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Data Derived Disease Diagnostics

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Abstract. Microscopic haematuria is a common incidental finding in childhood which often resolves on its own. However, it can be an early marker of genetic kidney disease. It is best practice to repeat testing to ensure resolution. Using data from the electronic medical record, we set out to find children without follow up, offer testing, and look for genetic kidney disease.

Keywords. Genetics; Kidney; Paediatrics; Haematuria; Health Analytics

1. Introduction

Microscopic haematuria occurs in the paediatric population at a rate of 0.3-4% [1,2]. It is often seen in the context of a fever or viral illness. Persistent microscopic haematuria is estimated to occur at approximately 1% [1]. In many children, this will resolve spontaneously. In a small number of children, it can be an early marker of genetic kidney disease. Alport syndrome is a heterogeneous group of genetic disorders resulting in renal failure, a progressive and irreversible hearing loss and eye problems [3]. If detected at an early stage, a medication can be commenced which delays the onset of renal failure by 15-20 years [4]. We sought to use data collected through the electronic medical record to identify children without follow up and offer appropriate testing.

2. Methods

We carried out a clinical audit of the electronic medical record to identify children between the ages of 0 and 15 with microscopic haematuria on urinalysis without a subsequent normal result. Our exclusions included Alport Syndrome in the problem list, Nephrology patients and deceased patients. We found 5531 children. 1959 had co-existing pyuria and were deemed likely infective in origin and excluded.

We stratified the remaining 3572 into 3 groups based on quantitative values of haematuria and focused on the group with >100 RBCs/ml (1247). We used the bulk communication and bulk order function within the electronic medical record to link a patient letter and pathology slip. Patients with persistent microscopic haematuria were referred to the Renal Genetic Clinic and offered genomic testing.

3. Results

There was a fault in the bulk communication function which resulted in patients not receiving their information letter and just receiving the pathology slip. This caused

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significant confusion and an increased number of calls to the hospital but no adverse clinical events. The processes to correct this were laborious and highly manual. We received repeat tests in 12.5% of patients. 8 children were found to have persistent microscopic haematuria. 7 have had genomic testing to date. 2 children have been diagnosed with Alport syndrome and both look likely to have family members who may also be affected. 3 children have a negative 5 gene Alport panel but have other clinically suspicious signs and have been sent for further genomic analysis. 2 children have no genomic abnormality identified.

4. Conclusions

There is huge potential to use the incidental data gathered in healthcare to allow us to be proactive in paediatric healthcare. Detecting genetic kidney disease at an early stage allows us to instigate treatment and enables reproductive planning for parents [5,6]. There are some barriers that need to be addressed to support this type of program. The reliance on postal communication within healthcare is archaic and inefficient[7]. We are currently testing the bulk communication function in the hope of re-attempting contact and seeking ethics approval to trial different modes of communication to improve the response rate including text, phone, patient portal and another letter. The interface between the hospital electronic medical record system and external pathology providers is also problematic resulting in delayed processing of external results. The prevalence of genetic kidney disease amongst children with microscopic haematuria is unknown. We can better understand this prevalence and improve rates of diagnosis and care if we can overcome the logistical challenges.Acknowledgements
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