CASE REPORT

An unusual association of autosomal polycystic kidney disease and multiple myeloma

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SUMMARY

Autosomal dominant polycystic kidney disease (ADPKD) is well known for its different renal and extrarenal complications. Chronic renal failure (CRF) from ADPKD is also reported in older patients. On the other hand, multiple myeloma (MM) can also cause renal damage by different mechanisms. A 38-year-old man presented with dull aching abdominal pain, recent onset respiratory distress along with bilateral pedal swelling. Initial investigations revealed polycystic kidney disease but subsequently it was found that the patient was also suffering from MM. This rare association of ADPKD and MM was responsible for aggravating the renal damage and perhaps resulted in early age of presentation with CRF. The patient was managed symptomatically but unfortunately succumbed before starting specific treatment for MM.

BACKGROUND

Autosomal dominant polycystic kidney disease (ADPKD) is the most common inherited kidney disease where renal function usually does not deteriorate for many years until the kidneys have approximately quadrupled in volume. It may lead to chronic kidney disease (CKD) over 60 years and dialysis remains the only treatment option.¹ But renal failure from multiple myeloma (MM) is more common due to various pathophysiologies. It is present in about 20% of patients with MM at diagnosis. It may be reversible in up to 50% of patients, while approximately in 10% of newly diagnosed patients with MM may require dialysis.² MM can also present with plasmacytoma where histology will help in diagnosis. But when both ADPKD and MM occur in the same setting, as in our case, this rare association may aggravate the renal injury simultaneously.

CASE PRESENTATION

A 38-year-old male patient with non-diabetes, non-hypertension, presented with progressively increasing dull aching diffuse pain abdomen for 6 months, respiratory distress for 2 weeks and bilateral pedal swelling for 10 days. There was also history of low-grade intermittent fever for the last few days along with nausea and anorexia. There was no history of cough, haemoptysis, chest pain or paroxysmal nocturnal dyspnoea. There were also no histories of yellowish discolouration of eyes or urine, abdominal distension, haematuria, burning micturition, rash or joint pain. The patient had no addiction and took paracetamol only as and when necessary for pain relief. On examination, there was

hypertension (blood pressure 160/96), tachycardia, tachypnoea along with facial puffiness, moderate pallor, bilateral pitting pedal oedema, no lymphadenopathy and some firm swellings over the lateral sides of the chest wall. Bilateral fine crepitations were present on chest auscultation, and abdominal examination revealed a mass over the left lumbar region. Other system examinations were within normal limits.

INVESTIGATIONS

Complete haemogram was—haemoglobin 7.2 g/dL (13–17 g/dL), total leucocyte count 9800 (4000–10 000/mm³), N74, L15, M11, platelet 2.01 lakhs/dL (1.5–4 L/mm³), packed cell volume 23.8% (40–50%), mean corpuscular volume 71 fL (83–101 fL), mean corpuscular haemoglobin 21.7 pg (27–32 pg), mean corpuscular haemoglobin concentration 30.3% (31.5–34.5%), erythrocyte sedimentation rate 45 mm (<30 mm). Fasting sugar was 106 mg/dL (70–110 mg/dL), urea 115 mg/dL (10–45 mg/dL), creatinine 5.2 mg/dL (0.5–1.5 mg/dL), Na 134 mEq/L (136–149 mEq/L), K 4.8 mEq/L (3.6–5.3 mEq/L). Liver function test was carried out that showed bilirubin total 0.7 mg/dL (0.2–1.0 mg/dL), aspartate aminotransferase 14 units/L (<37 units/L), alanine aminotransferase 16 units/L (<41 units/L), alkaline phosphatase 153 units/L (40–130 units/L), total protein 7.36 g/dL (6.6–8.3 g/dL), albumin 2.8 g/dL (3.5–5.5 g/dL), globulin 4.56 g/dL (2.0–3.5 g/dL). Serum calcium was 16 mg/dL (8.8–10.6 mg/dL) and phosphate was 5 mg/dL (2.5–4.5 mg/dL). Chest X-ray showed triangular opacity on the right mid zone, along with radiolucent spots over the ribs and scapula (figure 1). Ultrasonography of the whole abdomen revealed heterogeneous echo textured liver along with small cysts in both lobes. There was also bilateral renal enlargement (RK 16.3 cm, LK 17.3 cm) with multiple cysts of varying sizes in them (figure 2). Mild left-sided pleural effusion was also noted. Transthoracic echocardiography did not reveal any abnormality other than mild left ventricular hypertrophy and mild pericardial effusion. After 2 days his creatinine level reached to 8 mg/dL and started suffering from severe respiratory distress. So we went for dialysis. At first we thought that the renal impairment was due to polycystic kidney disease, but high serum calcium, some radiolucent spots over the ribs and firm swellings over the chest wall were hinting of some more hidden pathology.

We performed X-ray of the skull (figure 3) that revealed multiple punched out lesions and X-ray of the pelvic bones also revealed multiple lytic lesions. Fine-needle aspiration cytology was carried out

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Figure 1 Chest X-ray showing triangular opacity on the right mid zone, along with radiolucent spots over the ribs and scapula.

from firm swellings over the lateral chest wall, which showed round cells with scanty cytoplasm, clumped nuclear chromatin and scanty cytoplasm, arranged in sheets and rosette-like fashion along with lymphoplasmacytic cells. We carried out serum protein electrophoresis that demonstrated M spike in the γ -globulin region (γ -globulin 4.58 g/dL). We also carried out bone marrow biopsy that showed 90% plasma cells (figure 4). So considering all the facets of our investigations, there was underlying MM beside the ADPKD that made the scenario of renal failure.

DIFFERENTIAL DIAGNOSIS

- ▶ Renal cell carcinoma with renal failure.
- ▶ Other intra-abdominal malignancy with distant metastasis.
- ▶ Chronic renal failure with polycystic kidney disease.

TREATMENT

The patient was managed symptomatically on presentation.

OUTCOME AND FOLLOW-UP

Unfortunately patient succumbed to sudden cardiac death before starting specific therapy for MM. The patient's death can be related to arrhythmia due to dyselectrolytaemia. The patient party did not give consent for autopsy. So the exact cause of death remained undetermined.

Figure 2 Ultrasonography of the abdomen showing multiple cysts in both the kidneys.

Figure 3 Skull X-ray showing multiple punched-out lesions.

DISCUSSION

ADPKD is the most common inherited kidney disease. In most of the cases, there is family history but it may be absent in 5–10% cases.¹ While by using Ravine's ultrasonographic criteria renal cysts are evaluated, there may be cysts in other organs too such as liver, pancreas, brain, spleen, ovaries and testis in descending order. Besides this, there may be cardiac valvular disorders, abdominal and inguinal hernias, and aneurysms of cerebral or coronary arteries and aorta.³ It may be complicated by Potter's syndrome in the child, hypertension and excessive size of the kidneys causing respiratory dysfunction. By the age of 60, about 50% of the patients develop CKD. Non-renal complications include hepatic fibrosis, intracranial aneurysm rupture leading to subarachnoid haemorrhage, cardiac valvular lesions such as mitral valve prolapse, hernias and colonic diverticulosis.^{1–3} Cysts may rupture and give rise to haematuria. Owing to enlarged kidney, pain in the abdomen, intestinal obstruction and even necrosis have been reported.⁴ Gatalica *et al*⁵ have reported a case of papillary renal cell carcinoma (RCC) in association with ADPKD, which presented with lytic bone lesions

Figure 4 Bone marrow biopsy showing plasma cells.

and hypercalcaemia. Hajj *et al*,⁶ in their study, found 12.35% chance of RCC from ADPKD with end-stage renal disease and histologically they were clear cell carcinomas (58.3%) and tubulopapillary carcinomas (41.7%). Now, malignancies which can present with lytic bone lesions include carcinoma from thyroid, renal, adrenal, Wilm's tumour, Ewing's sarcoma, melanoma, etc. On the other hand, differential diagnosis of malignant small round cell tumour includes Ewing's sarcoma, peripheral neuroectodermal tumour, rhabdomyosarcoma, synovial sarcoma, non-Hodgkin's lymphoma, retinoblastoma, neuroblastoma, hepatoblastoma and nephroblastoma, small cell osteogenic sarcoma, undifferentiated hepatoblastoma, plasmacytoma, granulocytic sarcoma and intra-abdominal desmoplastic small round cell tumour.⁷ Kouno *et al*⁸ have reported a case of osseous plasmacytoma presenting as osteolytic tumours of the iliac bone and femur, found out to be a CD56 small round cell tumour. Terada⁹ has also reported a rare case of intracranial plasmacytoma with small round cell pattern of histology, preceding lytic skull lesions. MM can cause renal injury from hypercalcaemia, amyloidosis, urate nephropathy, light chain deposition, etc.

In the long run, ADPKD is reported to progress into CKD or renal malignancy, mostly in aged patients according to some case reports. But when this occurs early, as in the middle age of

our patient, some second insult must be thought of. In our case, the hidden MM with plasmacytomas over chest, being small round cell tumour on histology, perhaps accelerated the course to CKD. On the other hand, MM could itself damage the kidney by different mechanisms, which may worsen due to simultaneous presence of ADPKD. In spite of our extensive search, we have not found such a rare association of ADPKD and MM in the literature. So perhaps our case is so unique in that sense to present.

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Contributors SKM and SSM participated in treating physician; JG wrote the article and KS reviewed the literature.

Competing interests None.

Patient consent Obtained.

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Learning points

- In a background of autosomal dominant polycystic kidney (ADPKD), if features of early renal failure develop, then a second cause is always to be ruled out.
- Rare association with ADPKD and multiple myeloma may occur.
- Multiple myeloma may also occur in younger age group.

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