CASE REPORT

Neurofibromatosis 1 with Internal Jugular Vein Aneurysm and Thrombus

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ABSTRACT

Neurofibromatosis 1 (NF1) is a common genetic disorder. Vascular abnormalities have been defined in these patients, which are usually arterial and very rarely venous. Venous aneurysms are difficult to repair as the vessel walls are friable due to the invasion of the vessel wall with neurofibromas. We present the case of a woman with NF1 who had aneurismal dilation and thrombus of the internal jugular vein. The patient was managed conservatively and died a sudden death. Venous vascular abnormalities in NF1 are rare and when present usually involve the internal jugular vein. A thrombus in the internal jugular vein in these cases is dangerous. Surgical treatment can be complicated by life-threatening haemorrhage, while conservative management can be complicated by embolization into the pulmonary circulation.

Keywords: Neurofibromatosis 1, vascular abnormality, venous thrombosis

INTRODUCTION

Neurofibromatosis 1 (NF1) is an autosomal-dominant genetic disorder characterized by café-au-lait spots, axillary and inguinal freckling, multiple cutaneous neurofibromas, and iris Lisch nodules. Rarer manifestations include plexiform neurofibromas, gliomas, malignant peripheral nerve sheath tumours, scoliosis, tibial dysplasia and vasculopathy. Vascular abnormalities are rare and when present are usually arterial. Venous vascular abnormalities are exceedingly rare.

CASE REPORT

The patient was a 65-year-old female who presented to us with a 1-month history of a painless neck swelling. The neck swelling had been increasing since the onset. The swelling was localized on the right side of the neck, extending from the clavicle to the cricoid cartilage and measuring around 5 cm. (Fig. 1). The swelling was non-tender, non-pulsatile and elastic in character. There was no palpable thrill or bruit on auscultation. The patient was a known case of NF1 with multiple family members affected with the same. The patient had multiple neurofibromas and café au lait spots on her body (Fig. 2). The patient had no past episodes of similar illness or any vascular anomalies.

Fig. 1: Patient with the neck swelling as shown by the arrow. The neck swelling is due to the internal jugular vein aneurysm and thrombus.

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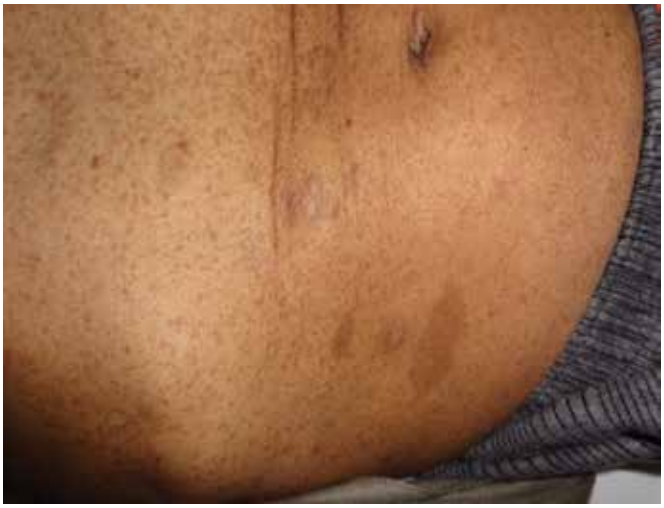

Fig. 2: A picture of the abdomen of the patient showing neurofibromas and multiple café au lait spots.

A Doppler ultrasonography of the neck vessels showed internal jugular vein thrombus and aneurysm, which was confirmed on contrast-enhanced computed tomography of the neck. (Fig. 3). The rest of the investigations were normal. The patient was planned for a surgery but declined to undergo the same after discussions. The patient was advised a regular follow-up and was started on anticoagulation. One month later, the patient was brought to our emergency with a sudden cardiac arrest and was declared dead. The cause of the death was suspected to be pulmonary embolism.

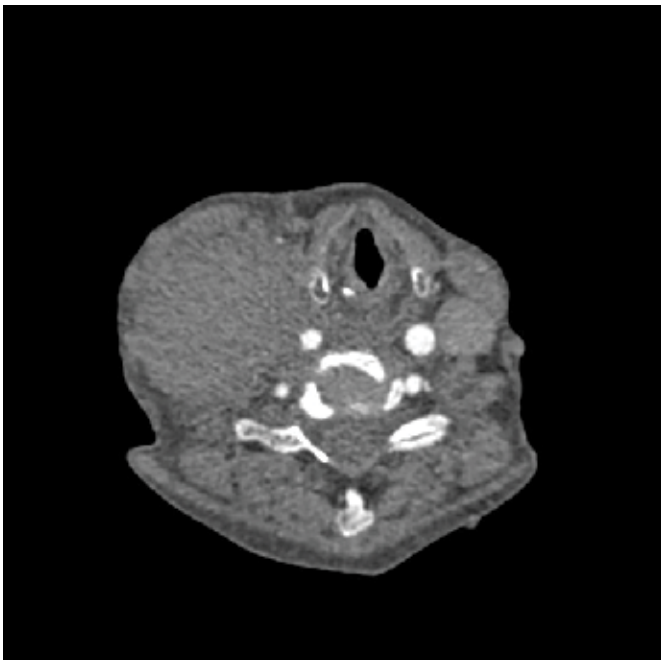

Fig. 3: Computed tomography scan showing the internal jugular vein aneurysm and thrombus on the right side.

DISCUSSION

Neurofibromatosis is a common genetic disorder. It is of two types: NF1 and NF2. Neurofibromatosis 1 has a characteristic cutaneous expression that includes benign neurofibromas, which are mixed tumours of all cell types found in the peripheral nerves; café-au-lait spots, which are hyperpigmented macules, axillary/inguinal freckling; and Lisch nodules, which are pigmented hamartomas of the iris. Neurofibromatosis 2, on the other hand, is mainly restricted to tumours of the central and peripheral nervous systems, which are rarely accompanied by skin disorders (1, 2). Neurofibromatosis 1 is an autosomal-dominant disorder. The penetrance approaches 100% by age 20, although expression is variable (3). The NF1 gene is located on chromosome 17q11.2 and encodes for the protein neurofibromin (4).

NF can involve multiple organs with its varied manifestations. Vascular abnormalities have been observed in NF1. Most of them are arterial and only rarely are venous. They most notably have been aneurysms or stenoses of the aortic, renal and mesenteric circulation (5). We found only four cases of jugular vein aneurysm associated with NF (5–8). Extreme fragility of both the vessel wall and the surrounding tissue with severe intraoperative bleeding were frequently seen during surgical repair. The aneurismal wall was found to be focally infiltrated by the neurofibroma, and also the surrounding tissue was widely infiltrated by the neurofibromas in these cases. In our case, we could not get the pathological specimen of the patient, but it is highly likely that similar findings might have been found. Therefore, before undertaking surgery on a vascular aneurysm in an NF1 patient, it should be kept in mind that the aneurysm may bleed massively during the surgery, and hence necessary precautions should be taken; also, the patient should be informed about this possible complication. Our patient had a sudden death, which could have been caused by an embolic complication or a haemorrhagic complication like sac rupture. This is a significant point to be kept in mind while starting the patient on anticoagulation. We suggest close monitoring of coagulation in patients being managed conservatively.

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