Discrepancy between the clinical and histopathologic diagnosis of soft tissue vascular malformations

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Background: Soft tissue vascular malformations are generally diagnosed clinically, according to the International Society for the Study of Vascular Anomalies (ISSVA) classification. Diagnostic histopathologic examination is rarely performed.

Objective: We sought to evaluate the validity of the current diagnostic workup without routinely performed diagnostic histopathology.

Methods: We retrospectively determined whether there were discrepancies between clinical and histopathologic diagnoses of patients with clinically diagnosed vascular malformations undergoing therapeutic surgical resections in our center (2000-2015). Beforehand, a pathologist revised the histopathologic diagnoses according to the ISSVA classification.

Results: Clinical and histopathologic diagnoses were discrepant in 57% of 142 cases. In these cases, the pathologist indicated the diagnosis was not at all a vascular malformation (n = 24; 17%), a completely different type of vascular malformation (n = 26; 18%), or a partially different type with regard to the combination of vessel-types involved (n = 31; 22%). Possible factors associated with the discrepancies were both clinician-related (eg, diagnostic uncertainty) and pathology-related (eg, lack of immunostaining).

Limitations: Retrospective analysis of a subgroup of patients undergoing surgery.

Conclusion: The large discrepancy between clinical and histopathologic diagnoses raises doubt about the validity of the current diagnostic workup for vascular malformations. Clear clinical and histopathologic diagnostic criteria might be essential for a uniform diagnosis. (J Am Acad Dermatol http://dx.doi.org/10.1016/j.jaad.2017.03.045.)

Key words: clinical; dermatopathology; diagnosis; histology; histopathology; ISSVA; vascular anomalies; vascular malformations.

In 1982, Mulliken et al[1] proposed a classification for vascular anomalies that separated vascular malformations from infantile hemangiomas and other vascular tumors on the basis of histologic endothelial characteristics, natural history, and physical findings. Since then, vascular malformations were recognized as abnormally developed vessels, a distinct entity with treatment options and prognoses different from vascular tumors.[2]

Since 1996, the International Society of the Study for Vascular Anomalies (ISSVA) has been providing a more specified, up-to-date classification of vascular

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anomalies.[3-6] In this classification, vascular malformations are further categorized based on blood flow characteristics (high flow versus low flow) and the type of vessels included (Table I), which can be veins (venous malformations [VM]), arteries (arteriovenous malformations [AVM] or arteriovenous fistulas), lymphatic vessels (lymphatic malformations [LM]), capillaries (capillary malformations [CM]) or a combination of these vessel-types.[5] These different vascular malformation types have different clinical presentations, therapeutic options, and presumably different responses to treatment.[3] Hence, a correct diagnosis is essential, not only to distinguish vascular tumors from malformations but also to diagnose vascular malformation subtypes.

CAPSULE SUMMARY

- Diagnostic histopathologic examination is not routinely performed in the diagnostic workup for patients with vascular malformations.
- In more than half of patients undergoing therapeutic surgical resections, the clinical and histopathologic diagnosis did not correspond.
- There is a need for clear diagnostic criteria for clinicians and pathologists.

In the current diagnostic workup, diagnoses are generally made clinically based on natural history and physical examination, usually combined with radiologic imaging.[7-9] Diagnostic histologic biopsies are only performed in atypical clinical cases, particularly when a malignant tumor is considered in the differential diagnosis. Because vascular malformations are benign lesions, clinicians tend to avoid invasive diagnostic procedures that could have harmful sequelae, like bleeding and scarring.

However, it is unknown whether routinely performed histopathologic examination is indeed unnecessary in the diagnostic workup. Hypothetically, if clinical and histopathologic diagnoses of vascular malformations are (nearly) identical, the current diagnostic workup (without the use of histopathology) should suffice. In the present study, we therefore determined if clinical and histopathologic diagnoses corresponded in patients with clinically diagnosed vascular malformations undergoing therapeutic surgical resections. To elucidate the reasons for a potential discrepancy, we identified the variables associated with discrepancy between the clinical and histopathologic diagnosis.

METHODS

Study design

We conducted a retrospective clinicopathologic evaluation in a vascular anomaly expert center. The institutional review board exempted this study from ethics approval and waived the need for informed consent.

Patient selection

Eligible patients were identified through operation room planning schedules (2000-2015). All patients with clinically diagnosed vascular malformations of skin or soft tissue, according to the ISSVA classification, who underwent surgical resections for therapeutic purposes were included. Although these surgical resections were nondiagnostic, the surgeon routinely sent the resected tissue for histopathologic examination for research purposes. Patients undergoing surgical resections or biopsies for purely diagnostic purposes were excluded.

Clinical diagnosis

Patients with suspected vascular malformations were evaluated by a multidisciplinary team consisting of a dermatologist, a plastic surgeon, an interventional radiologist, and a vascular surgeon who have been using the ISSVA classification for guidance in the diagnostic procedure since 1996. Clinical diagnoses were based on natural history and physical examination, usually in conjunction with radiologic imaging (Fig 1). Imaging is primarily performed to assess the lesion extensiveness and flow characteristics and might be omitted in superficial lesions. For this study, we used the clinical diagnosis as stated in the preoperative letter from the outpatient clinic and the histopathology request form.

Histopathologic diagnosis

The initial histopathologic diagnoses, as stated in the initial pathology reports, were reported by many different pathologists over the years and were presumably not all made using the ISSVA terminology.

Therefore, a revision of histopathologic diagnoses was performed by a pathologist (Dr A. C. van der Wal) who is subspecialized in vascular pathology, based on the latest ISSVA classification by Wassef et al[5] (Table I). This pathologist, blinded for the clinical diagnosis and the initial histopathologic diagnosis, re-examined all tissue sections. For the revision procedure, tissue sections of all cases were retrieved from the pathology archive. For all cases, hematoxylin-eosin and Elastic van Gieson stainings were available. Immunohistochemistry stainings, performed at the discretion of the initial handling pathologist, were also available in selected cases: glucose transporter 1 to exclude infantile