REVIEW

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An appropriate diagnostic workup for suspected vascular birthmarks

ABSTRACT

Birthmarks are common and commonly ignored by patients and primary care doctors. Yet they sometimes represent significant vascular anomalies that require diagnosis and treatment. We summarize when and how to work up a variety of vascular anomalies.

KEY POINTS

Knowing the vascular anomalies underlying birthmarks helps avoid missed or incorrect diagnosis or an extensive, unnecessary workup.

The history and physical examination provide enough information to make the diagnosis, but imaging is needed to evaluate the extent of deep tissue involvement and to help plan treatment.

Some arteriovenous malformations can progress rapidly, so the diagnostic workup should be completed as soon as possible.

BIRTHMARKS ARE common, but are commonly ignored by patients and primary care physicians. In most cases, this is fine. Yet birthmarks can sometimes represent significant vascular anomalies that require a diagnostic workup and treatment.

In this article, we summarize the features of potentially troublesome vascular anomalies and review the highlights of the diagnostic workup and treatment.

VASCULAR ANOMALIES CATEGORIZED

The naming of vascular anomalies has long been a confusing matter, probably contributing to inaccurate and untimely diagnosis and treatment. Mulliken and Glowacki[1] simplified the situation by classifying vascular anomalies biologically, as either hemangiomas (including congenital vascular tumors) or vascular malformations (TABLE 1).

Hemangiomas include infantile, congenital, noninvoluting, intramuscular, and kaposiform hemangioendothelioma, and each has a somewhat different presentation.

Vascular malformations are classified as high-flow or low-flow or a combination of the two. The distinction between high flow and low flow is important: high-flow anomalies are treated by transcatheter embolization, whereas low-flow lesions are treated by the percutaneous injection of sclerosing agents (sclerotherapy). Low-flow malformations include capillary, lymphatic, and venous types. High-flow malformations can be arterial or arteriovenous.

In this article, we focus on common hemangiomas and then on venous, lymphatic, arteriovenous, and combined vascular anomalies.

TABLE 1

A classification of vascular birthmarks

Hemangiomas and congenital vascular tumors
- Infantile
- Congenital
- Noninvoluting
- Intramuscular
- Kaposiform hemangioendothelioma

Vascular malformations
- High-flow
  - Arteriovenous malformation
  - Arteriovenous fistula
- Low-flow
  - Capillary malformation ("port-wine stain")
  - Venous malformation
  - Lymphatic malformation
- Combined
  - High-flow
    - Parkes-Weber syndrome: capillary, arterial, venous malformation in a limb, with limb overgrowth
  - Low-flow
    - Klippel-Trenaunay syndrome: capillary-lymphatic-venous malformation in a lower extremity with limb overgrowth
    - Maffucci syndrome: lesions resembling venous malformations, with enchondromatosis

HEMANGIOMAS

Hemangiomas tend to be small and to involve only the skin, and they involute (regress) without complications, leaving no sign or perhaps a small blemish.[2]

Typical features and course

The most common hemangioma is infantile hemangioma, which is easily recognizable by its appearance, as well as its patterns of growth and involution. It appears within the first few weeks of life, usually as a strawberry-like superficial lesion (FIGURE 1), and grows rapidly within the first year, reaching a plateau around the age of 1. After that, the hemangioma regresses until it gradually disappears in early childhood.[3–6]

Reassure concerned parents

Parents may find such lesions alarming and may push for treatment. In such cases, once the diagnosis is confirmed, they simply need to be reassured that the lesion will resolve, in most cases without a trace.[2]

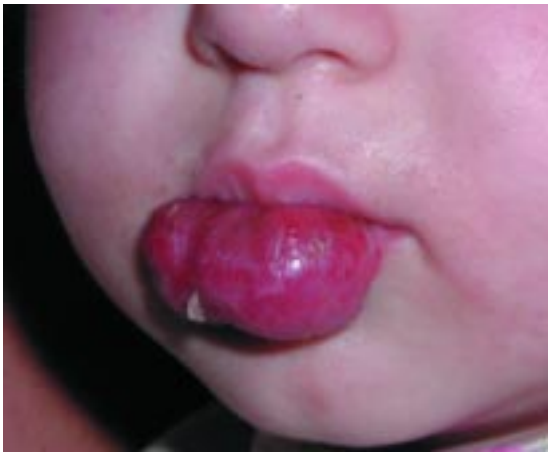

FIGURE 1. Typical infantile hemangioma in the lower lip. This is a soft mass that is not compressible. The diagnosis can be made without imaging in most of these patients.

Diagnosis

Diagnosis of hemangioma is based on the clinical presentation. No diagnostic imaging is indicated in most cases. In some patients, a hemangioma may resemble a "port-wine stain," a capillary malformation. However, a key difference is that a hemangioma regresses over time.[2]

Other situations may call for diagnostic imaging with magnetic resonance imaging (MRI) or, in some cases, Doppler ultrasonography,[7–10] performed by a radiologist experienced in vascular anomalies. MRI of hemangiomas helps differentiate them from other tumors and vascular malformations of infancy (TABLE 2). For example, extensive cervicofacial hemangiomas require MRI to rule out intracranial and intrathoracic abnormalities,[11,12] and suspected deep hemangiomas involving vital structures require MRI evaluation to confirm the type and extent of involvement.

Doppler ultrasonography is occasionally used to evaluate the nature of a hemangioma and to confirm the diagnosis.[13] Computed tomography (CT) and plain radiography have limited roles in hemangiomas.

Biopsy. For hemangiomas with atypical clinical or imaging presentation, biopsy may be indicated to rule out kaposiform hemangioendothelioma or malignancy.

TABLE 2

Typical imaging features of vascular anomalies

LESION	DIGITAL SUBTRACTION ANGIOGRAPHY	MAGNETIC RESONANCE IMAGING	DOPPLER ULTRASONOGRAPHY	COMPUTED TOMOGRAPHY
Hemangioma	Dilated arteries and veins; "cotton-wool" appearance	Homogeneous contrast enhancement; flow voids in and around the lesion	Solid mass with small arteries and veins in and around the mass	Uniformly enhanced mass with dilated vessels
Venous (low-flow) malformation	Opacification of the lesion during the venous phase	High T2 signal; opacification of the lesion with contrast; no flow voids	Compressible dilated venous spaces	Delayed opacification of the lesion; phleboliths
Lymphatic (low-flow) malformation	Lesion is avascular	High T2 signal; no or very minimal peripheral contrast opacification; no flow voids	Compressible dilated channels	Soft-tissue abnormality without contrast accumulation; no phleboliths
Arteriovenous (high-flow) malformation	Dilated arteries; nidus; early opacification of draining veins	Flow voids; no mass	Dilated arteries and veins, arterialized Doppler waveform in the draining veins	Dilated vessels in the area of the lesion

Other types

Less common hemangiomas include rapidly involuting congenital hemangioma, noninvoluting hemangioma, and intramuscular hemangioma. These usually present atypically and require further workup, including imaging or biopsy.[8,14,15]

Treatment

In most cases, hemangiomas require no treatment. However, 1 out of 10 patients may have a problematic lesion: eg, it causes marked distortion or interferes with vision, breathing, eating, or other normal functions.[2] If so, treatments are available, including steroids, embolization therapy, excision, and laser therapy.

VENOUS MALFORMATIONS

Venous malformations are generally present at birth and can present clinically as bluish skin discolorations and soft-tissue swelling (FIGURE 2). They are soft, easily compressible, and typically demonstrate engorgement when the body part involved is lower than the heart. Plain radiography usually shows venous calculi or concretions (phleboliths) within the involved soft tissues.

Diagnosis

CT has a limited role in the diagnosis of venous malformations (FIGURE 3). MRI is used to confirm the diagnosis and to evaluate the extent of the lesion for treatment planning. Because venous malformations are usually recognizable by their clinical presentation and appearance on imaging, biopsy is not usually required for diagnosis.

Treatment

Sclerotherapy is the first-line treatment for most venous malformations. There are a number of ways to apply the therapy and a number of sclerosing agents available. Factors that influence the approach include the location of the lesion (eg, whether it is close to a major nerve bundle) and the pattern of venous drainage.

Most hemangiomas resolve, leaving little or no mark

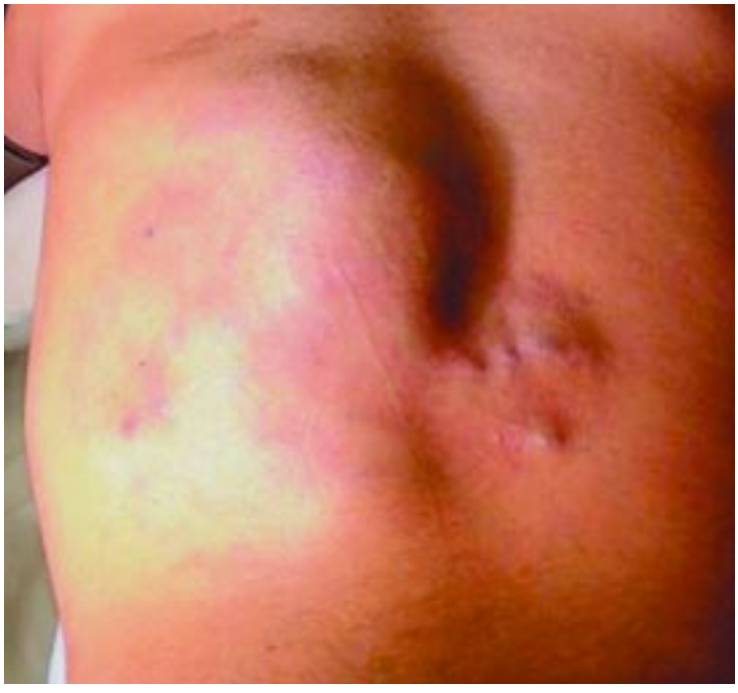

FIGURE 2. A large venous malformation in the posterior-lateral chest wall appears as a soft-tissue prominence with bluish skin color change. It was not pulsatile or sensitive on palpation and was easily compressible. Diagnosis was based on the clinical presentation alone.

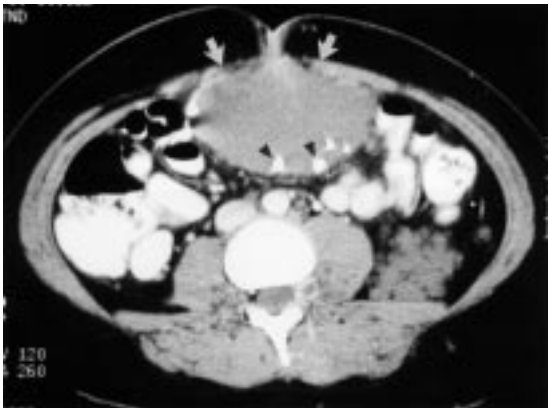

FIGURE 3. Axial computed tomography (CT) shows an intra-abdominal venous malformation that appears as an ovoid mass (arrows) anterior to the small bowel loops, with several phleboliths (calculi) (arrowheads). CT is not able to differentiate between the types of venous malformations. In this example, the multiple phleboliths are a defining feature; otherwise, the CT is not diagnostic.

MRI can be used to confirm the diagnosis or to guide therapy

LYMPHATIC MALFORMATIONS

Lymphatic malformations (the result of lymphatic vessel dysplasia) are typically noted at birth, grow gradually, and may or may not be associated with typical skin or mucosal vesicles (FIGURE 4). They are only rarely life-threatening.[2] They usually present as an easily compressible soft-tissue prominence with or without skin involvement. Lymphatic malformations can involve any part of the body, but they commonly occur in the head and neck. They may become more prominent and painful with infections.

These lesions tend to be either microcystic—which are the most common and were once referred to as "lymphangiomas"—or macrocystic (former name "cystic hygroma").[2]

Diagnosis

On physical examination, lymphatic malformations usually do not show engorgement even when lower than the heart. The skin may show signs of cellulitis and lymphangitis, with redness, pain, and swelling.[2]

MRI is commonly used to evaluate the tissue characteristics of the lesion (microcystic vs macrocystic) (FIGURE 5), which helps determine the type of therapy: macrocystic lymphatic malformations can be treated with sclerotherapy, while microcystic lesions can only be treated surgically.

Tissue sampling is usually not necessary, as the clinical presentation and MRI evaluation confirm the diagnosis.

ARTERIOVENOUS ANOMALIES

Arteriovenous malformations and arteriovenous fistulas may be recognized as focal, pulsatile soft-tissue abnormalities with associated bruit or continuous murmur (FIGURE 6). They are usually noticed in early childhood and grow with the child into adulthood, demonstrating more rapid change during growth spurts, at puberty, and after trauma, pregnancy, or surgery. They may present with high-output cardiac failure in infancy, but most evolve gradually, becoming symptomatic in late childhood or early adulthood. Ischemia caused by arterial "steal"—ie, arterial blood passing into the venous system via the lesion, before it reaches normal tissues—and venous hypertension can lead to pain, skin breakdown, and bleeding.

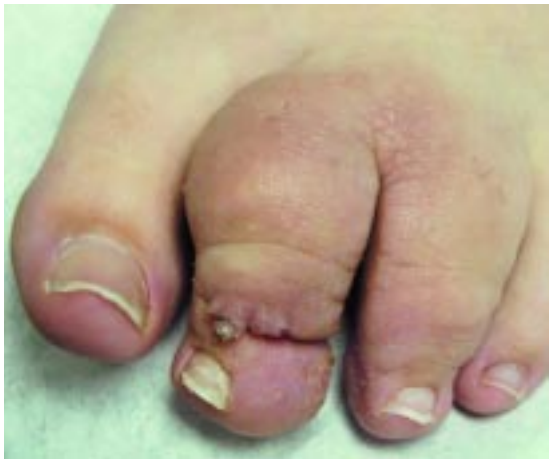

FIGURE 4. A lymphatic malformation appears as a soft-tissue mass on the foot with typical skin changes. Most of these lesions are not compressible on palpation. MRI confirms the diagnosis and, more importantly, identifies the morphologic type (microcystic vs macrocystic), which guides therapy.

Diagnosis

Although most arteriovenous malformations can be recognized clinically, MRI confirms the diagnosis, shows the extent of soft-tissue involvement, and provides a map of the feeding arteries and draining veins that can be used to guide embolization therapy.

In patients with suspected arteriovenous malformations, the diagnostic workup needs to be completed as soon as possible, because the problem may progress rapidly, making embolization therapy much more difficult.

Conventional arteriography is usually not used for the diagnosis of arteriovenous malformation. However, if a lesion is confirmed and embolization is selected as the treatment, arteriography is used to guide embolization therapy.

CT arteriography has improved significantly in recent years with the introduction of multislice CT scanners. The technique currently has great potential to evaluate the vascular architecture and should be expected to eventually replace MRI in the evaluation of these high-flow vascular malformations.[8]

Biopsy of an arteriovenous malformation may cause significant bleeding and should be avoided whenever possible.

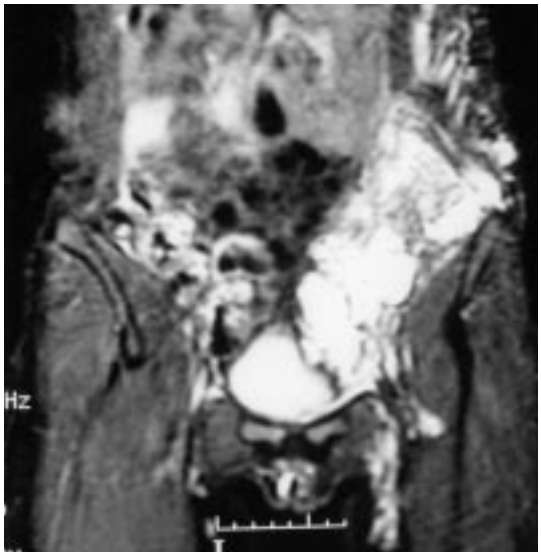

FIGURE 5. A lymphatic malformation in the pelvis as seen with T2-weighted coronal MRI. Note the large, hyperintense lesion in the left pelvic area involving the anteriolateral abdominal wall, consistent with a macrocystic lymphatic malformation. This lesion was treated with sclerotherapy.

COMBINED VASCULAR ANOMALIES

Combined vascular anomalies should undergo MRI to confirm the diagnosis, to evaluate the tissue characteristics of the lesion, and to search for additional occult abnormalities (**TABLE 2**). Among these conditions, Klippel-Trenaunay syndrome deserves special attention due to its relatively high incidence and typical clinical presentation. The deep venous system in the involved extremity needs to be evaluated for patency before any surgical intervention can be performed, so that the patient will not have venous drainage problems after removal of the abnormal yet functional superficial veins.

MRI is the ideal way to simultaneously evaluate the soft tissues and the vascular anatomy in these patients.[10] The other advantage of this test is to rule out Parkes-Weber syndrome, which is distinguished from Klippel-Trenaunay syndrome by the presence of arteriovenous shunts. Differentiation is important, because Klippel-Trenaunay syndrome generally requires sclerotherapy, where-

Avoid biopsy of arteriovenous lesions, as it can cause significant bleeding

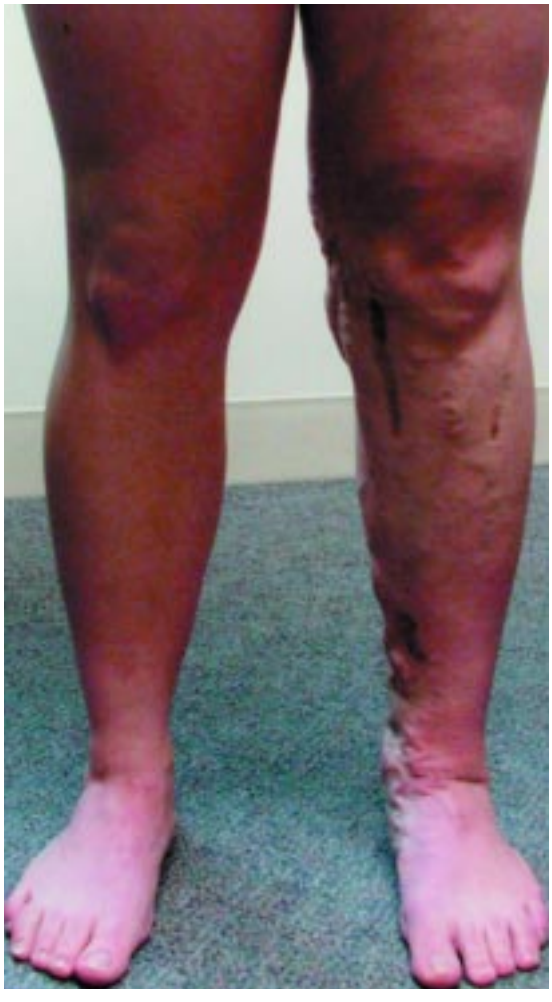

FIGURE 6. An extensive arteriovenous malformation in the left leg. The left leg appears larger than the right leg, with multiple large varicoid venous abnormalities and skin changes. A thrill was palpable over the varicosities due to rapid arteriovenous shunt. MRI confirmed the diagnosis and evaluated the extent of the lesion. Magnetic resonance angiography provided information about the vascular architecture of the lesion. This patient underwent transcatheter embolization.

as Parkes-Weber syndrome requires transcatheter embolization for treatment. Also, in patients with suspected Sturge-Weber syndrome (facial port-wine stain with leptomeningial vascular anomaly), MRI is used to evaluate possible intracranial vascular anomalies.

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