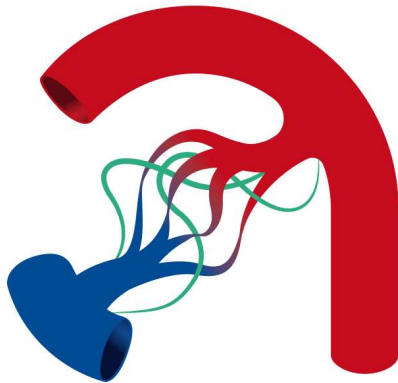




European Reference Network

for rare or low prevalence
complex diseases

 **Network**
Vascular Diseases
(VASCERN)



Venous malformation

Final Approved Patient Pathway by the Vascular
Anomalies (VASCA) Working Group – 29/04/2020

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Disclaimer

- This document is an opinion statement reflecting strategies put forward by experts and patient representatives involved in the Vascular Anomalies (VASCA) Rare Disease Working Group of VASCERN.
- It is preferable that patients be evaluated in a multidisciplinary center specialized in the diagnosis and management of vascular anomalies.
- This pathway is issued on 29/04/2020 and will be further validated and adjusted as needed.
- Responsibility for care of individual patients remains with the treating physician.

Dieses Dokument wurde am 05.12.2022 von Dr. Friedrich Kapp für das Zentrum für Gefäßfehlbildungen (Uniklinik Freiburg) für das FZSE Freiburg geprüft, es war keine Kommentierung notwendig. Eine gemeinsame Überprüfung und Diskussion wird unabhängig hiervon in VASCA-WG erfolgen.

Dieses Dokument wurde am 15.11.2023 von Dr. Friedrich Kapp für das Zentrum für Gefäßfehlbildungen (Uniklinik Freiburg) für das FZSE Freiburg geprüft und kommentiert. Eine gemeinsame Überprüfung und Diskussion wird unabhängig hiervon in der VASCA-WG erfolgen.

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Dieses Dokument wurde am 10.10.2024 von Dr. Friedrich Kapp für das Zentrum für Gefäßfehlbildungen (Uniklinik Freiburg) für das FZSE Freiburg geprüft und kommentiert. Eine gemeinsame Überprüfung und Diskussion wird unabhängig hiervon in der VASCA-WG erfolgen.



Suspected Venous Malformation

Histology :

- **VM** : dilated veins, sparse vascular smooth muscle cells
- **GVM** : veins surrounded by glomus cells
- **Maffucci syndrome** : spindle cell hemangioma
- **VVM**: clusters of venus-like channels, GLUT 1+

VM :Venous Malformations
 GVM: Glomuvenous Malformations
 VVM: Verrucous Venous Malformations

- 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

Sporadic Multifocal

Familial Multifocal

Combined and/or Syndromic

A

B

C

D

LEGEND:

Clinical evaluation

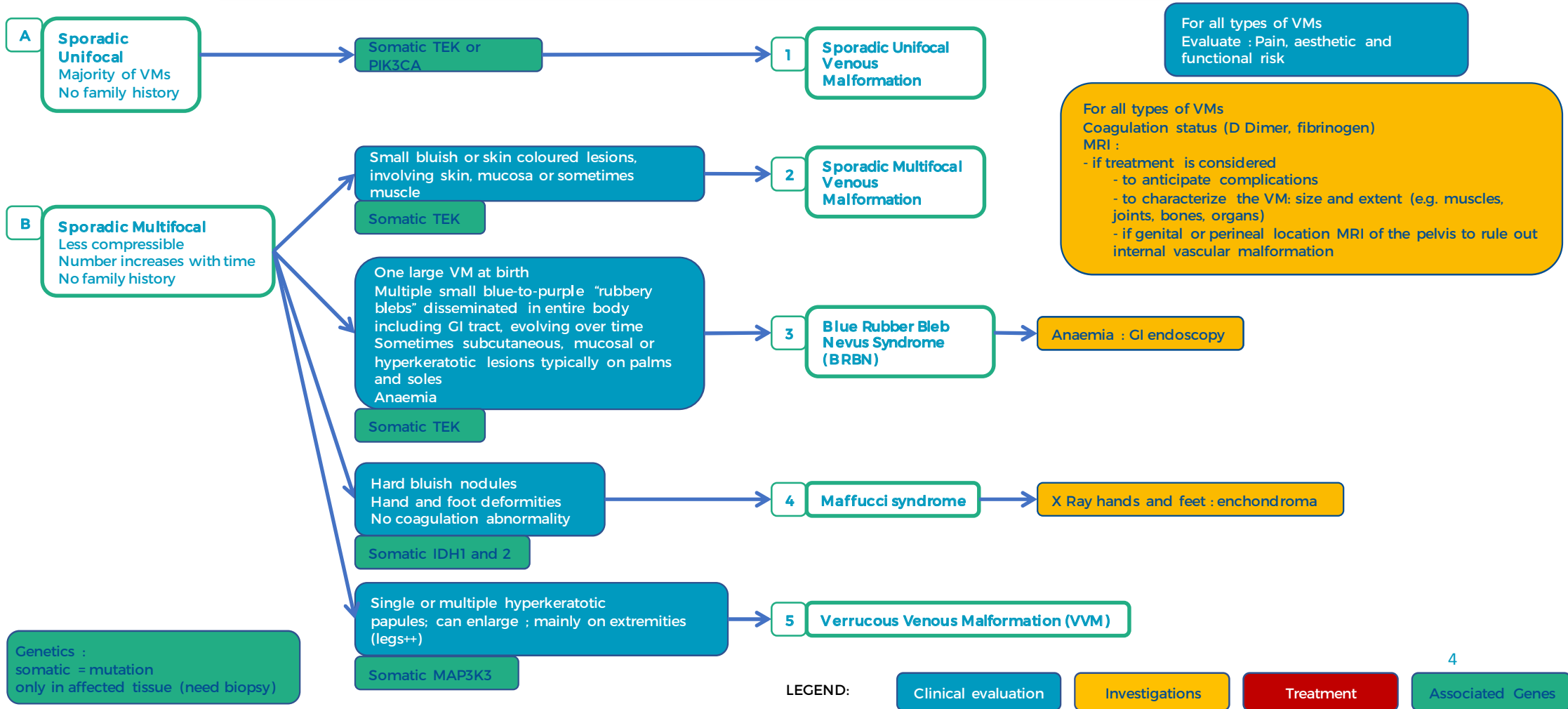
Investigations

Treatment

Associated Genes

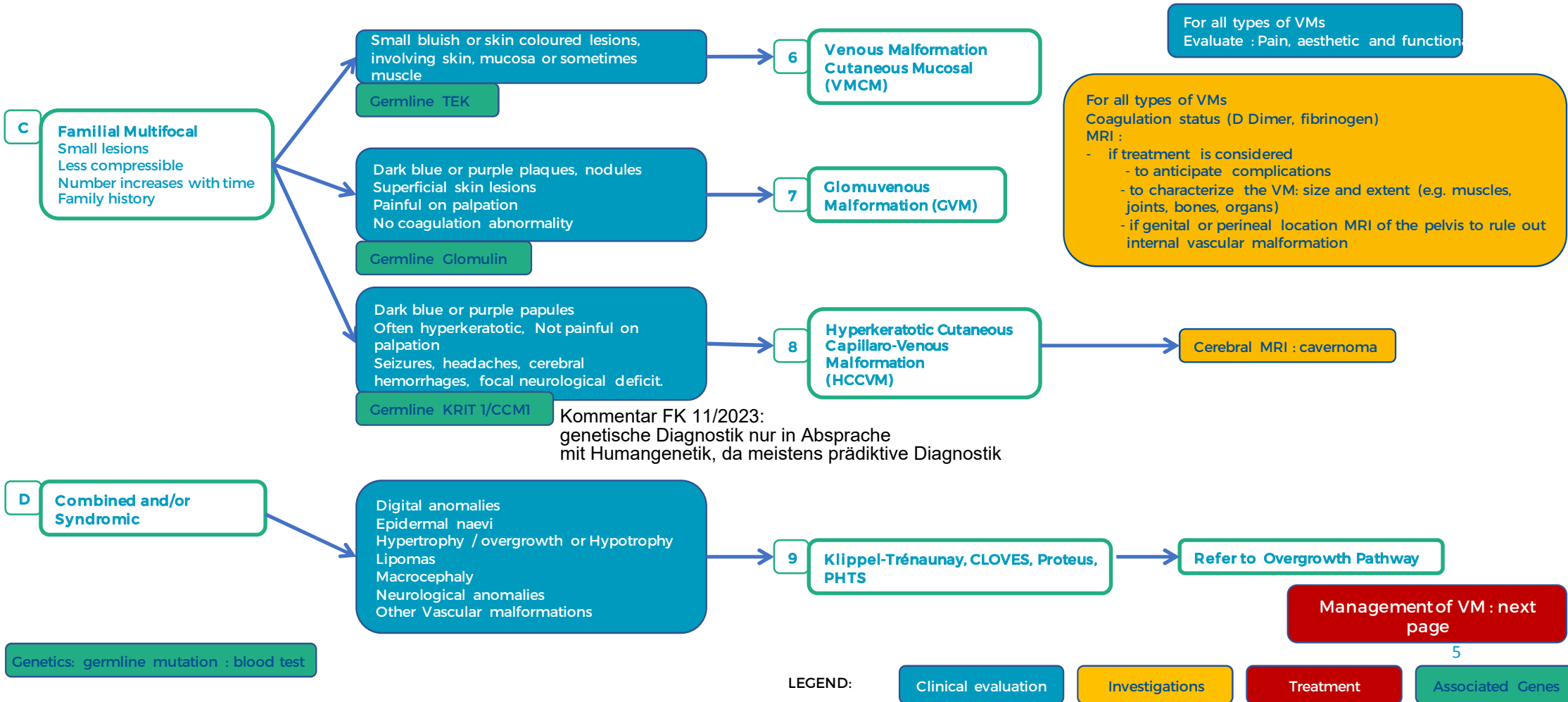


Venous Malformation Diagnostic Work-Up continued





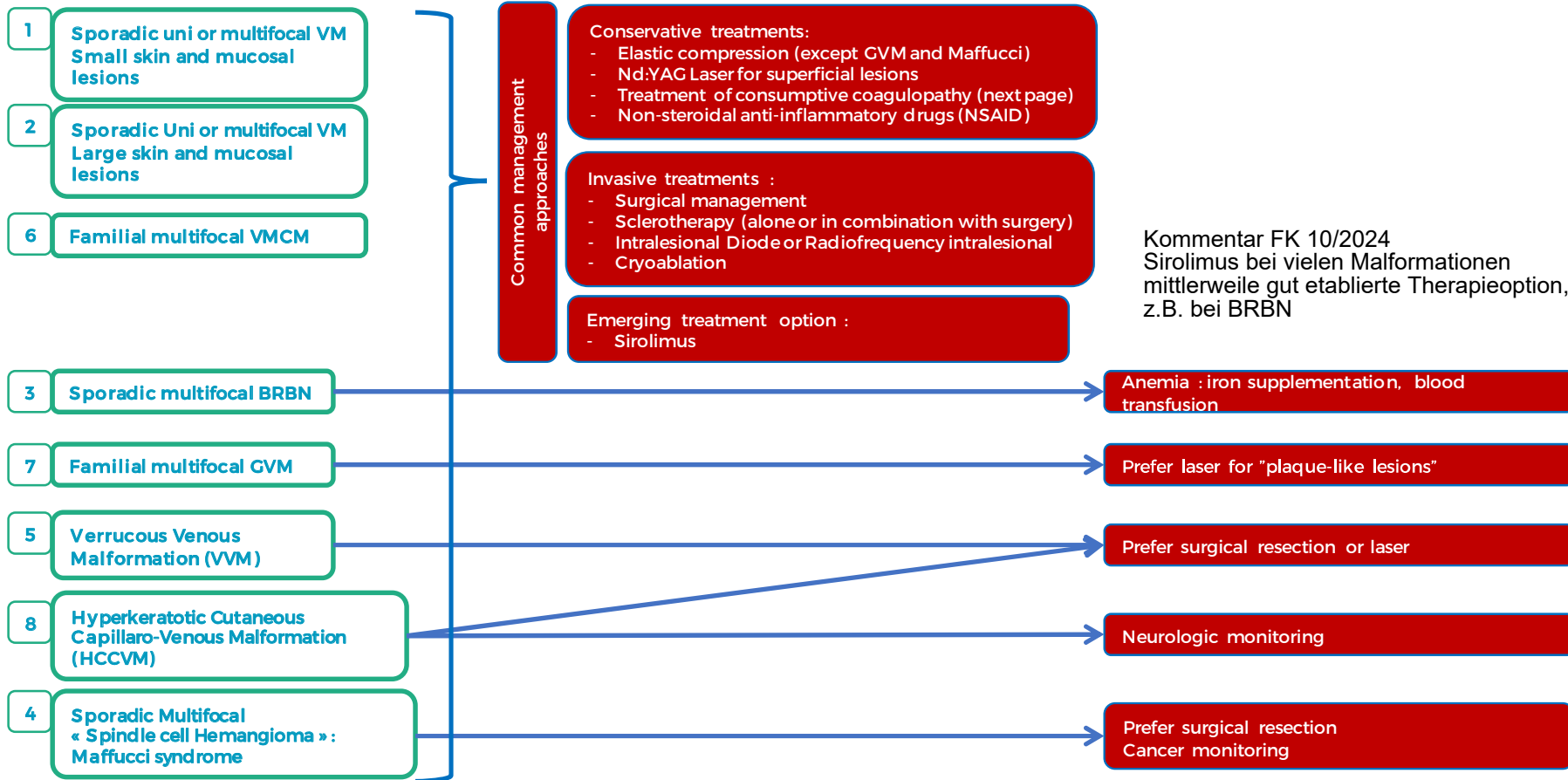
Venous Malformation Diagnostic Work-Up continued



Kommentar FK 11/2023:
genetische Diagnostik nur in Absprache mit Humangenetik, da meistens prädiktive Diagnostik



Venous Malformation Management/Treatment



Kommentar FK 10/2024
Sirolimus bei vielen Malformationen mittlerweile gut etablierte Therapieoption, z.B. bei BRBN

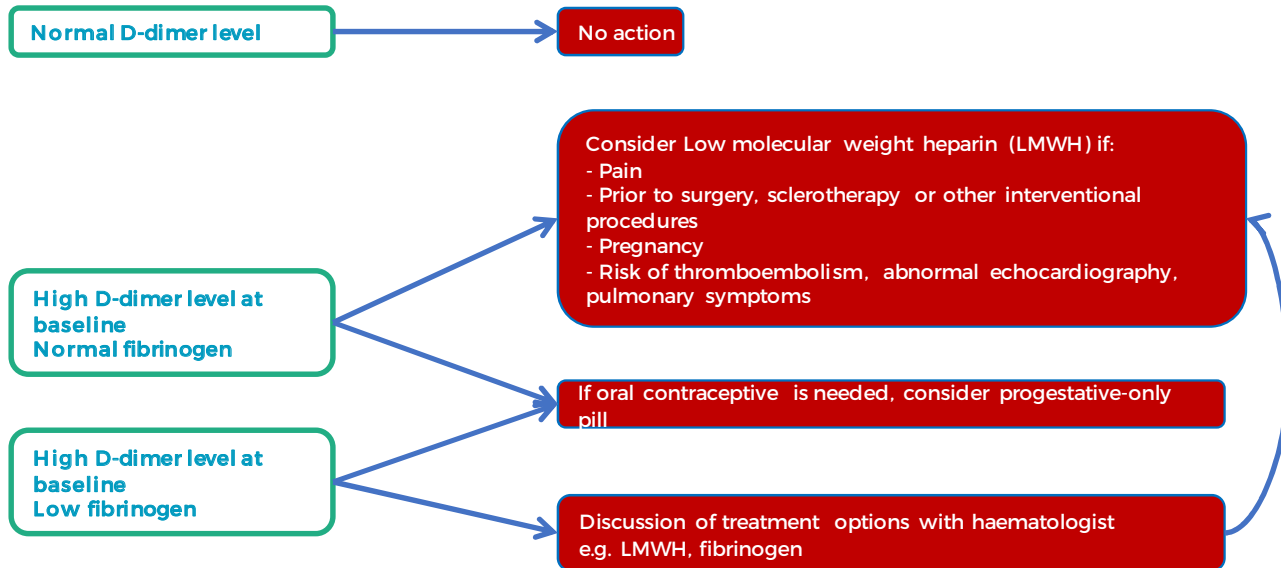
Kommentar FK 11/2023:
cMRT wenn suggestive Symptomatik, bei asymptomatischen Patienten nach gemeinsamer Abwägung mit Patienten

LEGEND:

Clinical evaluation
Investigations
Treatment
Associated Genes



Chronic Consumptive Coagulopathy Management



LEGEND:

Clinical evaluation

Investigations

Treatment

Associated Genes



VASCERN

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



VASCERN, the European Reference Network on Rare Multisystemic Vascular Diseases, is dedicated to gathering the best expertise in Europe in order to provide accessible cross-border healthcare to patients with rare vascular diseases (an estimated 1.3 million concerned). These include arterial diseases (affecting aorta to small arteries), arterio-venous anomalies, vascular malformations, and lymphatic diseases.

VASCERN currently consists of 30 highly specialised multidisciplinary Healthcare Providers (HCPs) from 11 EU Member States and of various European Patient Organisations and is coordinated in Paris, France.

Through our 5 Rare Disease Working Groups (RDWGs) as well as several thematic WGs and the ePAG- European Patient Advocacy Group, we aim to improve care, promote best practices and guidelines, reinforce research, empower patients, provide training for healthcare professionals and realise the full potential of European cooperation for specialised healthcare by exploiting the latest innovations in medical science and health technologies.

More information available at: <https://vascern.eu>

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