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A Rare Case of Von-Hipple Lindau Syndrome

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Abstract: ***Von Hippel-Lindau disease (VHL) is an inherited cancer syndrome with renal manifestations. VHL is an autosomal dominant condition caused by mutations in the VHL tumor-suppressor gene. VHL is localized to the primary cilia and is necessary for the formation of primary cilia. Like many autosomal dominant cancer syndromes, VHL is recessive at the cellular level: a somatic mutation in the second VHL allele leads to loss of VHL in the cell and abnormal growth. Kidney manifestations of VHL include multiple bilateral kidney cysts, and renal cell carcinomas. Non-renal features of VHL include pheochromocytomas, cerebellar hemangioblastomas, and retinal hemangiomas. Kidney cysts and carcinoma affects the majority of VHL patients. Diagnosis is typically made from a positive family history consistent with autosomal dominant inheritance and multiple kidney cysts bilaterally. Renal ultrasonography is often used for presymptomatic screening of at-risk subjects and for evaluation of potential living-related kidney donors from ADPKD families. We present a case of a 35 years old female having a history of recurrent urinary tract infection, uterine prolapse since 4 years presented with hematuria and burning micturition and fever associated complaint of back pain. Ultrasound of kidneys showed multiple cortical cysts noted throughout right kidney with increased echogenicity of intervening normal cortex, moderate to gross hydronephrosis with dilated ureter up to lower end. CT-KUB showed multiple cortical cysts in both cysts with pancreatic cyst, possibility of ADPKD von hipple lindau syndrome Patient was treated symptomatically with antibiotics according to culture and sensitivity and fluids with antipyretic were given, proper reposition of the uterus was done patient.***

Keywords: Von-Hipple Lindau Syndrome

1. Introduction

VON HIPPEL-LINDAU DISEASE

Von Hippel-Lindau disease (VHL) is an inherited cancer syndrome with renal manifestations. VHL is an autosomal dominant condition caused by mutations in the VHL tumor-suppressor gene. VHL is localized to the primary cilia and is necessary for the formation of primary cilia. Like many autosomal dominant cancer syndromes, VHL is recessive at the cellular level: a somatic mutation in the second VHL allele leads to loss of VHL in the cell and abnormal growth. Kidney manifestations of VHL include multiple bilateral kidney cysts, and renal cell carcinomas. Kidney cysts and carcinoma affects the majority of VHL patients. Non-renal features of VHL include pheochromocytomas, cerebellar hemangioblastomas, and retinal hemangiomas. While much rarer than ADPKD, it is important for this entity to be considered in the differential diagnosis of an individual with newly recognized kidney cysts.

Syndrome-von Hippel-Lindau syndrome
Chromosome (S)-3p25
Gene Protein-VHL von Hippel Lindau protein
Kidney Tumor Type-Clear cell
Additional Findings-Hemangioblastoma of the retina and central
Nervous system; pheochromocytoma; pancreatic
and renal cysts; neuroendocrine tumors

Clinical features:

- **Kidney:** hematuria, flank or abdominal pain, and a flank or abdominal mass
- **Pheochromocytoma**, Headaches, Profuse sweating, Palpitations and tachycardia, Hypertension, sustained or Paroxysmal, Anxiety and panic attacks, Pallor, Nausea, Abdominal pain, Weakness, Weight loss, Paradoxical response to antihypertensive drugs, Polyuria and polydipsia, Constipation, Orthostatic hypotension, Dilated cardiomyopathy, Erythrocytosis, Elevated blood sugar, Hypercalcemia. The clinical presentation of pheochromocytoma in VHL disease is similar to that in sporadic cases, except that there is a higher frequency of bilateral or multiple tumors, which may involve extra-adrenal sites in VHL disease.
- **Cerebellar Hemangioblastomas-**the cerebellum and spinal cord are the most frequently involved sites
- **retinal hemangiomas**
- **Pancreas-**The most frequent pancreatic lesions in VHL are multiple cyst-adenomas, which rarely cause clinical disease. However, non secreting pancreatic islet cell tumors occur in <10% of VHL patients, who are usually asymptomatic.

2. Evaluation

Diagnosis is typically made from a positive family history consistent with autosomal dominant inheritance and multiple kidney cysts bilaterally. Renal ultrasonography is often used for presymptomatic screening of at-risk subjects and for evaluation of potential living-related kidney donors from ADPKD families. The presence of at least two renal cysts (unilateral or bilateral) is sufficient for diagnosis among at-risk subjects between 15 and 29 years of age with a sensitivity value of 96% and specificity value of 100%. The presence of at least two cysts in each kidney and at least four cysts in each kidney, respectively, are required for the diagnosis among at-risk subjects aged 30–59 years and aged ≥60 years with a sensitivity value of 100% and specificity value of 100%. This is because there is an increased frequency of developing simple renal cysts with age. Conversely, in subjects aged between 30 and 59 years the absence of at least two cysts in each kidney, which is

associated with a false negative rate of 0%, can be used for disease exclusion. T scan and T2-MRI, with and without contrast enhancement, are more sensitive than ultrasonography and can detect cysts of smaller size. However, a CT scan exposes the patient to radiation and radiocontrast, which may cause serious allergic reactions and nephrotoxicity in patients with renal insufficiency. T2-MRI, with gadolinium as a contrast agent, has minimal renal toxicity and can detect cysts of only 2–3 mm in diameter.

3. Case Presentation

35 years old female having history of recurrent urinary tract infection, uterine prolapse since 4 years presented with hematuria and burning micturition and fever since 10 days, associated complaint of back pain.

Patient has family history of such symptoms in her mother who died of renal cell carcinoma and similar symptoms of hematuria and recurrent urinary tract infections with usg suggestive of polycystic kidney disease in her brother.

Patient was normally built and malnourished
On examination
Temperature-high (100 F) P-100/min BP-100/60 mm hg Rr-16/min
On respiratory examination-bilateral air entry present
Cvs-normal heart sounds
Cns-conscious oriented
Per abdomen-soft non tender

On blood investigation
Hb-8.3 mg/dl, Wbc-9.54, Apc-2.92
Urea-129 mg% Creat-4.6 mg%
Na-131 meq/L K-5.6 meq/L
Total protein-9.9 Albumin-4.4 Globulin – 5.5
UA-5.0

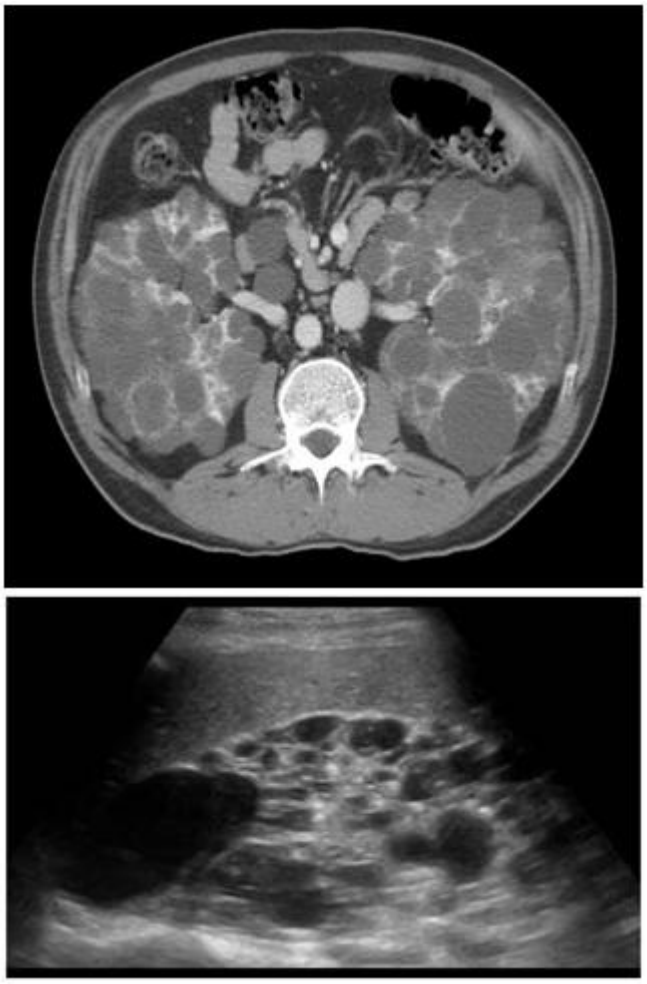
On ultrasound
Right kidney: multiple cortical cysts noted throughout right kidney with increased echogenicity of intervening normal cortex, moderate to gross hydronephrosis with dilated ureter upto lower end.

Left kidney: multiple small cortical cysts noted throughout left kidney with echogenicity of intervening normal cortex, moderate to gross hydronephrosis with dilated ureter upto lower end. Possibility of autosomal dominant polycystic kidney disease. Streak of perinephric fluid is seen.

Pancreas: 15*12 mm anechoic cystic lesion noted in head of pancreas.
Urinary bladder slightly thickened and irregular with max wall thickness 4 mm
Liver and spleen normal in size and morphology.

CT-KUB: Multiple cortical cysts in both cysts with pancreatic cyst, possibility of-
-ADPKD
-von hipple lindau syndrome

Patient was treated symptomatically with antibiotics according to culture and sensitivity and fluids with antipyretic were given, proper reposition of the uterus was done patient counselled for urosurgery for management of cyst in kidney.



Conclusion:
In our case patient had hematuria, recurrent urinary tract infection, pancreatic cyst and ct-kub finding suggestive polycystic kidney disease and from family history likely of von hipple lindau syndrome.