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RESEARCH ARTICLE

Concordance between referral and final diagnoses of pediatric patients with vascular malformations

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Abstract

Background: *Vascular malformations (VaM) are a heterogeneous group of disorders resulting from the dysmorphogenesis of blood vessels. Although correct classification is relevant to providing adequate treatment according to evidence-based medicine, diagnostic terminology may be misused or need clarification.* **Methods:** *We conducted a retrospective study to measure agreement and concordance between referral and final confirmed diagnoses of 435 pediatric patients with VaM newly referred to the multidisciplinary Vascular Anomalies Clinic (VAC) using Fleiss kappa (κ) concordance analysis.* **Results:** *We found fair concordance between referral and confirmed diagnoses of VaM (κ 0.306, $p < 0.001$). Lymphatic malformations (LM) and VaM associated with other anomalies showed moderate diagnostic concordance (κ 0.593, $p < 0.001$ and κ 0.469, $p < 0.001$, respectively).* **Conclusions:** *Continuing medical education strategies are required to improve physician knowledge and diagnostic accuracy in patients with VaM.*

Keywords: *Vascular malformation. Diagnosis. Clinical decision-making. Diagnostic errors. Pediatrics.*

Concordancia entre el diagnostico de referencia y final de pacientes pediátricos con malformaciones vasculares

Resumen

Introducción: *Las malformaciones vasculares (MVa) son un grupo heterogéneo de trastornos resultantes de la dismorfogénesis de los vasos sanguíneos. A pesar de que la correcta clasificación es relevante para brindar un adecuado tratamiento de acuerdo con la medicina basada en la evidencia, la terminología diagnóstica podría resultar confusa o utilizarse de manera inapropiada.* **Métodos:** *En este estudio retrospectivo se midieron el acuerdo y la concordancia entre los diagnósticos de referencia (o derivación) y los diagnósticos finales confirmados de 435 pacientes pediátricos con MVa recién remitidos a la Clínica de anomalías vasculares (CAV) multidisciplinaria, mediante el análisis de concordancia kappa de Fleiss (κ).* **Resultados:** *Se encontró una buena concordancia entre los diagnósticos de referencia (o derivación) y los diagnósticos confirmados de MVa (κ 0.306, $p < 0.001$). Las malformaciones linfáticas (LM) y las MVa asociadas con otras anomalías presentaron concordancias diagnósticas moderadas (κ 0.593, $p < 0.001$ y κ 0.469, $p < 0.001$, respectivamente).* **Conclusiones:** *Se requiere de estrategias de educación médica continua para mejorar el conocimiento de los médicos y la precisión diagnóstica de los pacientes con MVa.*

Palabras clave: *Malformaciones vasculares. Diagnóstico. Toma de decisiones clínicas. Errores de diagnóstico. Pediatría.*

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Introduction

Vascular malformations (VaM) are a heterogeneous group of disorders with endothelial cell dysmorphogenesis, classified as (1) simple, (2) combined, (3) of major named vessels, and (4) associated with other anomalies[1]. Their correct classification is relevant to provide adequate treatment according to evidence-based medicine. The International Society for the Study of Vascular Anomalies (ISSVA) periodically updates this classification to standardize terminology, including new genetic and clinical findings. However, physicians who are not familiar with these patients may need more clarification with diagnostic terminology to prevent inadequate evaluation, management, and prognosis[1-3].

This study aimed to measure the concordance between referral diagnoses of new pediatric VaM patients sent to the Vascular Anomalies Clinic (VAC) and the final confirmed diagnoses.

Methods

We conducted a retrospective study of 435 patients referred between 2012 and 2022 to our institution's Vascular Anomalies Clinic (VAC). First, we reviewed the diagnoses provided at the time of referral and then those established by our team according to the ISSVA after evaluating clinical, imaging, and histological findings. We used descriptive statistics and Fleiss kappa (κ) concordance analysis with the following interpretation: ≤ 0 indicated no concordance; 0.01-0.20, none to mild; 0.21-0.40, fair; 0.41-0.60, moderate; 0.61-0.80 substantial; and 0.81-1.00, near perfect concordance.

Results

Fifty-one percent of patients were female, with a mean age at initial diagnosis of 50.31 months (standard deviation (SD) ± 51.86). Simple VaMs were observed in 300 patients: 162 with lymphatic malformations (LM), 82 with venous malformations (VM), 46 with arteriovenous malformations (AVM), and ten with capillary malformations (CM). Combined VaMs were observed in 109 patients: 107 with slow-flow (CM ± LM ± VM) and two with high-flow. VaMs associated with other anomalies were identified in 23 patients, and provisionally not classified conditions were observed in three patients.

Fifty-eight percent of patients were correctly diagnosed with VaM. The rest of the patients were misdiagnosed, as they had a different condition: vascular tumor (17.4%), neoplasm (11.7%), cyst/abscess/infection (5.4%), and others (9.4%). Patients were referred by pediatricians (146), surgeons (146), dermatologists (61), and primary care physicians (30). Dermatology (62.2%), neonatology (50%), oncology (50%), surgery (42.8%), and pediatrics (38.3%) most frequently diagnosed patients correctly.

When analyzed by specific VaM subtypes, we found 53% of LM, 30.4% of VaM associated with other anomalies, 23.9% of AVM, 20% of CM, and 10.9% of VM were correctly diagnosed. Overall, the diagnostic concordance between referral and final diagnoses was fair (κ 0.306, standard error (SE) 0.064, $p < 0.001$); for LM and VaM associated with other anomalies, it was moderate (κ 0.593 and κ 0.469, respectively) (Figure 1).

Discussion

Suboptimal diagnostic concordance of VaM impairs communication, treatment, and research in this field[3]; a low yield of correct referral diagnoses of VaM has been reported. Greene et al. found that 45.6% of 3,645 patients were labeled correctly at referral, particularly LM (69.3%) and AVM (59.4%)[4]. LMs are frequent VaM[5] and, together with AVM, have distinctive clinical features, which may explain why they can be more frequently correctly identified. Only LM and VaM associated with other anomalies had moderate diagnostic concordance in our study.

Low diagnostic consistency has been reported even at centers with VACs. For example, Pahls et al. reported a concordance of 9%, reflecting physicians' lack of familiarity or resistance to adopting ISSVA terminology rather than insufficient knowledge[6]. Additionally, the correct diagnosis of VaM may be more problematic than tumors because they are less common, and physicians should be familiar with the full spectrum[4]. Vascular anomalies are only sometimes taught to medical students. Moreover, residents and specialty practitioners who care for these patients receive limited exposure and training[7].

The main problem of misdiagnosis is the impact on patient care. For example, patients with VaM may be misdiagnosed as infantile hemangiomas and prescribed ineffective medical treatment (Figure 2A) or diagnosed with neoplastic diseases and consequently treated with improper procedures (Figure 2B).

The following ways to fill this niche are proposed:

– Facilitating trainees' exposure to specialized multidisciplinary groups/clinics that care for patients with vascular anomalies, as they are the cornerstone of proper training.

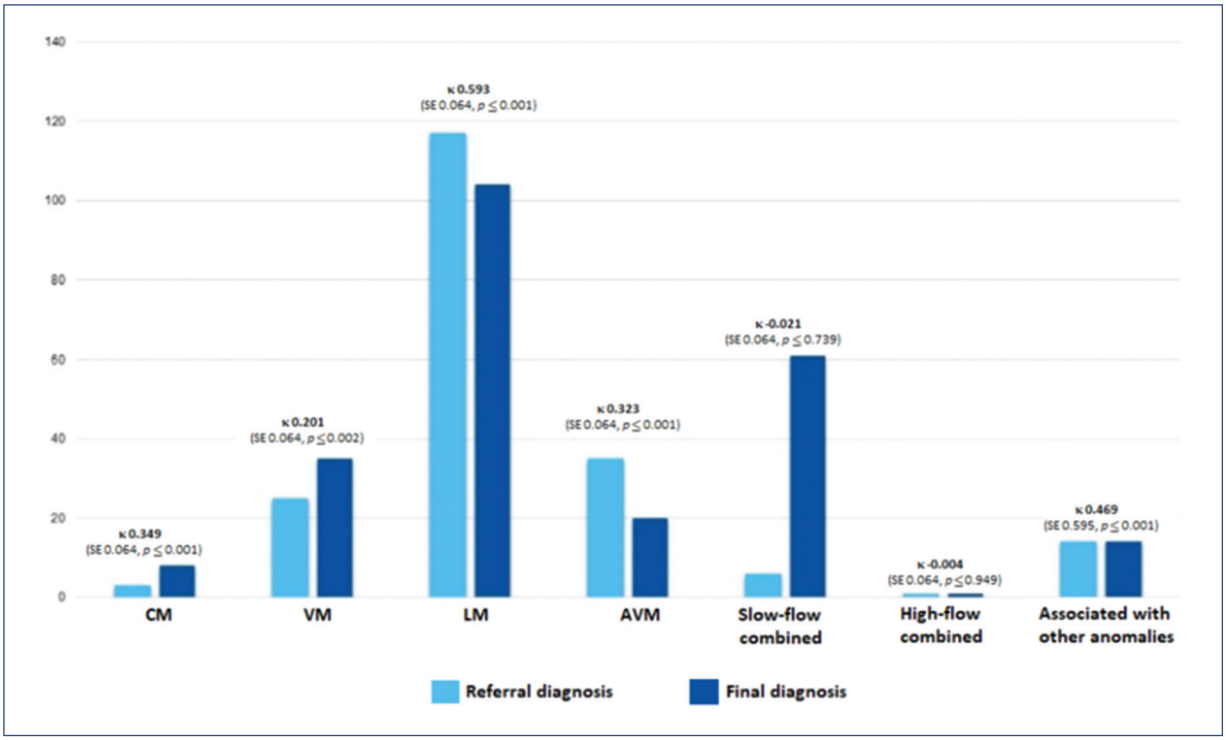

Figure 1. Fleiss kappa (κ) concordance analysis between referral and final diagnoses of patients with vascular malformations (VaM). AVM, arteriovenous malformations; CM, capillary malformations; LM, lymphatic malformations; SE, standard error; VM, venous malformations.

Figure 2. A. Patient with a venous malformation (VM) from birth on the lower lip that was diagnosed as a nevus. At 3 years of age, the lesion increased in size and volume and became painful. She was diagnosed with a hemangioma and treated with propranolol and topical corticosteroids for 6 months. Due to a lack of response, she was referred to the Vascular Anomalies Clinic (VAC), where she was successfully treated with polidocanol sclerotherapy. B. Patient with retroorbital venous-lymphatic malformations (VLM) observed at 2 years of age due to volume increase and proptosis after trauma. Computerized tomography findings suggested the probable diagnosis of alveolar rhabdomyosarcoma. A biopsy was performed, complicated by hemorrhage requiring drainage and tarsorrhaphy. Once diagnosed with VLM, the patient was referred to the VAC. Unfortunately, by this time, we found severe vision loss.

– Specialties involved in the care of these patients should advocate for this topic to be part of their conferences and journals.
– Development and distribution of specific guidelines or pathways, such as the European Vascular Anomalies (VASCA) Working Group[8].
– Inclusion of vascular anomalies as part of the dermatology curriculum for medical students.

Limitations of this study include the retrospective design and the limited number of patients with some types of VaM. However, we used the κ coefficient, considered the gold standard for measuring concordance by excluding random coincidences.

In conclusion, we found fair concordance between referral and final diagnoses of VaM. Continuing medical education strategies are necessary to improve adequate diagnoses and, therefore, the management of these patients.

Ethical disclosures

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this study.

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors have obtained the written informed consent of the patients or subjects mentioned in the article. The corresponding author has this document.

Conflicts of interest

The authors declare no conflicts of interest.

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