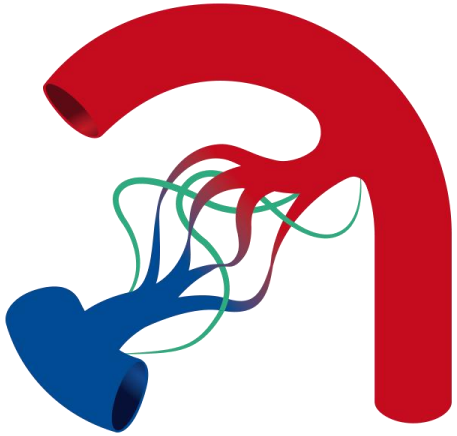
European Reference Network

for rare or low prevalence complex diseases

Network Vascular Diseases (VASCERN)



Arteriovenous malformations

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Disclaimer

- This document is an opinion statement reflecting strategies proposed 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 **February 20, 2026** and will be further validated and adjusted as needed.
- Responsibility for care of individual patients remains with the treating physician.
- Multidisciplinary team should re-evaluate treatment decisions regularly.

Arteriovenous Malformation (AVM)

It can arise in any area of the body

Symptoms differ based on the affected location.

It can be part of a syndrome with specific associated symptoms

It can be sporadic or hereditary

Work-up varies according to subtype and/or location

Based on associated clinical signs

- Hereditary Syndromic
- Sporadic Syndromic
- Sporadic non-syndromic

Based on location

- Central Nervous System
- Visceral and intracavitary
- Peripheral

Arteriovenous Malformation (AVM)

Clinical symptoms & work-up

Adult

Paediatric

General hemodynamic manifestations
CSF flow abnormalities

Neurological symptoms
Headaches
Cerebral or spinal hemorrhages

All

Dyspnea

Hemoptysis

Brain abscess or stroke

Abdominal pain

Blood in stool

Hematuria, proteinuria

Pelvic pain

All

Red warm mass or stain
Deformation or hypertrophy
Pulse, thrill, local swelling, ulceration, bleeding, functional limitation and pain

High output cardiac failure

CNS AVM

Intracranial AVM

Spinal AVM

Visceral AVM

Intracavitary AVM

Peripheral AVM

US

MRI and/or MRA and/or CTA and/or DSA

LEGEND: Clinical evaluation | Investigations

Arteriovenous Malformation (AVM): Associated symptoms

Hereditary Syndromic AVM

Curaçao criteria for HHT (multiple) visceral AVM (multiple) bAVM Spinal AVM

See corresponding pathway and HHT guidelines

- HHT-1 — *ENG*
- HHT-2 — *ACRVL1*
- Juvenile-Polyposis/HHT — *SMAD4*
- HHT-5 — *GDF2*

Peripheral AVM Macrocephaly

- Doppler ultrasound
- MRI and/or MRA and/or DSA
- Genetic counseling

PHTS — *PTEN*

→ Cancer screening

(multiple) CM with pale surrounding anemic halo (multiple) AVMs Parkes-Weber Syndrome*

- Doppler ultrasound
- Cerebral or spinal MRI and MRA and/or CTA and/or DSA
- Genetic counseling

- CM-AVM type 1 — *RASA1*
- CM-AVM type 2 — *EPHB4*

* Parkes-Weber Syndrome :
- Hereditary or sporadic
  - Hereditary caused by germline RASA1 or EPHB4 mutations
  - Somatic caused by mosaic RASA1 or KRAS mutations
- Clinical presentation :
  - Limb (lower>upper) hypertrophy
  - Diffuse erythema and warmth
  - "CM" (faint red stain (blush)) on affected limb
  - Multiple arteriolo-venular shunts

***HHT** : Hereditary Hemorrhagic Telangiectasia or Osler-Weber-Rendu syndrome*
***CM-AVM** : Capillary Malformation - Arteriovenous Malformation*
***PHTS** : PTEN Hamartoma Tumor Syndrome*

LEGEND: Clinical evaluation | Investigations | Genes

Arteriovenous Malformation (AVM): Associated symptoms

Sporadic Syndromic AVMs

Limb hypertrophy "CM", faint red stain blush Arteriolo-venular shunts No CMs outside the affected limb Spinal AVM	Facial +/- retinal +/- brain AVM (need min 2)	Back or lumbar "CM" Para-lumbar soft tissue AVM Spinal AVMs or AVFs	Lipomatous overgrowth Slow-flow VAs Epidermal Nevi Skeletal anomalies Spinal AVMs
- Doppler ultrasound - MRI and MRA (- CTA and/or DSA) - Orthopedic evaluation	- Cerebral MRI and MRA (- CTA and/or DSA) - Ophthalmic evaluation	- MRI and MRA (- CTA and/or DSA) (- US)	- MRI and MRA (- CTA and/or DSA) (- US)
Parkes-Weber Syndrome*	**Cerebro-facial Arteriovenous Metameric Syndrome (CAMS)**	**Spinal Arteriovenous Metameric Syndrome (SAMS)** *(Cobb syndrome)*	**CLOVES syndrome**
Somatic RASA1 *Somatic KRAS*	*Somatic KRAS*	*Somatic KRAS*	*PIK3CA*

***CLOVES** : Congenital Lipomatous Overgrowth, Vascular Malformations, Epidermal Nevi, and Skeletal/Scoliosis Anomalies*

LEGEND: Clinical evaluation | Investigations | Genes

Arteriovenous Malformation (AVM): Associated symptoms

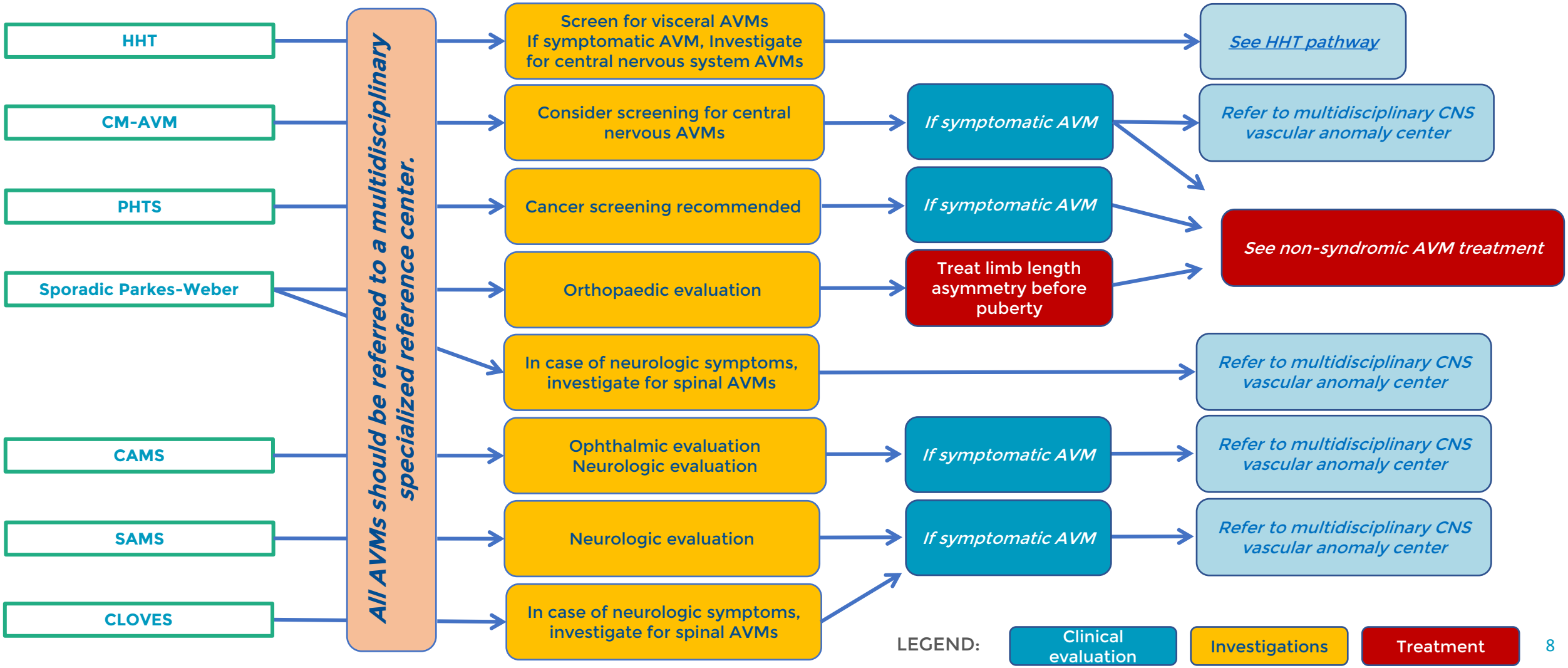
Sporadic Non-syndromic AVMs

Neurological symptoms Cerebral hemorrhage Headache, tinnitus, ...	Neurological symptoms Sphincterial dysfunctions	Dyspnoea, haemoptysis Melena, rectorrhagia Abdominal pain Cardiac failure	Abdominal pain Thoracic pain Cardiac failure	Mass, swelling, hypertrophy, warm vascular stain Warmth, pulsatility, thrill Pain, ulceration, functional impairment, Cardiac failure
- Cerebral MRI and MRA (- CTA and/or DSA)	- Spinal MRI and MRA (- CTA and/or DSA)	- US and/or CTA - MRI and MRA	- US and/or CTA - MRI and MRA	- US - MRI and MRA (- CTA and/or DSA)
Intracranial AVM *RASA1* *ENG* *EPHB4* *ACVRL1* *KRAS* *PTEN*	**Spinal AVM** *PTEN* *ENG* *RASA1* *ACVRL1* *EPHB4*	**Visceral AVM** *RASA1* *ENG* *EPHB4* *ACVRL1* *SMAD4* *GDF2*	**Intracavitary AVM**	**Peripheral AVM** *KRAS* *MAP2K1* *HRAS* *BRAF* *PTEN* *RASA1* *RIT1* *EPHB4*

LEGEND: Clinical evaluation | Investigations | Genes

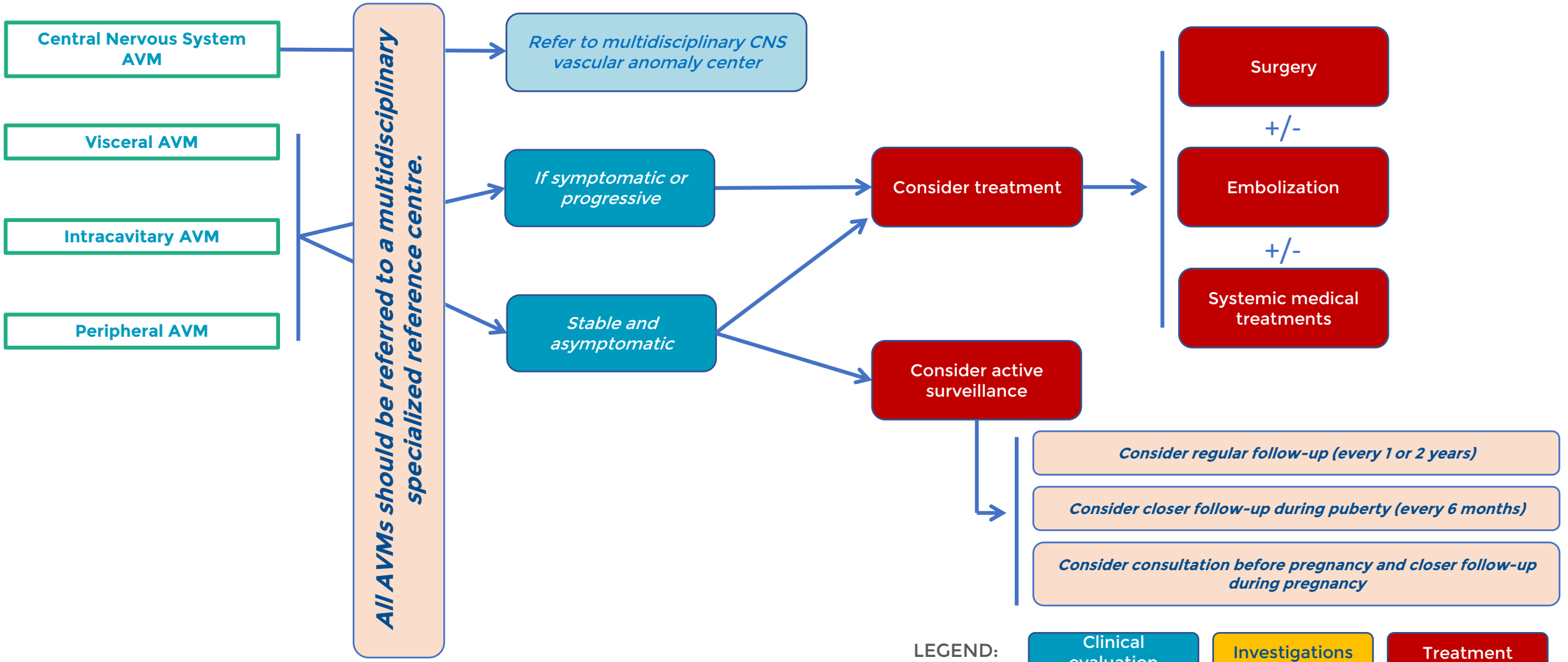
Arteriovenous Malformation (AVM): Management/Treatment

Syndromic AVMs



Arteriovenous Malformation (AVM): Management/Treatment

Non-syndromic AVMs



European Reference Network

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 gathers 48 expert teams from 39 highly specialized multidisciplinary HCPs, plus 6 additional Affiliated Partner centers, coming from 19 EU Member States, as well as various European Patient Organisations, and is coordinated in Paris, France.

Through our 6 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: www.vascern.eu

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