle

LeGENE Personal Genetic Centre
Suite 1411-1413 Central Building, 1 Pedder Street, Central HK
www.le-gene.com Tel: 2327 1411 Fax: 2327 4108 Email: cs@le-gene.com