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CASE REPORT

Neurofibromatosis Type-1 and Diffuse Intracranial Dolichoectasia

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ABSTRACT

Dolichoectasia implies abnormal elongation, dilation and tortuosity of arteries, and in the cranial vasculature the vertebrobasilar circulation is predominately affected. Neurofibromatosis type1 (NF-1) predisposes to dolichoectasia. We describe a patient with NF-1 and a unique pattern of symptomatic dolichoectasia involving the intracranial internal carotid, middle cerebral, vertebral and basilar arteries.

Introduction

Neurofibromatosis type-1 is associated with increased risk of Dolichoectasia, which classically involved posterior circulation. No prior reports of NF-1 associated dolichoectasia involving anterior and posterior circulation. Advanced dolichoectasia can cause severe complications and treatment options may result in catastrophic side effects.

Case Presentation

A 47-year-old Caucasian male known to have Neurofibromatosis type 1 (NF-1) presented with a 3 month history of progressive dysphagia and the more recent onset of shortness of breath. His neurologic exam was normal. Computed Tomographic Angiography (CTA) of the neck demonstrated a partially thrombosed right internal carotid artery (ICA) pseudoaneurysm measuring 5cm x 6.5cm x 6.8cm, with associated compression of the esophagus and trachea (figure 1). Cranial CTA also demonstrated dilation of the Internal Carotid Arteries (ICAs), Middle Cerebral Arteries (MCAs) and Vertebral and Basilar Arteries (VBAs). Using the CTA of the head and neck and performing measurements electronically via the Picture Archiving And Communication System (PACS) instrument, we calculated that both Vertebral Arteries (VAs) and the Basilar Artery (BA) fulfilled the Smokers diagnostic criteria for VBA dolichoectasia, with the BA diameter 6.8mm and VA diameters 6mm (Figure 4a,4b). There are no well-established criteria for anterior circulation cranial dolichoectasia, but it has been suggested that an MCA diameter more than 4mm and an ICA diameter more than 7mm are sufficient to establish the diagnosis. Our patient's CTA demonstrated a left ICA diameter of 7.7mm, right ICA 7.6mm, right MCA 6.4mm, and left MCA 7.3mm (Figure 4c,4d,4e). A tracheostomy was followed by a catheter arteriogram which confirmed the CTA findings and demonstrated extensive dilation and tortuosity of all four major intracranial arteries (figures 2,3). Stenting was

deferred due to severe right ICA tortuosity. The aneurysm was coiled, and the proximal right ICA was occluded with a vascular plug; there was good left-to-right filling of the right anterior cerebral artery and MCA through the Anterior Communicating Artery (ACoA). The patient was neurologically normal immediately following the procedure. Four hours later the patient was noted to be less responsive and to exhibit dense left hemiplegia and gaze deviation to the right. CTA of the head and neck demonstrated abrupt cut-off of the right MCA (M2 segment). As before, there was evidence of diffuse intracranial dolichoectasia involving both the anterior and posterior circulations (Figure 4). A catheter arteriogram was performed. Intra-arterial tissue plasminogen activator was administered via a left ICA approach, with access to the occluded right MCA through the ACoA. The right MCA occlusion persisted, and 24 hours following attempted thrombolysis the patient's neurologic deficits were unchanged.

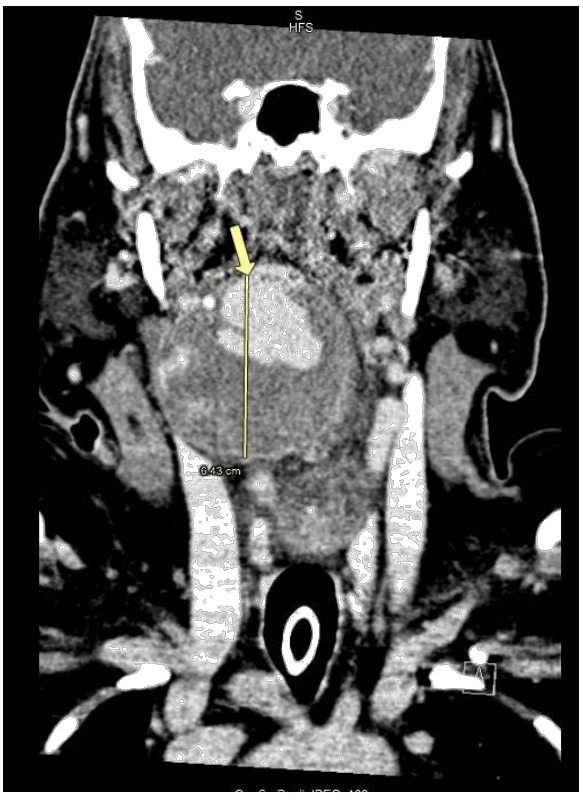


Figure 1: CTA of the neck demonstrates a large, partially thrombosed aneurysm with compression of adjacent midline structures.

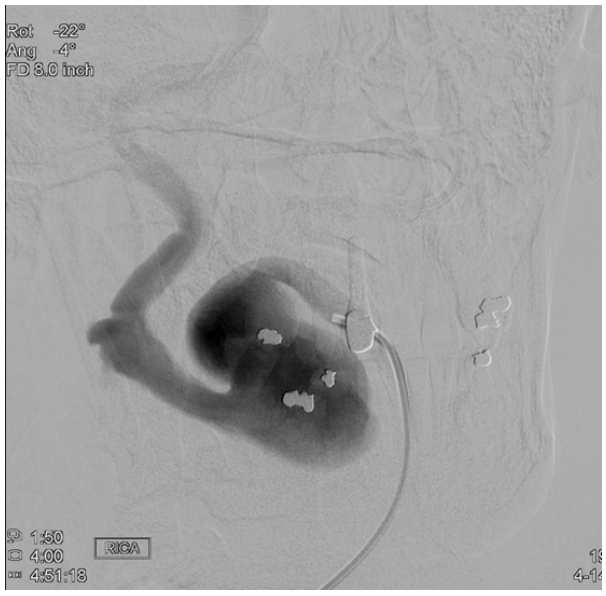


Figure 2: Catheter arteriogram demonstrates dilation and tortuosity of the right ICA.

Discussion

Dolichoectasia is a vasculopathy that involves the arterial wall and produces abnormal dilation, redundancy and tortuosity of the affected vessel. Loss of the internal elastic lamina results in reduction of normal vascular recoil [1]. Dolichoectasia's prevalence is 0.05-0.06% in the general population, and in the cranial arteries it occurs more commonly in the vertebrobasilar system than the carotid system [1-3]. The co-existence of intracranial anterior and posterior circulation dolichoectasia is apparently quite rare. Dolichoectasia is associated with a wide spectrum of clinical presentations, ranging from asymptomatic/incidental, the most common, to disabling and even fatal focal neurological deficits resulting from ischemia, haemorrhage or mass effect [4,5]. Neurofibromatosis is the most common neurocutaneous disorder affecting the central nervous system [6]. In NF-1, gene mutation leads to an increased incidence of benign and malignant tumors within and outside the nervous system. There is multisystem involvement, including but not limited to the cardiovascular, skeletal, renal and integumentary systems [7]. Cerebral vasculopathy resulting from NF-1 is manifested as dolichoectasia or vascular

stenosis/occlusion which may produce a moya-moya pattern of collateralization [8].

As with this vasculopathy generally, dolichoectasia occurring with NF-1 preferentially affects the vertebrobasilar circulation.

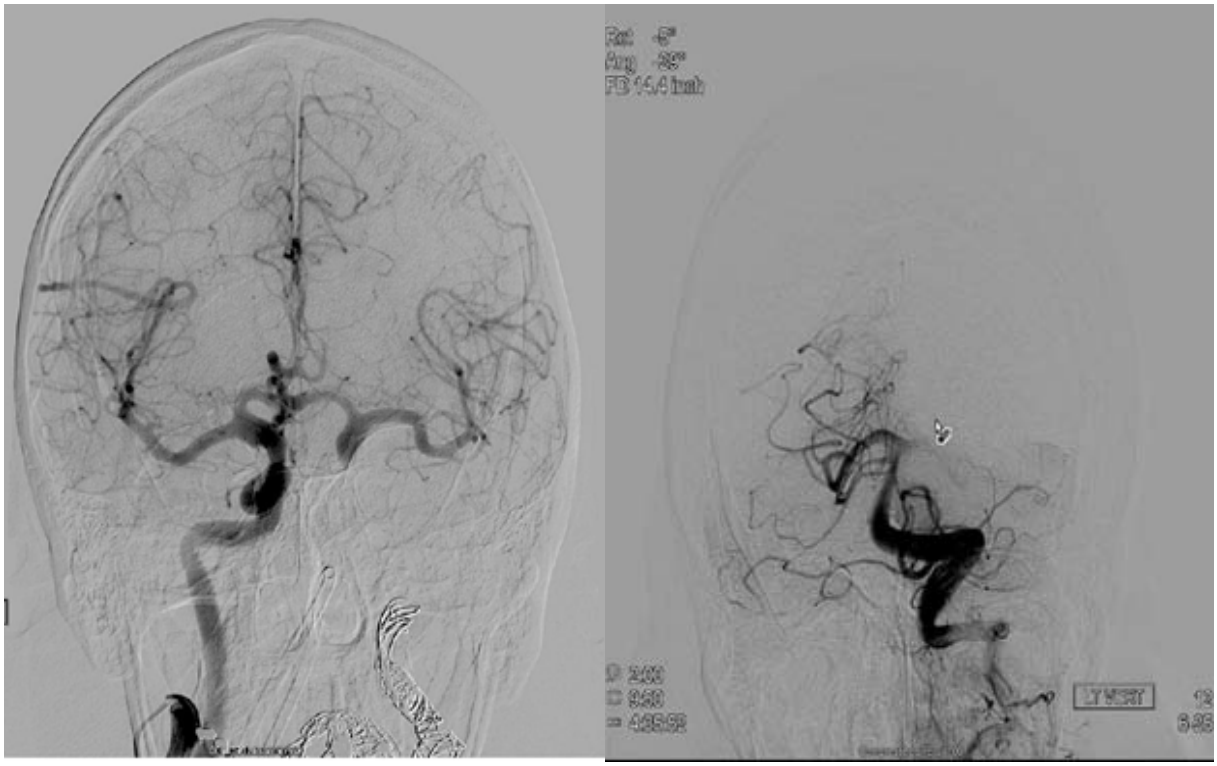
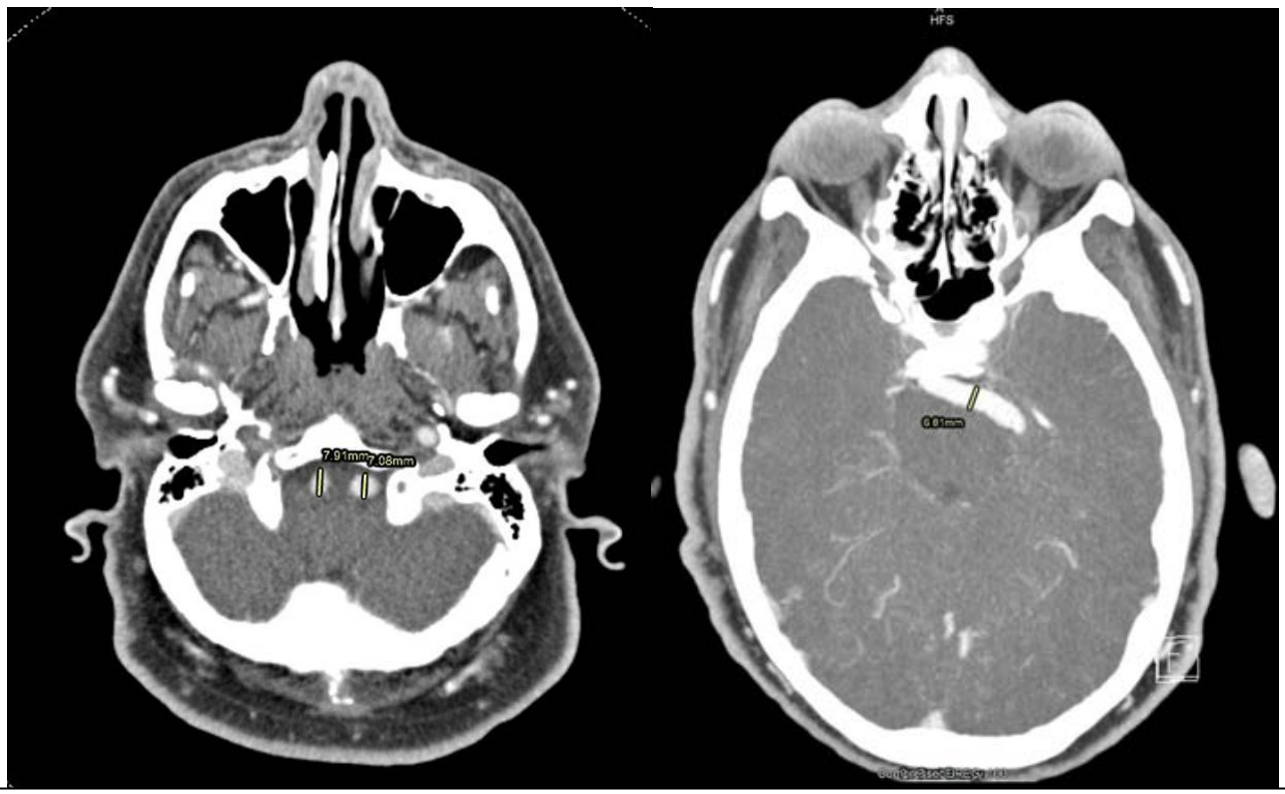
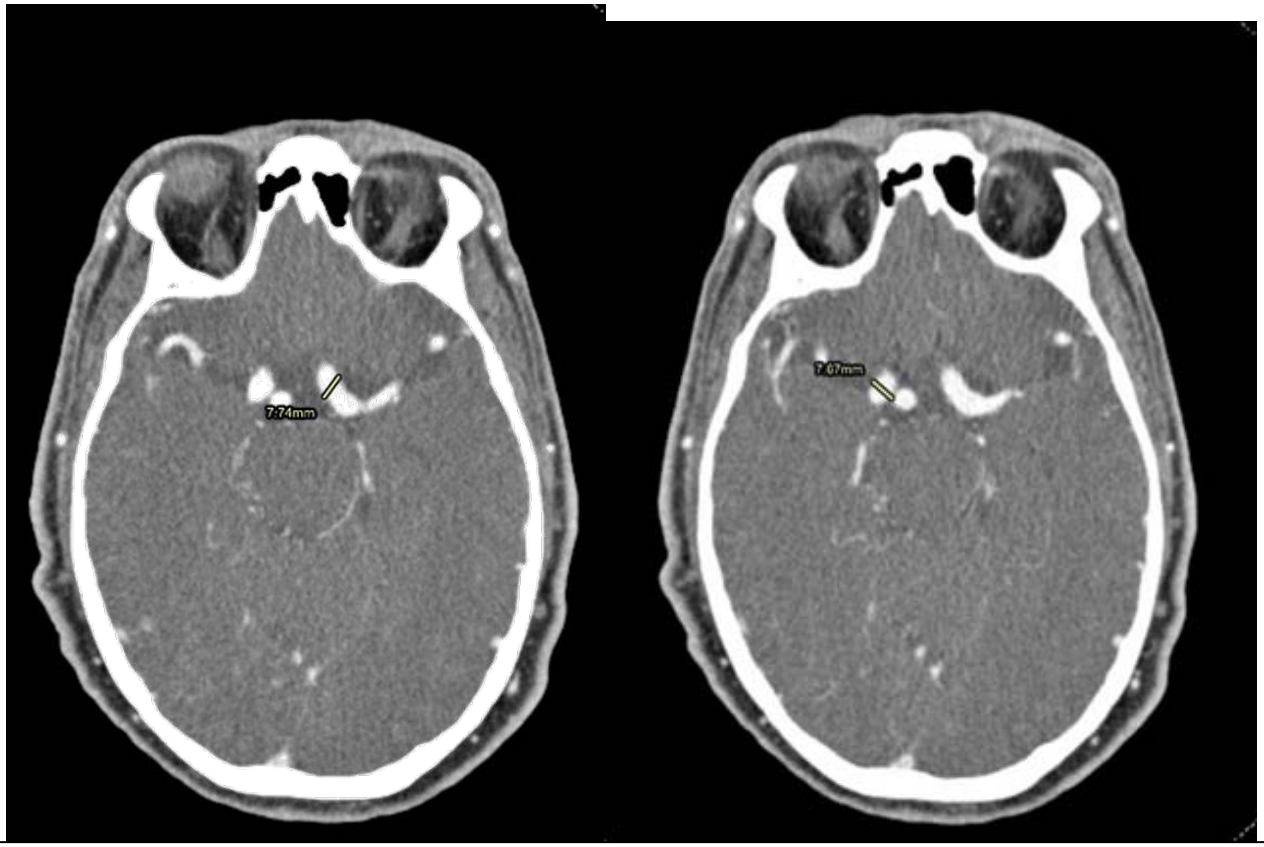


Figure 3: Cerebral arteriogram demonstrates dilation of the bilateral MCAs (a) and left vertebral artery (b)



(a) (b)



(c) (d)

(e)

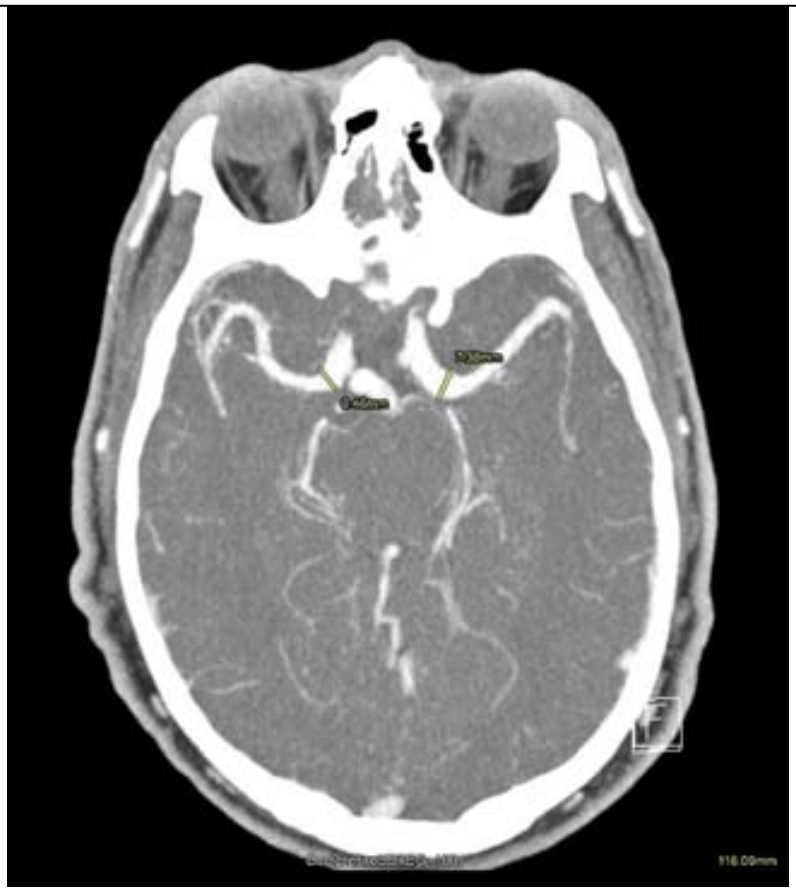


Figure 4: Cranial CTA demonstrating dilation of the arterial system of the intracranial anterior and posterior circulation

The cause of the cerebral vasculopathy in NF1 is not well understood. Histopathologic examination and electron microscopy have revealed intimal proliferation, smooth muscle nodules, thinning and fibrosis of the media and fibrosis of the adventitia in cerebral and other vessels [9-13]. In summary, we describe a patient with NF-1 who was found to have intracranial dolichoectasia affecting both the anterior and posterior circulations. Intracranial dolichoectasia has been associated with an increased incidence of cerebral small vessel disease, including lacunar strokes, leukoaraiosis, and status cribrosus [1,14]. There has been no report of dolichoectasia causing large vessel ischemic stroke, and in the case we present the patient's clinically severe stroke from MCA occlusion was likely to have been iatrogenic in origin. In summary, the cerebrovascular dolichoectasia that occurs with NF-1 can involve the anterior and posterior portions of the cerebral circulation, each independently or together, and produce serious neurological complications.Therapeutic intervention intended to treat those complications may in turn produce an adverse clinical outcome. An increased awareness of dolichoectasia and its association with NF-1 potentially could result in earlier detection and intervention, before significant complications take place.

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