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Short Communication

A Rare Nephrology Case Report-Gross Hematuria As A Presenting Feature Of Autosomal Dominant Polycystic Kidney Disease (Adpckd)

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Abstract

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is the most common hereditary cause of renal failure worldwide. Its hallmark is progressive cyst development in association with the renal parenchyma and other extrarenal tissues such as the liver, pancreatic, heart and arachnoid membranes. Although cyst development can start as early as childhood, and even in utero, severe symptoms seldomly develop until the third or fourth decade of life, highlighting its insidious presentation

Kew Words: autosomal dominant; polycystic kidney disease; haemodialysis

Introduction

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is the most common hereditary cause of renal failure worldwide. Its hallmark is progressive cyst development in association with the renal parenchyma and other extrarenal tissues such as the liver, pancreatic, heart and arachnoid membranes. Although cyst development can start as early as childhood, and even in utero, severe symptoms seldomly develop until the third or fourth decade of life, highlighting its insidious presentation. [1] Systemic features include the development of hepatic cysts, colonic diverticula, intracranial aneurysms, and valvular heart lesions highlighting the diversity of involvement of ADPKD. [2] Moreover, hypertension is present as a common manifestation in the great majority of ADPKD patients before the development of overt renal function decline. This sequence may reflect that each episode of progressive kidney enlargement represents an independent risk factor for end-stage renal disease. [3] ADPKD usually has an insidious onset and clinical heterogeneity that could delay disease recognition. Additionally, it can be challenging to diagnose as it initially is asymptomatic and there is not a specific diagnostic test, the reason it may often be overlooked or underdiagnosed.

This principle very strongly suggests that medical professionals should, when treating patients who have a family history of the disease, establish and maintain a high index of suspicion and integrate routine screening procedures into clinical practice. By identifying ADPKD at an early stage, early intervention and better management outcomes can be achieved. This can be achieved through raising awareness and expanding screening efforts. The significance of early identification and treatment of ADPKD is shown by this thorough comprehension. To lessen its effects and enhance long-term results for those who are impacted, early detection must be combined with prompt intervention and a multimodal approach to treatment.

Clinical presentation-

The case pertains to a 50-year-old Indian male patient with a background of hypertension. It was established that his mother was diagnosed with ADPCKD at an earlier time. At the time of presentation, he was undergoing treatment for hypertension for the past 3 years. His therapeutic regimen comprised of a combination drug of metoprolol and amlodipine, sodium bicarbonate and a syrup formulation of disodium hydrogen citrate as a urine alkaliser. His latest visit to the hospital was concerning an episode of painless, reddish discolouration of urine. This was the first and only such episode. The patient's admission to Department of General Medicine, L.G. Hospital, Ahmedabad facilitated a complete evaluation of his clinical status.

Diagnosis

Clinical investigations upon admission encompassed haemoglobin levels (14 mg/dL), platelet count (214,000/mm³) and total leukocyte count (9,550/mm³) with a neutrophilic predominance (82%). Renal function tests were carried out, establishing a low-normal serum sodium (132 mg/dL) alongside elevated creatinine (6.2mg/dl) and urea levels (128 mg/dl). The urinary albumin-to-creatinine ratio was astoundingly high (157.78), reflecting the gravity of renal filtration inefficiencies. Ultrasound revealed markedly enlarged kidneys with lobules, where normal kidney tissue is bilaterally replaced by multiple cystic shadows of varying sizes and echogenicity. Urinalysis showed slightly red urine, with 2+ proteinuria, 4+ haematuria, and 80-85 RBCs per high power field. These findings collectively support a primary diagnosis of autosomal dominant polycystic kidney disease. Considering the possibility of concurrent extrarenal manifestations, various other investigations were also conducted. An echocardiogram was carried out to assess valvular function, demonstrating normal valvular efficiency. Abdominal as well as neurological imaging was performed to determine the presence of any cystic components and aneurysms, respectively, which reflected normal anatomical structure. Collectively, these findings converged to make the primary diagnosis of ADPCKD presenting with hypertension and renal failure

Treatment

The patient's therapeutic regimen underwent meticulous adjustment to address and preserve his kidney function with careful regard to his ADPKCD diagnosis. The amended strategy encompassed the introduction of intravenous antibiotics to mitigate the risk of potential infections, the optimisation of his previous medication to ameliorate the symptoms brought on by the progression of renal failure, and the reinstatement of his antihypertensive medications, with comprehensive counselling over the importance of compliance to the prescribed medication, avoidance of alcohol and commonly available nephrotoxic medications and a strong recommendation to undergo periodic imaging to monitor the course of ADPCKD.

Discussion

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is a significant hereditary condition affecting the kidneys, characterized by the formation of numerous cysts. Early diagnosis is pivotal in managing this condition effectively. [4] Symptoms often manifest in various forms, with haemorrhage within the cyst, haematuria (blood in urine), and hypertension being common indicators. Cyst haemorrhage, occurring in about 60% of cases, can present as either gross or microscopic haematuria, particularly if the cyst connects with the collecting system. The sudden onset of hypertension in at-risk individuals warrants immediate attention and thorough evaluation. [5] Ultrasonography stands out as the primary diagnostic tool due to its accessibility and cost-effectiveness. Additionally, extra-renal complications, such as cardiovascular issues, demand careful screening, including blood pressure monitoring, echocardiography, and screening for intracranial aneurysms (ICAs) in high-risk individuals. [6]

Management of ADPKD involves a multi-faceted approach. Lifestyle modifications, including sodium and calorie restriction, hydration, and dyslipidaemia management, play a crucial role regardless of disease progression. Medications like angiotensin-converting enzyme inhibitors and angiotensin receptor blockers are frontline therapies for hypertension control. Tolvaptan, a promising drug, offers a disease-modifying treatment for patients at risk of rapid kidney function decline. It is a vasopressin receptor antagonist that specifically targets V2 receptors and disrupts the reabsorption of free water, influencing osmolarity as well as the expression of aquaporins, which are transmembrane osmoregulatory proteins, leading to effective reduction in cyst growth rate and total kidney volume. Certain studies also propose the use of mTOR inhibitors for controlling ADPKD, considering the role of polycystin-1 and polycystin-2 malfunctions and mutations activating mTOR and increasing cell growth. Rapamycin is a popular mTOR inhibitor which can be used in this regard. [7] For those requiring renal replacement therapy, transplantation remains the preferred option when feasible. Haemodialysis and peritoneal dialysis serve as suitable modalities when transplantation is not immediately available. Regular monitoring, early intervention, and a comprehensive treatment strategy are essential in managing ADPKD effectively, ultimately improving patient outcomes and quality of life. [8]

Conclusion

The insights gained from this case study aptly underscore the critical need for standardizing the management approach to hypertension concurrent with haematuria, ensuring comprehensive consideration of all potential aetiologies, including congenital or hereditary conditions, especially in cases with a strong familial predisposition. The array of symptoms stemming from a missed diagnosis or inadequate management of Autosomal Dominant Polycystic Kidney Disease (ADPCKD) encompasses a myriad of factors, encompassing individual behaviours and environmental influences. It is crucial to emphasize that this case study strongly advocates for the thorough evaluation of hypertensive patients presenting with haematuria to identify renal parenchymal pathologies. Nonetheless, this evaluation should not be limited solely to renal parenchymal aetiologies, owing to a distinct possibility of concurrent extrarenal manifestations of ADPCKD. These include mitral valve prolapse, cerebral aneurysms, diverticulosis, as well as hepatic and pancreatic cysts. Therefore, a clinical approach addressing both renal and extrarenal manifestations is essential for effective and efficient management.

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