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Case Report

Internal Medicine Section

Calcium on Mitral Valve: Decipher Aetiopathogenesis

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ABSTRACT

We hereby describe an unusual case of a 17-year-old female with severe mitral regurgitation secondary to heavily calcified immobile valve leaflets. Along with the mitral valve, corneas were also calcified, due to congenital systemic metabolic disorder, distal renal tubular acidosis. Histopathology proved that there was no intrinsic pathology of the mitral valve.

Congenital distal renal tubular acidosis with normokalemia presenting with severe mitral and corneal calcification is not known. This case notes important clinical features and is thought to add to the existing knowledge regarding the disease. Patient succumbed to her illness during mitral valve surgery and genetic analysis was not done prior. This is the limitation of our reporting. In this modern era, specific clinical features are also important and of equal value to try and understand molecular and genetic basics of the diseases.

Keywords: Acidosis, Calcinosis, Renal tubular

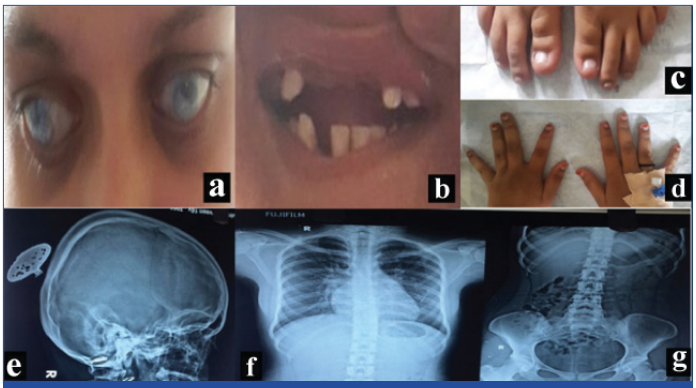
CASE REPORT

A 17-year-old female patient presented with complaints of dyspnoea on exertion which was present for four to five years and had worsened over past six months restricting even her daily household activities with paroxysmal nocturnal dyspnoea since past two months. Her family was more worried about her decreased vision. She could hardly visualize light beam. She was a full term born child with no significant past medical history. Her elder sibling had no significant medical history but her younger sibling died on first day of his life, the cause of which was not known to parents; rest of family history was non contributory. She had regular menstrual periods and fully developed secondary sexual characters.

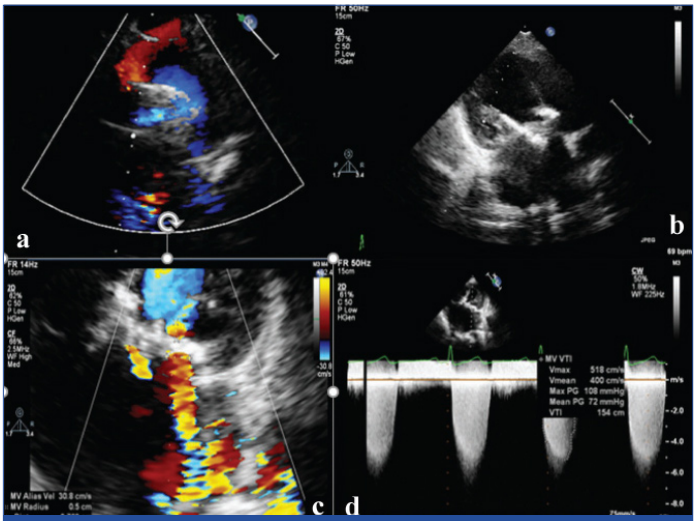
On general examination quite a few things were conspicuous as shown in [Table/Fig-1]. She had leucomatous opacity of both corneas more in the lower half, deformed secondary dentition but no deformity in small bones as might be expected with parathyroid, calcium or phosphorus related metabolic disorders. On auscultation, her heart sounds were soft but there was pan systolic murmur of mitral regurgitation.

Her blood investigation such as complete blood count, renal and liver function tests, serum and urinary calcium, potassium and sodium were normal. Her arterial blood gas analysis showed non-anion gap metabolic acidosis with positive urine anion gap. Her skull, chest and abdominal roentgenogram showed no evidence of any bony abnormality or calcium deposits and moreover ultrasound of kidneys was also unremarkable. Her echocardiography revealed severe calcification of mitral annulus causing almost fixed mitral valve with severe regurgitation and stenosis as well as specks of calcium in subaortic region of myocardium along with Patent Ductus Arteriosus (PDA) as shown in [Table/Fig-2] [Video-1-3]. Our diagnosis was distal renal tubular acidosis with normokalemia of genetic origin. She was posted for mitral valve replacement surgery along with closure of PDA, but unfortunately she succumbed during operation due to poor left ventricular function and lot of resection of mitral annulus to find suitable place for fixing prosthetic valve ring.

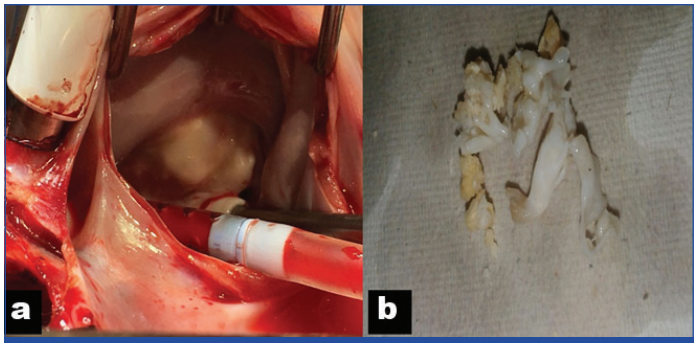
Parents were neither ready for academic post mortem nor affording for genetic analysis. Gross and histopathological examination of resected tissue proved the point that there was calcification in non-diseased heart valves as shown in [Table/Fig-3]. This is the most unusual description of genetic distal renal tubular acidosis with



[Table/Fig-1]: a) Blue sclera and dense calcification involving lower half of cornea; b) Loss of teeth of secondary dentition; c,d) Normal toes and fingers; e-g) Roentgenogram of skull lateral view, chest anteroposterior view and abdomen with pelvis with no calcium deposits.



[Table/Fig-2]: a) Color echocardiography suprasternal view still frame image showing blood flowing through PDA in diastole from descending aorta; b) Echocardiogram still frame image showing severe calcification of mitral valve, annulus extending upto aortic root; c) Color Doppler interrogation in apical 4 chamber view showing severe mitral regurgitation as evidenced by vena contracta of 0.7 mm and Effective Regurgitant Orifice (ERO) area of more than 0.4 cm^2 as calculated by proximal iso velocity surface area (PISA) method; d) Still frame image of apical 4 chamber view with Doppler interrogation across mitral valve showing severe mitral regurgitation as evidenced by V_{max} of more than 4 m/s.



[Table/Fig-3]: a) Intraoperative image of densely calcified mitral valve; b) Gross specimen of mitral valve tissue with almost normal looking chordas and leaflets with blobs of calcium attached to their surface.

calcification inside heart and involving the cornea with sparing of kidneys both anatomically and physiologically.

DISCUSSION

Renal tubular acidosis is a medical disorder of diverse aetiology that leads to acidemia as a result of the failure to acidify urine [1]. They are classified depending upon the site of lesion in renal tubules. Proximal Renal Tubular Acidosis (pRTA) results in glycosuria, aminoaciduria and loss of bicarbonates in urine while distal Renal Tubular Acidosis (dRTA) is due to failure of secretion of H+ ions in urine [2]. Albright F et al., in 1946 described the clinical syndrome of dRTA that consisted of hypokalemia, hyperchloremic metabolic acidosis, inability to lower urine pH below 5.5, nephrocalcinosis and nephrolithiasis [3]. dRTA can occur due to various pathological mechanisms such as impaired proton pump function, abnormal anion exchange, voltage defect or rate dependent dRTA, aldosterone deficiency or backleak of anions in tubules [4,5].

In patients with interstitial renal disease; however, a normokalemic form of dRTA may develop due to isolated failure of the H+-ATPase anion [6]. This patient had no clinical features such as nephrocalcinosis or osteomalacia typical of dRTA rather had intracardiac calcification amounting to severe valvular dysfunction along with PDA which points towards undiscovered genetic syndrome [7,8].

CONCLUSION

Today, in this modern world of medicine, diagnosis is being heavily relied upon sophisticated investigations. Physicians have almost detour from the art of history taking and examination. The case presented here cannot overemphasize the importance of physical examination. We must keep on searching known as well as unknown associations with genetic disorders. As the science of medicine evolves with human race, we might land up in completely different features of previously well recognized genetic disorder otherwise.

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