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Autosomal Dominant Polycystic Kidney Disease Associated with Liver Cysts and Hypothyroidism: A Rare Case Finding

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Abstract: ***Autosomal Dominant Polycystic Kidney Disease(ADPKD) is a multisystem, dominantly inherited type of complex disorder affecting adult patients with bilateral, multiple, variable sized renal cysts as the hallmark finding. The most prevalent extra-renal symptom of ADPKD is liver cyst, which is frequent incidental finding while hypothyroidism is less prevalent. Treatment with Tolvaptan has recently approved for the management of liver cysts. This case study presents a rare finding of Autosomal Dominant Polycystic Kidney Disease (ADPKD) associated with liver cysts and hypothyroidism in a 39-year-old female. The patient presented with symptoms including abdominal fullness, decreased appetite, pedal edema, and generalised weakness. On examination, massive hepatomegaly, hypertension, hypothyroidism, anemia, and increased serum creatinine were found. Patient was started on symptomatic treatment with regular screening and follow up. The study highlights the importance of considering liver cysts and hypothyroidism in patients presenting with such symptoms and emphasises the need for careful evaluation and monitoring of patients with polycystic liver disease to manage potential complications and optimise treatment outcomes.***

Keywords: ADPKD, Renal cysts, liver cyst, hypertension, hypothyroidism

1. Introduction

The most prevalent inherited kidney ailment, ADPKD, was showcased nearly 300 years ago and has an incidence of 1:500 to 1:1000 per live birth[1].The kidney in ADKPD is characterised by the development of multiple variable-sized cysts that eventually obliterate the normal structure, this results in interruption of the filtration and physiologic functions of the kidneys[2].There is hypertrophy of the remaining glomeruli and compensatory hyperfiltration, which maintains kidney function within a relatively normal range for decades[2].

In addition to the kidney, cysts can also develop in the liver, thyroid, pancreas, and spleen[3]. ADPKD is genetically diverse, with two genes, namely, PKD1 on chromosome 16p and PKD2 on chromosome 4q211[1,3]. PKD1 mutations cause renal and hepatic cysts to form, while PKD2 mutations cause the remaining 14% of cases[3]. In addition to genetic mutation, other factors such as age, female sex, and oral contraceptive usage also play a role in cystogenesis[1,3].

Ultrasound (USG), tomography, magnetic resonance imaging, and laparoscopy are the primary diagnostic techniques for hepatic involvement[4]. The treatment of liver cysts is directed as per patient complaints and ranges from no therapy in asymptomatic patient to drug intervention in symptomatic patient that inhibits cell dedifferentiation, apoptosis, fluid secretion, and dysregulated cellular signal cascades like Octreotide, Lanreotide, and Pasireotide as somatostatin analogues; Sirolimus and Everolimus as mTOR inhibitors; and, most recently, Tolvaptan, a vasopressin V2 receptor antagonist[3,4].

2. Case Report

A 39- year- old female presented with the chief complaints of abdominal fullness, decreased appetite, and generalised weakness for5 months, pedal edema for 4 days. She had no complaints of decreased urination, burning micturition, sore throat, fever, cough, cold, altered sleep pattern, or breathlessness. Treatment history included a diagnosis of ADPKD, hypothyroidism, and diabetes mellitus from a private hospital from which she was referred to SSGH. Obstetric history includes G2P2A0L2. Menstrual history showed no abnormality. On general examination, blood pressure and Random blood sugar were higher than normal, while other vitals were stable.

On systemic examination, Per-abdominal examination revealed massive hepatomegaly, while CVS, RS, and CNS findings were normal.

Laboratory investigations are as follows:

Parameter	Value	Reference range
Haemoglobin	**9.40 gm/dL**	(11.5 – 15 gm/dL)
Erythrocyte count	**3.47 * 10^6/ cmm**	(3.8 – 4.8 * 10^6/ cmm)
PCV(Packed Cell Volume)	**31.70%**	(36 -46 %)
MCV(Mean Corp. Volume)	84.80 fL	(83 – 101 fL)
MCH(Mean Corp. Hb)	**25.10 pg**	(27 – 32 pg)
MCHC(Mean Corp Hb Conc)	**29.70 gm/dL**	(31.5 – 34.5 gm/dL)
RDWcv	**17.80%**	(11.6 – 13.7 %)
WBC(Leucocyte Count)	6000/cmm	(4000 – 10000/ cmm)
Platelets	**148000/cmm**	(150000 -410000/ cmm)
S. Urea	20.00 mg/dl	14 - 40 mg/dl
S. Creatinine	**1.53mg/dl**	0.6 - 1.2 mg/dl
ESR	**30.00 mm**	1 - 12 mm

Peripheral smear revealed mild microcytic hypochromic anaemia. USG showed massive hepatomegaly with innumerable, variable-sized cystic lesions, a bilaterally distended kidney with multiple, variable-sized cysts, the right kidney displaced slightly inferiorly, a distended gall bladder, a pancreas obscured by bowel gas, and a lower abdomen full of bowel. The patient was kept on symptomatic treatment, and medications included Tab Thyroxine 25ugonce a day, Enalapril 5mg at bedtime, folic acid once daily, Vitamin C twice a day, Tab Calcium 500mg twice a day, and a Vitamin D3 sachet once a week. The patient was also given IV pantoprazole and ondensatron to control vomiting. Thepatient's condition improved after starting the treatment, and regular screening and follow up are advised.

3. Discussion

ADPKD is the most prevalent genetic kidney condition and a leading contributor to end-stage renal disease[5]. ADPKD is a systemic ailment, as evidenced by the numerous clinical signs and symptoms it exhibits, such as developing cysts in the kidneys, hypertension, headaches, hematuria, pain in the back and extrarenal consequences including liver cysts, cerebral aneurysms, and heart valve disease [2].In our case, the patient presented with chief complaints of abdominal fullness, decreased appetite, and pedal edema. The patient had associated comorbidities like Type 2 DM and hypothyroidism. The past and family history was insignificant. Obstetric and menstrual histories were also normal. On general examination, hypertension was detected; USG examination revealed massive hepatomegaly with multiple cystic lesion, pancreas obscured with bowel gas and lower abdomen distended with bowel. ESR and serum creatinine were increased.

Our aim in treatment was to control symptoms and reduce associated complications like hypertension, biventricular organ dysfunction, and organ damage. Medication included injections of Pentoprazole and Ondensetron, Enalapril to control blood pressure, Vitamin C, Tab Calcium and Vitamin D3.

In our case, liver cysts were found, which is a common ADPKD-associated finding in people over 35 years of age, predominantly in females, most probably due to oestrogen exposure during pregnancy or exogenous oestrogen pills[6]. It is generally asymptomatic to symptoms like abdominal pain and vomiting, depending on the size of cysts[3,7]. It might be associated with the PKD1 mutation, which is the most common extra-renal finding[8]. Another finding was hypothyroidism, which may be due to defective cilia signalling, thus causing thyroid lesions[9].

Other findings of ADPKD like saccular aneurysm, mitral valve prolapse, aortic regurgitation, colonic diverticula are rare and were not seen in our case[10].

In conclusion, the present case highlights the importance of considering hypertension along with abdominal pain and abdominal fullness as differential findings in presentation of ADPKD with associated liver cysts. Early diagnosis and treatment can reduce the risk of associated organ damage, end-stage kidney failure, expansion, development and infection of liver and kidney cysts.

4. Conclusion

This case report describes a rare case of a 39yearold female patient with ADPKD, presenting with liver cysts and hypothyroidism. The patients symptoms included abdominal dullness, bilateral pedal edema, and hypertension.The patient was started on a treatment plan consisting of enalapril, pantoprazole, ondensetron, folic acid, iron supplementation, calcium, and vitamin D3 supplements. Regular follow-up visits were scheduled to monitor the patient's progress and manage any complications. The case report emphasizes the need for careful evaluation and monitoring of patients with polycystic liver disease to manage potential complications and optimize treatment outcomes. Further research is needed to improve our understanding of this disease and develop more effective therapies.

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