Servelle-martorell syndrome with severe orthostatic hypotension in a pediatric patient

Yuri Igor López-Carrera, Max Bernal-Moreno¹, Oscar Colin², María Teresa García-Romero, Carola Durán-McKinster
Departments of Dermatology, ¹Radiology and ²Orthopaedic Surgery, National Institute of Pediatrics, Mexico City, Mexico

ABSTRACT

Servelle-Martorell syndrome (SMS) is an extremely uncommon vascular malformation characterized by venous malformations (VMs), limb overgrowth, and bone hypotrophy. The most common complications are venous thrombosis and pathological fractures. Ectasia and aneurysmal dilatations of the superficial veins may result in overgrowth of soft tissues but shortening of the affected limb. We report an 11-year-old Hispanic boy with an enlarged lower limb and VM. The patient suffered a fall at 9 years of age resulting in ankylosis of the knee without the possibility of limb extension or walking. When he tried to stand up, a severe orthostatic hypotension and almost loss of consciousness were present. An angio computed tomography and an arteriography demonstrated a complete absence of the deep venous system and bony hypotrophy compatible with SMS.

Key words: Bone hypotrophy, limb overgrowth, Servelle-Martorell syndrome, venous malformation

INTRODUCTION

Congenital vascular-bone syndrome is an alteration in limb growth caused by congenital vascular malformations. Limb overgrowth is observed particularly in Klippel-Trenaunay, Parkes Weber syndrome, and Servelle-Martorell syndromes (SMSs), which are characterized by capillary, venous, lymphatic or arterio-venous malformations.[1] The first two syndromes are associated with bone hypertrophy while SMS present with osteohypoplasia and limb foreshortening.[2,3] Belov[4] stated the relationship of bone shortening due to a global flow reduction in a limb.

SMS present with venous hypoplasia or aplasia in the deep system and a partial or complete lack of valves. Multiple phleboliths with severe bone destruction are observed. Most cases involve the lower limb; rarely, the upper limbs are affected.[5,6]

CASE REPORT

An 11-year-old Hispanic boy presented for evaluation in the clinic of vascular anomalies of our institute with an enlarged lower limb and VM. The patient suffered a fall at 9 years of age resulting in ankylosis of the knee without the possibility of limb extension or walking. When he tried to stand up, a severe orthostatic hypotension and almost loss of consciousness were present. An angio computed tomography and an arteriography demonstrated a complete absence of the deep venous system and bony hypotrophy compatible with SMS.

How to cite this article: López-Carrera YI, Bernal-Moreno M, Colin O, García-Romero MT, Durán-McKinster C. Servelle-martorell syndrome with severe orthostatic hypotension in a pediatric patient. Indian J Paediatr Dermatol 2017;18:299-302.
walking since then. The left limb was slightly shorter than the right leg and was occasionally painful. When the patient was asked to stand up, he felt dizzy and almost lost consciousness. His parents reported this happened frequently; thus, he avoided standing up.

Radiographs showed a right lower limb without abnormalities. In the anterior-posterior projection, the left lower limb presented important soft-tissue swelling. Hypotrophy and decreased bone density with irregular and thin cortical were evident as well as decreased joint spaces. There were multiple phleboliths in the entire limb, both in the superficial and profound soft tissues from 2 to 10 mm in diameter. The knee joint had severe arthrosis with a 70° flexion contracture. Levoconvex Cobb scoliosis of 30° with apex in thoracic vertebrae T9 was observed caused by the knee contracture.

The Doppler ultrasound showed multiple dilated tortuous anechoic gaps from the pelvic region to the entire left lower limb, especially in the subcutaneous fat and intramuscular tissues. Slow flow color Doppler showed no record saturation in larger vascular beds.

In the high caliber superficial veins, there was no evidence of valves. No patency of the deep venous system could be demonstrated.

An angio computed tomography showed confluent and countless superficial venous vessels in the subcutaneous tissues, which reinforced irregularly in the late phase of the intravenous contrast [Figure 1b]. All muscles were small and heterogeneous throughout the limb, including the foot. The diameter of long bones was decreased, shortened in length and curved [Figure 2]. There was no deep venous system. Arteries showed decreased diameter without abnormal reinforcement in the arterial phase [Figure 3]. There were multiple phleboliths, rounded, and well defined in the scrotum, hip, thigh, and leg.

An arteriography showed arterial hypoplasia in the common, superficial, and deep femoral system. The venous return was at expenses of small superficial veins. Low flux and blood stasis in the entire superficial system were evident. There was no evidence of permeable deep venous system.

With these clinical and radiological findings, the diagnosis of SMS was made based on the hypoplasia of the deep venous system, bone hypotrophy, and soft-tissue swelling of the entire left lower limb. Because of the 70° knee contracture and the
impossibility of leaning his foot for walking, important muscle atrophy of the entire affected limb was present as well as osteoporosis of most of the bones. The foot had an equine attitude, reducible with passive-active mobilization. Levoconvex Cobb scoliosis in T9 is also secondary to the knee flexion and pelvic balance.

Treatment: A trial of intramuscular injections of botulinum toxin was prescribed to improve muscle contraction without any benefit. The patient is receiving physical therapy, and the flexibility of his knee has improved slightly.

**DISCUSSION**

Bone growth depends both on bone-forming osteoblasts and on bone vasculature and is controlled by local and systemic factors. In SMS, also known as angio-osteohypotrophic syndrome, bone shortening occurs due to intraosseous dysplastic vessels and flow reduction, which destroys the spongiosa and cortical of the bone. Substitution and destruction of the epiphyseal cartilages may be observed.

Venous malformations (VMs) present with a wide range of clinical presentations. The association between congenital vascular malformations and altered bone growth, the so-called congenital vascular bone syndrome, has been proposed to describe abnormal enhancement or reduction of growth in long bones due to pathologic circulation during childhood. Limb lengthening is well known in Klippel–Trenaunay and Parkes Weber syndromes, while limb length difference but with shortening of the pathologic limb is characteristic of SMS. VMs are observed as compressible and tender bluish, ectatic and dilated veins on the skin of the affected limb, and with time, progressive overgrowth of soft tissues occurs at the same time as shortening of the limb and bone hypoplasia. In many cases, additional destruction of a joint is present. Involvement of part of limbs is more common and rarely involves whole of an extremity. Bhatnagar reported a 12-year boy who presented whole of left upper limb, scapular region, and axilla involvement.

Diagnosis is made from the clinical features and imaging studies. A simple radiograph may reveal multiple well-defined, radiopaque lesions consistent with phleboliths at the age of 2 or 3 years. Arteriography and phlebography are required to demonstrate the ectatic regions of the involved vessels. In the deep venous system, partial or complete lack of valves and venous aplasia or hypoplasia is the main characteristic. Magnetic resonance is the best examination to delimit vascular malformation.

In our patient, at 9 years of age, he presented a fall playing football, which resulted with hemarthrosis and followed by ankylosis and the impossibility to walk since then. When he tried to stand up, severe orthostatic hypotension resulted with elevated heart rate, pale mucous membranes, and dizziness, which normalized as soon as he sat down. We believe that because of the absence of a deep venous system, blood accumulates in the ectatic vascular canals of the limb causing consequent orthostatic hypotension.

The prognosis of this malformation is uncertain. The most common complication is venous thrombosis, and the thrombi may be palpated at the site of pain and correspond to phleboliths in the X-rays. Another possible complication is a pathological fracture of the bone due to severe osteoporosis. Less common is the development of consumption coagulopathy due to stasis in the ectatic vascular canals although it should be investigated before any invasive procedures.

Treatment must be conservative and includes external compression with graduated compressions stockings, especially if extensive edema is present. Surgical treatment should be done when aneurysmal complications are found. Arterial embolization is contraindicated because it may provoke tissue necrosis. Amputation could be necessary when the limb is grossly hypertrophied to improve life quality.

At this point, we will continue to treat our patient conservatively and, depending on complications which may arise, look for other options that may improve his quality of life.

We believe this case is important to recognize because patients with SMS are extremely uncommon, and frequently are confused with Klippel–Trenaunay or Parkes Weber syndromes.

**Financial Support and Sponsorship**

Nil.

**Conflicts of Interest**

There are no conflicts of interest.

**REFERENCES**

1. Mattassi R. Differential diagnosis in congenital vascular-bone syndromes. Semin Vasc Surg 1993;6:233-44.
2. Servelle M, Trinquecoste P. Des angiomes veineux. Arch Mal
Coeur Vaiss 1948;41:436-42.
3. Martorell F. Hemangiomatosis braquial osteolática. Angiologia 1949;1:219-23.
4. Belov S. Haemodynamic pathogenesis of vascular-bone syndromes in congenital vascular defects. Int Angiol 1990;9:155-61.
5. Karuppal R, Raman RV, Valsalan BP, Gopakumar TS, Kumaran CM, Vasu CK. Servelle-Martorell syndrome with extensive upper limb involvement: A case report. J Med Case Rep 2008;2:142.
6. Bhatnagar A, Deshpande M. Rare case of Servelle Martorelle syndrome. Kathmandu Univ Med J (KUMJ) 2012;10:91-4.
7. Vollmar JF, Paes E, Irion B, Friedrich JM, Heymer B. Aneurysmic transformation of the venous system in venous angiodysplasias of the limbs. Vasa 1989;18:96-111.
8. Weiss T, Mädler U, Oberwittler H, Kahle B, Weiss C, Kühler W. Peripheral vascular malformation (Servelle-Martorell). Circulation 2000;101:E82-3.
9. Mattassi R, Vaghi M. Vascular bone syndrome – Angio-osteodystrophy: Current concepts. Phlebology 2007;22:287-90.
10. Gibbon WW, Pooley J. Pathological fracture of the femoral shaft in a case of Servelle-Martorell syndrome (phleboeclastic osteohypoplastic angiodysplasia with associated arterio-venous malformation). Br J Radiol 1990;63:574-6.