Congenital Absence of Pericardium and Straight Back Syndrome: Do They Have Similarities?
M Ali Ostovan, R Mollazadeh

Citation

Abstract
The straight back syndrome, consisting of loss of normal upper thoracic spinal curvature is not an uncommon entity. In contrary congenital defects of the pericardium is a rare disease (1/10000 autopsies). In this report we present a case presented with chest pain whom after initial work up was presumed to have congenital absence of pericardium but further investigation uncovered the diagnosis as straight back syndrome. Here in we review the literature for the possible similarities between these two entities and the possible reason for our first misdiagnosis.

INTRODUCTION
The straight back syndrome, consisting of loss of normal upper thoracic spinal curvature associated with cardiac murmurs which was previously considered a form of 'pseudo heart disease', has been attributed to squashing of the heart in the reduced anteroposterior diameter of the chest. On the other hand complete or partial absence of the pericardium is an uncommon congenital anomaly with various complications and presentations. Herein we report a young girl who presented with chest pain and dyspnoea, whom we thought to have congenital absence of pericardium primarily but later found to suffer from straight back syndrome.

CASE PRESENTATION
A 16-year-old girl with no previous cardiac problems came to the outpatient clinic with ill-defined chest pain. She declared that she had this pain for many months. She complained of dyspnoea but not easy fatigability. Her cardiac examination was completely normal. Her electrocardiogram showed poor R wave progression. Postero-anterior chest-X ray showed cardiac levoposition with borderline cardiac size (Figure 1).

Figure 1
Figure 1: Posteroanterior chest X-ray of the patient. Heart is moved to left hemithorax, cardiac size is enlarged, left heart border is straightened.

Unusual echocardiographic views (Figure 2) and paradoxical septal motions (Figure 3) during transthoracic echocardiography augmented the suspicion of congenital absence of pericardium, so magnetic resonance imaging was performed. Contrary to our expectation, pericardial contour was intact (Figure 4).
Figure 2
Figure 2: Transthoracic echocardiogram of the patient in apical 4-chamber view that has abnormal view; right atrium and right ventricle falsely seem to be dilated.

RA: Right Atrium, RV: Right Ventricle, LA: Left Atrium, LV: Left Ventricle

Figure 3
Figure 3: M-mode transthoracic echocardiography of the patient in parasternal short axis view that shows paradoxical septal motion.

Figure 4
Figure 4: ECG gated T weighted spin echo magnetic resonance image showing no interposition of lung between aorta and pulmonary artery (arrow): Ao: Aorta, PA: Pulmonary Artery

A thorough physical re-examination revealed flat back; a lateral chest x-ray confirmed the presence of obliteration of thoracic kyphosis of the spine.

DISCUSSION
Except for one report of coexistence of congenital absence of pericardium and straight back syndrome, these two entities seem to be unrelated. Herein we review the literature for their similarities and differences.

ETIOLOGY
Congenital pericardial absence is due to premature atrophy of the left common cardiac vein with insufficient blood supply to the pleuropericardium leading to its agenesis. Male/female ratio is 3:1. This entity was described in the context of different syndromes. HLA typing in straight back syndrome indicates that it is inherited as an autosomal dominant condition and suggested that the antigenic determinants may be located on chromosome 6.

CLINICAL MANIFESTATIONS
Congenital absence of the pericardium is an uncommon finding ((1/10,000 in autopsies) comprise partial left (70%), partial right (17%) or total bilateral (extremely rare) pericardial absence). Most patients are asymptomatic are discovered incidentally during cardiac surgery for an unrelated condition or at postmortem examination. Symptomatic patients may experience non-exertional paroxysmal stabbing chest pain. However homolateral cardiac displacement and augmented heart mobility impose
an increased risk for traumatic aortic dissection. Partial left
sided defects can be complicated by cardiac strangulation
caused by herniation of the left atrial appendage, atrium or
ventricle through the defect (chest pain, shortness of breath,
syncope, or sudden death). About 30% of patients have
additional congenital abnormalities. Partial absence of
pericardium is often associated with other cardiac anomalies
including atrial septal defect, bicuspid aortic valve, mitral
valve stenosis, patent ductus arteriosus, tetralogy of Fallot
or pulmonary malformation. Coexistence with recurrent
pulmonary infections and endocarditis are also reported.

On the contrary, straight back syndrome usually comes to
attention due to cardiac murmurs. Referring diagnosis
include atrial septal defect, pulmonic stenosis and rheumatic
heart disease. In one study half the patients were
asymptomatic; others had dyspnea after brisk effort,
palpitation and atypical chest pain.

PHYSICAL FINDINGS
In patients suffering from pericardial absence clinical
examination may show a significantly displaced apical
impulse, which may be palpated in the anterior or
midaxillary line. Basal ejection murmurs, apical midsystolic
clicks and murmurs may also be found. Increased splitting of
the second heart sound may be due to the presence of a right
bundle branch block that is frequently associated with this
defect.

In straight back syndrome decrease in antero-posterior chest
dimensions due to loss of thoracic kyphosis. Other findings
in order of frequency are: palpable pulmonary artery
pulsation, ejection systolic murmurs in pulmonic area,
palpable impulse in left lower sternal border, accentuation
and delay in tricuspid valve closure, exaggerated inspiratory
splitting of the second heart sound, increase in amplitude of
aortic and pulmonary closure. None of patients had thrill in
pulmonic area nor did any of them have diastolic
murmurs. Apical systolic click and murmurs are usually due
to coexistent mitral valve prolapse.

ELECTROCARDIOGRAM
Electrocardiographic manifestations of pericardial absence
are: bradycardia, right bundle branch block, poor R-wave
progression secondary to leftward displacement of the
precordial transitional zone, and prominent P-waves in the
mid-precordial leads which denote right atrial overload.

ECG manifestations of straight back syndrome are right
bundle branch block in V1 and small terminal r waves in aVR

CHEST X RAY
Chest X-ray findings due to pericardial absence are
levoposition of the heart, flattening and elongation of the left
ventricular border, and a lucent area between the diaphragm
and the heart, or aorta and pulmonary artery due to lung
interposition.

Surprisingly patients with straight back syndrome may have
some findings in the Chest X ray: pancake appearance
simulating cardiomegaly, levoposition of heart, prominence
of the main pulmonary artery. In all patients both the
antero-posterior diameter and the ratio of antero-posterior to
transsthoracic diameter were below the normal mean. On
the lateral chest radiograph the distance from the middle of
the anterior border T1 to a vertical line connecting T4 and T12
was found to be significantly reduced compared to controls,
and a value less than 1.2 cm is the established criteria for
straight back.

ECHOCARDIOGRAPHIC FINDINGS
Due to pericardial absence heart forms a “teardrop”
appearance due to elongated atria and relatively bulbous
ventricles because of suspension of the heart from its basal
pedicle. Findings in order of decreasing frequency are:
unusual windows (a higher and more lateral window is
required), cardiac hypermobility (cardioptosis), abnormal
swinging motion, paradoxical or flat systolic motion of the
ventricular septum with normal systolic thickening. Right
ventricular overload with reduction in right atrial dimension
may be evident with decreased tricuspid valve excursion.
Increased diastolic wave velocities in both the superior vena
cava and pulmonary veins on Doppler examination with
decreased caval systolic wave velocity suggest accentuation
of changes in systemic over pulmonary venous return (esp.
in partial defects).

Echocardiograms were normal in 36% and abnormal in 64% of
patients (mitral valve prolapse (MVP) in (58%) and
bicuspid aortic valve in (6%)) of patients with straight back
syndrome. Strong association of MVP with this syndrome
was proposed previously. One of the important issues on the
long-axis view of tomographic echocardiography is the
aorto-septal angle of patients with straight back syndrome
which was wide and thought to be due to clockwise rotation
of the heart rather than right ventricular enlargement.

11.
OTHER TECHNIQUES

ECG-gated MRI and spiral chest CT-scan are two other diagnostic tools to diagnose pericardial absence. ECG gated T1 weighted spin echo magnetic resonance image is highly sensitive and is capable of demonstrating the absence of the preaortic pericardial recess in pericardial absence which is present in normal hearts.

TREATMENT

Complete cases of pericardial absence require no intervention unless complications occur. Patients with debilitating symptoms may benefit from pericardioplasty. However, in partial defects, when herniation occurs or is threatened, extension of the defect by the pericardiectomy, or pericardioplasty may be performed but in straight back syndrome therapy is directed toward specific coexisting conditions (e.g. MVP).

CONCLUSION

Although congenital absence of the pericardium and straight back syndrome seem to be unrelated, it is important to know their similarities: usually asymptomatic, basal systolic murmurs, right bundle branch block in ECG, levoposition of heart in postero-anterior chest X-ray. Furthermore we want to propose that echocardiography of the patient with straight back syndrome could have abnormal view and paradoxical septal motion probably due to clockwise rotation of heart. Not knowing these issues lead us to misdiagnose patient as congenital absence of pericardium primarily.

CORRESPONDENCE TO

R.Mollazadeh, Nemazei Hospital, Shiraz, Iran E mail: mollazar@yahoo.com Tel: +98 917-313-3749 Fax: +98-711-6261089 P.O. Box: 71435-1414

References

Author Information

Mohammad Ali Ostovan
Cardiology Department, Nemazee Hospital

Reza Mollazadeh
Cardiology Department, Nemazee Hospital