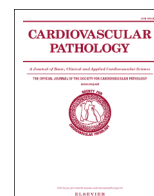




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Case Report

A rare case of internal jugular vein aneurysm with massive hemorrhage in neurofibromatosis type 1

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ABSTRACT

Neurofibromatosis Type 1 (NF1) is a relatively common autosomal dominant disorder. Vascular involvement is a well-recognized manifestation of NF1, but venous aneurysm associated with NF1 is extremely rare. We present a case of an NF1 patient with a left internal jugular vein aneurysm with massive hemorrhage occurring during surgery. Due to the extreme fragility of both the aneurismal wall and the surrounding tissue, the patient developed severe intraoperative bleeding. Pathological examination confirmed aneurismal wall infiltration of the neurofibromatosis. Physicians should be aware that hemorrhagic complication in NF1 can occur and be fatal.

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1. Introduction

Neurofibromatosis Type 1 (NF1), otherwise referred to as von Recklinghausen disease, is an autosomal dominant disorder affecting one in 3000 individuals. NF1 can involve any organ, but mainly connective and nerve tissues are affected [1]. In NF1, vascular complications represent the second most common cause of death, after malignant peripheral nerve sheath tumor [2]. However, vascular involvement is relatively uncommon in NF1, with an estimated prevalence ranging from 0.4% to 6.4% [3].

A literature review of the vascular involvement in NF1 by Oderich et al. [4] found predominantly arterial involvement, with 41% occurring in the renal artery. Other involvement sites include the neck and head (19%), extremities (12.9%), and the abdominal aorta (12%). Involvement of the venous system is rare. Only three cases have been identified in the literature with aneurismal lesions in the venous system, and all of these lesions were localized in the internal jugular vein [4–6].

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2. Case report

A 60-year-old man with neurofibromatosis presented with a 3-day history of tenderness and an enlarged left cervical mass. Physical examination revealed multiple neurofibromas over his face, trunk, and extremities, associated with café-au-lait spots. There was a soft elastic mass without pulsation, 8 cm in diameter, extending from the left mandibular angle to above the left clavicle (Fig. 1).

A contrast-enhanced computed tomography scan demonstrated a cystic mass, 6 cm in diameter, in the left submandibular space. Magnetic resonance imaging (MRI) revealed an internal jugular vein aneurysm with a thrombus. In addition, contrast-enhanced MRI revealed irregular enhancement in both the aneurismal wall and the surrounding fat tissue (Fig. 2).

At preoperative blood tests, blood counts and activated partial thromboplastin time were normal. The prothrombin time was 13.6 s (reference range 9.4 to 12.5 s). The other clotting tests, including antithrombin III, fibrin degradation products, and D-dimer were not examined.

After obtaining the informed consent, the patient underwent surgery. The internal jugular vein aneurysm was partially filled with an organizing thrombus and was surrounded by well-vascularized and extremely fragile tissue. Due to the fragile nature of both the vessel wall and the surrounding tissue, venous and arterial bleeds were difficult to control. Since the intraoperative blood loss was 12,195 ml, the patient received 27 units of packed erythrocytes and

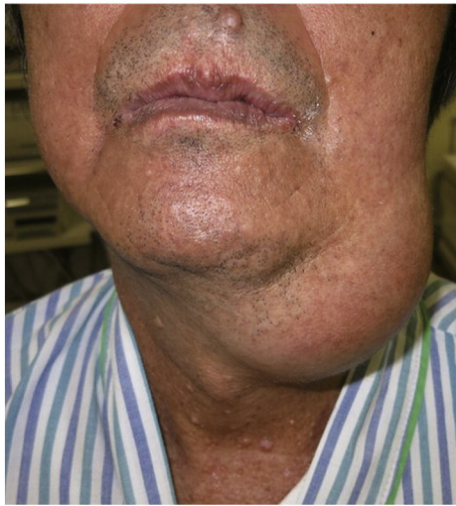


Fig. 1. Photograph of the neck showing a left cervical mass and multiple neurofibromas.

10 units of frozen plasma during the operation. No arteriovenous communication was detected. The aneurysm and a part of the left internal jugular vein were analyzed for pathology.

The aneurysm was 5.5×5×2 cm (Fig. 3A–B) and was adherent to the left parotid gland and to the surrounding fibrous and fat tissue. The aneurismal wall showed irregular thinning or thickening with fibrosis, and the aneurysm was partially filled with an organizing thrombus (Fig. 3C).

The paraffin-embedded tissue was sectioned and stained with hematoxylin-eosin and Elastic–Masson's stains. An immunohistochemical study was performed on the paraffin-embedded tissue using a standard avidin-biotin immunoperoxidase technique and S-100 protein (DAKO) antibody.

Histological examination revealed that the aneurismal wall had a reduction of elastic fibers in the tunica media, with a few residual smooth muscle fibers (Figs. 4A–B, 5A–B). An organizing thrombus with recanalization was observed in the aneurysm. The surrounding tissue of the aneurysm showed diffuse proliferation of spindle-shaped cells with wavy nuclei and a myxoid change of the stroma, which focally infiltrated the aneurismal wall. Immunohistochemi-

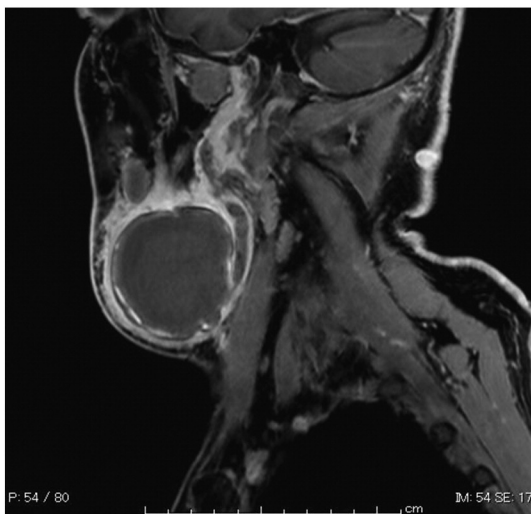


Fig. 2. Contrast-enhanced MRI, which revealed an internal jugular vein aneurysm with thrombosis and irregular enhancement in both the aneurysmal wall and the surrounding fat tissue.

cally, S-100 expression was observed in the cytoplasm of the proliferating cells (Figs. 4C, 5C). These findings indicated that there was infiltration of the neurofibroma in the aneurismal wall. The wall of the small veins and arteries in the surrounding tissue and the wall of the left internal jugular vein were also infiltrated by the neurofibroma.

3. Discussion

In our case, the patient developed an internal jugular vein aneurysm causing a tender neck mass. In general, venous aneurysms are rare and can be the result of several processes, including tumor growth, inflammation, and trauma, or they can appear spontaneously [7,8]. On review of the literature, we found several cases of jugular vein aneurysm [7,8], but only three cases were associated with NF1 [4–6]. Extreme fragility of both the vessel wall and the surrounding tissue, with severe intraoperative bleeding, presented in two of these patients [5,6]. We had similar problems with our patient as well. On pathological examination, we found that the aneurismal wall was focally infiltrated by the neurofibroma and also that the surrounding tissue was widely infiltrated by the neurofibroma. In the aneurismal wall infiltrated by the neurofibroma, there was a reduction of both elastic fibers and smooth muscle fibers in the tunica media, which we suggest are associated with the fragility of the aneurismal wall.

Arterial dysplasia is another type of vascular lesion associated with NF1 that Greene et al. suggested represented mesodermal dysplasia [9]. This lesion is characterized by an accumulation of mucoid substance and proliferation of myointimal cells in the intima of the arteries [9,10]. However, in our case, these types of dysplastic features were not detected in the aneurysm and in the surrounding small vessels.

Therefore, we suggest that the vascular infiltration by the neurofibroma was primarily responsible for the difficulty in maintaining hemostasis and thus led to severe intraoperative bleeding.

Despite the vascular infiltration of the neurofibroma, there is no histological evidence of malignancy, such as cellularity, cellular pleomorphism, or mitoses.

In conclusion, patients with NF1 can present with various levels of vascular involvement, including a jugular vein aneurysm. The infiltration of the vessel wall by a neurofibroma can cause extreme fragility of both the aneurismal wall and the surrounding tissue and result in massive bleeding during the surgery. Since the hemorrhagic complication in NF1, especially with a venous aneurysm, can be fatal, both clinicians and pathologists should be aware of this possible complication.

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