Case 4

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Case 4

• 36-year-old man
• lower back pain with right sciatica of one year duration.
• after 3 months, progressive onset of a mass in the right buttock.
• radiograms and CT-scan showed small calcifications in the soft tissues
• on MRI serpiginous T1 and T2 hypersignals in the subcutaneous fat and muscles.
• the gross specimen was 16 x 15 x 6 cm
• fatty appearance with muscle strands
• poorly demarked lesion with vessels and spongiform nodules, containing dark blood.
not present on the submitted specimen
Case 4
Diagnosis
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Diagnosis

Angiomatosis of soft tissues
after Rao and Weiss
Angiomatosis of Soft Tissue

An Analysis of the Histologic Features and Clinical Outcome in 51 Cases

Vasantha K. Rao, M.D., and Sharon W. Weiss, M.D.

a histologically benign vascular lesion that affects a large segment of the body in a contiguous fashion either by vertically involving multiple tissue types (e.g., subcutis, muscle, bone) or by involving similar tissue types (e.g., multiple muscles).
Young patients  77% ≤ 30 years
Female predominance  68%

N=  40
Lower limb  25 (62%)
Upper limb  7
Face and neck  6
Trunk  2

- mass, firm, no volume variation
- pain (32%) often severe
- mass or pain of sudden onset
  => biopsy (n=8) to exclude malignancy
- deep lesions, only 5 with subcutis involvement (12.5%)
  1 case with skin lesions

Rao & Weiss paper

multinodular / stellate vascular lesion with fat tissue
Veins with muscular pads and fissures (77%)
Slit-like thin-walled channels radiating from larger veins
honeycomb or pulmonary alveoli like pattern (97%)
thick-walled vessels with onion bulb pattern (80%)
tissular or intravascular lymphocytes (92%)
Podoplanin (D2-40) expression
extra-vascular smooth-muscle bundles (70%)
Phleboliths
Differential Diagnosis

• Other vascular malformations
  – mainly common venous malformations
  – +/- lymphatic malformations
Differential Diagnosis

- Other vascular malformations
  - mainly common venous malformations
  - +/- lymphatic malformations

- Intramuscular capillary hemangioma
### Provisionally unclassified vascular anomalies

<table>
<thead>
<tr>
<th>Condition</th>
<th>Causal genes</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intramuscular hemangioma *</td>
<td></td>
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<tr>
<td>Angiokeratoma</td>
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<tr>
<td>Sinusoidal hemangioma</td>
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<tr>
<td>Acral arteriovenous &quot;tumour&quot;</td>
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<tr>
<td>Multifocal lymphangioendotheliomatosis with thrombocytopenia / cutaneovisceral angiomatosis with thrombocytopenia (MLT/CAT)</td>
<td>PTEN</td>
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<tr>
<td>PTEN (type) hamartoma of soft tissue / &quot;angiomatosis&quot; of soft tissue (PHOST)</td>
<td>PTEN</td>
</tr>
<tr>
<td>Fibro adipose vascular anomaly (FAVA)</td>
<td>PIK3CA</td>
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</tbody>
</table>

* Distinct from infantile hemangioma, from intramuscular common VM, PHOST/AST, FAVA and AVM some lesions may be associated with thrombocytopenia and/or consumptive coagulopathy [see details](#).
PTEN Hamartoma of Soft Tissue: A Distinctive Lesion in PTEN Syndromes

Kyle C. Kurek, MD,* † Emily Howard, MD,* L.B. Tennant, MD,* Joseph Upton, MD,†
Ahmad I. Alomari, MD,† † Patricia E. Burrows, MD,† † Kim Chalache, BA,†
David J. Harris, MD,† † Cameron C. Trenor III, MD,† † Charis Eng, MD,† †
Steven J. Fishman, MD,† † John B. Mulliken, MD,† †
Antonio R. Perez-Atayde, MD,* and Harry P. W. Kozakewich, MD*†

Am J Surg Pathol 2012
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Am J Surg Pathol 2012
Luks VL, et al.
Lymphatic and other vascular malformative/overgrowth disorders are caused by somatic mutations in PIK3CA.
J Pediatr 2015
A series of vascular lesions with features of AST
- all with anomalies of PIK3CA or PTEN
- a majority of PIK3CA anomaly
- a minority of patients with PTEN germline mutation
The histological diagnosis of angiomatosis of soft tissue (AST) seems to correspond to either

PTEN hamartoma of soft tissue (PHOST)
or
fibro adipose vascular anomalie (FAVA)

A diagnosis of AST should prompt testing for PTEN germline mutation or PIK3CA mosaicism

- PTEN patients are at risk for cancer
- Targeted therapies for PIK3CA lesions exist
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