Opinion Article

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Neurofibromatosis Type 1: Endovascular Inner Catching of Burst Occipital Conduit Pseudo Aneurysm

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Introduction

An autosomal dominant mutation on chromosome 17q11.2 known as Neurofibromatosis Type 1 (NF-1a) alters the production of the protein neurofibromin and downregulates the ras tumor suppressor oncogene. Neurofibromas, Lisch nodules, axillary freckling, and café au lait spots are just a few of the distinct phenotypes that can result from these genetic changes. Anomalies in the vascular system that cause spontaneous bleeding are uncommon in this condition. We describe a patient with NF-1 who developed a spontaneous thigh hematoma. Since there were no obvious arterio-venous malformations found during the operation, it was determined that the patient probably had arterial dysplasia and a neurofibroma that had invaded nearby. The dermatologic and neurologic features of Neurofibromatosis 1 (NF-1) that are crucial to the diagnosis of this rare autosomal dominant genetic condition are frequently the focus of discussions. For instance, for a diagnosis to be made there must be evidence of at least two of the following: neurofibromas, Lisch knobs, axillary freckling, and bistro au lait spots. Anomalies of the cardiovascular system are another rare association that has been described in NF-1 patients. These lesions, which include aneurysms, stenosis, and arteriovenous malformations, are referred to as NF-1 vasculopathy. The lack of cases portraying vascular pathology and confusions in NF-1 patients is fundamentally because of these discoveries staying asymptomatic for most of patients. The authors describe a case of a spontaneous thigh hematoma in a patient with NF-1 that eventually required surgical intervention in order to provide additional detail and assist others in comprehending this uncommon aspect of an uncommon illness.

Harmless Neurofibromas

RH is a 51-year-old man who presented to the emergency department with the primary complaint of right thigh swelling. In addition to having neurofibromatosis, hypertension, hypercholesterolemia, thyroid disease, scoliosis, and gallstones, RH also has hypertension and hypercholesterolemia. The patient was in good health when they arrived and noticed what appeared to be an insect bite on the right lateral aspect of the thigh. The patient noticed a small amount of pus when she popped the lesion. Following a couple of moments the modest quantity of expanding that was there at first had expanded to the size of a baseball. The swelling doubled throughout the day, prompting the patient to seek medical attention. The patient denied taking any blood thinners and only took 81 milligrams of aspirin per day, according to additional information gathered in the emergency department. Additionally, the patient denied receiving any blunt force on the affected area. The patient had never experienced anything like this before. Upon admission, vital signs and laboratory results were stable and within normal limits. During the physical exam, a large, round mass-like lesion on the anterior proximal thigh that extended laterally to the right gluteus was found. The mass was about 20 centimeters in size and barely tender. Skin necrosis, lesions, erythema, or warmth were absent. In the emergency department, a CTAb, followed by a CTA with right lower extremity runoff, revealed an 18 cm x 6.4 cm hematoma with active contrast extravasation. Interventional radiologists were consulted regarding a potential coil embolization based on the findings of this study; however, there was no obvious vessel feeding the hematoma, so the patient was given a compressive bandage, admitted to the hospital, and had a series of overnight lab tests.

Haemodynamic Instability

Von Recklinghausen was the first person to describe neurofibromatosis in 1821. The two most common types of neurofibromatosis are Types I and II. Type I (chromosome 17q11.2) and type II (chromosome 22q12) can be distinguished based on the location of the genes. Patients with type I neurofibromatosis might have different cutaneous discoveries, for example, bistro au-lait spots, harmless neurofibromas, and iris hamartomas.1 Blood vessel contribution has additionally been noted, ordinarily as stenoses and most often of the renal supply route. Blood vessel aneurysms have been less regularly portrayed. A ruptured thyrocervical aneurysm was the cause of our patient's life-threatening neck hemorrhage. Ishizu et al. reported the only other similar case in which a NF-I patient presented with neck swelling as a result of a ruptured thyrocervical branch. A ruptured intercostal artery aneurysm or aneurysmal branch of the subclavian artery has been linked to haemothorax in neurofibromatosis patients. The fact that our patient had skin flaps on the same side may have contributed to the unusual presentation of neck haematoma rather than

haemothorax. Nonetheless, the past reconstructive medical procedure made translation of the angiography testing. As a result, it was challenging to determine whether the ruptured vascular abnormality was a genuine NF-1 aneurysm or a pseudoaneurysm caused by the previous reconstructive surgery. We were faced with a number of dilemmas by this patient. The need for arterial control in a difficult neck region with abnormal anatomy and complex previous surgery was eliminated by our initial endovascular approach. Although endovascular embolization was carried out, the microcatheter was unable to fully extend beyond the aneurysm into the distal artery. Time was of the essence because the patient was unstable. Although every effort was made to embolise as far distally as possible, fortunately there was no retrograde flow to fill the aneurysm. The "chicken and egg" challenge of determining whether the inotrope requirements were caused by ongoing bleeding or whether open drainage of the tense haematoma would further stress her overtly impaired cardiac function was posed by the change in cardiac enzymes and ongoing haemodynamic instability. The CT check appearance has likewise shown pressure of the left normal carotid supply route, which could likewise be an expected reason for inotrope reliance. The neck flaps made the surgical access for decompression difficult, so the vascular and maxillofacial teams eventually worked together to perform the procedure. However, controlling bleeding from the abnormal veins was not easy. As a result, our case demonstrates that, despite the increasing use of endovascular embolization in the treatment of acute bleeding, open surgical and endovascular approaches may occasionally be required.