

TITLE: Congenital Vascular Malformations

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Vascular Malformations

“Vascular malformation” is a generalized term used to describe a group of lesions, present at birth, formed by an anomaly of angiovascular or lymphovascular structures. There has been historical confusion over which lesions should be included, or excluded, from the category of vascular malformation, as well as over a suitable classification scheme within the category. Virchow, and his student, Wegener, in 1880, first separated vascular lesions into angiomas and lymphangiomas. They classified these as simplex, cavernosum, or racemosum. This original classification was based on histological appearance, and did not take into account biological behavior. It wasn't until 1982 when Mulliken and Glowacki described a classification based upon structure, as well as behavior, that a practical clinical approach to these tumors was possible. Mulliken's dichotomy separated vascular lesions into hemangiomas and vascular malformations with two main clinical characteristics defining the two:

Hemangiomas

Hemangiomas are usually not present at birth, but become apparent during the first few weeks of life. This appearance is usually followed by rapid progression during the first two years of life, followed by slow involution. Vascular malformations, on the other hand, usually are present at birth. They then grow proportionately with the child, showing no signs of spontaneous resolution. On a cellular level, hemangiomas and vascular malformations are quite different as well. While hemangiomas are true tumors, characterized by hyperproliferation of vascular endothelium, malformations display normal endothelium with a progressive dilation of vascular channels.

Vascular malformations, now clearly differentiated from hemangiomas, are described and classified according to their major vascular component. The major categories are lymphatic malformation, capillary malformation, venous malformation, and arteriovenous malformation. Although these categories are helpful for description, many malformations contain elements from more than one type, thus being described as mixed lesions. Burrows, in conjunction with Mulliken, in 1983, further described malformations as either high flow, having a connection to

the arterial or capillary system or low flow, having a connection to the venous or lymphatic system.

Whereas high-flow vascular anomalies, such as arteriovenous fistulas and arteriovenous malformations, are adequately addressed by means of transarterial embolization, low-flow malformations found to be solitary or combined in capillary, venous, or lymphatic vessels are successfully treated with sclerotherapy. In 1988, the Hamburg classification was created, delineating lesions into truncular malformations, derived from a differentiated embryological vascular truncus, and extratruncular malformations, derived from remnants of primitive capillaries.

Lymphangiomas

Diagnosis of lymphatic malformations is made using physical examination, aided by radiographs. Numerous non-invasive and invasive radiologic studies aid in diagnosis. Noninvasive studies include MRI, duplex ultrasound, whole body blood pool scintigraphy, transarterial lung perfusion scintigraphy, lymphoscintigraphy, and CT scan. 3 non-invasive tests are sufficiently accurate and obviate the need for invasive studies.

Invasive procedures, such as traditional arteriography are usually reserved for treatment planning. Ultrasound is generally the initial test performed. It is readily available, inexpensive, and can give the physician an idea on the extent of the lesion.

More detailed information is gleaned from MRI studies. Studies should include T1- and T2-weighted spin-echo imaging in multiple planes, fat-saturated T1-weighted imaging with the intravenous administration of a gadolinium-based contrast agent, and gradient-recalled echo (GRE) imaging. T2-weighted images are used mainly to evaluate the extent of the abnormality. GRE images are used to identify the hemodynamic nature of the condition (high- vs low-flow lesions); and contrast-enhanced images are used to determine the extent of the malformation and to distinguish low-flow vascular anomalies (venous malformation versus lymphatic malformation). For any vascular anomaly, the basic approach is first, to evaluate fat-suppressed T2-weighted images to determine the extent of the anomaly, and second, to evaluate the GRE images to decide whether the anomaly is a high-flow lesion.

If the anomaly is a low-flow lesion, arteriovenous malformation, arteriovenous fistula, and hemangioma can be excluded from the differential diagnosis. Low-flow vascular anomalies (venous malformation, lymphatic malformation, capillary-lymphatic-venous malformation) can be further differentiated on the basis of their morphologic appearances and contrast-enhancement patterns. If the anomaly has no contrast enhancement or a minimal degree of peripheral contrast enhancement (rings and arcs), lymphatic malformation should be considered foremost in the differential diagnosis. If the anomaly has easily noticeable patchy areas of contrast enhancement, venous malformation should be suspected. If the lesion is a high-flow anomaly,

In hemangiomas, fast-flow vessels are usually at the periphery of the mass, and the mass usually enhances homogeneously. A mass lesion is not expected in an arteriovenous malformation. If there are any remaining questions, the high-flow nature of an arteriovenous

malformation can be easily confirmed with Doppler examination, which reveals high-flow, low-resistance arteries and an arterialized waveform in the draining veins.

Lymphatic malformations generally are documented at birth. The microcystic variant, or lymphangioma, presents as clusters of vesicles on the buccal mucosa, tongue, and conjunctiva. The vesicles can be clear, red or black as a result of microscopic bleeding. Macrocytic lesions, or cystic hygromas, are often located below the level of the mylohyoid muscle and present as cervical cystic swelling, often with the overlying skin having a bluish hue. Microcystic forms tend to be associated with adjacent bone and soft tissue hypertrophy. CT Imaging reveals an isodense mass that obscures tissue planes. Macrocytic forms are less invasive and appear as cystic structures with sharp demarcations of loculations and ring enhancement. Diagnosis of these malformations can be aided by staining for vascular endothelial growth factor receptor 3, which is found within the endothelial cells.

Treatment

Lymphatic malformations are treated with either sclerotherapy or surgical resection. Drainage of these lesions results only in temporary shrinkage. Sclerotherapy is accomplished with ethanol, sodium tetradecyl sulfate, doxycycline, or OK-432 (a killed strain of group A *Streptococcus pyogenes*) Macrocytic lesions are ideally removed in one procedure, because repeated excisions are complicated by fibrosis and anatomic distortion. Microcystic lesions are often difficult to resect, because there are no distinct tissue planes between the malformed and normal structures. Repeated procedures are necessary, and complete removal is almost impossible. In planning such a procedure, restrictions should be set for the extent of dissection. The most common complication of resection in the neck is nerve palsies.

Venous Malformations

Venous malformations (VM) account for 2/3 of all vascular malformations. There are designated as low-flow lesions. Venous malformations are present at birth, grow proportionately with the child, and often enlarge during puberty. VMs present in a spectrum, ranging from an isolated skin varicosity or localized spongy mass to complex lesions infiltrating various tissue planes. They are common in the skin and subcutaneous tissue of the head and neck region. They may present in skeletal muscle, most commonly in the intramasseteric area. VMs may also occur in the craniofacial skeleton, and are most commonly in the mandible, less frequently in the maxilla, and rarely in the nasal and cranial bones.

Diagnosis

Mandibular venous anomalies can present with increased mobility of the teeth, expansion of the buccal cortex, or spontaneous bleeding. The overlying skin may be normal, or it may exhibit a bluish tinge caused by involvement of the dermis. Intraorbital VMs cause expansion of the orbital cavity and can cause exophthalmia when the head is dependent and enophthalmia when the patient is upright. The VM is a soft, compressible nonpulsatile mass with rapid refilling. Expansion will occur on compression of the jugular vein or Valsalva's maneuver or with the head in a dependent position. Sluggish flow and stasis lead to phlebothrombosis, which presents clinically as recurrent pain and tenderness. Phleboliths can appear in patients as young

as 2 years of age. Characteristic phleboliths can be palpated and seen on radiographic examination.

The plain radiographic appearance of an intraosseous VM demonstrates a localized hypolucency with a honeycombed or *soap bubble* appearance. Profile or tangential films show spicules of bone radiating in a sunburst pattern. MRI is the most useful radiographic study soft tissue VM. They are T₂-hyperintense lesions and differ from LMs by the presence of contrast enhancement of the contents of the vascular spaces. Phleboliths or thrombi can be seen as signal voids. Stagnation within the VM causes a localized intravascular coagulopathy.

Treatment

Treatment of VM is based on the location, appearance, and complications such as pain, bleeding, and associated functional problems. The treatment options are sclerotherapy and resection. Sclerotherapy is potentially dangerous and requires the skills of an experienced interventional radiologist. A small cutaneous or oral mucosal VM can be injected with an agent such as sodium tetradecyl sulfate; for larger VMs, ethanol (100%) is used. Often, multiple sclerotherapy sessions are needed because of the propensity for recanalization and recurrence. Local complications include blistering, full-thickness cutaneous necrosis, and nerve damage. More severe and systemic complications include hemolysis, renal toxicity, and cardiac arrest. Numerous papers have been published concerning sclerotherapy for venous malformations. In 2005 Boll published a study in *Radiology*, describing MR-guided sclerotherapy of low flow vascular malformations with ethanolamine. In this study, patients with lesions of the head and neck presented with chief complaint of either bleeding into oral cavity or cosmetic deformity.

After therapy, all patients reported resolution of bleeding and an improvement in cosmetic appearance. In this study of 15 patients, no severe complications—such as skin necrosis, neuropathy, muscle atrophy and contracture, deep venous thrombosis, pulmonary embolus, disseminated intravascular coagulation, or cardiopulmonary collapse—were observed. Surgical resection is indicated, usually after completion of sclerotherapy for large or symptomatic VMs. Under most circumstances, total extirpation is impossible, and a subtotal resection is indicated to reduce bulk and improve contour, function, or relieve pain. VMs of the jaw, nasal bones, and zygoma are managed by curettage and packing with a hemostatic agent.

Capillary Malformations

Capillary malformations are present at birth and change slowly with time to a purple color in adulthood. CMs are often associated with hypertrophy of the soft tissue and underlying skeleton and when in the cervicofacial area may be associated with enlargement of the affected lip, gingiva, maxilla, and mandible. Skeletal overgrowth may not be obvious at birth, but progress in childhood. Patients with capillary staining of the ophthalmic (V1) and maxillary (V2) dermatome. may have Sturge-Weber syndrome, with capillary, venous, and arteriovenous anomalies of the leptomeninges.^[46] This anomalous circulation is responsible for the progressive degeneration and atrophy of the cerebral hemispheres causing seizures, contralateral hemiplegia, and delayed motor and cognitive skills. There is also an increased risk for retinal detachment, glaucoma, and blindness associated with choroidal vascular abnormality. Fundoscopic

examination and tonometry are essential in the evaluation of these patients and throughout their care.

Treatment

Tunable flashlamp pulsed-dye laser (585-nm wavelength) pulsed dye laser is widely regarded as the optimum treatment for these disfiguring lesions. An overall improvement with lightening of the stain and flattening of the area is expected in approximately 70% of patients. Theories exist on reasons for persistence of lesions after laser thermolysis. In 2005, Sivarajan et al. published a study investigating changes in capillary depth and diameter, within lesions, that occur with laser therapy. Their findings show that persistent vessels in capillary malformations after treatment are deeper and narrower than those in untreated lesions. The authors of this study suggest that since depth and diameter are crucial to the most effective wavelength and pulse duration, respectively, of a therapeutic laser, adjusting these laser parameters for treating resistant lesions may be effective. In selected cases, older patients, there may be a consideration for excision in the esthetic facial units. The potential problems after excision and grafting include scarred hypertrophy at the junction of the graft and normal skin and unpredictable pigmentation within the skin graft itself. Soft tissue and skeletal hypertrophy often require surgical correction. Orthognathic and orthodontic procedures are required for maxillary and mandibular overgrowth.

Arteriovenous Malformation

Arteriovenous malformation (AVM) is usually noted in birth, but is rarely symptomatic during infancy. Many lesions have a warm erythematous blush and can be mistaken for "hemangioma" or mislabeled as a "port-wine stain." Local infection, trauma, hormonal changes, and puberty may trigger expansion and often manifest during childhood, adolescence, or even adulthood. Local warmth, thrill, and a bruit confirm the diagnosis. Shunting of blood diminishes nutritive flow, which may result in skin necrosis, ulceration, bone destruction, and bleeding. The patient often seeks management for swelling, pain, or sudden hemorrhage. The diagnosis is confirmed by ultrasonography and color Doppler examination. MRI and magnetic resonance angiography (MRA) are used to assess the extent of the lesion and involvement of vital structures. The natural history of AVMs is documented by a clinical staging system introduced by Schobinger: Stage I (quiescence), Stage II (expansion), Stage III (destructive), Stage IV (decompensation).

Treatment

Rarely is treatment indicated for an asymptomatic AVM. Once the diagnosis is made, the child should be closely followed every 6 months or yearly. However, in rare instances and after careful consideration, resection may be performed for a well-localized, Stage I AVM. Often intervention should be delayed until there are signs and symptoms of pain, bleeding, ulceration, infection, or concern for endangering vital structures (Schobinger stage II–III). There is no place for ligation or proximal embolization of feeding vessels. This will lead to rapid recruitment of flow from nearby arteries and denies access for embolization. Superselective arterial or retrograde venous embolization may have a role in palliation, or it may be used as primary therapy for surgically inaccessible AVM. The only therapy that carries any hope for long-term

success is total resection of the tissue involved with the AVM. Leaving behind residual and dormant anomalous channels only invites further collateral formation, shunting, and expansion.

Preoperative superselective embolization will not diminish the extent of the resection. However, it will minimize intraoperative bleeding. Embolization must be in the nidus, or epicenter, of the AVM and is carried out 24 to 72 hours before the resection. Often a two-team approach (for resection and reconstruction) is useful for these lesions. The critical decision is how extensive the resection must be to include all of the involved tissue. Reconstruction often necessitates closure and soft-tissue replacement with microvascular tissue transfer. Given proper indications and with careful planning, extensive resection may be justified.

Conclusion

Much knowledge is now known about the radiographic appearance, natural history, and response to treatment of vascular malformations. This knowledge now allows clinicians, using a team approach to form appropriate treatment plans. Clinicians in private practice who suspect their patient has a vascular malformation should refer them to a tertiary center where a multidisciplinary team can treat the malformation successfully, while minimizing complications.

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