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Promoting Sustainable Kidney Health: The Asia-Pacific and Beyond

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Extrarenal clinical features are reported for most genes implicated in genetic kidney disease

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Objectives : Genetic kidney disease is typically suspected where there is a positive family history or extrarenal features. This study examined how often these criteria might be expected in genetic kidney disease.
Methods : Two hundred and fifty-five genes from the Genomics England 'green lists' for Congenital Kidney Anomalies of the Kidney and Urinary Tract (CAKUT)(n=57), the Ciliopathies and Cystic kidney diseases (n=90), Haematuria (n=5), Renal proteinuria (n=55) and the Renal Tubulopathies (n=48) were examined for mode of inheritance and, in OMIM, for reported clinical features in different systems (neurological, cardiac etc) that would be obvious on history or physical examination.
Results : Biallelic inheritance alone was recorded for 148 of the 248 genes (60%) with an OMIM entry. Extrarenal features were associated with 221 genes (89%) including those for Haematuria (5, 100%), Renal ciliopathies (86, 97%), CAKUT (52, 91%), Renal tubulopathies (41, 85%) and Proteinuric renal disease (37, 76%). The median number of affected systems was 4 (range 0 – 10). More extrarenal features were associated with CAKUT (4, 0 – 10) and the Ciliopathies (5, 0 – 9) than for Haematuria (2, 2-5), Proteinuria (3, 0-7) and the Tubulopathies (3, 0-7) ($p<0.00001$). The commonest systems affected were growth and musculoskeletal (164, 66%), neurological (147, 59%), and ocular (133, 54%).
Conclusions : The biallelic inheritance means that a family history is absent for many genes affected in genetic kidney disease. While extrarenal features are reported with most genes, extrarenal anomalies are more common in CAKUT and the ciliopathies. However data are limited on how often features occur in affected individuals.