SHORT REPORT

Axillary Artery Aneurysm in Tuberous Sclerosis—a Report of Two Cases

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Introduction

Tuberous sclerosis is an autosomal dominant disease characterised by the clinical triad of seizures, mental retardation and adenoma sebaceum. Although dysplasia and hamartomatous proliferation of small vessels including haemangiomas are common, disease of the medium and large sized blood vessels are rare.1 We report two cases of tuberous sclerosis presenting with axillary artery aneurysm.

Case Report

Case 1

A three month old male child with known tuberous sclerosis presented with ischemia affecting the tips of the fourth and fifth fingers of the left hand. A pulsatile axillary mass was felt, but with no clinically palpable pulse at the wrist. The hands were symmetrical in size but the left hand was cold, with delayed capillary filling at the pulp of the medial two fingers.

A colour duplex scan and a selective left subclavian angiography were performed. These demonstrated a smooth stenosis of the axillary artery followed by an aneurysm of the proximal brachial artery. (Fig. 1). The brachial artery was occluded in the region of the antecubital fossa. Collaterals reformed the proximal few centimetres of the radial and the ulnar artery, which were distally occluded in the mid forearm. The distal forearm and the hand were supplied by collaterals, but no radial, ulnar or palmar arteries could be identified.

Exploration of the brachial artery and its bifurcation was performed because of clinical deterioration. The arterial wall was markedly thickened causing narrowing of the lumen. There was weak, non-pulsatile forward flow, with very little back bleeding. In the absence of any distal run off, no arterial bypass was feasible. The aneurysm was left undisturbed because there was no risk of distal embolization. Histology of the brachial artery revealed almost complete occlusion of the lumen by fibroelastic intimal hyperplasia.

Five months after the operation, there was mumification and dry gangrene of the medial four digits, requiring amputation at the level of metacarpophalangeal joints.

Case 2

The second case was a two year old boy with known tuberous sclerosis, who presented with a two week history of a progressively enlarging swelling in the right axilla. Examination confirmed the presence of a 5 cm mass with expansile pulsation. The distal pulses were palpable, and the right hand was slightly larger than the left hand.

An angiogram showed two aneurysms involving the axillary artery (Fig. 2). The aneurysms were
excised and an interposition bypass with a reversed basilic vein graft was performed. The first part of the axillary artery, although not aneurysmal, had a grossly thickened wall. Pathological examination of the excised artery showed aneurysmal dilatation, associated with thickening of the vessel wall caused by dense cellular collagenous tissue. After seven years of follow-up the graft remained patent.

Discussion

Tuberous sclerosis is characterised by the presence and growth of hamartomatous lesions in almost every organ of the body. Brain lesions are consistently observed, in the form of calcified nodules that are clearly depicted by MRI or CT scan. Dysplasia and hamartomatous proliferation of small vessels are also seen.¹

Involvement of large and medium sized arteries has been reported, and this usually takes the form of aneurysm formation or ectasia. There have been reported cases of aneurysm formation of the thoracic aorta, the abdominal aorta and the intracranial vessels.¹,² Arterial stenosis is rare, and has been reported to affect the iliac, renal and mesenteric vessels.³

Pathologically, large muscular and musculoelastic arteries show fibrocytic and myofibrocytic intimal proliferation resulting in severe luminal stenosis.¹,³ The wall of the aneurysm sac shows thickening of the intima and atrophy of the media. There is also extensive degeneration of the medial smooth muscles and elastic fibres, which leads on to aneurysm formation.²,³ The cause of this medial degeneration is not clear. It has been proposed that intimal hyperplasia in the vasa vasorum of a large musculo-elastic artery may lead to ischemia of the vessel wall, which contributes to medial degeneration and the formation of an aneurysm.³

In tuberous sclerosis, aneurysm of the peripheral arteries are rare. Only two cases of axillary artery aneurysm have been reported. The aneurysm was excised in one case, with no bypass operation being performed because of good collateral blood supply to the hand.⁴ In the other case, resection of the axillary artery aneurysm followed by a bypass was performed.⁵ We do not know of any reported
case of peripheral extremity aneurysm with associated stenosis and occlusion, in patients with tuberous sclerosis.

Our two cases illustrate the spectrum of vascular involvement in tuberous sclerosis. The first patient had a combination of aneurysmal and occlusive disease, with the occlusive component being more widespread and predominant. This resulted in poor collateral blood supply to the hand, and also precluded any surgical reconstruction. The second patient had predominantly aneurysmal disease, in which a resection with interposition graft could be performed.

References


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