

Homocystinuria presenting as a calcified right atrial mass

Tahleel Altaf Shera, Naseer Ahmed Choh, Faiz Altaf Shera, Azher Maqbool Khan

Department of Radiodiagnosis and Imaging, Sher-I-Kashmir Institute of Medical Sciences, Srinagar, Jammu and Kashmir, India

ABSTRACT

Homocystinuria is a genetic inborn error of metabolism due to the deficiency of cystathionine β -synthase resulting in increased serum homocysteine and methionine and decreased cysteine which predisposes affected individuals to arterial and venous thromboembolic phenomena. We present a case of homocystinuria who presented to us as a calcified right atrial mass during the evaluation for lower respiratory tract infection. Our case reveals an unusual mix of findings using imaging with multiple detector computed tomography and radiographs.

Keywords: Calcified cardiac mass, cardiac thrombus, homocystinuria

INTRODUCTION

Cardiac calcifications may be nonspecific or they may be indicators of some local or systemic pathology. Cardiac calcifications are most often seen in coronary atherosclerotic disease. Other causes include vascular, toxic, inflammatory, and degenerative disease. In children, causes of cardiac calcifications include chromosomal disorders, intrauterine infections, hyperparathyroidism, and inborn errors of metabolism. Calcifications may also be seen in cardiac tumors, most commonly in myxomas, fibromas, and teratomas.^[1] Intracavitary tumors of the right atrium are rare and may be primary or metastatic. Metastatic tumors of the heart are more common than primary. Metastases usually involve the pericardium and rarely produce intracavitary masses.^[2] In children, the metastatic cardiac disease is even rarer than adults with sarcoma, lymphoma, testicular cancer, and Wilms tumor being common primaries.^[3] Transesophageal imaging frequently adds additional important information to the assessment of mass lesions and should always be considered when image quality is inadequate or pertinent clinical questions remain unanswered with surface imaging.^[4] Computed tomography and cardiac magnetic resonance imaging are being increasingly used in the characterization and evaluation of cardiac masses.

CASE REPORT

A 13-year-old boy was referred to our hospital for the evaluation of mental retardation due to poor performance at school. The parents gave a history of delayed milestones as compared to siblings and multiple respiratory tract infections. There was no history of visual disturbances, trauma, or central venous catheterization. The patient had two elder sisters, both of whom had no known medical issues. There was no history of mental retardation or sudden cardiac death in the first degree relatives of the mother or father. Physical examination revealed a weight for age and height for age below 3rd centile with a high arched palate, slender extremities, and long thin fingers. “Thumb sign” was positive. No kyphosis or scoliosis was present. Ophthalmological examination revealed bilateral ectopia lentis. Chest examination revealed bilateral basal crepts. Chest radiograph showed a calcified mass in the right hilar region and prominent bilateral bronchovascular markings. The patient underwent contrast-enhanced computed tomography for the evaluation of the hilar mass which was found to be a densely calcified mass in the right atrium with a calcified tail extending into superior vena cava (SVC)

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

For reprints contact: reprints@medknow.com

How to cite this article: Shera TA, Choh NA, Shera FA, Khan AM. Homocystinuria presenting as a calcified right atrial mass. *Ann Pediatr Card* 2018;11:214-6.

Access this article online

Quick Response Code:



Website:

www.annalspc.com

DOI:

10.4103/apc.APC_174_17

Address for correspondence: Dr. Tahleel Altaf Shera, Department of Radiodiagnosis and Imaging, Sher-I-Kashmir Institute of Medical Sciences, Soura, Srinagar, Jammu and Kashmir, India. E-mail: tahleel149@yahoo.co.in

and the right subclavian vein. Bronchiectasis was seen in both lungs [Figures 1 and 2]. Echocardiography performed subsequently showed a nonmobile right atrial mass adherent to the endocardium with dense posterior acoustic shadowing. The lack of mobility, the absence of myocardial continuity, no relation to foramen ovale, and morphology of the mass, especially the extension into the SVC suggested that the mass represented a calcified thrombus. Coagulation studies showed prolongation of PT and aPTT with elevation of fasting plasma homocysteine (235 $\mu\text{mol/l}$) and elevated methionine (139.3 $\mu\text{mol/l}$). Radiographs of his hands showed diffuse osteopenia with long fingers [Figure 3]. Elevated levels of homocysteine may be found in deficiencies of vitamin B12 or folic acid and in other disorders of methionine metabolism; however, the severity of homocystinemia with coexisting methioninemia, Marfanoid features, and mental retardation suggest the diagnosis of homocystinuria. The patient was then started on pyridoxine, folic acid, and vitamin B12, and then referred to a higher center for the surgical management of the right atrial mass.

DISCUSSION

Homocystinuria is a genetic inborn error of metabolism due to the deficiency of cystathionine β -synthase resulting in increased serum homocysteine and methionine and decreased cysteine.^[5] Patients with this disorder are initially normal at birth. During infancy, nonspecific features are present, which include developmental delays, and failure to thrive.^[6] Typical manifestations occur later and include mental retardation, osteoporosis, marfanoid characteristics, lens dislocation, and thrombophilic tendencies.^[7] The course of the disease is such that approximately 40% experience a vascular event by the time they are 20 years of age.^[8]

Atrial thrombosis has been reported in patients with central venous catheters, ventriculoatrial shunts, and pacing electrodes.^[1,9] Atrial thrombi are more common than myxomas; however, in cases, where the thrombus is pedunculated, it may be difficult to distinguish between the two on imaging.^[10]

Calcified cardiac masses not related to thrombosis have been reported and are descriptively termed calcified amorphous tumor of the heart.^[11] This patient had a known predisposing factor to thrombus formation, and therefore, the mass likely represented a calcified thrombus. To the best of our knowledge, such a presentation of homocystinuria as a calcified right atrial mass has not been reported. Calcified thrombi are important differentials to be considered when evaluating hyperdense cardiac masses, and an assessment for deranged coagulation profile should be performed in such patients.

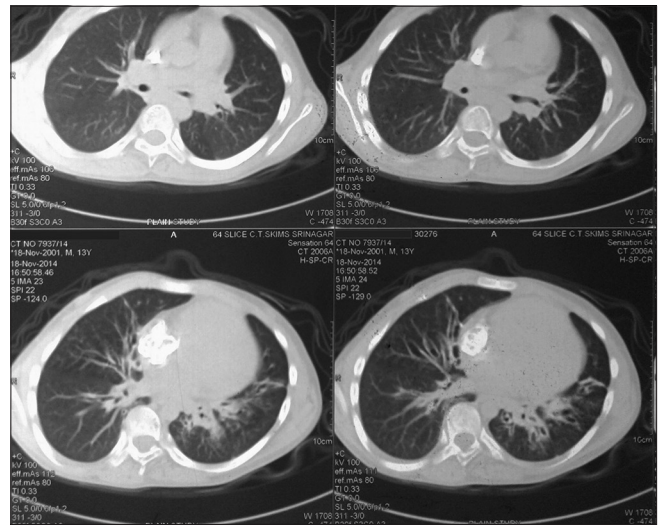


Figure 1: Computed tomography showing the calcified right atrial mass with a calcified tail extending into superior vena cava, bronchiectasis seen in the right middle lobe

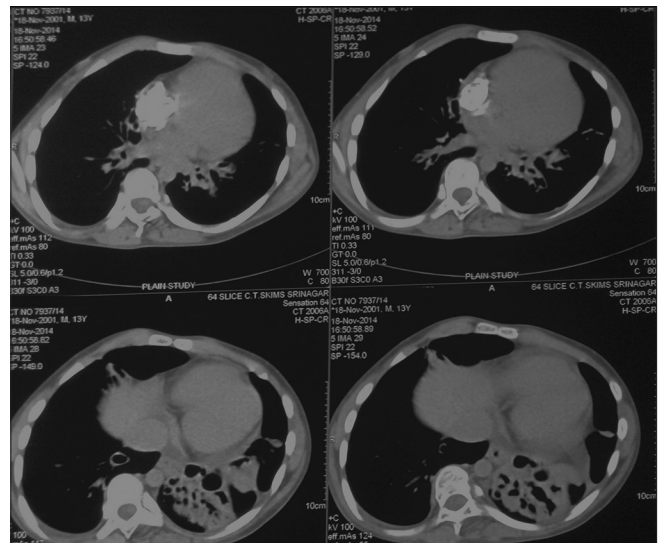


Figure 2: Computed tomography showing bronchiectasis in both lower lobes, with adjacent consolidation on the left side



Figure 3: Diffuse osteopenia in radiograph of both hands

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. Watts MA, Innes BJ. Calcified atrial thrombus: Complication of central venous hyperalimentation. *Tex Heart Inst J* 1984;1s1:76-9.
2. Panidis IP, Kotler MN, Mintz GS, Ross J. Clinical and echocardiographic features of right atrial masses. *Am Heart J* 1984;107:745-58.
3. Tao TY, Yahyavi-Firouz-Abadi N, Singh GK, Bhalla S. Pediatric cardiac tumors: Clinical and imaging features. *Radiographics* 2014;34:1031-46.
4. Peters PJ, Reinhardt S. The echocardiographic evaluation of intracardiac masses: A review. *J Am Soc Echocardiogr* 2006;19:230-40.
5. Mudd SH, Skovby F, Levy HL, Pettigrew KD, Wilcken B, Pyeritz RE, *et al.* The natural history of homocystinuria due to cystathionine beta-synthase deficiency. *Am J Hum Genet* 1985;37:1-31.
6. Eslamiyeh H, Ashrafzadeh F, Akhondian J, Beiraghi Toosi M. Homocystinuria: A Rare disorder presenting as cerebral sinovenous thrombosis. *Iran J Child Neurol* 2015;9:53-7.
7. Mudd SH, Levy HL. Disorders of transsulfuration. In: Stanbury JB, Wyngaarden JB, Frederickson DS, Goldstein JL, Brown MS. (editors). *Metabolic and Molecular Bases of Inherited Disease*. New York: McGraw-Hill; 1983.
8. vd Berg W, Verbraak FD, Bos PJ. Homocystinuria presenting as central retinal artery occlusion and longstanding thromboembolic disease. *Br J Ophthalmol* 1990;74:696-7.
9. Gabriele OF, Clark D. Calcified thrombus of the superior vena cava. Complication of ventriculoatrial shunt. *Am J Dis Child* 1969;117:325-7.
10. Scheffel H, Baumüller S, Stolzmann P, Leschka S, Plass A, Alkadhi H, *et al.* Atrial myxomas and thrombi: Comparison of imaging features on CT. *AJR Am J Roentgenol* 2009;192:639-45.
11. Reynolds C, Tazelaar HD, Edwards WD. Calcified amorphous tumor of the heart (cardiac CAT). *Hum Pathol* 1997;28:601-6.