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Congenital musculoskeletal abnormalities associated with aortic, pulmonary and iliac aneurysms

Salvatore Lentini[1], Concetta Zito[1], Antonio David[2], Roberto Gaeta[1]

[1]Clinical and Experimental Department of Medicine and Pharmacology, University of Messina, Italy

[2]Department of Cardiovascular and Thoracic Anesthesia and Intensive Care, Policlinico Universitario G. Martino, University of Messina, Italy

Abstract

We present the case of a young patient with severe congenital musculoskeletal abnormalities. associated with different pathologies, with involvement of the arterial tree on both the systemic and pulmonary circulation, and involvement of the lungs due to thoracic hypoplasia. The presence of such extensive pathology raises the question of the suitability of such patients to undergo major corrective cardiac surgery procedures. (Cardiol J 2010; 17, 4: 412–414)

Key words: musculoskeletal abnormalities, upper extremity deformities, congenital, aorta, aneurysm

Introduction

In the literature, the presence of multiple arterial aneurysms has been reported associated with congenital musculoskeletal abnormalities. However, the case we present is particular for the association of different pathologies, for the extensive nature of congenital deformities, the extensive involvement of the arterial tree on both the systemic and pulmonary circulation, and the lungs involvement due to thoracic hypoplasia. The presence of such extensive pathology raises the question of the suitability of such patients to undergo major corrective cardiac surgery procedures.

Case report

A 27 year-old patient presented with multiple congenital abnormalities of the left side of the body, associated with multiple arterial aneurysms. The patient had normal height. He had hypoplasia of the left side of the chest, associated with agenesia of the left upper limb with focomelia, and deformity of the left side of the face. No neurological abnormalities were present. Previously, the patient had reconstructive spine surgery with stabilization plates (Fig. 1) in our institution. This surgery allowed the patient to maintain an erect position. However, this required a long stay in the post-surgical intensive care unit with tracheotomy and long term mechanical ventilation. The patient remained with severe spine scoliosis and hypoplastic chest wall, compromising the respiratory functions (Fig. 2). A right iliac artery aneurysm was also present (Fig. 3). The patient was referred to our clinic for associated aneurysms of the ascending aorta (6.0 cm in diameter) and aneurysm of the main pulmonary artery (5.7 cm in diameter) (Fig. 4). No other cardiac anomalies, congenital or acquired, were present.

At examination, the clinical general conditions were poor, with chronic renal failure. Hypoplastic chest wall was associated with hypoplastic lungs

Address for correspondence: Salvatore Lentini, MD, FESC, Cardiovascular and Thoracic Department, Policlinico G. Martino, Università di Messina, Via Consolare Valeria, Messina, Italy, tel: +39 090 2217086, fax: +39 090 2217086, e-mail: salvolentini@alice.it

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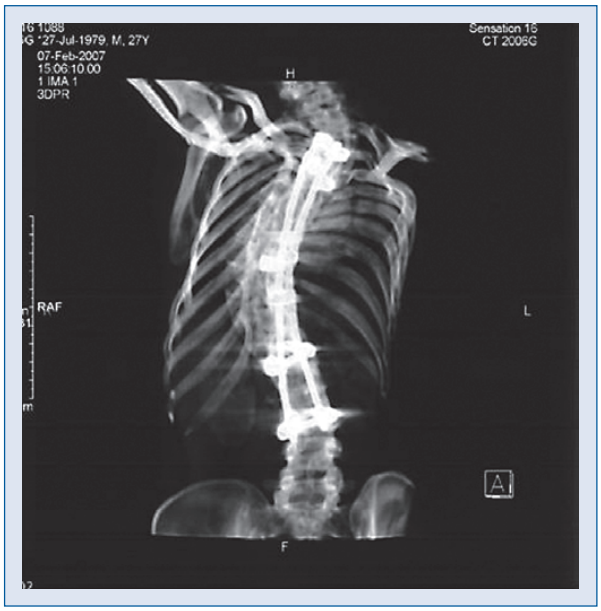

Figure 1. Skeletal computed tomography scan demonstrating severe spine deformities, persisting after spine surgery with stabilization plates, agenesia of left upper limb with focomelia.

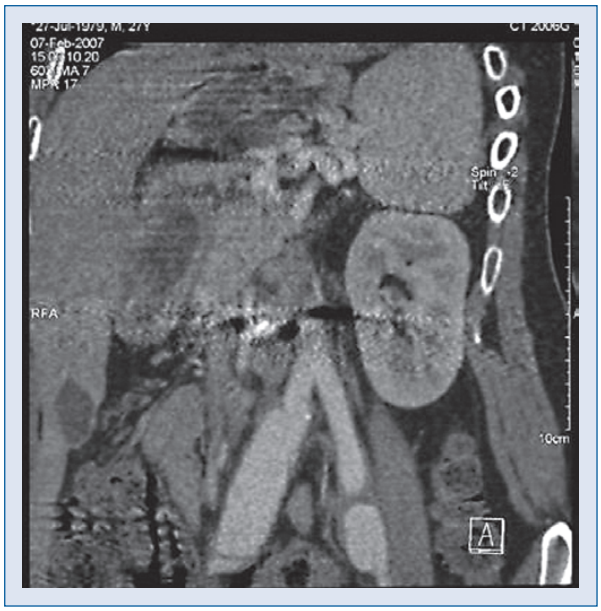

Figure 3. Right iliac artery aneurysm.

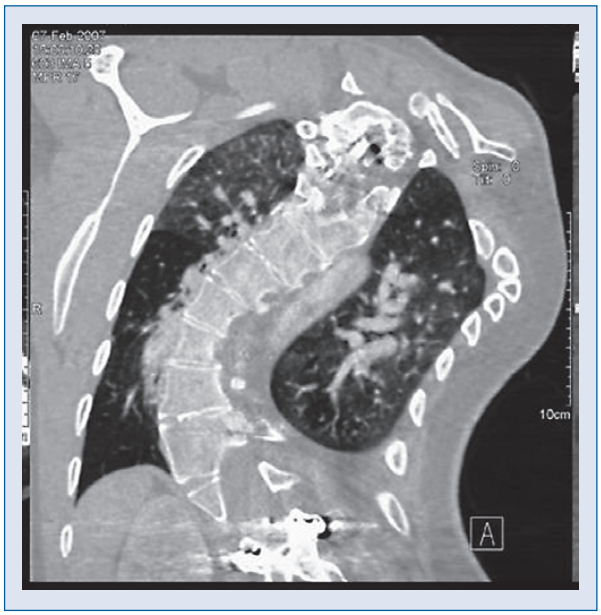

Figure 2. Severe spine scoliosis compromising respiratory function.

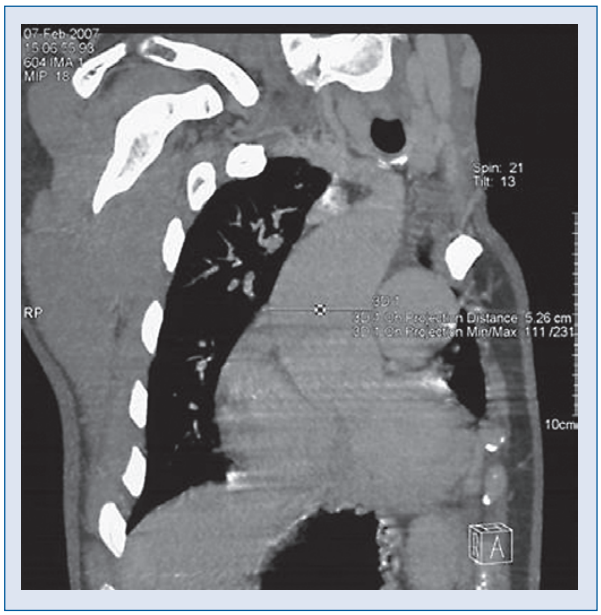

Figure 4. Aneurysm of the ascending aorta (6.0 cm in diameter) and aneurysm of the main pulmonary artery (5.7 cm in diameter).

(Fig. 5), resulting in severe chronic lung insufficiency. Respiratory function tests were severely impaired with restrictive disease features, and with a reduced forced expiratory volume in one second (FEV1) of less than 15% of the expected value. The patient was on home ventilation support and continuous oxygen therapy. The surgical option on the ascending aorta and pulmonary artery was considered

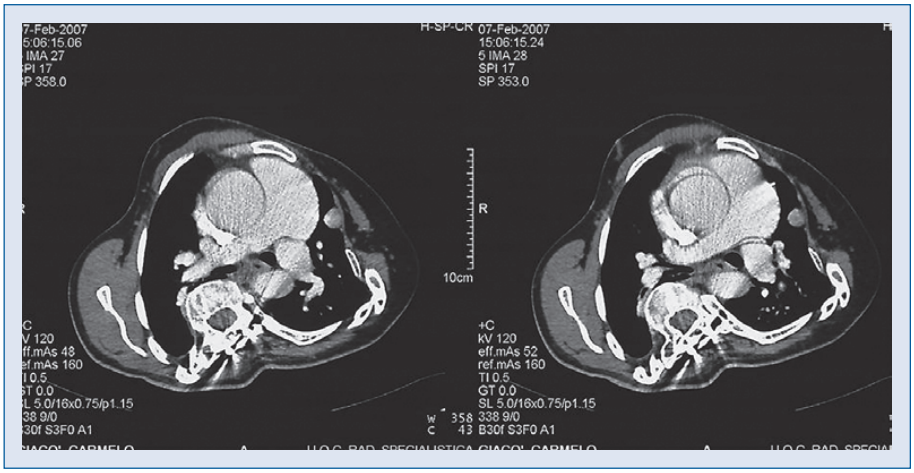

Figure 5. Computed tomography scan showing — dilated ascending aorta and pulmonary artery, and hypoplastic left chest including hypoplasia of the left lung.

to be high risk. The patient refused any surgery, both elective and in emergency when he presented six months later with type A aortic dissection.

Discussion

This case shows an association between arterial aneurysms and congenital malformation of the chest wall and one side of the body. The patient and his family didn't report any history of pre-natal infection. There were not chromosomal abnormalities, and no family history of congenital inherited diseases. Tissue biopsy would have been of value to discover any concomitant connective disorder.

The presence of arterial aneurysms has been associated with several diseases. This patient does not show the phenotypic characteristics of a Marfan, or vascular Ehlers-Danlos syndrome [1], or Loeys-Dietz (Loeys) syndrome [2].

In the presented case, congenital chest abnormalities with severe involvement of respiratory function influenced the surgical decision on major surgery. The patient's psychological involvement was also an important factor, especially because previous surgery was complicated by chronic long stay in the intensive care unit for severe respiratory failure.

Acknowledgements

The authors do not report any conflict of interest regarding this work.

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