ISSN No:-2456-2165 https://doi.org/10.5281/zenodo.14979741

A Case Report of Acute Leukoencephalopathy as an Initial Neurological Presentation of Autosomal Dominant Polycystic Kidney Disease

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Publication Date: 2025/03/11

Abstract: Autosomal Dominant Polycystic Kidney Disease (ADPKD) is a common hereditary disorder characterized by progressive renal cyst formation and extrarenal manifestations. While neurological complications such as intracranial aneurysms and hypertensive encephalopathy are recognized, acute leukoencephalopathy remains an extremely rare presentation. This case report discusses a 38-year-old male presenting with acute neurological symptoms, including hemiparesis, dysarthria, and encephalopathy, ultimately diagnosed as ADPKD with uremic leukoencephalopathy. Neuroimaging revealed bilateral white matter abnormalities suggestive of acute leukoencephalopathy. The patient's condition improved significantly after initiation of hemodialysis, highlighting the reversibility of metabolic encephalopathy in ADPKD. This report underscores the importance of recognizing atypical neurological symptoms in ADPKD patients, emphasizing the need for early diagnosis and timely intervention to prevent long-term complications.

How to Cite: Dr. Jale Venkat; Dr. Anwar. H. Mujawar (2025) A Case Report of Acute Leukoencephalopathy as an Initial Neurological Presentation of Autosomal Dominant Polycystic Kidney Disease. *International Journal of Innovative Science and Research Technology*, 10(2), 1921-1923. https://doi.org/10.5281/zenodo.14979741

I. INTRODUCTION

Autosomal Dominant Polycystic Kidney Disease (ADPKD) is a prevalent genetic disorder affecting approximately 1 in 400 to 1 in 1000 live births worldwide. It is characterized by the progressive formation of fluid-filled cysts in the kidneys, leading to increased kidney size and gradual decline in renal function. In addition to renal involvement, ADPKD is associated with extrarenal manifestations such as hepatic and pancreatic cysts, cardiovascular abnormalities, and cerebrovascular complications. Neurological involvement, although less common, is primarily associated with intracranial aneurysms, hypertensive encephalopathy, and cerebrovascular events. Acute leukoencephalopathy, however, is an exceedingly rare neurological complication of ADPKD. This report presents a case of acute leukoencephalopathy as the initial neurological manifestation of ADPKD, discussing its diagnostic challenges and management considerations.

II. PATHOPHYSIOLOGY OF ADPKD

ADPKD results from mutations in either the PKD1 or PKD2 genes, which encode polycystin-1 and polycystin-2 proteins, respectively. These proteins are crucial for maintaining renal tubular cell function and structural integrity. Mutations in these genes lead to abnormal cellular proliferation, increased fluid secretion, and extracellular matrix remodeling, contributing to progressive cyst formation and renal dysfunction.

❖ *Clinical Features of ADPKD*

Patients with ADPKD often present with a spectrum of renal and extrarenal symptoms, including:

➢ *Renal Manifestations:*
- Hypertension
- Hematuria
- Proteinuria
- Progressive decline in renal function

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➢ *Extrarenal Manifestations:*
- Hepatic and pancreatic cysts
- Cardiac valvular abnormalities
- Colonic diverticulosis

➢ *Neurological Complications:*
- Intracranial Aneurysms: Increased risk of subarachnoid hemorrhage due to vascular wall fragility.
- Hypertensive Encephalopathy: Secondary to chronic hypertension associated with ADPKD.
- Stroke and Intracerebral Hemorrhage: Increased susceptibility due to vascular abnormalities.

Acute leukoencephalopathy is an extremely rare neurological presentation in ADPKD and is believed to be associated with uremia, electrolyte imbalances, and metabolic encephalopathy.

III. CASE REPORT

❖ *Patient Presentation*

A 38-year-old male with no known prior medical conditions presented to the emergency department with the following acute neurological symptoms:
- Right-sided hemiparesis (weakness of the right upper and lower limbs)
- Dysarthria (difficulty speaking)
- Confusion and altered mental status, indicative of encephalopathy

❖ *Additional complaints included:*
- Bilateral pedal edema for one month
- Occasional flank pain

The patient had no history of fever, seizures, head trauma, or recent infections.

➢ *Clinical Examination*
- Vital Signs: Normal blood pressure, regular heart rate, and afebrile.
- Neurological Examination:
- Right-sided hemiparesis with hyperreflexia
- Dysarthria without cranial nerve involvement
- No signs of meningeal irritation or cerebellar dysfunction
- Abdominal Examination: Mild bilateral flank tenderness without palpable masses.
- Cardiovascular and Respiratory Systems: Normal findings.

❖ *Diagnostic Workup*

➢ *Laboratory Investigations*
- Renal Function Tests: Elevated blood urea nitrogen and creatinine, suggesting azotemia.
- Electrolytes: Hyponatremia, hyperkalemia, and metabolic acidosis, consistent with uremic encephalopathy.
- Urinalysis: Mild proteinuria.
- Complete Blood Count: Normal hemoglobin and platelet levels.

❖ *Imaging Studies*

➢ *Abdominal and Pelvic CT Scan (Figures 1 & 2)*
- Bilateral enlarged kidneys with multiple cysts
- Hepatic cysts, confirming the diagnosis of ADPKD

➢ *Brain MRI (Figure 3)*
- Symmetric T2 hyperintensities in the bilateral centrum semiovale
- Diffusion restriction in the parieto-occipital white matter and splenium of the corpus callosum
- No evidence of hemorrhage or ischemic stroke
- Findings consistent with acute leukoencephalopathy secondary to uremic encephalopathy

❖ *Diagnosis*

The patient was diagnosed with ADPKD complicated by uremia and acute leukoencephalopathy. His neurological symptoms were attributed to metabolic encephalopathy caused by uremia and electrolyte imbalances.

❖ *Management Approach*

➢ *Urgent Hemodialysis*
- Initiated immediately to manage uremia, acidosis, and electrolyte imbalances.
- Significant improvement in neurological symptoms observed post-dialysis.

➢ *Correction of Electrolyte Imbalances*
- Gradual correction of hyponatremia and hyperkalemia to prevent osmotic demyelination syndrome.
- Intravenous bicarbonate administered for metabolic acidosis.

➢ *Supportive Care*
- Neurological monitoring for encephalopathy and hemiparesis recovery.
- Blood pressure control with antihypertensives to prevent hypertensive encephalopathy.
- Nutritional optimization and fluid balance management.

➢ *Long-term Follow-up and Management*
- Nephrology and Neurology Follow-up: Regular monitoring of renal function and neurological status.
- Lifestyle Modifications: Blood pressure control, dietary changes, and renal function surveillance.
- Genetic Counseling: Recommended for family screening and early detection of ADPKD.

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IV. DISCUSSION

➢ *Neurological Complications in ADPKD*

While intracranial aneurysms are the most frequently observed neurological complications in ADPKD, acute leukoencephalopathy is exceptionally rare. Possible mechanisms include:

- Blood-brain barrier dysfunction due to uremia
- Vasogenic edema
- Metabolic disturbances affecting cerebral autoregulation

➢ *Differential Diagnosis Considerations:*

- Posterior Reversible Encephalopathy Syndrome (PRES)
- Hypertensive Encephalopathy
- Metabolic and Toxic Encephalopathy
- Ischemic Stroke or Demyelinating Disorders

This case is unique because acute leukoencephalopathy was the initial neurological presentation of ADPKD, which is rarely documented. The significant neurological recovery post-dialysis highlights the reversible nature of metabolic encephalopathy in ADPKD.

V. CONCLUSION

Acute leukoencephalopathy is an uncommon yet reversible neurological complication in ADPKD, often linked to uremia and metabolic disturbances. This case emphasizes the importance of early neuroimaging and prompt hemodialysis in managing neurological symptoms in ADPKD patients. Physicians should maintain a high index of suspicion for atypical neurological presentations in ADPKD and initiate timely intervention to prevent permanent neurological impairment.

➢ *Clinical Implications*

- Early recognition of neurological symptoms in ADPKD can lead to timely intervention.
- Neuroimaging is crucial in distinguishing metabolic encephalopathy from other neurological disorders.
- Hemodialysis plays a pivotal role in reversing uremic encephalopathy.
- Multidisciplinary care is essential for long-term management of ADPKD.

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