

# Klinik und Einteilung vaskulärer Malformationen

Daniel Staub

Angiologie

Universitätsspital Basel

[daniel.staub@usb.ch](mailto:daniel.staub@usb.ch)

**Table I:** Clinical assessment

1.	<b>Patient and family history</b> <ul style="list-style-type: none"><li>– Lesion present at birth?</li><li>– Familial cluster?</li><li>– Earlier images (clinical photos, X-rays, etc.)</li></ul>
2.	<b>Physical examination</b> <ul style="list-style-type: none"><li>– Palpation (warmth, thrill, painful, etc.)</li><li>– Hand held doppler to distinguish between high flow vs. low flow</li></ul>
3.	<b>Duplex ultrasound</b> <ul style="list-style-type: none"><li>– Vascular malformation (vessels) vs. vascularized tumor (mass)</li><li>– High flow vs. low flow</li><li>– Structural involvement</li><li>– Typical imaging features (e. g. pleboliths in VMs?)</li></ul>
4.	<b>Planing of further imaging</b> <ul style="list-style-type: none"><li>– MRI T2 fat-saturated (with/without contrast): Feeding and draining vessels, extension</li></ul>

# Duplexsonographie

Primäre first-line Bildgebung (oder als Ergänzung)

- Vaskuläre Malformation oder vaskulärer Tumor
- Kompressibel oder nicht kompressibel
- Schneller oder langsamer Fluss (PRF erhöhen, PW-Doppler)
- Abklärung der Umgebungsstrukturen
- Zuführendes/abfließendes Gefäß
- Zugang für die Behandlung

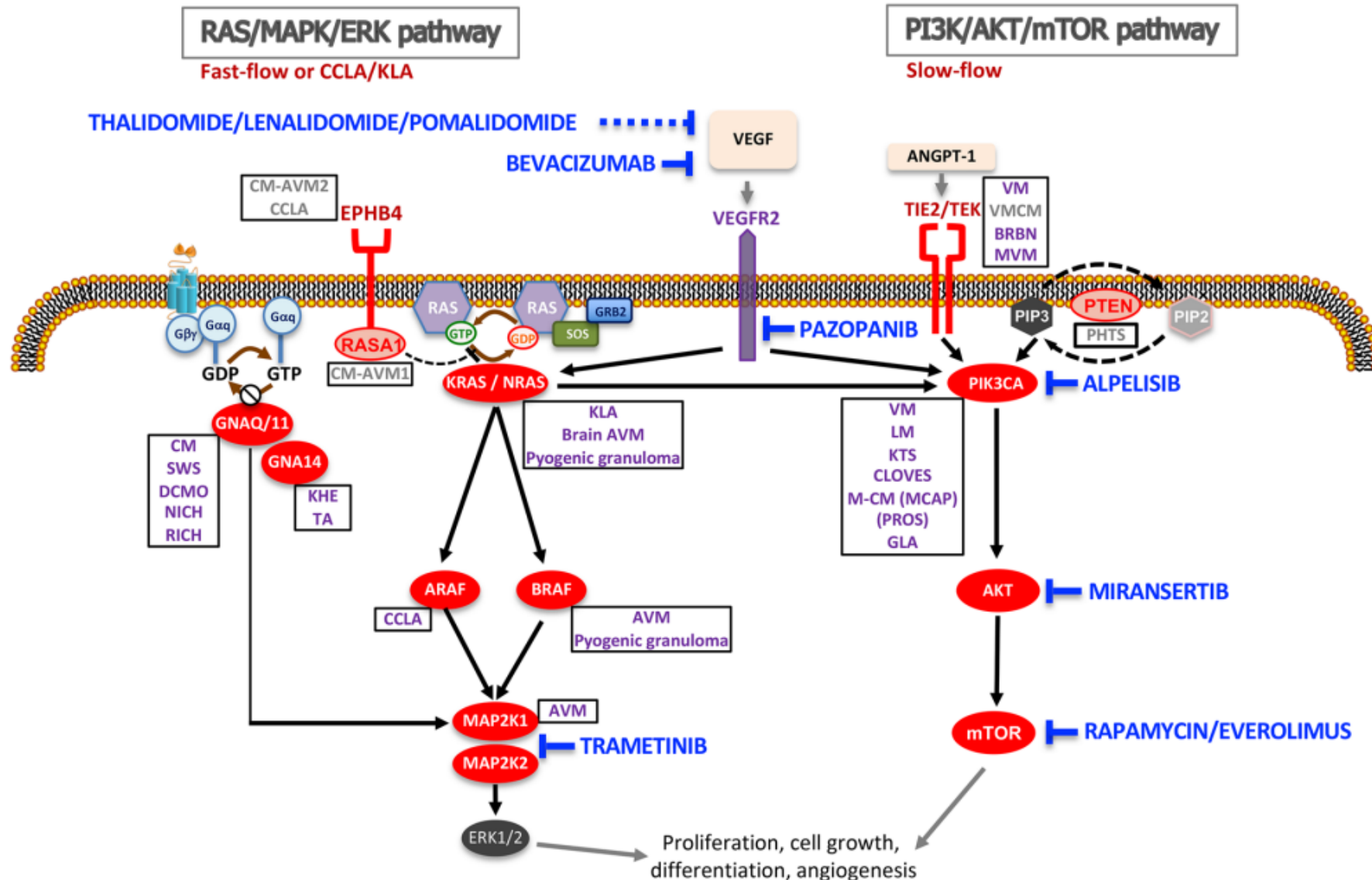
Vascular Anomalies	
Tumors	Malformations
<ul style="list-style-type: none"> <li>• <b>Benign:</b></li> <li>• Infantile Hemangioma (IH)</li> <li>• Rapidly involuting congenital hemangioma (RICH)</li> <li>• Non-involuting congenital hemangioma (NICH)</li> <li>• Pyogenic granuloma (lobular capillary hemangioma)</li>   <li>• <b>Borderline:</b></li> <li>• Kaposiform Hemangioendothelioma (KHE)</li>   <li>• <b>Malignant:</b></li> <li>• Angiosarcoma</li> <li>• Epithelioid Hemangioendothelioma (EHE)</li> </ul>	<ul style="list-style-type: none"> <li>• <b>Low flow:</b></li> <li>• Capillary malformation (CM)</li> <li>• Venous malformation (VM)</li> <li>• Lymphatic malformation (LM)</li>   <li>• <b>High flow:</b></li> <li>• Arteriovenous malformation (AVM)</li> <li>• Arteriovenous fistula (AVF)</li>   <li>• <b>Combined and syndromic</b></li> </ul>

<b>Combined vascular malformations*</b>		
CM + VM	capillary-venous malformation	CVM
CM + LM	capillary-lymphatic malformation	CLM
CM + AVM	capillary-arteriovenous malformation	CAVM
LM + VM	lymphatic-venous malformation	LVM
CM + LM + VM	capillary-lymphatic-venous malformation	CLVM
CM + LM + AVM	capillary-lymphatic-arteriovenous malformation	CLAVM
CM + VM + AVM	capillary-venous-arteriovenous malformation	CVAVM
CM + LM + VM + AVM	capillary-lymphatic-venous-arteriovenous m.	CLVAVM

## Vascular malformations associated with other anomalies

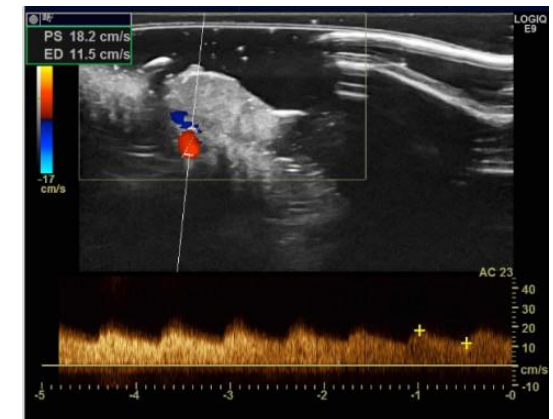
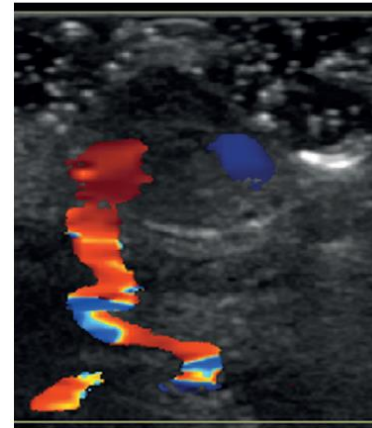
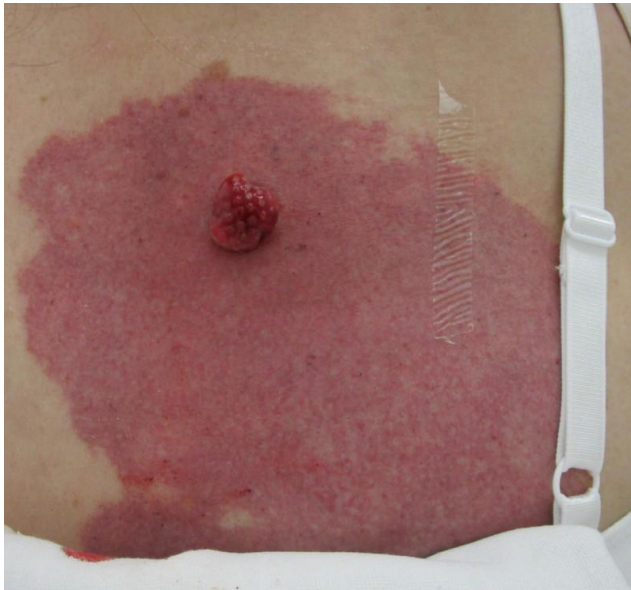
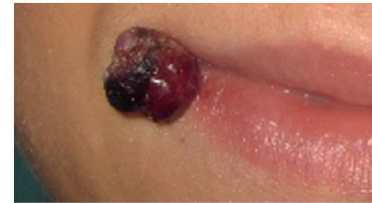
Klippel-Trenaunay syndrome: *	CM + VM +/- LM + limb overgrowth	PIK3CA
Parkes Weber syndrome:	CM + AVF + limb overgrowth	RASA1
Servelle-Martorell syndrome:	limb VM + bone undergrowth	
Sturge-Weber syndrome:	facial + leptomeningeal CM + eye anomalies +/- bone and/or soft tissue overgrowth	GNAQ
Limb CM + congenital non-progressive limb overgrowth		GNA11
Maffucci syndrome:	VM +/- spindle-cell hemangioma + enchondroma	IDH1 / IDH2
Macrocephaly - CM (M-CM / MCAP) *		PIK3CA
Microcephaly - CM (MICCAP)		STAMBP
CLOVES syndrome: *	LM + VM + CM +/- AVM + lipomatous overgrowth	PICK3CA
Proteus syndrome:	CM, VM and/or LM + asymmetrical somatic overgrowth	AKT1
Bannayan-Riley-Ruvalcaba sd:	AVM + VM +macrocephaly, lipomatous overgrowth	PTEN
CLAPO syndrome: *	lower lip CM + face and neck LM + asymmetry and partial/generalized overgrowth	PIK3CA

# Neue gezielte Therapie für vaskuläre Malformationen



# Malformation vs. Tumor?

- “Pyogenes Granuloma”
- Gestielter Tumor
- Zentrale Gefäße
- Schneller Fluss
- Blutungen
- Elektrische Schlinge, Histologie





## Venöse Malformation, low-flow (VM)

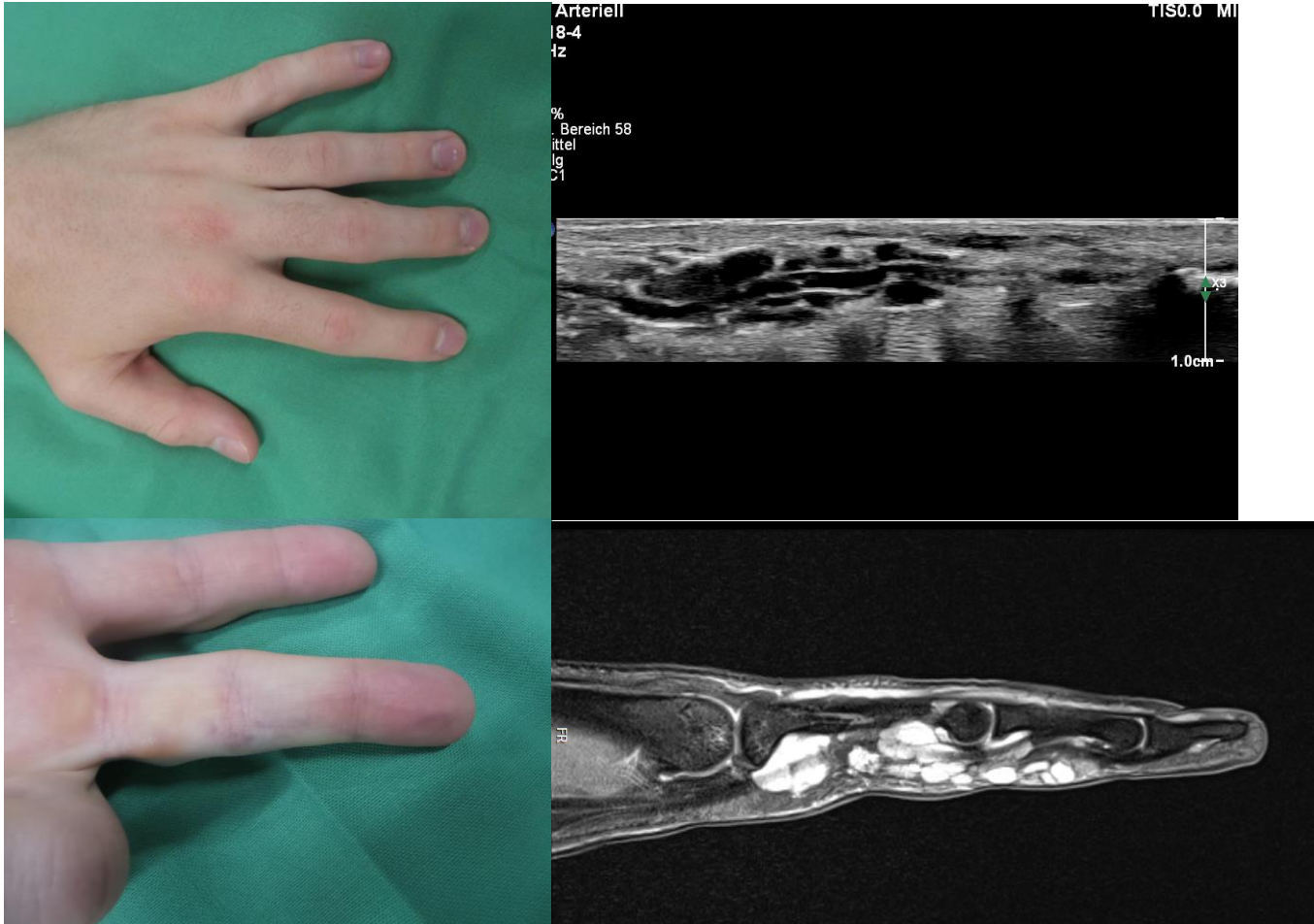
- Häufigste langsam fließende Malformation
- Kongenital (wie alle Malformationen)
- 90% mit Hautbeteiligung
- Spongiform, phlebektatisch, aneurysmatisch, retikulär
- Duplex: kompressibel (vs. LM nicht-kompressibel),  
Phlebolit
- Somatische Mutation

**Table II:** Classification of venous malformations

1.	<b>Spongiform type</b> <ul style="list-style-type: none"><li>– Most common type</li><li>– Often singular lesion</li><li>– Spongiform venous network</li></ul>
2.	<b>Phlebectatic type</b> <ul style="list-style-type: none"><li>– Large diameter</li><li>– Combined vascular malformations</li><li>– Increased risk of deep venous thrombosis/pulmonary embolism</li></ul>
3.	<b>Aneurysmatic type</b> <ul style="list-style-type: none"><li>– Involvement of large veins (IVC)</li><li>– Increased risk of deep venous thrombosis/pulmonary embolism</li></ul>
4.	<b>Reticular type</b> <ul style="list-style-type: none"><li>– Spiderweb-like appearance</li><li>– Combined vascular syndromes (fibro-adipose vascular anomaly)FAVA</li></ul>

## Klinische Beispiele

16-jähriger Patient, Schmerzen Digitus III  
beim Hantelheben und Karate



22-jähriger Patient

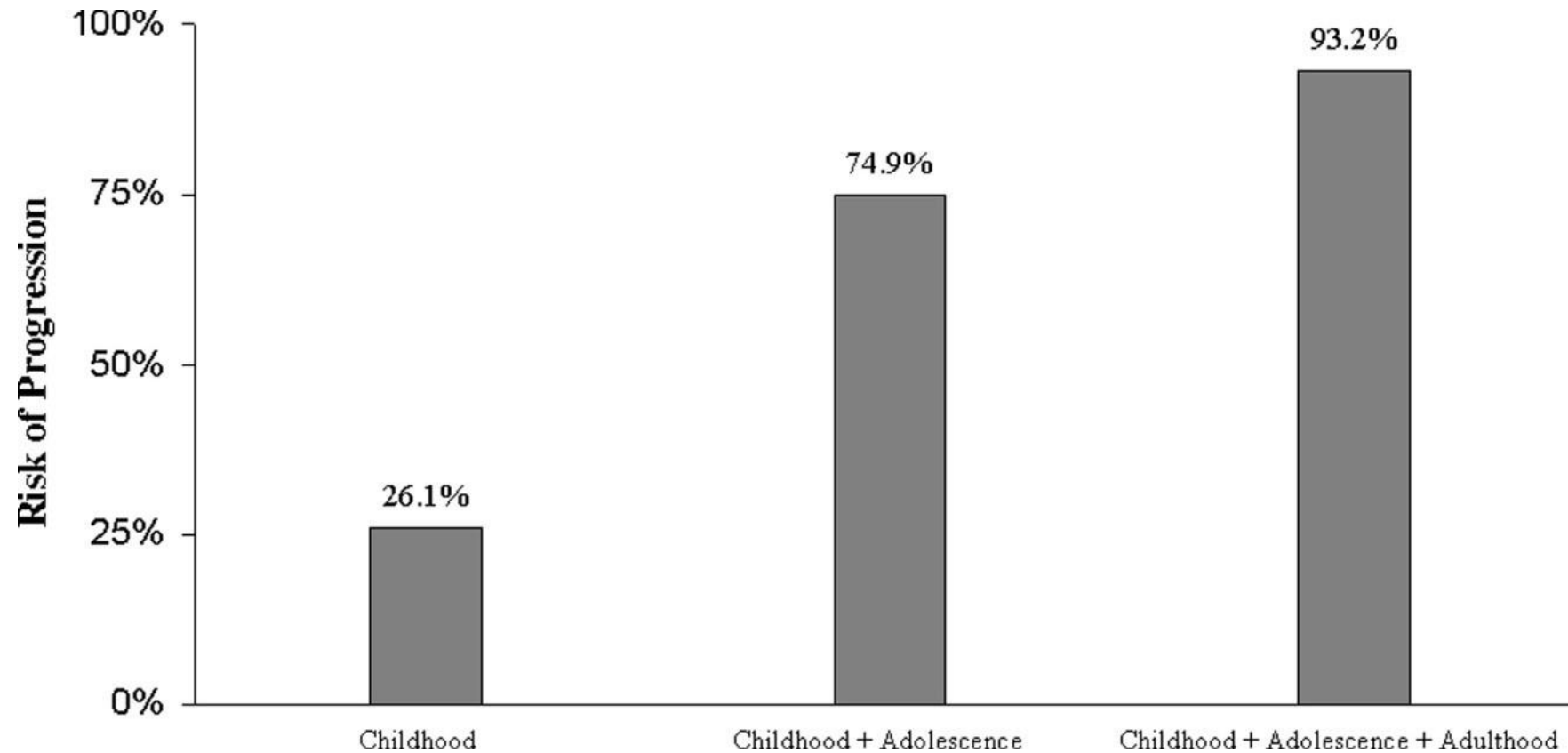


*Blue Rubber Bleb  
Naevus Syndrome*

9-jähriger Patient

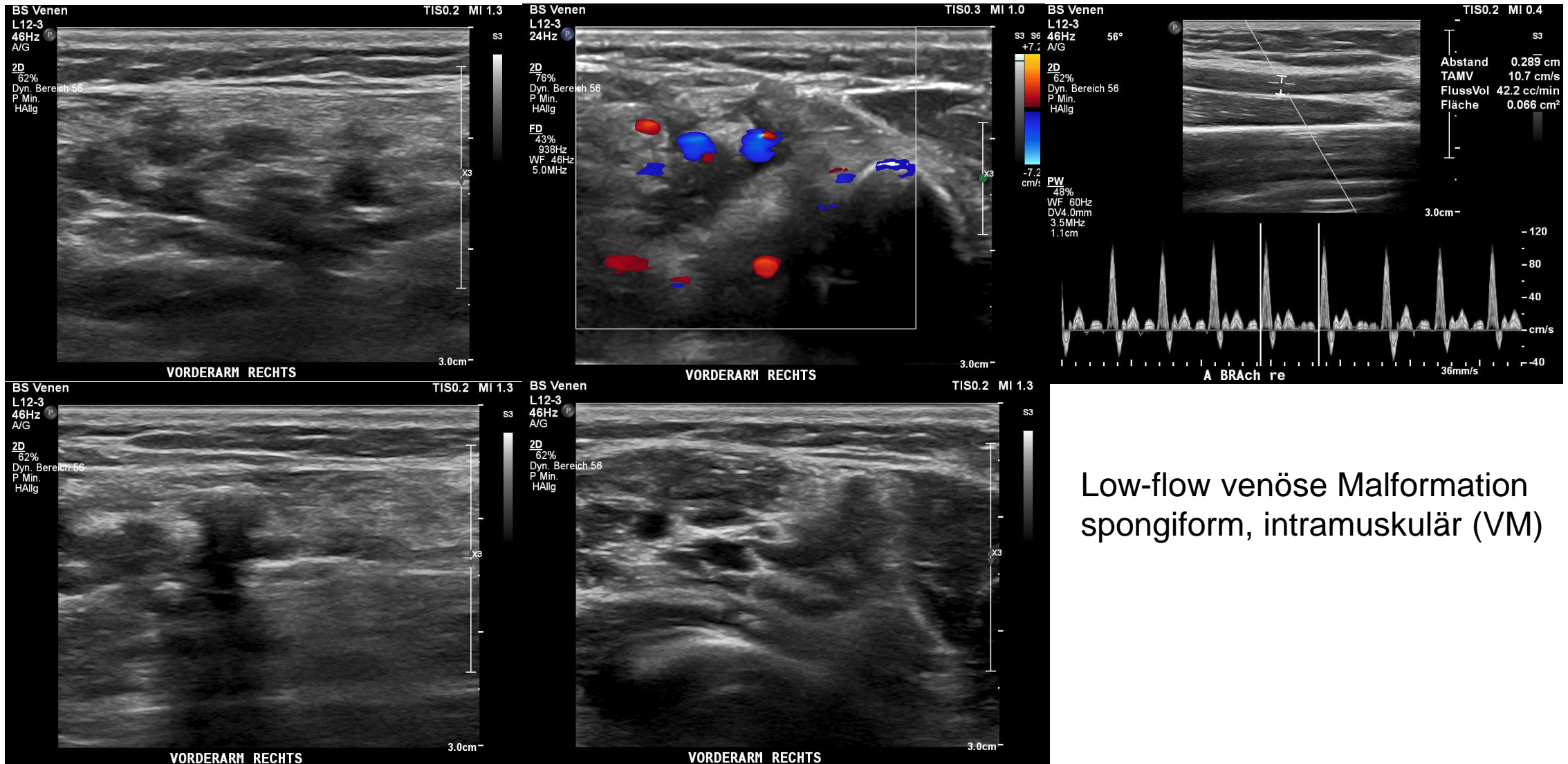


## Progression VM



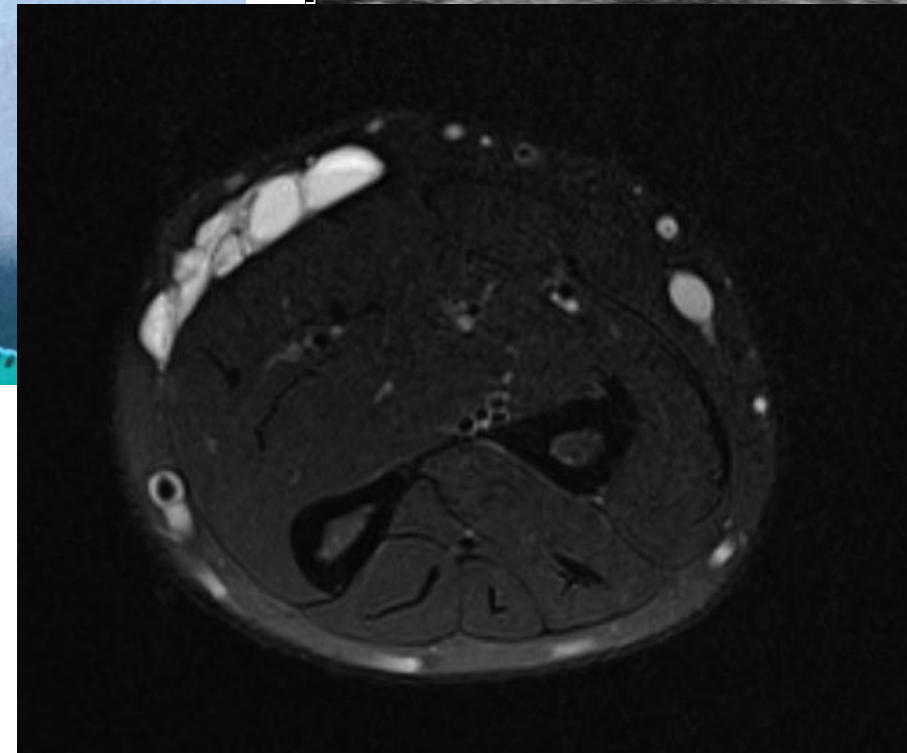
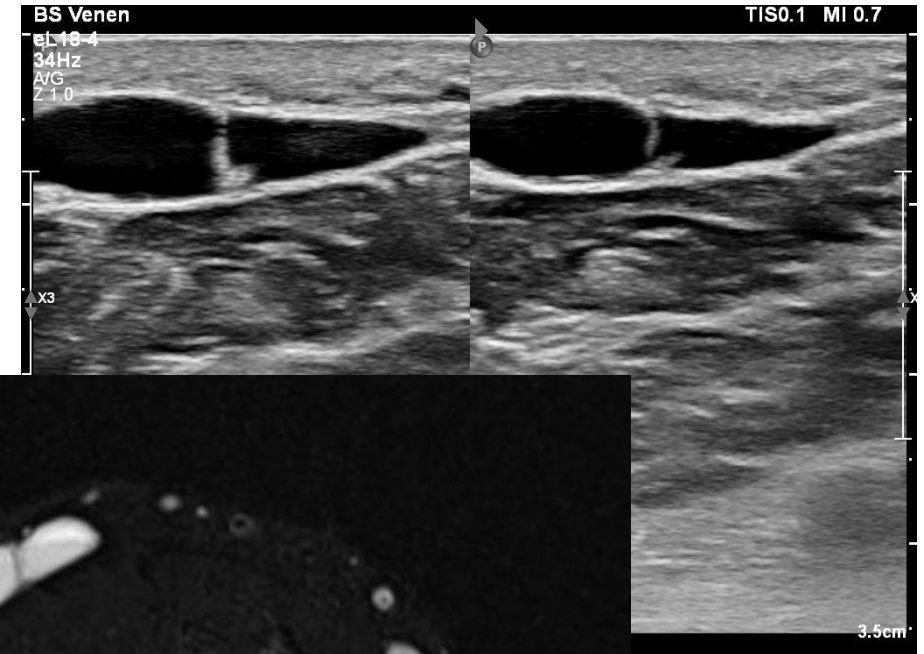
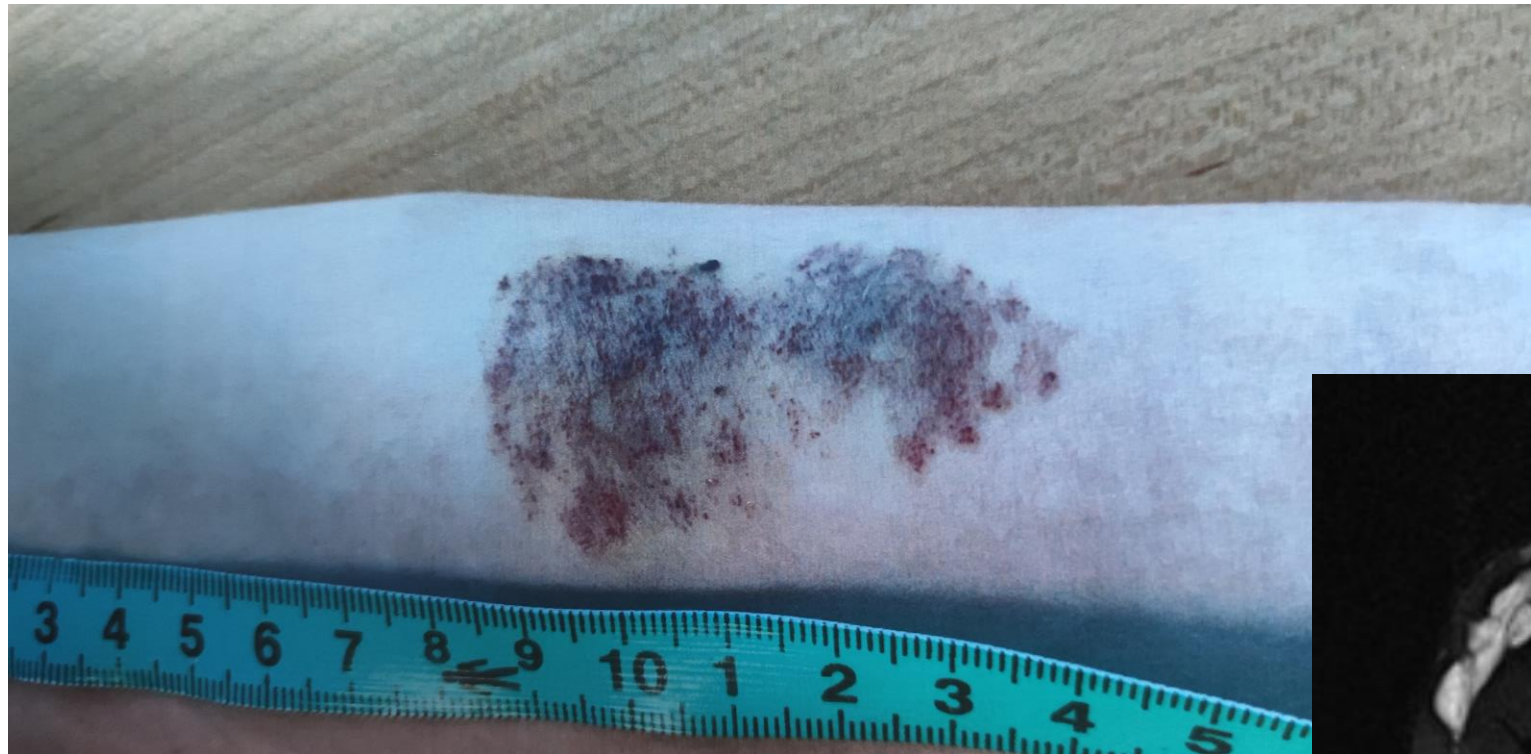
HASSANEIN AH, ET AL. VENOUS MALFORMATION: RISK OF PROGRESSION DURING CHILDHOOD AND ADOLESCENCE. *ANN PLAST SURG.* 2012

# 28-jähriger Patient, Schwellung und Schmerzen am Vorderarm



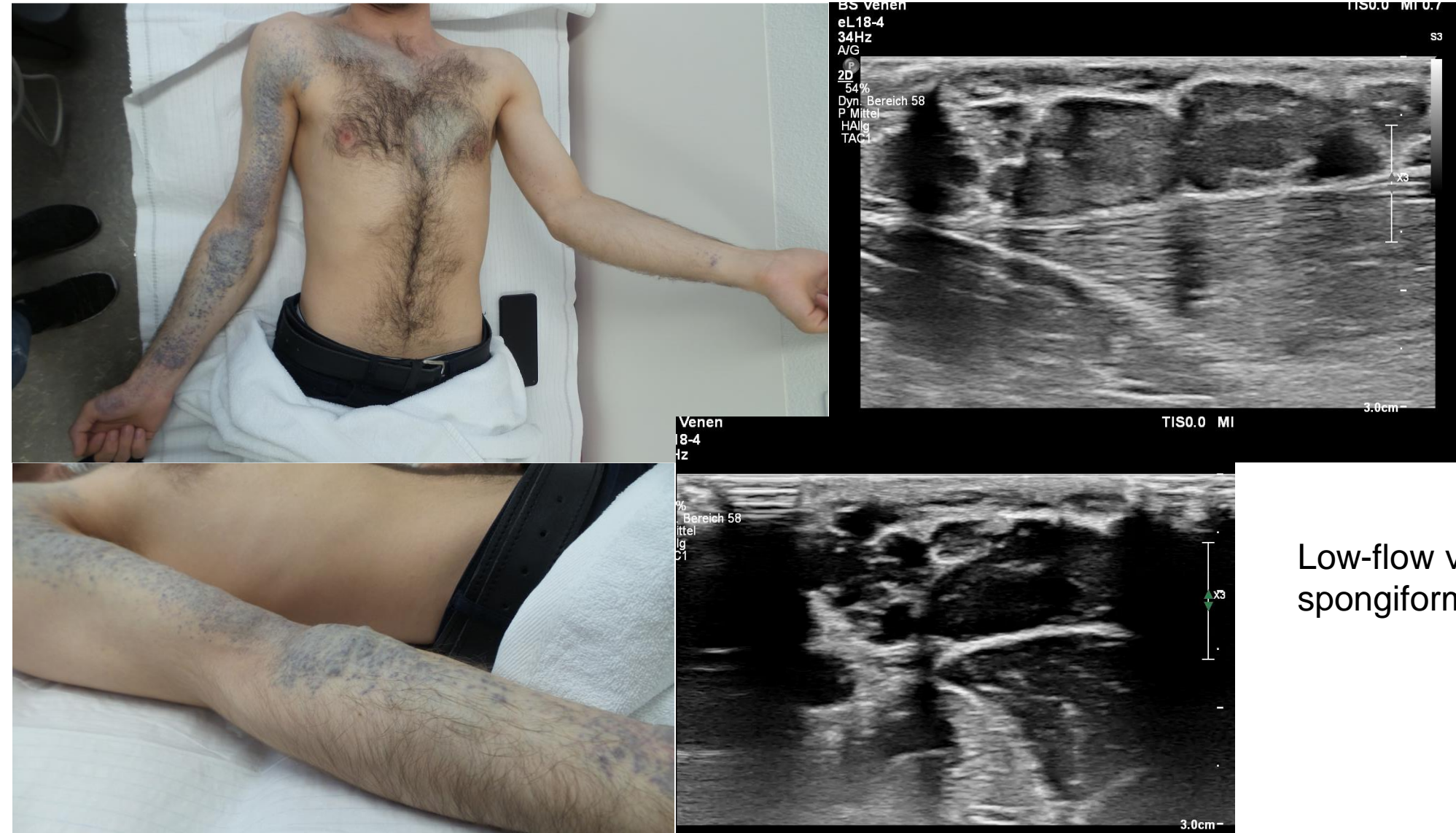
Low-flow venöse Malformation  
spongiform, intramuskulär (VM)

# 13-jähriger Patient, seit der Geburt Hautveränderung am Vorderarm, Schwellung und Schmerzen seit Monaten



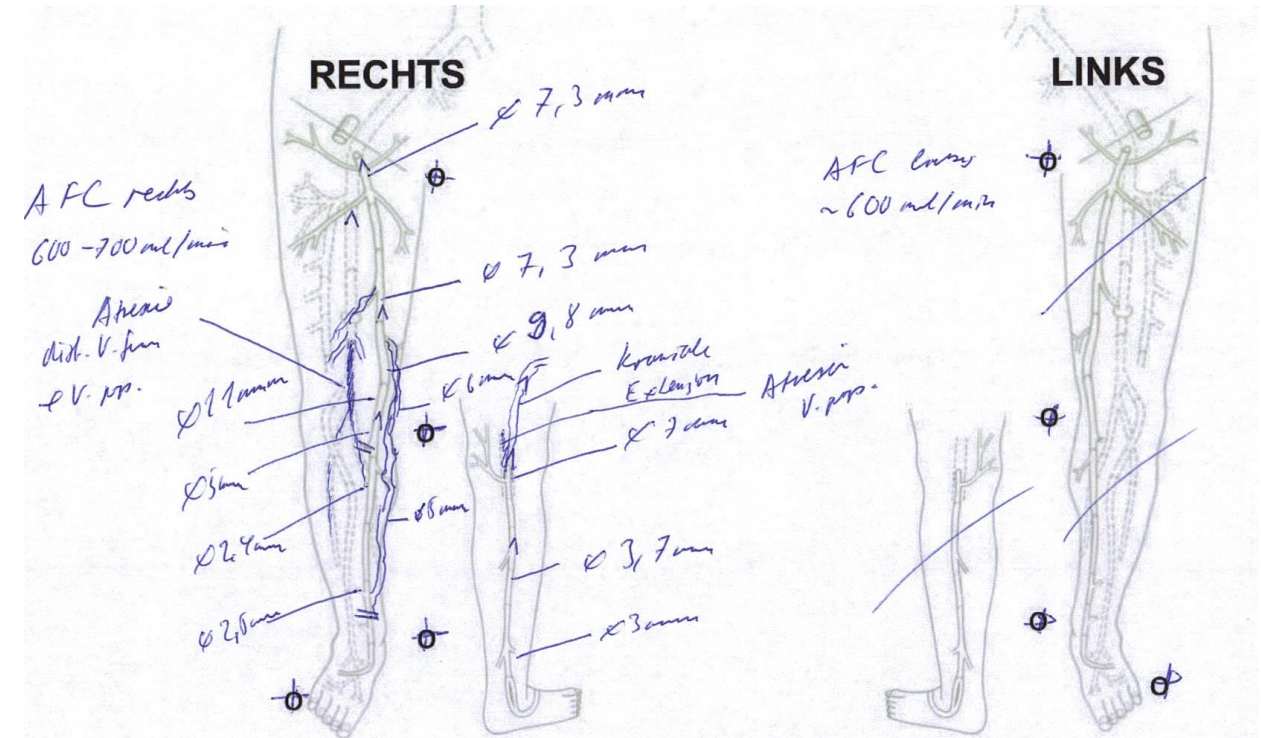
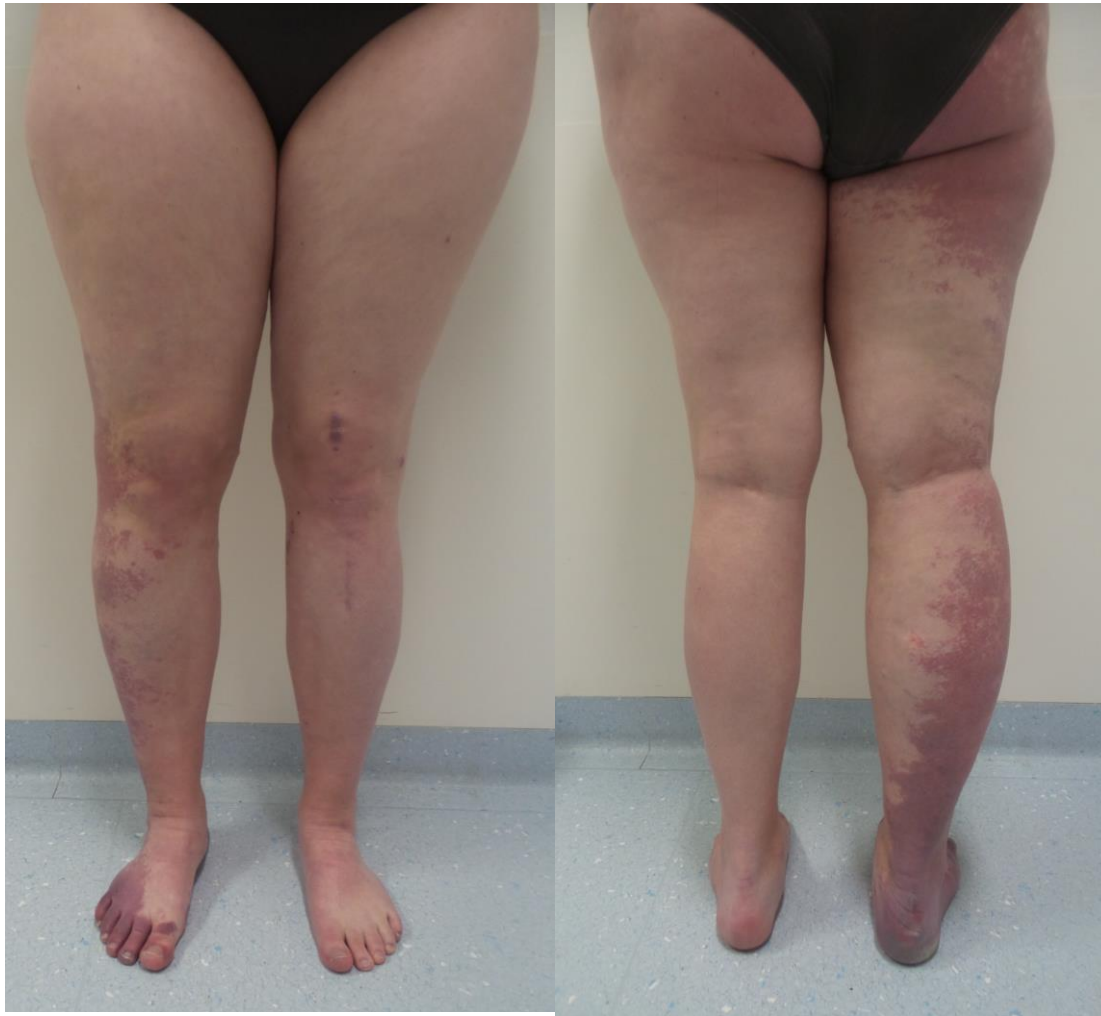
Low-flow makrozystische, lymphatische  
und kapilläre Malformation (CLM)

## 22-jähriger Patient, Flüchtling aus Afghanistan



Low-flow venöse Malformation  
spongiform, phlebectatisch (VM)

## 20-jährige Patientin, vaskuläre Malformation, Beinlängendifferenz links (operiert)

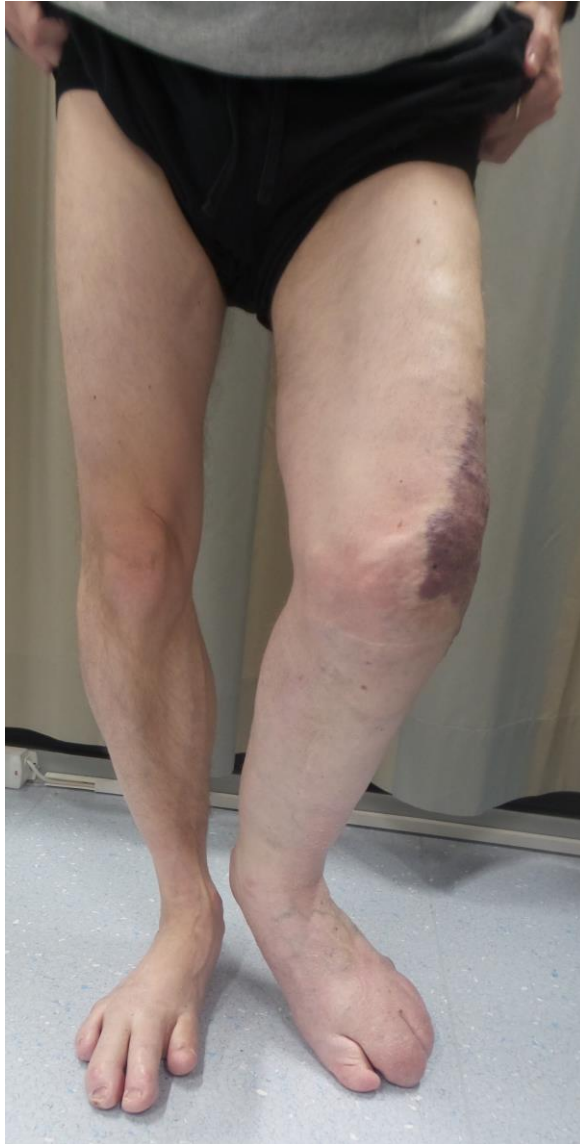


### Low-flow Malformation Typ Klippel-Trénaunay (CLVM)

- Kapilläre Malformation (CM) (Naevus flammeus)
- Venöse Malformation (VM) (Atresie des tiefen Venensystems)
- Lymphödem (LM)



## 46-jähriger Patient, vaskuläre Malformation, limb overgrowth (operiert)



### Low-flow Malformation Typ Klippel-Trénaunay (CLVM)

- Kapilläre Malformation (CM) (Naevus flammeus)
- Diffuse venöse Malformation (VM) (normales tiefes Venensystem) (operierte Marginalvene)
- Lymphödem (LM)

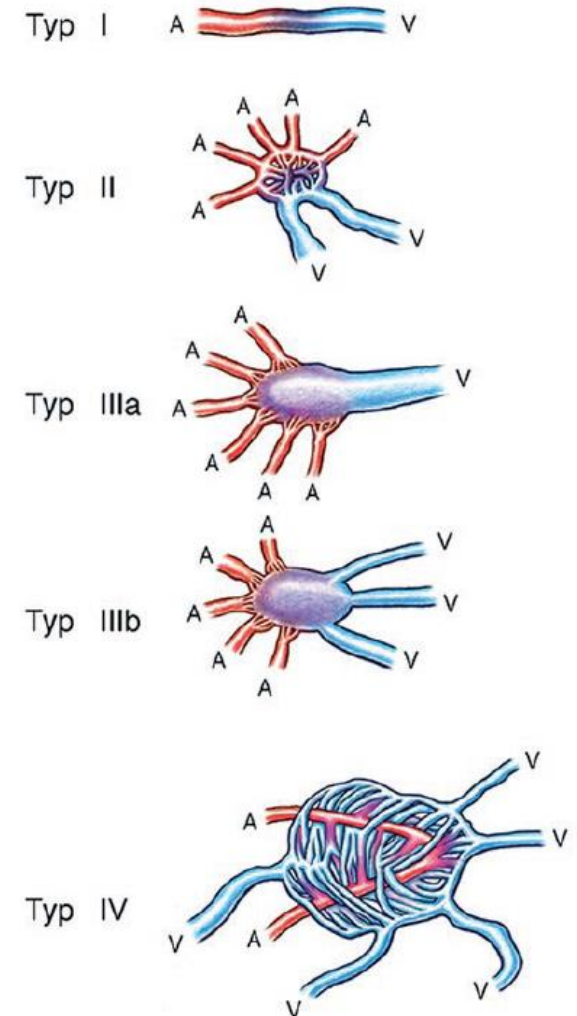
## Arterio-venöse Malformation (AVM)

**Table IV:** Schobinger classification of AVMs [70]

<b>Stage I: Quiescent</b>	Cutaneous blush/warmth, no relevant swelling
<b>Stage II: Expansion</b>	Lesion expansion with swelling and palpable bruit, no pain
<b>Stage III: Destruction</b>	Pain, ulceration, bleeding, infection
<b>Stage IV: Decompensation</b>	Additional high-output cardiac failure

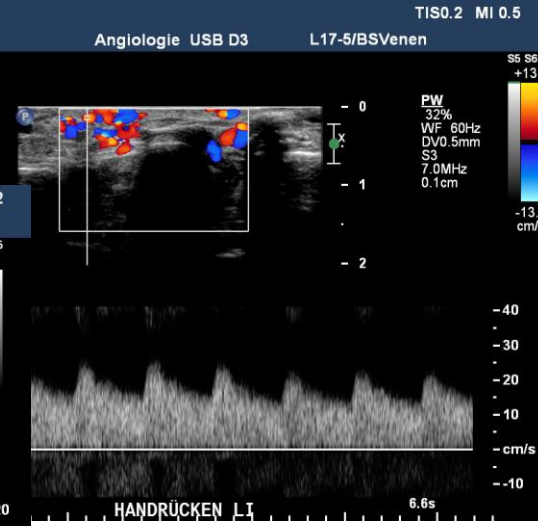
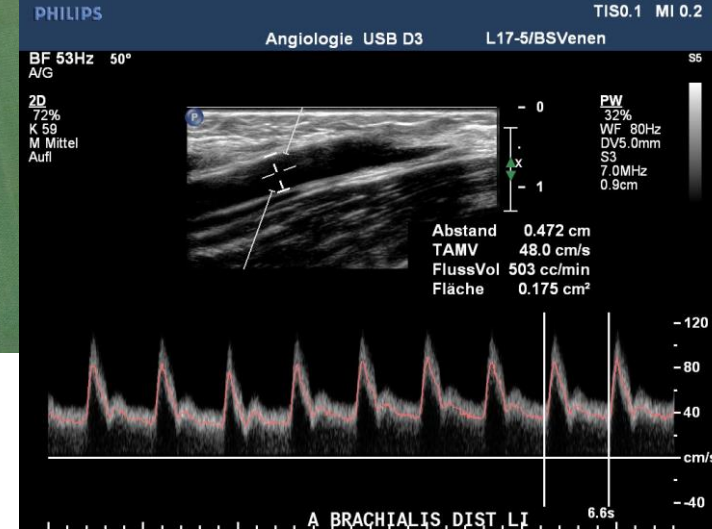
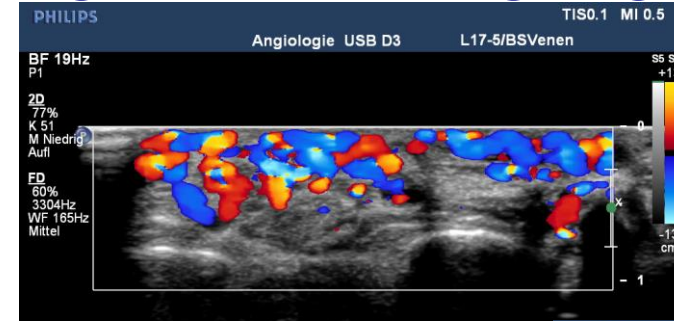
**Table V:** Angiographic classification of AVMs according to Cho et al. [71]

<b>Type I:</b>	No more than 3 arterial feeders shunt to 1 draining vein
<b>Type II:</b>	Multiple arterioles with plexiform appearance shunt to a single draining vein
<b>Type IIIa:</b>	Multiple arteriovenous fistulae with non-dilated fistula appearing as fine striation on angiography
<b>Type IIIb:</b>	AMultiple arteriovenous fistulae with dilated fistula appearing as complex vascular network on angiography



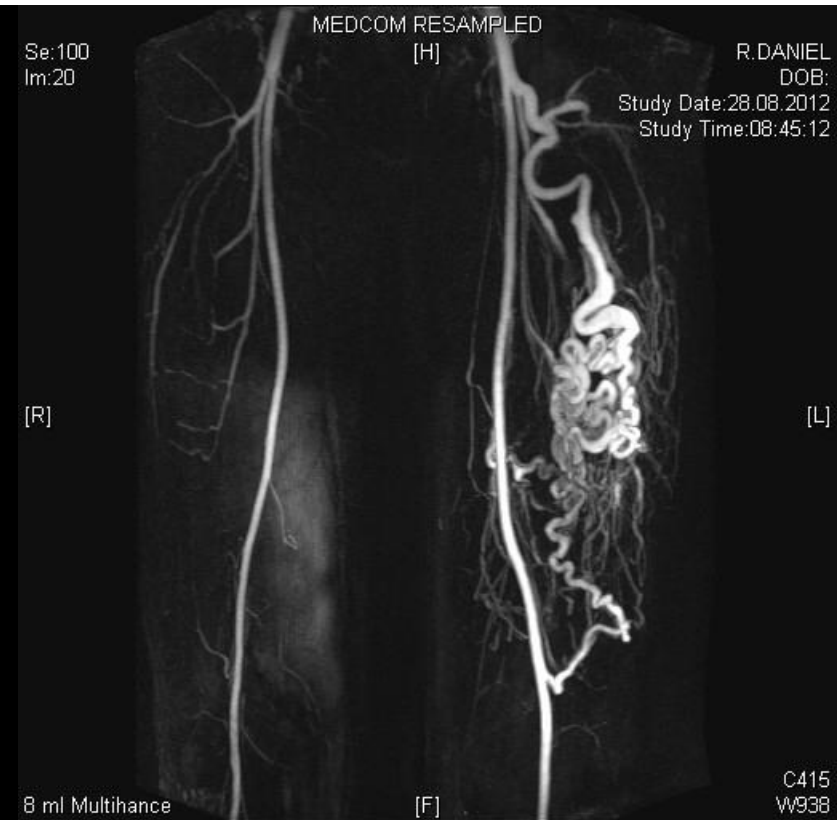
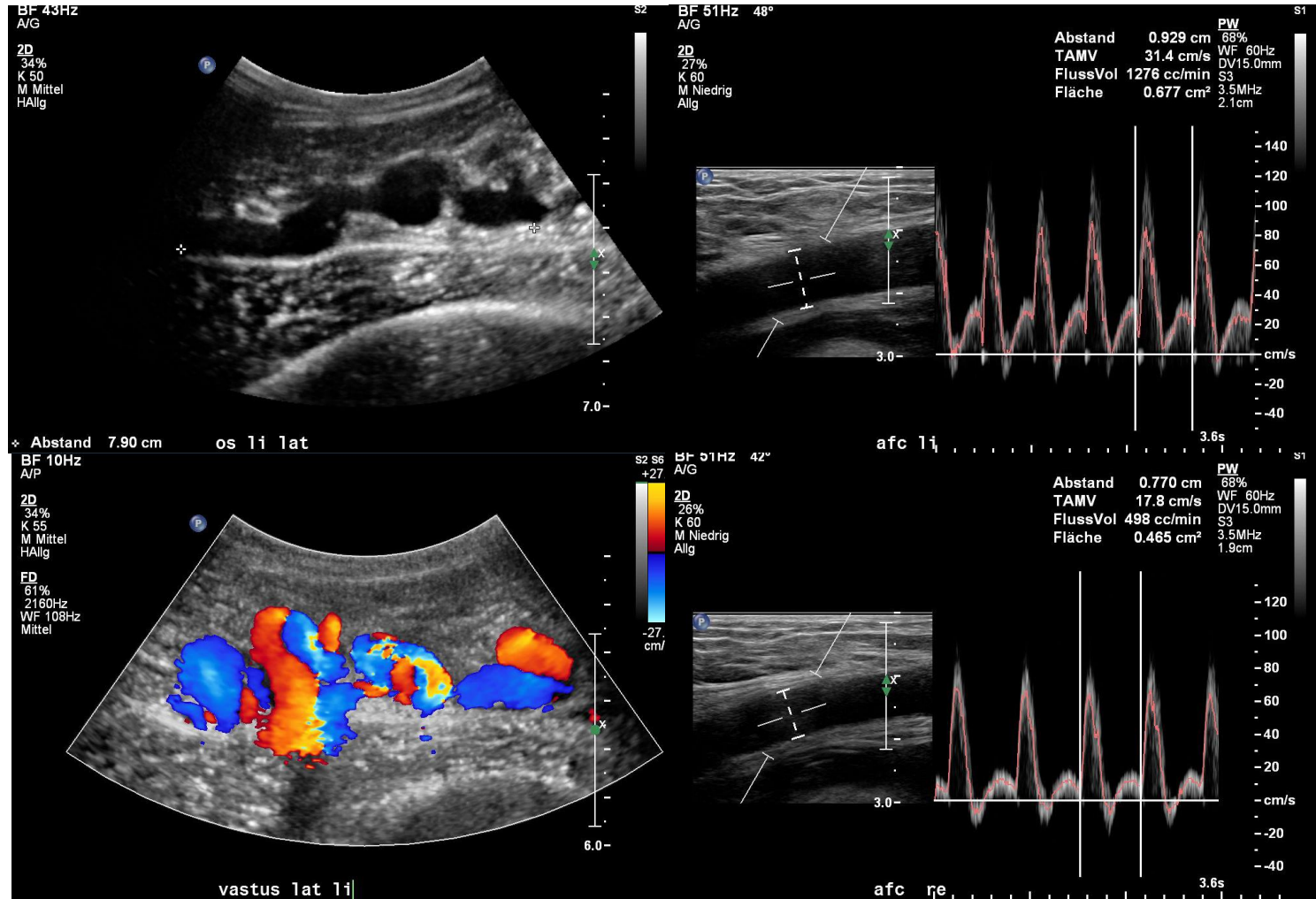
# 63-jährige Patientin, bekannte AVM der Hand, St.n. lokale Exzision und Embolisation,

## Druckgefühl und zunehmendes Wärmegefühl, Bewegungseinschränkung



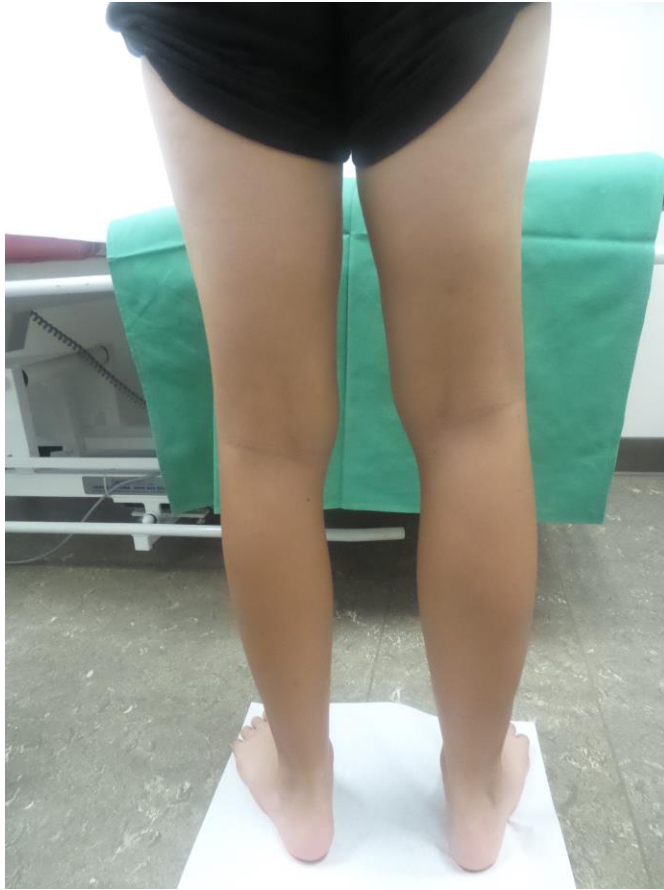
high-flow AV-Malformation (AVM) der Hand

# 35-jähriger Patient, Schwellung und Schmerzen am Oberschenkel

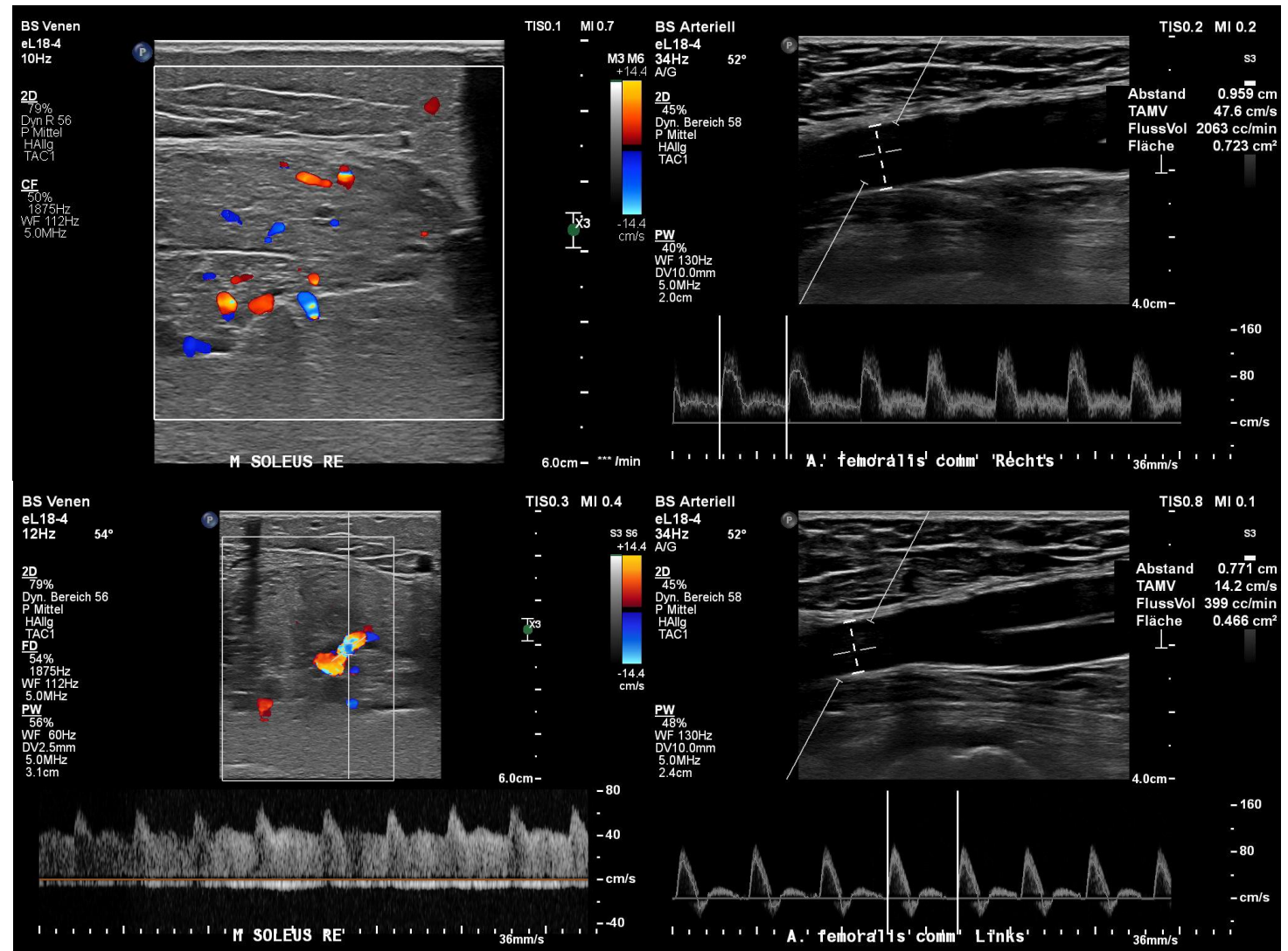


high-flow Malformation (AVM) am Oberschenkel

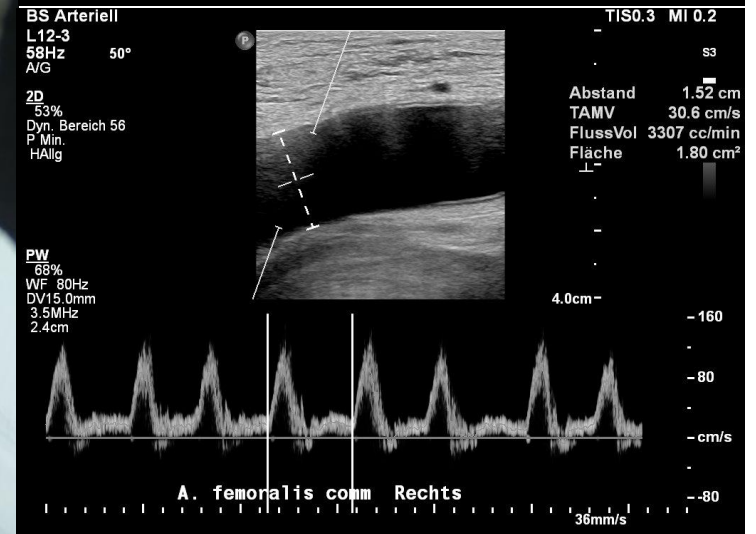
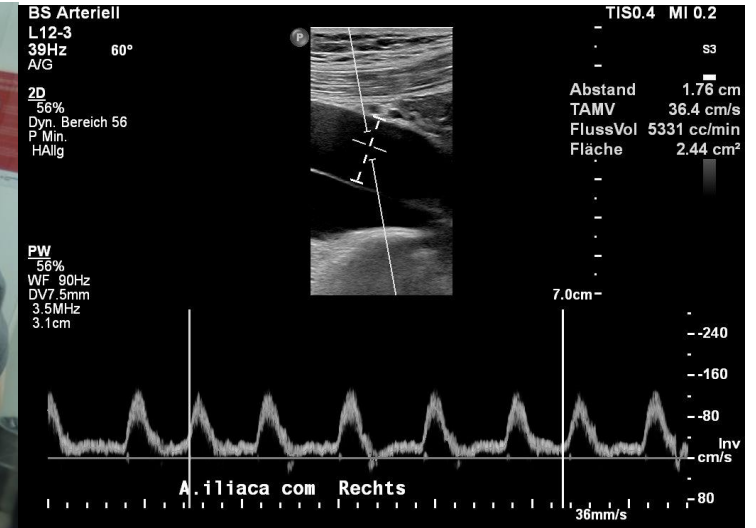
# 10-jähriger Patient, Beinlängendifferenz rechts, Umfangvermehrung, gut sichtbare oberflächliche Venen am rechten Bein



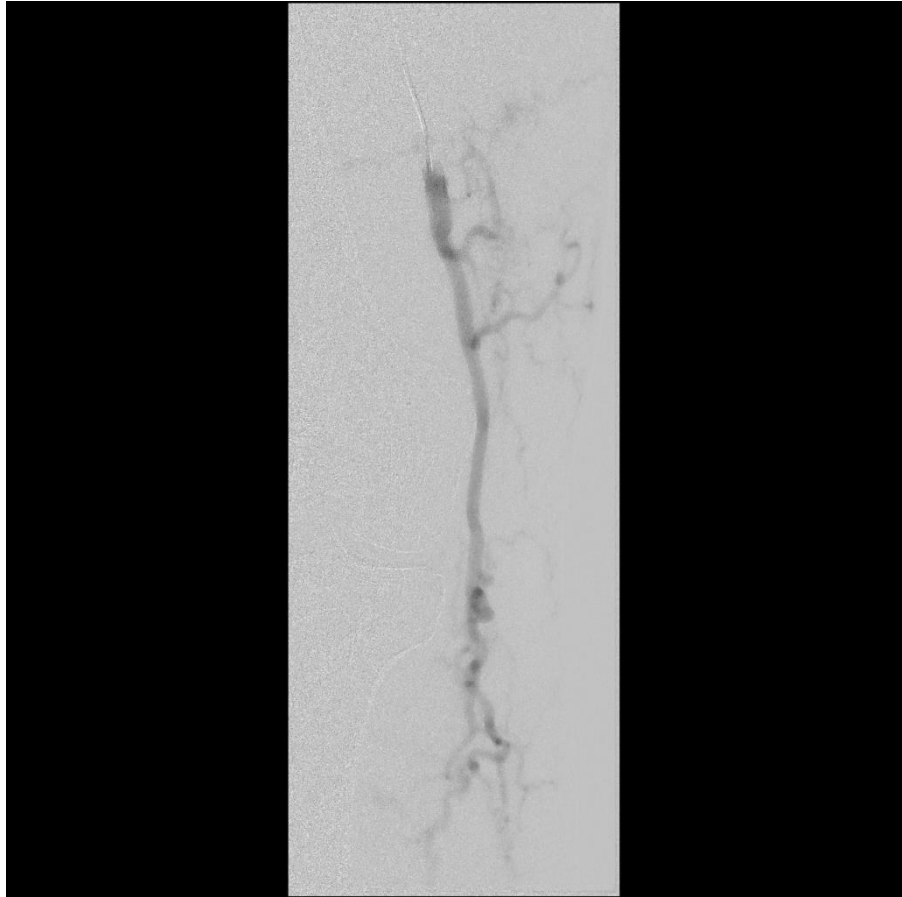
high-flow AV-Malformation,  
diffus in der Wadenmuskulatur (AVM)



# 55-jähriger Patient, bekannte vaskuläre Malformation Typ Parks-Weber, verschiedene Operationen in der Vorgeschichte



## 55-jähriger Patient, bekannte vaskuläre Malformation Typ Parks-Weber, High-Output Heart Failure (MRI cardiac output 13L/min)



- Kapilläre AV-Malformation (micro shunt)  
«Parkes-Weber-Syndrome» (capillary AVM)
- Naevus flammeus (CL)
- Lymphödem (LM)
- Limb overgrowth

Biopsie der kutanen kapillären  
Malformation (RASA 1 positiv)

Systemtherapie: Trametinib (Proteinkinase-  
Inhibitor verwendet in der Onkologie bei  
BRAF-Mutationen)

# Zusammenfassung

- Duplex als first-line tool oder als Ergänzung
- B-Bild: Tumor oder vaskuläre Malformation
- Duplex: Schneller oder langsamer Fluss, kompressibel, nicht-kompressibel
- Beschreibende Diagnose
- Seitenvergleich
- Ergänzende Bild- und Labordiagnostik
- Frage nach Behandlungsmöglichkeiten
- Chronische Erkrankung, multiple Behandlungen



# Vielen Dank

Für ihre Aufmerksamkeit!

