Clinical Imaging 39 (2015) 677–681

Contents lists available at ScienceDirect

Clinical Imaging

journal homepage: http://www.clinicalimaging.org

Cervical dural arteriovenous malformation and large epidural venous varices in a rare adult presentation of congenital vascular bone syndrome

Vasant Garg [a], Sunil Manjila [b], Mark Corriveau [c], Nicholas C. Bambakidis [b], Jeffrey L. Sunshine [a,*]

[a] Interventional Neuroradiology, Department of Radiology, Neurological Institute, University Hospital Case Medical Center, Cleveland, Ohio
[b] Department of Neurosurgery, Neurological Institute, University Hospital Case Medical Center, Cleveland, Ohio
[c] Case Western Reserve University School of Medicine, Cleveland, Ohio

ARTICLE INFO

Article history:
Received 7 September 2014
Received in revised form 27 December 2014
Accepted 14 January 2015

Keywords:
Cervical dural AV malformation
Venous varices
Congenital vascular bone syndrome
Servelle–Martorell syndrome
Klippel–Trenaunay syndrome

ABSTRACT

61-year-old male presented with shortness of breath and chest pain. Workup for acute myocardial infarction was negative; however, computed tomography angiography visualized what a vascular malformation within the cervical spinal canal. Given the patient's history of Servelle–Martorell syndrome, neurovascular imaging was performed. We present the unique vascular findings of a right thyrocervical trunk-based dural arteriovenous malformation (dAVM) and a large epidural venous varix. The cervical dAVM induced intervertebral foraminal widening and polyradiculopathy, representing a rare adult case of congenital vascular bone syndrome.

1. Introduction

The description of congenital appendicular and axial vascular lesions resulting in soft tissue and osseous changes has been recently termed *congenital vascular bone syndrome*[1]. Numerous eponyms were initially coined to describe specific clinical presentations, of which Klippel–Trenaunay, Parkes Weber, and Servelle–Martorell are identified by varicose veins and limb hypertrophy. Both Klippel–Trenaunay and Parkes Weber syndromes were originally thought to be associated with limb lengthening; these two syndromes are differentiated by cutaneous capillary malformations in Klippel–Trenaunay and the presence of arteriovenous malformations (AVMs) and shunting in Parkes Weber syndrome. Servelle–Martorell, on the other hand, was thought to consist of soft tissue enlargement but with osteohypoplasia and limb foreshortening. However, due to inconsistencies in presentations with patients not demonstrating all of the expected clinical and/or radiological characteristics for a particular syndrome as well as overlap of signs among the different diagnoses, more recent researchers have characterized the presentations under the umbrella term *congenital vascular bone syndrome*. All of these vascular lesions are typically congenital and usually present in children and young adults. We report a rare presentation of the congenital vascular bone syndrome spectrum, wherein an adult patient presented with a cervical dural AVM (dAVM) and a large epidural varix, resulting in polyradiculopathy.

2. Case history

A 61-year-old male with systemic hypertension and the previous diagnosis of Servelle–Martorell syndrome presented to our hospital complaining of shortness of breath and central chest pain radiating to his right shoulder and down his right arm. Electrocardiogram and cardiac enzymes were negative; however, systolic blood pressure was elevated to over 210 mmHg. Although the patient's blood pressure subsided with clonidine and metoprolol, his chest pain persisted. An emergent computed tomography angiography (CTA) of the chest was performed, which was negative for vascular pathologies such as aortic dissection. Instead, the CTA partially visualized an enhancing structure extending into the spinal canal at the vertebral cervicothoracic junction, suggestive of a vascular pathology. This finding was concerning for a vascular malformation and thought to correlate to the patient's history of Servelle–Martorell syndrome.

His physical exam revealed bluish discoloration overlying a bulging compressible diffuse lesion on the patient's bilateral upper extremities. Neurological exam demonstrated evidence of right triceps weakness and 50% sensory loss in the right C7 dermatome. There were no neurological findings suggestive of myelopathy (Figs. 1 and 2).

Subsequent magnetic resonance angiography delineated a complex cervical spinal vascular pathology associated with a large varix extending from approximately the C6 level down to the T1 vertebral body, for which a radiological differential diagnosis of dAVM versus AVM was entertained. Conventional catheter angiogram confirmed a complex dural-based AVM with a large nidus measuring 18.5×24 mm located to the right of midline and spanning the C5–C6 vertebral levels, predominately supplied by the right thyrocervical trunk. In addition,

* Corresponding author. Department of Radiology, University Hospital Case Medical Center, Cleveland, Ohio 44106. Tel.: +1 216 844 3112; fax: +1 216 844 3905.
E-mail address: Jeffrey.Sunshine@UHHospitals.org (J.L. Sunshine).

http://dx.doi.org/10.1016/j.clinimag.2015.01.012
0899-7071/

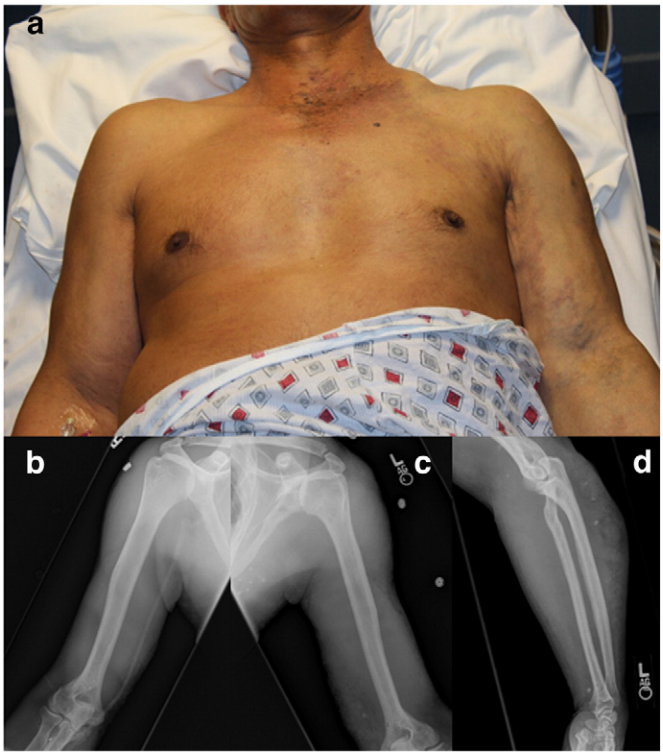

Fig. 1. (a) Compressible soft tissue mass lesions with bluish discoloration overlying the patient's bilateral upper extremities, left greater than right, and left supraclavicular region, concerning for venous malformations. (b, c, and d) Plain radiographs of the right humerus, left humerus, and left forearm with soft tissue enlargement and scattered calcifications amongst serpiginous nodular densities suggestive of phleboliths and associated vascular malformations, respectively.

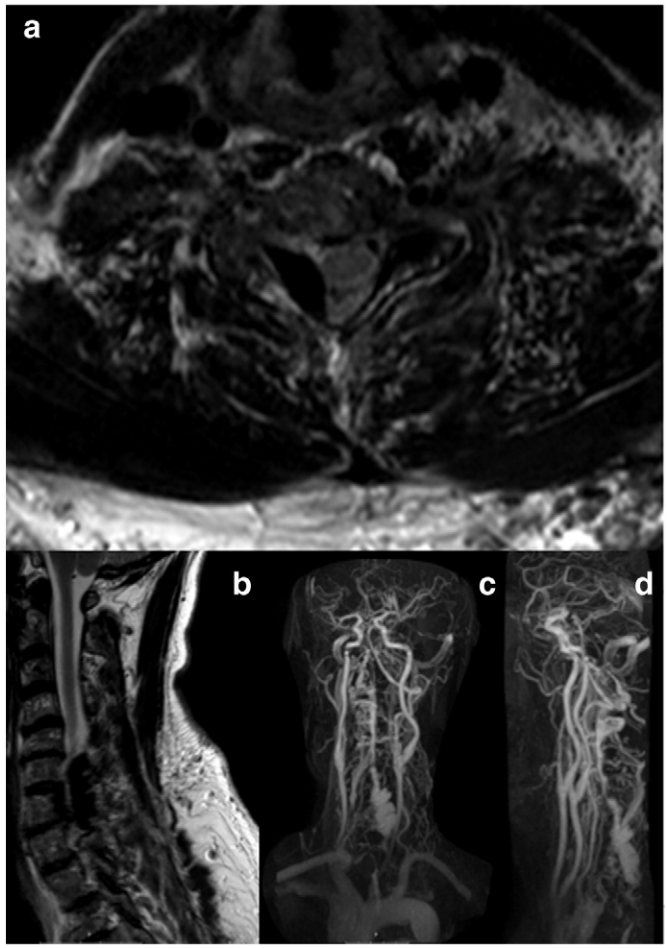

Fig. 2. (a, b) Magnetic resonance imaging T2 axial and sagittal slices of the neck with a large epidural varix extending from the level of C6 down to T1 vertebral body. (c, d) Magnetic resonance angiography coronal and left sagittal projections confirmed a large arteriovenous fistula without full discrimination of its complex angioarchitecture.

we noted a large venous varix in the posterior right epidural space communicating with a dilated posterior vertebral venous plexus (Figs. 3 and 4).

Medical records from a prior admission for an episode of noncardiac chest pain revealed that the patient's Servelle–Martorell syndrome was first diagnosed in 2008. Along with anemia and leukopenia, he was found to have erosive skull lesions on CT head concerning for multiple myeloma. Concern for an oncologic process warranted an extensive workup with immunoglobulin assays, which returned negative. Detailed physical exam during that admission had further revealed a bluish discoloration overlying diffuse compressible lesions on the patient's bilateral upper extremities concerning for venous malformations. This was further corroborated with radiography of the upper extremities revealing limb hypertrophy with prominent soft tissues calcifications suggestive of phleboliths and AVMs. A review of Medline publications at that time suggested that the patient's presentation fit the Servelle–Martorell syndrome. The patient remained relatively asymptomatic except for occasional mild left shoulder pain until the current admission.

Considering the presence of a symptomatic complex cervical dAVM with lateralizing neurological deficits and its natural history including disease progression, the overall clinical prognosis was discussed with the patient and his family. The patient refused any interventional treatment, returned to his home outside the United States of America, and hence was lost to clinical follow-up.

3. Discussion

The term Klippel–Trenaunay syndrome was coined after French neurologists Klippel and Trenaunay who in 1900 described a series of patients demonstrating a single hypertrophic extremity with superimposed varicose veins and ipsilateral cutaneous hemangioma, typically presenting as a port-wine nevus [2]. Shortly thereafter, in 1907, Parkes Weber described additional cases with a similar constellation of findings but with the presence of high-flow lesions within the extremity such as arteriovascular malformations, and therefore arose Parkes Weber syndrome [3]. Servelle in the 1940s and 1950s described 80 cases of Klippel–Trenaunay syndrome, of which fibrous bands or aberrant arteries were found to compress deep venous channels resulting in varicosity in 79 of the patients; however, in one deviant case, no obstructing lesion was present and instead agenesis was present [3]. His research in combination with that of Martorell was later felt to potentiate a third clinical entity characterized by limb hypertrophy, varicose veins, and capillary malformations but osseous hypotrophy and limb foreshortening [4,5].

Over the last century since the first cases of Klippel–Trenaunay were illustrated, multiple factors have confused the definition of each eponym. Each of the original authors presented cases with inexact characteristics and often anecdotal exceptions. Klippel and Trenaunay acknowledged "incomplete" cases without one of the three signs [6]. Subsequent researcher have "published cases of Klippel–Trenaunay syndrome with a–v fistulas, cases of Parkes Weber syndrome without fistulas,… [and] cases of Klippel–Trenaunay syndrome with shortening of the limb" [7]. As a result, additional terms have been published including congenital angiectactic hypertrophy, infantile angiectactic osteohyperplastic syndrome, osteoangiohypertrophic syndrome, and