Venous Malformation: Truncular Form

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Subtyping of venous malformation (VM) into truncular and extra-truncular form derives from the embryological background in Hamburg classification of CVM (see Chap. 9). In the International Society for the Study of Vascular Anomalies (ISSVA) classification, it is classified as "vascular anomaly of major named vessels" which is referred to "truncular form" malformations [1].

To understand the truncular congenital vascular malformation (CVM), it is important to know the pathogenesis of truncular VM. Primitive vascular tissue in the limb bud first appears in the third week of gestation. During the first stage (undifferentiated stage) of vasculogenesis, only a capillary network appears. In the second stage (retiform stage), large plexiform structures can be seen. In the third stage (maturation stage), it developed to larger channels and differentiate to the arteries, veins, and lymphatics [2].

Truncular VM lesions are believed to occur as a result of developmental arrest in the "later" stage of the vasculogenesis. Since this developmental arrest occurs after an early stage (reticular stage) of the vasculogenesis, these lesions are also known as "post-truncal lesions." In contrast to "pre-truncal or extra-truncular lesion, truncular VM lesions do not have characteristics of an early embryonic vascular tissues, which can proliferate after birth by certain stimuli [3].

An International Interdisciplinary Consensus Committee on Venous Anatomical Terminology [4] defined venous abnormality as follows in Table 15.1.

It is reported that VM is the most common type of CVM, and deep vein anomaly presents in 47% of VM patients including various types of deep vein anomalies: *venomegalia* (phlebectasia) (36%), aplasia or hypoplasia (8%), venous aneurysm (8%), and avalvulia (congenital absence of valves in the venous trunks, 7%) [6].

The prevalence of deep venous anomalies is reported even higher in patients with mixed form of CVM which affects extremity veins, skin capillary (CM), lymphatic system (LM), and rarely arteriovenous malformation (AVM) [7].

The prevalence can be variable depending on the type of investigation and the definition of the anomaly. According to a study with fresh, non-embalmed cadavers by Jean-François Uhl et al. [8], they found that femoral venous system was unitruncular in 91% and bitruncular in 9%. Unitruncular femoral vein includes normal configuration (88%), deep femoral trunk (2%), and axio(sciatico)-femoral trunk (1%). Human

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Table 15.1 Terminology regarding abnormal caliber of the extremity vein

Anomaly	Definition
Dysplasia [4]	Complex abnormality of development of a vein or of a group of veins that greatly differ from the normal conditions in size, structure, and connections
Atrophy [4]	Decrease in size or wasting away of a normally developed vein or segment of a vein, following a degenerative process
Venous aneurysm [4]	Localized dilation of a venous segment, with a caliber increase ≥50% compared with normal
Venomegalia [4]	Diffuse dilation of one or more veins with a caliber increase ≥50% compared with normal
Agenesis [5]	Complete absence of a vein or of a segment of a vein
Aplasia [5]	Lack of development of a vein or of a segment of a vein. The vein is present but diminutive in size, and its structure is similar to that in the embryo
Hypoplasia [5]	Incomplete development of a vein or of a segment of a vein

Caggiati et al. [4]

leg venous system finishes their embryologic development through the evolution and involution. In the case of axio(sciatico)-femoral and deep femoral trunk, the femoral vein is hypoplastic, and the axial (sciatic) vein or deep femoral vein works as the main draining trunk of the thigh. The venous duplication itself (e.g., double femoral veins) or patent but variant venous anatomy such as axio-femoral or deep femoral trunk does not cause hemodynamic abnormality in clinical practice.

According to the SMC database, venous predominant CVM accounts for 51.5% of all CVMs, and truncular VM accounts for 14% of all VM lesions affecting the extremity (Table 15.2).

Persistent marginal vein (MV) is not uncommon in patients with a lower extremity VM. It is an atypical truncular vein that failed to regress and characterized by superficially located dilated vein along the lateral aspect of the lower extremity with variable extension from the calf to the buttock.

Table 15.2 Frequency of truncular VM and various deep venous anomalies (at SMC, 1994–2015)

All CVM patients	*N* = 3063
Venous malformation	1576 (51.5%)
Pure VM	1378 (87%)
Venous predominant mixed CVM	198 (13%)
VM affecting the extremity	1019 (65%)
Extra-truncular VM	949 (93%)
Truncular[a] VM	146 (14%)
Venous aplasia	25 (17%)
Venous hypoplasia	18 (12%)
Venous aneurysm	1 (0.6%)
Venomegalia (phlebectasia)	12 (8%)
Persistent marginal vein	108 (74%)

VM venous malformation, *CVM* congenital vascular malformation

[a]Diagnosis of truncular VM was made with duplex ultrasonography, MRI, whole-body blood pool scintigraphy, and/or venography

Port-wine skin lesion (capillary malformation) and limb hypertrophy often coexist with MV as a combined type CVM. In addition to cosmetic problem due to varicosity and associated skin lesion, the clinical significance of MV is chronic venous insufficiency due to congenital absence of a venous valve in the MV. Besides, MV remains as a source of pulmonary embolism. MV is often associated with varying range of deep venous anomalies from aplasia or hypoplasia to intrinsic defects such as webs, stenosis, or defective valves of deep vein system. Weber et al. [9] classified marginal vein in five types with venographic study (Fig. 15.1).

As described above, the main clinical features of the truncular VM are venous hemodynamic consequences due to steno-occlusive (aplasia, hypoplasia), dilated lesions (venous aneurysm, venomegalia) of deep venous system in the extremity and venous reflux due to congenital absence of venous valve.

Since truncular VM is often combined with other forms of CVM such as capillary malformation (CM, skin red spot), lymphatic malformation (LM, lymphedema due to aplasia or hypoplasia of the main lymphatic trunks, lymphatic cyst or bleb, lymphorrhea), limb hypertrophy, or limb length discrepancy, the patients may present with

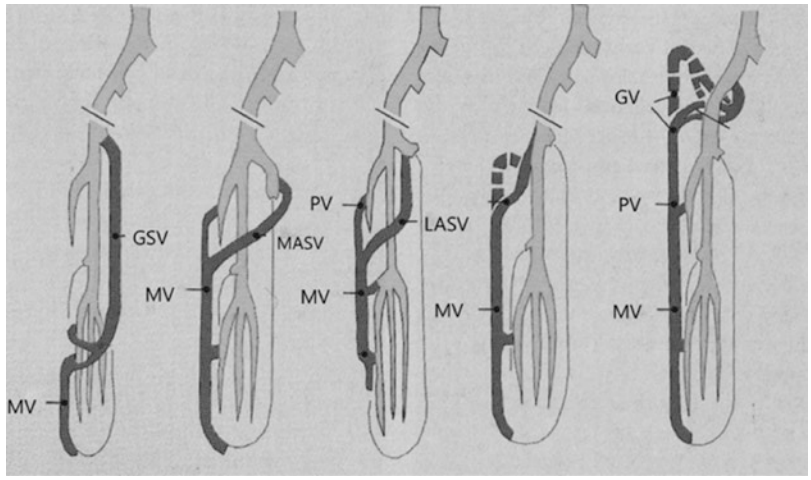

Fig. 15.1 Venographic classification of the marginal vein (Reprinted with permission [9]). *MV* marginal vein, *GSV* great saphenous vein, *MASV* medial accessory saphenous vein, *PV* perforator vein to deep femoral vein, *LASV* lateral accessory saphenous vein, *GV* gluteal vein

clinical features of complications of VM and LM such as cellulitis, venous thrombosis or pulmonary embolism, and stasis ulcer.

According to the extent and anatomic site of the VM lesion and combined CVM or its complication, clinical features are variable.

Because of sluggish blood flow through the anomalous venous channels, venous thrombosis is frequently formed causing local pain and tenderness. When the thrombus is superficially located, it may be palpable on physical examination and detectable on CT or MRI. Constant leg heaviness or acute intermittent episodes of pain may be a prominent feature of the VM patients. Various causes of leg pain are suggested in patients with mixed-type veno-lymphatic malformation (Klippel-Trenaunay syndrome), which include chronic venous insufficiency, cellulitis, growing bone pain, thrombophlebitis or venous thrombosis, phlebolith, intraosseous VM, arthritis, and neuropathic pain [10].

Phlebolith is a calcified organized venous thrombus, which is a characteristic feature of extra-truncular VM lesion. Diagnosis of VM in general is usually made with physical finding and either MRI or duplex ultrasonography. Deep vein anomaly (truncular VM) can be easily detected with duplex ultrasonography (US). Duplex US gives us an excellent morphological (caliber, depth, extent of deep vein abnormality or marginal vein) and hemodynamic information (connections with perforators and deep veins, flow direction) and presence of coexisting extra-truncular VM lesion.

For more detailed assessment of the associated VM lesion, MRI can be recommended. MRI is the most informative tool in the diagnosis and follow-up examinations of VM patients. Its advantages over duplex US are its capability to visualize the anatomic relationships between VM lesion and adjacent tissue (nerve, soft tissue, organ, bone, and joint). T2-weighted images are mainly used to evaluate the extent of the VM lesion, while gradient recalled echo (GRE) images are used to identify the lesion's flow characteristics. Gadolinium-enhanced MRI is used to determine the extent of the VM lesion and to distinguish slow-flow VM from lymphatic malformation (LM). And 3D, T1-weighted, contrast-enhanced MR image is similar to a venogram and readily identifies feeding and draining vessels [11].

Catheter venogram is not routinely performed as a diagnostic tool for patients with VM. However, venography is an essential road map during an embolo-sclerotherapy of an associated VM lesion because adjacent MV can provide a pathway of

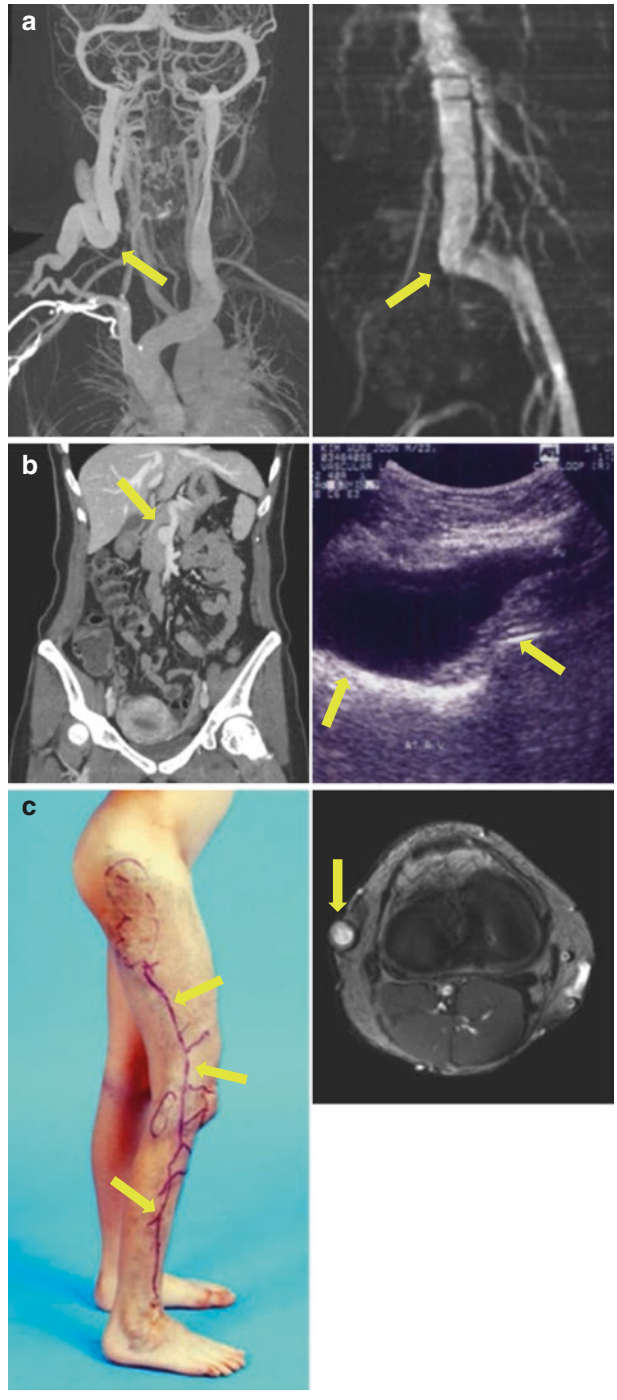

Fig. 15.2 Various clinical features of truncular VM. (**a**) Agenesis (aplasia): (*left*) right brachiocephalic vein agenesis on MR venogram and (*right*) right iliac vein agenesis on MR venogram. (**b**) Aneurysmal dilatation: (*left*) superior mesenteric vein aneurysm on CT venogram and (*right*) popliteal vein aneurysm on ultrasonography. (**c**) Persistent marginal vein along the lateral aspect of the right leg: (*left*) medical photo of the marginal vein in the right leg and (*right*) marginal vein in the right leg on contrast-enhanced CT scan

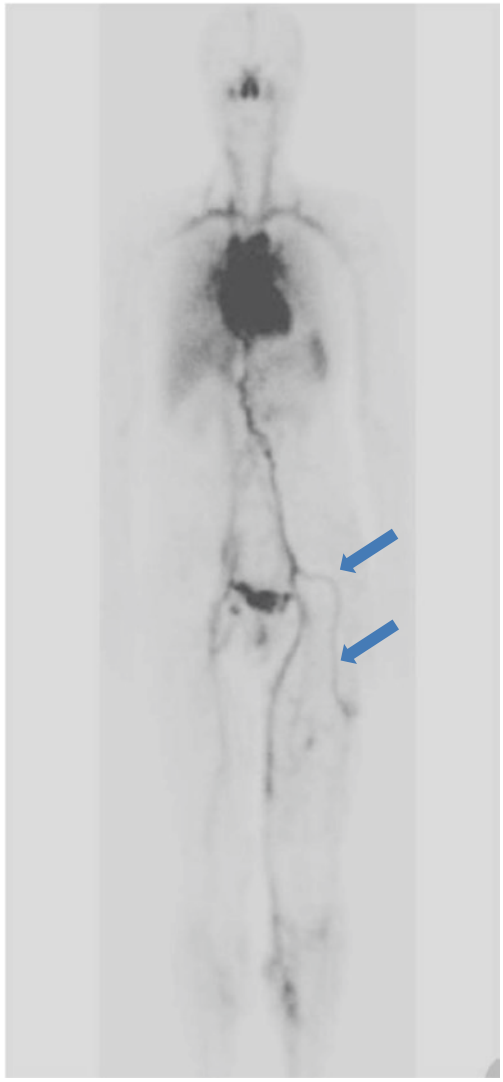

Fig. 15.3 Tc-99 m RBC whole-body blood pool scintigraphy (WBBPS). WBBPS in a 44-year-old male patient with venous malformation (VM) in the left leg, which shows uptake defect in the deep vein and increased uptake in the great saphenous vein and marginal vein (*arrow*)

pulmonary embolism from the preexisting venous thrombus in VM or embolization material.

We perform whole-body blood pool scintigraphy (WBBPS) to confirm the distribution of all VM lesions. WBBPS is a whole-body imaging obtained with dual-head gamma camera after intravenous injection of the radiolabeled erythrocytes (99mTc-RBC). High sensitivity (97%) is reported in the diagnosis of VM [12].

It can provide not only an abnormal blood pooling or lack of normal blood filling in the whole body (Fig. 15.3) but also interval changes of VM lesions with semiquantitative assessment of the amount of abnormal blood pooling in the VM lesion. Furthermore, WBBPS can also be an excellent tool to distinguish VM lesion from LM in patients with mixed form CVM.

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