Case Series

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Vascular Malformations in Paediatric Age Group: Learning through Mistakes

Paediatrics Section

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ABSTRACT

Vascular malformations were known in all paediatric age groups including neonates to adolescence. These were known since very long time. Many such cases were missed earlier due to lack of diagnostic imaging techniques. Proper diagnosis of such lesions is very challenging. So, proper identification and early treatment is of utmost importance. Any misdiagnosis can lead to inappropriate management and outcome. With recent advances in imaging techniques and better interventional facilities, the rate of diagnosis has improved but still the rate of misdiagnosis remains high. Though the newer treatment modalities are available, but their availability is restricted to tertiary care multifacility hospitals. Therefore, even after proper diagnosis, the management remains inadequate. The present series is about three different patients of vascular malformations, belonging to different paediatric age groups. The clinical presentations, diagnosis and management were varied, but diagnosis was possible due to high level of clinical suspicion and modern imaging techniques. The first patient initially seemed to have a post-traumatic swelling or juvenile idiopathic arthritis, but was later diagnosed as peripheral vascular malformation. The second patient was suspected to have haemangioma, but final diagnosis based on radiographical findings revealed low flow vascular malformation of face. The third patient was diagnosed with vascular malformation of head, but was clinically thought to be a sebaceous cyst.

Keywords: Management protocols, Misdiagnosis, Newer diagnostic imaging, Sebaceous cyst

INTRODUCTION

Vascular malformations occur due to error in morphogenesis of arterial, venous and lymphatic vascular channels in any combinations. They are present since birth and increases in size with growth of the child without showing any tendency to involute spontaneously [1]. About 60% of paediatric vascular malformations are found in head and neck region [2]. Epidemiological data of vascular malformations remains a confusing entity through decades. To have correct incidence and prevalence data of vascular malformations, one need to have more accurate and elaborate classification [3]. The natural history of arteriovenous malformation is uncertain, as few resolve on their own and few lead to severe complications. Paediatric vascular malformations have higher rate of rupture, and are majorly diagnosed after rupture [4]. The classification is complex and treatment remains controversial [5]. Peripheral vascular malformations can have intraarticular and intraosseous extensions that would pose greater therapeutic challenges [6]. Several classification systems have been proposed. But few are clinically significant. The Milliken and Glowacki classification (1982) differentiated vascular anomalies into haemangiomas and vascular malformations, based on endothelial characteristics. The classification by the International Society for the Study of Vascular Anomalies (ISSVA) is widely accepted [1]. Vascular anomalies can be high flow or low flow [7].

A variety of imaging techniques are available. Most commonly used are doppler Ultrasonography (USG) and Magnetic Resonance Imaging (MRI). The doppler ultrasound should be the initial imaging modality for recognising vascular tumours from vascular malformations. Computed Tomography (CT) and MRI are the best imaging modality for evaluation of extent of the lesion prior to treatment and also, for follow-up [6]. This helps to evaluate the nature, extent and complexity of the malformations and this helps to plan appropriate treatment [6]. The treatment requires multidisciplinary approach. The first line therapy should include image-guided (check globally) percutaneous and interventional treatment [Table/Fig-1].

Vascular anomalies	Treatment
Capillary	None
Haemangioma	None or propranolol
Slow flow venous and lymphatic malformations	Percutaneous sclerotherapy
Fast flow Arteriovenous malformation (AVM) and Arteriovenous fistula (AVF)	Transarterial embolisation

[Table/Fig-1]: Treatments of vascular malformations according to flow dynamics [6].

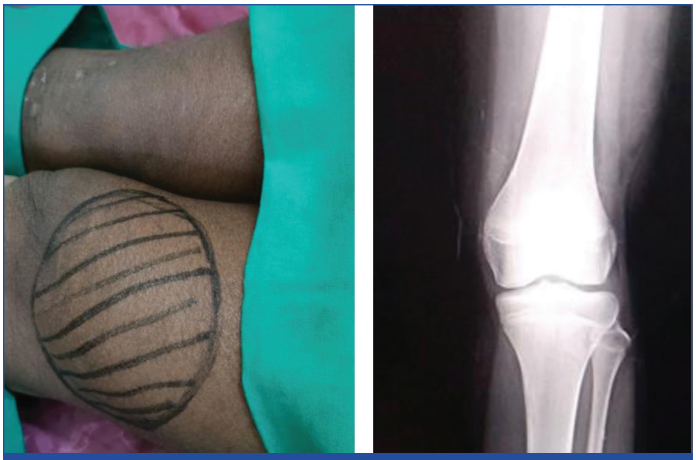
After procedure, immediate clinical monitoring is recommended. Imaging studies such as colour Doppler USG, contrast CT and MRI are used for assessing immediate results and long-term management [6]. The present case series is about three different patients of vascular malformations, belonging to different paediatric age groups.

CASE SERIES

Case 1

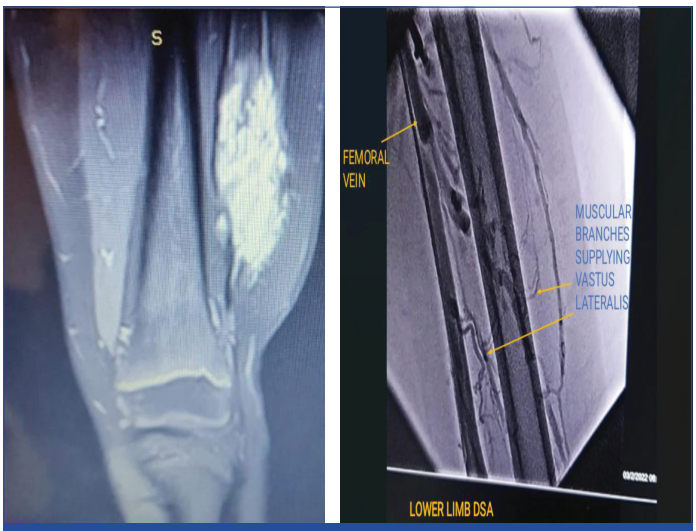
A 13-year-old girl was admitted with complaints of gradually progressive swelling over the left thigh just above the left knee joint along with mild pain, since last six months. The swelling was present in the anterolateral side of lower aspect of thigh above the knee joint. There was a history of blunt trauma over the affected region due to fall from cycle, about six years back. Immediately there was pain which was treated with some analgesics. Gradually swelling appeared with occasional pain, especially during exercise. No joint swelling and deformity were noted. None of the other joints in the body were involved. Joint mobility was not restricted. Range of movement of left knee joint was within normal limit. There was no history of spontaneous bleeding from any site.

On physical examination, vital signs were stable. The swelling was 10×10 cm, soft, having smooth surface, mildly tender with restricted mobility [Table/Fig-2]. There were no fluctuations, pulsatility or bruit over the swelling. There were no systemic abnormalities like fever, rash, lymphadenopathy, etc. The musculoskeletal and neurological examinations were also normal. X-ray left knee joint did not show any fracture [Table/Fig-3]. USG of left knee joint did not show any haematoma or abscess. It suggested some altered echogenicity in



[Table/Fig-2]: Swelling of the knee. **[Table/Fig-3]:** X-ray of left knee joint showing no fracture or bony deformity. (Images from left to right)

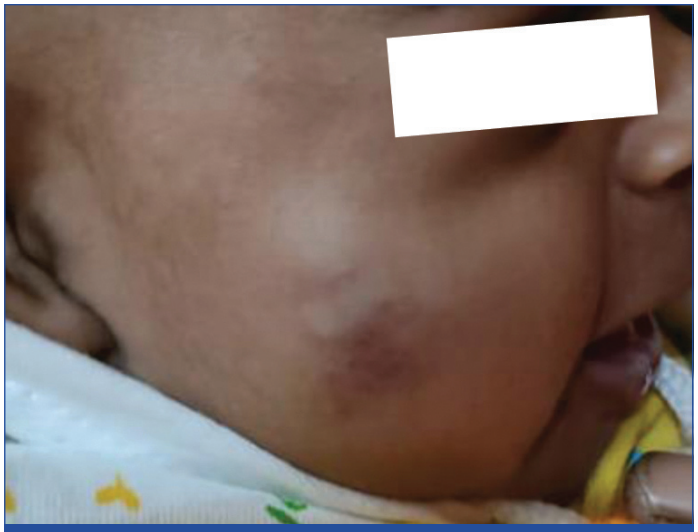
the vastus lateralis muscle. The inflammatory markers like C-Reactive Protein (CRP), Antinuclear Antibodies (ANA), Rheumatoid Arthritis (RA) factor were all within normal limit. MRI left thigh with contrast revealed altered high intensity area noted in the caudal part of vastus lateralis muscle, showing multiple tortuous T2 hyperintense channels. Multiple hypointense flow voids were also noted within the lesion. Lesion showed communication with the superficial subcutaneous vascular channels. Contrast study showed enhancement with interspersed flow voids. Features were suggestive of arteriovenous malformations [Table/Fig-4]. Digital Subtraction Angiography (DSA) was performed which showed low flow vascular lesion [Table/Fig-5]. The child was referred for vascular surgery to a higher centre.



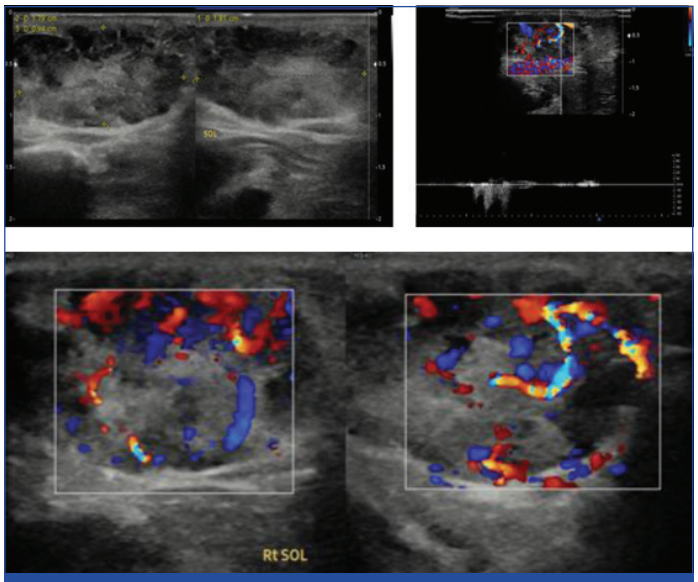
[Table/Fig-4]: MRI with contrast showing altered signal intensity area, hyperintense, in the caudal part of vastus lateralis muscle, internal multiple tortuous T2 hyperintense vascular channels.
[Table/Fig-5]: Lower limb DSA showing muscular branches, arising from the femoral vein, supplying the vastus lateralis muscle-suggestive of low flow vascular lesion. (Images from left to right)

Case 2

A two-month-old girl presented with swelling over the right cheek for six weeks [Table/Fig-6].The swelling was not apparent at birth. It was an isolated swelling which gradually increased in size. The birth history and developmental history was uneventful. No similar birth lesion was present in the family members. The child was asymptomatic. The size of the swelling was about 1.5×3 cm. The margins were not well-defined. It was soft and cystic in nature, but was non compressible. There was no pulsation over the swelling. Overlying skin was healthy, it was not associated with fever, any tenderness or any trauma. The provisional diagnosis was haemangioma. USG with Doppler study of right cheek showed a low flow arteriovenous malformation [Table/Fig-7]. The child was advised surgery and referred to Cardiothoracic and Vascular Surgery (CTVS) Department for further management.



[Table/Fig-6]: Cheek swelling.



[Table/Fig-7]: USG with Doppler study of right cheek showing a low flow arteriovenous malformation.

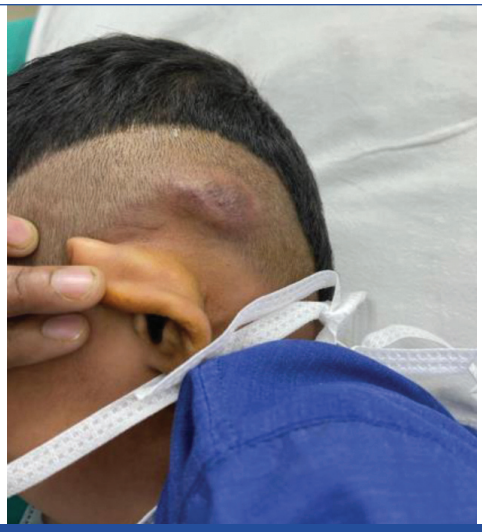
Case 3

A five-year-old boy presented with complaint of swelling over the left-side of the scalp above the ear, associated with mild itching. The swelling was slowly progressive since birth, and was without any tenderness [Table/Fig-8]. On examination, the size of the swelling was 3×5 cm. It was mildly compressible, but non pulsatile. The child did not have any such similar lesion in any other part of body. The child did not had any symptoms suggestive of any intracranial lesion (no convulsion, hemiparesis). The child also did not have any facial dysmorphism or stridor or any bleeding manifestations. It was not associated with any systemic features like fever or any other swelling in the body. It was suspected to be a lipoma/dermoid/sebaceous cyst/simple cyst.

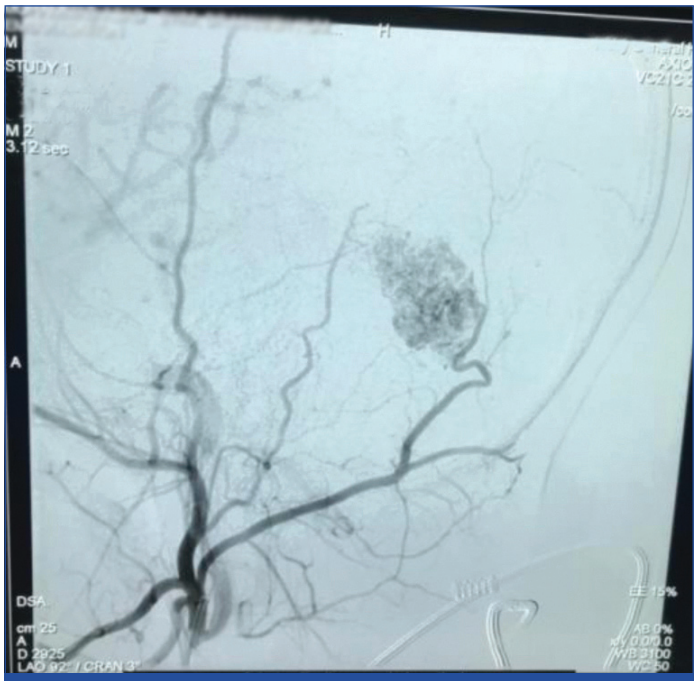
DSA was done which revealed a low flow vascular malformation [Table/Fig-9]. It was treated surgically by endovascular embolisation, followed by excision done under general anaesthesia [Table/Fig-10]. The child recovered uneventfully, after surgery and was discharged with conservative management.

DISCUSSION

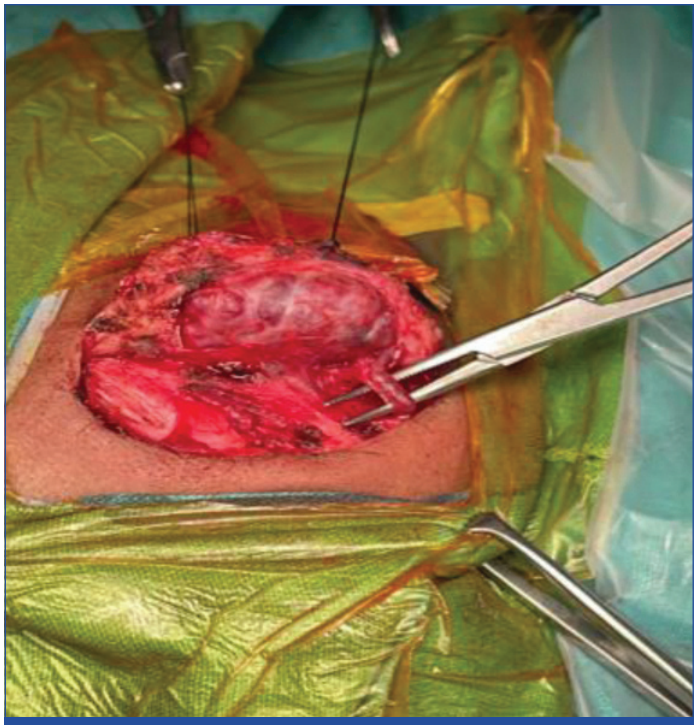
Vascular anomalies broadly fall into two categories: a) vascular tumour, (b) vascular malformations. Vascular tumours include Infantile Haemangioma (IH), Congenital Haemangioma (CH), Kaposiform Haemangioendothelioma (KH). IH is most common among them. The management of vascular tumours mainly includes conservative approaches, oral medications and surgical intervention depending on their type, location and associated complications. The main



[Table/Fig-8]: Scalp swelling.



[Table/Fig-9]: DSA showing the nidus and feeding vessels.



[Table/Fig-10]: Perioperative image.

treatment options for vascular malformations are endovascular or surgical obliteration [2,8]. If a vascular lesion is wrongly diagnosed, then can lead to wrong management and thus lead to complications like massive bleeding.

The above three cases of vascular malformations were seen in children of different age groups ranging from infant to adolescent. Though they can be present since birth, these gradually become apparent as the size increases with age [2]. In the present, two out of three patients had involvement of the head and neck region [1]. As per literature, majority of vascular malformations are detected only after rupture causing bleeding symptoms [2]. Fortunately, in the present series, all of them were detected early and properly identified with the help of appropriate imaging technique. An early diagnosis prevented them from serious complications like bleeding. The symptoms and imaging appearances can be complicated which can further complicate the diagnosis. Thus, from tthe present series, it can be seen that a vascular malformation can be wrongly diagnosed as vascular tumours (IH, CH, KH), complicated arthritis Juvenile Idiopathic Arthritis (JIA), traumatic or infective lesions (abscess, cyst, haematoma etc.,) or muscular or bony tumours (sarcomas etc.,). Many such misdiagnosed cases, even received management of JIA with steroids, methotrexate.

The complications of vascular malformations depend on the location like overgrowth of limbs, disfigurement, especially of facial areas, pain, respiratory difficulty, dysphagia, if lesions are inside organ systems, bleeding (internal or external) [9]. Complications may also arise after surgical treatment. The prognosis of vascular malformations also depends on its type, location, time of presentations. Many such lesion have very good prognosis, if treated, at earliest. However, malformations present near vital organs like brain have high propensity to bleed, cause ischaemic brain injury, seizures etc., with a high mortality rates of about 10% [10]. Complications of cerebral AVM include strokes, intracerebral haemorrhages (9% of subarachnoid haemorrhages are due to AVM), unprovoked seizure, headaches and even brain abscess [11]. Even pulmonary AVM leads to CNS complications (stroke or brain abscess). A total of 70% patients of pulmonary AVM had neurological complications. Even death can occur as a result of such complications. These can also lead to severe hypoxia [12]. Newer developments in diagnostic modalities and high-end interventional procedures, have made the management easy, thus reducing potentially serious complications [6].

CONCLUSION(S)

In all the above cases, the initial impression appeared to be different from the actual diagnosis. A multicentric approach was made to establish the correct diagnosis and define accordingly the appropriate treatment and follow-up. Vascular malformations may be misdiagnosed, unless modern imaging techniques are used. Vascular malformations of knee can mimic JIA. Similarity, of the two in clinical presentation and imaging finding can be confusing and may lead to misdiagnosis. Here, in the present case series, the initial diagnosis was very different from the actual diagnosis made later. Ultimately, with the help of modern imaging techniques we were able to diagnose and treat them appropriately. Peripheral vascular malformations are rare but they form an important spectrum of abnormalities. Identification of these lesions can be challenging. Clinical suspicion and good radiological and interventional back-up are needed for accurate diagnosis and management.

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