Congenital vascular malformations – a quick recap

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Received: 30 April 2020

Accepted: 5 July 2020

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DOI 10.5001/omj.2021.46

Abstract

We present a glimpse of vascular malformations seen at Sultan Qaboos University Hospital from July 2014 to December 2019.The cases are sporadic in nature. Molecular genetic studies can be conducted in patients with a family history of vascular malformations. Cultural practices relating to dress code may lead to delay in presentation. A diagnostic /management algorithm about vascular malformations would help healthcare professionals diagnose, counsel, refer appropriately and a national registry would further enhance research and patient support groups.

Keywords: Vascular, Malformation, AVM, Venous, Park-Weber, Klippel –Trennunay , Pediatric, Oman

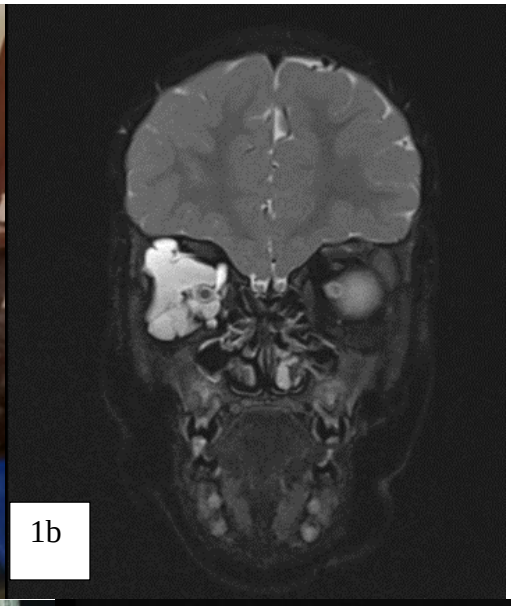
Introduction:

Vascular anomalies are soft tissue lesions that affect up to 10% of the newborns and were first classified by Mulliken and Glowacki in 1982 into congenital vascular malformation (CVM) and vascular tumors [1,2,3] Despite the advancements in this field CVMs remain a diagnostic and therapeutic challenge to the treating physicians, therefore requiring a multidisciplinary approach. [4]

Case 1

Eight year old girl, born to consanguineous parents presented with two years history of on and off proptosis, redness and pain of the right eye [Figure 1a] . These symptoms were provoked by an upper respiratory tract infection – three episodes in two years. An MRI done at initial presentation showed a lympho-venous right eye CVM [Figure 1b]. The patient was managed with intravenous steroids and sclerotherapy, followed by eye drops [Brinzolamide+Timolol]. Follow up MRI has shown regression and she has been asymptomatic for three years.

1a



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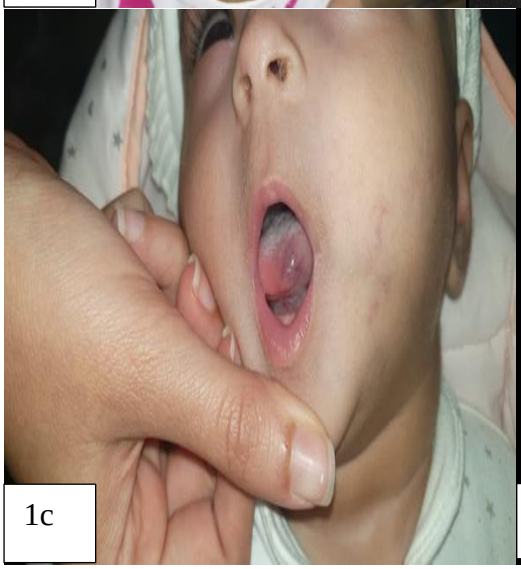

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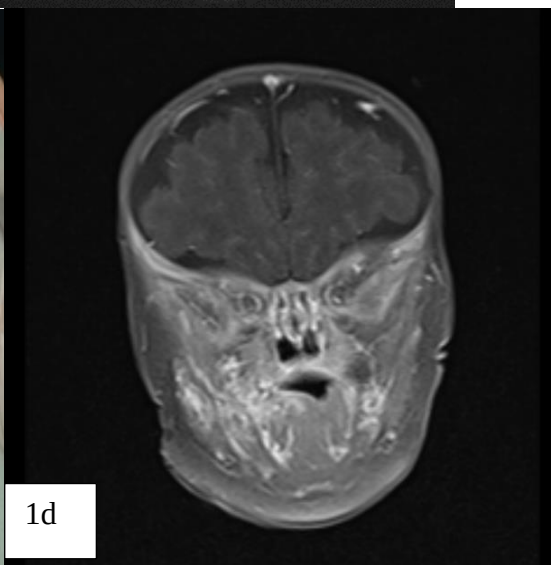

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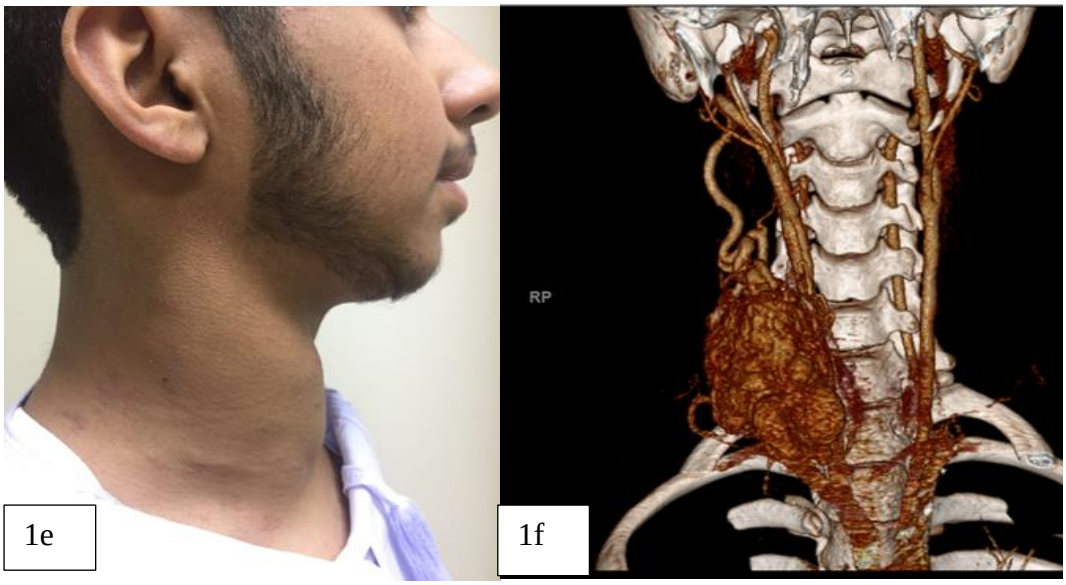


Figure 1: 1a: Lymphovenous CVM, right eye, 1b : MRI Head ; lymphovenous CVM, right eye, 1c: VM of the right cheek and the tongue, 1d : MRI head ,VM , right side of the face, 1e : Right lower neck pulsatile mass, 1f: CT angiogram ; high flow AVM

Case 2

Four month old girl diagnosed with multiple development anomalies - ASD (3.5 mm), pulmonary stenosis, subtle dysmorphism, developmental dysplasia of the hip & generalized tonic clonic seizure , presented with bluish discoloration of the skin around the cheek and tip of the tongue since birth , with multiple venous malformation (VM's)on the right side of cheek which become more prominent as the child cries [Figure 1c]. MRI at initial presentation showed thrombosis of the right transverse and sigmoid venous sinuses with multi-spatial heterogeneous ill-defined lesion along the right side of the tongue and cheek representing a VM [Figure 1d] .Patient is on a daily dose of enoxaparin 6.5 mg for cerebral venous thrombosis. If symptoms of pain or bleeding develop, sclerotherapy would be offered for the VM.

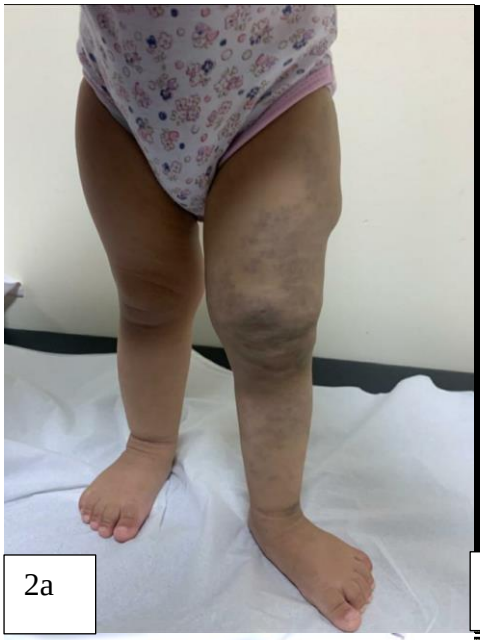
Case 3

Fourteen year-old boy presented with three year history of right lower neck swelling that increased in size gradually, was pulsatile and associated with mild pain. He had no history of trauma. Examination revealed a 12 x10 cm non-tender, pulsatile swelling at the carotid triangle

with engorged veins, a thrill and bruit [Figure 1e]. A CT angiogram [Figure 1f] confirmed a high flow arterio-venous malformation [AVM], with multiple feeders from the external carotid artery, thyrocervical trunk and the vertebral artery draining into the subclavian and internal jugular veins. An ECHO showed LVH in keeping with the high flow AVM. He underwent five sessions of angioembolization using glue and onyx injection and is planned for surgical excision.

Case 4

Fifteen months old girl presented with an irregular blue patch on her left lower extremity since birth, with hemihypertrophy of the limb [Figure 2a] .She started to walk with a limp. Galeazzi test is positive for discrepancy between the tibia . A diagnosis of Klippel-Trennauay syndrome [KTS] was made based on the presence of capillary, venous,lymphatic malformations seen on MRI [Figure 2b]. Custom made grade -2 compression hosiery for the affected limb upto the waist and height correction foot wear has been adviced. The child will be followed up regularly intervals and imaging planned once she is 18 years of age or earlier if she develops a complication - e,g bleed, thrombosis, infection.



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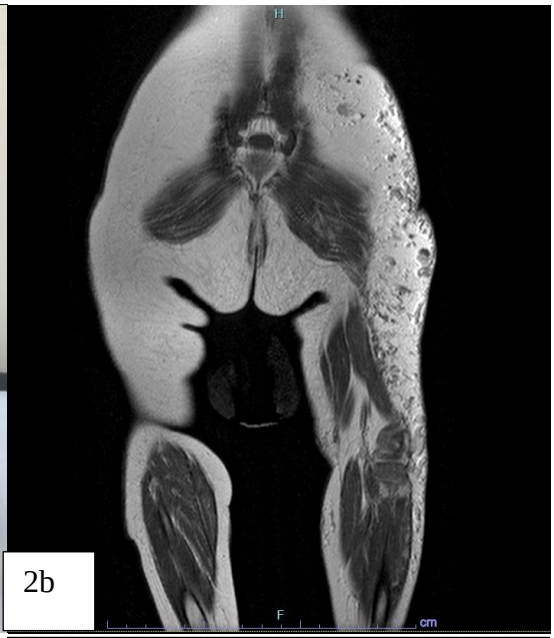

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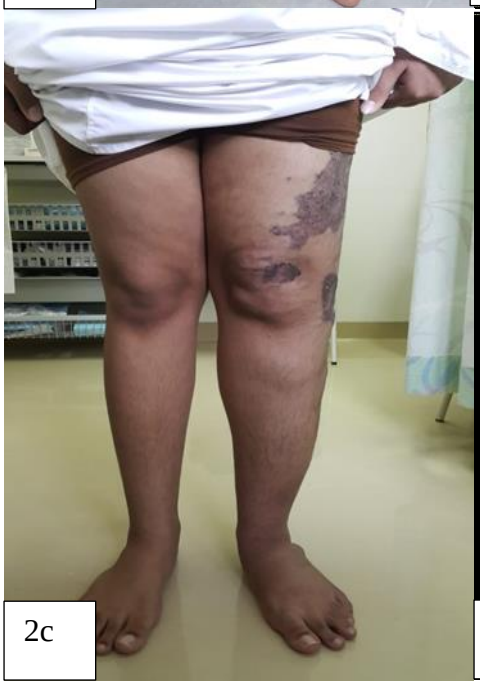

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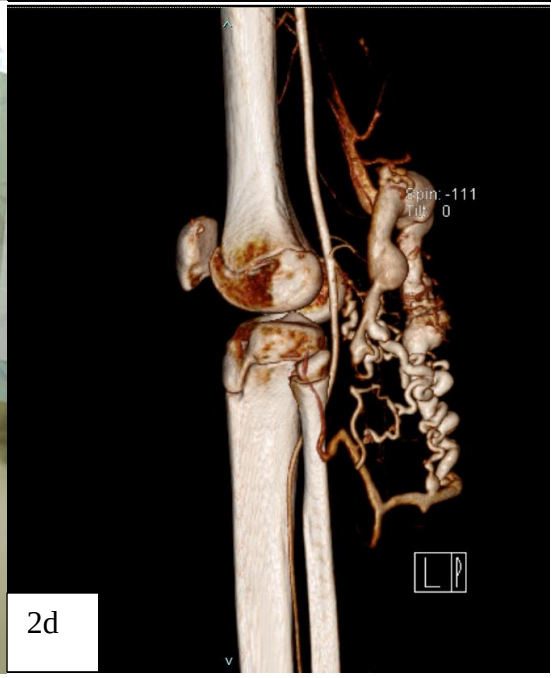

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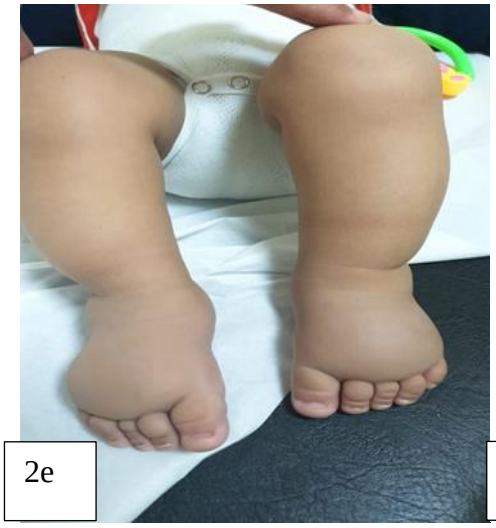

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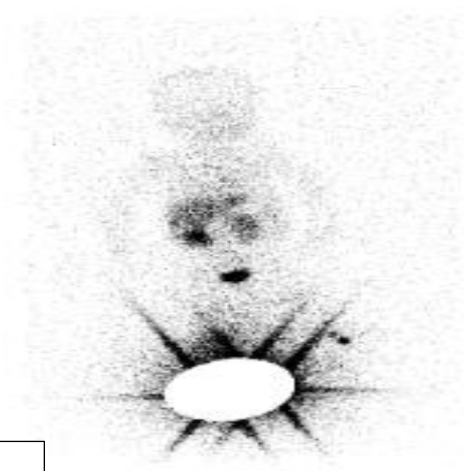

2f

Figure 2: 2a: Left lower limb KTS ; limb hypertrophy, 2b: MRI lower limb showing ; capillary, venous & lymphatic malformations ;KTS, 2c: Angiomatous lesion of the left thigh, PWS, 2d : MRI lower limbs ; left thigh capillary malformation and AVF, 2e: Primary congenital lymphedema of lower limbs, 2f : Lymphoscintigraphy ; no uptake.

Case 5

Fifteen year-old boy presented with left lower limb swelling with an overlying angiomatous lesion in the thigh [Figure 2c] .He was treated as a case of KTS; underwent radiofrequency ablation, sclerotherapy and attempt at varicose vein surgery abroad. During the excision of varicose veins over the popliteal fosssa, he had significant intraoperative bleeding that required blood transfusions and surgery abandoned. MRI [Figure 2d] and angiography , however, showed presence of arteriovenous fistula consistent with Park–Weber Syndrome[PWS]. Following 2 sessions of glue embolization of the fistulas he had reduction in edema and pain. Above knee grade-2 compression stockings continue to be used.

Case 6

Eight-month-old boy born to double consanguineous parents presented with swelling of both feet since birth. On examination he had bilateral pitting edema confined to the dorsal aspect of both feet[Figure 2e]. Lymphoscintigraphy suggested primary congenital lymphedema as there was no uptake of the tracer [Figure 2f]. Custom made grade 2 below knee stockings were prescribed and parents were taught about manual lymphatic drainage (MLD).

Below is a summary of the six cases ; [Figure 3].

Figure 3: Summary of patients gender, age, diagnosis, imaging finding and management.

Case	Age	Gender	CVM	Imaging	Finding	Management
1	8 years	Female	Rt Eye lympho venous malformation	MRI	Large lobulated insinuating lesion noted within the coronal compartment of the right orbit	Intravenous steroids , sclerotherapy, followed by eye drops [Brinzolamide+Timolol]
2	4 months	Male	VM of the rt cheek & tongue	MRI	Multi-spatial heterogeneous ill-defined lesion along the right side of the tongue and cheek	Conservative ; If symptoms of pain or bleeding develop, sclerotherapy would be offered
3	14 years	Male	AVM of the rt lower neck	CT angiogram	High flow AVM 'multiple feeders from the external carotid artery, thyrocervical trunk and the vertebral artery draining into the subclavian and internal jugular vein	Five sessions of angioembolization using glue and onyx injection , planned for surgical excision.
4	15 months	Female	KTS , left lower extremity	MRI	Capillary, venous,lymphatic malformations of the left lower extremity	Conservative ; Custom made grade -2 compression hosiery for the affected limb upto the waist and height correction foot wear .
5	15 years	Male	PWS , left lower extremity	MRI Angiography	Arteriovenous fistula	Two sessions of glue embolization ; above knee grade-2 compression stockings
6	8 months	Male	LM , feet	Lymphoscintigraphy	No uptake	Custom made grade 2 below knee stockings ; MLD

Discussion:

CVM's result from an arrest in the development of blood vessels during embryogenesis. Cells of mesodermal origin that arrest in the early stages of embryogenesis develop into extra-truncular CVM and manifest when there is a triggering factor –e.g. menarche, pregnancy, trauma or surgery; these lesions can recur after treatment. Truncular CVM's on the other hand arise when embryogenesis arrests during the formation of the vascular trunk and they tend to grow with the child, have minimal risk of recurrence and can be associated with hemodynamic compromise.[5] The classification of vascular anomalies by Mulliken and Glowacki in 1982 into congenital vascular malformation (CVM) and vascular tumors has been revised in 1997 by the International Society for the Study of Vascular Anomalies (ISSVA). The authors prefer to - the modified Hamburg classification subdivides CVM into capillary, venous, lymphatic, arterio-venous and combined malformations. Name-based eponyms has been used for many years and which continue to be used such as Klippel–Trenaunay syndrome (KTS), Parkes–Weber syndrome (PWS), Servelle–Martorell syndrom, etc. have been replaced by the modified Hamburg classification. [5, 6, 7, 8, 9] Readers can get further information from the references mentioned.

Venous malformations (70 %) are considered the most common CVM, followed by lymphatic malformations (12 %), arterio-venous malformations (8 %), combined malformation syndromes (6 %) and capillary malformations (4 %). Majority of these lesions are sporadic in nature. The importance of family history cannot be understated. [5, 6, 7]

Below is a diagnostic and management algorithm to help the reader diagnose, explain options and appropriately refer to a higher center (figure 3) [6].

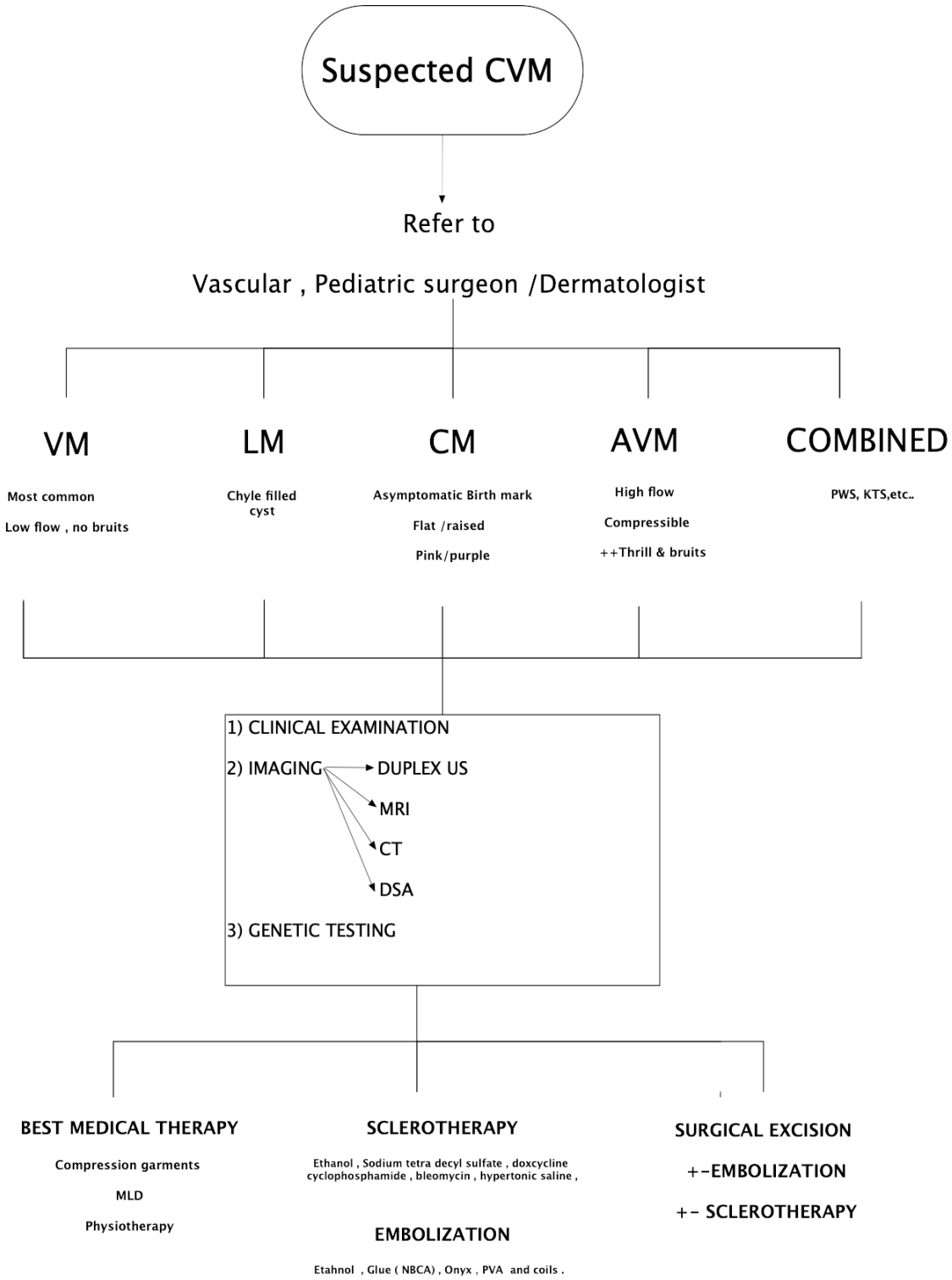

Figure 4: Diagnostic and treatment algorithm of congenital vascular malformations.

CVM = Congenital vascular malformation, VM = Venous malformation CM = Capillary malformation , LM = Lymphatic malformation, AVM = Arteriovenous malformation, MRI = Magnetic resonance imaging, US = Ultrasound , CT = Computed tomography , MLD = Manual lymphatic drainage , DSA =Digital Subtraction Angiography , NBCA= N-Butyl cyanoacrylate, PVA=Polyvinyl alcohol

Conclusion:

CVM's in the Omani population are not as uncommon as thought to be. Knowing the different clinical features, characteristic imaging findings and management options of theses lesions would help health care professional diagnose, counsel and refer appropriately. Molecular genetic studies can be conducted in patients with a family history of CVM. A multi-disciplinary team is needed to treat these patients in a holistic manner and a national registry should be started.

Acknowledgment:

We thank the parents for permitting us to take the pictures of the patients and publish them .

Conflict of interest:
None

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