# 2 Predominantly Venous Malformation

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## 2.1 Introduction

Vascular anomalies are divided into vascular tumours and vascular malformations. Vascular malformation classification is based on the anomalous channels: arterial, venous, lymphatic, or capillary. The most frequent vascular anomalies are venous malformations. The incidence is estimated to be around 1 in 10,000 [1]. The reader must be made aware of the numerous confusing misnomers such as glomangioma, cavernous haemangioma, and haemangioma that have also been used in prior literature.

## 2.2 Genetics

Venous malformations are sporadic in most cases but can be inherited. A locus for autosomal-dominant multiple cutaneous and mucosal venous malformations, VMCM1, was identified on chromosome 9p21 [2–4]. A mutation was found in the endothelial cell-specific receptor tyrosine kinase TIE-2 [5]. This mutation is likely to occur in vascular malformations.

## 2.3 Clinical Features

Generally, venous malformations are noted at birth but can also appear during infancy. They involve any tissue or organ in all anatomic locations. Venous malformations grow with the patient. Exacerbations can occur during puberty or pregnancy. Indeed, hormonal modulations of venous malformations can be seen during the menstrual cycle or under anovulant therapy. Surgery or trauma can also lead to the progression of venous
malformations. At clinical exam, venous malformations appear as soft tissue lesions that are compressible, and increase with Valsalva manoeuvre or gravity. The overlying skin can be normal, bluish, or purple in colour. The lesion is cold and not pulsatile. Bleeding is uncommon. Hematologic evaluation is important and must include complete blood count, fibrinogen, and d-dimer dosage. Localized intravascular consumption can be observed, especially in extensive venous malformations [6]. Coagulopathy can be exacerbated by sclerotherapy or surgery [7–9]. Extensive facial venous malformations are frequently associated with intracranial developmental venous anomalies [10].

2.4 Focal and Diffuse Venous Malformations

Venous malformations are either focal or diffuse, and may involve the skin, the mucosae, the muscles, the bones. In cases of extensive venous malformations, associated distal sites of involvement are likely.

2.5 Associated Syndromes

2.5.1 Blue Rubber Bleb Nevus Syndrome

Blue rubber bleb nevus syndrome, a rare disorder, is characterized by continuous development of multiple focal venous malformations of the skin, musculoskeletal tissue, and mucosa throughout the body, including the gastrointestinal tract. These venous malformations can be treated by sclerotherapy or surgery. In some cases of blue rubber bleb nevus, it has been shown that the mutation may occur on chromosome 9p.

2.5.2 Mucocutaneous Familial Venous Malformations

The characteristics of mucocutaneous familial venous malformations are the same as those of the blue rubber bled nevus syndrome except for the lack of gastrointestinal involvement.

2.5.3 Glomovenous Malformations

Glomovenous malformations are venous malformations associated with glomus cells. The smooth layer is formed by glomus cells, which are smooth muscle precursor cells [11]. Glomovenous malformations frequently recur. Sclerotherapy can be useful for their treatment.

2.5.4 Maffucci’s Syndrome

Maffucci’s syndrome consists in the association of venous malformations and multiple enchondromas. Intraosseous venous malformations as well as enchondromas are responsible for the bony defects [12].

2.6 Coagulopathies

Localized intravascular coagulopathy is reported in venous malformations. Most patients with venous malformations have no clinical signs or symptoms at presentation. A chronic form of consumptive coagulopathy is however possible as shown by positive d-dimer levels, and normal or low platelets and fibrinogen values. Consumptive coagulopathy must be corrected prior to initiating the treatment of vascular malformations. Low-molecular-weight heparin and elastic stockings are recommended [8]. Administration of cryoprecipitate, platelets, or fresh frozen plasma to patients with chronic coagulopathy has proved to be helpful before performing sclerotherapy or embolization in order to obtain a successful thrombosis [9].

2.7 Histopathology

Venous malformations are due to the abnormal development of the vein wall. Microscopy shows multiple thin-walled vessels with flattened endothelial cells, lacking proliferative features [1] (quiescent endothelium), and wall smooth muscle deficiency. The deficient smooth muscle layer leads to the inability of the affected veins to constrict normally,