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Review Article

Congenital Venous Malformations: A Clinico-Pathological Review

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ABSTRACT

Vascular anomalies have been classified in different ways based on clinical, pathological and embryological factors. In 1996, the International Society for the study of vascular anomalies proposed a classification based on that originally published by Mulliken and Glowacki, dividing these vascular lesions into vascular anomalies and tumors. Vascular tumors include hemangiomas and other proliferative lesions. They proliferate largely by endothelial hyperplasia. Vascular malformations may be simple and combined. They never regress and persist and enlarge during later life. Amongst the vascular lesions venous malformations (VM) are distinct lesions originating embryologically from primitive vasoformative tissue which later differentiate into venous system. These lesions mostly occurring subcutaneously and intramuscular are often misdiagnosed as hemangiomas and A-V malformations. They have definite clinical features and can be diagnosed clinically and confirmed by MRI imaging. They can be treated well either by sclerotherapy, Nd-YAG laser application or Surgical excision with good results. Intracranial and gastrointestinal lesions often present with bleeding and require adequate and appropriate care based on the location of the lesions.

Our particular stress in this review which is based on our personal experiences is for appropriate clinical diagnosis of these vascular malformations, which may be confirmed by MRI imaging and can be adequately and safely treated by surgical excision.

We observed that these lesions are very often misdiagnosed clinically due to two reasons: 1) Even though they are of congenital and embryonic origin they report during 2nd or 3rd decade. 2) The overlying skin doesn't show any changes except some bluish discoloration due to prominent dermal veins. 3) Sign of blanching, sign of compressibility, local warmth or pulsations seen in majority of vascular lesions are not usually seen in these cases.

Key words: Congenital Vascular Malformations, Vascular Tumors, Hemangiomas, Venous Angiomas.

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INTRODUCTION

The past two decades have seen great advances in understanding of the pathophysiology, classification, nomenclature and treatment of vascular lesions.[1-4] Venous malformations are most common of all vascular lesions with incidence of 1-4 % of the population. There is no sex predilection. They are often misdiagnosed as cavernous hemangiomas or A-V malformations.

These are low-flow lesions present since birth with thin walls. Most are solitary but multiple lesions can occur.

Venous malformations are always present at birth.[5] They may manifest clinically in infancy, childhood or adulthood; most commonly during the 2nd decade. They present as blue soft swellings subcutaneous, intramuscular, gastrointestinal tract or intracranial. Dilated anomalous intradermal venous channels account for blue discoloration. Often present as soft, cystic swelling with sign of compressibility. But in all our cases sign of compressibility is absent. Many are associated with Phlebothrombosis leading to phleboliths and calcification. Most of these patients complain of pain and tightness after getting up from sleep in the morning probably dueto venous stasis and microthrombi. Becomes prominent with dependency and decreases with elevation.

CLINICAL FEATURES[6-9] (Figures 1-4)

They present as soft swellings, subcutaneous or intramuscular in skeletal muscles. They are present since birth but usually noticed in adolescent or young adult age. Bluish in appearance even though skin is not affected and usually painless. Occasionally

tender due to phlebothrombosis and phleboliths. Those with intramuscular lesions particularly in the limbs complain of pain while doing manual work.

There are no pulsations and sign of compressibility is often absent. Often incorrectly diagnosed as cavernous hemangioma.

Sometimes prominent veins are seen surrounding the swelling. Usually the lesions are single, but multiple lesions can occur like in Blue-rubber bleb syndrome with multiple lesions on the trunk, palms, soles of feet as well as lesions in gastrointestinal tract which may bleed.

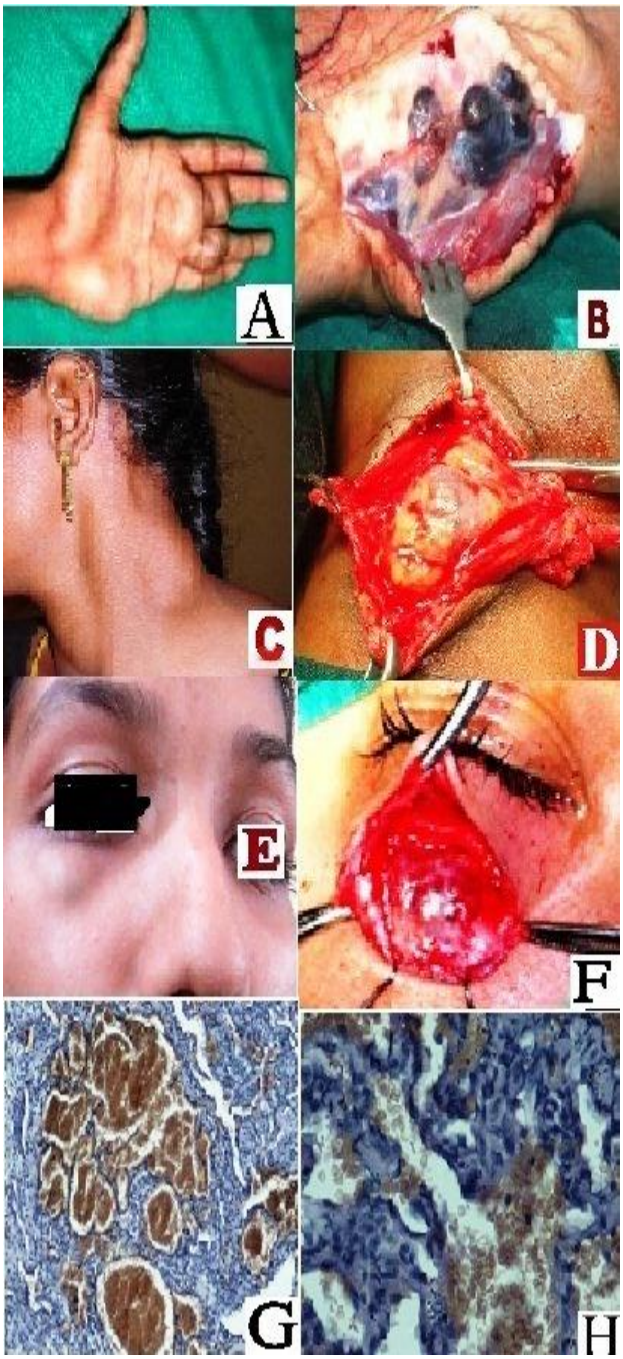

Figure 1: (A) Case 1 Preoperative; (B) Case 1 Per operative; (C) Case 2 Preoperative; (D) Case 2 Per operative; (E) Case 3 Preoperative; (F) Case 3 Per operative; (G) Microphotograph H & E X 10 collection of thin walled dilated veins filled with hemorrhage; (H) Microphotograph H&E X 40 Veins lined by flattened endothelium and break in the wall.

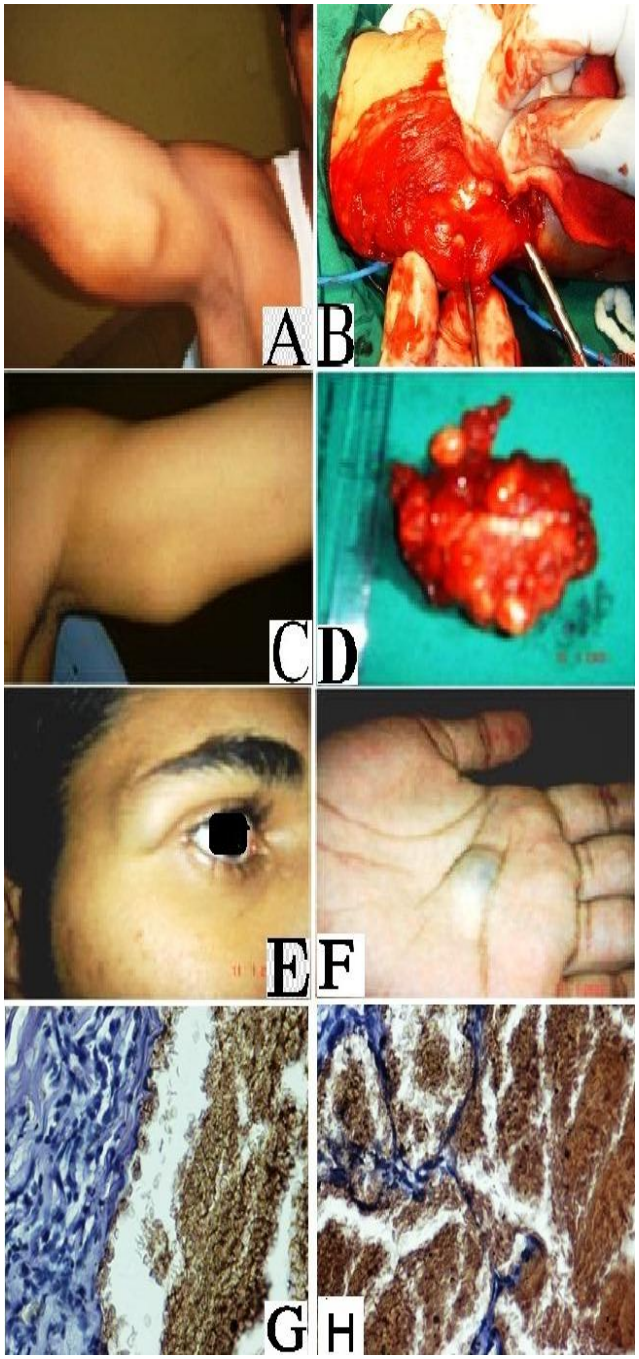

Figure 2: (A) Case 4 Preoperative; (B) Case 4 Peroperative; (C) Case 5 Preoperative; (D) Case 5 Peroperative; (E) Case 6 Preoperative eyebrow; (F) Case 6 Preoperative Lt. Palm; (G) Microphotograph H & E X 40 - Veins lined by flattened endothelium; (H) Microphotograph H & E X 40 - Composed of one or more dilated veins.

CASE HISTORIES

Case-1: Female aged 18 yrs. Having swelling in the left hand palmar aspect, in the hypothenar region extending into middle and ring fingers, present since birth, gradually increasing in size and are painful for the last 3 months.

Case-2: Female 20 yrs. Referred from another Tertiary care institution with a swelling left side of the neck in the posterior triangle present since childhood and increasing in size with age. It is painful occasionally for the last 5 yrs.

Case-3: Female child 13 yrs. Having swelling right lower eyelid since childhood, gradually increasing in size and occasionally painful. Referred from General surgery and Ophthalmic outpatient department.

Case-4: Male 18 yrs. Came to the hospital with a painful swelling in the left arm posterior aspect which is present since birth and gradually increasing in size. More painful while working and relieved after taking rest. Clinical examination showed the swelling situated in the posterior belly of the triceps muscle.

Case-5: Male 30 yrs came to the outpatient department with a swelling in the posterior aspect of the right arm present since birth which was gradually increasing in size. Getting more pain while working and relieved with rest.

Case-6: Male 17 yrs. Came with multiple bluish swellings present since childhood 1. Right eyebrow outer aspect 2. Left palm 3. Occipital region and 4.left cheek.

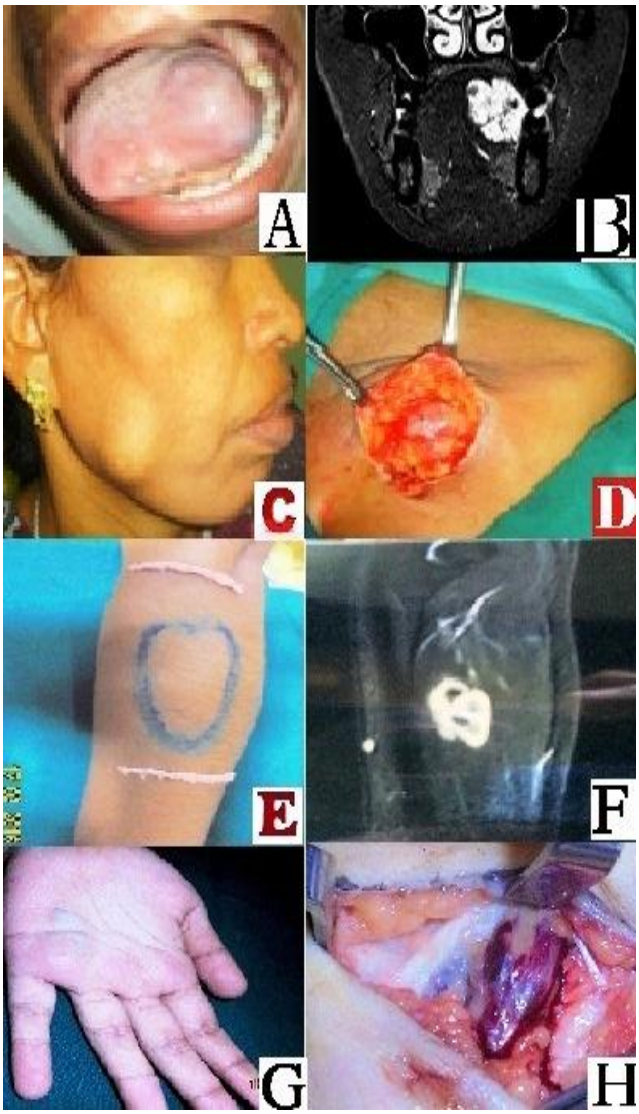

Figure 3: (A) Case 7 Preoperative; (B) Case 7 MRI Picture; (C) Case 9 Preoperative; (D) Case 9 Peroperative; (E) Case 10 Preoperative; (F) Case 10 MRI Picture; (G) Case 11 Preoperative; (H) Case 11 Peroperative

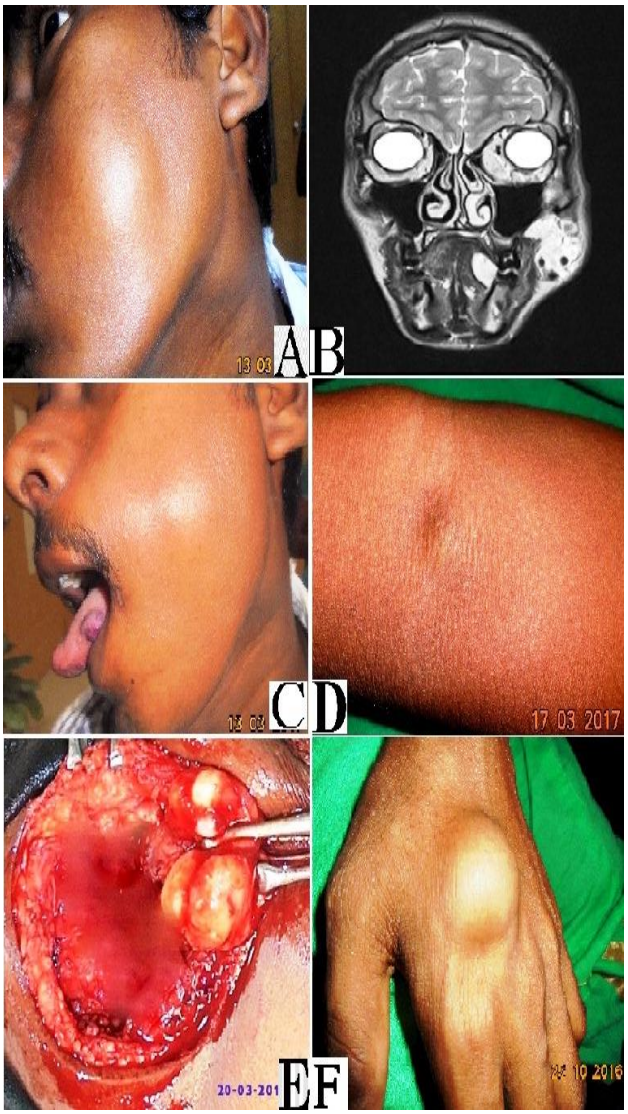

Fig 4: (A) Case 13.Preoperative; (B) Case 13: MRI face showing the lesion; (C) Case 13.showing lesions in the Neck, toungue; (D) Case13:lesion in the cubital fossa;(E) Case13:per operative with phleboliths; (F) Case 14. Preoperative.

Case-7: Female 23 yrs came to the outpatient department with congenital tongue tie and swelling left side of the anterior two-thirds of the tongue present since childhood. She complains of pain early in the morning after getting up from sleep.

Case-8: Male child 10 yrs age was brought to the hospital with a painful swelling in the left index finger, present since birth, gradually increasing in size. Referred from pediatric surgeon as hemangioma and Ultrasound diagnosis as ganglion or lipoma.

Case-9: Female 44 years with a swelling in the right masseteric region and occasionally painful for the last 6 months. Subcutaneous, soft superficial to masseter muscle. Referred from general surgery department.

Case-10: Female aged 23 yrs came with a swelling in the upper part of the left forearm intramuscular present since birth, but gradually increasing in size and painful while working.

Case-11: Female 15 yrs with a bluish swelling in the left palm also extending on to the dorsal aspect of 6 yrs duration and painful over the last 6 months

Case-12: Female child aged 12 yrs with swelling lower lip right side extending on to the cheek and right angle of the mouth, bluish and occasionally painful. Referred from pediatric surgery.

Case-13: Male aged 22 yrs came with multiple congenital swellings of left cheek, upper neck lt, tongue and rt cubital fossa. He underwent surgery for the left cheek lesion in childhood, which recurred within a short time.

Case-14: Female child 12 yrs came with a solitary, soft, cystic, bluish swelling over the dorsum of the left hand present since birth, but is painful since 6 months and increasing in size.

Even though all these lesions are congenital and embryonic in origin, all the patients came to the hospital for consultation during the 2nd or 3rd decade and all of them because of increasing size of the swelling for cosmetic reasons and associated pain. The patients with intramuscular lesions in the arm used to have pain while doing manual or agricultural work.

All the lesions were found to be confined deep to the skin, in the subcutaneous tissue or intermuscular. The overlying skin is normal. Bluish discoloration is seen in all the cases. Almost all the cases were diagnosed as hemangiomas or A-V malformations and referred to us.

Table I: Case Details

No.	Sex	Age	Site	Duration	Referral Diagnosis	Treatment
1	**Female**	18 yrs	Lt. hand palmar aspect	Since childhood - painful since 3 months	Cavernous Hemangioma	Excision
2	**Female**	20 yrs	Lt. side of the neck posterior triangle	Present since childhood - Painful last 05 years	? AV malformation Hemangioma	Excision
3	**Female**	13 yrs	Rt. lower eye lid	Since childhood - (Referred from Ophthalmology)	Hemangioma	Excision
4	**Male**	18 yrs	Lt. arm post aspect (intramuscular)	Since child hood - painful while working	Cavernous Hemangioma	Excision
5	**Male**	30 yrs	Post aspect of rt. arm (intramuscular)	Present since childhood - Painful in the morning and while working	Cavernous Hemangioma	Excision
6	**Male**	17 yrs	Multiple rt. eyebrow, scalp, lt. cheek, lt. palm	Since Childhood - painful since 6 months and increasing in size	Hemangioma	Excision of eyebrow & lt palm lesions
7	**Female**	23 yrs	Lt. side of anterior 2/3 of tongue	Since Childhood (Pain during early morning)	? Cavernous Hemangioma	Excision
8	**Male**	10 yrs	Lt. index finger (middle phalangeal region)	Present since childhood - Painful while working with the hand .Ref from gen.surgery/paed.surgery	Ganglion Hemangioma	Excision
9	**Female**	44 yrs	Subcutaneous, rt. masseteric region, intramuscular	Present since childhood - Painful since 6 months	Lipoma	Excision
10	**Female**	23 yrs	Lt. forearm intramuscular	Since 6 years - Painful in the night and early morning	AV Malformation	Excision
11	**Female**	15 yrs	Lt. hand palmar aspect	Since 6 years - increasing in size and painful since 6 months	Hemangioma	Excision
12	**Female**	12 yrs	Lower lip and cheek (rt. side)	Present since birth - increasing in size with pain for the last 2 years	Cavernous Hemangioma	Excision
13	**Male**	22 yrs	Multiple, lt cheek, neck, tongue, cubital fossa rt.	Present since birth-was operated in childhood-recurred-painful	Multiple hemangiomas	Excision of lt cheek lesion.
14	**Female child**	12 yrs	Solitary soft cystic bluish lesion dorsum of lt hand	Present since birth-painful for the last 6 months	Cavernous hemangioma	Excision.

MANAGEMENT

After clinical evaluation and necessary investigations like ultrasound scan or MRI wherever necessary we performed surgical excision. We did not encounter any problem during or after surgery. In cases of intramuscular lesions we repaired the muscle defect. All the patients had uneventful recovery and had no subsequent problems or recurrences. We strongly believe and advocate surgical excision as mainstay of treatment particularly in localized lesions, since it is safe and gives relief in one stage in a short duration. Even in cases -2,4, and 5 where the lesions were nearer to the brachial vessels in the arm and carotid sheath in the neck, we did not encounter any problems.

HISTOPATHOLOGY (Figure. 1G, H & 2G,H)

Histological specimens are stained with Hematoxylin and eosin. Histological examination of these lesions are characterized by malformed, compactly arranged vessels with partly degenerated walls.[10-13] There are thin walled dilated sponge like abnormal channels with surrounding smooth muscle distorted into clumps. In some cases there is evidence of Phlebothrombosis. Phleboliths with calcification and fibrofatty tissue surrounding the lesion. Also seen are areas of hemorrhage and a layer of hemosiderin laden macrophages around the blood vessels.

Cellular characteristics are flat endothelium, normal mast cell count, dysplastic walls with thin basement membrane.

Urinary basic fibroblastic growth factor will be low in these cases (BFGF).[14]

HEREDITY FACTORS

A mutation for venous malformations was identified in a gene that codes for an endothelial receptor on chromosome 9 p and Turner syndrome can be associated with multiple venous malformations and multiple glomangiomas with autosomal dominant transmission. It could also be familial.[15-17]

RADIOIMAGING[18-20] (Figure. 3 B, F & 4B)

Plain X-ray shows phleboliths as radioopaque shadow. MRI is the most useful tool. Brighter signal than fat on T2 Weighted sequences with more uniform enhancement seen on MRI and phleboliths can manifest as signal voids.

MANAGEMENT OF VENOUS MALFORMATIONS

Treatment is indicated for the following reasons

Pain, Swelling, Bleeding, Functional Deficit & Cosmetic reasons.

The patients usually come during second or third decade of age for treatment because of (A) Increasing size of the swelling and cosmetic reasons, (B) pain and (c) inability to work in some cases due to functional deficit.

MODALITIES OF TREATMENT

1) The useful therapeutic modalities are percutaneous sclerotherapy. Sclerotherapy requires multiple stages and take prolonged period for the desired results. The frequently used sclerosant agents are as follows[21-25]
   Absolute ethanol, Hypertonic saline, Bleomycin**,** Doxycycline, 3% sodium tetradecyl sulfate and OK-432.
2) Custom made elastic garments particularly for the lesions of the extremities and surgical excision alone or in combination with sclerotherapy.
3) Laser therapy is useful in selected cases. Argon or YAG laser has been used in the lesions of the oral cavity[26]
4) Surgery is indicated in isolated, symptomatic lesions particularly in the head and neck region and results are quite satisfactory provided the lesion is completely excised. In general Surgery or Sclerotherapy is more successful in dealing with pure venous malformations than when dealing with mixed vascular malformations. In all the cases, which are pure or localized, surgical treatment gave excellent results without any incidence of recurrence in all our cases.[27,28]

PROGNOSIS AND OUTCOMES

The results and prognosis are directly related to the size of the lesion and location of the lesion. There is likelihood of perioperative morbidity and recurrences with more diffuse vascular malformations and those intimately related to important neurovascular structures.

CONCLUSIONS

Isolated venous malformations (VM) are slow-flow vascular malformations and are distinctive clinical and pathological entities and are underreported often mistaking to hemangiomas. They never regress and often go for medical consultation due to pain during 2nd or 3rd decade.

They can be diagnosed clinically, confirmed by Magnetic Resonance Imaging (MRI)or MRV and by Histopathology. Even though they are of congenital and embryonic origin most of them seek medical advice in 2nd or 3rd decade because of swelling and pain associated with phlebothrombosis and phleboliths. Different protocols of treatment like sclerotherapy, use of Nd-YAG laser etc. are useful; Surgical excision remains the best choice of treatment. Carefully planned surgical excision gives very good and complete relief to the patients in a single stage.

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