

Klinik und Einteilung vaskulärer Malformationen

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Table I: Clinical assessment

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

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

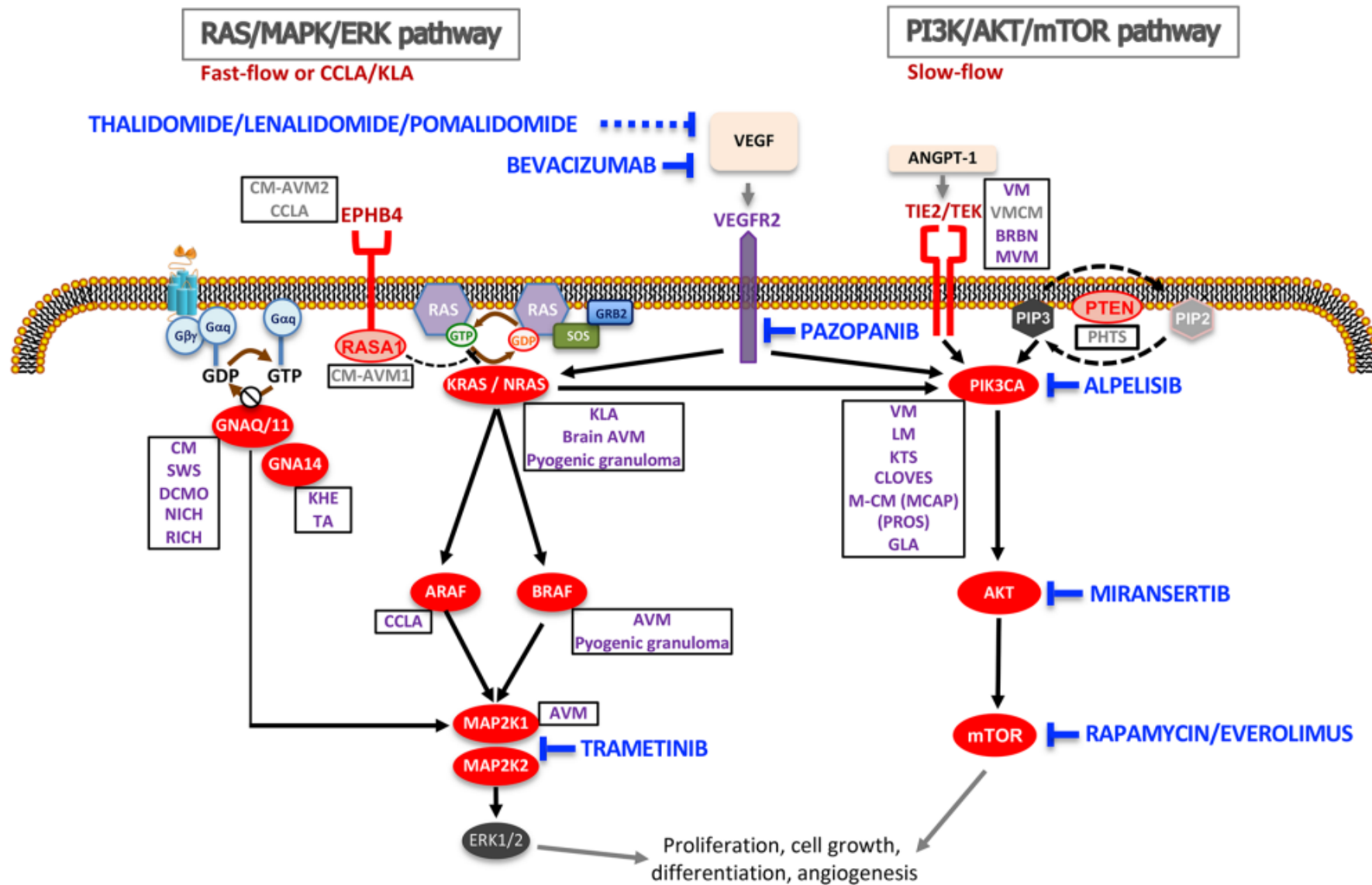
Vascular Anomalies	
Tumors	Malformations
<ul style="list-style-type: none"> • Benign: • Infantile Hemangioma (IH) • Rapidly involuting congenital hemangioma (RICH) • Non-involuting congenital hemangioma (NICH) • Pyogenic granuloma (lobular capillary hemangioma) • Borderline: • Kaposiform Hemangioendothelioma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

Combined vascular malformations*		
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

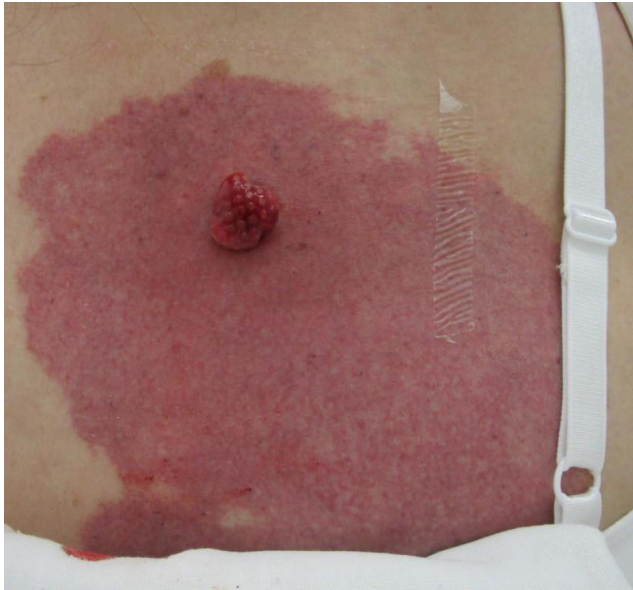
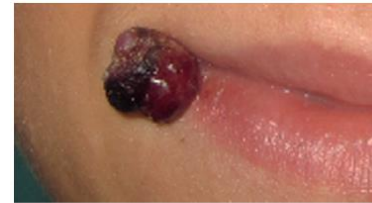
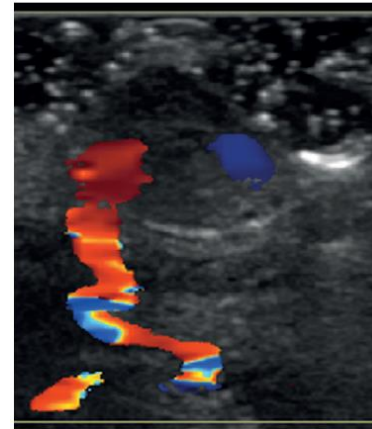
Klippel-Trenaunay syndrome: *	CM + VM +/- LM + limb overgrowth	PIK3CA
Parkes Weber syndrome:	CM + AVF + limb overgrowth	RASA1
Servelle-Martorell syndrome:	limb VM + bone undergrowth	
Sturge-Weber syndrome:	facial + leptomeningeal CM + eye anomalies +/- bone and/or soft tissue overgrowth	GNAQ
Limb CM + 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)

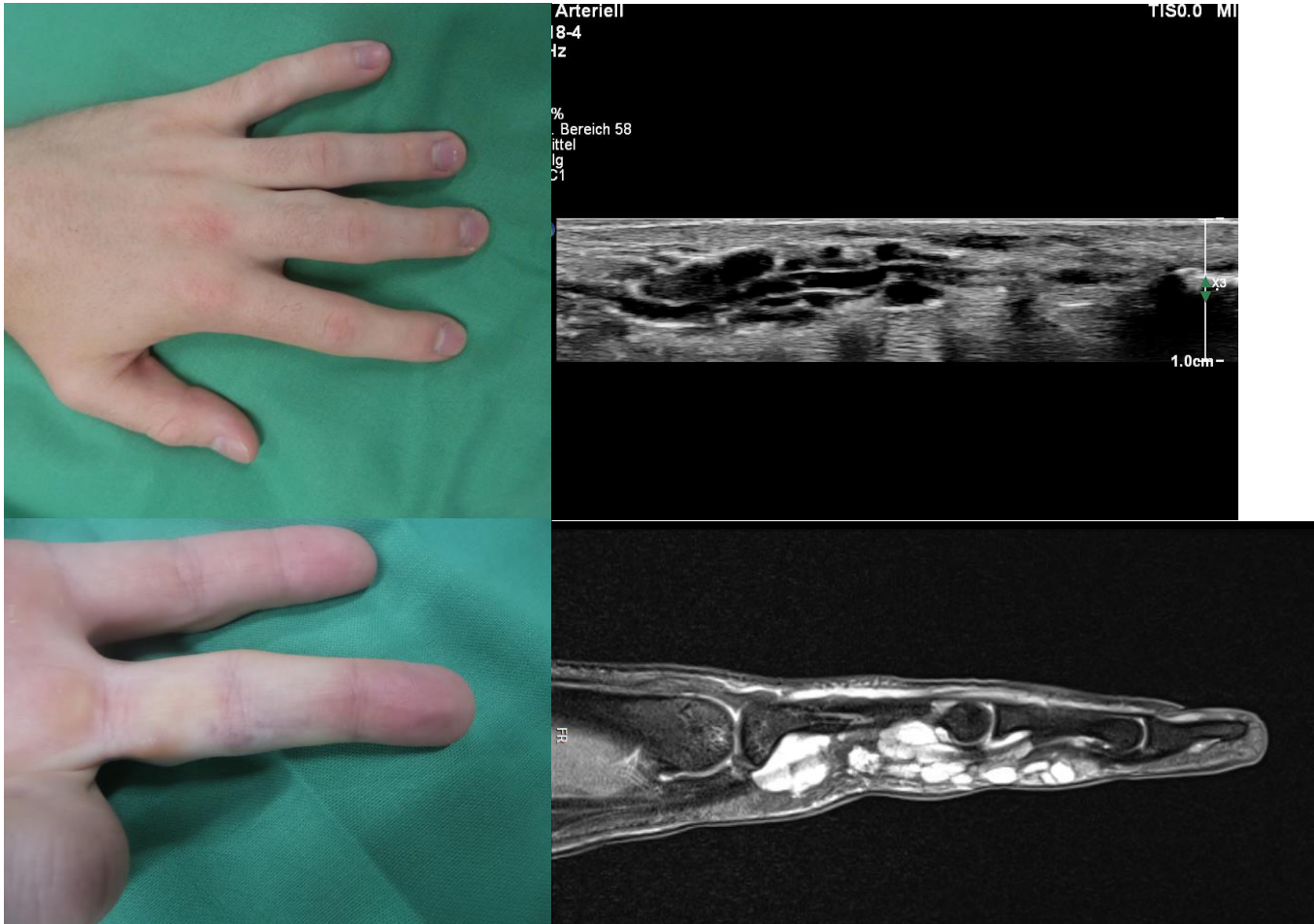
- Häufigste langsam fließende Malformation
- Kongenital (wie alle Malformationen)
- 90% mit Hautbeteiligung
- Spongiform, phlebektatisch, aneurysmatisch, retikulär
- Duplex: kompressibel (vs. LM nicht-kompressibel),
Phlebolit
- Somatische Mutation

Table II: Classification of venous malformations

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

Klinische Beispiele

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



22-jähriger Patient

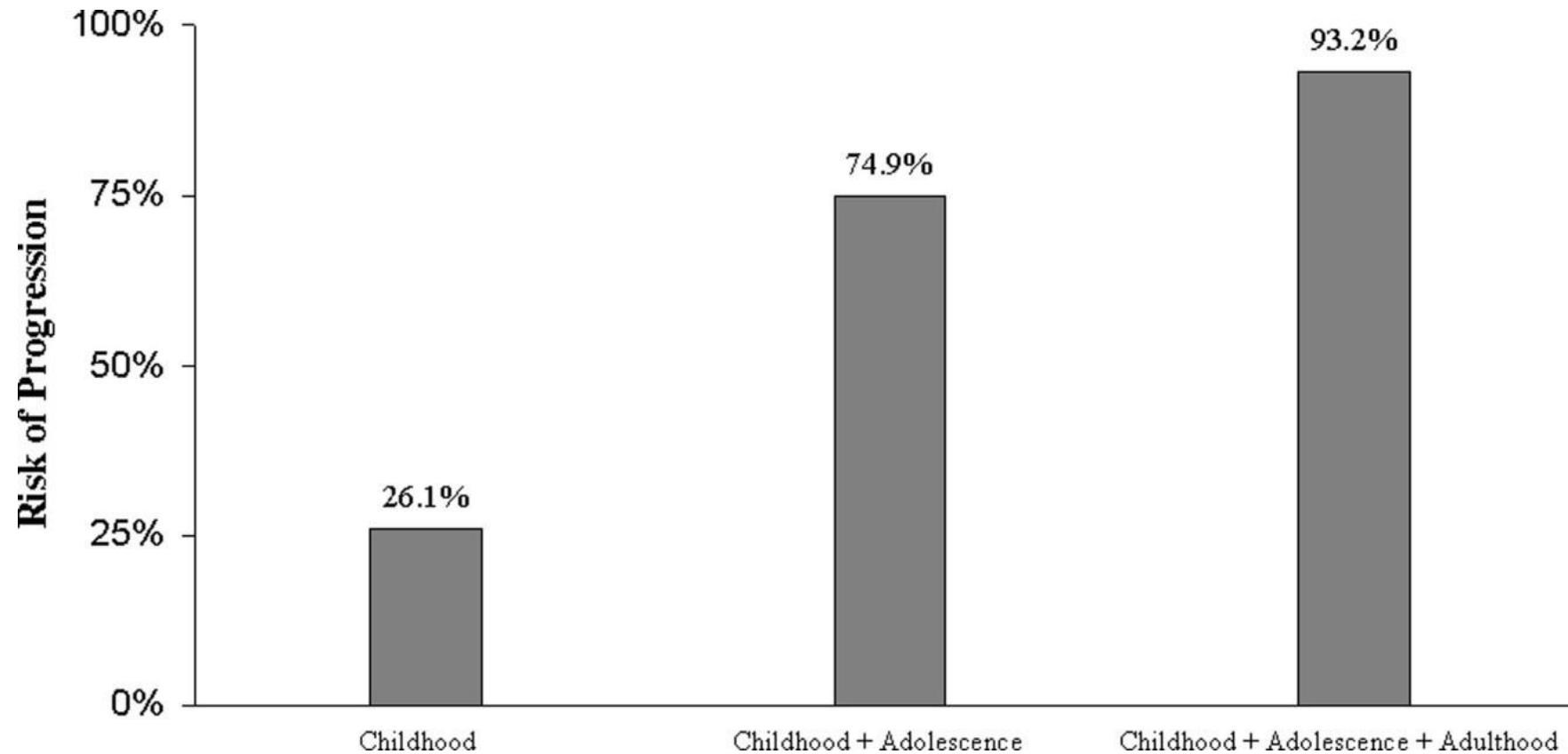


*Blue Rubber Bleb
Naevus Syndrome*

9-jähriger Patient

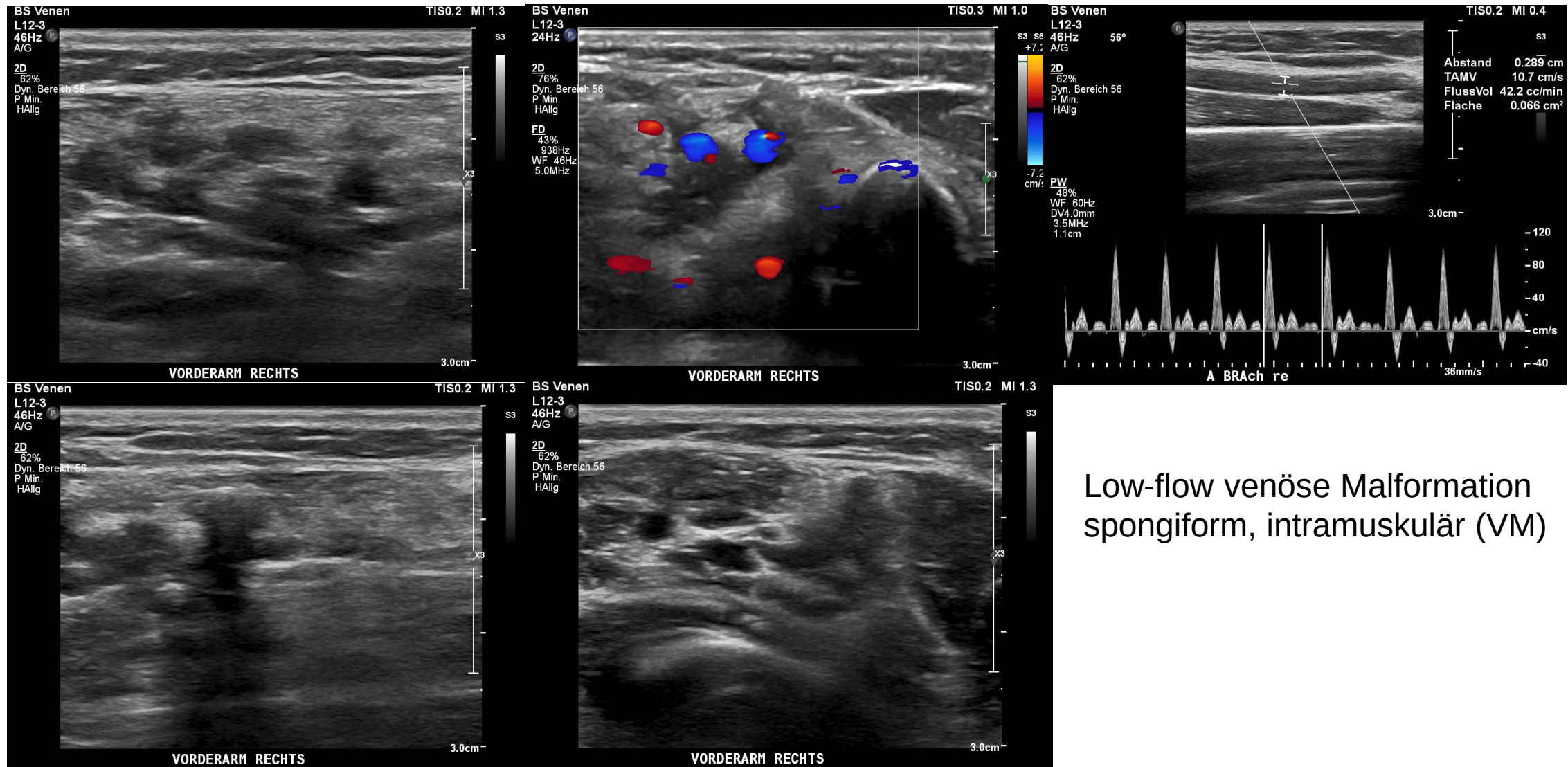


Progression VM

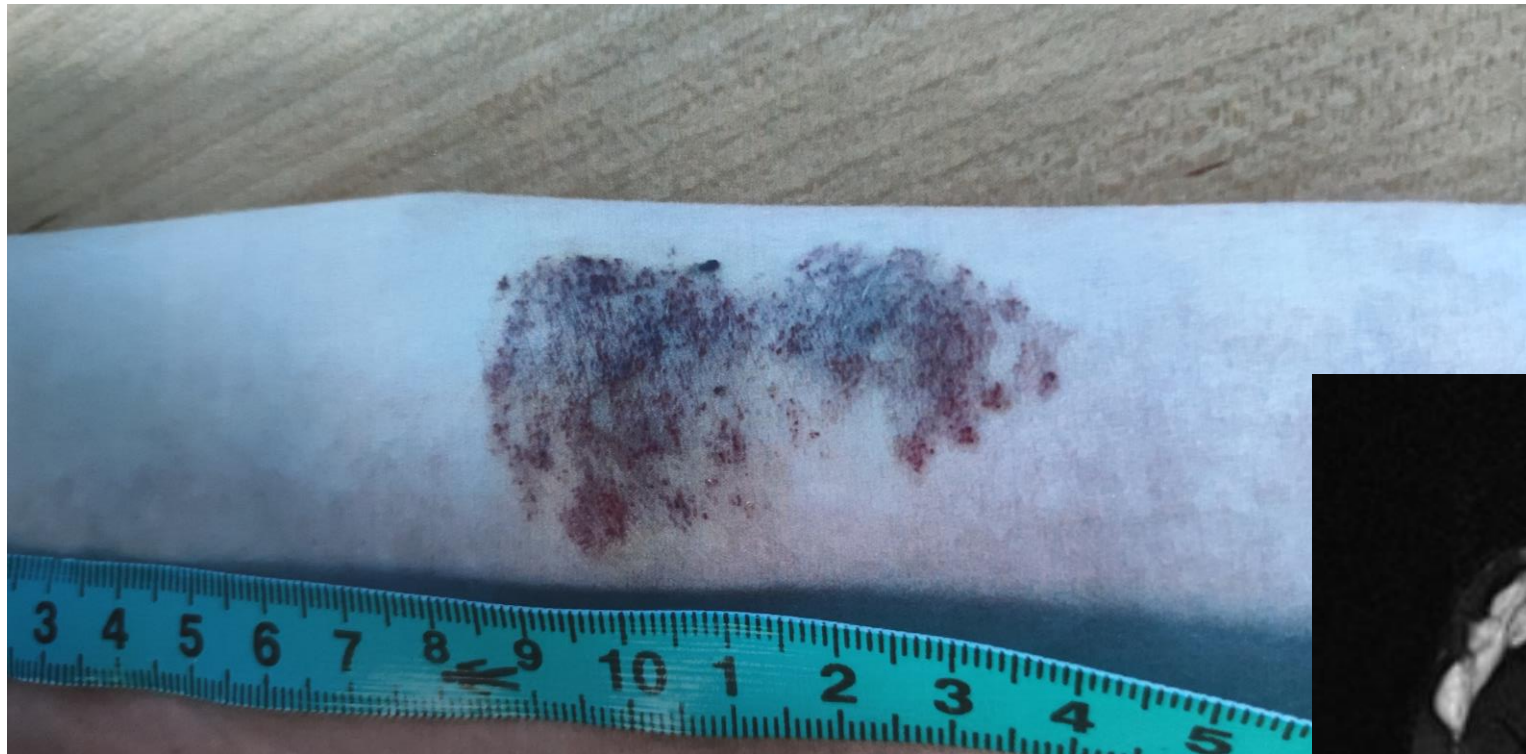
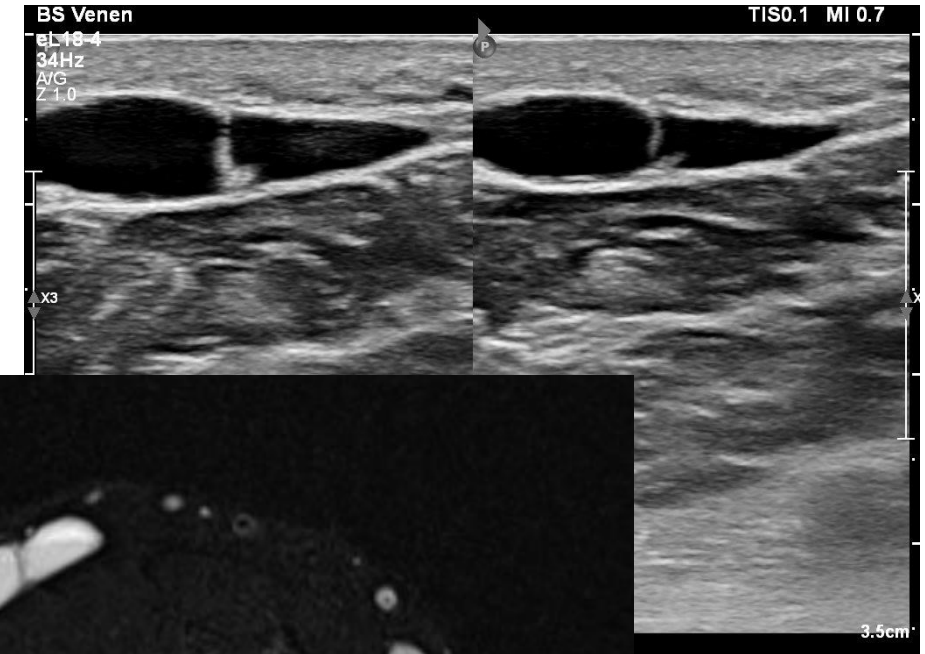
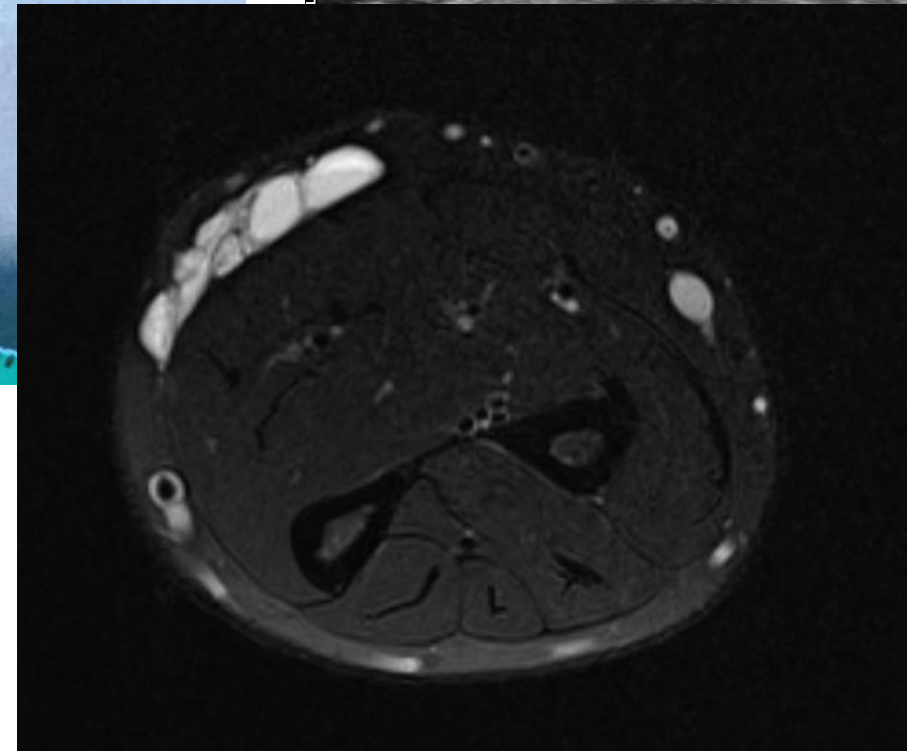


HASSANEIN AH, ET AL. VENOUS MALFORMATION: RISK OF PROGRESSION DURING CHILDHOOD AND ADOLESCENCE. *ANN PLAST SURG.* 2012

28-jähriger Patient, Schwellung und Schmerzen am Vorderarm



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



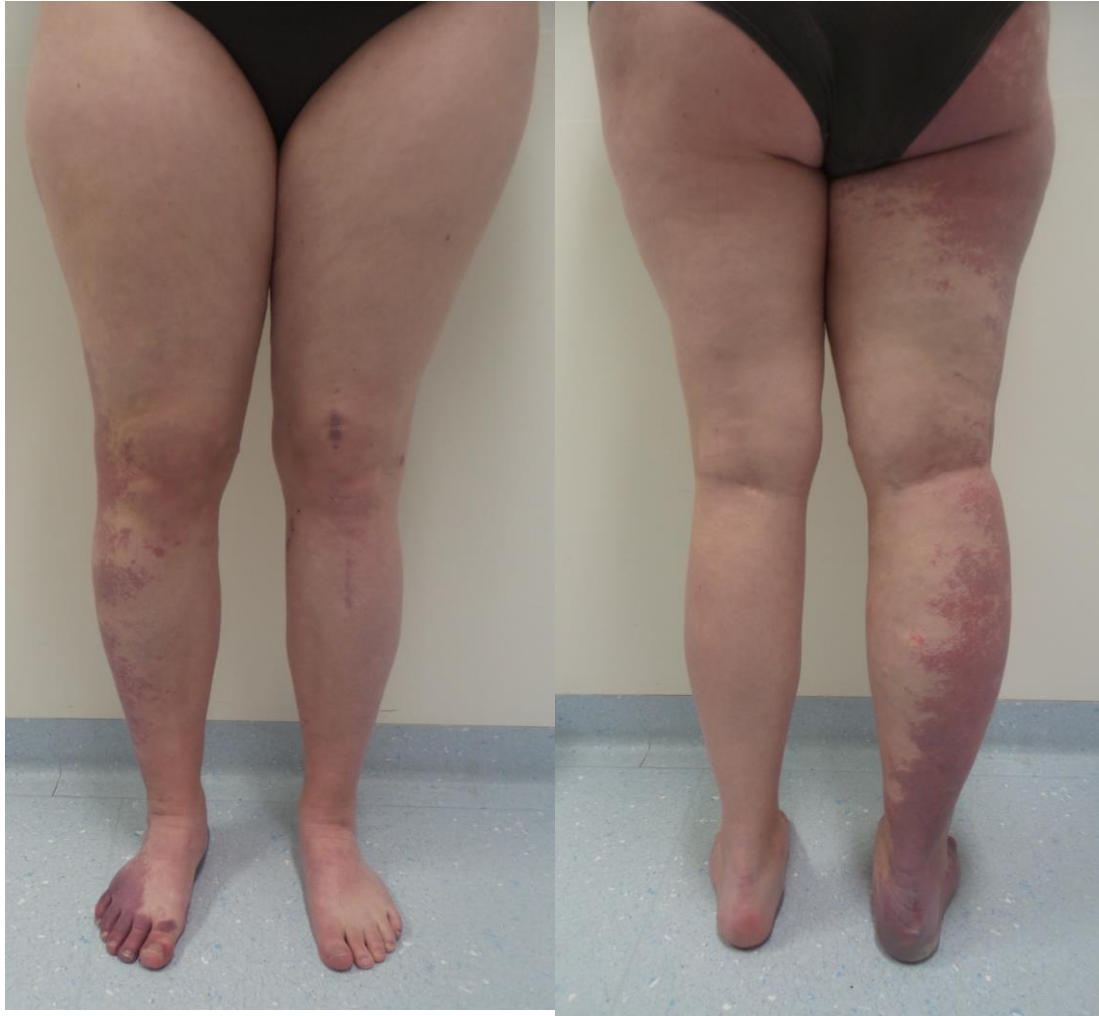
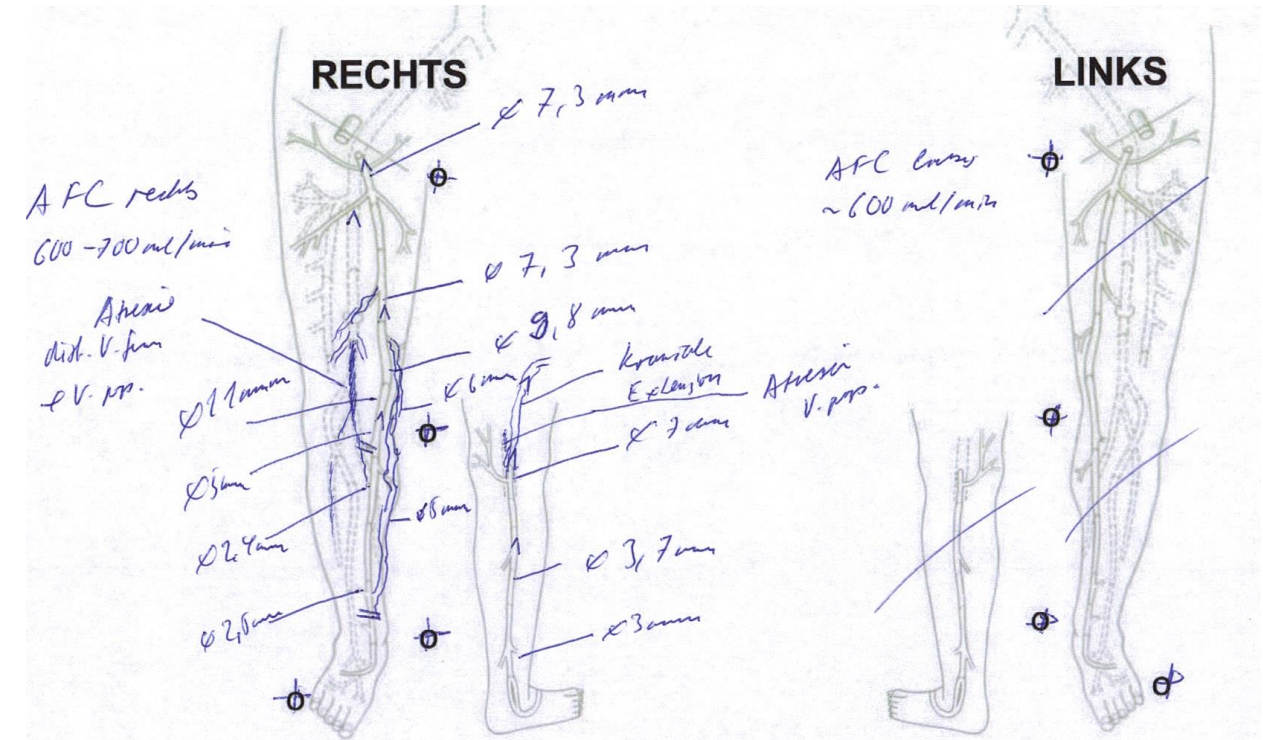
Low-flow makrozystische, lymphatische
und kapilläre Malformation (CLM)

22-jähriger Patient, Flüchtling aus Afghanistan



Low-flow venöse Malformation
spongiform, phlebectatisch (VM)

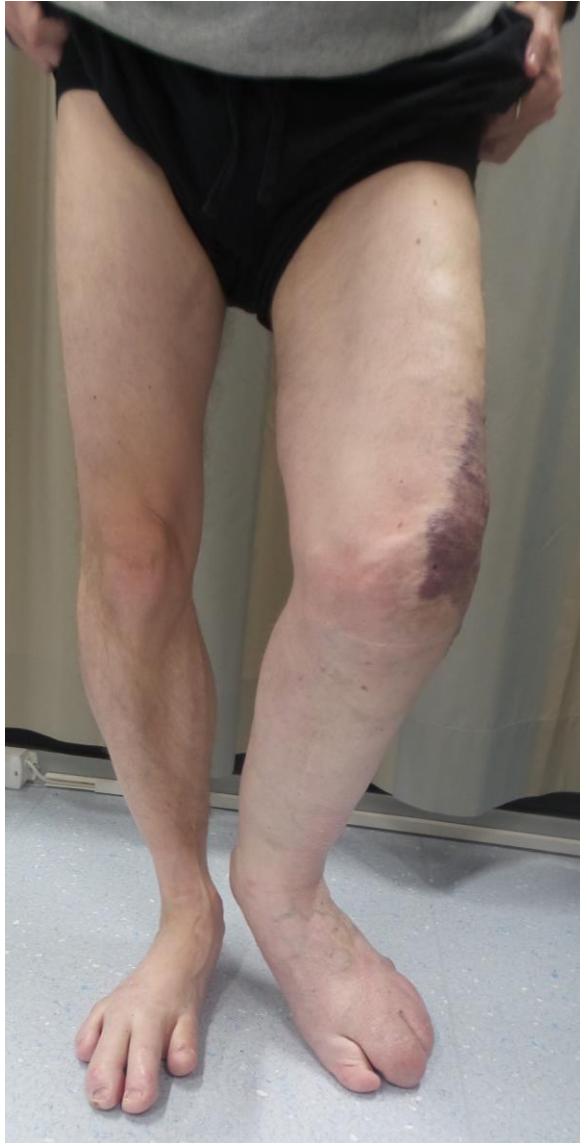
20-jährige Patientin, vaskuläre Malformation, Beinlängendifferenz links (operiert)



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)

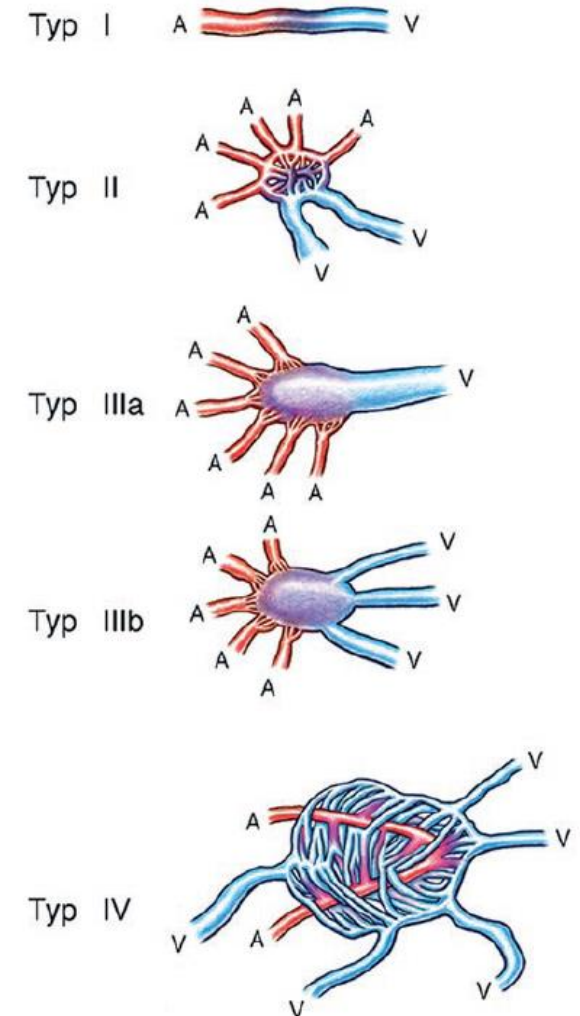
Arterio-venöse Malformation (AVM)

Table IV: Schobinger classification of AVMs [70]

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

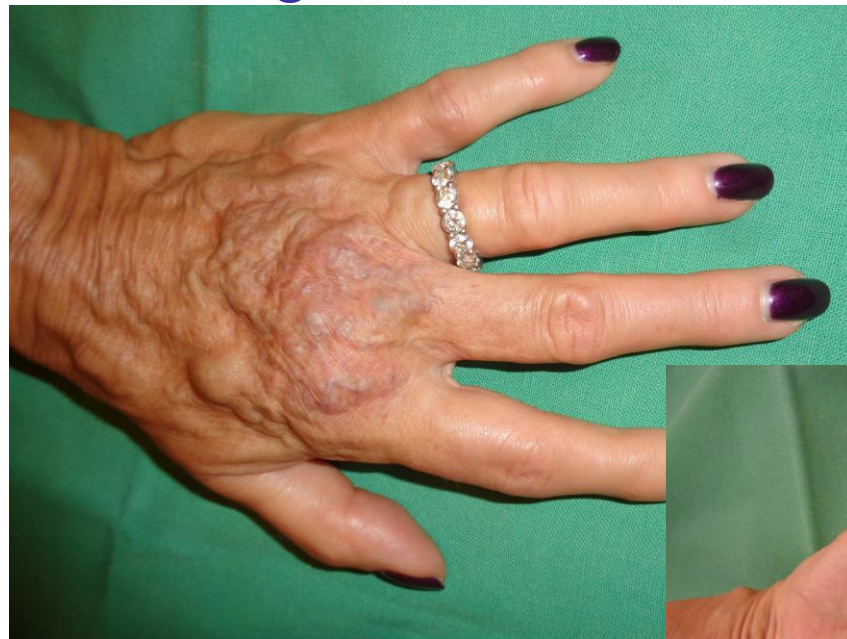
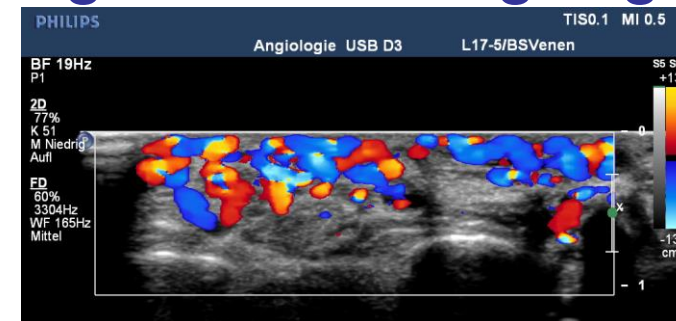
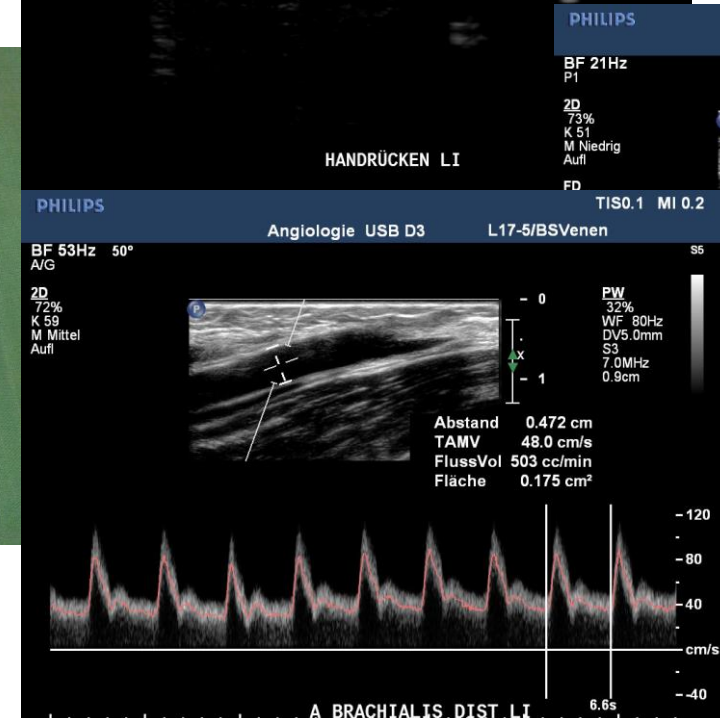
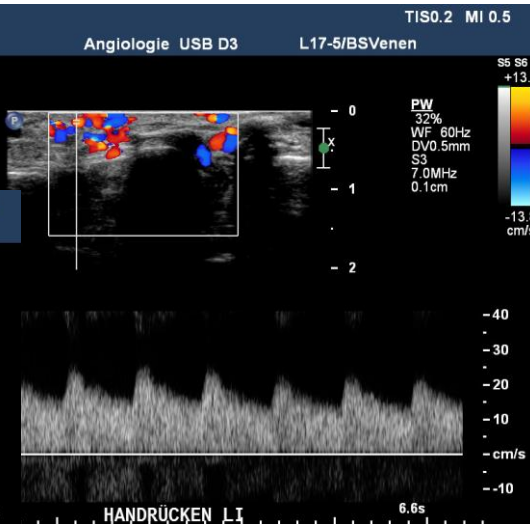
Table V: Angiographic classification of AVMs according to Cho et al. [71]

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



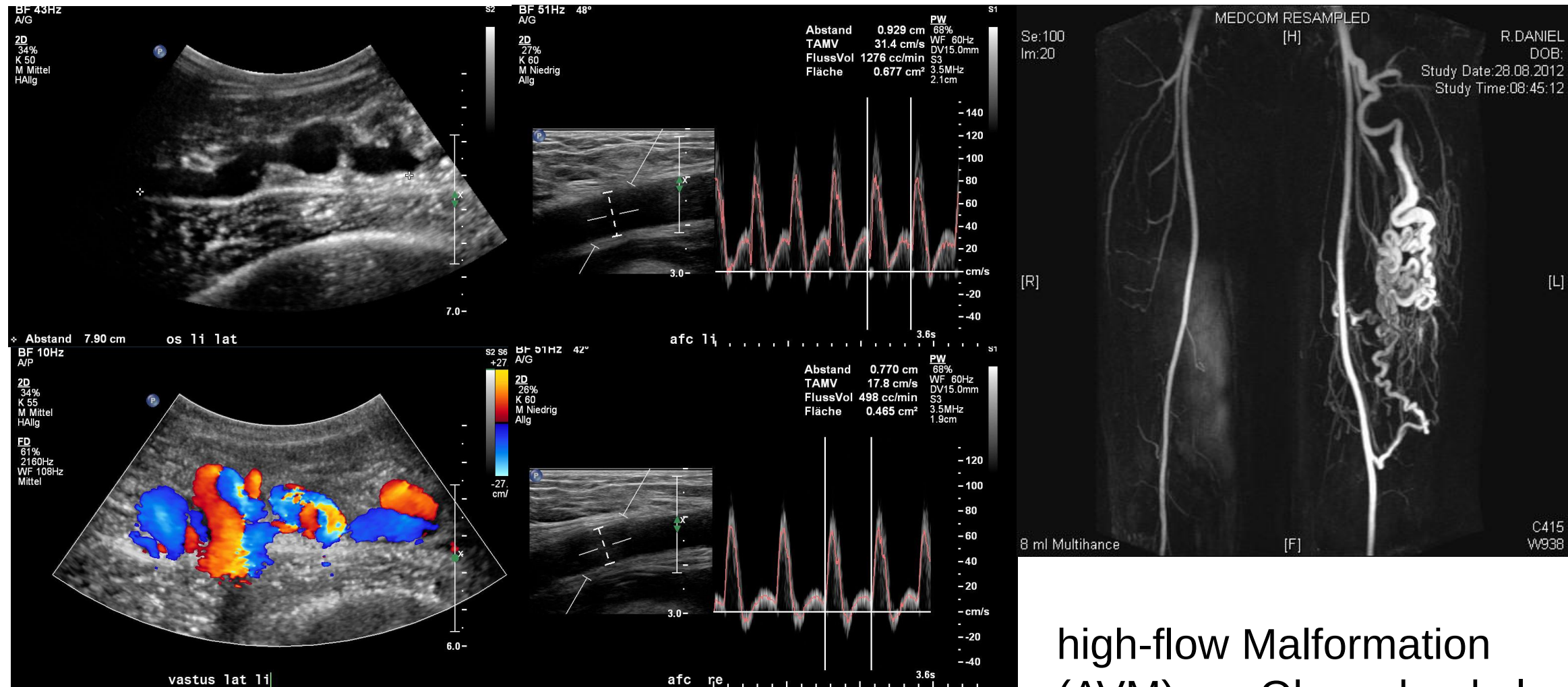
63-jährige Patientin, bekannte AVM der Hand, St.n. lokale Exzision und Embolisation,

Druckgefühl und zunehmendes Wärmegefühl, Bewegungseinschränkung



high-flow AV-Malformation (AVM) der
Hand

35-jähriger Patient, Schwellung und Schmerzen am Oberschenkel

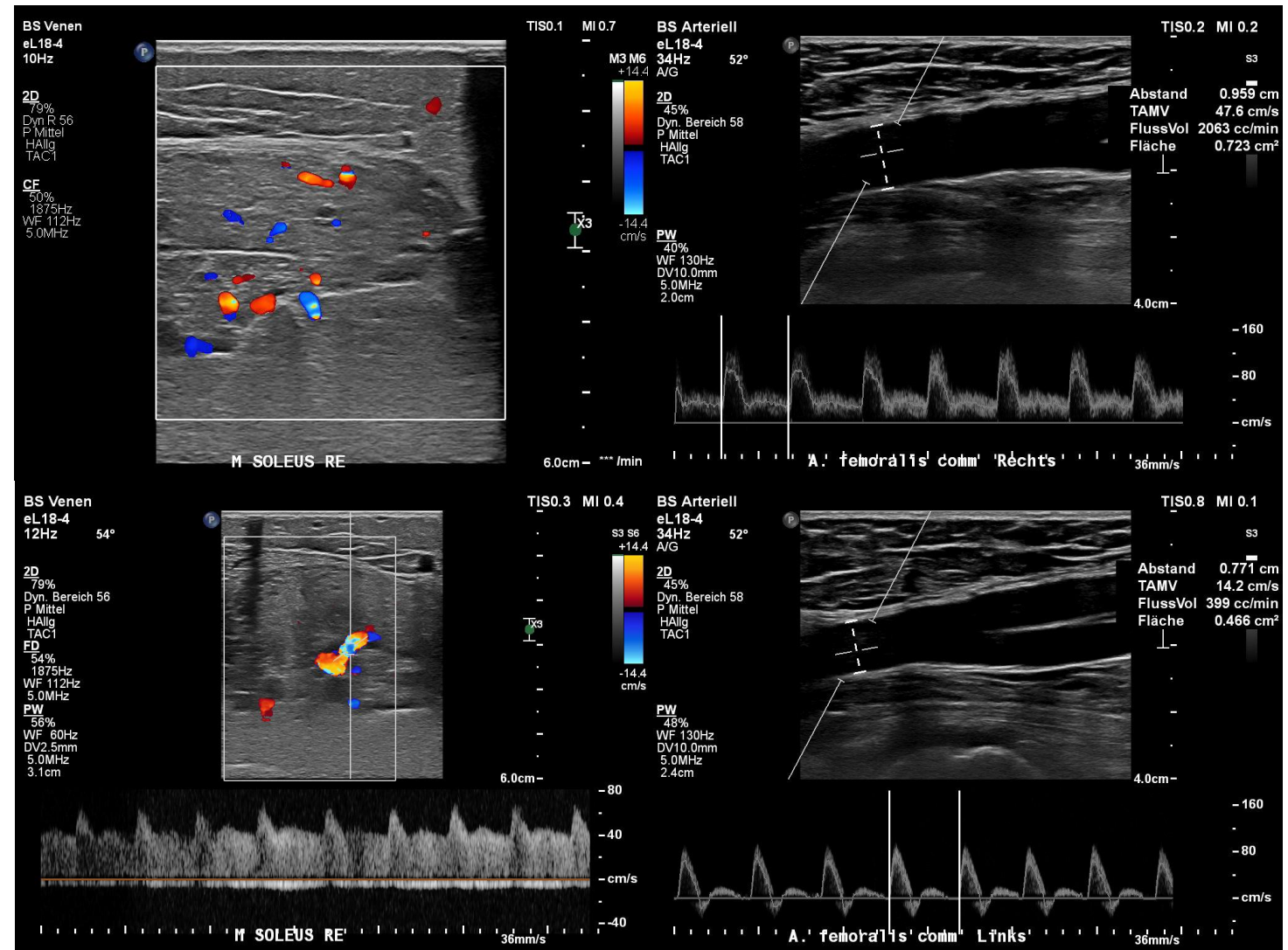


high-flow Malformation
(AVM) am Oberschenkel

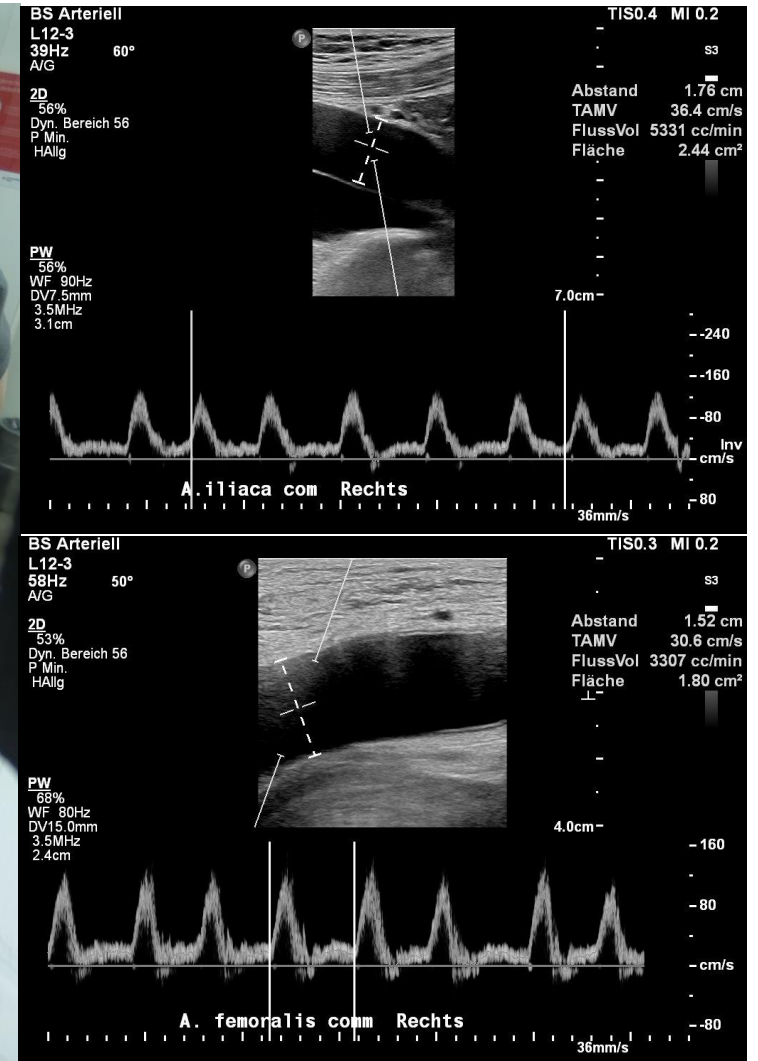
10-jähriger Patient, Beinlängendifferenz rechts, Umfangvermehrung, gut sichtbare oberflächliche Venen am rechten Bein



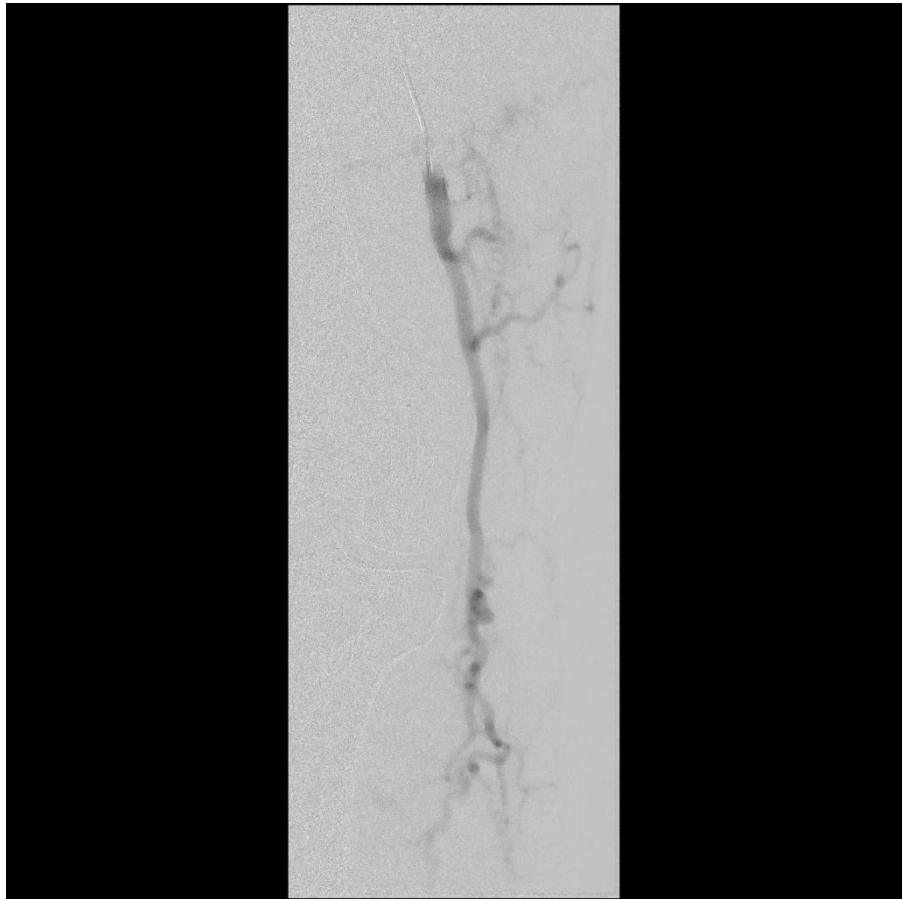
high-flow AV-Malformation,
diffus in der Wadenmuskulatur (AVM)



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 (RASA1 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!

