Capillary 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 specialising 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.
Capillary malformation (Vascular stain) Diagnostic Work-Up

Start

- Multiple oval to round small lesions +/- peripheral halo
  - Family history of similar lesions
  - Fast flow lesions
  - Consider baseline cranial and spinal MRI to check for AVM
  - Germline genetic testing
  - RASA1, EPHB4

- Microcephaly
  - Multiple oval to round, small, prominent, dark red lesions
  - Neurology
  - Germline genetic testing
  - STAMBP

- Linear or reticulated purple stain with skin atrophy
  - Cutis marmorata telangiectatica congenita (CMTC)

- Warm, pulsatile stain with bruit
  - Arteriovenous malformation (AVM)

- Pink to red-purple larger macules
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Capillary malformation (Vascular stain) Diagnostic work up continued

- **Pink to red-purple larger macules**
  - **Lateralized or bilateral +/- midline separation**
    - **YES**
      - Early referral to ophthalmology and neurology
      - Cranial MRI at 1-2 years of age or if symptomatic
    - **ABNORMAL**
      - Sturge-Weber Syndrome
        - GNAQ
    - **NORMAL**
      - Regular ophthalmological check up

- **Face**
  - **Not V-shaped, atypical, darker and wider**
    - **YES**
      - Cranial MRI
      - Neurology
      - Mosaic genetic testing
    - **NO**
      - Non-syndromic capillary malformation (Port-wine stain)
        - GNAQ
      - Megalencephaly-capillary malformation (MCAP)
        - PIK3CA

- **OR**

- **Elsewhere**
  - **Lower lip**
    - Check for oral/neck lymphatic anomalies and local overgrowth.
  - **Continues on page (5)***

- **Central face (glabella, philtrum)**
  - **YES**
    - Cranial MRI
    - Neurology
    - Mosaic genetic testing
  - **NO**
    - **ABNORMAL**
      - Nevus simplex (salmon patch)
    - **NORMAL**
      - CLAPO syndrome
        - PIK3CA

*Continues on page (5)*
Capillary malformation (Vascular stain) Diagnostic work up continued

Elsewhere

Trunk/Extremities

- Hypotrophy
- Overgrowth
- Normal growth

OR

- Vertex, nape, lumbosacral area

Capillary malformation with hypotrophy

Hypotrophy

- Overgrowth

Overgrowth

- Normal growth

Normal growth

Non-syndromic capillary malformation (Port-wine stain)

GNAQ

No other abnormalities but may have digital abnormalities

- Macrocephaly
- Lateral marginal veins
- Lymphatic vesicles
- Venous malformations
- +/- Lymphedema

Macrocephaly

Lateral marginal veins

Lymphatic vesicles

Venous malformations

+/- Lymphedema

Klippel-Trénaunay syndrome (KTS)

- Digital abnormalities
- Scoliosis
- Lipomatosis
- Other vascular malformation(s)

PIK3CA

CLOVES or PROS not otherwise specified

PIK3CA

Cerebriform connective tissue nevus

+/- Other vascular malformations

Hypotrophy

Scoliosis

Digital abnormalities

Proteus syndrome

PIK3CA

AKT1

Go to next page: Management/Treatment of Capillary malformation (Vascular stain)
Management/Treatment of Capillary malformation (Vascular stain)

Capillary malformation-arteriovenous malformation (CM-AVM)
- Doppler ultrasound and/or MRI of any suspected AVM
- Consider repeated cranial and spinal MRI at puberty
- Germline genetic testing
- Response to and safety of laser not known
- Treatment of AVM (refer to AVM pathway)

Sturge-Weber Syndrome
- Ophthalmology at regular intervals
- Annual neurology and repeat cranial MRI if symptomatic
- Consider early laser
- Consider acetylsalicylic acid or prophylactic antiepileptic drugs if severe leptomeningeal involvement

Facial capillary malformation (Port-wine stain) w/o Sturge-Weber Syndrome
- Consider early laser

CLAPO syndrome
- Check for oral or neck lymphatic anomalies and facial overgrowth. Consider somatic Genetic test
- Consider early laser

Microcephaly capillary malformation syndrome (MIC-CAP)
- Refer to neurologist
Management/Treatment of Capillary malformation (Vascular stain) continued

Nevus simplex (salmon patch)
- Consider laser if persistent after 2-3 years
- Consider spinal ultrasound (<6 months) or MRI (>6 months) of lumbosacral area in case of other signs of dysraphism

Cutis marmorata telangiectatica congenita (CMTC)
- Consider late laser, if persistent. Results are variable.

Overgrowth Syndromes
- Localised or diffuse capillary malformation with overgrowth (DCMO)
- Megalencephaly-capillary malformation (MCAP)
- KTS, CLOVES or PROS not otherwise specified, Proteus syndrome

Orthopedics
- Consider somatic genetic test

LEGEND:
- Clinical evaluation
- Investigations
- Treatment
- Genes

Refer to the VASCERN Combined and Syndromic Vascular Malformation Patient Pathway
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.

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