Case report – Thoracic general

Fatal right spontaneous haemothorax in Von Recklinghausen’s disease

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Abstract

Spontaneous massive haemothorax is uncommon and usually occurs as a result of pulmonary infarction, arteriovenous fistula, neoplasm, ruptured aortic aneurysm, rupture of pleural adhesions or pleural endometriosis. Massive haemothorax in Von Recklinghausen’s disease occurs rarely but with potentially fatal results in spite of surgery. We present a case of a spontaneous massive exsanguinating haemothorax in a patient with neurofibromatosis type 1 caused by rupture of a branch of the right subclavian artery. Bleeding was probably due to neurofibromatous invasion of the arterial wall.

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1. Introduction

Von Recklinghausen’s disease or neurofibromatosis type 1 (NF 1) is an autosomal dominant disorder characterized by multiple skin tumours and abnormal cutaneous pigmentation [1,2]. Different associated complications have been described in this pathology such as central nervous system and osseous system involvement, benign and malignant schwannomas and other types of malignant tumours [1]. Vascular lesions in this entity are rare, but sometimes fatal [1,3]. Massive spontaneous haemothorax in patients with Von Recklinghausen’s disease has been occasionally reported in the literature [1,2,5,6]. We describe a patient with NF 1 who died because of a massive spontaneous haemothorax following the rupture of a branch of the right subclavian artery.

2. Case report

A 33-year-old white man with Von Recklinghausen’s disease presented to the emergency service of another hospital complaining of a sudden right thoracic pain. Simultaneously he began to experience progressive dyspnea. Past medical history was significant for an intervened cerebral low-grade astrocytoma 14 years previously. Past medical history was significant for an intervened cerebral low-grade astrocytoma 14 years previously. Physical examination revealed a hypophonesis of the right haemithorax and abnormal cutaneous pigmentation [1,2]. Different associated complications have been described in this pathology such as central nervous system and osseous system involvement, benign and malignant schwannomas and other types of malignant tumours [1]. Vascular lesions in this entity are rare, but sometimes fatal [1,3]. Massive spontaneous haemothorax in patients with Von Recklinghausen’s disease has been occasionally reported in the literature [1,2,5,6]. We describe a patient with NF 1 who died because of a massive spontaneous haemothorax following the rupture of a branch of the right subclavian artery.

The post-mortem examination revealed a right haemothorax of about 500 ml with a collapsed right lung. There was a mediastinal haematoma of about 6 cm in diameter in the right upper pole of the caval vein and a rupture of the first branch of the right subclavian artery. Microscopic study demonstrated a dense spindle-cell proliferation involving the right subclavian artery (Fig. 1). The cellular proliferation was of neural type not showing nuclear anaplasia or mitoses. Medial degeneration and etc.) and multiple cutaneous neurofibromas associated with café-au-lait spots and axillary freckling. There was no history of previous trauma. A frontal chest X-ray showed a complete right pleural effusion and midline shift of the mediastinal structures to the left. An intercostal drainage tube was inserted and approximately 1500 ml of blood was removed. He suffered progressive haemodynamic instability so he was then urgently transferred to our centre for surgical evaluation.

He was admitted with clinical signs of hypovolaemic shock and an active blood discharge from the thoracic drainage tube. An emergency thoracotomy was indicated to determine the source of the bleeding. Surgical exploration demonstrated about 6000 cc of blood in the right thoracic cavity. A mediastinal haematoma near the superior caval vein was appreciated with minimal pleural rupture. When opening the mediastinal pleura, an active haemorrhage from the retrocaval region near the origin of the subclavian vein was noted. The patient suffered a hypovolaemic cardiac failure and he died despite of intensive cardiopulmonary resuscitation.

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disruption of the elastic layers accompanied the neurofibromatous invasion within the arterial wall (Fig. 2). These changes were also observed in several adjacent small veins. Neurofibromas could not be seen at the bleeding point. The soft tissue of the neck external to the haematoma demonstrated small vessels surrounded by neurofibromatous and ganglioneuromatous tissue.

There was generalized NF 1-associated arterial dysplasia. The abnormal small arteries showed an intimal expansion and stenosis due to an accumulation of mucoid material in the intima and myointimal cell proliferation. There was also a replacement of elastic tissue and the muscular layer by intimal cells.

A right adrenal pheochromocytoma of 4.5 cm in diameter and an appendicular carcinoid tumour (0.4 cm) were found.

3. Discussion

The incidence of vascular lesion in NF 1 has been reported to be only 3.6% [1]. Arterial lesions associated with Von Recklinghausen’s disease are classified into two categories in relation to the diameter of the vessels. Larger vessels such as the aorta, carotid, etc. are surrounded by neurofibromatous or ganglioneuromatous tissue whose proliferation weakens the media and injures the elastic tissue, leading to aneurysms formation or stenosis. On the other hand, a NF 1-associated arterial dysplasia involving smaller vessels and not related to neural malformation has been described [1].

Miura et al. [1] reported 12 cases of spontaneous haemothorax in patients with NF 1 in Japan. They occurred more frequently on the left side, with the intercostal artery and the subclavian artery being the most ruptured vessels. Only a case of affectation of the right subclavian artery was described. Pathological changes were observed in seven cases and a relationship between the vascular injury and the neurofibroma was proven in five of them. Neurofibromatous invasion of the vascular wall was noted in four cases and neurofibromatous proliferation surrounded an artery causing ischaemic changes in one case. Ten patients underwent surgery and half of them expired due to blood loss in spite of treatment.

In our case, bleeding was due to a rupture of the first branch of the right subclavian artery producing an important mediastinal haematoma and massive haemothorax. Microscopically the ruptured point could not be clearly appreciated because of the extensive haematoma. Histologic study of the neighbouring subclavian artery revealed involvement of the vascular wall by neurofibromas, so rupture is likely related to neurofibromatous invasion.

The prognosis is poor in cases when the loss of blood is massive and the patient falls into hypovolaemic shock. An immediate surgical intervention is recommended in those patients with Von Recklinghausen’s disease who develop spontaneous haemothorax. This procedure may be life-saving even when the suitable access route is not easy to establish (lesion of intercostal artery or subclavian artery?). If the clinical situation permits it, the best option is to make a radiological exploration in order to find the source of the bleeding. The existence of a mediastinal haematoma indicates an injury of the subclavian vessels, and sternotomy should be recommended. In cases when haemothorax is not accompanied by mediastinal haematoma an intercostal vessel injury is expected, so patients should undergo thoracotomy.
References


