CASE STUDIES

Venous malformation of lower limb with hypertrophy of limb, early terminalization of hair and hyperhidrosis

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Abstract

Vascular anomalies are the commonest developmental disorders. Venous malformations (VM) result due to dysmorphogenesis in the development of veins. Most commonly these disorders are localized to skin and subcutaneous tissue. Deeper venous malformation may affect the underlying muscle, bone and joints. Early terminalization of hair and increased sweating in VM plaque has been rarely reported. The present study deals with the case of a 15-year-old boy who had VM involving skin, subcutaneous tissue and bones with terminal hair and increased sweating over the plaque. Radiological examination showed bone hypertrophy with slow flow channels and phleboliths due to superficial and deep VM. He was treated with ethanol sclerotherapy.

Keywords: Venous malformation, intra-osseous involvement, early terminalization of hair, hyperhidrosis

Introduction

Vascular anomalies are the commonest developmental defect. Mulliken and Glowacki in 1982 divided vascular anomalies into two broad groups.[1] International society for study of vascular anomalies (ISSVA) accepted this classification in 1996 and later expanded it. Venous malformations (VM) are vascular anomalies involving the veins. Extremities are the commonest site for VM.[2] Intraosseous VM are relatively uncommon. Association of VM with cutaneous appendages have been rarely reported. The present study discusses the case of a 15-year-old boy who had VM with intra-osseous extension leading to limb hypertrophy and increased terminal hair and sparing of xerosis at the involved site.

Case report

A 15-year-old male child born of non-consanguineous marriage presented to the outpatient clinic of dermatology with pain while walking, bluish discoloration over skin for 6 months and history of progressive enlargement of the left lower limb since birth. There was no history of any ulceration, paraesthesia, trauma, drug intake, fever, shortness of breath or any other systemic complaints prior to increase in size of the lesion. There was history of excision of a growth present over the leg, which was reported as fibroepithelial polyp 6 weeks prior to patient's presentation. There was no history of similar lesion in the family. Physical examination revealed the presence of ill-defined bluish plaque of size 7 cms x 4 cms over anterior aspect of left leg (Fig.1a). It was soft and compressible with no palpable thrill or temperature change. Healed excision scar was present. Peripheral pulses were normal. There was no visible dilation of lymphatics, capillary or veins. There was generalized xerosis but area of malformation was spared (Fig. 1a). Terminal hairs were present over this area, while rest of the lower limbs was covered by vellus hair only (Fig. 1a). The difference between the limb girth 5 cms below tibial tuberosity was 2 cms (left more than right, Fig. 1b). There was no limb length discrepancy. X-ray of the left lower limb showed soft tissue hypertrophy as well as cortical tibial thickening (Fig. 2a). MRI imaging revealed tortuous venous channels in left anterolateral aspect of left leg with few vascular channels present in bone and muscle (Fig. 2b and 2c). Hence, a final diagnosis of VM with extension into muscle and bone was made. Sclerotherapy with ethanol was done. There was no post-sclerotherapy complication. Patient was asked to return for follow-up after 2 months.

Discussion

Vascular anomalies comprise of group of diseases showing abnormal development of angio-vascular or

Fig.1: a) Bluish plaque with terminal hair and hyperhidrosis on left leg in lying position. b) Left leg hypertrophy while standing

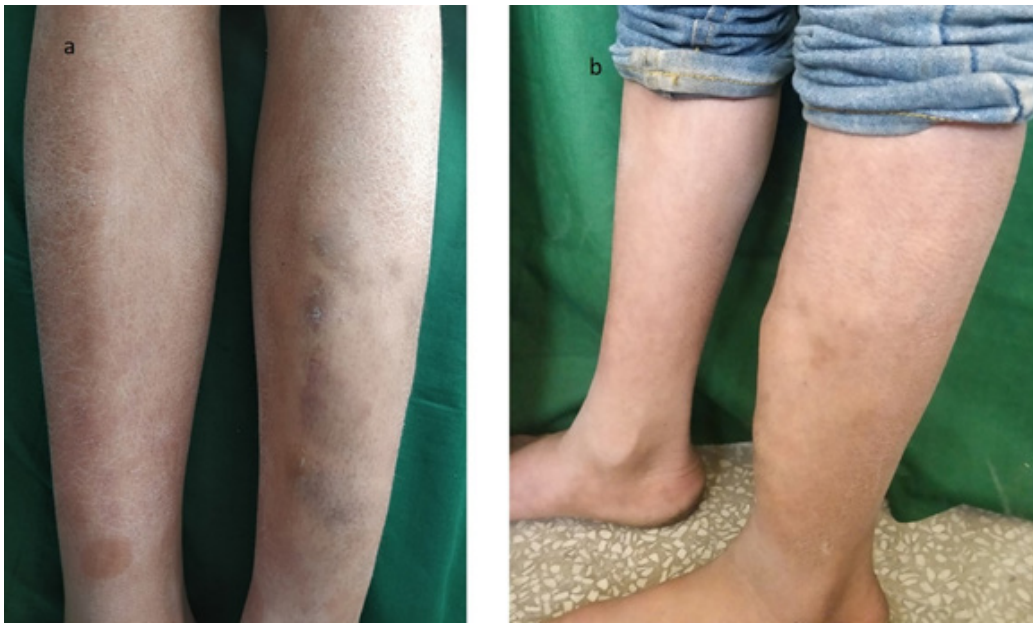
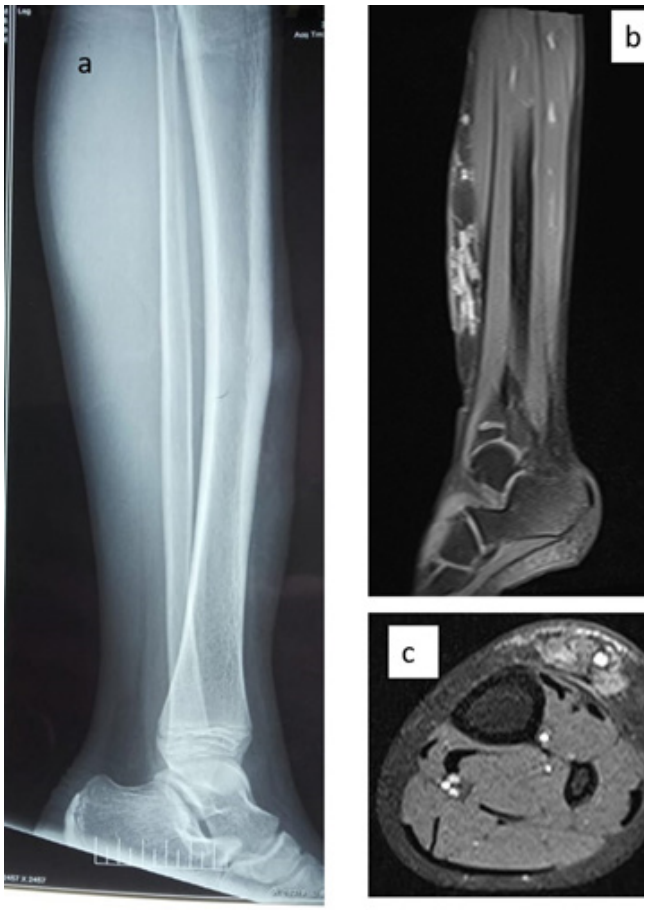


Fig. 2: a) Cortical tibial thickening visible on plane X-ray. b) Axial view on MRI showing venous malformations in skin, subcutaneous tissue and tibia c) coronal view showing venous malformations in skin and subcutaneous tissue



lympho-vascular structures. They are divided into two major types vascular tumors and congenital vascular malformations (CVM). In 2018, ISSVA revised classification of CVM and further divided them on the basis of vessel involved and hemodynamic status.[3] Vascular tumors arise during infancy due to endothelial proliferation. They grow for sometime, followed by later regression. While CVM show dysmorphogenesis. So they are present since birth, and progress with age without regression. CVM may be simple, combined with major vessels or associated with other anomalies. Simple CVM are defined by the blood vessel involved into capillary, lymphatic, venous, or arterio-venous malformations. Arteriovenous malformations are of slow flow type, while arteriovenous fistula is of fast flow type.

VM are one of the most common forms of congenital vascular malformations (CVM), second to lymphatic malformations only. Incidence of VM is 1:10,000.[4] The main locations of VM are the head and neck (40%), extremities (40%), and trunk (20%).[2] Clinically VM are soft, compressible, asymptomatic blue-to-purple subcutaneous non-pulsatile masses that grow in proportion to a child's growth and stabilize by adult life with no regression. They increase with Valsalva maneuver and gravity. Hormonal changes like puberty, pregnancy, hormone therapy and trauma or surgery act as triggering factors. The symptoms may depend on the site and size of VM.

VM are slow-flow malformations composed of simple dilated, dysplastic or inappropriately connected veins. Involvement may be of single or multiple veins. It may be diffuse or localized. It may be truncular (during late embryogenesis) or extratruncular (early embryogenesis, so retains mesodermal character and proliferative capacity).[5] Venous malformations may be superficial or deep. Superficial ones are localized to skin and subcutaneous tissue. Deep venous abnormalities can result in aneurysmal dilatation, aplasia, hypoplasia, duplications and venous incompetence. The deeper variant may invade any structure including abdominal viscera, muscle bone or joint. Intraorally VM can cause obstruction of airway, speech disturbance, distortion of teeth and bleeding or ulceration. Muscle involvement leads to pain in morning or muscle contraction. Joint involvement manifests as hemarthrosis. VM of bone can cause chronic pain and discomfort, limb hypertrophy or structural weakening leading to deformities and fractures. Our patient also had tibial involvement and hypertrophy. VM may get congested or thrombosed, leading to pain, hemorrhage or ulceration. Slow flow type of CVM increases the risk of phlebolith formation. Pulmonary embolism is not generally seen, as the thrombosed vessels get sequestrated from normal circulation.

Though most commonly sporadic, occasionally familial and multifocal VMs have been reported. Loss of function mutation on angiopoietin receptor gene TIE2/TEK located on chromosome 9p is thought to be the genetic basis of VM.[6] There is upregulation of several growth factors like tissue growth factor beta (TGF-beta) and basic fibroblast growth factor (beta-FGF).[7] The increase in growth factors could be responsible for terminal hair, fibroepithelial polyp and increased activity of localized sweat gland activity seen in our patient. Progesterone receptors present on vessels in VM result in sensitivity to hormonal changes.[8]

Plain radiological examination may show soft tissue and bone hypertrophy. Doppler helps in differentiating low and high flows. VM are often found as heterogenous hypoechoic areas and phlebolith as hyperechoic masses. Computerized tomography and MRI help in visualizing the extent of VM. Before sclerotherapy, the competence of deep venous system should be assessed. Various invasive studies, e.g., ascending, descending, and percutaneous phlebography, are required for treatment and excluding other vascular tumors.

Conservative management like limb elevation and compression stockings help in decreasing hydrostatic pressure. Intervention is required, if there is recurrent swelling, pain, deformity, interference with physical activity, hemorrhage thromboembolic complication, chronic venous hypertension or sepsis, or VM is aesthetically unacceptable.[9] Either sclerotherapy or excision is performed. Sclerotherapy is done by injecting sclerosant (ethanol and sotradecol) into sequestrated vessels. It is also effective for managing intra-osseous VM. Cutaneous necrosis, hemolysis, peripheral nerve injury, venous thromboembolism, and delayed muscle fibrosis and contractures are the various complications reported. Sclerotherapy is avoided if there is thrombosis or neurological involvement. Sclerotherapy is not recommended for distal lesions. Usually, sclerotherapy is avoided in very young child (<6 years of age), but early intervention is required in lower extremity vascular-bone syndrome.[10]

Competing interests
The authors declare that they have no competing interests.

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