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Anemia in a Young Female with Multiple Cysts in Bilateral Kidney and Liver: A Case Report

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Abstract:
Background*: Polycystic Kidney Disease (PCKD) is a rare inherited disorder in which non-cancerous round cysts containing fluid are present in kidney which can also develop in liver and elsewhere in body. In early stages PCKD can lead to rise in erythropoietin levels which are not accompanied by the rise in hemoglobin concentration in blood.*
Case Report*: Here we present a case of a 36 years old young Muslim woman with the complaint of weakness, lower backache and discharge per vaginum. After detailed history and general examination done, blood investigations, ultrasonography (USG) whole abdomen, Chest X ray PA view were adviced. Ultrasound revealed hepatomegaly, multiple cystic lesions in both lobes of liver of variable size, gall bladder distended with 10 mm size single calculus, and multiple small cysts in B/L kidney. General blood picture (GBP) suggestive of microcytic hypochromic anemia along with tear drop cells and elliptocytes.*
Discussion*: This is a case of young Muslim woman presented with weakness, pain in right side flank. Multiple hematological and radiological test were done which were showing microcytic hypochromic anemia and tear drop cells with multiple cysts in bilateral kidney and both lobes of liver. The findings were suggestive of polycystic kidney and liver disease. It is a rare inherited disorder which is associated with chromosomal mutation. It is responsible for chronic kidney disease and leading to failure in later stage.*
Conclusion*: Polycystic kidney and liver disease are rare inherited disease; cyst may be present in spleen, lungs, brain and other visceral organs. Patient's present with minor complaints or mainly asymptomatic symptoms. Symptoms develop in advanced conditions.*
Keywords*: weakness, microcytic hypochromic anemia, multicystic kidney, multicystic liver*

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I. Introduction:

Polycystic Kidney Disease (PCKD) is a rare inherited disorder in which cluster of cysts develops primarily within kidneys, which may be adult – (Autosomal Dominant, AD) and Childhood (Autosomal Recessive, AR), causing them to enlarge and loose function over time. Cysts are non cancerous round sacs containing fluid. These may vary in size; cysts can also develop in liver and elsewhere in the body. The disease can cause serious complications, including high blood pressure and kidney failure. Prevalence of PCKD is one per 1000 in general population and that of multicystic liver disease is one per 100,000 in general population (1). In early stage of PCKD, there is a significant rise in serum erythropoietin (EPO) levels that is not accompanied by a similar elevation of hemoglobin. In later stages, there is a continuous fall in hemoglobin. In early stages of PCKD of other etiologies, there is a significant negative correlation between EPO and hemoglobin that is lost in stages 4 and 5, but no correlation was found, in any stage, in patients with PCKD. Kidney enlargement resulting from the expansion of cysts in patients with AD-PCKD is continuous and quantifiable and is associated with the decline of renal function. Renal anemia is caused by a relative EPO deficiency. Due to difficult interpretation of serum EPO concentrations adapted to anemia and renal function, the diagnostic value of measuring serum EPO concentrations is limited(2).

Polycystic liver disease (PCLD) is an inherited disorder estimated to affect around 1 in 100,000 people. It is characterized by the progressive growth of cysts of various sizes scattered throughout the liver (3). People affected by this condition tend to have more and larger cysts as they age and usually start to have symptoms

around age 50, although symptoms can begin to occur earlier. However, many affected individuals do not have symptoms. Males and females are affected in equal numbers, but most patients with symptoms and with severe disease are women. The suggested cause of this difference is that female sex hormones, such as estrogen, contribute to growth of liver cysts. Oral contraceptives and estrogen replacement therapy are also associated with more severe disease as estrogen promotes cysts growth, it is recommended for women diagnosed with PCLD to stop hormonal contraceptives or estrogen replacement therapy(1). EPO is present inside and outside the cyst. The intracystic EPO concentration is insensitive to the change of Hb concentration and oxygen saturation in blood (1).

II. Case Report:

A 36 yrs old young Muslim female, presented with the complaint of weakness which has been persistent since 8-9 yrs, pain in right side of back which was dull aching in nature, insidious in onset, persistent, mild to moderate in severity, non radiating, non migratory with no precipitating and relieving factors associated with discharge per vaginum which is intermittent since 8-9 years , watery to curdy-whitish in consistency , moderate in amount, foul smelling and not associated with lower abdominal pain, bleeding per vaginum, burning micturition, increase in frequency of urination, fever and not associated with any variation in time and season or with menstrual cycle .

After detailed history and physical examination it was elicited that she was a young married female, mother of four children with normal built, with good nutritional status having belonging to upper middle class having mild pallor and no other co-morbidities and with normal menstrual history and with no history of any contraceptive use or any drug allergy.

On detailed clinical examination: tenderness on back in right side at the level of 12^{th} rib and rest per abdomen and detailed gynecological examination suggests no abnormal findings.

General Blood tests were performed which suggested red blood cells of moderate anisopoikilocytosis with mixed population of microcytic hypochromic anemia with tear drop cells and elliptocytes. The differential diagnosis includes-Lymphoblastic leukaemia, Myelodysplastic Syndrome (MDS) – Congenital, Chemo & Radiation Induced, Metastatic Neoplasm, Splenic Abnormality, Primary Myelofibrosis, Megaloblastic anaemia and Thalassaemia. Serum iron profile was deranged which was suggestive of iron deficiency anemia and also folic acid deficiency. Serum Erythropoietin levels were normal. Liver and Kidney function tests were within normal limits.

Regarding radiological tests, chest x-ray P/A view was normal with no visible abnormality and USG W/A was suggestive of enlarged multicystic liver with lesions in both the lobes of liver of varying sizes, gall bladder distended with normal wall and single calculi noted in the lumen of gall bladder. Multicystic and enlarged kidneys with anechoic cysts and mildly bulky uterus, b/l ovaries were normal rest all structures were within normal limits.

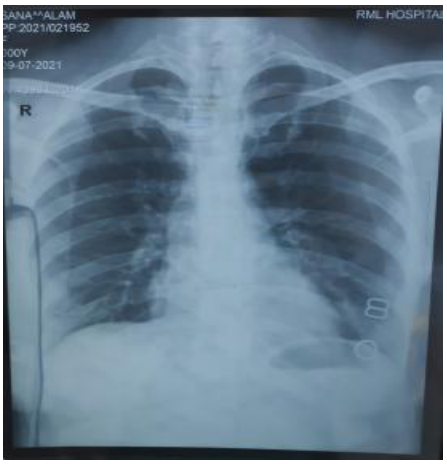

Figure 1
Figure showing normal chest X ray PA view

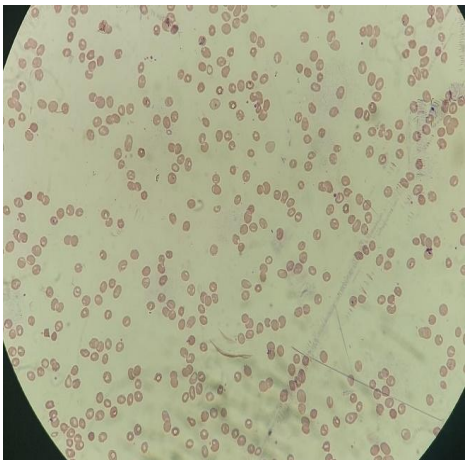

Figure 2
GBP showing moderate anisopoikilocytosis
Tear drop cells and few elliptocytes seen
(Leishman stain at 40 X)

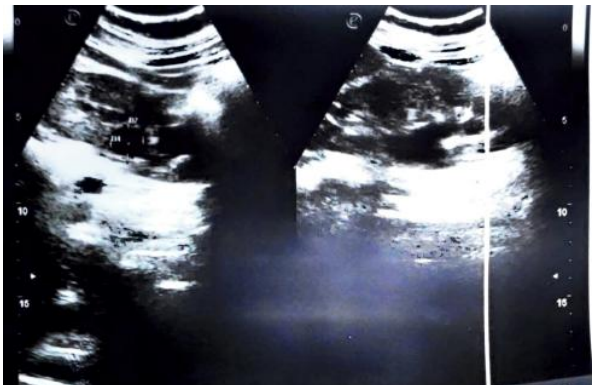

Figure 3 a
Figure showing multiple cysts in B/L kidney in USG

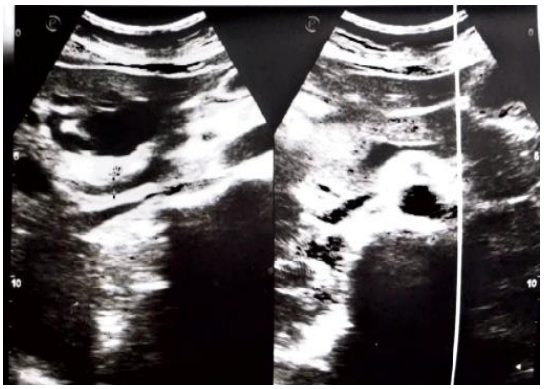

Figure 3 b
Figure showing multiple cysts in both lobes of Liver in USG

III. Discussion:

This is an interesting case of a young Muslim woman who presented with the complaint of weakness and pain in right side of flank; multiple tests were done blood, urine (routine & microscopic) which was normal. Peripheral blood smear was suggestive of microcytic hypochromic anemia, Ultrasound Whole abdomen was done and it showed a coincidental finding of multiple cysts in bilateral kidney and both the lobes of liver of varying sizes. The findings are suggestive of a case of polycystic kidney and liver disease; it is a rare inherited disorder which is associated with mutation in PKD1 in chromosome 16p13.3 in polycystin-1 and in PKD2 in chromosome 4q21 in polycystic 2(4). Polycystic liver disease is associated with mutation SEC63 and PRKCSH mutation (3).

Polycystic kidney disease is responsible for chronic kidney disease and leading to failure in late stages. Currently recommended medical treatment for Polycystic kidney diseases are for signs, symptoms and complications in early stages and in later stage kidney transplant may be required(5). There are several ongoing trials for the management of polycystic kidney (PCKD) and liver disease (PLD) like Vasopressin 2 receptor antagonist (TOLVAPTAN), Somatostatin receptor antagonist (OCTREOTIDE), M-TOR blocker (SIROLIMUS), VEGF antagonist BEVACIZUMAB(6).

IV. Conclusion:

Polycystic kidney and liver disease are rare inherited disorders that are usually asymptomatic and can present with very minor symptoms or largely asymptomatic. They may present symptomatically in advanced conditions. Hence it is advisable to perform routine chest radiography and ultrasound whole abdomen to rule out any underlying pathology, so that, detect these cases as early as possible.

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