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Vascular Malformations: Journey through The Maze- A Case Series

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Abstract

Vascular malformations belong to the spectrum of orphan diseases involving all segments of the vascular tree: arteries, veins and capillaries, and similarly the lymphatic vasculature. These malformations are a common cause of soft tissue masses and often referred for ultrasonographic evaluation. The ultrasonographic and doppler imaging findings can help to categorize these malformations into slow-flow or fast-flow lesions which forms the basis of further workup and management. Further evaluation with magnetic resonance imaging (MRI) or computed tomography (CT) helps in delineation of lesion extent whereas MR or CT angiography helps in mapping of vascular supply of the lesion. We present an overview of the diagnostic workup of these vascular malformations, describe their imaging characteristics and verify the role of ultrasound and doppler examination in the initial diagnosis of these lesions.

Keywords: Vascular malformations, arteriovenous malformations (AVM), capillary malformations, lymphatic malformations (LM), venous malformations (VM), ultrasonography (US), doppler.

Introduction

Vascular malformations are vascular spaces lined by flat epithelium with an estimated prevalence of 4.5% in the population[1,2]. These malformations present as a cutaneous lesion or a deeper palpable soft tissue mass and can involve any part of the body, most commonly the head and neck followed by the extremities. These lesions can be diffuse or focal, simple or combined based on the subtype of the vessel involved. Ultrasound (US) and doppler has been the first investigating modality utilized in the diagnosis of patients with vascular malformations due to its low cost, non-ionizing technology, superficial spatial resolution and its ability to allow the assessment of its fluid, flow characteristics and vascularity[3,4,5].

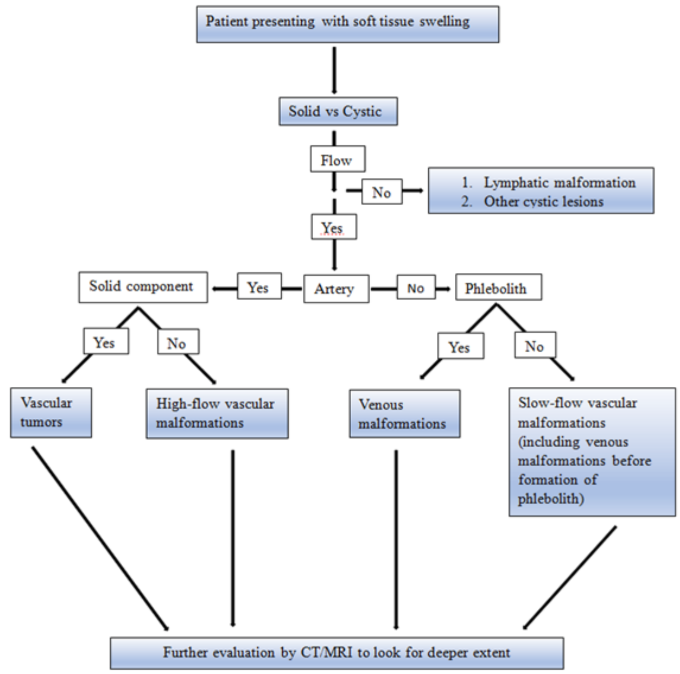
Imaging may be employed to delineate the deeper extent of the lesion, to differentiate vascular malformations from vascular tumors and for proper treatment planning by depicting the anatomy of inflow feeder, nidus and draining vessel of the malformation [6,7].

Discussion

In 1996, the International Society for the Study of Vascular Anomalies (ISSVA) released a classification system for vascular anomalies, which was updated in 2014 and revised in 2018 [8,9,10]. According to this classification, vascular anomalies are classified into vascular tumors and vascular malformations. Based on the flow dynamics, the vascular malformations are further divided into "high-flow" or "slow-flow" groups. High-flow lesions contain an arterial component such as arterial malformation, arterio-venous malformation, aneurysm/pseudoaneurysm and arteriovenous fistula. The slow-flow lesions are all other lesions that do not contain an arterial component including capillary malformation, venous malformation and lymphatic malformation [10,11].

Imaging is indicated in vascular anomalies to confirm the clinical diagnosis, characterize the lesion as high or low flow, know the complete extent of deep seated, infiltrative or wide-spread lesion and know the vascular supply of the lesion. USG is first line imaging modality along with colour and spectral doppler and can accurately diagnose most of the cases when interpreted along with the clinical features[10,12,13]. Contrast-enhanced ultrasound combines all the advantages of the USG with the evaluation of the microvasculature of the lesion[14]. MRI is frequently the next step in the assessment to evaluate the deeper extent of the lesion and its relation with the vital structures. CT also plays an important role in selected cases where urgent evaluation is to be done or when the MRI is not available or the child cannot be sedated for MRI [15].

Diagnostic Approach in a suspected case of vascular Malformations [4,16]



When an arterial flow is detected within the lesion, the following evaluations should be done[10,17,18]

1. Vessel density: area of greatest vascularity within the lesion is identified. Less than 2 vessels/cm sq. is considered as low density, 2-4 vessels/cm sq. is considered as moderate density and 5 or more vessels/cm sq. is considered as high density. High vessel density is a characteristic of high-flow vascular malformations[17].
2. Peak systolic velocity (PSV): Peak velocity of arterial flow of the lesion is measured. High values of PSV are seen in high-flow vascular malformations.
3. Resistive Index (RI): The high-flow malformations show low resistive index. In some cases of slow flow malformations and lymphatic malformations, arterial flow is demonstrated within the septae/walls of the constituent cyst which typically shows high resistive index.

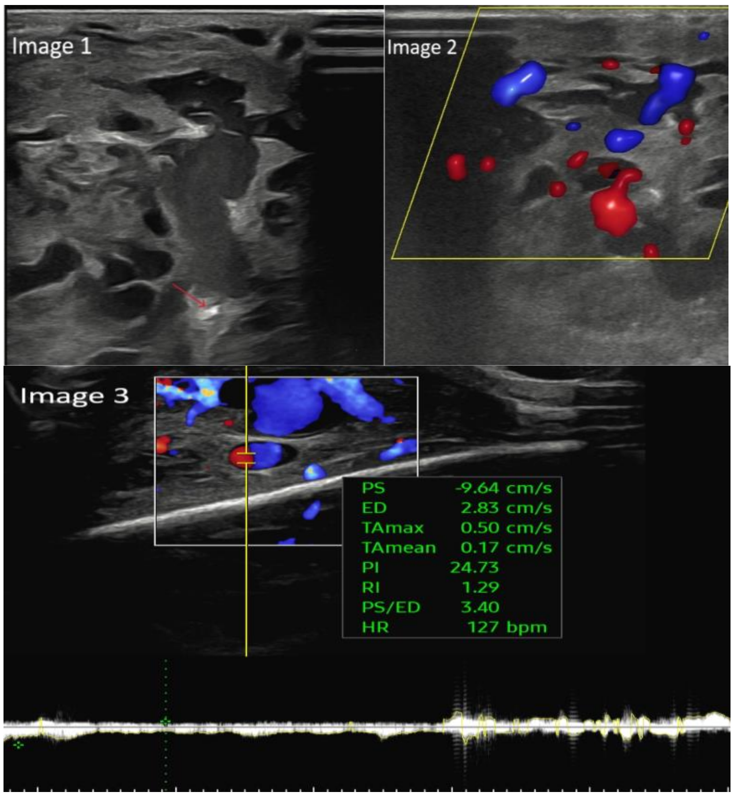
4. Arterialization of venous flow: High venous peak velocities seen in cases of arterio-venous malformations or arterio-venous fistula.

A. SLOW-FLOW MALFORMATIONS

1. Venous Malformations (VM)

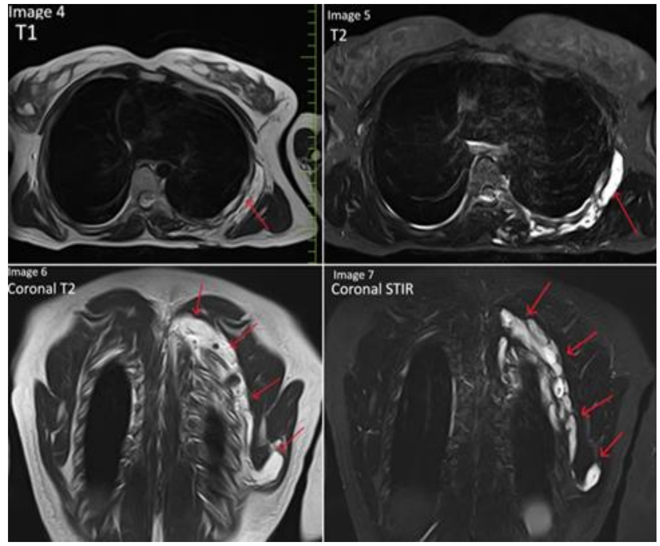
Case 1

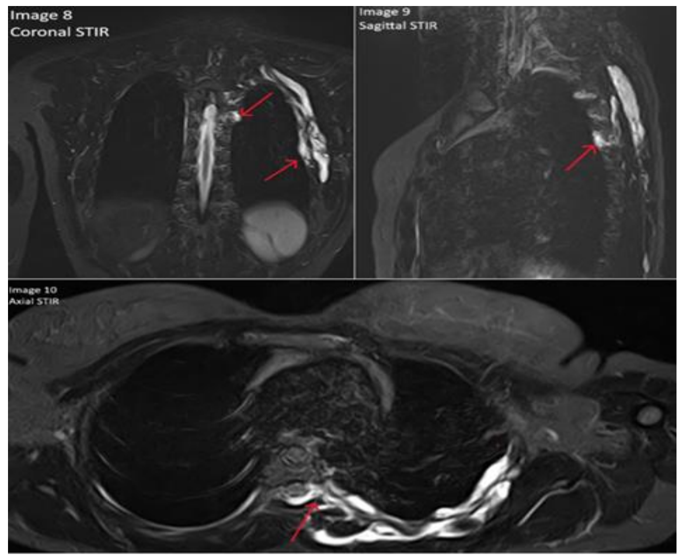
A 20-year-old female patient presented with complains of swelling over back on the left side subjected to USG (Figure-1).



USG demonstrated multiple dilated tubular channels with multiple echoes and few hyperechoic phleboliths within (image 1) and doppler demonstrated color flow (image 2) with predominantly low-velocity venous waveform (image 3).

Patient was followed up with MRI (Figure 2 and 3).

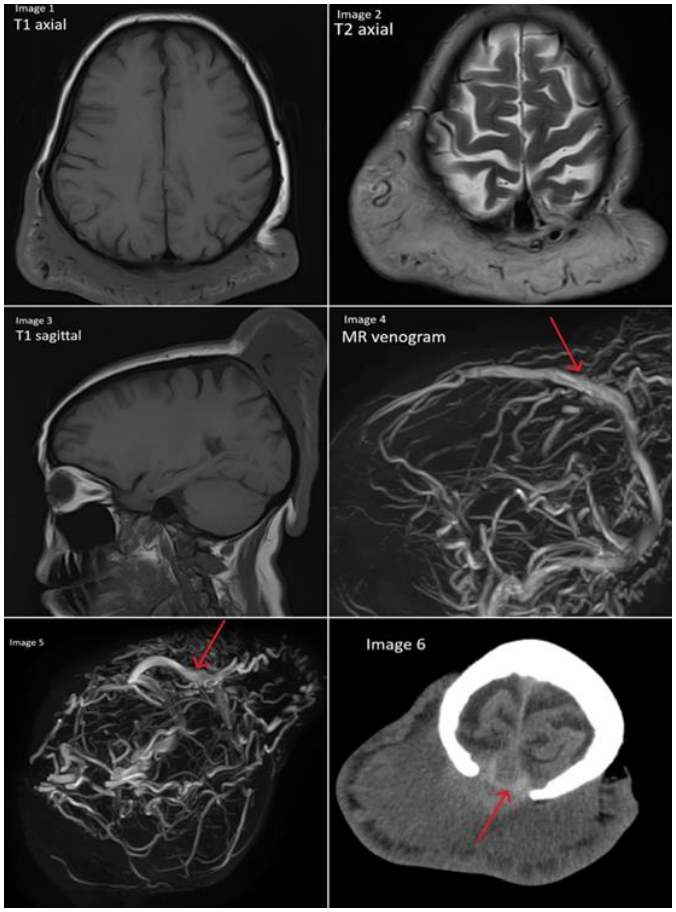




MRI demonstrated a large relatively well-defined area appearing isointense on T1W (image 4) and heterogeneously hyperintense on T2W/STIR images (images 5, 6 and 7) involving the superficial and deep inter and intramuscular compartments of left postero-lateral chest wall involving latissimus dorsi, serratus anterior, internal intercostal muscles and left paraspinal muscles. They were seen extending cranio-caudally from D1-D10 vertebral levels on the left side and medially into the intercostal rib spaces with intra-thoracic extension from D5-D8 vertebral levels (image 8). There was suspicious extension of this lesion noted into the spinal canal on left side at D4 vertebral level (images 9 and 10). A diagnosis of venous malformation was made.

Case 2

A 30-year-old female patient presented with swelling at scalp since birth increased in size over 2-3yrs (Figure 4).



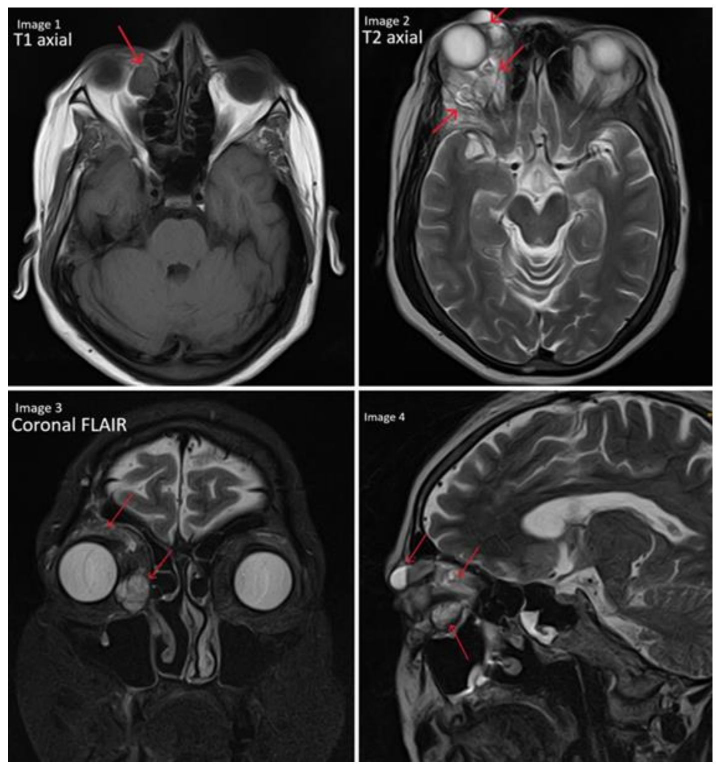
There was a large well-defined lesion noted involving the subgaleal soft tissues of bilateral parieto-occipital regions with multiple serpiginous T1W and T2W hypointense flow voids within (images 1, 2 and 3). There was a bony defect noted involving the calvarium of high parietal bones (image 6) with an abnormal communication between the extracranial flow voids and the mid third of superior sagittal sinus (images 4 and 5). However, no extension into the brain parenchyma was seen.

A diagnosis of sinus pericranii was made.

Venous malformations show prevalence of 1% in general population [19]. These malformations can be found anywhere in the body more prevalent in head, neck and extremities. The severity of symptoms depends on the size, extent and the involvement of adjacent structures. US shows multiple venous spaces or ectatic/dysplastic veins with slow-flow within it. Doppler helps to differentiate between venous and lymphatic malformations [20]. Phleboliths are seen in cases of venous malformations as echogenic focus with posterior acoustic shadowing. CT venography can be used to identify underlying obstructed veins, anatomical variation and extent of venous thrombosis [21]. MRI can be used in evaluation of large or infiltrative lesions [22].

Case 3

A 32-year-old female patient presented with swelling over right eye with diminution of vision subjected to MRI brain + Orbits (Figure 5).

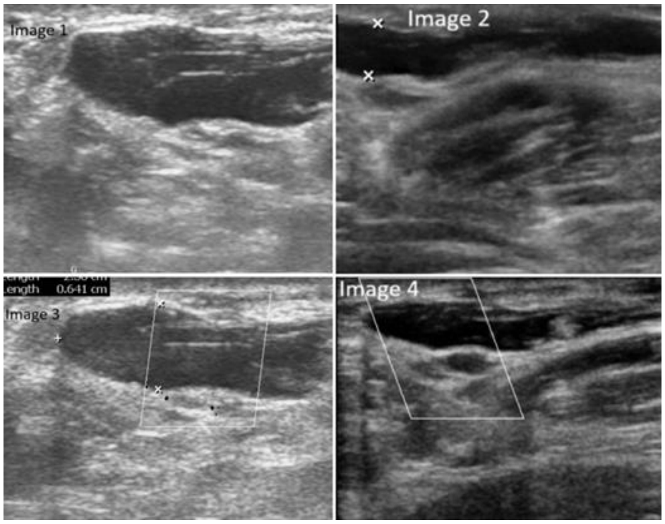


Multiple well-defined lobulated T1W isointense (image 1) and T2/FLAIR inhomogenously hyperintense lesions (images 2, 3 and 4) were noted involving the intra and extra conal compartments of right orbit causing proptosis and lateral displacement of right globe. A diagnosis of slow-flow vascular malformation was made.

2. Lymphatic Malformations (LM)

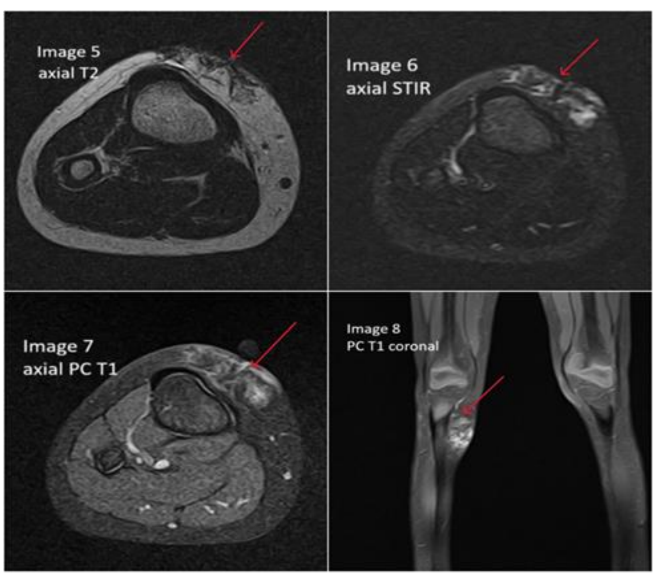
Case 4

A 6-year-old female patient presented with painless swellingon medial aspect of right leg increasing in size gradually (Figure 6).



USG demonstrated dilated anechoic cystic and tubular spaces (images 1 and 2) with no color flow on doppler interrogation (images 3 and 4).

Patient was followed up with MRI (Figure 7)



MRI showed an altered signal intensity lesion involving the subcutaneous plane of antero-medial aspect of right leg (image 5) appearing hyperintense on STIR (image 6) showing vivid post-contrast enhancement (images 7 and 8). No deeper extent with normal underlying bony cortices were noted.

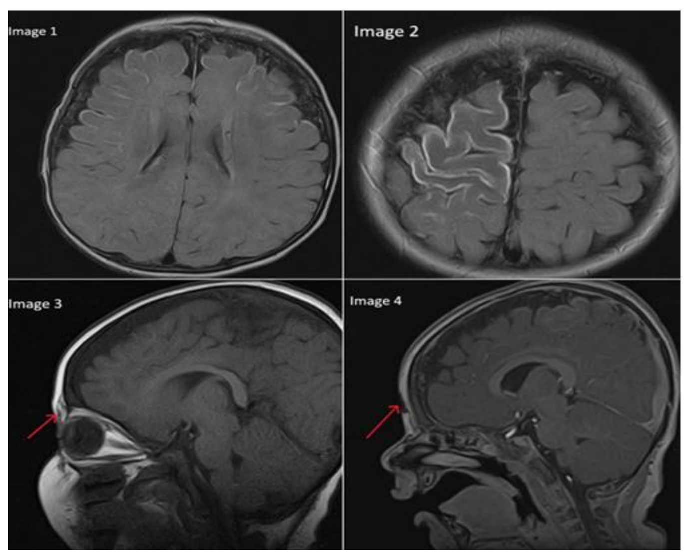
A diagnosis of lymphatic malformation was made.

Lymphatic malformations account for 2.8/100,000 population and are composed of vessels or large chambers lined by endothelial cells[23]. Most of the cases involve head and neck. LM are of two types: Macrocystic type showing multiple cystic spaces of 2cm or more appearing as soft tissue masses [24]. Microcystic type shows lesions infiltrating tissues, can present with limb swelling and can be complicated by secondary infections [25]. US shows multiple anechoic cystic spaces with absent flow[26]. MRI is the investigation of choice in diagnosis of LM appearing hypo to iso-intense on T1W images and hyperintense on T2W images with absence of flow voids.

3. Capillary Malformations (CM)

Case 5

A 1-year-old male patient presented with history of left sided weakness, seizures and port wine stain on forehead subjected to MRI brain (Figure 8).



MRI brain demonstrated serpentine leptomeningeal enhancement along the bilateral frontal and right high parietal lobes – pial angiomatosis (images 1 and 2). Serpentine flow voids were noted at the site of port-wine stain on forehead – capillary malformation (images 3 and 4).

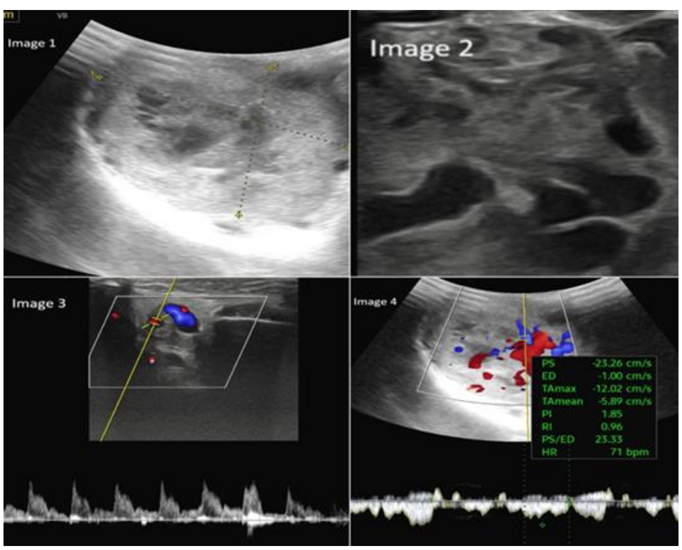
A diagnosis of Sturge-Weber syndrome was made.

Capillary malformations are found in 0.1-2% of the population [6]. They are the anomalies of capillary network involving the skin and mucous membranes and can be isolated or associated with syndromes such as Sturge-Weber syndrome or Klippel-Trenaunay syndrome [32].CMs present clinically as cutaneous lesions and imaging has no significant role in the diagnosis. Also, they regularly do not require endovascular therapies and no corresponding imaging treatment plan.

B. HIGH-FLOW MALFORMATIONS

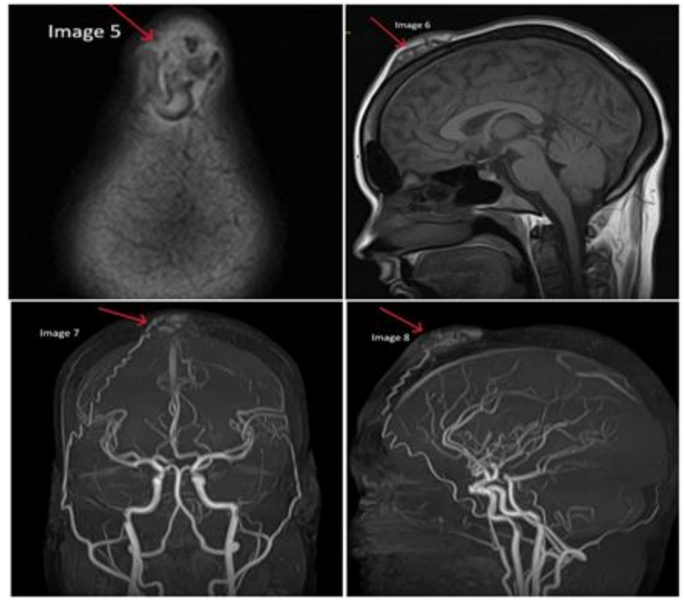
1. Arterio-venous Malformations

Case 6: A 27-year-old male patient presented with swelling on the scalp on high frontal region subjected to USG (Figure 9).



USG demonstrated a well-defined heterogeneously hypoechoic lesion (image 1) with multiple cystic areas and serpiginous channels within (image 2). Doppler demonstrated mixed arterial and venous flow (images 3 and 4).

Patient followed up with MRI (Figure 10).

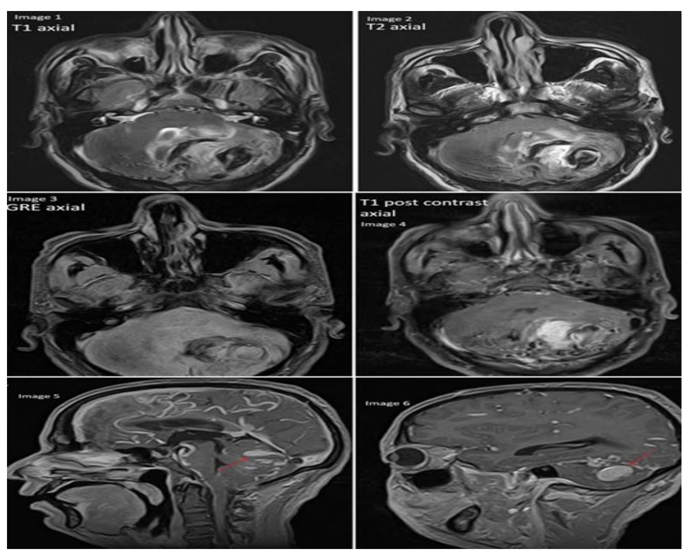


MRI brain demonstrated a well-defined lesion involving the midline high frontal region showing multiple serpiginous flow voids within it (images 5 and 6). No underlying bony defect was noted with no intracranial extension. The MR Angiogram demonstrated feeding artery arising from bilateral external carotid arteries (images 7 and 8).

A diagnosis of arterio-venous malformation was made.

Case 7

A 55-year-old male patient presented with seizures subjected to MRI brain (Figure 11).



There were numerous T1 and T2 hypointense serpiginous flow voids involving the left cerebellar hemisphere (images 1 and 2) indicating a nidus. There was a T1 and T2 hyperintense area adjacent to the nidus (images 1 and 2) showing blooming on GRE (image 3) indicating late subacute intraparenchymal hemorrhage. On post contrast study, the nidus showed vivid enhancement (image 4). MR venogram demonstrated with multiple engorged venous channels involving the entire brain parenchyma and the nidus (images 5 and 6).

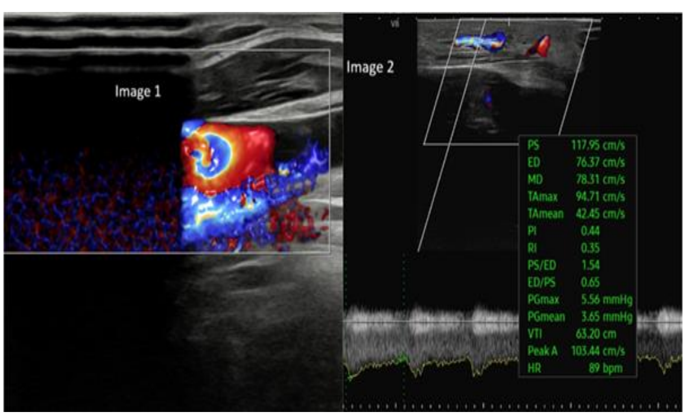
A diagnosis of cerebellar arterio-venous malformation was made.

AVM is composed of dysplastic arteries communicating with veins with absence of capillary bed in between. They contain a nidus between arterial and venous bed. On US and doppler examination, AVM presents as pulsatile lesion showing vessels with high systolic and diastolic flow and prominent arterio-venous shunting [12]. On MRI, it presents as network of arteries and veins connected by a shunt, presence of flow voids with gradient images demonstrating rapid flow [27]. Diagnostic angiography shows dilated arteries with early opacification of dilated veins [28].

2. Arterio-venous Fistula

Case 8

A 55-year-old male patient, a known case of chronic kidney disease on hemodialysis with brachio-cephalic fistula on the left forearm (Figure 12)



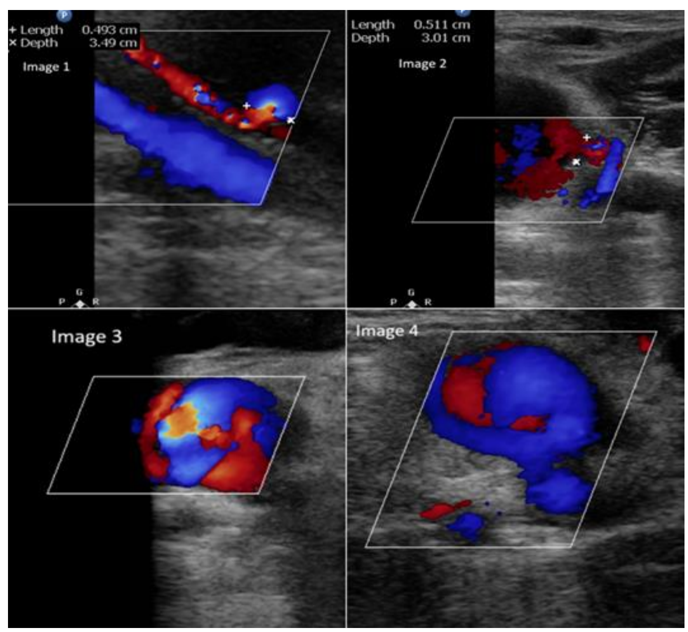
Doppler demonstrated aliasing of color flow at the fistula site (image 1) with arterialization of venous flow showing high peak velocity in the cephalic vein distal to the fistula site (image 2).

They are usually acquired due to trauma or iatrogenic and lack the nidus present in AVM. Like other vascular malformations, US is the first investigation in evaluation of AVFs showing high frequency, low resistance continuous flow with elevated diastolic velocities. The fistula site shows characteristic aliasing due to mixing of arterial and venous blood. MRI shows serpiginous signal voids without a focal mass with presence of arterial and venous component[29]. Angiography shows enlarged vessels with early venous drainage.

3. Aneurysm / Pseudo-aneurysm

Case 9

A 56-year-old male with history of arterial puncture of right femoral artery subjected to right lower limb arterial doppler (Figure-13)



USG demonstrated a saccular outpouching arising from superficial femoral artery (image 1) connected to it by a narrow neck measuring 5mm in diameter (image 2).

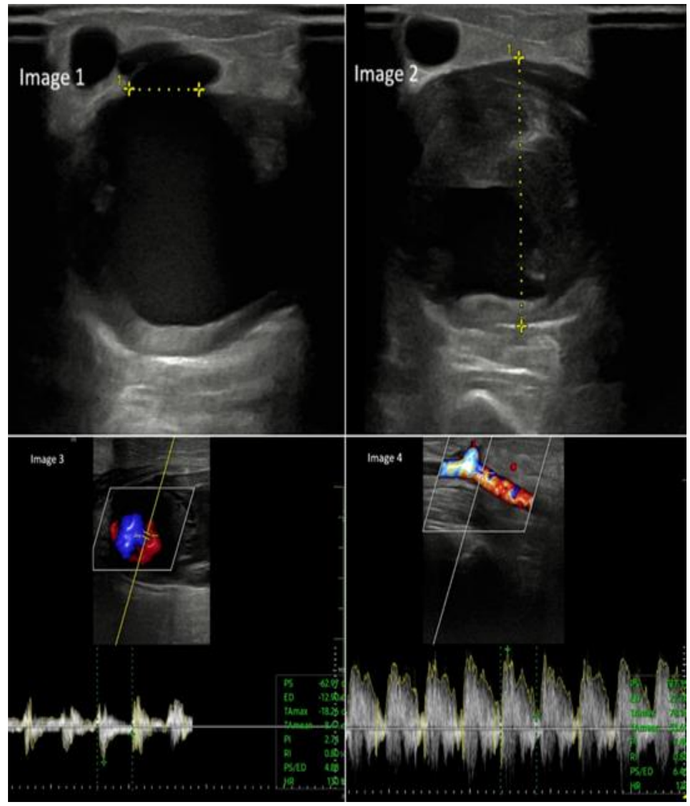
Color doppler demonstrated "ying-yang" sign within the lesion (images 3 and 4).

Patient was followed-up with lower limb CT angiogram (Figure-14)

CT angiogram confirmed the presence of pseudoaneurysm arising from right femoral artery showing contrast opacification.

Case 10

A 39-year-old female patient, a known case of CKD on hemodialysis with brachio-cephalic fistula on left arm presented with diffuse swelling of entire left arm subjected to left upper limb arterio-venous doppler (Figure 15).

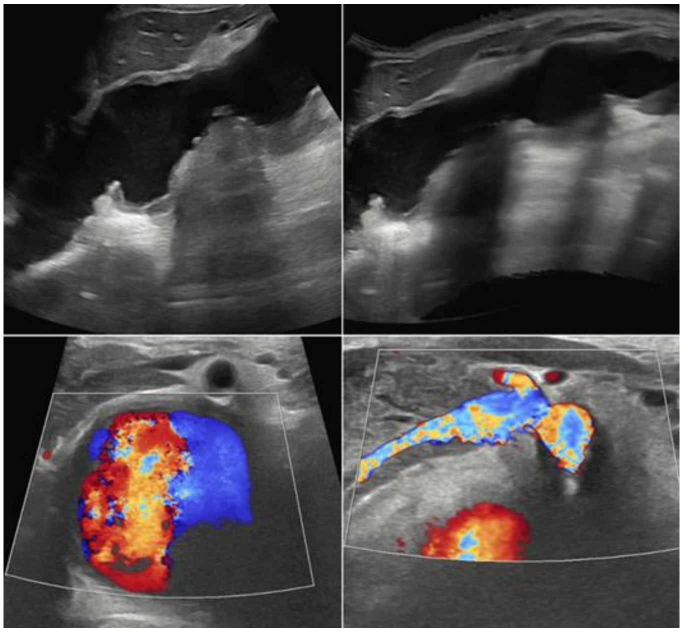


USG demonstrated a saccular outpouching arising from cephalic vein just proximal to the site of fistula site with a neck measuring 9mm (image 1) showing a hyperechoic partial lumen occluding thrombus within in the distal 2/3rd part (image 2). The patent proximal 1/3rd portion shows swirling echoes within and color doppler demonstrated positive "ying-yang" sign (image 3) suggestive of partially thrombosed pseudoaneurysm arising from cephalic vein.

Fistula is noted between brachial artery and cephalic vein with presence of aliasing and arterialization of cephalic venous flow demonstrating high peak velocity of 137 cm/sec (image 4).

Case 11

A 50-year-old female patient presented with history of hypertension subjected to USG abdomen (Figure 16).



USG abdomen demonstrated multiple aneurysmal dilatations involving the abdominal aorta with turbulent flow within.

These malformations result from the abnormal focal dilatation or outpouching of the artery. The true aneurysm is bounded by all 3 layers of vessel wall (intima, media and adventitia) whereas the pseudoaneurysm is bounded only by the tunica adventitia, the outermost layer of the arterial wall. Pseudoaneurysm occurs when there is breach in the arterial wall by trauma or infection in which blood leaks through the inner wall but contained by the adventitia and surrounding soft tissues[30]. US demonstrated dilated anechoic lesion arising from a vessel wall communicating with it by a narrow neck. Pseudoaneurysm typically shows a characteristic ying-yang sign on color doppler due to turbulent and backward flow and "to-and-fro" pattern on pulsed doppler. CT angiogram can help define the relation to the surrounding structures. Angiography is the gold standard in diagnosis of cardiac and visceral pseudoaneurysms with a sensitivity of 85%[31].

The management of vascular malformations require a multi-disciplinary approach and interventional radiology techniques have an important role to play in the management of these lesions[33]. The mainstay of treatment for AVM is embolization focused on obliteration of the nidus which can be done percutaneously, via an endovascular approach or surgically[34]. The VMs and LMs are managed by percutaneous sclerotherapy or surgery[35].

Conclusion

Imaging plays an important role in diagnosis of vascular malformations in conjunction with clinical history and detailed clinical examination. US is the initial imaging modality and can accurately diagnose these lesions. Color and spectral doppler help in categorizing these anomalies into high-flow and slow-flow malformations which forms the basis of further management. It also helps in guiding initial management decisions and monitoring the response to interventional, medical or surgical treatment. CT/MRI plays an important role in assessment of the deeper extent of these lesions and their relation to the adjacent structures. Angiographic evaluation also helps in assessment of the high flow malformations, evaluation of the feeder vessel guiding the further intervention by embolization.

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