Commentary

Neurofibromatosis type 1 (NF1), or Von Recklinghausen disease, is a group of heterogeneous diseases that present with multiorgan involvement. This disease is an autosomal dominant genetic disorder that includes an abnormality of the long arm of chromosome 17. The prevalence of this disease is about 1 in 3,000 births.[1] Diagnosis of this disease is according to the following criteria: Café-au-lait spots, plexiform neurofibroma, other forms of neurofibroma, iris hamatomas, fleckling, optic glioma, abnormality of bone lesions, and presents in the first degree relation.[2]

The tumor-related NF1 includes neurofibroma, glioma, malignant peripheral nerve sheath tumor, non-lymphomatic leukemia, and pheochromocytoma.[3] However, the most common tumor-related disease is neurofibroma. Vascular involvement, including occlusion, stenosis, aneurysm, or ectasia, is rare. The cause of vasculopathy associated with NF1 is described as: (1) Direct vascular invasion from the adjacent neoplasms, and (2) vascular dysplasia with the thickening and concomitantly reduced strength of the arterial wall causing aneurismal formation.[4,5]

Massive hemothorax is a fatal complication of NF1. The cause is usually a spontaneous rupture of arterial involvement due to wall fragility. The most common arterial involvements are subclavian and intercostal arteries. The other arterial involvements include the thyrocervical trunk, internal thoracic artery, phrenic artery, and vertebral artery. Abdominal aorta involvement was also reported.[6] The source of arterial bleeding could be determined from several previous articles due to the spontaneous rupture of the pathologic artery.[2,4-6] However, some reports could not identify the source of bleeding. In this case report, the patient had a spontaneous massive right hemothorax, but the source of bleeding could not be found by either CT scan or thoracoscopy. In my opinion, the cause of bleeding in this case was possibly due to a ruptured neurofibroma or a direct invasion of the artery at the apical lung, evidenced by a sentinel clot adjacent to the tumor.[7] Treatment of spontaneous massive hemothorax in NF1 depends on the etiology of the bleeding. Endovascular treatment with embolization is one choice in the case of an arterial rupture related to NF1.[2] In contrast, cases of a ruptured tumor should be treated by surgery.

In summary, spontaneous massive hemothorax in NF1 is not common, but becomes a life-threatening condition because of massive blood loss and respiratory discomfort. Early management should be performed to decrease morbidity and mortality.

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