

Case Report

Servelle-Martorell Syndrome with Extensive Upper Limb involvement: A case Report

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Abstract:

Servelle-Martorell syndrome is characterized by venous or rarely arterial malformations with limb hypertrophy and bony hypoplasia. Extensive involvement of the upper limb is a rare feature of Servelle-Martorell syndrome. Cases with minimal upper limb involvement have been described in the literature. A young man presented with gradually increasing multiple swellings over the right upper limb and periscapular region with functional difficulty. The arm muscles were atrophic. The bones of forearm and hand were hypoplastic and tender. We report a case of Servelle-Martorell syndrome with extensive involvement of the entire right upper limb and periscapular region.

Key Words: Venous malformation.

Introduction:

Servelle-Martorell syndrome is characterized by limb hypertrophy owing to venous and rarely, arterial, malformations with skeletal abnormalities (hypoplasia). Similar conditions such as Klippel-Trenaunay, Parkes-Weber and Blue rubber bleb nevus syndromes can present with limb and bone hypertrophy. Magnetic resonance is the best imaging method for diagnosis. Adequate radiological investigations with collaborative clinical findings are crucial to establish correct diagnosis. The prognosis of this disorder is uncertain. Therapy is predominantly conservative. In the presence of aneurysmal complications or severe shunting, surgery may be indicated. Servelle-Martorell syndrome has been reported rarely in the literature.

Venous malformations (VM) are present at birth, although not always evident. They are bluish, soft and compressible and can be localized or extensive. They are usually present on the face, limbs or trunk. However, VM can also involve oronasopharynx, genitalia, bladder, brain, spinal cord, liver, spleen, lungs, skeletal muscles and bones.

Extensive involvement of whole right upper limb is very uncommon. We present a rare case of extensive venous malformations involving right periscapular region and upper limb.

Case Report:

A 23 year old male presented with history of gradual diffuse enlargement of right upper limb for last 15 years and functional difficulty. The swelling was small to begin with and progressively increased in size. Patient often complained of pain and stiffness in the affected areas. On clinical examination multiple swollen areas of various sizes were seen over right upper limb. They were soft and compressible. The right forearm was shorter in length than left side.

Plain radiographs of the right upper limb showed soft tissue masses and areas of calcification. The forearm bones revealed hypoplasia on right side.



Fig.I: Photograph showing extensive nodular soft tissue swellings involving whole right upper limb and periscapular region

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

Color Doppler study showed no flow within the lesion but, while performing a Valsalva maneuver, there was sluggish flow within the lesion suggestive of dilated tortuous venous channels involving the superficial venous system. The arterial system appeared normal.

Discussion:

Servelle-Martorell syndrome is also known as phlebectatic osteohypoplastic angiodisplasia (Weiss et al, 2000). The ectasia and aneurysmal dilatation of the superficial veins may result in a monstrous deformity of the extremity. An abnormal vein location, partial or complete lack of valves, and/or venous hypoplasia or aplasia has been observed in deep venous system.. Intra-osseous vascular malformations may lead to hypoplasia of the bone with the destruction of spongiosa and cortical bone, resulting in shortening and hypoplasia of the limb (Weiss et al, 2000). Intra-osseous vascular ectasias may lead to joint destruction. Radiographs can demonstrate multiple soft-tissue swellings, hypoplasia of the bones and multiple phleboliths in the venous ectasias. The prognosis of this disorder is uncertain.



Fig. II : AP Radiograph of right arm showing soft tissue swelling & multiple calcifications.



Fig. III: AP Radiograph of both Hands showing soft tissue swelling with multiple calcifications on right side. Ulna and first, second metacarpal bones are hypoplastic

Venous vascular malformations span 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. These are frequently and incorrectly termed 'cavernous hemangiomas'. Pure venous malformations usually exhibit blue coloration of the skin or in the overlying mucosa, while the combined venous malformations and capillaries exhibit a hue that ranges from dark-red to violet (Enjolras & Mulliken, 2000; Fishman & Mulliken, 1993).

The venous malformations revealed low flow. The condition deteriorates with pregnancy or trauma (Enjolras & Mulliken, 1998; Burns et al, 1991). There may be demineralization, hypoplasia or lytic changes in the underlying bones in up to 71% of cases (Enjolras & Mulliken, 1998).

Venous thrombosis is a regular complication, and the thrombi may be palpated at the point of pain. Another possible complication is the development of consumption coagulopathy due to stasis in the ectatic vascular canals. The possibility of consumption coagulopathy must be investigated prior to undertaking any invasive procedures (Fishman & Mulliken, 1993; Burns et al, 1991).

Diagnosis is made in majority of cases by clinical features. A simple radiograph may reveal phleboliths and bone hypoplasia at the age of 2 or 3 years. Magnetic resonance is the best investigation to delimit vascular malformation (Enjolras & Mulliken, 1998).

The majority of the reported cases had a limited area of involvement (Weiss et al, 2000). The extensive involvement of the entire right upper limb and the periscapular region made this case rare.

Nonoperative management is adequate for most patients with Servelle-Martorell syndrome. This includes external compression with graduated compression stockings and garments. Compression therapy can be helpful in protecting the limb, even from minimal trauma that can cause bleeding of the large superficial malformations. Patients with significant edema of the lower limbs can be treated with diuretics.

Sclerotherapy with local injection of 95% alcohol or 1% sodium tetradecyl sulphur may be used for small lesions. Surgical resection may then be performed following successful obliteration. The embolization of arteries sustaining the malformation is contraindicated since it may provoke tissue necrosis.

Patients with recurrent attacks of cellulites 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 problems with bleeding.

Surgery should not be done to improve cosmetic look at the expense of function. Aneurysmal complications or severe shunting may be an indication of the requirement for surgery. Surgical excision is the definitive therapy, often rendered impossible, however, by anatomic, esthetic and functional limitations (Enjolras & Mulliken, 1998). 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 is seldom 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 lymph

edema (Cliff & Mortimer, 2000).



Fig. IV: High resolution ultrasound of Right arm with 12 Mhz probe showing anechoic spaces with calcification

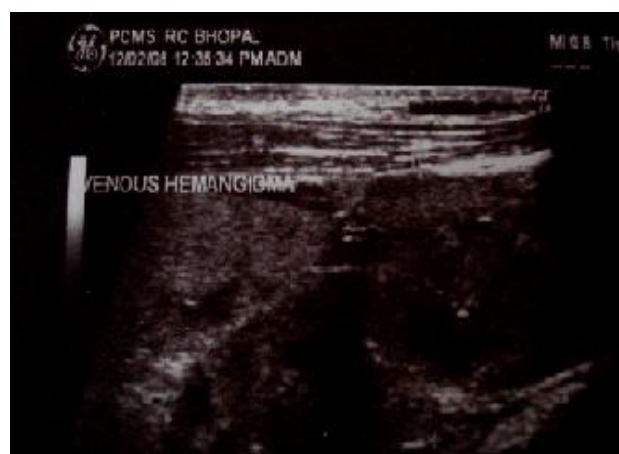


Fig. V: Sonography reveals irregular anechoic spaces with calcification.

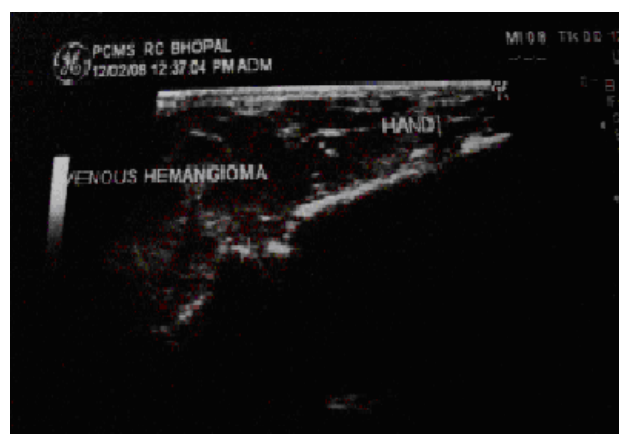


Fig. VI: Sonography of dorsal aspect of hand reveal irregular anechoic spaces.

Conclusion:

Servelle-Martorell syndrome is a rare condition, the diagnosis of which can be confused with Klippel-Trenaunay, Parkes-Weber and blue rubber bleb nevus syndromes. Venous malformations are present in all these conditions; bony hypoplasia is characteristic of Servelle-Martorell syndrome. Although it is rare, extensive limb involvement may be seen in Servelle-Martorell syndrome. Magnetic resonance imaging is useful in assessing the extent 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.

To conclude, it is very important to have a correct diagnosis for the vascular anomalies before the treatment. Proper understanding of the natural course and multidisciplinary approach is essential to tackle these complex anomalies. Management of large VM require role of expert interventional Radiologist. It is difficult to cure these lesions, and the aim is to control them and treat the associated complications.

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