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Massive Pericardial Effusion in Down Syndrome with Supravalvar Pulmonary Stenosis Associated to Left Pulmonary Collapse

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Abstract

Background: Pericardial effusion (PE) in Down syndrome (DS) patients usually occurs secondary to hypothyroidism, but we have no report of massive pericardial effusion in euthyroid Down syndrome patient with supravalvar pulmonary stenosis. **Case Presentation:** We reported an 11-month-old male with Down syndrome with massive pericardial effusion that had conservative management with levothyroxin who presented with cyanosis, respiratory distress and dominantly left upper lobe pulmonary collapse. There was no response to medical management by antibiotic therapy, O₂ therapy with hood and chest physiotherapy after five days. Pericardiocentesis guided echocardiography performed on the sixth day after admission and 180 cc transudated pericardial fluids has extracted. Clinical and paraclinical findings were relieved dramatically 12-24 hour after pericardiocentesis. However, Angiography showed supravalvar pulmonary stenosis (SPS). **Conclusion:** We assume that the chronic compression effect of massive pericardial effusion may be a major cause of SPS. [GMJ. 2014;3(1):54-58]

Keywords: Down syndrome; pericardial effusion; supravalvar pulmonary stenosis; pulmonary collapse; Pericardiocentesis

Introduction

Down syndrome (DS), which is normally caused by trisomy 21, is the most common chromosomal defect, and its incidence in the United States is one per 733 live births [1]. Due to the extensive number of chromosome 21 genes [2], there is an extremely high incidence of congenital anomalies such as important cardiac and gastrointestinal malformations in individuals with DS [3]. Pericardial effusion is an abnormal accumula-

tion of fluid in the pericardial cavity that can lead to negative affect on heart function. Massive pericardial effusion in Down syndrome (DS) predominantly correlated with congenital hypothyroidism [4], but in the pediatric field, it may have correlation with infectious diseases, usually viral infections [5], acquired and secondary to severe Primary Pulmonary Hypertension (PPH) that is associated with right heart failure [6]. Dinleyici *et al.* report four infants with DS who have PE due to congenital hypothyroidism that PE was complete-

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ly resolved in all cases with thyroxin therapy without pericardiocentesis [7]. However, according to our literature investigations we have no report of massive pericardial effusion in euthyroid Down syndrome patient without viral infection and PPH with supravalvar pulmonary stenosis (SPS).

Case Report

An 11-month-old infant with Down syndrome admitted to Be'sat hospital of Hamadan because of cyanosis and respiratory distress. In physical examinations pectus carinatum, tachypnea, (Respiratory rate=66/min), tachycardia (Heart Rate= 180beats/min), and cyanosis at crying (SPO2 75%-80%) were revealed. The peripheral pulse and blood pressure (90/60 mmHg) were normal and pulsus paradoxus was negative and negative Ewart sign. On auscultation, the left lung sound was decreased and the heart sounds were distant. He had history of admission with similar symptoms in another center at 8.5 months of age that he underwent pericardiocentesis and Polymerase Chain Reaction (PCR) of adenoviruses and enteroviruses were negative. In spite of the normal thyroid, function test

Levothyroxin has started this time.

In addition, he had one admission because of upper respiratory infection in this center three weeks ago, who had overt cardiomegaly with normal lung field in chest X-ray (Figure-1) and moderate to severe pericardial effusion in echocardiography.

At first, the patient managed by O2 4-5 lit with Hood, Intravenous therapy, Ceftriaxone 75mg/kg. Laboratory test showed a normal Cell Blood Count (CBC), normal thyroid function test (TFT), electrolyte and renal function test. The ECG depicted sinus tachycardia, pulsus alternant, Right axis deviation, and QR pattern in the right pericardial leads (significant Right Ventricular Hypertrophy; RVH). The chest x ray illustrated considerable left hemi thorax opacity and cardiomegally with shift to the left mediastan and probably mild interstitial pulmonary edema. Conventional echocardiography showed significant RVH in figure-2 (TR gradient was 65 mmHg), no congenital heart disease and moderate to severe pericardial effusion, (No tamponad evidence). Thoracic Computer tomography (CT) scan demonstrated totally left upper lobe and partially left lower lobe collapse with considerable pericardial effusion (Figure-3).

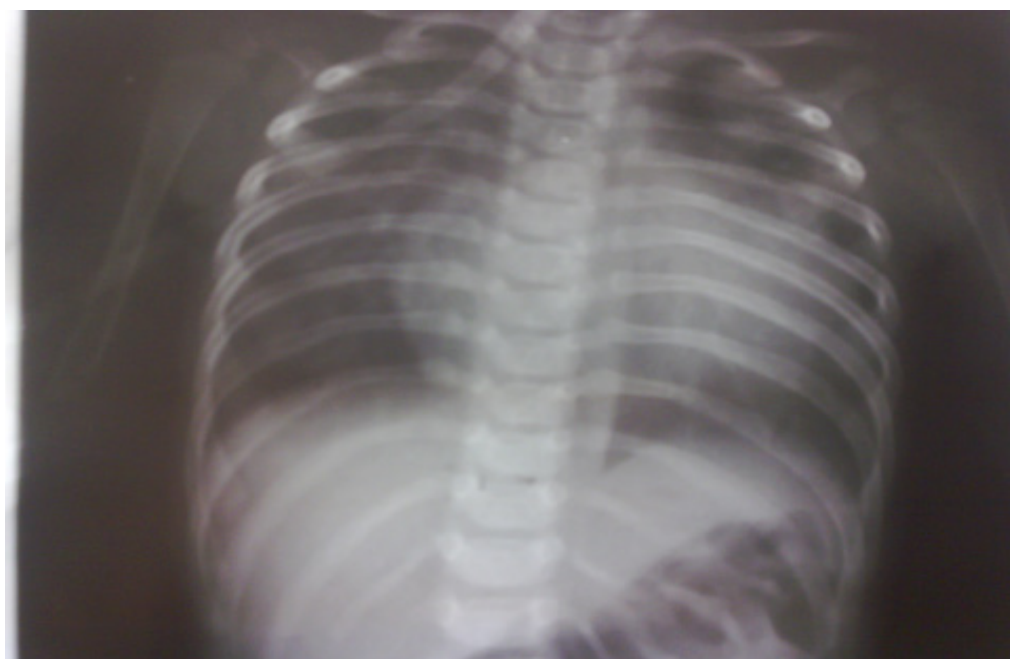


Figure1. Overt cardiomegaly with normal lung field in an 11 months old boy case of Down's syndrome



Figure 2. An 11-Month-Old Infant with Down Syndrome, Significant Right Ventricular Hyperplasia.

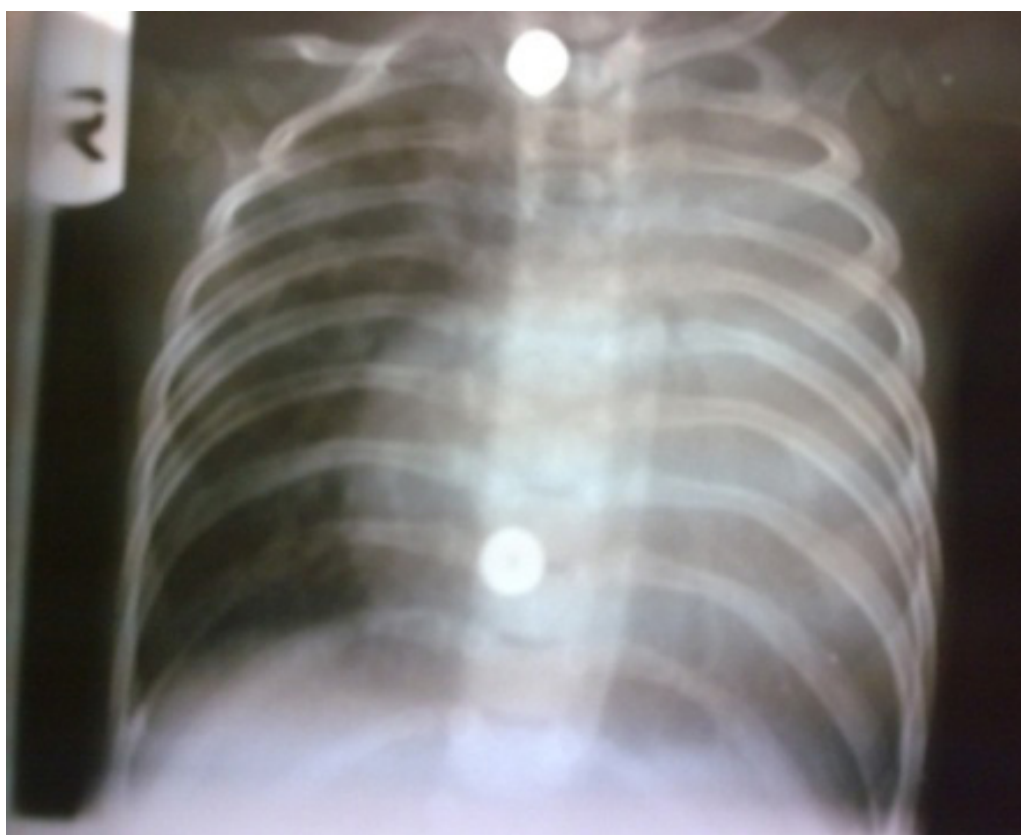


Figure 3. An 11-Month-Old Infant with Down Syndrome, Totally Left Upper Lobe and Partially Left Lower Lobe Collapse with Considerable Pericardial Effusion

There was no response to medical management and chest physiotherapy after five days. Pericardiocentesis guided echocardiography performed on the sixth day after admission and 180 cc transudated pericardial fluids has extracted. Clinical and paraclinical findings were dramatically cured 12-24 hrs after pericardiocentesis.

Conventional echocardiography showed significant RVH and because of high prevalence of PE in patients with severe PPH we suspected to PPH [6], so right ventricular angiography, as the gold standard of diagnosis of PPH, was done, but the pressure of right ventricle was 100/65 mmHg and left pulmonary artery pressure was 35/15 mmHg that rule out PPH, but RV injection at AP and lateral views showed supra-valvar pulmonary stenosis (type 1 of peripheral pulmonary stenosis; Figure-4).

Discussion

Down syndrome (DS), which is normally caused by trisomy 21, is the most common chromosomal defect, and its incidence in the United States is one per seven-hundred and thirty-three live births [1]. Cardiac and gastrointestinal malformations are important anomalies in individual with DS [3]. Massive pericardial effusion in Down syndrome (DS) predominantly correlated with congenital hypothyroidism [4], but in the pediatric field, it may be correlated with infectious diseases, usually viral infections [5]. This DS

patient had PE with an unusual presentation. In this case the first working diagnosis was post viral pericardial effusion so diagnostic pericardiocentesis done in another center at 8.5 month ago that PCR of adenoviruses and retroviruses were negative, then thyroid function test (TFT) was done that it was normal but levothyroxin was started since that time. In this admission, our patient had revealed Pectus carinatum, tachypnea, tachycardia, and cyanosis at crying. The combination of pericardial effusion and bradycardia might suggest hypothyroidism [8], but our patient had PE and tachycardia and laboratory test showed a normal TFT. Dinleyici et al. report four infants with DS who have PE due to congenital hypothyroidism that PE was completely resolved in all cases with thyroxin therapy without pericardiocentesis [7]. Williams et al. report a child with DS and long-standing severe hypothyroidism had a massive pericardial effusion without cardiac tamponade. The effusion completely resolved with medical treatment without pericardiocentesis [9]. In our case, PE completely resolved after pericardiocentesis and there was not recurrence with levothyroxin admission after one year, we propose a probability that pericardial effusion is initially occurs secondary to hypothyroidism with suboptimal treatment and exaggerated with some other factors. On the other hand, the ECG and conventional echocardiography showed sinus tachycardia and significant RVH. Considerable left hemi-tho-

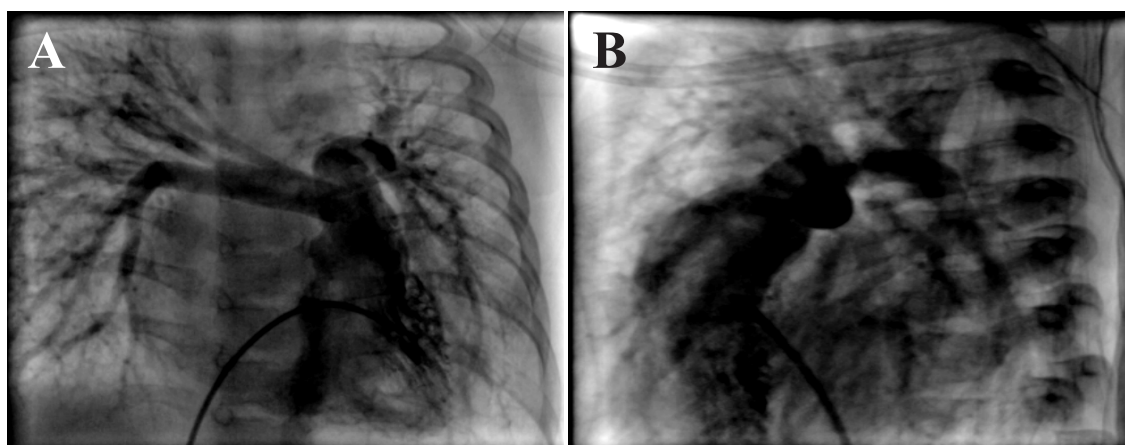


Figure 4. Right Ventricular Injection at AP (A) and Lateral (B) Views Showed Supra-valvar Pulmonary Stenosis (type 1 of peripheral pulmonary stenosis)

rax opacity and cardiomegaly with shifting to the left mediastin have seen in the chest X-ray and thoracic CT scan showed totally left upper lobe and partially left lower lobe collapse with massive PE with mild interstitial edema in the right lung. At first, we thought that severe pulmonary hypertension might be the reason for RVH and PE that were associated to pectus carinatum leading to compression effect on upper lobe of left lung. While in the normal chest, massive pericardial effusion has been associated with left lower lobe compression and positive Ewart sign [10]. After the evaluations just showed only RVH, we considered that PE exacerbation following end stage pulmonary hypertension, due to high prevalence of PE in patients with severe PPH and associated with right heart failure [6]. But surprisingly, Angiography and catheterism showed that the pressure of pulmonary vessels were normal which ruled out PPH, but RV injection at AP and lateral views showed LPA stenosis. Although the cause of the LPA stenosis is un-

clear here, we think chronic compression effect of PE may be the cause of LPA stenosis.

Conclusion

Hypothyroidism is a common cause of symptomatic PE in DS patients, so any child with DS who present with cardiomegaly without CHD should suspected of having PE due to hypothyroidism and echocardiography examination and TFT should be performed immediately because of prevalence of hypothyroidism and symptomatic pericardial effusion in these patients. However, if the TFT was normal and we cannot find any underlying disease, it is necessary to searching for other ominous cause of PE and we can use of angiography and catheterism if there was evidence of PPH. We must know if PE was massive and does not resolve with medical therapy that we must use pericardial drainage as soon as possible because of the risk of tamponade and chronic compression effect.

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