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Title: Servelle-Martorell Syndrome With Extensive Upper Limb Involvement - A Case Report

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Version: 5 Date: 29 January 2008

Author's response to reviews: see over
Servelle-Martorell Syndrome With Extensive Upper Limb Involvement - A Case Report

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ABSTRACT:
Angio-osteohypotrophic syndrome is also known as Servelle-Martorell angiodysplasia. It is characterized by venous or rarely, arterial malformations and bony hypoplasia. There will be aneurysmal dilatation of the superficial veins which may result in limb hypertrophy and bony hypoplasia. We report a case of Servelle-Martorell syndrome with extensive involvement of the entire upper limb and periscapular region. Extensive limb involvement is rare in Servelle Martorell syndrome. Cases of only minimal upper limb involvement have been described in the English literature. This article highlights Servelle-Martorell syndrome as one of the causes of angiodysplastic limb hypertrophy.

Introduction
Servelle-Martorell syndrome is characterized by limb hypertrophy due to venous and rarely, arterial malformations with skeletal abnormalities (hypoplasia)\(^1\). The similar conditions like Klippel-Trenaunay, Parkes-Weber and Blue rubber bleb nevus syndromes can present with limb and bone hypertrophy. Adequate imagings with corroborative clinical findings are crucial to establish the correct diagnosis. MRI is the best imaging method for diagnosis\(^1\). The prognosis of this disorder is uncertain. Therapy is
predominantly conservative. In the presence of aneurysmal complications or severe shunting, surgery may be indicated. A very few cases of this rare syndrome were reported in English literature, to the best of our knowledge.

**Case Presentation**

A 21-year-old male presented with enlargement of right upper limb and difficulty in using the right upper limb since birth. There was occasional pain in the right upper limb. Pain and the soft tissue enlargement aggravated on keeping the limb dependent. Physical examination showed multiple swellings involving the whole of upper limb extending to the axilla and periscapular region (Fig.1). The swellings were of different sizes, soft in consistency and compressible and it reduced in size on elevating the limb. The palm had bluish discoloration. Other parts of the body showed no abnormalities. The right clavicle, arm, forearm and hand were shorter than the left side. The peripheral pulses were palpable with equal volume on both sides. Forearm and hand bones were hypoplastic and tender. His pectorals, periscapular, deltoid, biceps and triceps muscles were palpable though atrophic. There was no sensory deficit. His motor power of right upper limb was MRC grade III to IV. No bruits or thrills were found. No temperature difference was observed. His elbow had a fixed flexion deformity of 80° with further full flexion. Cardiovascular system was normally functioning.
Investigation revealed a normal blood picture. Radiographs showed multiple soft-tissue swellings and hypotrophy of the bones of right upper limb. There were multiple well-defined radio opaque lesions consistent with phleboliths in the affected upper limb and periscapular region (Figure 2).

Musculoskeletal ultrasound showed multiple dilated tortuous anechoic lesions involving the upper limb and the periscapular region. Echogenic lesions with shadowing suggestive of phleboliths were seen inside the anechoic lesions. The forearm muscles were thinned out and replaced by these anechoic lesions.

Color Doppler study - showed no flow within the lesion but while doing Valsalva maneuver there was sluggish flow within the lesion suggestive of dilated torturous venous channels involving the superficial venous system. Proximal part of the deep venous system appears normal but distal part was not visualized. The arterial system appeared normal.

MRI study showed multiple dilated veins in the superficial aspect of right upper limb (Fig.3). Muscles of the limb were replaced by abnormal signal. Triceps, biceps and deltoid were partially involved. No intraosseous venous malformation was seen. Arteries were normal.
We managed this case non-operatively by external compression with graduated compression stockings. Compression therapy helped to diminish his symptoms of venous insufficiency. Our patient does not have any of the complications like venous thrombosis, consumption coagulopathy, recurrent cellulitis, or recurrent bleeding.

**Discussion**

Servelle-Martorell syndrome is also known as phlebo ecstatic osteohypoplastic angiodysplasia. The ectasia and aneurysmal dilatation of the superficial veins may result in a monstrous deformity of the extremity. In the deep venous system - an abnormal vein location, partial or complete lack of valves, and/or venous hypoplasia or aplasia has been observed. Intraosseous vascular malformations may lead to hypoplasia of bone with destruction of spongiosa and cortical bone, resulting in shortening and hypoplasia of the limb. Intraosseous vascular ectasias can result in joint destruction. Multiple phleboliths can be found in venous ectasias on the radiograph. The prognosis of this disorder is uncertain.

Venous vascular malformations span over a wide spectrum, varying from isolated cutaneous ectasias to voluminous lesions involving manifold tissues and organs. They are soft and compressible, and show no alteration in skin
temperature, thrill or bruits. They are frequent and wrongly called cavernous hemangiomas. Pure venous malformations usually exhibit blue coloration on the skin or in the overlying mucosa, while combined venous and capillaries exhibit a hue that ranges from dark-red to violet\textsuperscript{1,3}. The venous malformations are hemodynamically inactive, with a low flow. The condition deteriorates with pregnancy or trauma\textsuperscript{4, 5}. Absence of overgrowth of the limbs distinguishes it from combined vascular malformations, such as Klippel-Trenaunay syndrome. There may be demineralization, hypoplasia or lytic changes in the underlying bones in up to 71\% of cases\textsuperscript{4}.

Venous thrombosis is a regular complication, and the thrombi may be palpated at the point of pain, but in our case there was no complications related to the venous thrombosis. Another possible complication is the development of consumption coagulopathy due to stasis in the ectatic vascular canals\textsuperscript{6}. The possibility of consumption coagulopathy must be investigated prior to undertaking any invasive procedures\textsuperscript{3, 4, 5}.

The diagnosis is from clinical features for most cases, but a simple radiography may reveal phleboliths and bone hypoplasia at the age of two or three years. Magnetic resonance is the best examination to delimit vascular malformation\textsuperscript{1}.\n
Based on the observations of 47 cases of angiodysplasia of types Parkes-Weber, Klippel-Trenaunay and Servelle-Martorell, Langer M et al demonstrated that differentiation of these 3 syndromes is possible by taking standard X-rays of the extremities (both sides) which are examined under direct magnification (0.1-01 mm). In this way, the Weber syndrome should be suspected if bone lengthening is seen in association with loss of substances from the skeleton. In the Klippel-Trenaunay syndrome the bony lesions do not accompany lengthening. In the Servelle-Martorell syndrome bony lesions go hand in hand with limb hypertrophy\textsuperscript{1,4}.

Arteriography is not required in Klippel-Trenaunay type of angiodysplasia whereas arteriography as well as phlebography is necessary in the Servelle-Martorell type of angiodysplasia, to demonstrate ectatic regions of the involved vessels\textsuperscript{10}.

Majority of the reported cases were having limited area of involvement\textsuperscript{2}. Extensive involvement of the entire upper limb and the periscapular region made our case as a rare one.

Management in most patients with Servelle-Martorell syndrome should be non-operative. Nonoperative management includes external compression with graduated compression stockings and garments. Compression therapy
can be very helpful to protect the limb, even from minimal trauma that can cause bleeding of the large superficial malformations. It has no effect, however, on the ultimate size of the limb. Patients with significant edema of the lower limbs can be treated with diuretics. Sclerotherapy, with a local injection of sclerosing solutions, like 95% alcohol or sodium tetradecyl sulfur 1% can be used for small lesions. Surgical resection may be performed after successful obliteration by sclerotherapy. The embolization of arteries sustaining the malformation is counter indicated since it may provoke tissue necrosis.

Patients with recurrent attacks of cellulitis may benefit from prophylactic antibiotic therapy. Anticoagulants are indicated after deep vein thrombosis or pulmonary embolus. Patients with recurrent superficial thrombophlebitis frequently require daily administration of aspirin or ibuprofen. However, this may promote bleeding problems.

Surgery should not be done to improve cosmesis at the expense of function. Presence of aneurysmal complications or severe shunting may be an indication for surgery. Surgical excision is the definitive therapy, often rendered impossible however by anatomic, esthetic and functional limitations.¹.⁸ Amputation of a grossly hypertrophied, poorly functioning
digit may be necessary but a more proximal foot, hand, or limb amputation is rarely required. Symptomatic varicosities or localized venous malformations can be removed in selected patients with good results provided that there is a functioning deep vein system. It should be recognized that complete excision of extensive malformations with debulking procedures seldom is possible. Debulking procedures can damage venous and lymphatic structures and lead to increased edema of the affected part, scar formation, recurrence, chronic wound infection, and chronic weeping lymphoedema.

Conclusion

Servelle-Martorell syndrome is a very rare condition, which makes a diagnostic confusion with, Klippel-Trenaunay, Parkes-Weber and blue rubber bleb nevus syndrome. Venous malformations are present in all these conditions; bony hypoplasia is characteristic in Servelle-Martorell syndrome. Though it is rare, extensive limb involvement may be seen in Servelle-Martorell syndrome. MR imaging is very useful in assessing the extension of venous malformations. Conservative treatment is recommended in most cases. Sclerotherapy with or without surgery is recommended in cases of functional impairment, even if recurrences are frequent.
Declaration of competing interests:

There is no competing interest exists in our interpretation of data or presentation of information, since there is no personal or financial relationship with other people or organizations. Authors do not have any financial competing interests and also any non-financial competing interests.

References:


Figures Legend

Figure 1 Multiple soft tissue swelling involving the entire upper limb, axilla & periscapular region.
Figure 2 - multiple soft-tissue swellings, hypotrophy of the bone, and multiple well-defined radio opaque lesions consistent with phleboliths (X-ray)

Figure 3 - Multiple dilated veins in the superficial aspect of right upper limb with bone hypoplasia (MRI Scan)

Abbreviation List

MRI: Magnetic Resonance Imaging
MRC: Medical Research Council

Authors contributions:

Raju Karuppal:
Made substantial contributions to conception and design, acquisition of data, analysis and interpretation of data

Rajendran V Raman
Made substantial contributions to analysis and interpretation of the investigations

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Gopakumar T S
Made substantial contributions to conception and design, revised the manuscript critically for important intellectual content

Chathoth Meethal Kumaran
Made substantial contributions by revising and given final approval of the version to be published

Chembu Kara Vasu
Made substantial contributions to revise the manuscript critically for important intellectual content

Acknowledgment:

Authors deeply acknowledge Dr Anwar Marthya, Senior Lecturer in Orthopaedics, for his substantial contribution in design, drafting the manuscript and revising it critically.
Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

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