CASE REPORT
Korean Circulation J 2005;35:411-414
ISSN 1738-5520

A Case of Complex Congenital Anomaly Combined with Congenital Pseudoarthrosis of the Left Clavicle: Is it a New Syndrome?

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ABSTRACT

Congenital pseudoarthrosis of the clavicle is a rare disorder of the shoulder girdle, with only approximately 200 individual cases having been reported in the world literature to date. A persistent left superior vena cava (SVC) is the most common thoracic venous anomaly, and has been observed in 0.3% of the general population. Mesocardia and an aneurysm of the main pulmonary artery, associated with bicuspid pulmonary valves, are both extremely rare entities. We report the first case of a 23 year-old man with the above mentioned complex congenital anomaly. **(Korean Circulation J 2005;35:411-414)**

KEY WORDS : Pseudoarthrosis ; Superior vena cava ; Pulmonary valve ; Aneurysm.

Introduction

Congenital pseudoarthrosis of the clavicle is a rare disorder, with only approximately 200 individual cases having been reported in the world literature to date. It should be differentiated from the more common form of pseudoarthrosis that occurs secondary to a fractured clavicle.[1)2)] The presence of a left superior vena cava (SVC) has been reported to occur in approximately 0.3% of the general population. They drain via the coronary sinus to the right atrium in more than 90% of patients, but rarely to the left atrium when associated with other congenital heart diseases.[3-5)] The prevalence of mesocardia is 2 in every 1000 deliveries (0.2%), so is very rare.[6)] Congenital abnormalities of the pulmonary valve are rare; 21 in 3600 (0.58%) consecutive autopsies. There are very few reports of bicuspid pulmonary valves, the majority of which are associated with congenital heart disease,[7)8)] combined with a pulmonary artery aneurysm.[9)]

We report the first case of a 23 year-old man with the above mentioned complex congenital anomaly.

Received : January 28, 2005
Accepted : March 14, 2005
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Case

A 23 year-old man, a soldier, presented with numbness of the left arm when he shouldered a pack. There was no history of trauma or other diseases. On examination, he was of average build; with a blood pressure of 110/80 mmHg and pulse rate of 52 beats/minute. The breath sounds in both lung fields were clear, and regular heart beats, without murmur, were noted on auscultation. Chest palpation revealed the absence of the distal 3/4 of the left clavicle (Fig. 1). There was no limitation of motion in the left shoulder. A chest X-ray showed mesocardia (Fig. 2). The 12-lead ECG revealed a normal sinus rhythm and right axis deviation. Tests of his serum VDRL, ANA, Anti-dsDNA and rheumatoid factor were all negative, and the CRP was also within the normal range. Transthoracic and transesophageal echocardiography revealed bicuspid pulmonary valves, a normal pulmonary artery pressure, mild pulmonary regurgitation and aneurysmal dilatation of the main pulmonary artery, without stenosis (Fig. 3A). No other cardiac abnormality was observed, with the exception of a huge coronary sinus along the posterior side of the left atrium (Fig. 3B). A 3-dimensional CT scan showed a dilated main pulmonary artery, with a diameter of 4.4 cm (Fig. 4), and a persistent left SVC draining into the coronary sinus (Fig. 5). There were no stenotic lesions of the subclavian artery and vein.

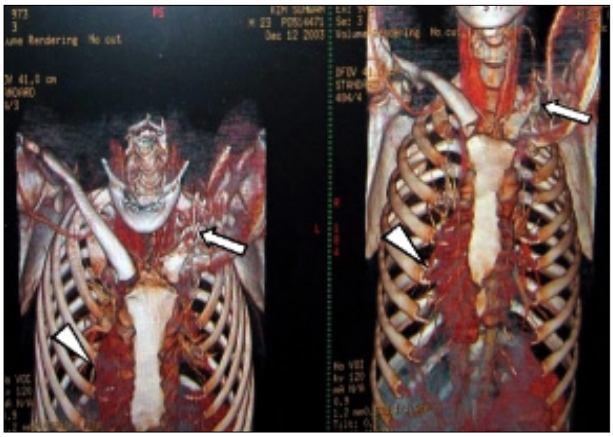

Fig. 1. The 3D-Chest CT: the apical lordotic and AP views, showing congenital pseudoarthrosis of the left clavicle. The distal 3/4 of the left clavicle is absent (arrow) and mesocardia is shown (arrow head).

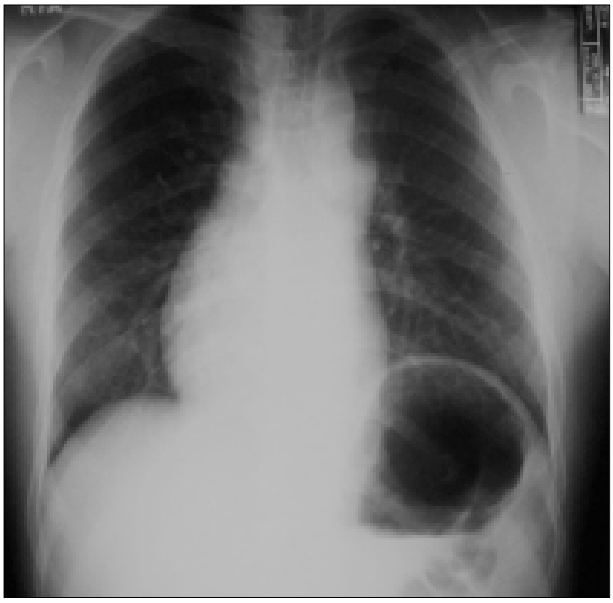

Fig. 2. Chest PA. Mesocardia with situs solitus.

Discussion

The etiology of congenital pseudoarthrosis is obscure, but some theories have been proposed. One, namely the vascular theory, proposes that it is due to the pressure exerted by the subclavian artery, but another suggests it is due to the separation of the two primary ossification centers.[2)10)] The abnormality occurs almost entirely on the right side, with involvement of the left side usually only occurs with dextrocardia and situs inversus. Bilateral cases are typically reported with genetic problems.[1)10-12)] Surgical management may be chosen due to their unaesthetic appearance and the development of thoracic outlet syndrome. Our approaches; however, varied in terms of the indications for surgery, the type of surgery and the timing of the reconstruction.[1)2) 10)12)]

In the early stages of embryological development, a left SVC is present as a counter part of a normal right-

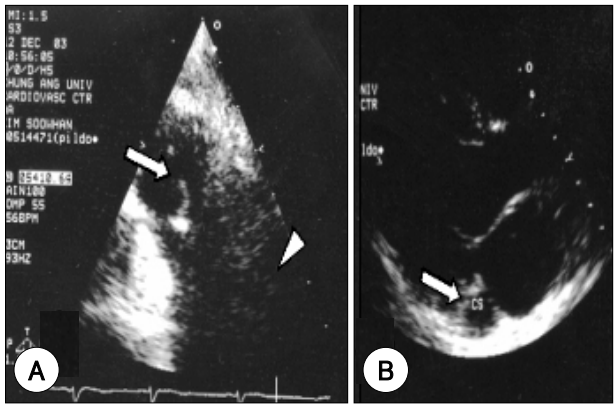

Fig. 3. The 2D-Echocardiogram. A: parasternal short axis view; thickened, dysplastic partially bicuspid pulmonary valve (arrow) and marked dilatation of the main pulmonary artery (arrow head). B: parasternal long axis view; huge coronary sinus (arrow) along the posterior side of the left atrium.

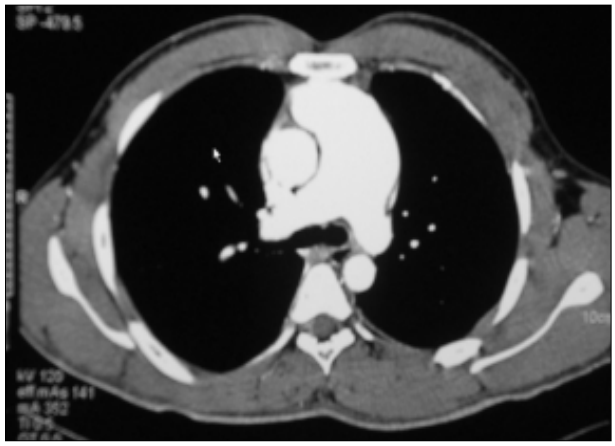

Fig. 4. A 3D-Chest CT. Marked aneurysmal dilatation of the main pulmonary artery.

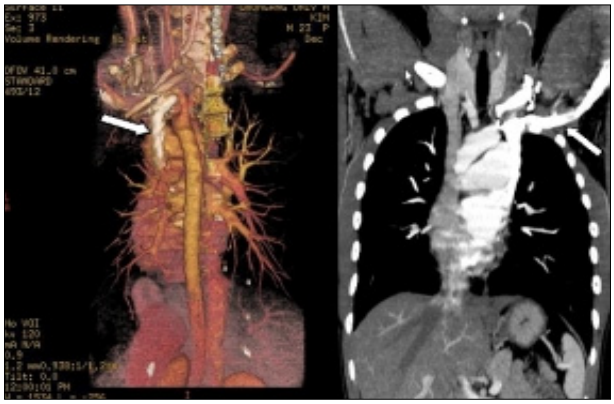

Fig. 5. A 3D-Chest CT. Persistent left superior vena cava (arrow) draining into the coronary sinus.

sided SVC. However, it normally obliterates and forms the 'Ligament of Marshall' in adulthood.[3)5)] Residual persistence of the left SVC in adult life is normal in rabbits and some other mammals, but it is a rare abnormality in humans. Moreover, when associated with a congenital disease, its more relevant clinical implication

is its association with disturbances of cardiac impulse formation and conduction.[3)13)14)] Developmentally, the sinus node, atrioventricular (AV) node and the His bundle may be heavily influenced by the lack of regression of the left SVC. The persistent presence of the left SVC alters the anatomic location and histology of the conducting system, paradoxically speaking. This pathologic substrate may predispose the patient to arrhythmias and sudden death.[3)15)] The prevalence of the left SVC in patient with congenital cardiac abnormalities is much higher than in the general population, ranging from 2.8 to 4.3%. In addition, about 10% of these subjects do not have a right SVC.[3)13)16)] Nsah et al.[5)] reported significantly more frequent associations between a persistent left SVC and AV canal defects, cor triatriatum and mitral atresia. When not associated with other congenital cardiac anomalies, it is usually asymptomatic and hemodynamically insignificant.[4)] However, a persistent left SVC has important clinical implications under certain circumstances, such as the positioning of a left-sided pacemaker or implantable cardioverter-defibrillator (ICD), the placement of central venous lines for therapeutic purposes and hemodynamic monitoring, and cardiopulmonary bypass in patients undergoing cardiac surgery procedures.[3)4)17)]

Mesocardia means that the heart is in the middle of the thorax, with a prevalence of 4 in 1716 autopsied cases (0.2%); therefore, is a very infrequent diagnosis, and 10 different types of congenital mesocardia were found in 17 autopsied cases.[6)18)]

A pulmonary artery aneurysm is an uncommon vascular anomaly. Deterling and Clagett[19)] reviewed 109,571 autopsied cases, and only 8 cases of a pulmonary artery aneurysm were recorded. Its etiology and pathogenesis are not well known, but can be categorized as idiopathic and secondary types. More than 50% of pulmonary artery aneurysms are combined with congenital heart diseases, such as patent ductus arteriosus (PDA), ventricular septal defect (VSD) and atrial septal defect (ASD). Other causes, such as infection, collagen vascular disease, bronchial tumor, iatrogenic (Swan-Ganz catheter), and bicuspid pulmonary valves, may also be related to the pathogenesis of a pulmonary artery aneurysm.[9)20)] A pulmonary artery aneurysm is diagnosed with a measured diameter more than 28 mm, which can be achieved by a combination of echocardiography, conventional CT scan, MRI and angiography. Recently, a 3-dimensional CT scan has also been widely used as an accurate, noninvasive diagnostic tool.[20)] The most common causes of death in the presence of an aneurysm of the pulmonary artery are congestive heart failure or aneurysm rupture.[19)] Correction is the most usual management of a pulmonary artery aneurysm of primary cause. Surgical intervention should be considered when in the presence of an aneurysmal progression in size, pulmonary hypertension and dissection. Recently, the benign clinical courses have been reported, with uneventful long-term follow-up. Thus, periodic monitoring of the pulmonary artery aneurysm diameter and pressure, and right ventricular dysfunction are recommended in the long-term follow-up.[9)20)]

Our patient had four rare ontogenic anomalies; congenital pseudoarthrosis of the clavicle, a persistent left SVC, mesocardia and a pulmonary artery aneurysm combined with bicuspid pulmonary valves. We just observed and followed up, without specific treatment as the patient had no esthetic complaint, clinical symptoms or hemodynamic instability. We report for the first time this new case of a complex congenital anomaly. It is a new syndrome or not, that is question and needed observation.

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