Case Report

Pulmonary Arterial Hypertension in a 14-Year-Old Girl: Diagnostic Challenges and Management

Efe Abolodje[1], Efemena Edoja

[1] Department of Paediatrics, DELSUTH, Oghara
Email: efexabo@gmail.com

ABSTRACT

Pulmonary hypertension carries a high morbidity and mortality especially when presentation occurs late. Due to its subtle manner of presentation and high cost of care, diagnosis is often delayed and treatment is beyond the reach of most affected Nigerians. A 15-year-old girl who presented with easy fatigability of 2 years duration and dypnoea on exertion of 3-weeks duration. She had features of systemic and pulmonary congestion on examination. An echocardiogram done revealed supra-systemic pulmonary pressure with a massively dilated and poorly functional right ventricle. A large secundum ASD with bi-directional flow of blood was also noted. She was commenced on anti-failure regimen and treatment for pulmonary hypertension. Due to high cost of care, combination therapy for pulmonary hypertension could not be sustained and patient eventually succumbed to the illness.

INTRODUCTION

Pulmonary hypertension (PHT) is defined as mean pulmonary arterial pressure greater than 25mmHg at rest.[1] The condition is characterized by exercise intolerance in the early stage and right ventricular failure in the late stage.[2] It is one of the most challenging aspects of cardiology with limited success in management especially with late presentation. Pulmonary hypertension carries a high morbidity and mortality especially when presentation occurs late.[3] Its management is very complex, expensive and requires specialized cardiac centres. The diagnosis is often made late due to its non-specific presentation. The index case is a 15-year-old girl that presented with right ventricular failure due to pulmonary hypertension with poor response to medical therapy.

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Case Presentation

A 15-year-old girl with easy fatigability of 2 years duration, fast breathing, orthopnoea and paroxysmal nocturnal dyspnoea of 3 weeks duration. Past medical history revealed that from her early childhood she had several admissions for cough and difficulty in breathing. She was observed to be sluggish and not quite as active as other children. However, conditions worsened in the last 2 years when minor daily activities like walking and singing resulted to breathlessness with worsening orthopnoea that requires her leaning forward on a chair to sleep.

Clinical findings

She was wasted and cyanosed with grade 4 digital clubbing and generalized pitting oedema.

The PR was 120bpm, BP-90/60mmHg, JVP was 12cm, and apex beat was displaced to 7th intercostal apace along the anterior axillary line. Additional findings included right parasternal heave, S3 gallop rhythm and a pan-systolic murmur loudest at the left lower sternal border. The RR-40cpm, dyspnoeic at rest with bi-basal crepitation. She had ascites demonstrable by fluid thrill with tender hepatomegaly. Her SPO_2- 70% and on oxygen appreciated to about 80%.

Diagnostic assessment

Chest radiograph showed cardiomegaly and on echocardiogram revealed severely dilated right ventricle, flattening of the interventricular septum, a large pulmonary artery, dilated IVC, measuring 20.0mm with poor collapse during inspiration, a large Secundum ASD measuring 39.6mm with bidirectional flow. The mean pulmonary pressure estimated from the pulmonary regurgitant jet was 46.85mmHg and the sPAP estimated from tricuspid regurgitant jet and RAP was 102mmHg (sPAP + RAP). Right ventricular function was poor with TAPSE of 11.0mm and fractional area change (FAC) of 10.0%. The left ventricle appears squashed by the supra-systemic pulmonary pressure; however, the ejection fraction was adequate and no significant left valvular heart disease. Screening for HIV and schistosomiasis were negative. Chest CT scan and Right Heart Catheterization were not done due to financial constraints.

The diagnosis was right ventricular failure 2^0 pulmonary arterial hypertension with background large ASD (?Eisenmenger Syndrome). Figure 1-4 show the findings on echocardiogram.

Therapeutic intervention

Initial treatment was with antibiotics for pneumonia, diuretics and ACE inhibitors for heart failure and Sildenafil for PHT. The D dimer was elevated and as a result patient was commenced on anticoagulation therapy with heparin and warfarin. The SPO_2 improved to 91%, however, pulmonary pressures remained supra-systemic. Patient was discharged at WHO functional class III for follow up in the cardiology clinic. After 6 months of monotherapy with sildenafil, borsetan was introduced. However, due to the high cost of the medications, it was stopped after 2 weeks. Parents who were subsistence farmers were counselled on the poor prognosis of the child's condition and the need for heart-lung transplants. The condition was also discussed in a webinar session with a US based NGO that partners with some health facilities in Nigeria to provide surgical treatment for children with heart conditions. Due to the difficulty with donors, and the challenges of management, the case did not get the needed attention during the discussions. The patient continued to decompensate with features of systemic and pulmonary congestion with poor response to anti-failure therapy. Patient succumbed to the illness after about a year of treatment.

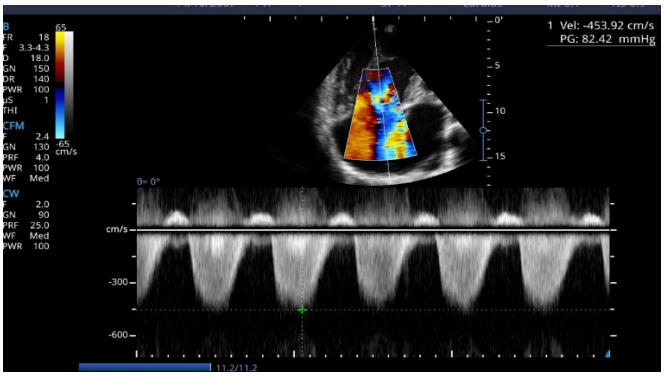

Figure 1: TR jet from Apical 4 Chamber View

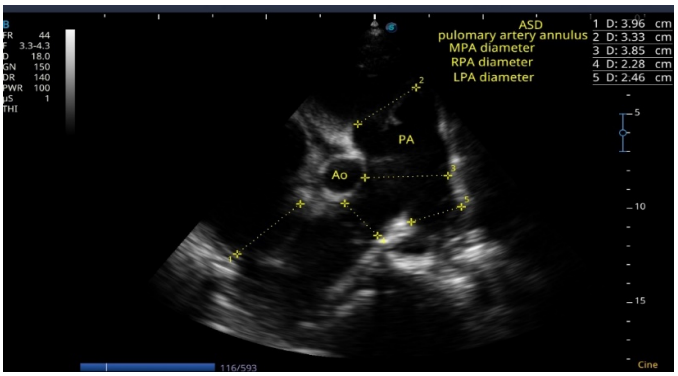

Figure 2: Pulmonary Artery from Parasternal Short Axis View

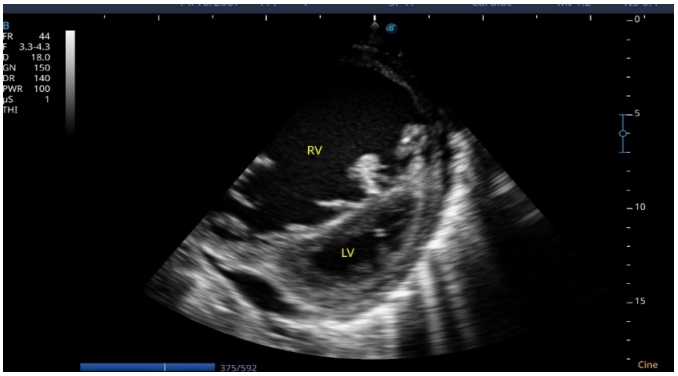

Figure 3: Right Ventricle versus Left Ventricle

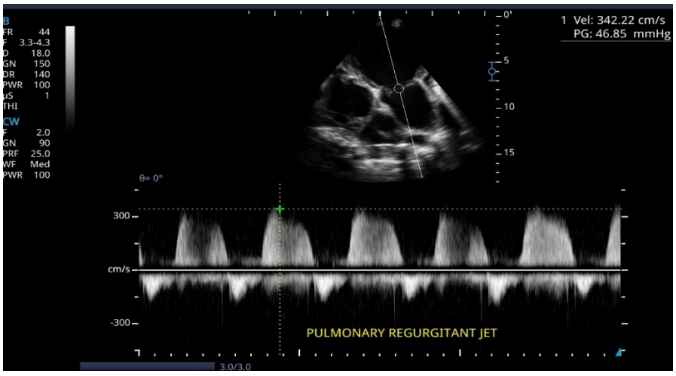

Figure 4: Pulmonary Regurgitant Jet for Mean Pulmonary Artery Pressure Estimation

DISCUSSION

Pulmonary hypertension (PHT) is a bag of several diseases. It encompasses various conditions that share the common outcome of high blood pressure in the pulmonary arteries, ultimately leading to RV failure and death.[4] The WHO classifies PHT into 5 categories and groups share histology, pathophysiology and likely specific treatment or prognosis.[4] Early stage of the disease often goes unnoticed until irreversible damage has been done to the pulmonary vasculature and the right ventricle. The index case presented in the terminal stage of the disease, a stage that poorly responds to medical therapy.

Pulmonary arterial hypertension (PAH) is defined as a mean pulmonary arterial pressure greater than 25 mmHg at rest, with a normal pulmonary artery wedge pressure less than 15 mmHg and an increased pulmonary vascular resistance greater than 3 Wood/m^2. There are few studies that have reported the prevalence/incidence of PHT in children and they are mostly in the developed world. The incidence of idiopathic PAH in the national registry from the United Kingdom was 0.48 cases per million children per year and the prevalence was 2.1 cases per million children.[5] In the Netherlands, annual incidence and point prevalence averaged 0.7 and 2.2 cases per million children respectively.[6] The prevalence is higher in conditions such as HIV infection, sickle cell anaemia and in congenital heart diseases (CHD).[7] The prevalence of PHT in HIV is reported to be about 2,500 times higher than the general population.[8]

The index case screening for HIV and schistosomiasis was negative. However, a large secundum ASD was discovered on echocardiogram. Atrial septal defects, due to their insidious presentation, can go undetected because these patients do not present with overt heart failure in childhood due to the fact that the atria are low pressure chambers. Though pulmonary hypertension can occur at any age in life following a CHD, they commonly occur in the 4th decade of life.[9] The condition is also described as Eisenmenger syndrome. It is not quite clear if the pulmonary hypertension was due to the background ASD or if it was idiopathic

A similar case has been reported in the literature in 35-year-old women who was known to have OS-ASD measuring 23mm. Patient developed PAH with PASP-85*mmHg*, MAP-55*mmHg* and a PVR of 3.38woods. She had combination therapy with ambrisentan and tedalafil for 3 months with significant improvement in her WHO functional class (from IV to II) and her 6-minute walk (110-255m). The response to the combination therapy of endothelin receptor antagonist and 5[1]- phosphodiesterase inhibitor occurred after 3-months.[10] Another case reported was a 19-year-old boy with OS-ASD that was complicated by severe PAH, reported in Nigeria.[11] This case was lost to follow-up after discharge, most likely due to financial constraint. The index case could not afford combination therapy and did not respond to monotherapy. Guidelines for managing patients with PAH that failed to respond to monotherapy state that two or three agent combination therapy with 5-phosphodiesterase inhibitor, endothelin receptor antagonist and prostacyclin analogues should be used.

Several investigations are required to make the diagnosis of PAH due to the fact that there are many other secondary causes. As part of evaluation, patients are required to undergo a right heart catheterization and a vasoreactivity test using inhaled nitric oxide or intravenous epoprostenol during the procedure. Those who respond are commenced on calcium channel blocker, however, our patient did not have a RHC due to lack of funds. There are also limited centres that provide this level of care in Nigeria with few specialists. Additionally, invasive procedures such as RHC require general anaesthesia, and these can be catastrophic for patients with PHT. Mortality can be as high as 23% during some major procedures.[12] A collaborative team has to pre-evaluate, risk assess & optimize and give parents enough time to weigh the risk of procedures. PAH in children without intervention has a

median survival of about 10 months after diagnosis.[13] Heart lung transplantation which is the definitive management in terminal stage of the disease is challenged by availability of donors, lack of expertise, the risk of surgery and cost of care.

CONCLUSION

Management of PAH requires a team of dedicated experts in the field of cardiology in a specialized centre. The diagnosis is often made late but exertional dyspnoea with no apparent cause can serve as a clue. ECG which is relatively available in most settings and affordable can serve as a ready screening tool

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