A rare newly described overgrowth syndrome with vascular malformations- Cloves syndrome

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Abstract

There are many overgrowth syndromes described in the literature. Few are associated with vascular malformations. We describe a rare, newly described syndrome with features of overgrowth and vascular malformations.

Key words: Cloves, scoliosis, venous malformation

Introduction

There are many overgrowth syndromes described in the literature. Few are associated with vascular malformations. We describe a rare, newly described syndrome with features of overgrowth and vascular malformations.

Case Report

A 16-year-old boy presented with swellings in the anterior abdominal wall of both lumbar regions [Figure 1A]. There was also a large swelling over the back of left shoulder [Figure 1B]. The anterior abdominal wall swellings were gradually increasing in size for 15 years. These swellings were associated with discoloration over the skin surface. There was a recent history of spontaneous bleeding from the right anterior abdominal swelling. On examination, these swellings were soft and non-pulsatile.

General examination revealed flat foot [Figure 1A and C] wide gap between the first and second toes [Figure 1D]

bilaterally. Radiographs of both feet also confirmed the widened interspace between the first and second toes and the flat foot [Figure 2A]. On examination of the spine, there was scoliosis at the thoracic level.

Local examination revealed swellings in the anterior abdominal wall in the lumbar region. Multiple black and reddish discolorations were noted in the anterior abdominal wall swelling. There was also swelling in the back of the left shoulder. The skin over the swelling in the back appeared normal. USG doppler of the anterior abdominal wall showed dilated venous spaces in the subcutaneous plane [Figure 2B]. The patient underwent Computed tomography (6-slice CT scanner (Brilliance, Philips Medical Systems, Netherlands) and magnetic resonance imaging (MRI 3-TESLA MRI scanner, INTERA ACHIEVA Philips Medical Systems, Netherlands) of the abdomen. CT brain was also done.

MRI showed heterogeneous high signal in the anterior abdominal wall [Figure 3A and B] and in the paraspinal region [Figure 3C]. There was asymmetric increased fat deposition in the posterior abdominal wall [Figure 3A and B]. MRI also confirmed the scoliosis with apex to the right [Figure 3C].

Sclerotherapy was done for right anterior abdominal swelling. Sclerotherapy was planned to treat bleeding. Approximately 3 ml of sodium tetradecyl sulfate was injected into the right anterior abdominal wall swelling [Figure 4]. The patient came for follow-up after a period of 6 weeks. There were no episodes of bleeding.

Figure 1 (A-D): On general examination, there are. (A) swellings in the anterior abdominal wall (asterisk) (B) swellings over the back (long white arrow) (C) flat foot (short white arrow) (D) wide gap in between the first and second toes (black arrow)
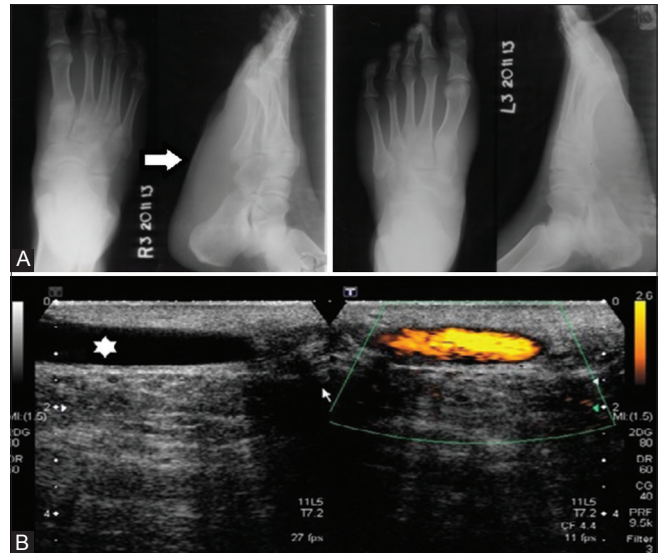

Figure 2 (A and B): (A) Radiographs of the feet show widened space between the first and second toes and bilateral flat foot (white arrow) (B) USG and color Doppler images showing venous malformation (Asterisk)
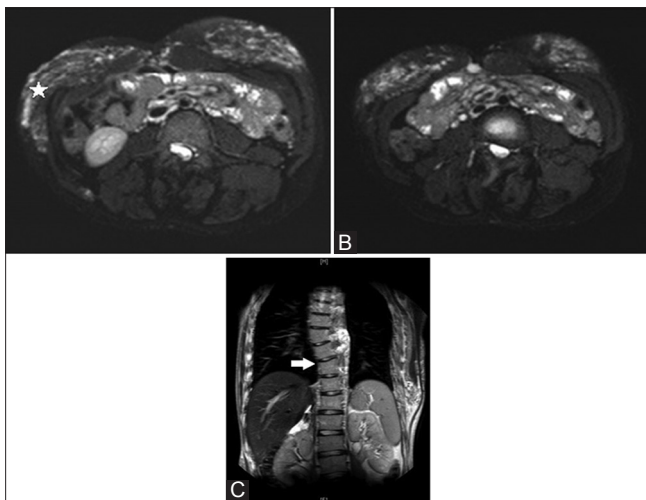

Figure 3 (A-C): (A and B) MRI-T2 STIR axial sections showed multiple heterogeneous high signal foci in the anterior abdominal wall (asterisk) (C) T2 coronal section showed scoliosis of the thoracic spine (arrow)

Diagnosis

The diagnosis made was CLOVES syndrome. Congenital Lipomatous Overgrowth, Vascular malformations, Epidermal nevi, and Skeletal anomalies are the key components of this syndrome. Only less than 20 cases have been described in the literature. This syndrome was described independently by Saap *et al.* and Alomari.[1,2] A similar case was already reported by Harit and Aggarwal[3] from India.

Discussion

Overgrowth syndromes with complex vascular anomalies are a rare group of syndromes which usually consists
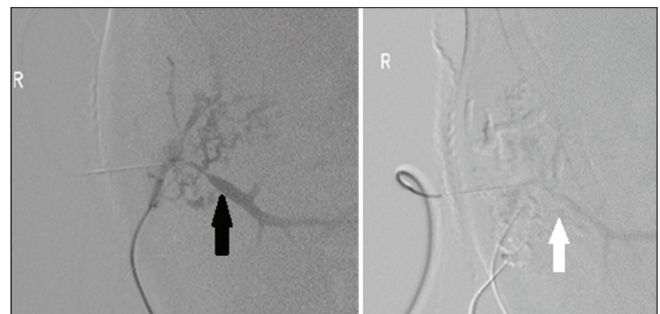

Figure 4: Sclerotherapy for the right anterior abdominal wall swelling. Black arrow points to venous channels filling with contrast. White arrow shows sclerosant filling the venous channels

of hypertrophy of one or more regions of the body and multifocal vascular anomalies.

There are many overgrowth syndromes. The common overgrowth syndromes are neurofibromatosis, Beckwith-Wiedemann syndrome, Proteus syndrome, and Sturge-Weber syndrome. Few of these syndromes are associated with vascular anomalies.

This syndrome can also be detected during prenatal period.[4] Features described in the prenatal period are truncal cystic mass and body and acral anomalies. Differentials for combination of these findings would include Klippel-Trenaunay, Proteus syndrome, and CLOVES syndrome.

Patients with CLOVES syndrome showed lipomatous masses (which is one of the component of the syndrome) causing asymmetric hypertrophy of the trunks and hemihypertrophy.

Central nervous system (CNS) involvement is also described in this syndrome. The features include polymicrogyria, non-contiguous abnormalities of the gray and white matter, a four-layered cortex, partial agenesis of corpus callosum, and ventriculomegaly.[5] Neural tube defects and tethered cord were also described by Saap *et al.* and Alomari.[2] The patient described here did not have any CNS malformations.

Musculoskeletal and acral anomalies include leg length discrepancy, chondromalacia patellae, dislocated knees, scoliosis, wide hands and feet, furrowed sole, wide sandal gap macrodactyly, and talipes. The patient discussed in this report had scoliosis, wide sandal gap, and talipes.

Vascular malformations are part of this syndrome. Vascular malformations included in this syndrome are epidermal nevus and low-flow malformations like venous and lymphatic malformation, and there are case reports in AJNR which also describe fast-flowing paraspinal arteriovenous malformations.[6]

The case discussed here had venous and lymphatic malformations in the anterior abdominal wall and lipomatous masses in the back.

We diagnosed this entity as CLOVES syndrome as there were congenital lipomatous overgrowth, vascular malformations, and skeletal findings.

References

1. Sapp JC, Turner JT, van de Kamp JM, van Dijk FS, Lowry RB, Biesecker LG. Newly delineated syndrome of congenital lipomatous overgrowth, vascular malformations, and epidermal nevi (CLOVE syndrome) in seven patients. Am J Med Genet A 2007;143A: 2944-58.

2. Alomari AI. Characterization of a distinct syndrome that associates complex truncal overgrowth, vascular, and acral anomalies: A descriptive study of 18 cases of CLOVES syndrome. Clin Dysmorphol 2009;18:1-7.

3. Harit D, Aggarwal A. CLOVE syndrome with nevus unis lateris: Report of a case. Pediatr Dermatol 2010;27:311-2.

4. Fernandez-Pineda I, Fajardo M, Chaudry G, Alomari AI. Perinatal clinical and imaging features of CLOVES syndrome. Pediatr Radiol 2010;40:1436-9.

5. Gucev ZS, Tasic V, Jancevska A, Konstantinova MK, Pop-Jordanova N, Trajkovski Z, *et al*. Congenital lipomatous overgrowth, vascular malformations, and epidermal nevi (CLOVE) syndrome: CNS malformations and seizures may be a component of this disorder. Am J Med Genet A 2008;146A: 2688-90.

6. Alomari AI, Chaudry G, Rodesch G, Burrows PE, Mulliken JB, Smith ER, *et al*. Complex spinal-paraspinal fast-flow lesions in CLOVES syndrome: Analysis of clinical and imaging findings in 6 patients. AJNR Am J Neuroradiol 2011;32:1812-7.