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Case Report

Autosomal Dominant Polycystic Kidney Disease: A Risk Factor for Berry Aneurysm

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Abstract

Autosomal dominant polycystic kidney disease is a genetical disorder caused by changes occurred in PKD1 and PKD2 genes and is characterized mainly by the growth of multiple kidney cysts. It is progressive disorder with symptoms of high blood pressure, pain in the back and the sides, and also headache. Patients having ADPKD are having a high risk of formation of intracranial aneurysms. Presenting a case of ADPKD associated with subarachnoid hemorrhage with berry aneurysm and discussed about the diagnostic approach with imaging modalities in this patient and the treatment.

Key words: Autosomal dominant polycystic kidney disease, Aneurysms, Subarachnoid hemorrhage

INTRODUCTION

Autosomal dominant polycystic kidney disease is a disorder with a genetic cause of changes occurred in PKD1 and PKD 2 genes. Age specificity of this disease distribution is between ages of 30 and 40. The most common symptoms of this disease are pain in the back and sides, headaches, urinary tract infection symptoms, high blood pressure, blood in the urine, and fullness of abdomen. The other manifestations are liver and pancreatic cysts, abnormal heart valves, kidney stones, and brain aneurysms. Berry aneurysms are intracranial saccular aneurysms which has a well-known association with ADPKD. CT brain with angiogram being the imaging modality to show the berry aneurysms initially and digital subtraction angiography for the precise diagnosis. Treatment options for berry aneurysms being three: Surgical clipping, endovascular coiling, or conservative management.

CASE REPORT

A 45-year-old male presented to the emergency department with the chief complaints of vomiting, more than 10–15 episodes per day for 2 days, vomiting was not blood stained and not preceded by nausea. Furthermore, he had complaints of back pain and severe headache both in frontal and occipital regions associated with neck pain. The patient does not have a history fever, abdominal pain, loose stools, and blurring of vision. The patient was a known case of systemic hypertension for 5 years and was on regular medication. On examination, the patient was thin built, conscious, oriented to time, place, and person and was afebrile. Systemic examination and cardiovascular system examination-S1, S2 were normal, no murmurs and split. On respiratory system examination, bilateral air entry was equal, no added sounds were heard. Abdomen was soft and non-tender, non-distended. Central nervous system examination revealed no focal neurological deficit. On checking vitals, his blood pressure was 180/110 mmHg, pulse rate was 68 and regular and was maintaining normal oxygen saturation.

Routine blood investigations were done showed normal complete blood count analysis and serum electrolytes. There was mild elevation in values of urea and serum creatinine such as urea – 57 mg/dl and serum creatinine was 1.6 mg/dl, respectively. Nephrologist opinion was obtained in view of elevated renal function parameters and USG abdomen was taken showed multiple cysts of various sizes in both the kidneys indicating adult polycystic kidney disease. In view of persistent severe headache, MRI brain was taken showed features of raised

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intracranial hypertension and subarachnoid hemorrhage in basal cisterns, both the Sylvian fissures, and posterior subarachnoid space over the thoracic region.

USG Abdomen showing multiple cysts in the kidneys [Figure 1].

CT brain angiogram showing subarachnoid hemorrhage, few berry aneurysms in the right MCA, and proximal ACA and also "puff of smoke" like appearance (Moyamoya disease) [Figure 2].

Neurologist opinion was obtained and was treated with steroids such as, antiepileptics to prevent seizures, antihypertensives, calcium channel blockers to prevent vasospasm, and other supportive drugs. Moreover, as per neurologist suggestion, CT brain with angiogram was done and neurosurgeon opinion was obtained. CT brain angiogram showed, few small approximately 5 mm berry aneurysms in the right middle cerebral artery and its branches and proximal anterior cerebral artery and also the appearance of Moyamoya disease. As per neurosurgeon advice, digital subtraction angiography was done and treated surgically with neurosurgical clipping of the aneurysm. Post-surgery patient was under intensive care unit for some period, was shifted to ward once general condition was stable and the patient was discharged after symptomatic improvement.

DISCUSSION

Autosomal dominant polycystic kidney disease (ADPKD) is a genetic disease caused by changes in the genes such as PKD1 and PKD2. Polycystic kidney disease (PKD) is of two variants depending on the inheritance such as autosomal dominant and autosomal recessive. Autosomal dominant variant causes cysts to grow only in the kidneys and it is also called as adult PKD and patients with this variant may not notice any of the symptoms until the age of 30–50. Autosomal recessive variant causes cysts to grow both in kidneys and liver, which is also named as infantile PKD, as the babies may show symptoms during their 1st months of life. ADPKD being the common variant affects 1 in 500–1000 people all over the world. As it causes progressive growth of cysts in kidney, it causes the kidneys to enlarge and damage the kidneys and reduces its function, leading to ESRD in many cases. It is fourth important cause for end-stage renal disease. PKD is mostly always inherited either from one of the parents or both the parents. People affected are of both genders, all age groups, nationalities, and ethnicities equally. It is that if there is family history of ADPKD in a blood relative, its more likely to have ADPKD. However, it can also occur without a family history of ADPKD because of the new genetic change that occurs in the gene that causes ADPKD.

Polycystin 1 is responsible to regulate tubular epithelial cell adhesion and for its differentiation and polycystin 2 usually functions as ion channel along with mutations that cause cysts fluid secretion. These protein mutations alter the main function of renal cilia that is to enable the tubular cells for sensing the flow rates. Moreover, this ciliary dysfunctions lead to the cystic transformation. Hemorrhage may also occur in these cysts causing hematuria. During the initial period, patients do not develop any symptoms. The common symptoms being pain in the back, flank pain, and headache and signs such as hematuria, proteinuria, high blood pressure, and polyuria. Other manifestations may be development of liver cysts, pancreatic cysts colonic diverticula, and hernias, can lead to valvular heart disorders most often mitral valve prolapse and also aortic regurgitation, coronary artery aneurysms, and the most serious being the cerebral aneurysms which are about 5% in young adults and to a range of 10% in aged patients.[1]

Diagnosis is suspected in patients with a positive family history, with symptoms or signs. Diagnosis is usually made by USG abdomen imaging which shows extensive cystic changes throughout the kidneys which are usually enlarged. If USG results are not conclusive, then the next step being CT or MRI, whereas these both tests are very sensitive. Other routine tests such as urinalysis, renal function tests, and complete blood count were done. Genetic testing is done for patients who are suspected to have PKD but not with family history, where imaging studies are not definitive and in more young patients.[2,3] Treatment part being strict control of blood pressure with ACE inhibitors or ARBs which reduces the renal scarring and decreases the progression of loss of kidney function. Other drugs such as tolvaptan, a vasopressin receptor 2 antagonist, used to slow the increasing renal volume and declining kidney function but it may cause severe liver failure and is contraindicated in liver disease patients. If symptoms of urinary tract infection persists, then it is to be treated and if severe symptoms occur due to massively enlarged kidneys then nephrectomy option is being choosed. Hemodialysis, peritoneal dialysis, or transplantation of kidney are done for patients who develop chronic kidney disease.[2]

Most serious complication being cerebral aneurysms and is noted in 9–12% of ADPKD patients. Aneurysmal walls are mainly featured by the elastic tissue disruption and the vascular smooth muscle cells loss. Thus, the decreased level of polycystin in the vascular smooth muscle which is caused by the mutations in polycystic kidney disease facilitates the development of aneurysms. Berry aneurysms also named as saccular aneurysms represent more than 90% of the cerebral aneurysms. There is a ballooning that happens from the weak area in the wall of the concerned blood vessel of brain. Symptoms range from no symptoms to intracranial hemorrhage may be subarachnoid in some

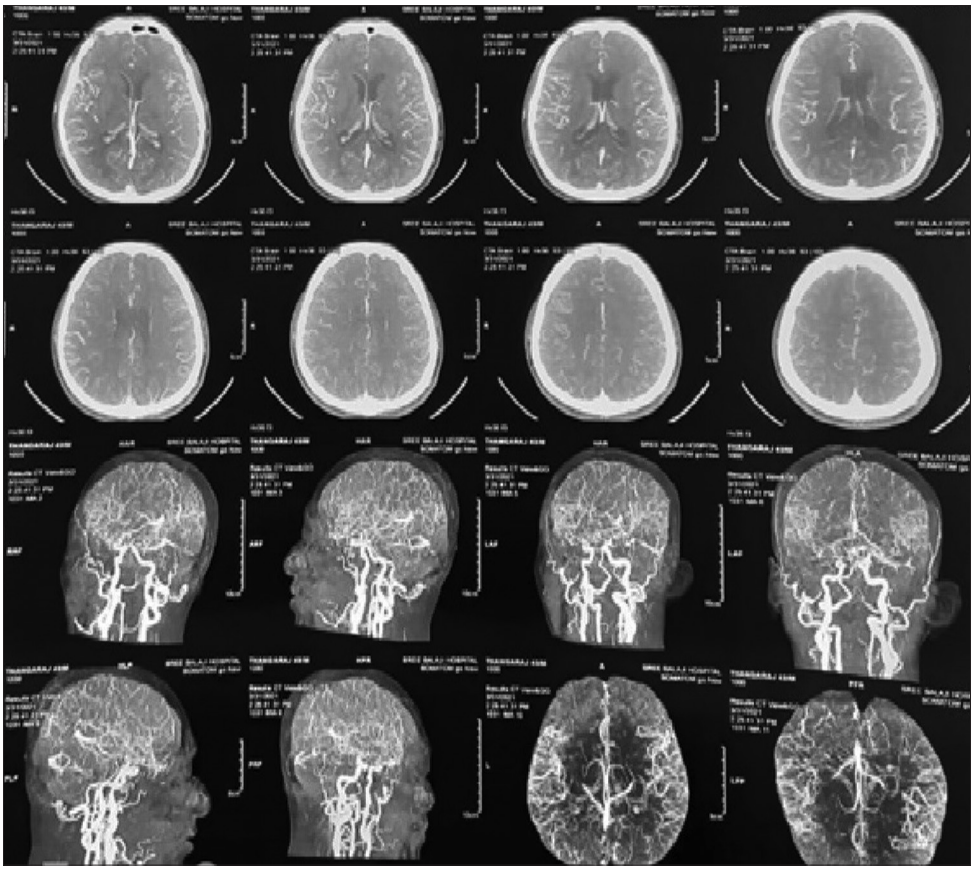

Figure 1: CT brain angiogram showing subarachnoid haemorrhage, few berry aneurysms in right MCA and proximal ACA and also "puff of smoke" like appearance (MoyaMoya disease)

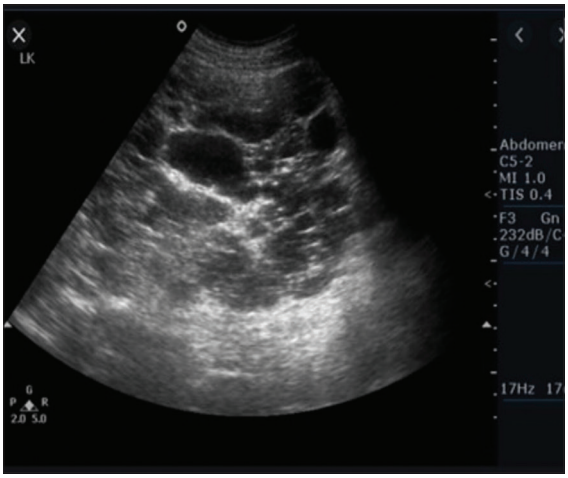

Figure 2: USG abdomen showing multiple cysts in the kidneys

cases. The causes of berry aneurysms commonly are connective tissue disorders, polycystic kidney disease and arteriovenous malformations, and less commonly cigarette smoking, drugs abuse, alcoholism, and trauma.[3] In that, aneurysmal rupture is the most fearing consequence and its about 0.7% rupture rate annually. Aneurysmal subarachnoid hemorrhage happens at the rate of 6–16 persons per 100,000 population. Moreover, this rupture of aneurysms accounts for about 0.5% of all deaths. Around 20–30% of patients with aneurysms will have more than 1 aneurysm.

More than 85% of aneurysms are in anterior circulation. Usually these aneurysms are located in the anterior circulation on the Circle of Willis that is in the junction of anterior communicating artery with anterior cerebral artery, and in the junction of posterior communicating artery with the internal carotid artery and also in the division of middle cerebral artery. In posterior circulation, basilar apex is the most common area to be involved. Important symptoms being headache, change in consciousness level, seizure episodes, neck pain and stiffness, speech disturbances, and visual disturbances also.[4] Physical examination should include cranial nerve testing, limb sensations, and power and signs for meningismus, Kernig's and Brudzinski's.

Diagnosis is made by CSF analysis to rule out other differential diagnosis, imaging techniques such as NCCT of brain, MRI brain CT angiogram of brain, and digital subtraction angiography. Medical management includes control of high blood pressure to reduce the risk of rebleeding, with nicardipine and labetalol, adequate pain control, nimodipine the drug to be given in the first 96 h mainly to help decrease the vasospasm along

with anti-seizure medications given.[5] Surgical options are chosen in patients who had rupture of aneurysm or who are at risk for rupture. Three methods are being used such as neurosurgical clipping of the aneurysmal artery, endovascular coiling of the aneurysm and the newer option being flow diverters, and tubular stent-like implants to divert blood flow away from aneurysm. Prognosis depends on age factor, comorbidities, previous neurological conditions, location site of aneurysm, and bleeding extent in aneurysmal rupture patients. Near about 25% with ruptured aneurysms may not survive the 1st day of diagnosis and some patients around 25% may die in the next 6 months of diagnosis due to complications.

CONCLUSION

The prevalence rate of intracranial aneurysms in patients with ADPKD is 9–12%. In many cases, patients with ADPKD may develop intracranial aneurysms and diagnosis of intracranial aneurysms is made only after the symptoms become worse and severe. As the diagnosis gets delayed, in some cases, aneurysms are diagnosed after rupture which is a more serious condition and the cause for death in many patients. As ADPKD being the identifiable risk factor for intracranial aneurysms, the brain imaging screening is recommended in all the cases of ADPKD for early diagnosis and to prevent further complications and mortality.

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