Fibroma-like PEComa
A Tuberous Sclerosis Complex-related Lesion

Ana B. Larque, PhD,* Richard L. Kradin, MD,* Ivan Chebib, MD,* G. Petur Nielsen, MD,*
Martin K. Selig, BA,* Elizabeth A. Thiele, PhD,† Anat Stemmer-Rachamimov, MD,*
Miriam A. Bredella, MD,‡ Pawel Kurzawa, MD, § and Vikram Deshpande, MD*

汇报人：黎郁平

指导老师：李侠

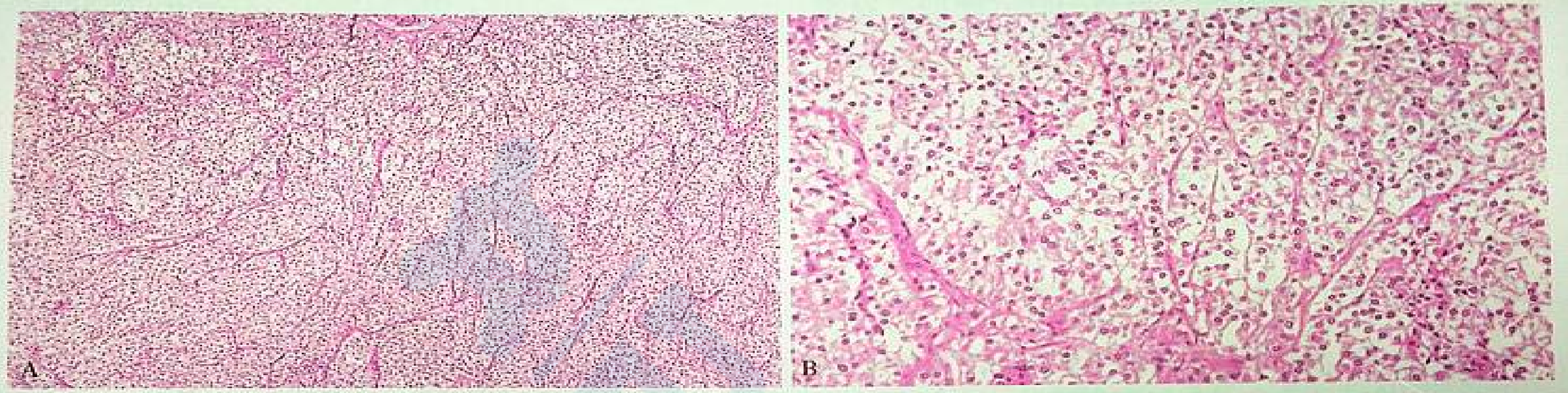
关键词:

- PEComa
- perivascular epithelioid cell
- fibroma
- tuberous sclerosis
- HMB-45

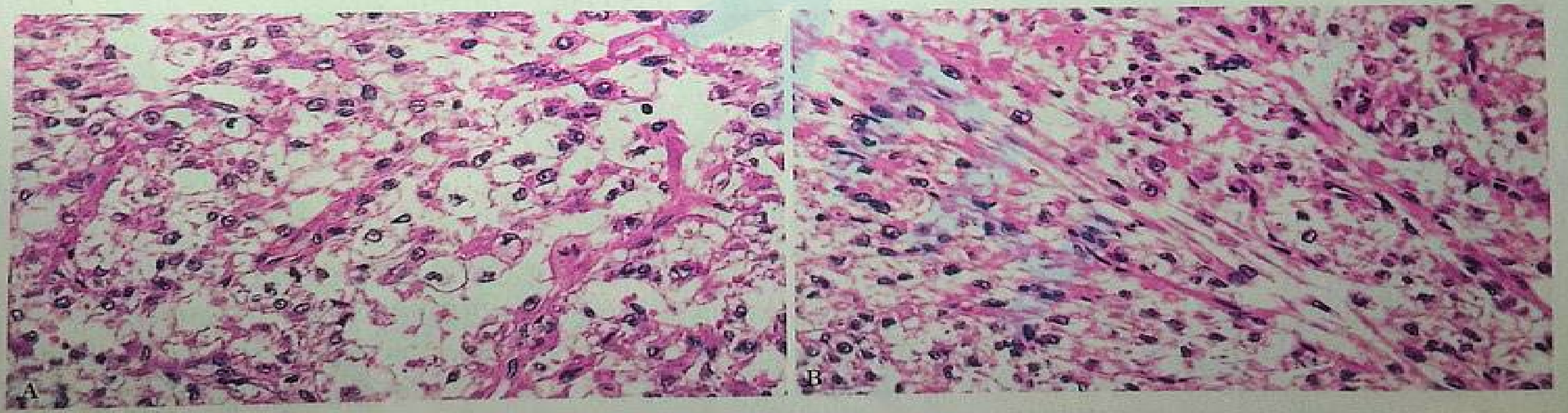
2002年WHO将PEComa定义为：

具有组织学和免疫表型特征的血管周上皮样细胞构成的间叶性肿瘤。

PEComas家族肿瘤包括：血管平滑肌脂肪瘤(AML)，肺透明细胞“糖”瘤（CCST），淋巴管平滑肌瘤病(LAM)，镰状韧带透明细胞肌黑色素细胞瘤（CCMMT）和少见的发生于胰腺、直肠、腹膜、子宫、阴道、大腿和心脏的透明细胞肿瘤。



PEComa。**A.** 典型的巢状结构。**B.** 多数病例肿瘤细胞胞浆透明，类似透明细胞癌。



A. PEComa 中的上皮样细胞。**B.** 由胞浆透明或微嗜酸的梭形细胞构成的 PEComas。

结节性硬化症(TSC):

是一种常染色体显性遗传的神经皮肤综合征，与TSC1和TSC2基因突变有关，也有散发病例。多由外胚叶组织的器官发育异常，可出现脑、皮肤、周围神经、肾等多器官受累，临床特征是面部皮脂腺瘤、癫痫发作和智能减退等。

TABLE 1. Panel of Antibodies Used in This Study

Antigen	Clone	Dilution	Source
SMA	Alpha sm-1	RTU	Leica PA0943
Desmin	DE-R-11	RTU	Leica PA0032
HMB-45	HMB-45	RTU	Leica PA0027
S100	Polyclonal	RTU	Leica PA0900
MART-1	M2-7C10	1:200	Cell Marque 281M-95
MiTF	D5	1:200	Dako M3621
TFE3	Polyclonal	1:50	Cell Marque 354R-16

TABLE 2. Clinical Features of Fibroma-like PEComas

Case	Age (y)/ Sex	Site	Size (cm)	Duration Before Diagnosis	Presenting Features	Recurrence	Metastasis	Follow-up (mo)	Original Diagnosis
1	4/F	Wrist	5.9	24 mo	Painless bump, getting progressively larger	None	None	ANED, 6 mo	Fibroma, tuberous sclerosis-related fibroma
2	25/F	Chest wall	2.8	None	Incidental	None	None	ANED, 29 mo	Desmoplastic fibroblastoma
3	51/F	Foot	5	<12 mo	Painless bump getting progressively larger	None	None	ANED, 131 mo	Fibroma of tendon sheath

ANED indicates alive with no evidence of disease; F, female.

TABLE 3. Clinical Manifestation of 3 TSC-related Fibromas

Case 1	Case 2	Case 3
Developmental delay, mild	Subependymoma	Carotid sinus aneurysm
Seizure disorder	Micronodular pneumocyte hyperplasia	Cerebral vascular malformation
Cardiac rhabdomyoma		Renal cell carcinoma
Simple renal cyst		Bilateral renal AMLs
		Lung LAM
		Angiofibroma (paranasal skin)
		Subungual fibromas

AML indicates angiomyolipoma; LAM, lymphangioleiomyomatosis.

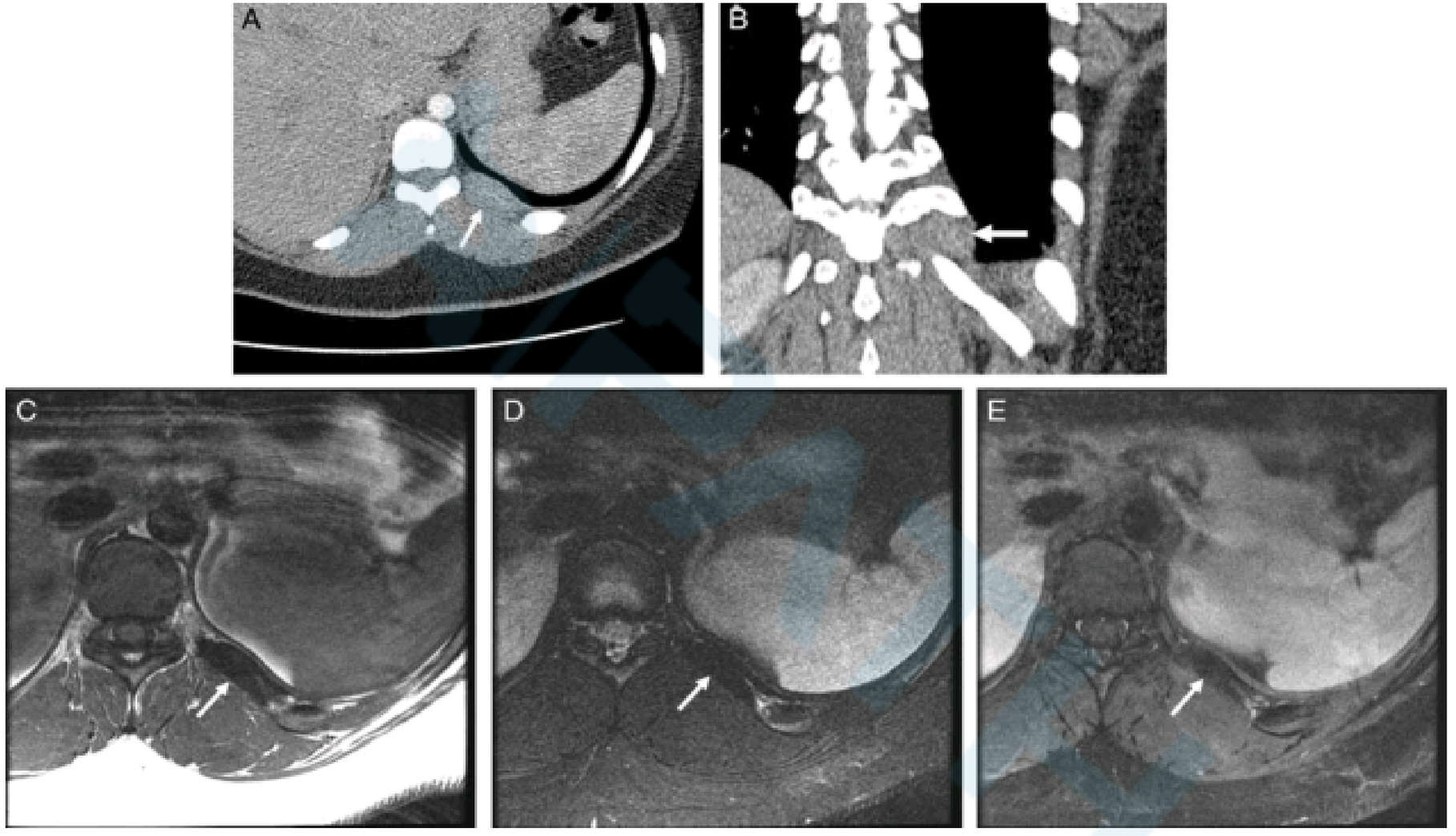


FIGURE 1. Case 2. Axial (A) and coronal reformatted (B) computed tomography images of the chest wall demonstrate a mildly hyperdense soft tissue mass, measuring 1.1 cm anteroposterior×3.2 cm transverse×2.0 cm SI, located between the left 10th and 11th ribs along the posterior-medial aspect of the chest wall, abutting the pleura (arrows). On magnetic resonance imaging, the mass is hypointense on axial T1 (C) and fat-suppressed T2 (D) weighted images and demonstrates no enhancement (E) (arrows). There is no evidence of invasion into the adjacent osseous or soft tissue structures.

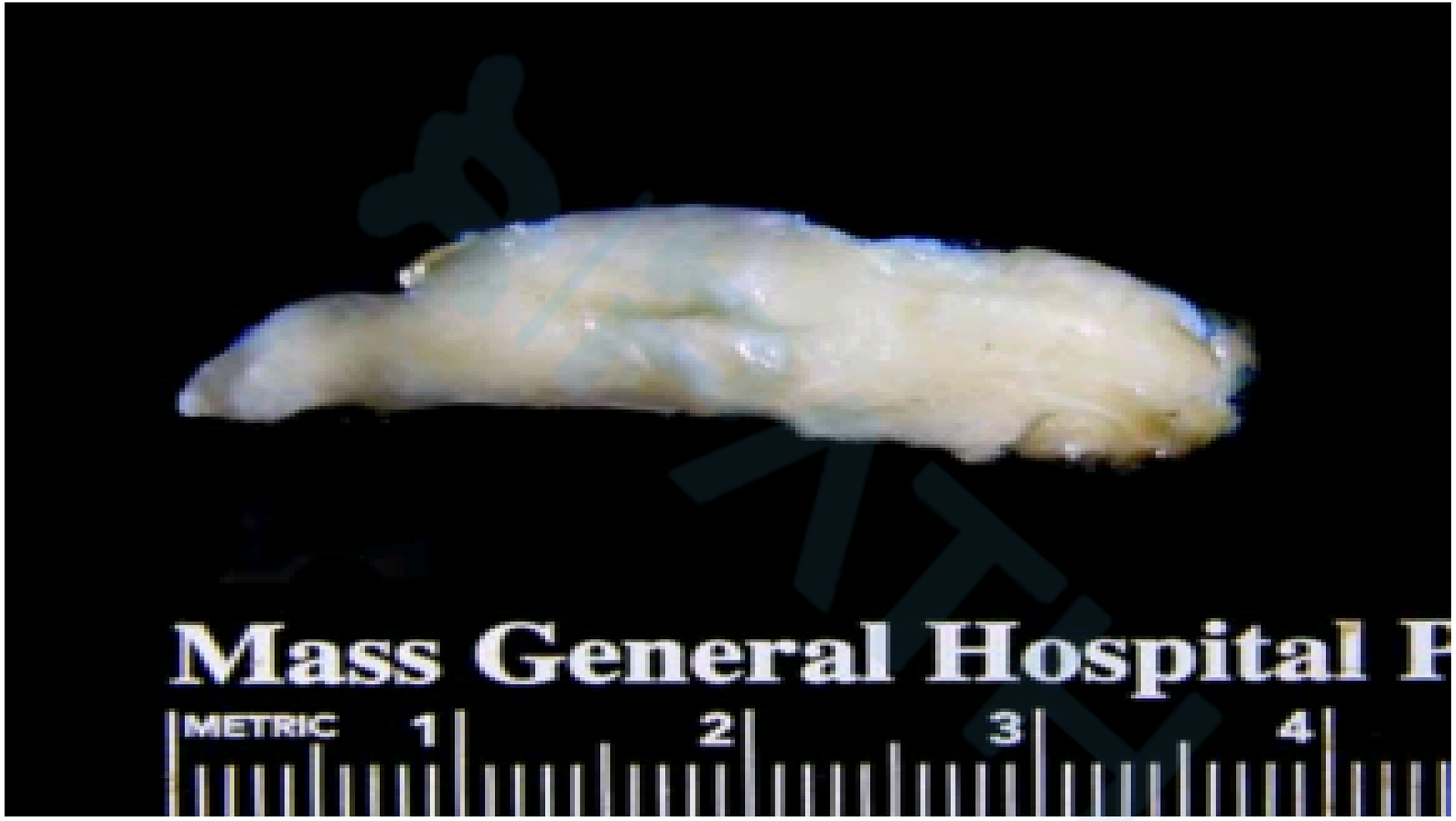


FIGURE 2. Gross image of tumor (case 1) showing homogenous yellow-gray glistening surfaces.

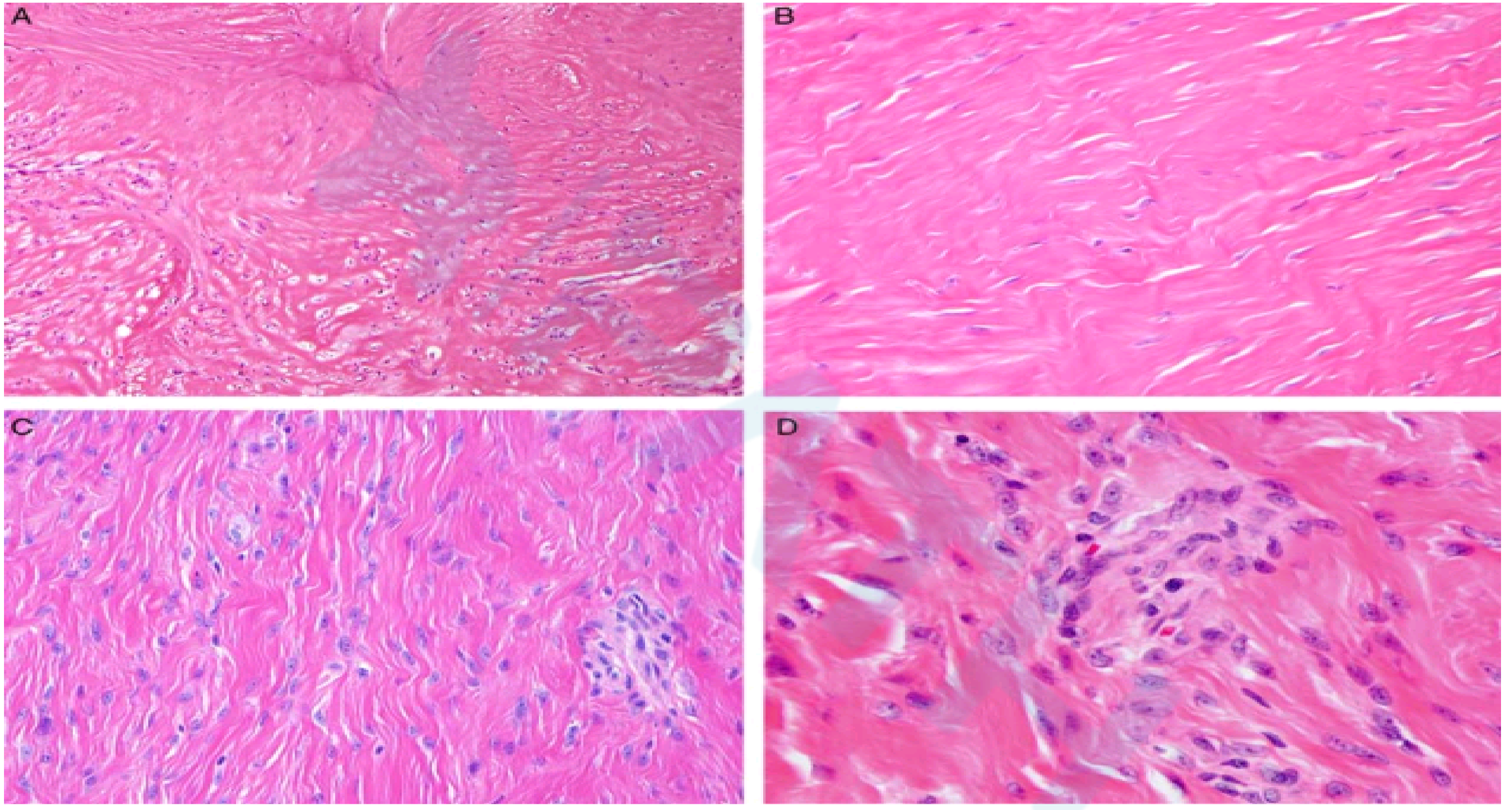


FIGURE 3. Fibroma-like PEComas composed of (A, B) bland stellate-shape or spindle-shape cells embedded in a hyalinized stroma (A, case 3; B, case 2). A moderately cellular tumor (C, D) with perivascular accentuation (case 3).

TABLE 4. Immunohistochemistry Results

Case	HMB-45	MART-1	MiTF	S100	Desmin	SMA	TFE3
1	4+	4+	0	0	4+	2+	0
2	4+	0	1+	0	0	1+	0
3	2+ wk	NA	NA	NA	NA	NA	NA

0 indicates no staining; 1+, <5% tumor cells reactive; 2+, 5% to 25%; 3+, 26% to 50%; 4+, > 50% tumor cells reactive; NA, not available; wk, weak.

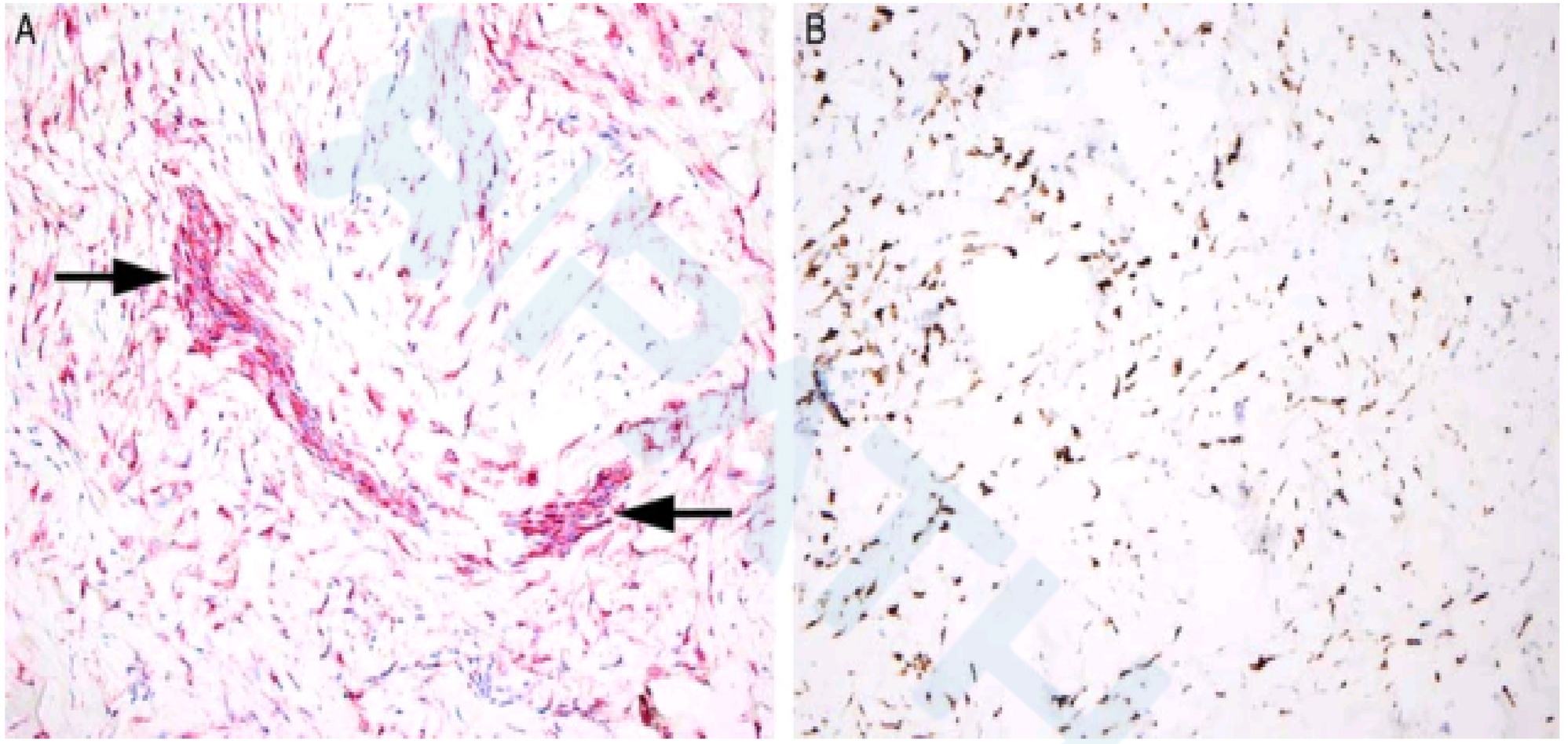


FIGURE 4. Immunohistochemical stain shows diffuse expression of HMB-45 (A) and desmin (B) by tumor cells (case 3). Note the perivascular accentuation of the tumor cells in (A) (arrows).

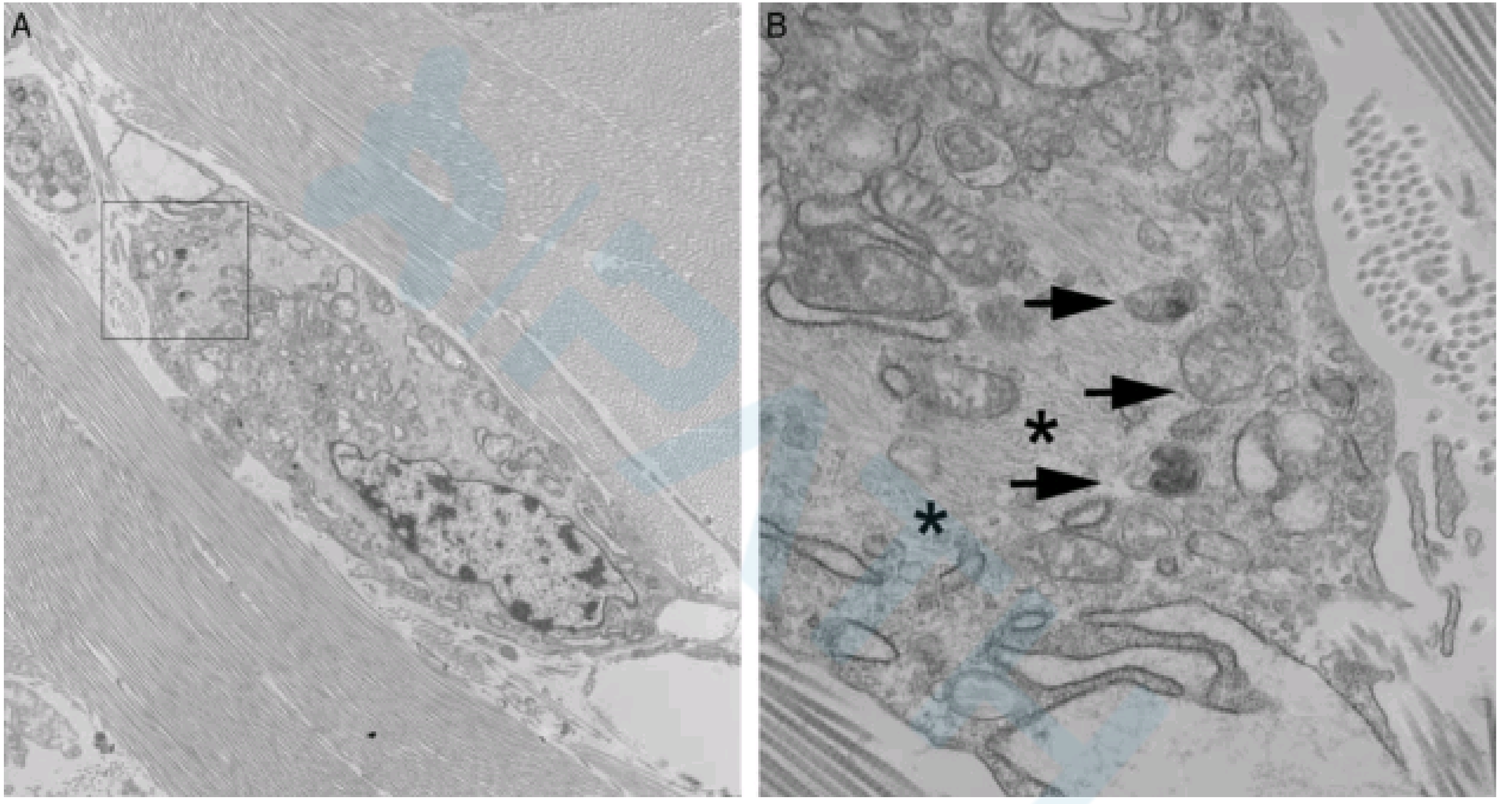


FIGURE 5. A and B, Ultrastructural findings. Cytoplasm of a tumor cell containing aggregates of intermediate filaments (*) and numerous membrane-bound granules (arrow). The portion of the cell within the box (A) is magnified in image B.

讨论

- 患者在TSC基因中显示了胚系突变。
- 表现出了血管周围聚集的上皮样细胞特点。
- 表达了黑素细胞标记HMB-45。
- 电镜下能看到膜结合颗粒。

Thank you !

謝謝！