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P329 -Experience of genomics through 100,000 Genomes Project for paediatric renal rare disease patients at a specialised hospital.

Ms Maria Kokocinska[1], Dr Larissa Kerecuk
[1]*Birmingham Children's Hospital, Birmingham, United Kingdom*

Background

Paediatric renal rare diseases presenting to mainstream clinicians frequently have a genetic component, are poorly defined producing significant burden of morbidity and mortality. These conditions can be kidney-specific but the majority are multisystemic with highly variable phenotype-genotype relationships. The emergence and advancements in genomic and computational technologies present an opportunity for a greater insight in these conditions at molecular level, new gene discovery and potential for development of targeted therapies, precision medicine and empowering decision making in healthcare. Genomic information obtained from next generation sequencing analysis is already utilized in clinical practice for diagnosis of some rare renal disorders such as Bartter's syndrome (1), steroid-resistant nephrotic syndrome (2), autosomal dominant polycystic kidney disease (3), Alport's syndrome (4) to name a few.

Objective

The 100,000 Genomes Project (100kGP) aimed to implement genomics medicine into the mainstream NHS as part of patients' routine care (5,6). The findings from the project could realise the potential of genomics for some renal patients as certain renal rare conditions were included in the project's eligibility criteria (Table 3). The results will be analysed by Genomics England Clinical Interpretation Partnership (GeCIP) renal domain with the objective of identification of new genes causing kidney disease and utilizing these discoveries in genomic testing.

Local approach

Our centre started recruiting renal rare disease patients to 100kGP in June 2015. The genomics team at our centre has developed a streamlined process which incorporated recruitment to project at the same time as patient's appointment. Thereby, offering all identified, eligible patients the chance to be recruited without increasing the work burden for the clinicians or the number of appointments for the family.

Results

Our centre was the highest single contributor of paediatric renal patients (570) in the UK outside London. Renal patients formed 40% of all those that took part in the 100kGP. There were 62% males and 38% females. All the ethnic and age groups were well represented (Table 1 and 2). Table 3 shows the distribution of conditions that recruited patients were affected with, 75% of those had CAKUT.

Conclusion

The approach used at our centre to recruit to the 100kGP shows that renal patients affected by rare renal disorders and their families are very keen to take part in genomics research. It is hoped that with clinical and

scientific expertise employed in data analysis and validation of the project's results will lead to new diagnosis, personalised management and improved outcomes for the renal patient and the NHS.