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. Meso-cardia 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.

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 aneurysm 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.
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. 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. 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.

In the early stages of embryological development, a left SVC is present as a counterpart of a normal right-sided SVC. However, it normally obliterates and forms the ‘Ligament of Marshall’ in adulthood. 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. 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. 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. 

Our patient had four rare ontogenic anomalies: congenital pseudoarthrosis of the clavicle, left superior vena cava with a coronary sinus connection, persistent left superior vena cava and atrial septal defect. 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.

REFERENCES

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