

Malformations vasculaires

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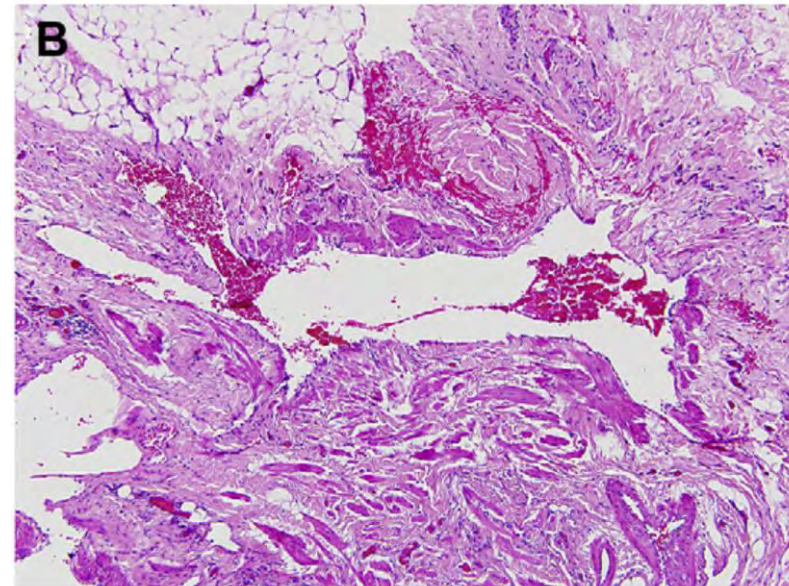
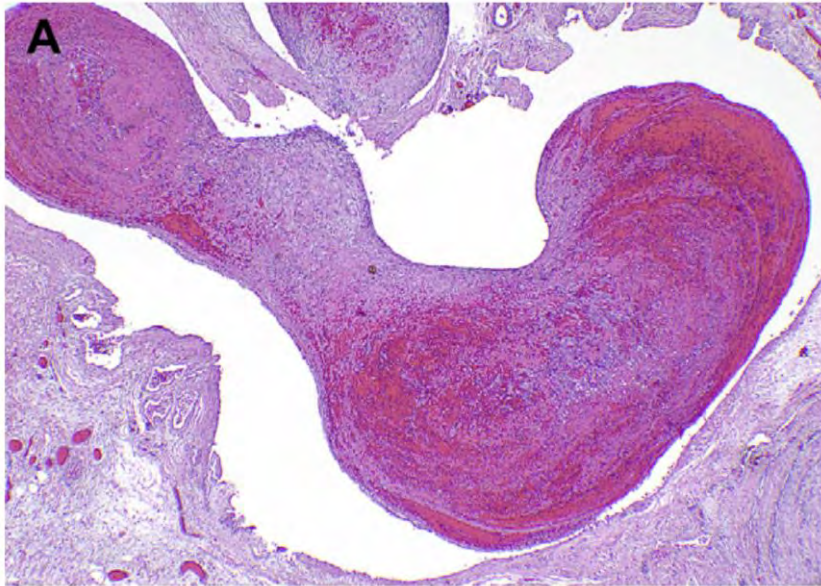
12e symposium valaisan des maladies vasculaires
7.9.2023

Malformation vasculaire

Formation congénitale anormale de vaisseaux sanguins, bordée de cellules endothéliales avec turnover normal

Dysplasie (\neq hyperplasie - tumeur vasculaire)

Prévalence de $\sim 1\%$



Objectifs

Définitions

Classification ISSVA

Approche diagnostique de la malformation veineuse (VASCERN)

Traitements systémiques spécifiques

Equipe multidisciplinaire

Définitions

«ANGIOME» – *just say no !*

Hémangiome caverneux → malformation veineuse

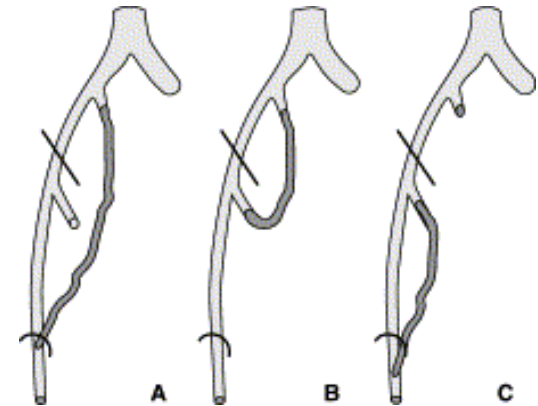
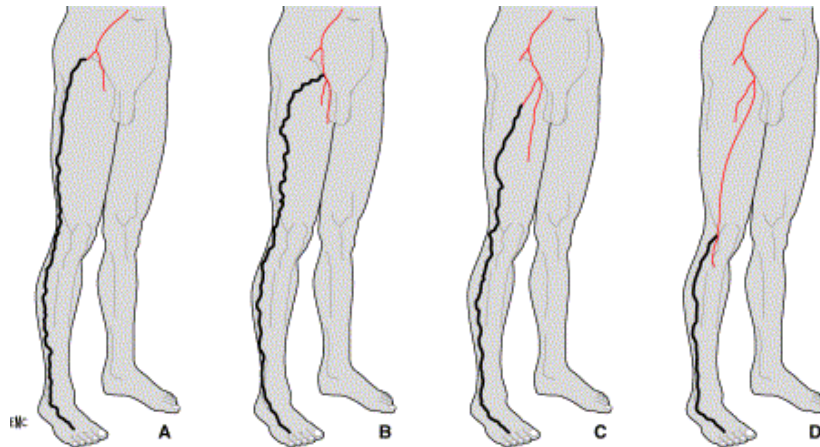
Anomalie EXTRATRONCULAIRE

Stade précoce de l'embriogénèse -> cluster de tissu vasculaire amorphe ne concernant pas les troncs vasculaires

Anomalie TRONCULAIRE

Stade tardif de l'embriogénèse, sur les vaisseaux déjà formés

Vaisseau supplémentaire ; aplasie ; anévrisme ; avalvulie





ISSVA classification for vascular anomalies ©

(Approved at the 20th ISSVA Workshop, Melbourne, April 2014, last revision May 2018)

This classification is intended to evolve as our understanding of the biology and genetics of vascular malformations and tumors continues to grow

Overview table

Vascular anomalies				
Vascular tumors	Vascular malformations			
	Simple	Combined °	of major named vessels	associated with other anomalies
Benign Locally aggressive or borderline Malignant	Capillary malformations Lymphatic malformations Venous malformations Arteriovenous malformations* Arteriovenous fistula*	CVM, CLM LVM, CLVM CAVM* CLAVM* others	See details	See list

° defined as two or more vascular malformations found in one lesion

* high-flow lesions

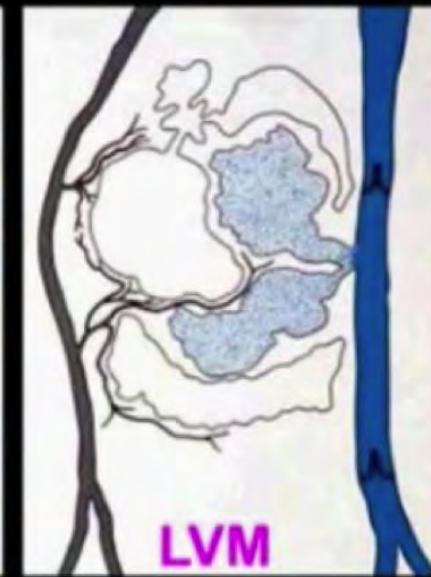
A list of causal genes and related vascular anomalies is available in [Appendix 2](#)

The tumor or malformation nature or precise classification of some lesions is still unclear. These lesions appear in a [separate provisional list](#).

[Abbreviations used](#)

For more details, click on the underlined links

Classification of Vascular Anomalies



Malformation vasculaire

```
graph TD; A[Malformation vasculaire] --> B[Simple]; A --> C[Combinée]; A --> D[Tronculaires]; A --> E[Associée (syndrome)];
```

Simple

Combinée

Tronculaires

Associée
(syndrome)

Simple

Capillaire

Lymphatique

Veineuse

Artério-veineuse



AP géographique
PIK3CA
malformations
veineuses+lymphatiques
hypertrophie

AP « tache de vin »
GNAQ
Malformations
veineuses
hypertrophie

Simple

Capillaire
(flux lent)

Lymphatique
(flux lent)

Veineuse
(flux lent)

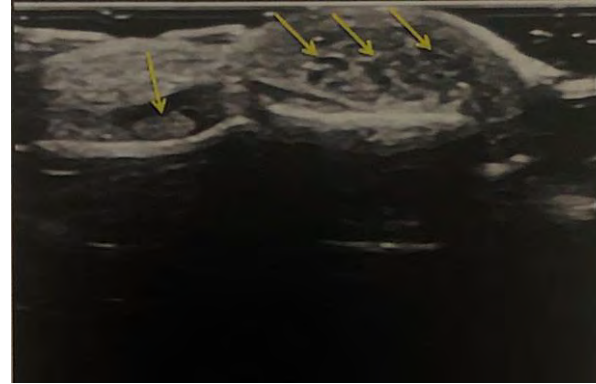
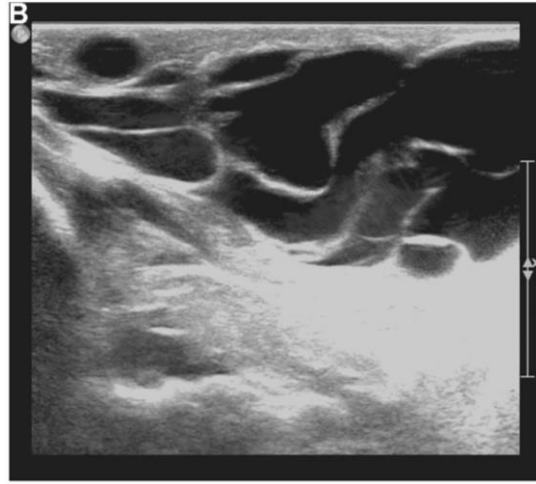
Artério-veineuse
(flux rapide)



Microkystique vs. Macrokystique (>1cm)

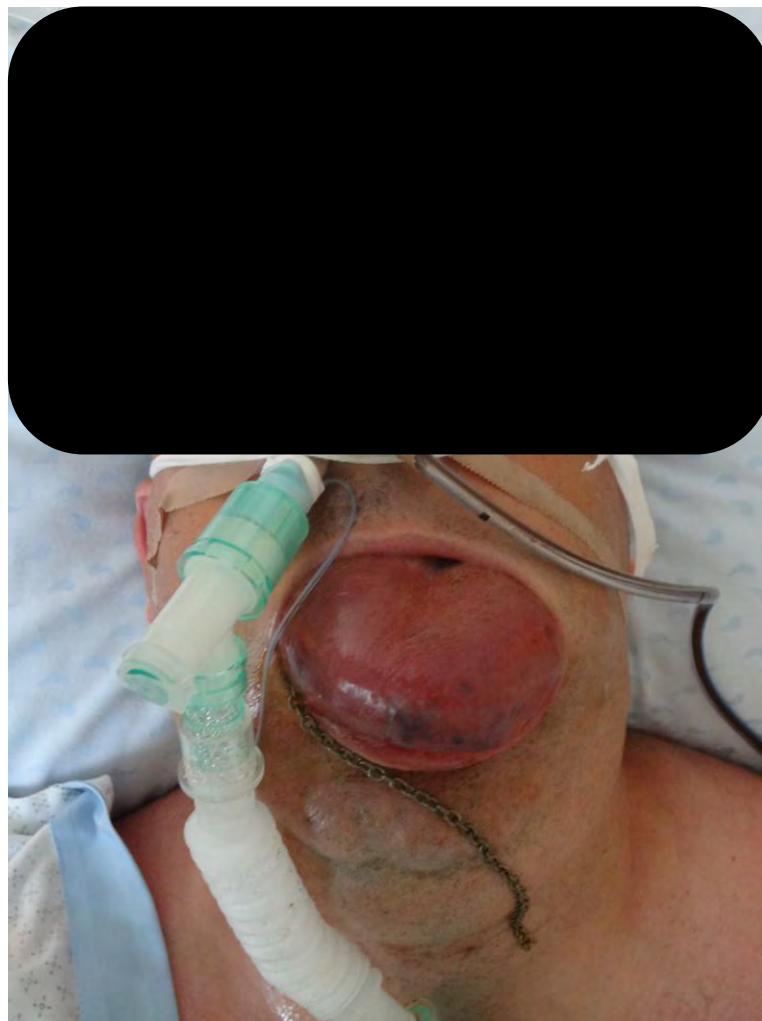
Malformation lymphatique simple

Tuméfaction incolore, non compressible, non fluctuante selon la position / Valsalva
Vésicules cutanées ou peau sp



Complications:

1. Hémorragie
2. Tuméfaction aiguë (para)-infectieuse
3. Compression des voies aériennes (naissance)



Malformation lymphatique simple

Traitement si symptomatique

1. Sclérothérapie

- Doxycycline
- Bléomycine (microkystique)
- Sodium Tetradecyl Sulfate (Thrombovar)
- Picibanil (OK-432)

2. Chirurgie

3. Traitement systémique (inhibiteur mTOR)

4. Laser (surface)

5. Laser (endovasculaire)

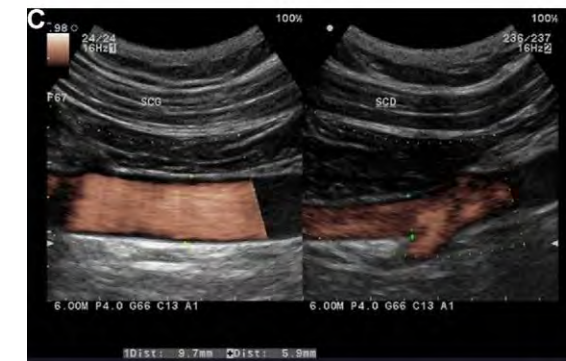
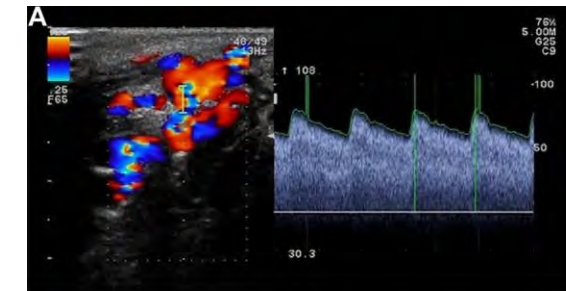
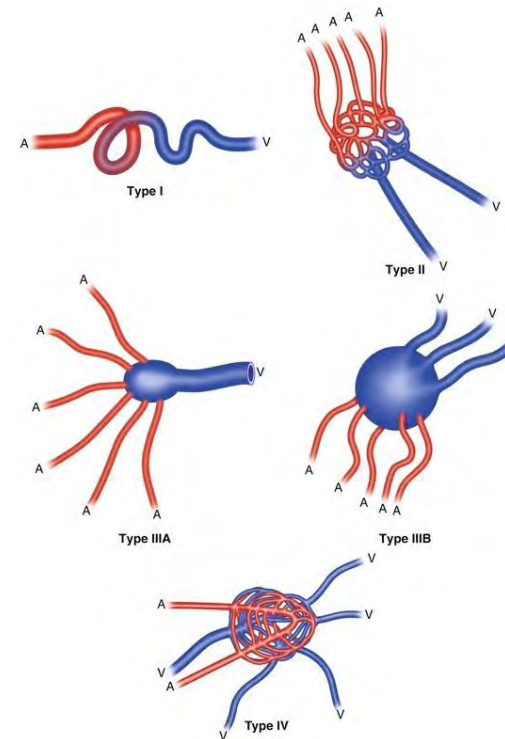
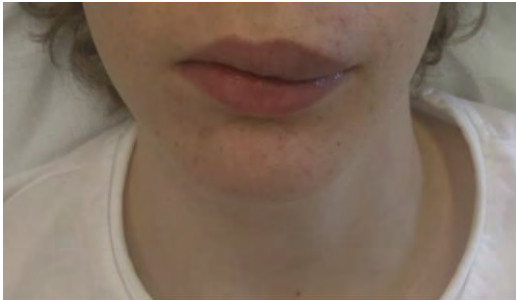
Simple

Capillaire
(flux lent)

Lymphatique
(flux lent)

Veineuse
(flux lent)

Artério-veineuse
(flux rapide)



Simple

Capillaire
(flux lent)

Lymphatique
(flux lent)

Veineuse
(flux lent)

Artério-veineuse
(flux rapide)



VASCERN

Gathering the best expertise in Europe
to provide accessible cross-border healthcare
to patients with rare vascular diseases



<https://vascern.eu/>

Suspected Venous Malformation

- Blue or skin coloured swelling; sometimes mucosal lesion
- Typically empties with compression and fills up in dependent position
- Painful at awakening or after exercise. For extended period if local thrombosis (1 to 2 weeks)
 - Sometimes : firm, painful on palpation and/or palpable phleboliths
 - No thrill, not warm
- Number of lesions, associated anomalies and/or limb hypo / hypertrophy
 - Family history

- Doppler ultrasound to confirm diagnosis & exclude fast flow
 - MRI if diagnosis unclear
- Biopsy rarely needed for differential diagnosis

Sporadic
Unifocal

A

Sporadic
Multifocal

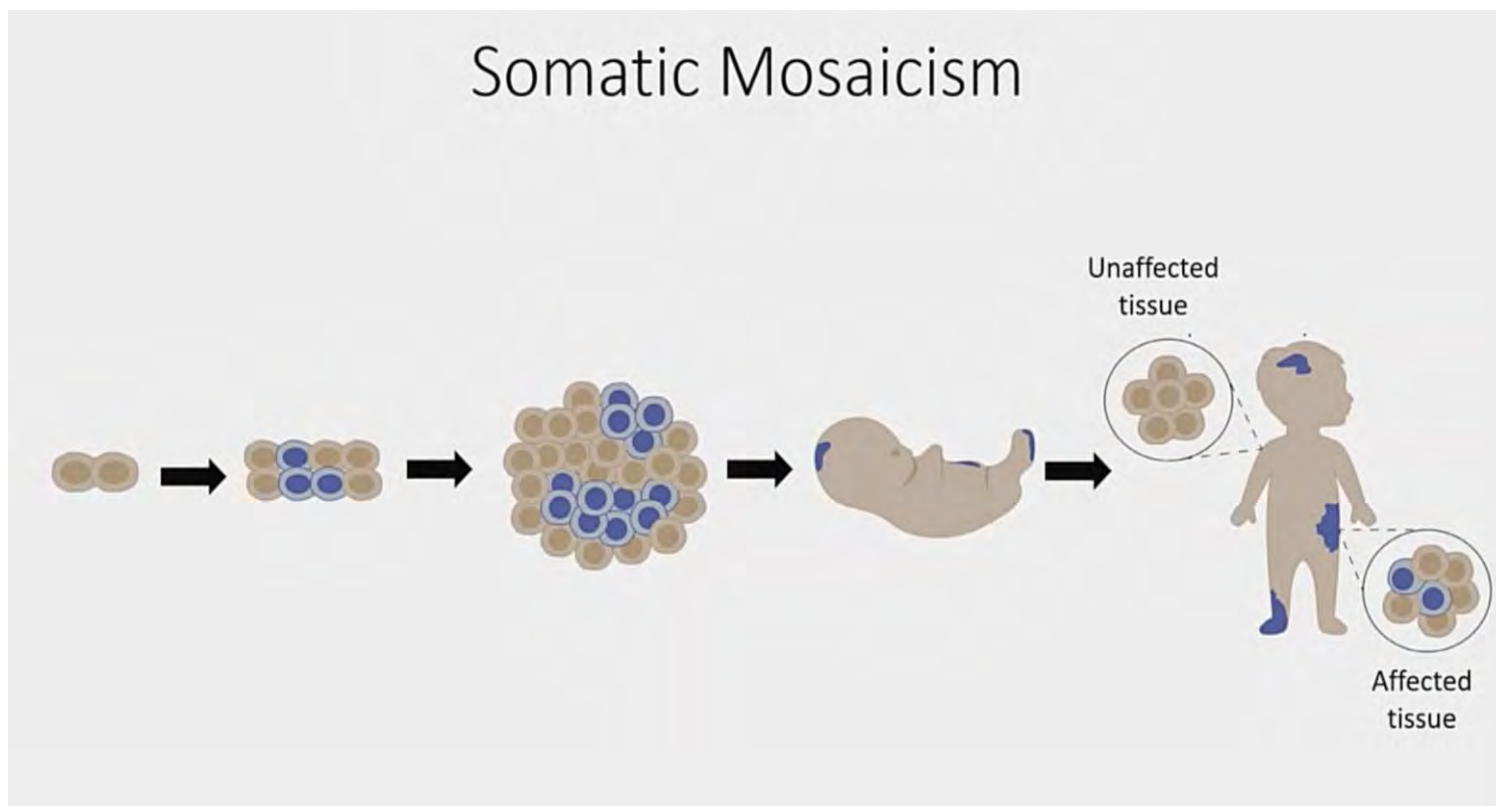
B

Familial
Multifocal

C

Combined and/or
Syndromic

D

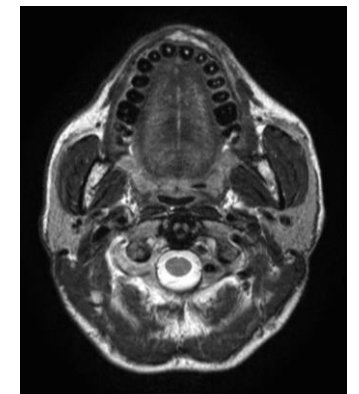
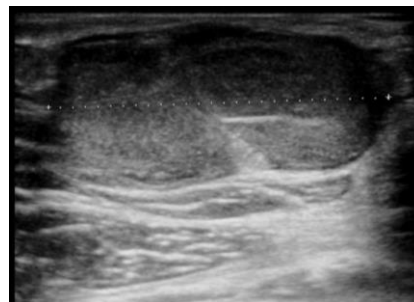
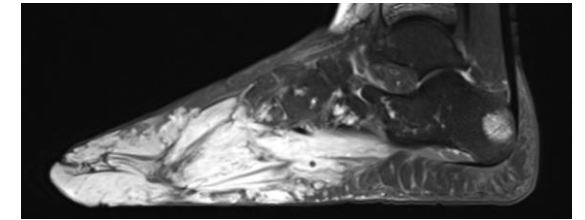
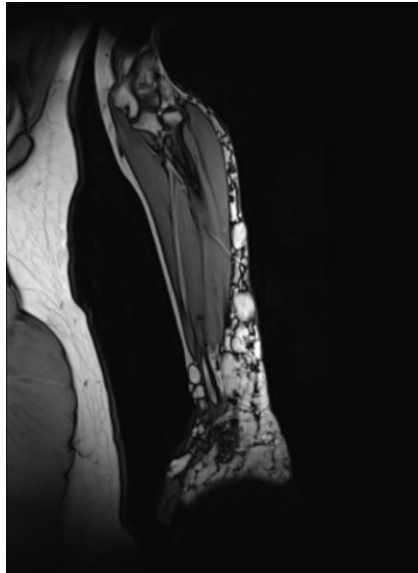


Sporadic
Unifocal
Majority of VMs
No family history

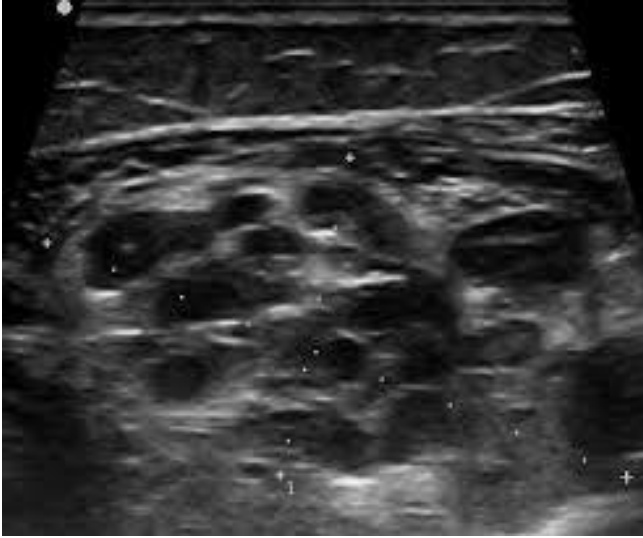
Somatic TEK or
PIK3CA

1

Sporadic Unifocal
Venous
Malformation



MV veineuses – écho-Doppler



Lacis veineux anéchogènes en amas (CAVE thrombose)

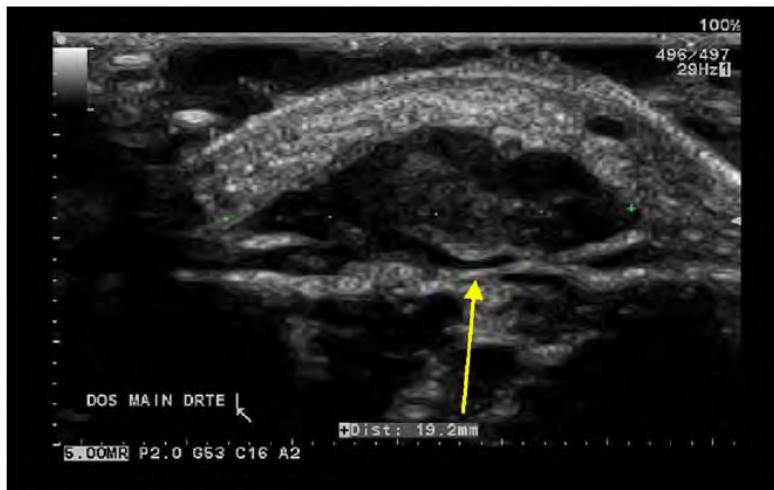
Dépressibles

Flux lents si compression

Phlébolithes

Bilan:

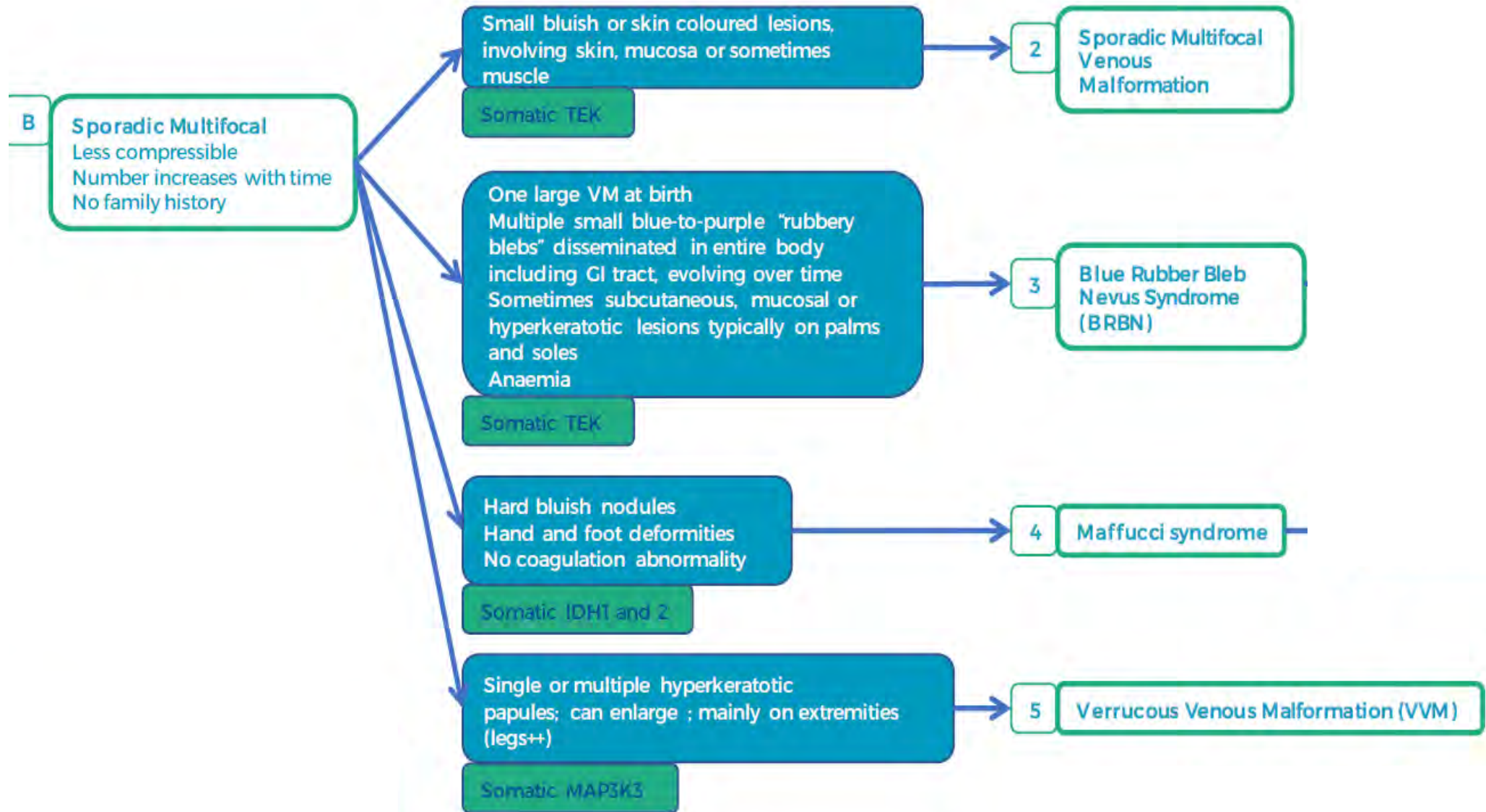
- veines de drainage
- anatomie vasculaire locale / du membre
- Dépistage 4 membres + cou (F)

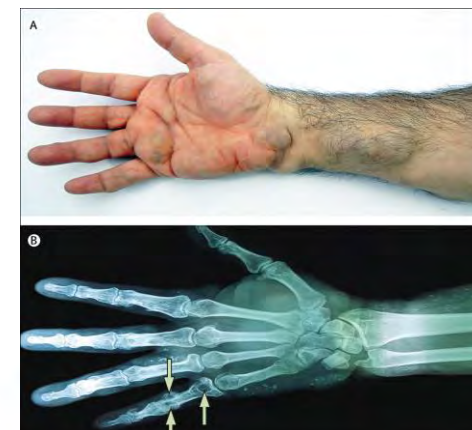
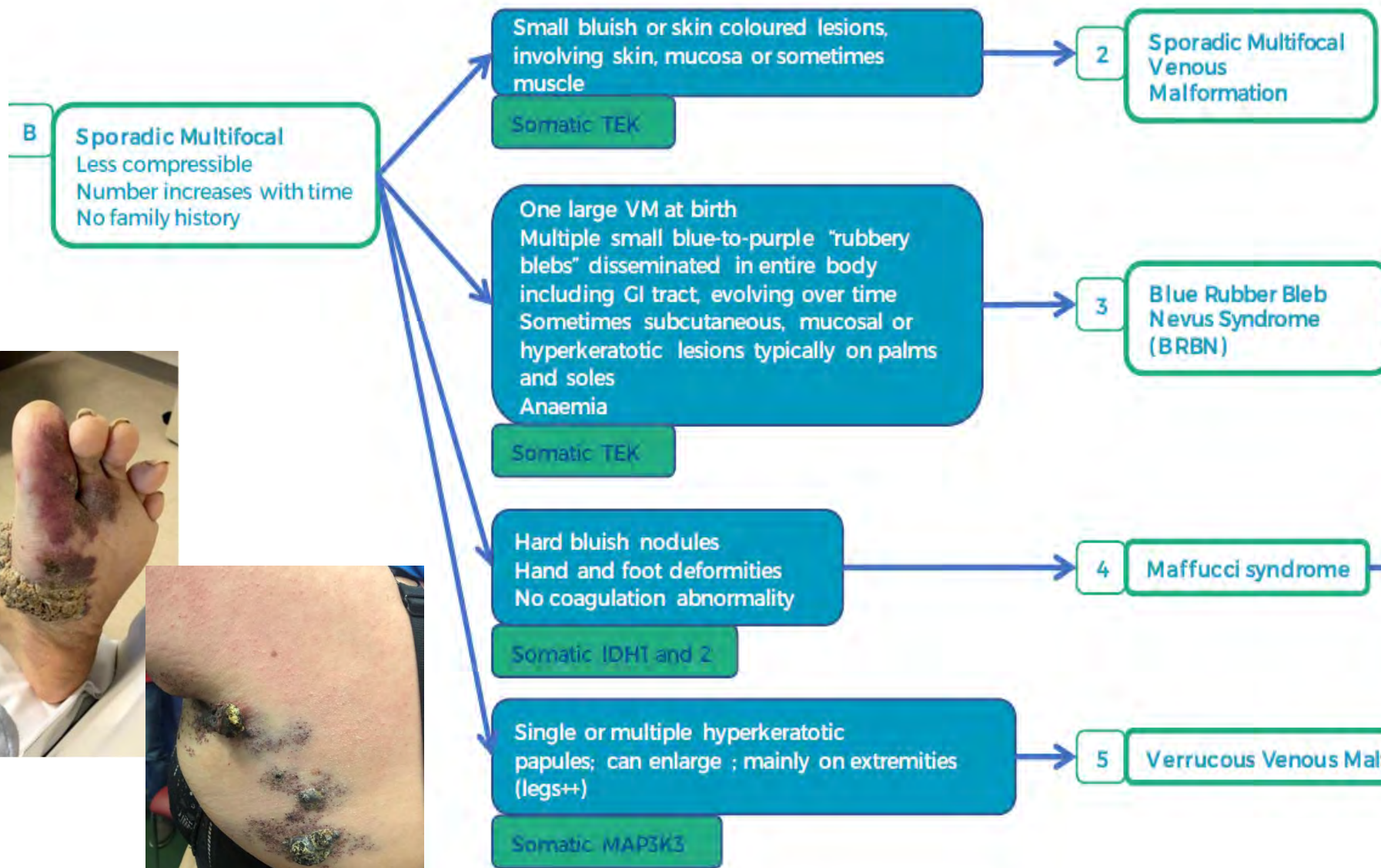


Sensibilité limitée par la profondeur / la taille de la lésion

Très explorateur-dépendant

En général insuffisant pour planifier le ttt





Blue rubber bleb nevus (BRBN) syndrome

1 grande MV + multiples MV circonscrites

«Rubbery»

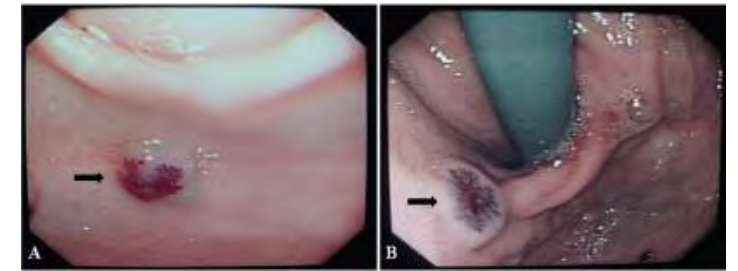
Extrémités / GI > muscle > organique > os /CNS

AF négative

± asymptomatique

Atteinte gastro-intestinale (bouche → anus)

- Spoliation chronique / hémorragie aiguë
- Intussusception / volvulus



Formes familiales



Malformations glomu-veineuses

Petites MV diffuses, souvent en plaques

Palpables, hyperkératosiques, «pavés»

Douloureuses

Absence d'atteinte mucosale

Génétique: mutation de *glomuline* (aut. dominant)

Ttt chirurgical (\neq compression)



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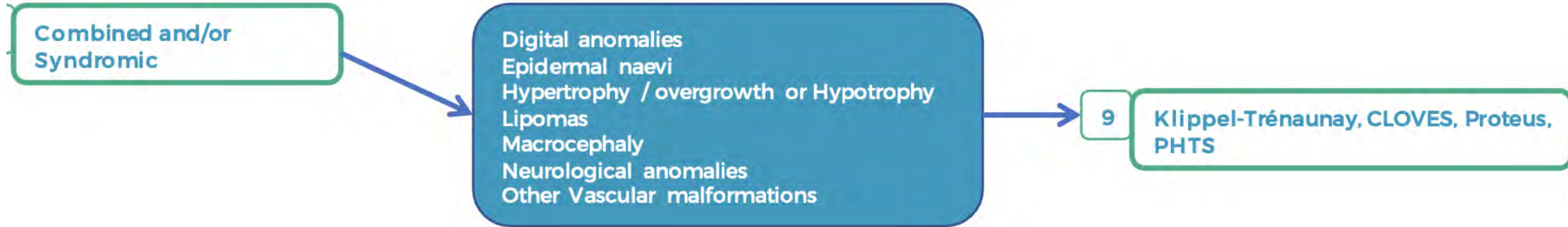
Familial
Multifocal

C

Combined and/or
Syndromic

D

Formes syndromiques



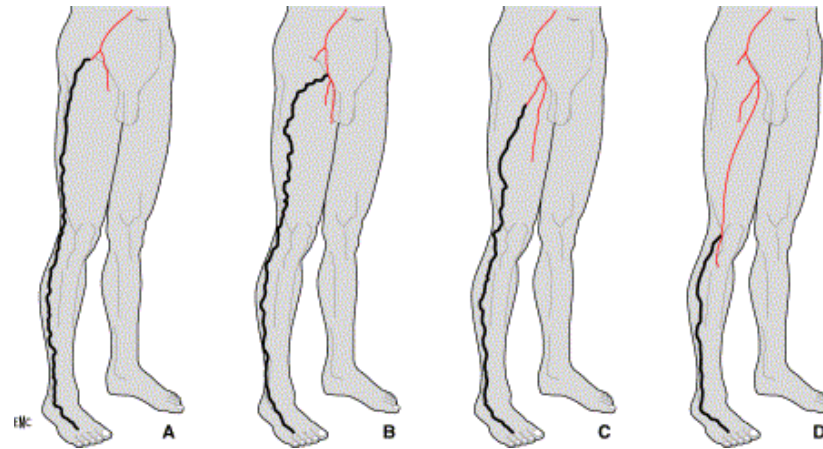


PIK3CA Related Overgrowth Syndromes (PROS)

Syndrome de **KLIPPEL-TRENAUNAY**

malformation capillaire + veineuse / lymphatique + hypertrophie

- Dépister la coagulopathie
- Dépister la veine marginale latérale (Serrvelle)



- Gestion de la grossesse (MTEV + HPP)

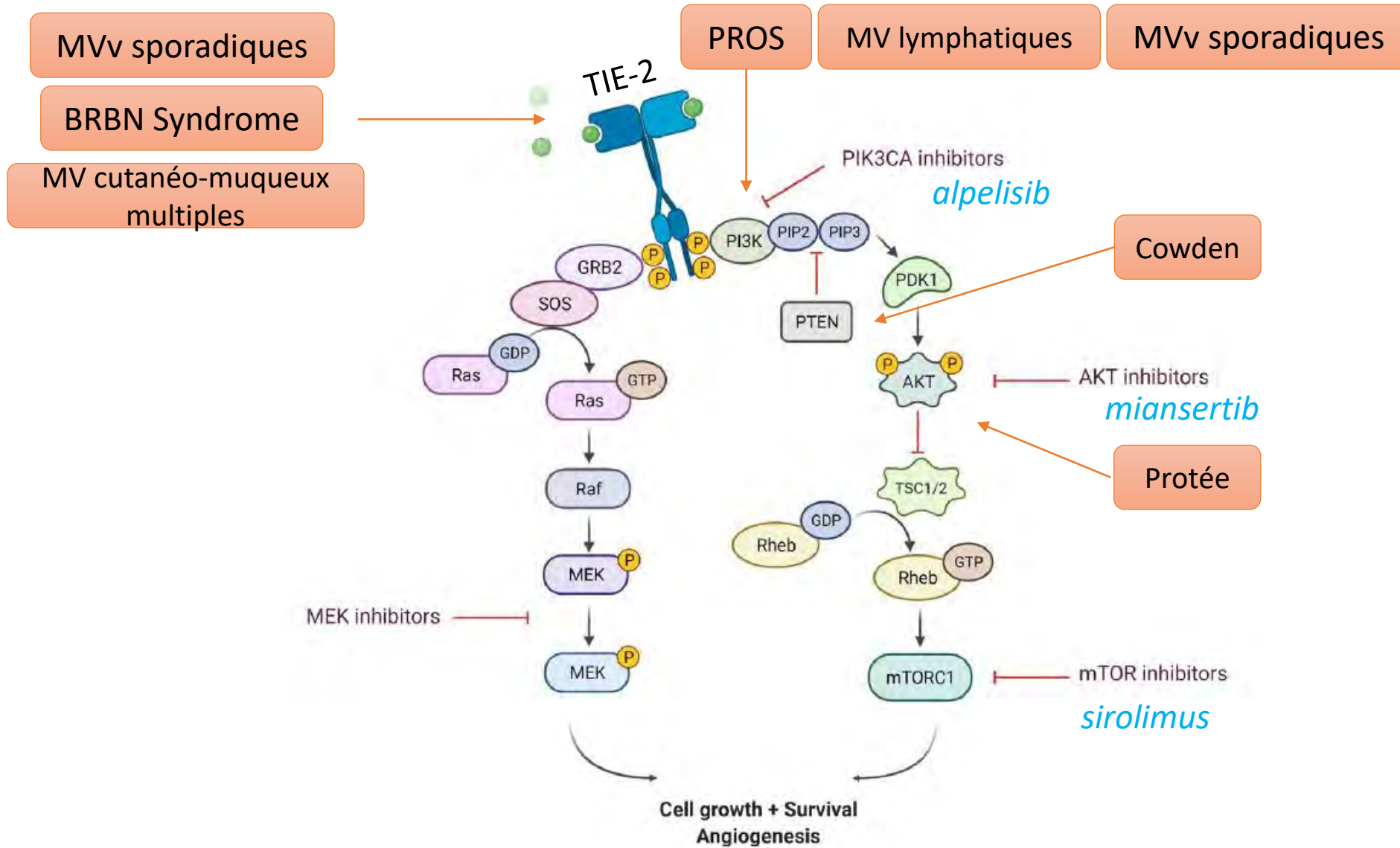
Malformation veineuse - traitement

Traitement si symptomatique

1. Sclérothérapie
 - Bléomycine
 - Sodium Tetradecyl Sulfate (Thrombovar), polidocanol, aetoxysclérol
 - polidocanol
 - Ethanol
2. Antithrombotiques (ASA, anticoagulants)
3. Compression
4. Chirurgie
5. Laser (surface)
6. Laser (endovasculaire)
7. Traitement systémique (inhibiteur mTOR)

TRAITEMENTS SPECIFIQUES

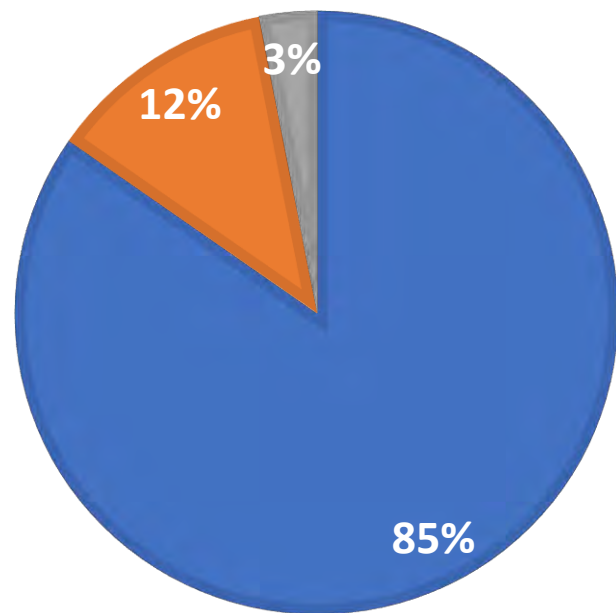
MV à flux lents



Efficacy and Safety of Sirolimus in the Treatment of Complicated Vascular Anomalies

Denise M. Adams, MD,^{a,b} Cameron C. Trenor III, MD, PhD,^c Adrienne M. Hammill, MD, PhD,^{a,b} Alexander A. Vinks, PhD,^{a,b} Manish N. Patel, DO,^{a,b} Gulraiz Chaudry, MBChB,^c Mary Sue Wentzel, MSN,^a Paula S. Mobberley-Schuman, MS,^a Lisa M. Campbell, MS,^a Christine Brookbank, MEd,^a Anita Gupta, MD,^{a,b} Carol Chute, APRN,^a Jennifer Eile, CPNP,^c Jesse McKenna, MPH,^c Arnold C. Merrow, MD,^{a,b} Lin Fei, PhD,^a Lindsey Hornung, MS,^a Michael Seid, PhD,^a A. Roshni Dasgupta, MD,^{a,b} Belinda H. Dickie, MD,^{a,b} Ravindhra G. Elluru, MD,^d Anne W. Lucky, MD,^a Brian Weiss, MD,^{a,b} Richard G. Azizkhan, MD^e

■ Réponse partielle ■ Progression ■ Stabilité ■



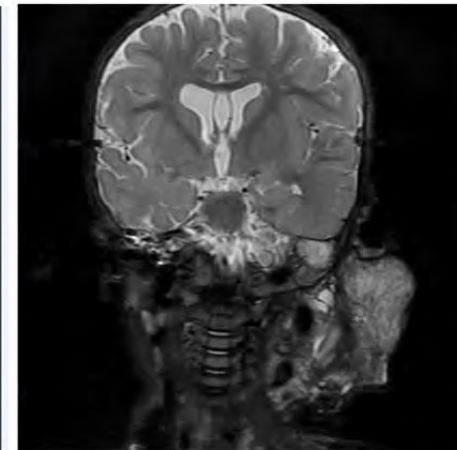
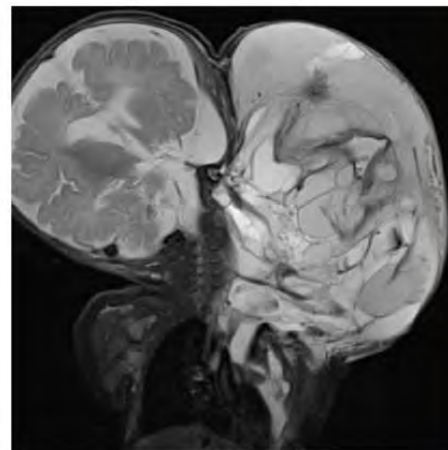
Generalized lymphatic anomaly ($n = 7$)
 Gorham syndrome ($n = 3$)
 Kaposiform lymphangiomatosis ($n = 7$)
 Microcystic lymphatic malformation ($n = 5$)
 KHE with KMP ($n = 10$)
 KHE without KMP ($n = 3$)
 Capillary lymphatico/venous malformation ($n = 13$)

Abnormalities of the central conducting lymphatic channels ($n = 3$)
 PTEN/AVM ($n = 2$)
 PTEN/overgrowth/VA ($n = 4$)
 Venous lymphatic malformation ($n = 3$)

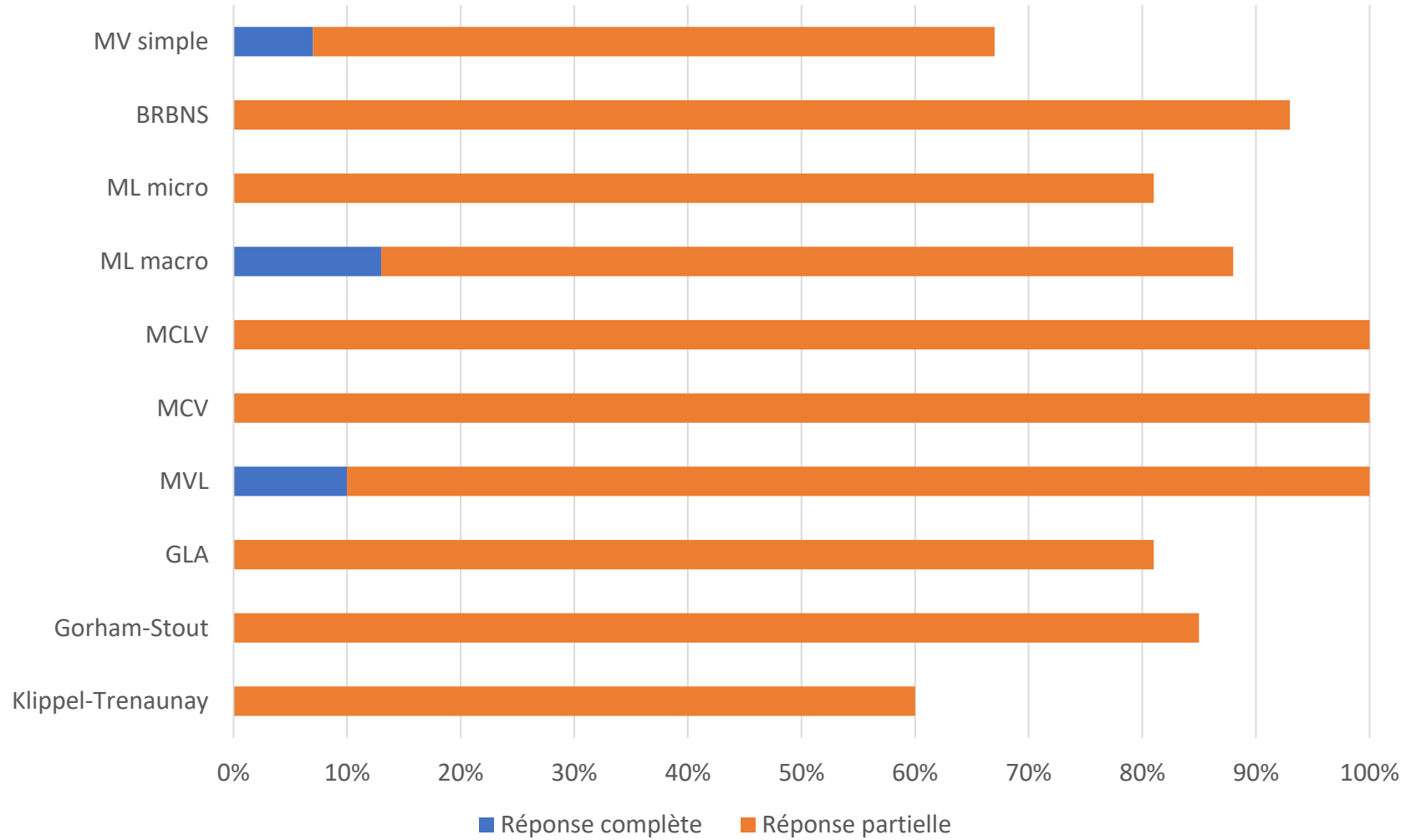
A

Prestudy

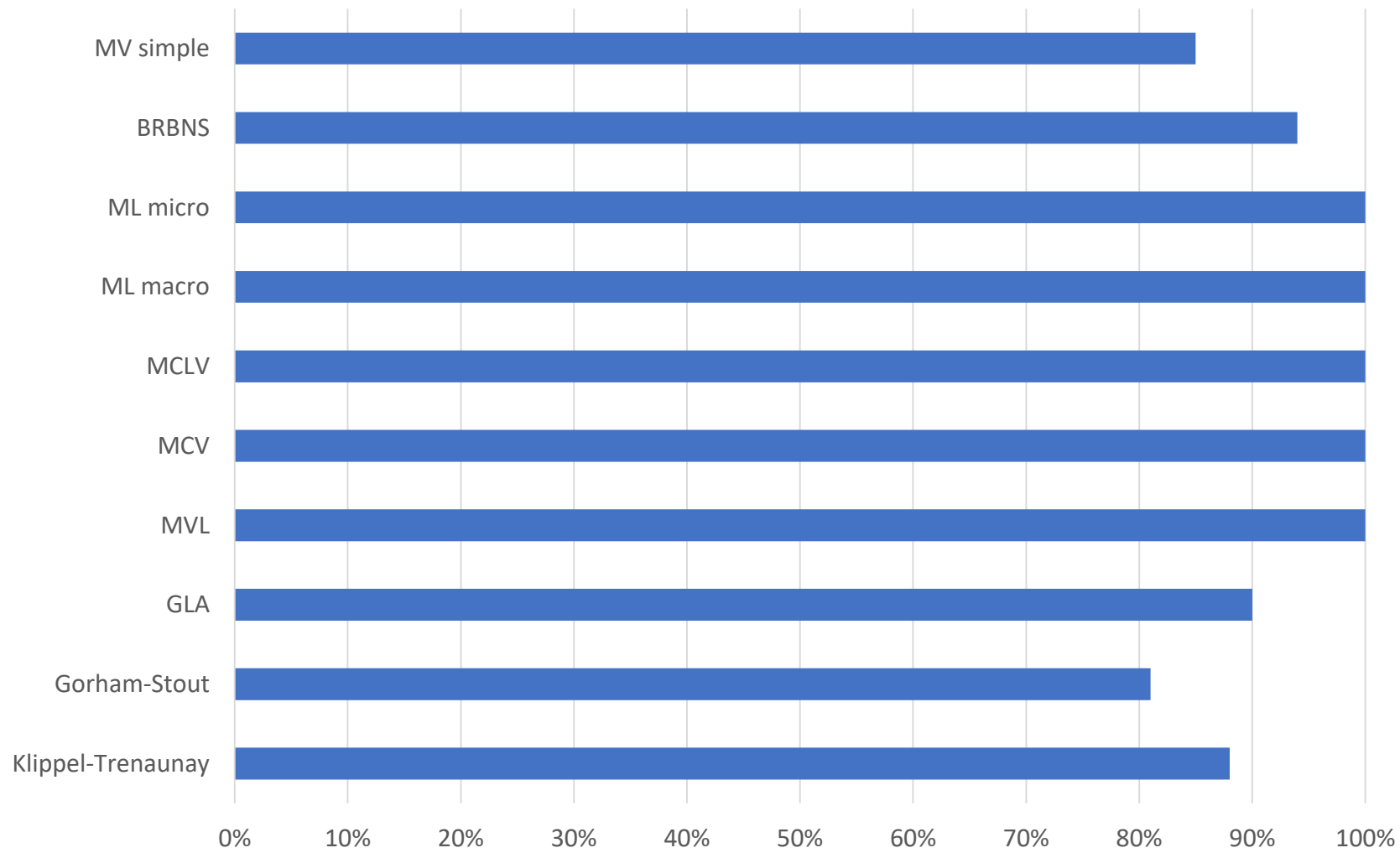
Poststudy



Taille (6 mois)



Symptômes (6 mois)



Restriction fonctionnelle
QoL
Douleur
Saignement
Infection

Sirolimus - tolérance

- 32% mucosite orale (aphtose)
- 17% dyslipidémie
- 12% leucopénie
- 10% symptômes digestifs (nausées-diarrhées)
- 8% rash-eczema
- 6% complication infectieuse

Céphalées

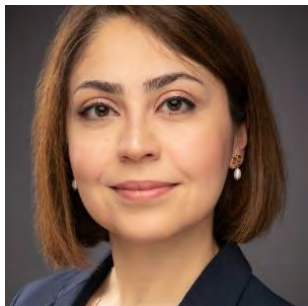
Sirolimus – indication

- Off-label → évaluation multidisciplinaire + demande assécurologique
- Malformations vasculaires à flux lent
- Malformations complexes
 - Ex: anomalie lymphatique généralisée
- Echec de traitement classique
 - Ex: MV veineuses étendues d'un membre
- MV et traitement classique risqués
 - Ex: MV lymphatiques ORL

Suivi intensif (1-3-6-9-12 mois)

Consultation multidisciplinaire MV - HUG

DERMATOLOGIE



Yassaman Alipour

NEURORADIOLOGIE



Hasan Yilmaz



Andrea Rosi

CHIRURGIE PLASTIQUE



Patricia Engels

CHIRURGIE PLASTIQUE PEDIATRIQUE



Giorgio La Scala

GENETIQUE



Géraldine Van Winkel

RADIOLOGIE



Matthieu Papillard

ANGIOLOGIE



Marc Blondon

PSYCHIATRIE



Alexis Berginc

Consultation mensuelle
80-100 patients/an

Consultation.angiologie@hcuge.ch

Consultation multidisciplinaire reconnue par l'International Society for the Study of Vascular Anomalies (2022)

Malformations vasculaires

Challenge des maladies rares

Terminologie à uniformiser («hémangiome»)

Clinique + US-Doppler +/- IRM +/- biopsie

Evaluation multidisciplinaire pour diagnostic et prise en charge

Révolution des traitements spécifiques

- sirolimus
- alpelisib
- MAV -> thalidomide / trametinib (anti-MEK)