Stadtspital Zürich

When Common Diagnoses Fail: A Challenging Case of Arteriovenous Malformation of the Thumb

Wessmann S, Dietrich M, Sproedt J
Stadtspital Zürich, Klinik für Orthopädie, Hand- und Unfallchirurgie, Zürich

Introduction

Arteriovenous malformations (AVMs) are rare congenital vascular anomalies, accounting for approximately 5 to 613 per 100.000 of the population[2]. They arise from errors in embryonic vascular development, leading to direct artery-to-vein shunts. Although present from birth, AVMs often remain clinically silent during childhood and become apparent later in life due to hormonal influences, trauma, infection, or progressive vessel enlargement[2]. Regarding the hand, AVMs are particulary uncommon and their presentation is often misleading, mimicking inflammatory or infectious disorders. In our patient, first symptoms occurred in adulthood, highlighting how AVMs may stay undetected until growth accelerates or external triggers provoke symptomatic progression.

Case Presentation

A 24-year-old healthy female presented with persistent pain, swelling, and warmth of the right thumb. Initial treatment for suspected cellulitis failed, and MRI showed soft tissue edema. At our institution, ultrasound showed signs of tenosynovitis (Figure 2). She was then referred to an external rheumatology, where CRPS was excluded. A subsequent MRI confirmed flexor pollcis longus (FPL) tenosynovitis, prompting corticosteroid infiltration of the flexor tendon sheath. Angiological assessment showed no venous outflow obstruction or arterial abnormalities. Over time, symptoms gradually migrated proximally. Clinically, a superficially pulsating artery became evident (Figure 1), prompting targeted ultrasound, followed by MR angiography. This was the patient´s third MRI, but the first with angiographic sequences, which finally revealed a high-flow arteriovenous malformation (AVM) of the palmar thumb (Figure 3). Following the decision of the Vascular Anomalies Board, an external biopsy confirmed the diagnosis and identified a somatic BRAF mutation. An interventional radiology review at Universitätsspital Zürich in July 2025 found no current indication for an invasive procedure given her clinical status and stage; annual ultrasound follow-up is planned. Hematology consultation is arranged to evaluate candidacy for targeted therapy. Since then, our patient has transitioned from carpentry to office work and is preparing for vocational retraining. She reports moderate daily symptoms with localized tenderness, using a protective hand splint for activities ouside the home.

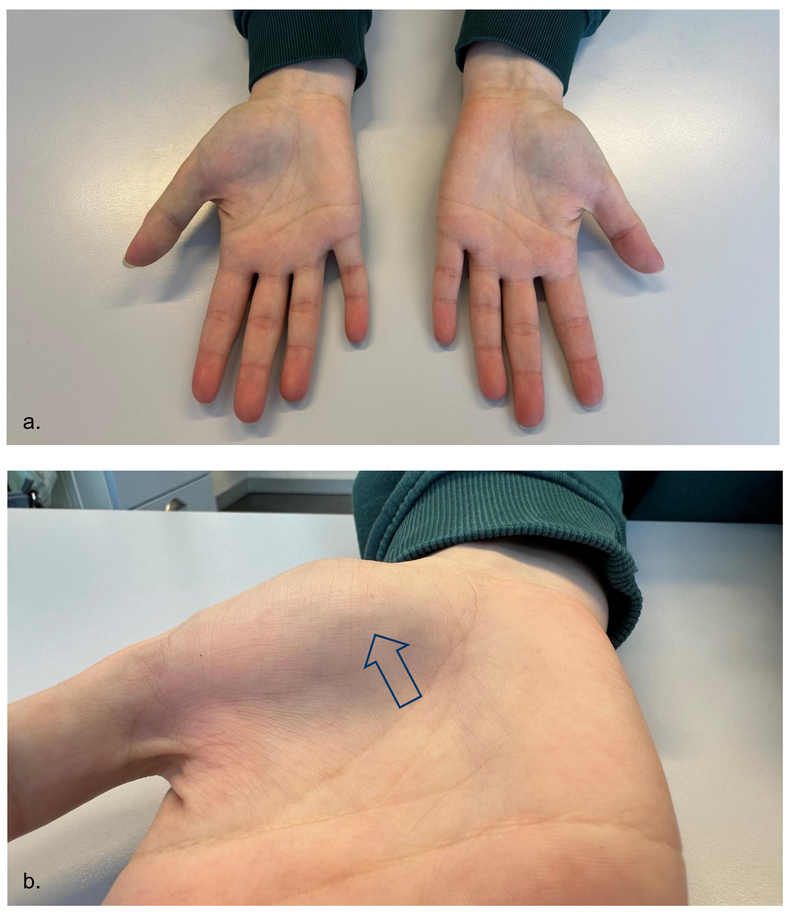

Figure 1
Clinical presentation of patient´s AVM
a. AVM in comparison to the left hand
b. Close-up of the right thenar

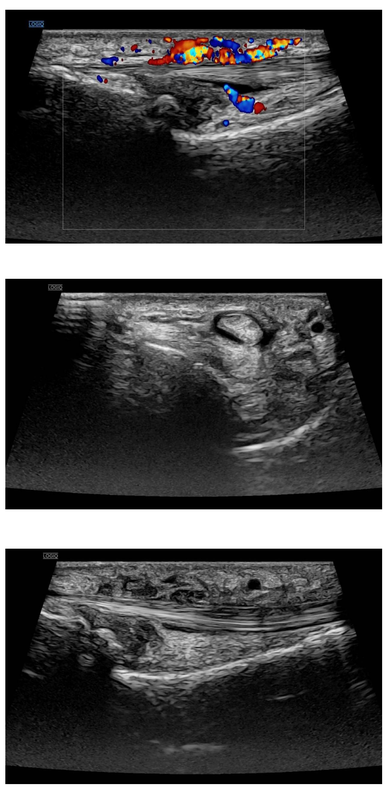

Figure 2
Initial ultrasound with signs of FPL tenosynovitis, mild joint effusion, and periarticular hypervascularity, withoud evidence of AVM.

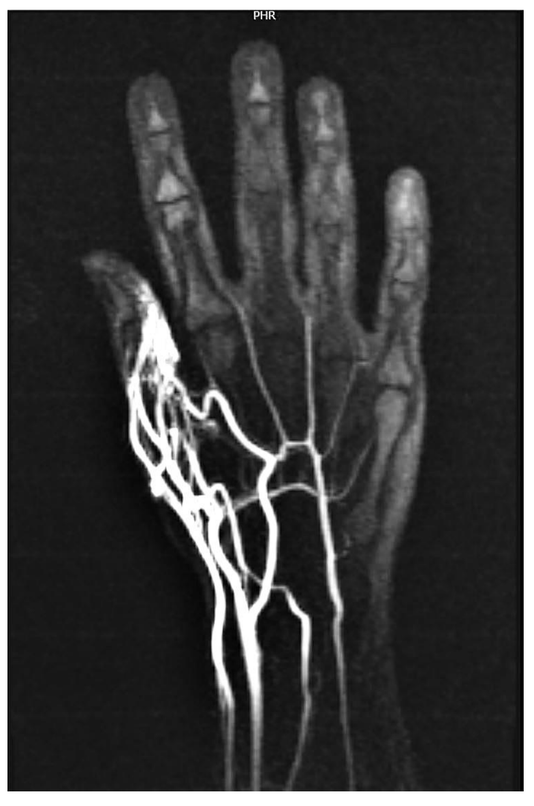

Figure 3
MR angiography of palmar AVM of the thumb

Discussion

In this case, molecular genetics have revealed AVM pathogenesis: biopsy showed a somatic BRAF mutation, linking AVMs to MAPK-pathway alterations[1]. Here, BRAF acts as a driver of abnormal vascular proliferation, not as a hereditary cancer marker. While KRAS mutations are more common, BRAF variants are increasingly recognized in AVMs[1,3]. Therapeutically, MAPK-pathway targeting with a BRAF inhibitor (dabrafenib) plus a MEK inhibitor (trametinib) has shown case-report efficacy, reducing flow and lesion size[4,6]. These approaches remain investigational but offer promise for progressive or surgically challenging AVMs, underscoring the value of molecular diagnostics in vascular anomalies.

This case highlights diagnostic pitfalls of hand AVMs, which can mimic inflammatory or rheumatologic disease[7]. AVMs are staged by Schobinger (I quiescent; II expansion; III destruction, IV decompensation). Our patient`s stable stage I/II supported conservative management[5]. Surgery was not recommended and no interventional therapy is currently indicated.

Early recognition with targeted imaging are essential to avoid diagnostic delay. Retrospectively, the initial ultrasound (Figure 2) likely reflected the malformation - later corroborated by a newly palpable pulsatile vessel at the thenar eminence. In our case, the interval from first presentation to definitive diagnosis was about one year. Detecting AVMs early enables safer, more effective management, whereas advanced disease is more difficult to treat and associated with higher complication rates[5].

Conclusion

Palmar AVMs are rare but potentially disabling lesions[7]. Retrospectively the main lesson is to consider vascular malformations earlier when symptoms persist and perform MR angiography. Molecular insights, such as BRAF mutations, further open perspectives for future targeted therapies[3,4,6].

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