## **CR11**

## A patient with thrombophilia and constrictive pericarditis – case report

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INTRODUCTION/OBJECTIVES: The diagnosis of constrictive pericarditis is difficult. Cardiac catheterization with intracavitary pressure curves analysis is considered as a gold standard, but other methods should also be included. This condition can be complicated with comorbidities.

CASE PRESENTATION: A patient was admitted to hospital with symptoms of right heart congestion and atrial fibrillation, known for 4 years. Anticoagulant therapy with dabigatran was started. Suddenly he felt breathless and thoracic MSCT showed bilateral incapsulated pleural effusion, bronchial deformities and a small pericardial effusion that measured 0.7 cm. MSCT pulmonary angiography did not show pulmonary embolism. Fiberoptic bronchoscopy found only nonspecific mucopurulent substrate. Quantiferon test was negative. Dyspnea and pericardial effusion were worsening. Echocardiography showed dilatation of both atria with indirect signs of high right atrial pressure and constrictive hemodynamic and thrombus in right atrium (RA). Another MSCT revealed multiple thrombi in RA, segmental PA, LA auricula. Genetic analysis confirmed thrombophilia. Cardiac MR confirmed constrictive pericarditis. Despite anticoagulant therapy he had another thromboembolic episode; inferior and superior caval vein thrombosis and right iliac artery embolism. Iliac artery thromboendarterectomy was performed. PET CT revealed metabolic active pericardium, so tuberculostatic treatment was started despite of negative Quantiferon test. Pericardiectomy was done partially due to adherent thick pericardium. Pathohistological analysis of pericardium did not confirm specific inflammation - tuberculostatic therapy was stopped. Patient was discharged in improved condition.

CONCLUSION: The diagnosis of constrictive pericarditis can be challenging, sometimes without known etiology. Comorbidities should be treated simultaneously.

## **CR12**

## Congenital hypotonia of an unclear origin

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INTRODUCTION/OBJECTIVES: Congenital hypotonia is a rare condition characterized by a reduced muscle tone and strength. Hypotonias can be central, originating from the central nervous system or peripheral, related to the disorders of the peripheral nerves and/or muscles. The diagnosis process consists of physical examination, lab tests, and gene analysis CASE PRESENTATION: We present an 8-months old male infant referred by a primary pediatrician under the suspicion of hypotonia. Upon first inspection, the given motor response was deficient on supination, lower extremities were crossed, with lower general muscle tone. Occasional tremor of the upper limbs was noted, and when set in a four-legged position, the patient could not maintain the posture which always led to falling. When placed in a sitting position, the patient balanced himself with anteflexion and leaning on his hands. Psychosocial development was concordant with age. Physical therapy was started. Six months later, the follow-up showed great improvement of the general physique. The patient was able to seat himself, manipulate objects with both hands, move to the four-legged position on his own, and stand on hit feet with assistance. The pincer-grasp was not present till that time.

CONCLUSION: The reported case is an extraordinary example of significant progress, even when the diagnosis is not established on time. Early diagnosis and an early start of regular physical exercise are vital parts of managing patients with hypotonia. Although the condition is limiting, many functions could be acquired that could ensure somewhat of a normal life.