



# SESSIÓ DE NOUS PATÒLEGS

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- HTA.
- DLP.
- SAOS amb CPAP.
- Hiperuricemia.

09/21

INGRÉS A  
NEUROLOGIA

•  
**AIT  
VERTEBRO  
-BASILAR**

•  
TROMBOLISI

•  
ALTA



5-6 días  
prèvis

FEBRICULA  
+  
ASTENIA  
+  
HIPORÈXIA  
+  
NAUSEAS  
SENSE  
VÒMITS



**NEGA:**

Simptomes  
respiratoris,  
urinaris,  
abdominals,  
neurològics,  
ginecològics i  
exterioritza-  
cions.

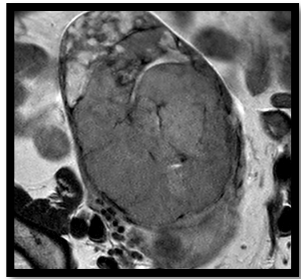
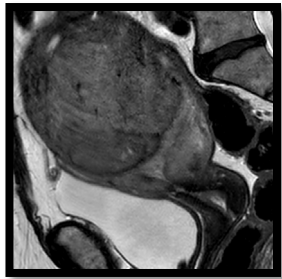


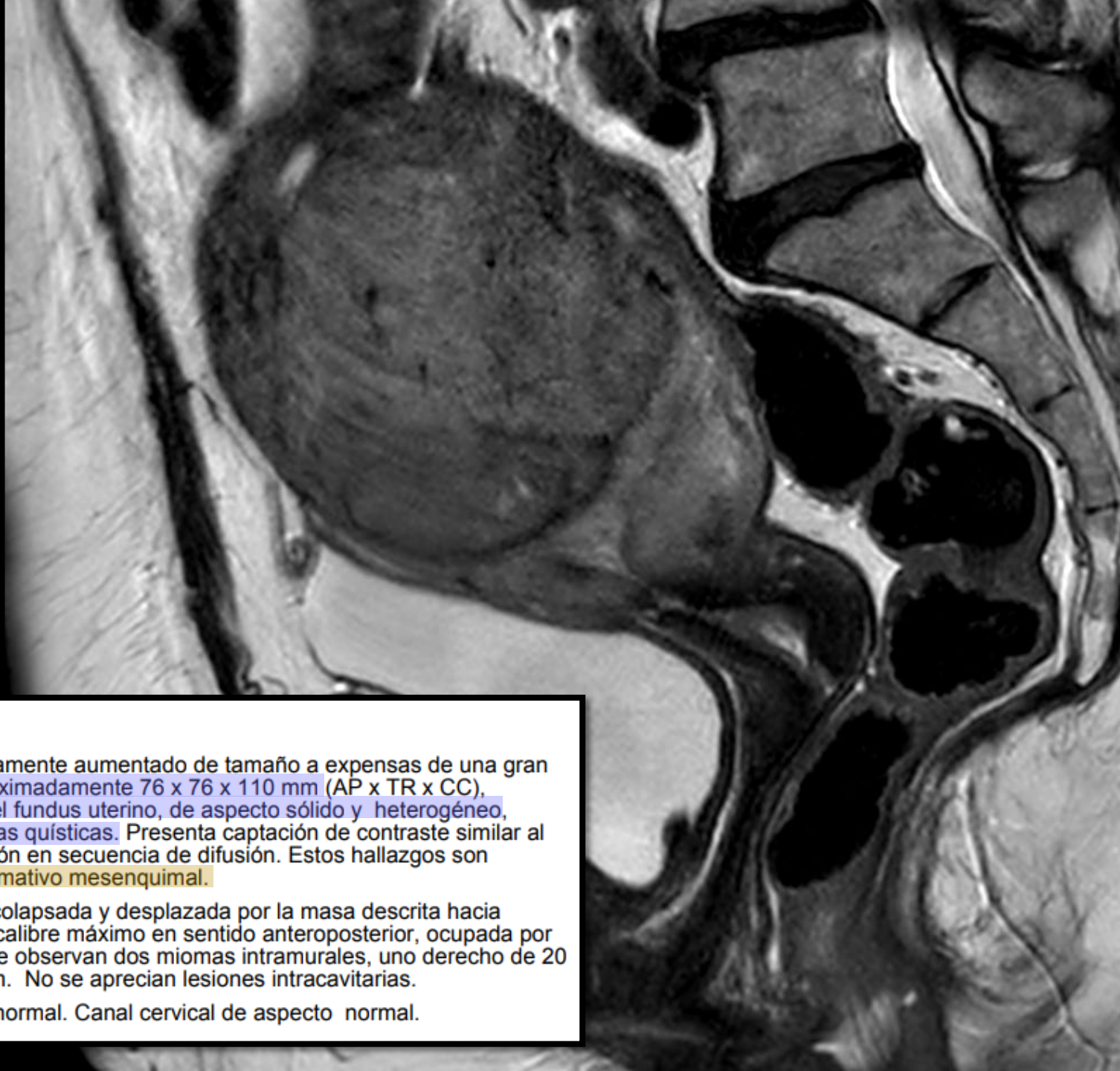
**ANALÍTICA**

LEU 15000  
NEU 14000  
PCR 120  
VSG 100

**ATB**





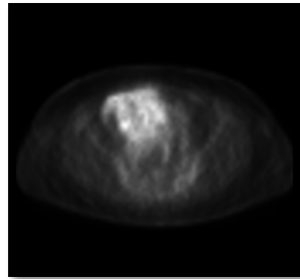
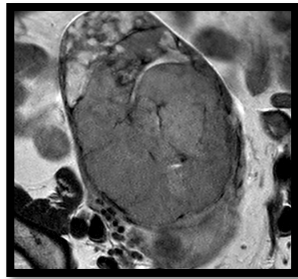
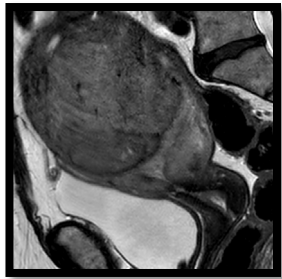


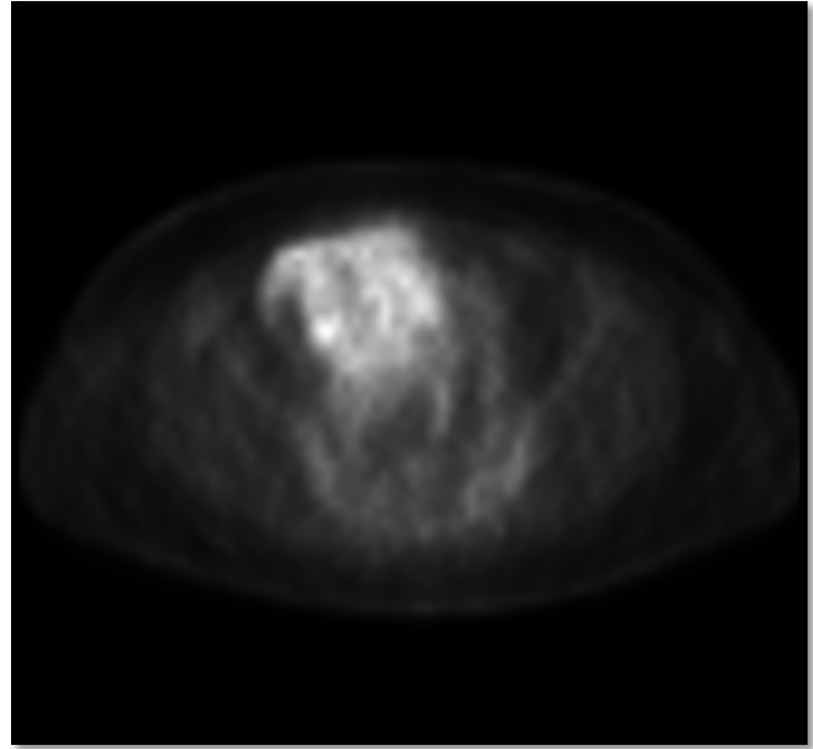
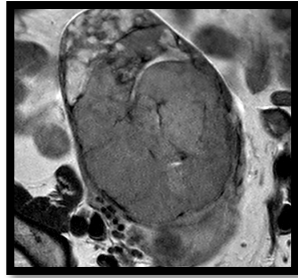
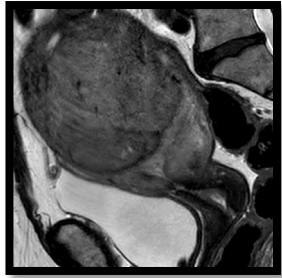
#### HALLAZGOS:

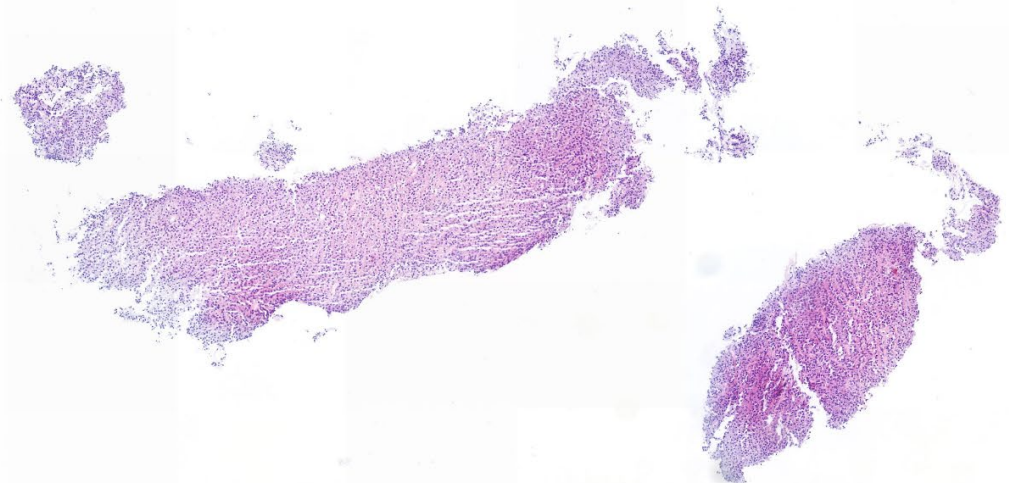
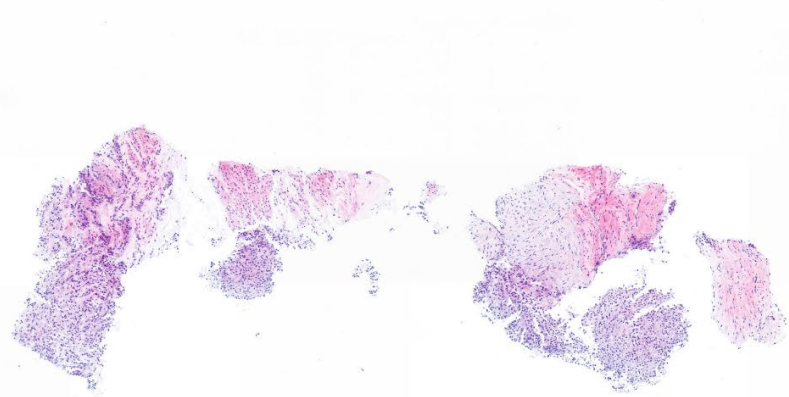
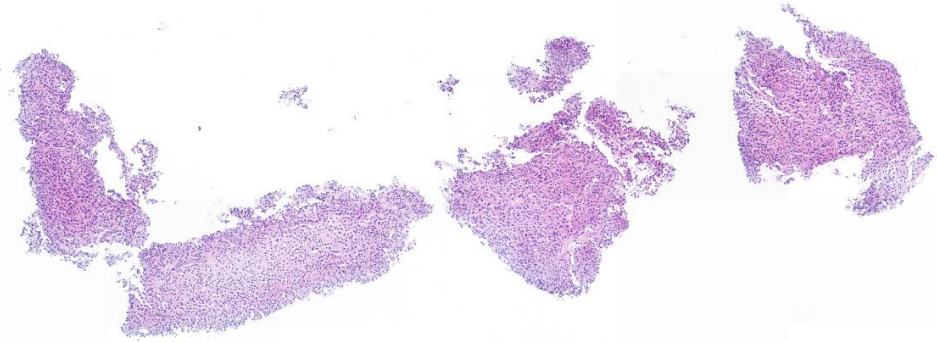
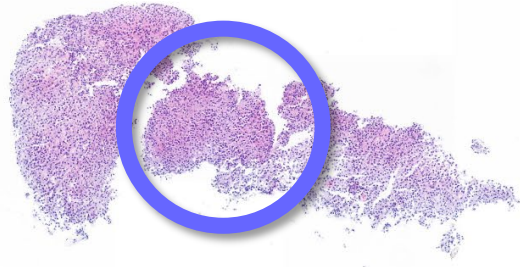
Útero en anteversión marcadamente aumentado de tamaño a expensas de una gran masa bien delimitada de aproximadamente 76 x 76 x 110 mm (AP x TR x CC), centrada a nivel miometrial del fundus uterino, de aspecto sólido y heterogéneo, polilobulada, con algunas áreas quísticas. Presenta captación de contraste similar al miometrio y marcada restricción en secuencia de difusión. Estos hallazgos son sugestivos de **proceso neofornativo mesenquimal**.

La cavidad endometrial está colapsada y desplazada por la masa descrita hacia posterior, con unos 4 mm de calibre máximo en sentido anteroposterior, ocupada por escasa cantidad de líquido. Se observan dos miomas intramurales, uno derecho de 20 mm y otro izquierdo de 18 mm. No se aprecian lesiones intracavitarias.

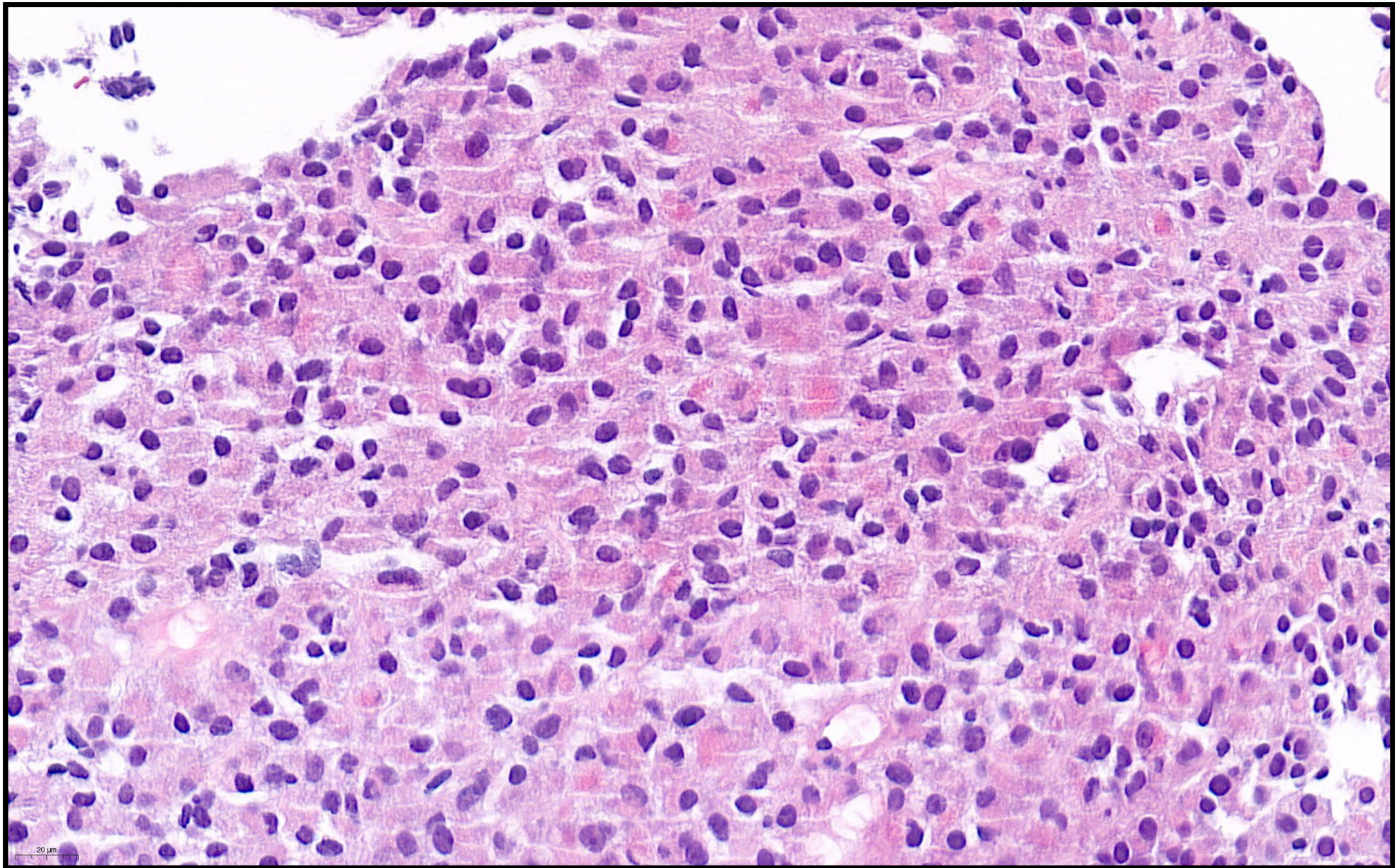
Cérvix de morfología y señal normal. Canal cervical de aspecto normal.

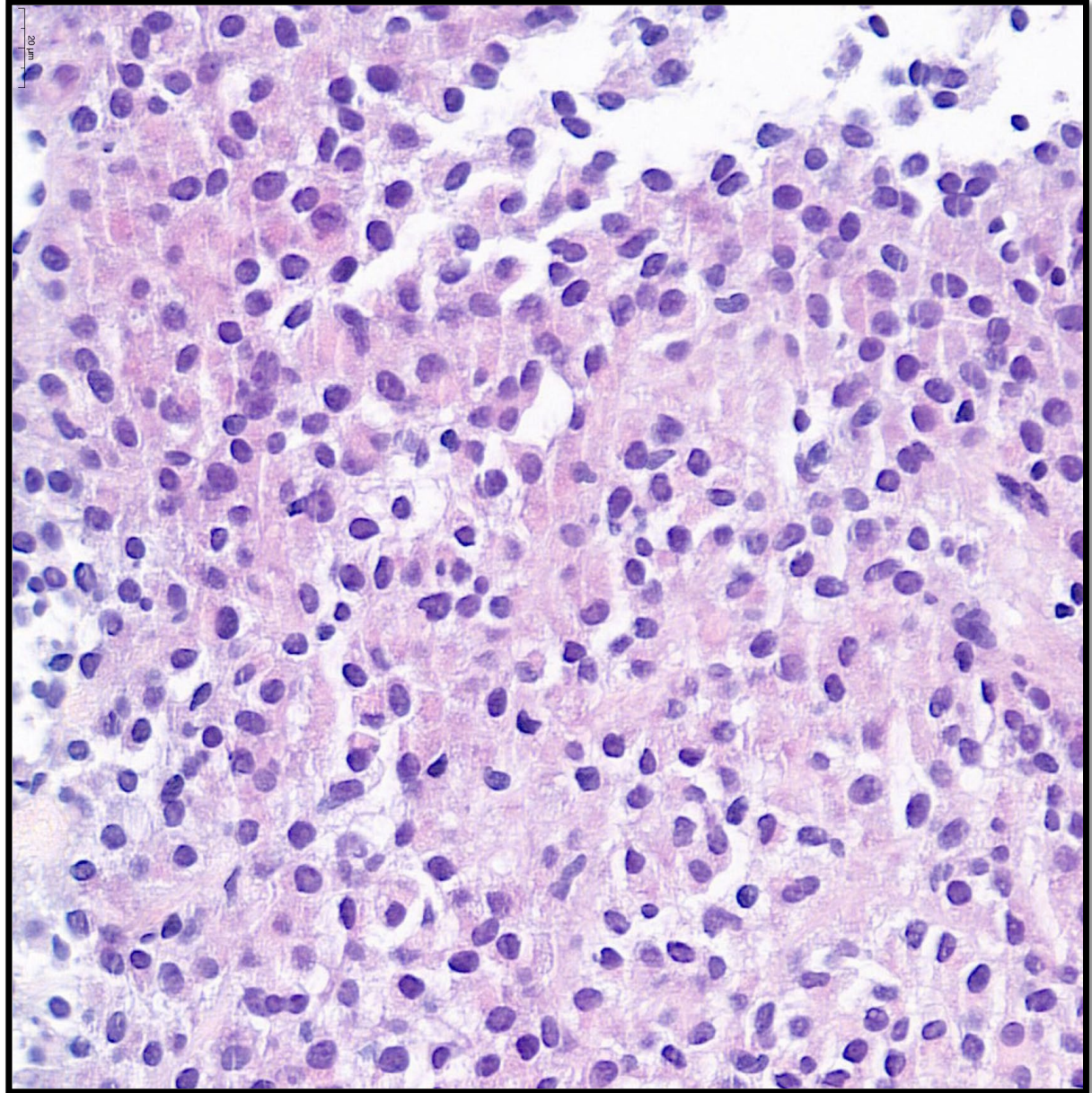
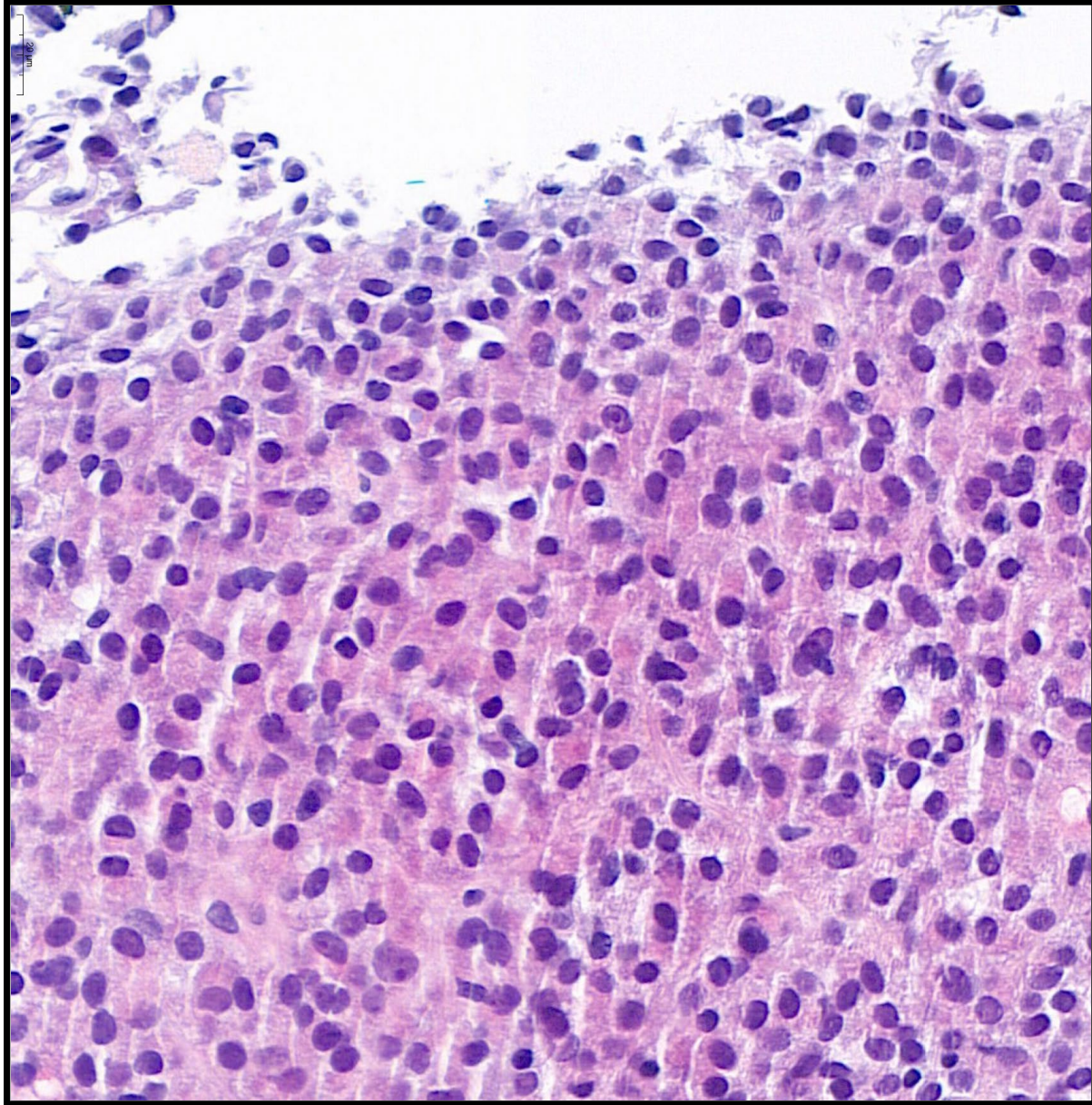


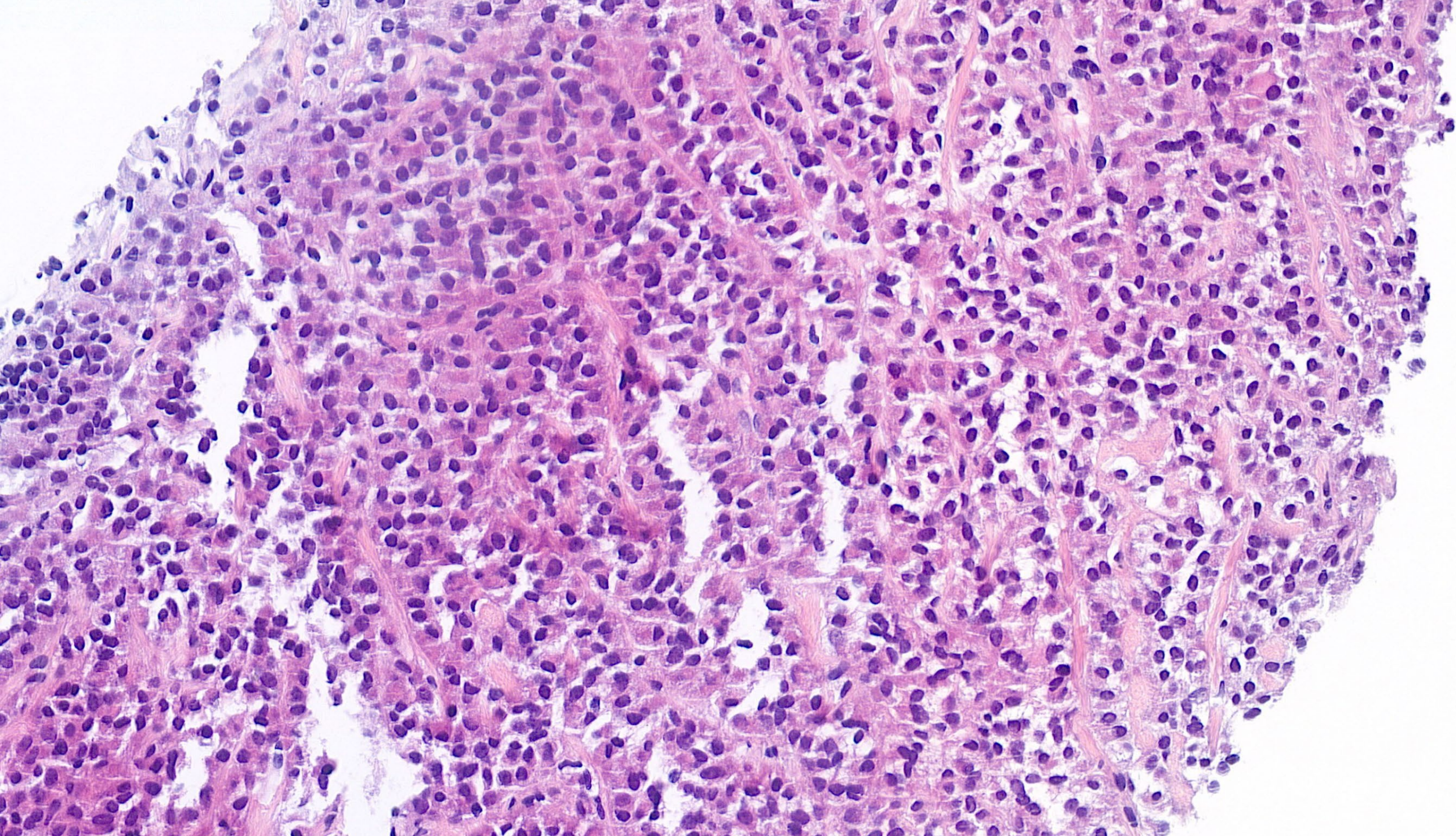


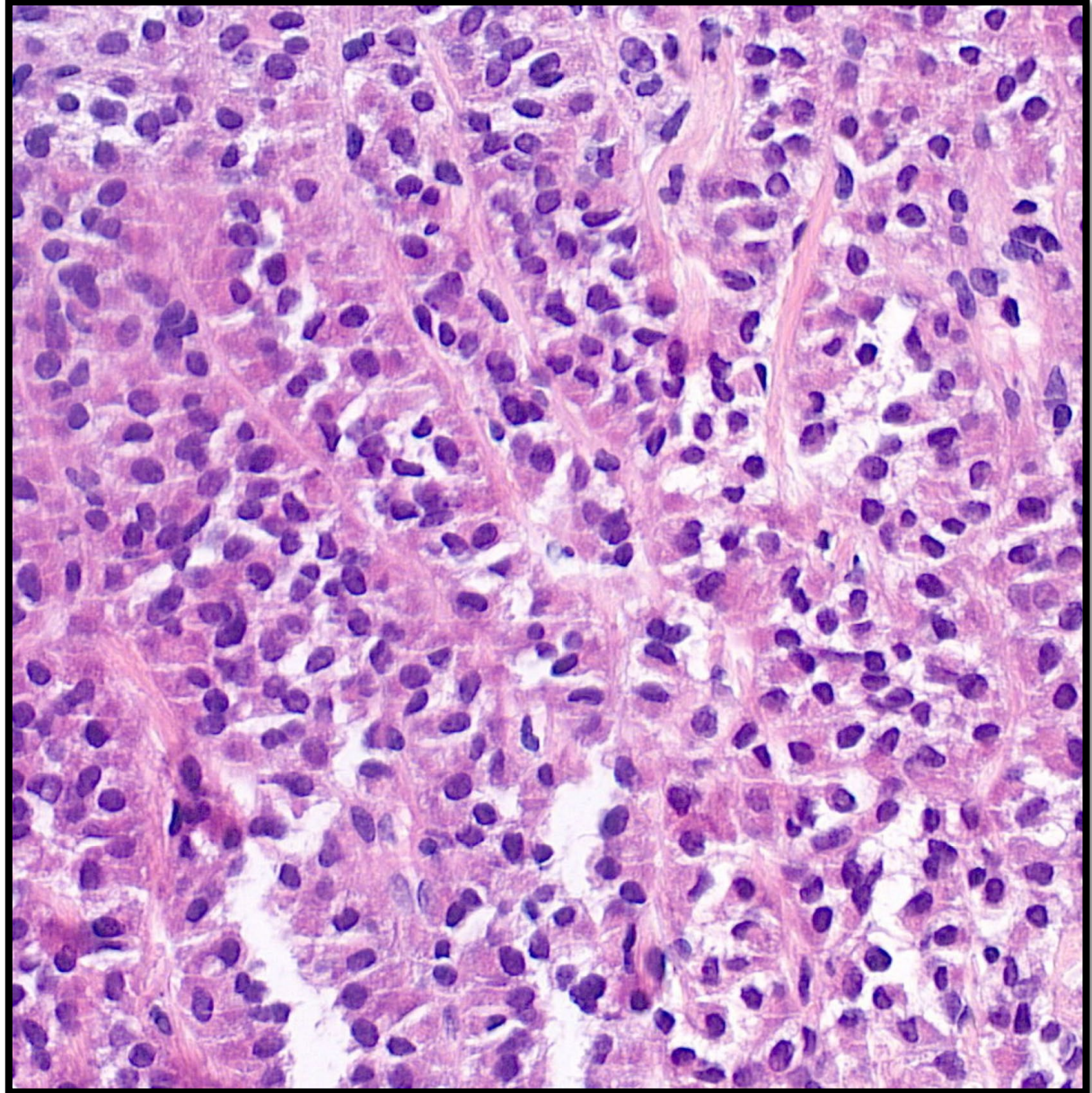
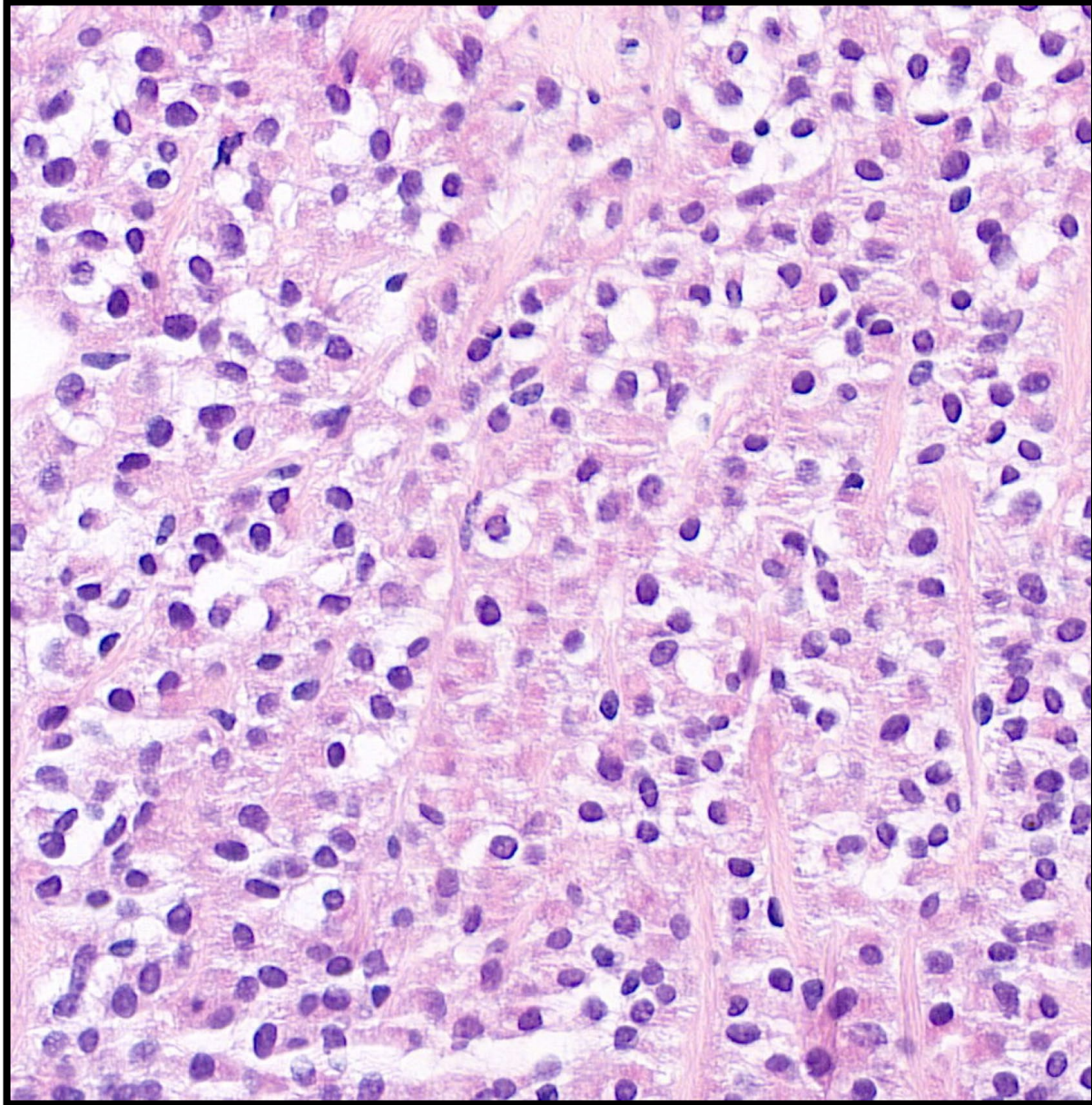












**NEOPLÀSIA DE MÚSCUL LLIS**

**NEOPLÀSIA DE CÈL·LULES PLASMÀTIQUES**

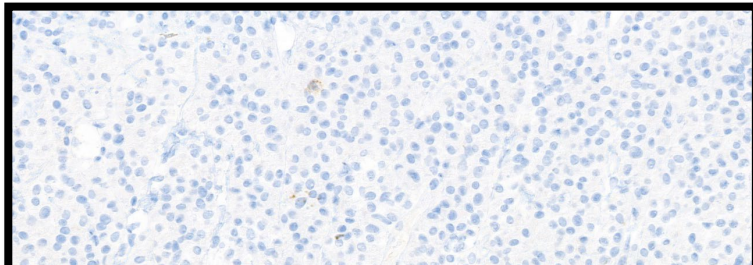
**TUMORS DE L'ESTROMA ENDOMETRIAL**

**TUMOR UTERÍ SEMBLANT A TUMORS DE CORDONS SEXUALS OVÀRIC  
(*UTROSC*)**

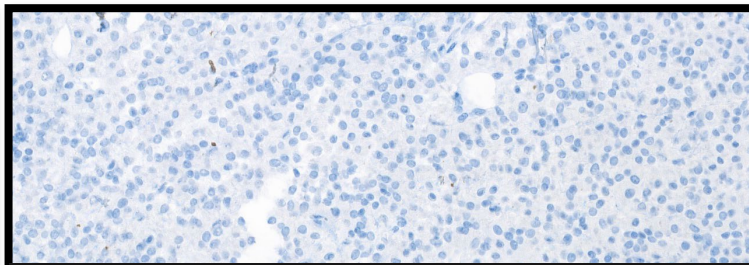
**PEComa**

**MELANOMA**

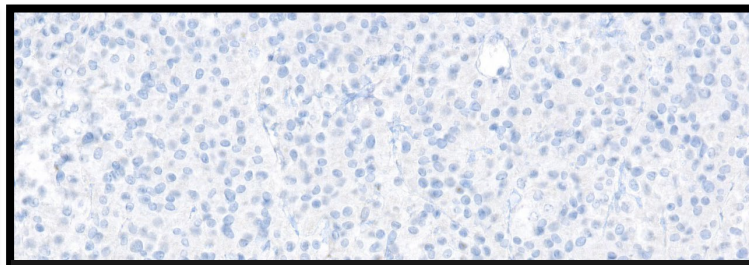
**CARCINOMA**



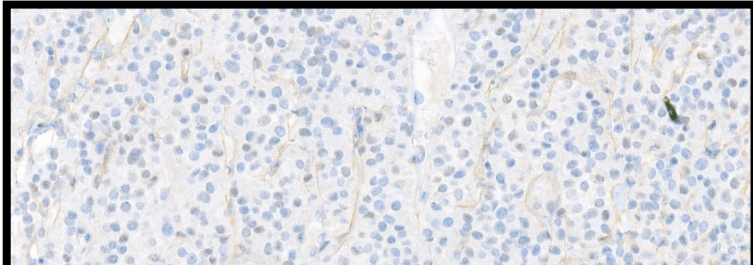
CK AE1/AE3



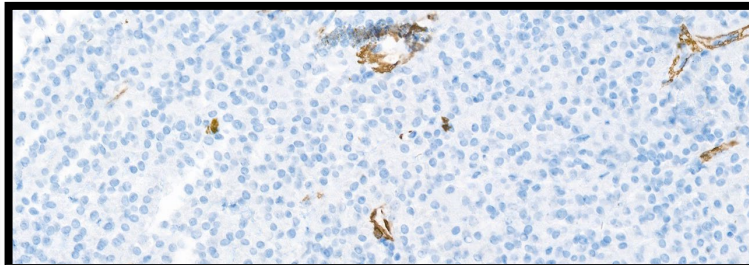
DESMINA



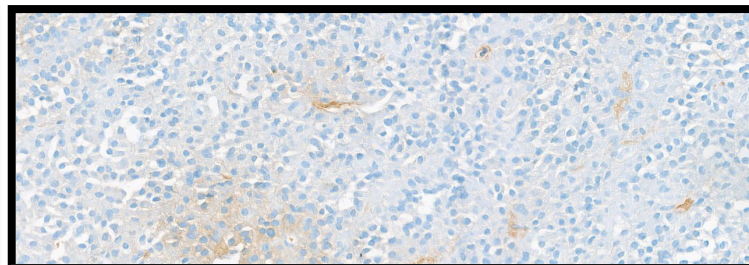
CD 138



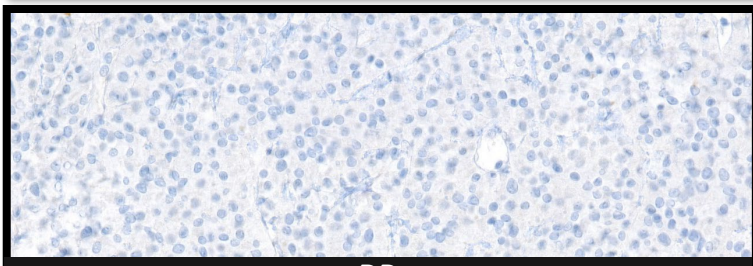
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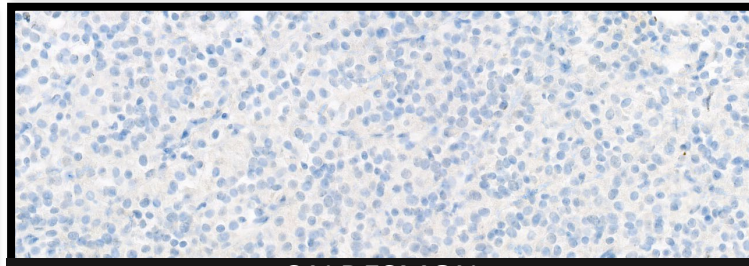
ACTINA



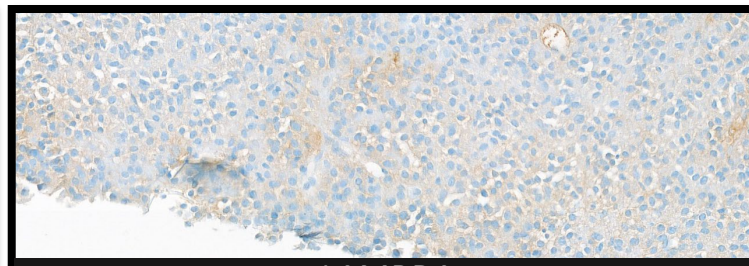
KAPPA



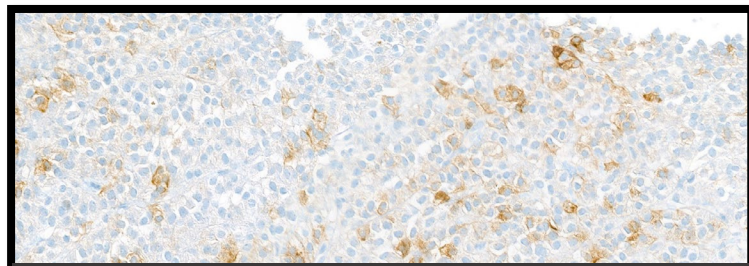
RP



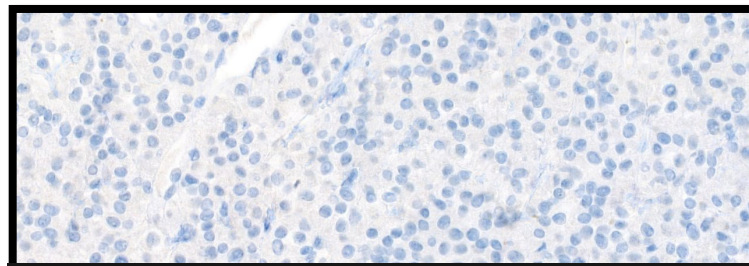
CALDESMON



LAMBDA



CD10



INHIBINA

NEOPLÀSIA DE MÚSCUL LLIS

NEOPLÀSIA DE CÈL·LULES PLASMÀTIQUES

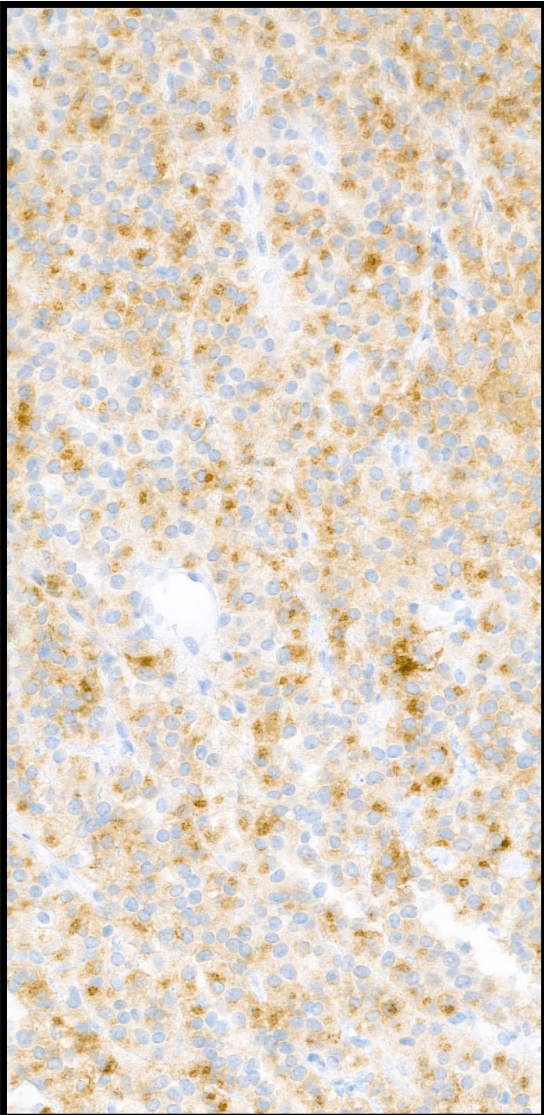
TUMORS DE L'ESTROMA ENDOMETRIAL

TUMOR UTERÍ SEMBLANT A TUMORS DE CORDONS SEXUALS OVÀRIC (UTROSC)

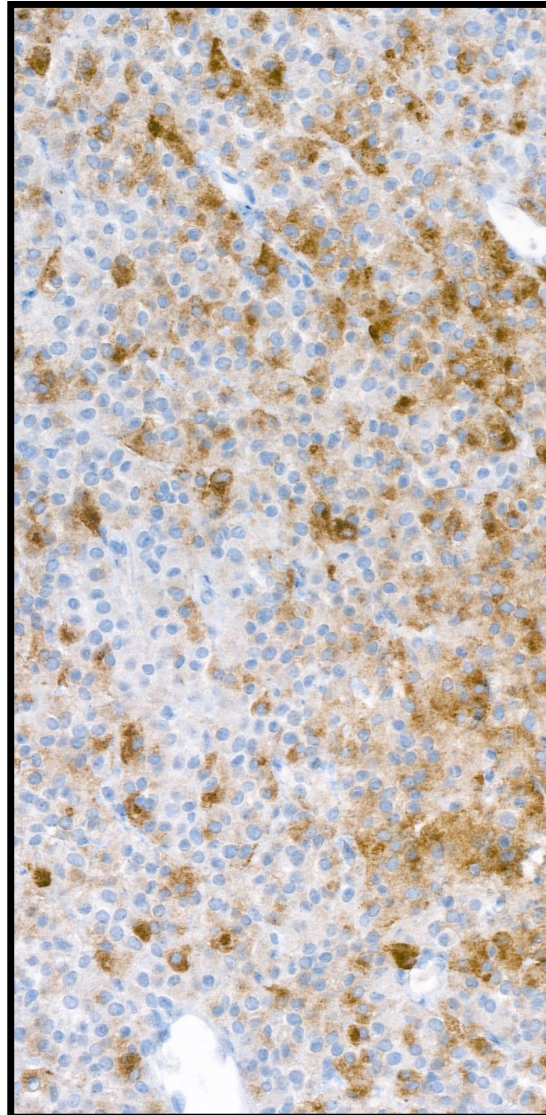
PEComa

MELANOMA

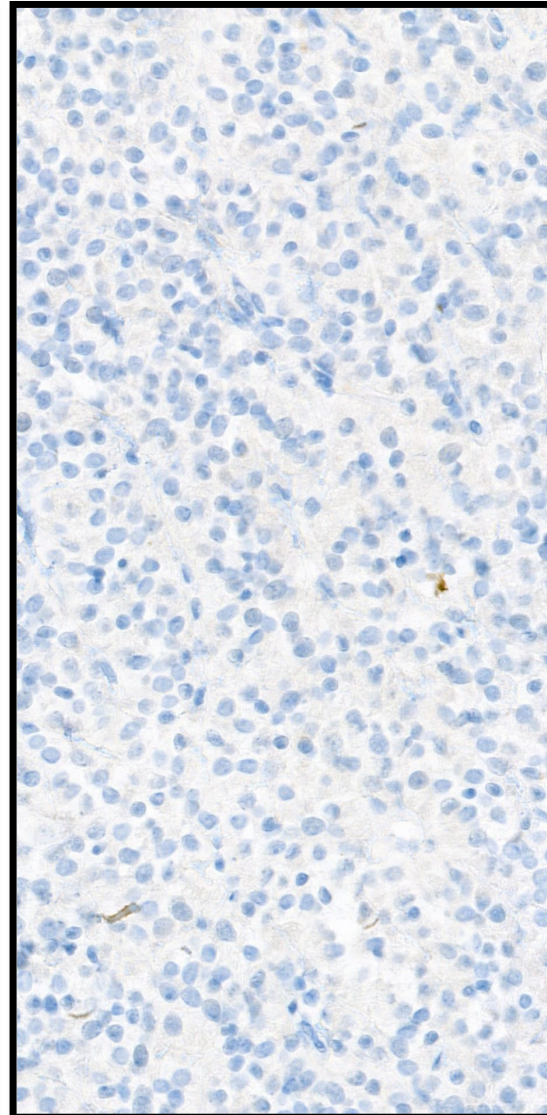
CARCINOMA



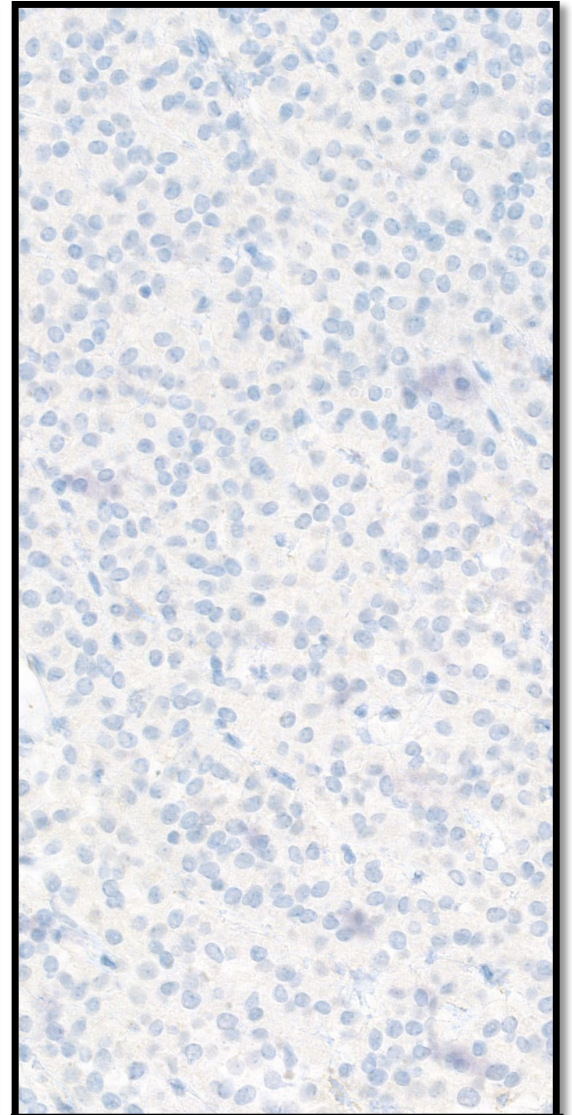
MELAN A



HMB 45

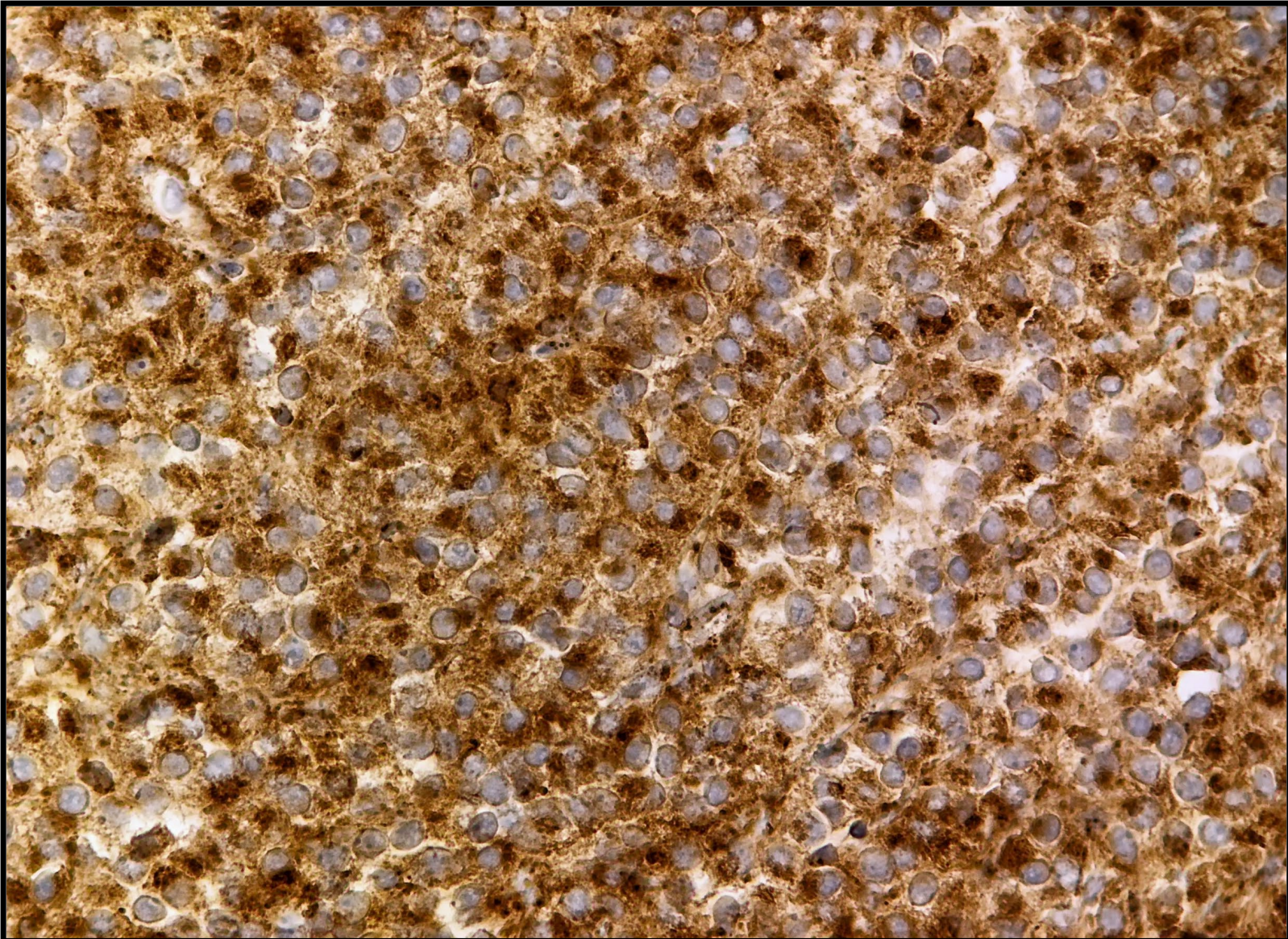


S100



SOX10

PEComa  
MELANOMA



CATEPSINA K



## DIAGNÒSTIC

Úter, biòpsia:

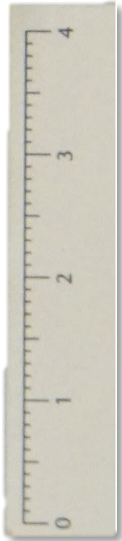
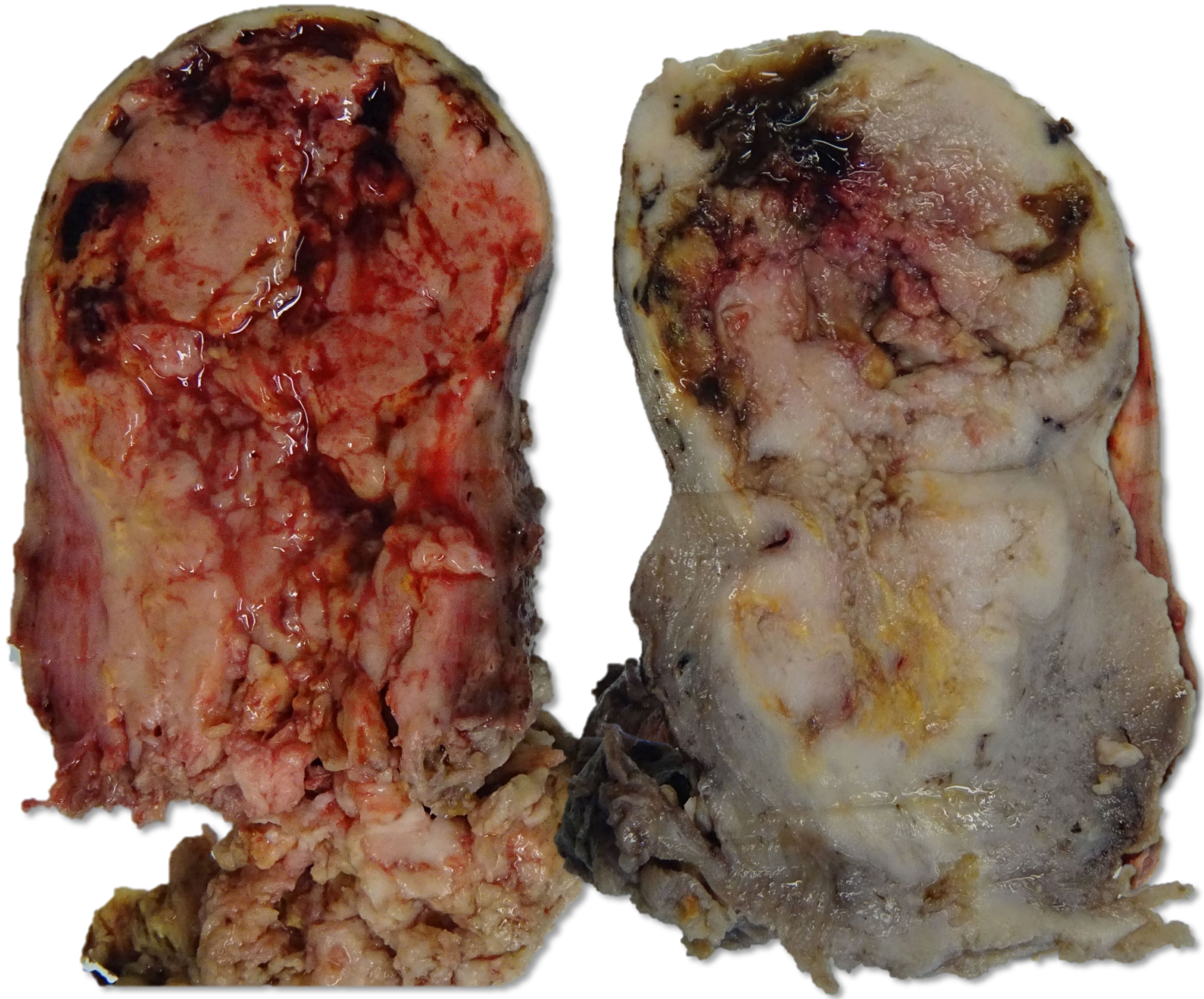
- TUMOR MESENQUIMAL DE MORFOLOGIA EPITELIODE ( VEURE COMENTARI)

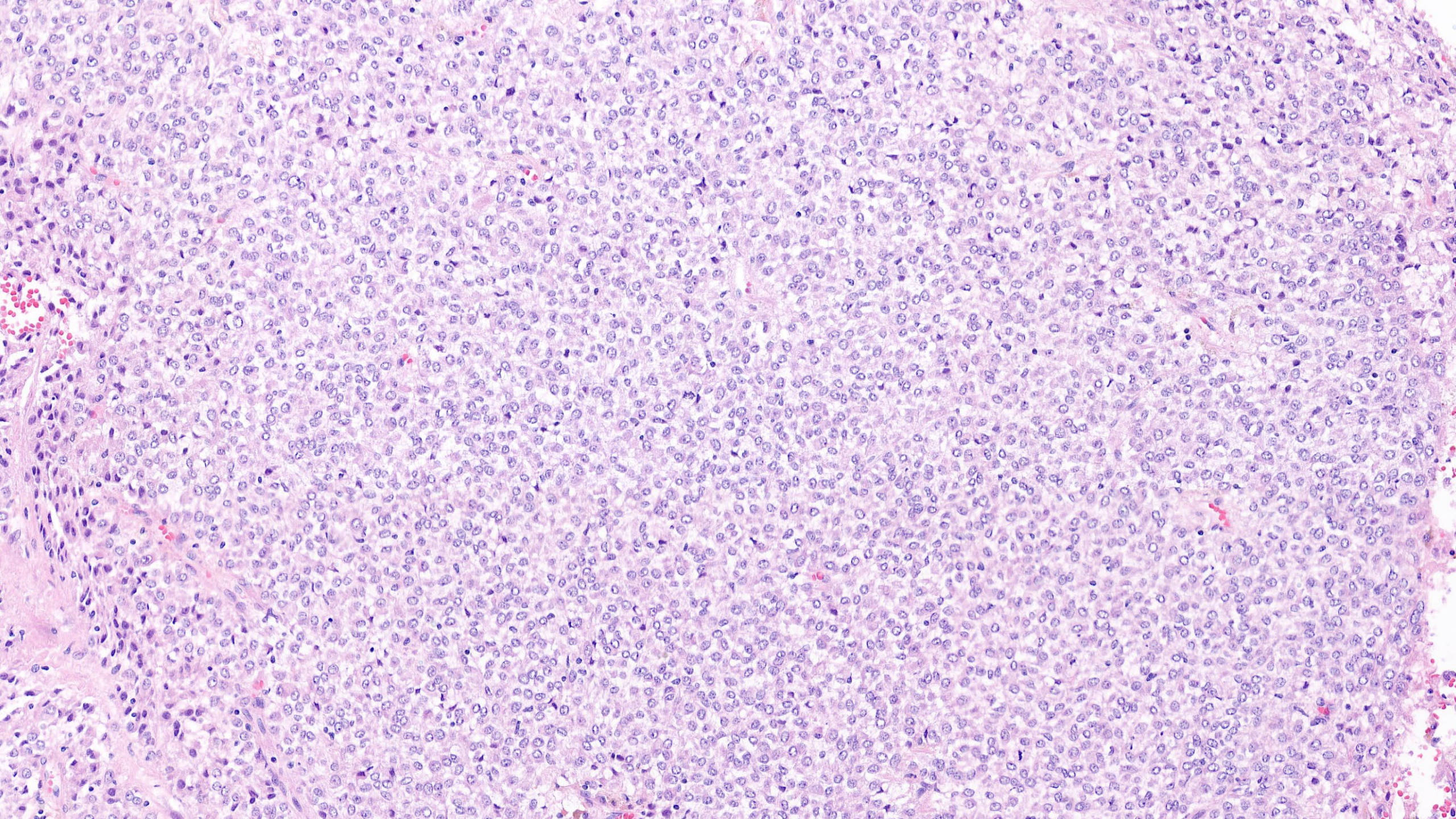
### ESTUDI IMMUNOHISTOQUÍMIC:

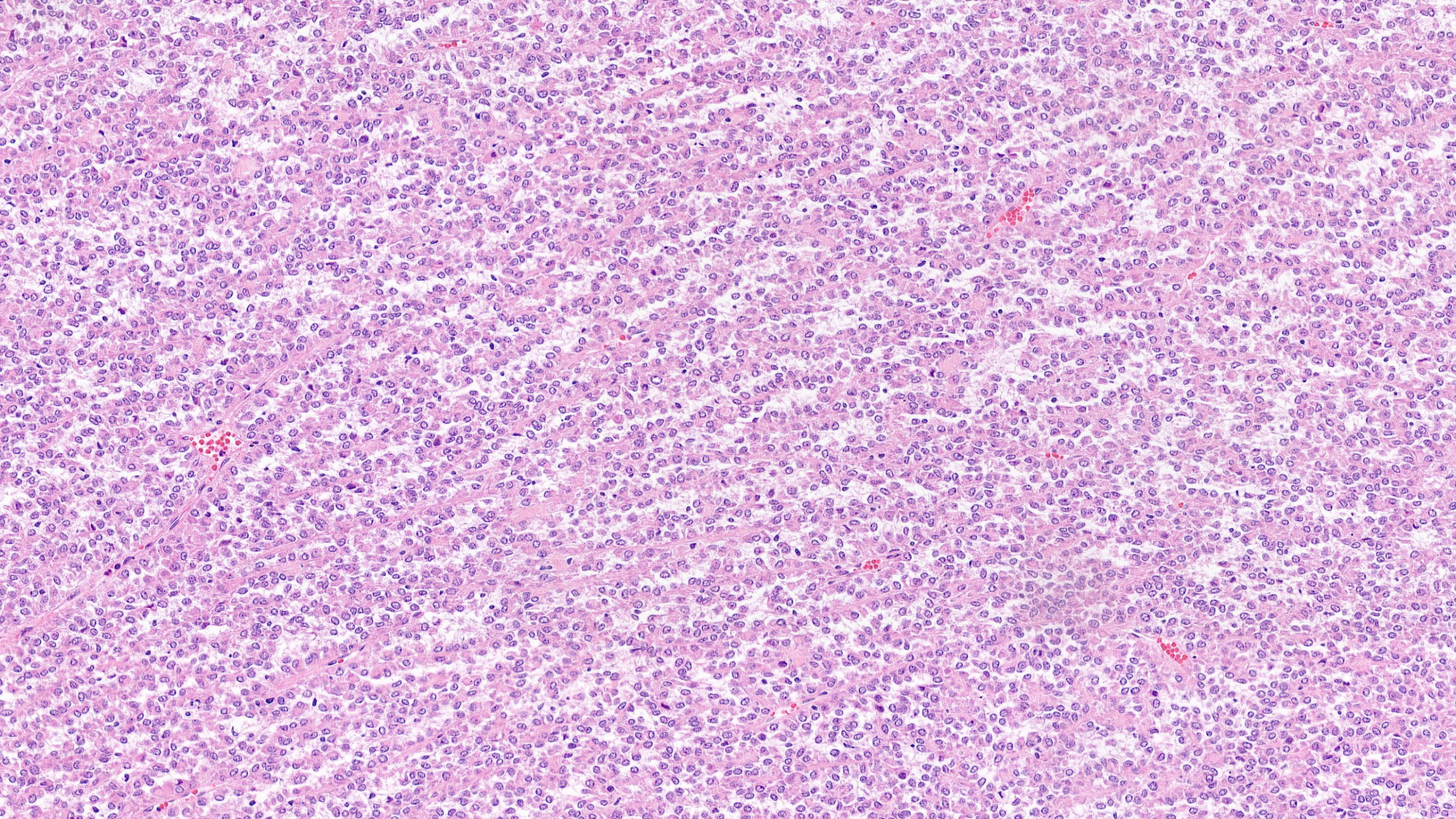
Melan-A (clon A103, Agilent-Dako): Positiu focal  
HMB45 (clon HMB45, Agilent-Dako): Positiu focal  
Desmina (clon D33, Agilent-Dako): Negatiu  
Actina de múscul llis (clon 1A4, Agilent-Dako): Negatiu  
CD10 (clon SP67, Ventana): Positiu focal  
Receptors d'estrògens (clon SP1, Agilent-Dako): Positiu focal  
Catepsina K (clon 3F9, Gennova): Positiu  
TFE3 (clon EPR11591, Gennova): Positiu  
Caldesmon (clon h-CD, Agilent-Dako): Negatiu (CPS < 10%)  
S100 (policlonal, Agilent-Dako): Negatiu  
SOX10 (clon BC-34, Biocare Medical): Negatiu  
Citoqueratina AE1 / AE3 (clon AE1/AE3, Agilent-Dako): Negatiu  
Alfa-Inhibina (clon R1, Agilent-Dako): Negatiu  
Receptors de progesterona (clon PgR1294, Agilent-Dako): Negatiu  
CD138 (clon MI15, Agilent-Dako): Negatiu  
CD163 (clon EDHu-1, BIO-RAD): Negatiu  
Kappa (policlonal, Ventana): Negatiu  
Lambda (policlonal, Ventana): Negatiu

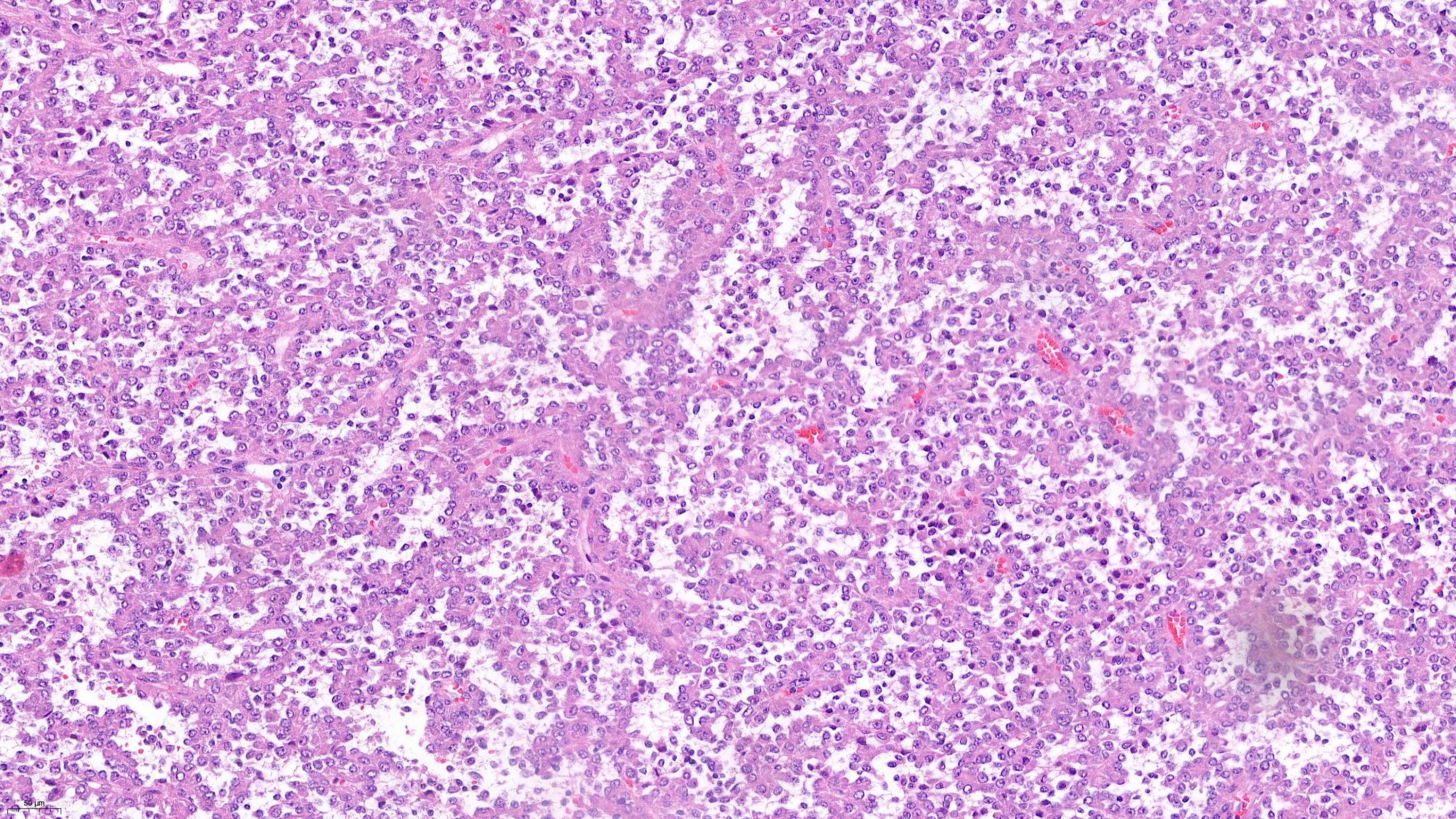
## COMENTARI I NOTES

Es tracta d'una tumoració que mostra un patró de creixement en nius constituïts per cèl.lules de morfologia epitelioide , amb un citoplasma eosinofílic i granular i nuclis monòtons amb atípia lleu. El perfil immunohistoquímic de la tumoració ( la positivitat per marcadors melanocítics i catepsina-K) i la seva la morfologia suggerixen que pugui tractar-se d'un tumor de cèl.lules epitelioides perivascular (PEComa).

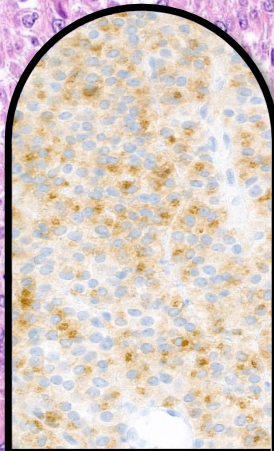
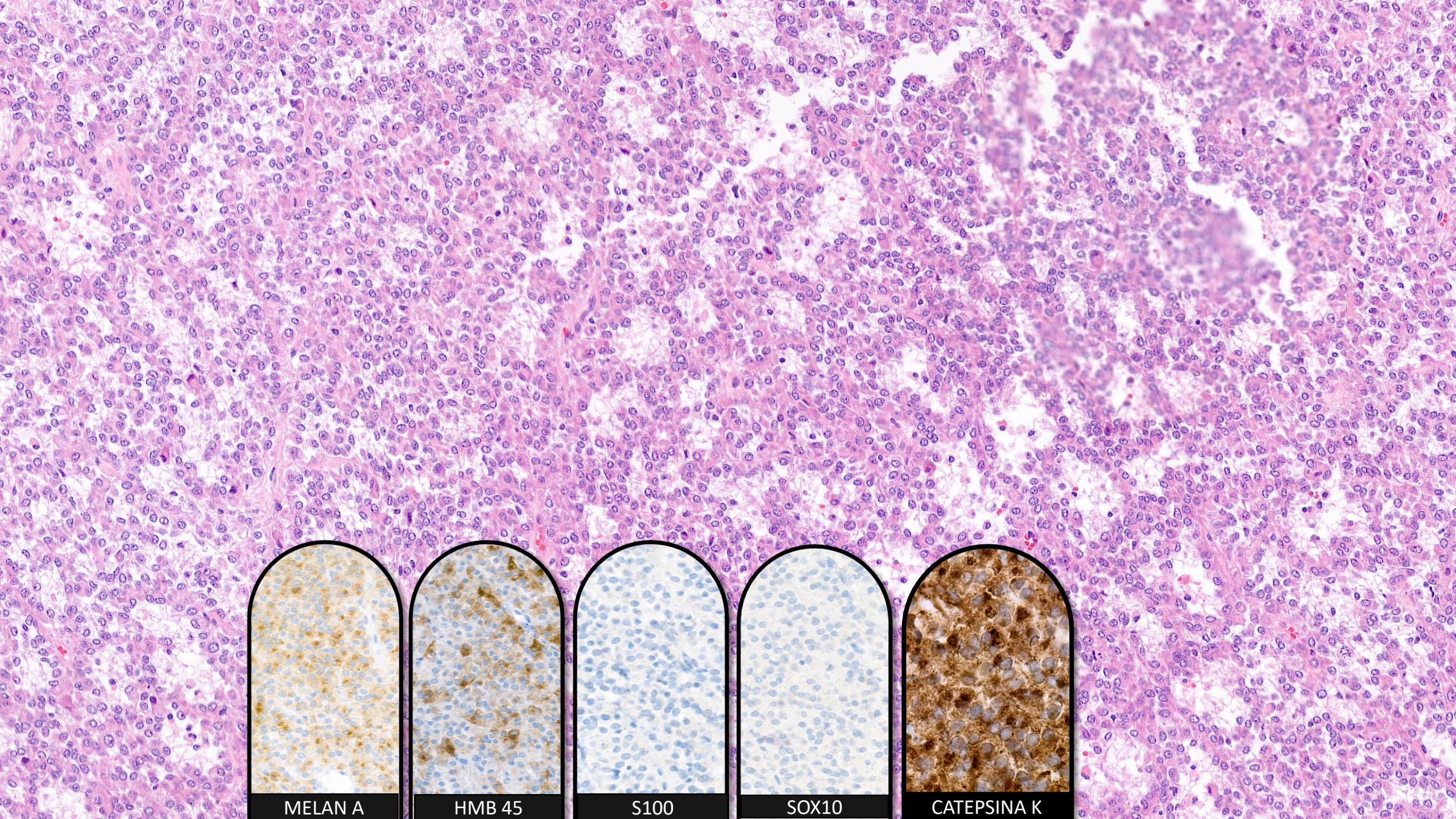




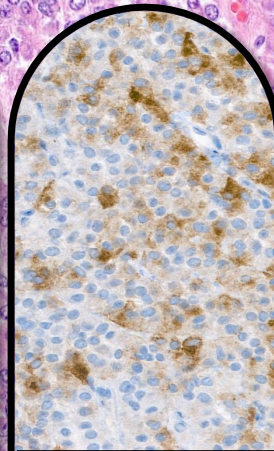




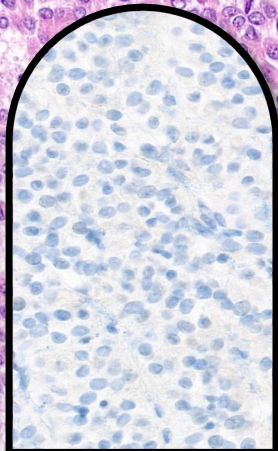
50  $\mu$ m



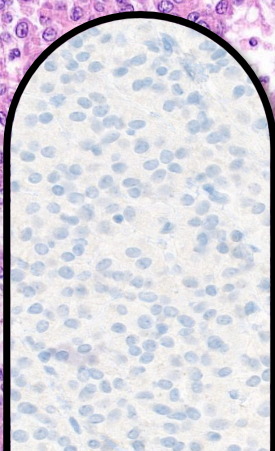
MELAN A



HMB 45



S100



SOX10

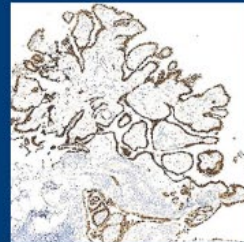
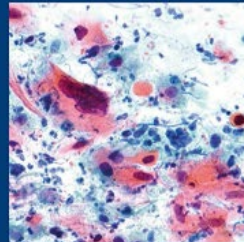
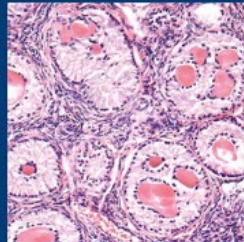
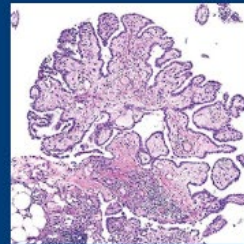
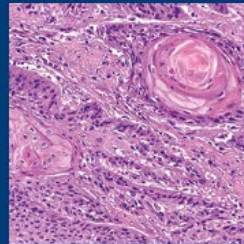
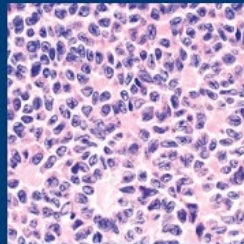


CATEPSINA K

WHO Classification of Tumours • 5th Edition

# Female Genital Tumours

Edited by the WHO Classification of Tumours Editorial Board



International Agency for Research on Cancer





Perivascular epithelioid cell tumour

# PEComa

## LOCALITZACIÓ

Els PEComas sorgeixen amb més freqüència en el **cos uterí** i amb menys freqüència en el coll uterí, la vagina, l'ovari i el lligament ample.

## CARACTERÍSTIQUES CLÍNIQUES

Els pacients solen presentar-se amb una **massa pèlvica**, sagnat anormal o dolor abdominopelvic.

## EPIDEMIOLOGIA

L'edat del pacient oscil·la entre els 16 i els 77 anys (**mitjana: 51 anys**).

## ETIOLOGIA

La majoria dels casos són esporàdics, però un subconjunt (~ 10%) s'associa amb l'esclerosi tuberosa.

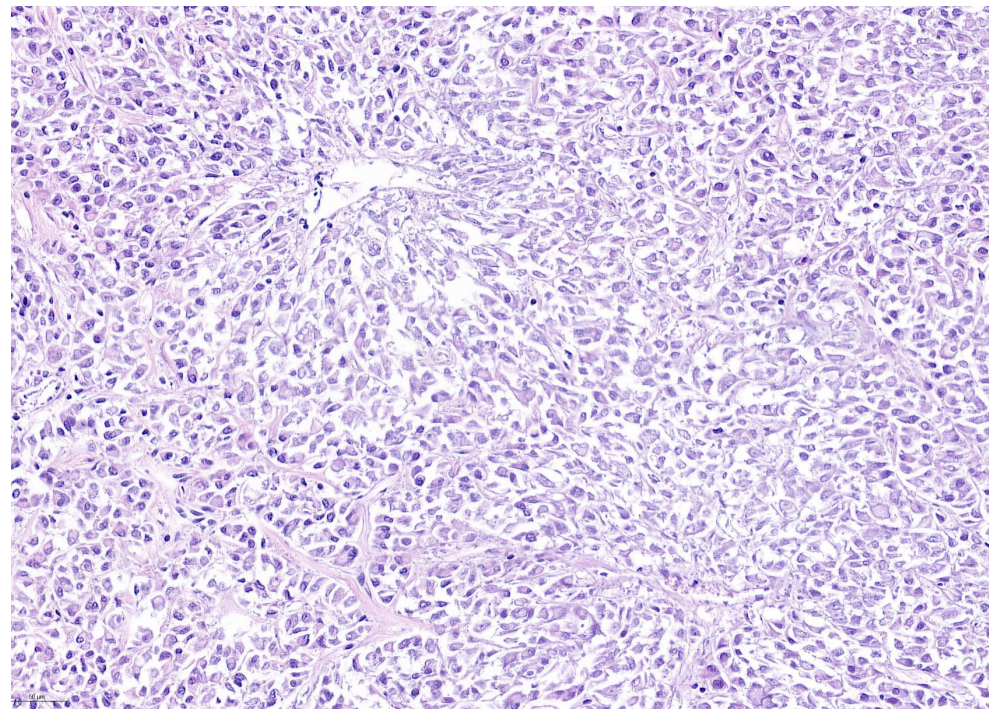
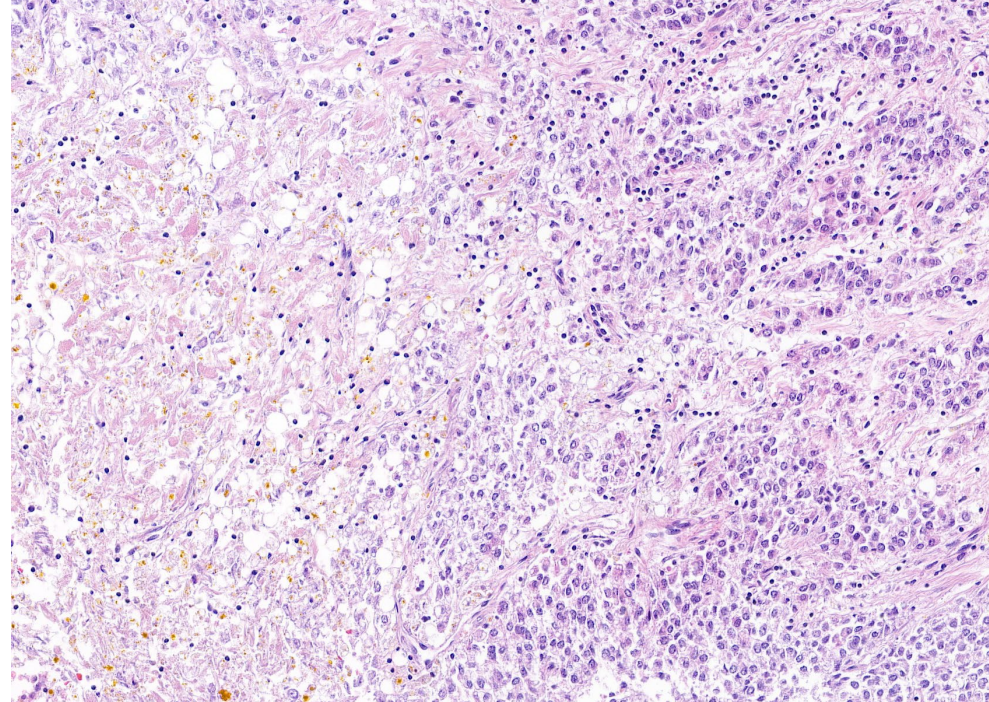
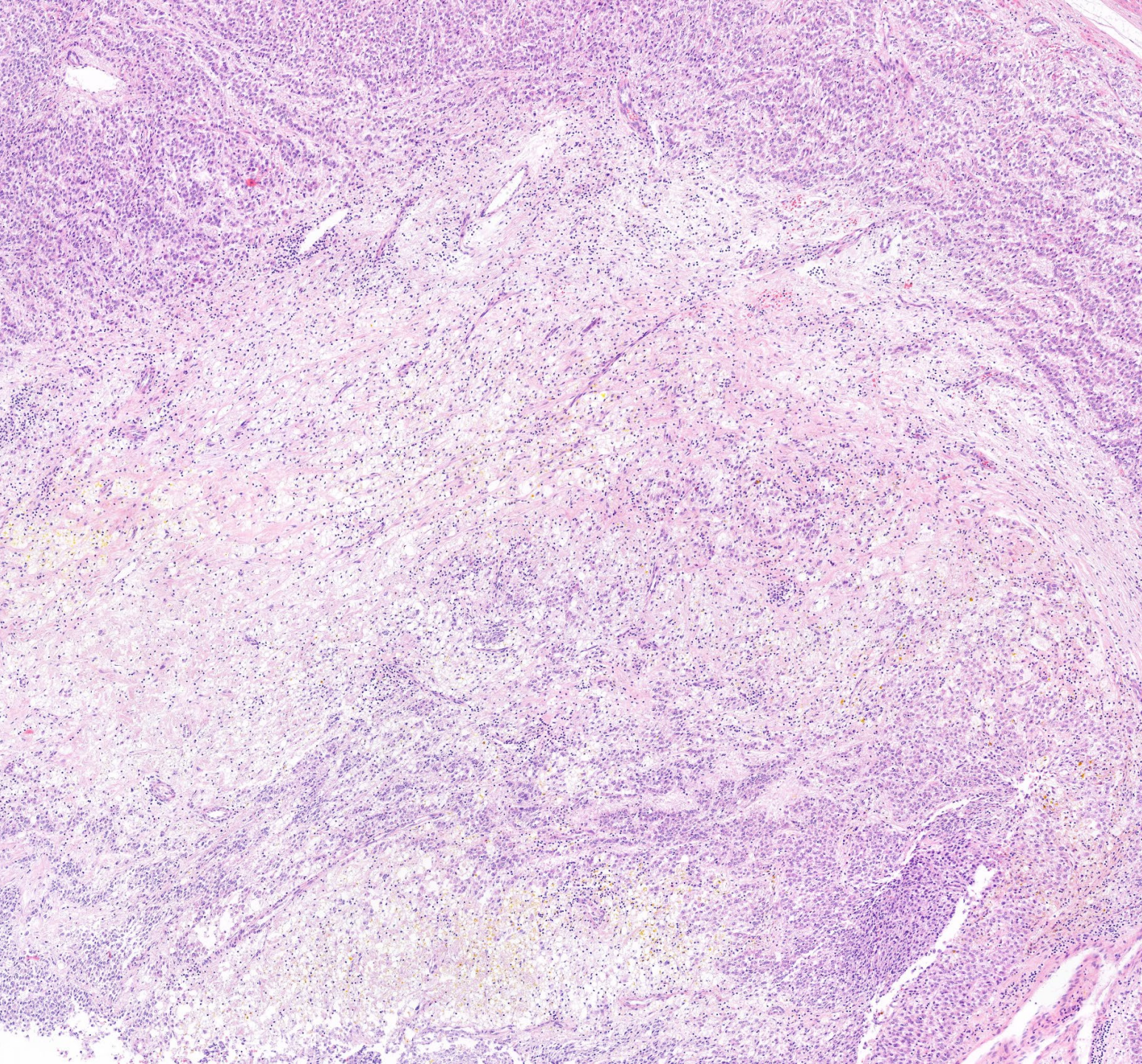
## PATOGÈNESI

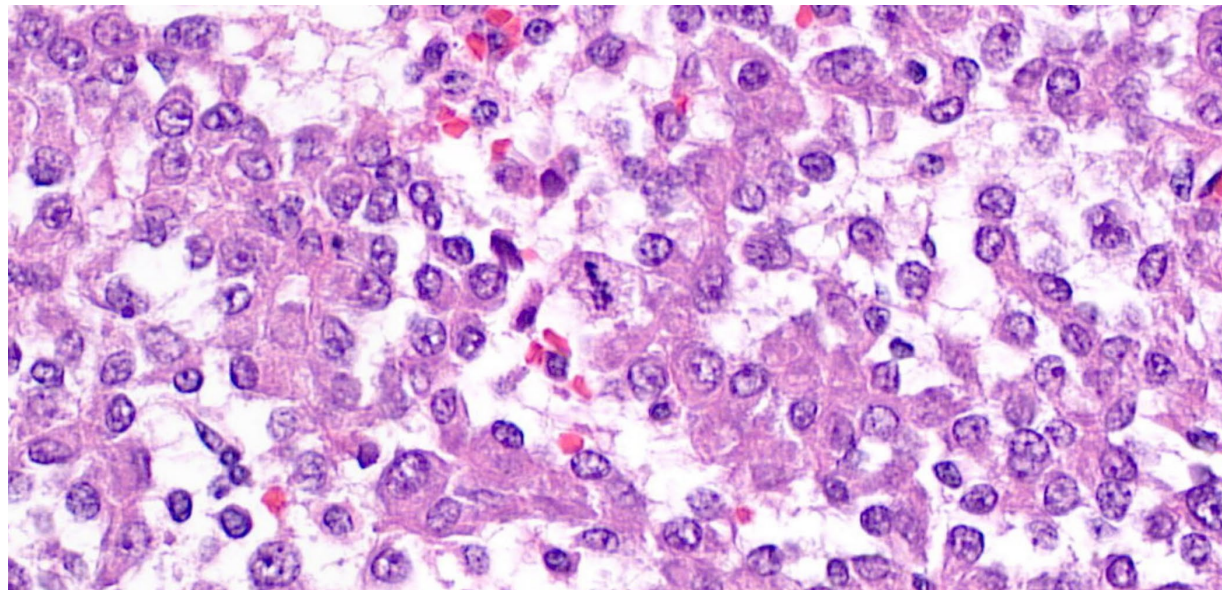
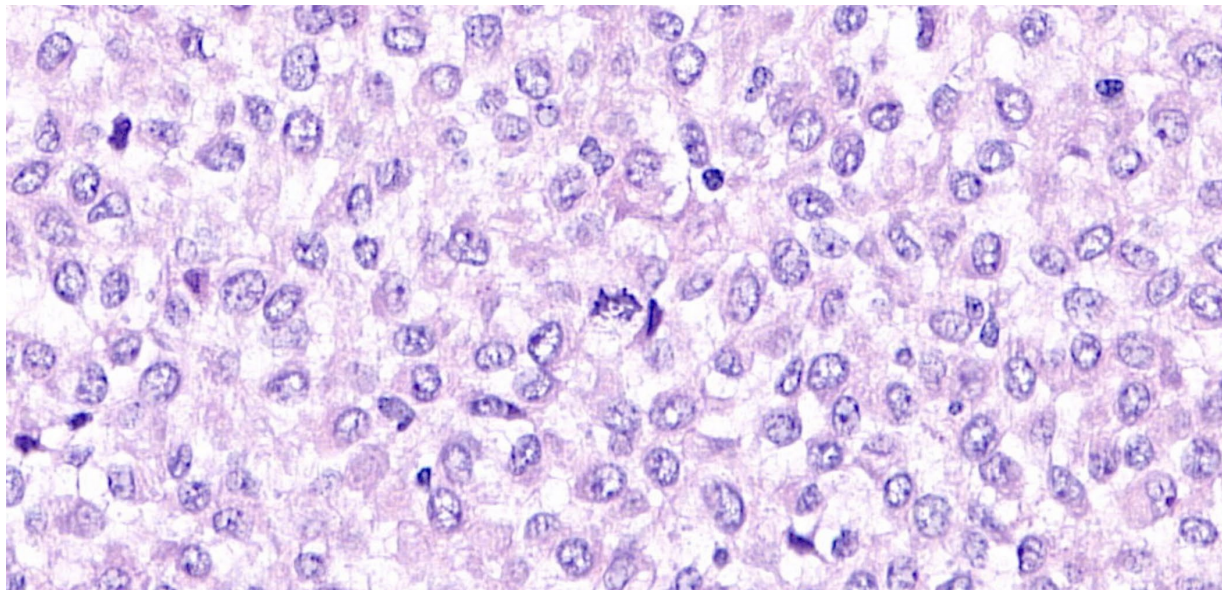
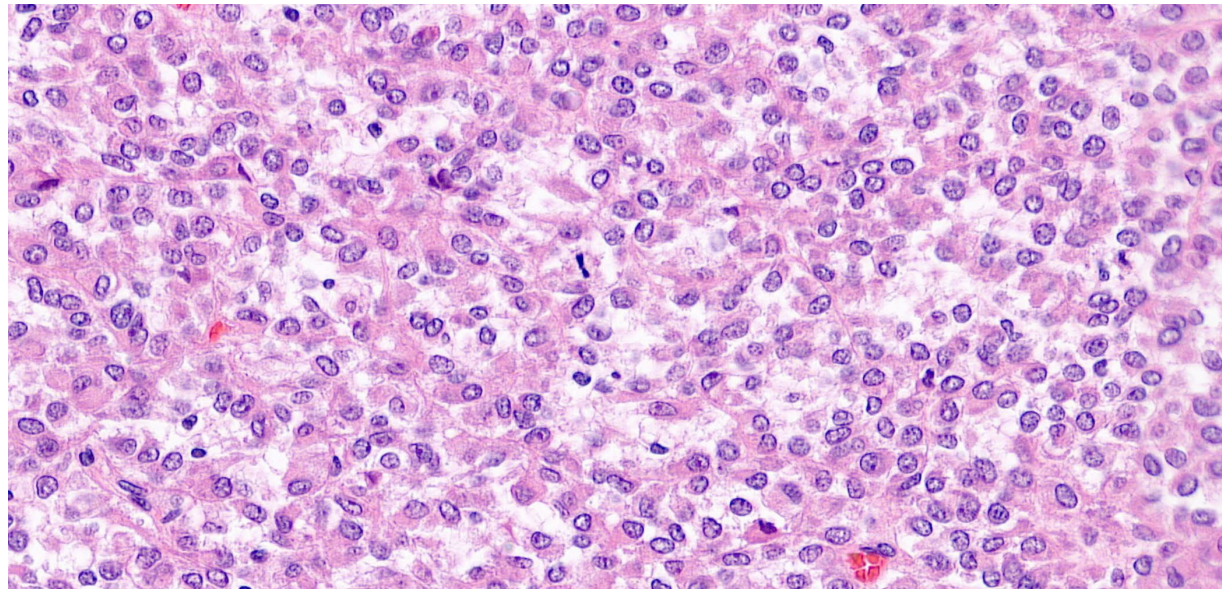
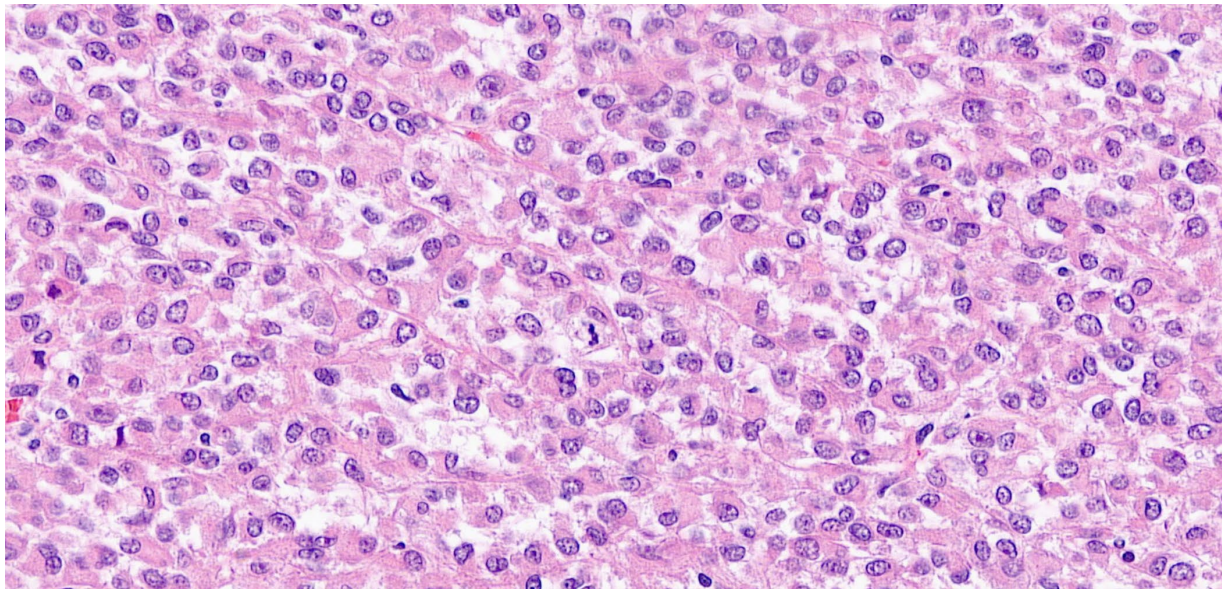
Les mutacions inactivants de **TSC1/TSC2** condueixen a sobrerregulació de senyalització mTOR. Alguns tumors **tenen fusions TFE3, RAD51B o HTR4-ST3GAL1**. Les mutacions TSC i les fusions TFE3 són **mútuament excloents**.

**ESSENCIAL:** Cèl·lules amb **citoplasma granular clar a eosinòfil; vasos de parets primes que envolten nius de cèl·lules; expressió d'HMB45 o melan-A, així com almenys un marcador de diferenciació mioide.**

**DESITJABLE:** confirmació de la reordenació o fusió de TFE3 en tumors reordenats per TFE3.









## HHS Public Access

Author manuscript

*Am J Surg Pathol.* Author manuscript; available in PMC 2019 October 01.

Published in final edited form as:

*Am J Surg Pathol.* 2018 October ; 42(10): 1370–1383. doi:10.1097/PAS.0000000000001119.

### Uterine PEComas: A Morphological, Immunohistochemical, and Molecular Analysis of 32 Tumors

Jennifer A. Bennett<sup>1</sup>, Ana Costa Braga<sup>2</sup>, Andre Pinto<sup>3</sup>, Koen Van de Vijver<sup>4</sup>, Kristine Cornejo<sup>5</sup>, Anna Pesci<sup>6</sup>, Lei Zhang<sup>7</sup>, Vicente Morales-Oyarvide<sup>8</sup>, Takako Kiyokawa<sup>9</sup>, Gian Franco Zannoni<sup>10</sup>, Joseph Carlson<sup>11</sup>, Tomas Slavik<sup>12</sup>, Carmen Tornos<sup>13</sup>, Cristina R. Antonescu<sup>7</sup>, and Esther Oliva<sup>14</sup>



## Classification of Uterine PEComas Using Non-Gynecologic Specific (Original) Criteria

Classification	Definition	PEComas With Aggressive Behavior Meeting Criteria	PEComas Without Aggressive Behavior Meeting Criteria *
<b>Benign</b>	< 5 cm, non-infiltrative, non-high-grade atypia, mitoses $\leq$ 1/50 HPFs, no necrosis, no LVI	0/0 (0%)	1/19 (5%)
<b>Uncertain Malignant Potential</b>	1) Nuclear pleomorphism/multinucleated giant cells or 2) > 5 cm	0/0 (0%)	1/19 (5%)
<b>Malignant</b>	2+ features (> 5 cm, infiltrative, high-grade atypia, mitoses > 1/50 HPFs, necrosis, LVI)	11/11 (100%)	6/19 (32%)

\* 11 tumors without known malignancy could not be classified using the above criteria

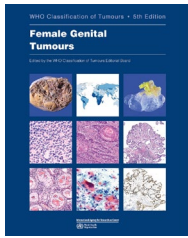
HPF=high-power field; LVI=lymphovascular invasion

## Classification of Uterine PEComas Using Gynecologic-Specific Criteria

Classification	Definition	PEComas With Aggressive Behavior Meeting Criteria	PEComas Without Aggressive Behavior Meeting Criteria
<b>Benign/Uncertain Malignant Potential</b>	< 4 features ( $\geq$ 5 cm, high-grade atypia, mitoses > 1/50 HPFs, necrosis, LVI)	4/11 (36%)	19/19 (100%)
<b>Malignant</b>	$\geq$ 4 features	7/11 (64%)	0/19 (0%)

HPF=high-power field; LVI=lymphovascular invasion





## Perivascular epithelioid cell tumors (PEComa) of the gynecologic tract

Jennifer A. Bennett MD<sup>1</sup> | Esther Oliva MD<sup>2</sup>



Subsequently, in 2018, Bennett and colleagues applied both of these algorithms to their study of 32 uterine PEComas.<sup>17</sup> Application of the original criteria resulted in similar findings to Schoolmeester et al., with all aggressive PEComas being classified as malignant, but 37% of benign tumors were misclassified as malignant and 37% were not able to be classified. Using the modified criteria, 36% (4/11) of aggressive PEComas were misclassified as benign/uncertain malignant potential, with three of these tumors showing three worrisome features. Thus, they proposed a revised system with a threshold of three atypical features for the diagnosis of “malignant”. Furthermore, they recommended elimination of the word “benign” in the first category as one of their aggressive PEComas had only one atypical feature (size >5 cm), but still recurred. This revised classification has since been adopted by the 2020 WHO classification of female genital tract tumors.<sup>91</sup>





Perivascular epithelioid cell tumour

# PEComa

	General criteria { 16327428 }	Modified gynaecology-specific criteria { 24418852 ; 30001237 }
<b>Benign</b>	< 5 cm, non-infiltrative, non-high nuclear grade, mitotic count of ≤ 1 mitosis/12 mm <sup>2</sup> , no necrosis, no vascular invasion	
<b>Uncertain malignant potential</b>	Nuclear pleomorphism / multinucleated giant cells or > 5 cm	< 3 of the following features: ≥ 5 cm, high nuclear grade, mitotic count of > 1 mitosis/12 mm <sup>2</sup> , necrosis, vascular invasion
<b>Malignant</b>	≥ 2 of the following features: > 5 cm, infiltrative, high nuclear grade, mitotic count of > 1 mitosis/12 mm <sup>2</sup> , necrosis, vascular invasion	≥ 3 features



## HHS Public Access

Author manuscript

Mod Pathol. Author manuscript; available in PMC 2022 March 31.

Published in final edited form as:

Mod Pathol. 2022 April ; 35(4): 515–523. doi:10.1038/s41379-021-00855-1.

### Uterine PEComas: Correlation Between Melanocytic Marker Expression and *TSC* Alterations/*TFE3* Fusions

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TSC Alteration

Classic  
PEComa

In summary, we identified *TSC* or *TFE3* alterations in most (18/19; 95%) uterine tumors morphologically and immunohistochemically compatible with PEComa, and 14/17 (82%) had > 50% HMB-45 expression. We also recognized that most malignant PEComas harbor alterations in *TP53*, *ATRX*, or *RB1*, and rare PEComas are mismatch repair protein deficient/microsatellite unstable. Finally, detection of identical mutations (*ATRX*, *ADGRVI*) in a morphologically and immunohistochemically biphasic tumor that subsequently acquired different alterations (*TSC1* mutation versus single copy deletions) in the different components, concludes this is a clonal neoplasm that underwent two pathways of evolution. Further studies are necessary to explore this finding and ultimately determine the relationship between PEComas and smooth muscle tumors.

Meso  
Neop  
Myom  
Differ

No *TSC*, *TFE3*, or  
LMS\* alterations  
and <50% HMB-45

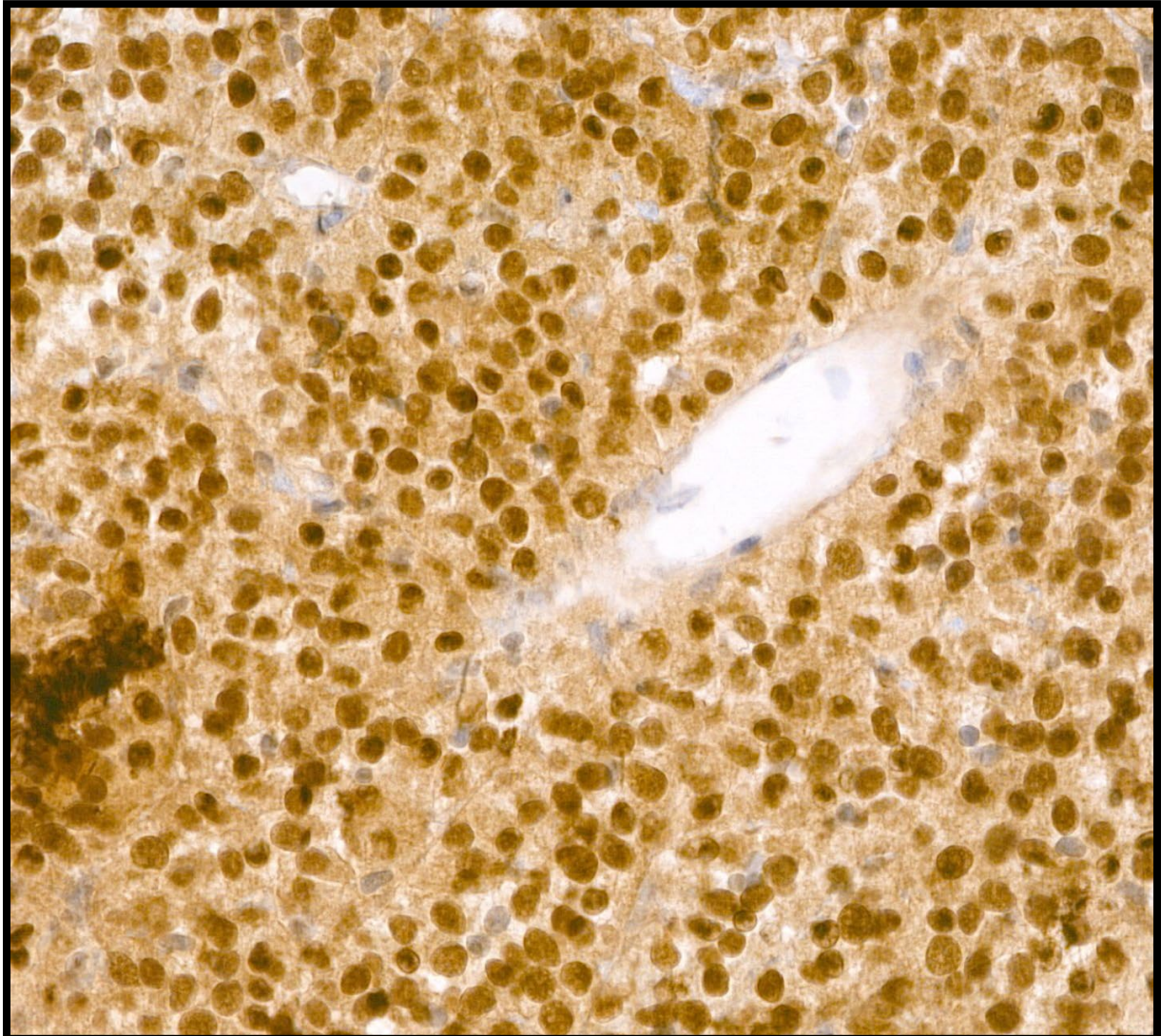
Descriptive  
Diagnosis



\**TP53*, *ATRX*, *RB1*, and/or *MED12* alterations in the absence of *TSC/TFE3* alterations

#### Figure 4.

Proposed algorithm for evaluation of a uterine tumor with myomelanocytic differentiation by immunohistochemistry.



TFE3

NGS







Perivascular epithelioid cell tumour

# PEComa

## LOCALITZACIÓ

Els PEComas sorgeixen amb més freqüència en el corpus uterí i amb menys freqüència en el coll uterí, la vagina, l'ovari i el lligament ample.

## CARACTERÍSTIQUES CLÍNIQUES

Els pacients solen presentar-se amb una massa pèlvica, sagnat anormal o dolor abdominopelvic.

## EPIDEMIOLOGIA

L'edat del pacient oscil·la entre els 16 i els 77 anys (mitjana: 51 anys).

## ETIOLOGIA

La majoria dels casos són esporàdics, però un subconjunt (~ 10%) s'associa amb l'esclerosi tuberosa.

## PATOGÈNESI

Les mutacions inactivants de **TSC1/TSC2** condueixen a l'upregulació de senyalització mTOR. Alguns tumors **tenen fusions TFE3, RAD51B o HTR4-ST3GAL1**. Les mutacions TSC i les fusions TFE3 són **mútuament excloents**.

## ESTADIATGE

El PEComa maligne s'estadifica com els sarcomes uterins.

**ESSENCIAL:** cèl·lules amb citoplasma granular clar a eosinòfil; vasos de parets primes que envolten nius de cèl·lules; expressió d'HMB45 o melan-A, així com almenys un marcador de diferenciació mioide.

**DESITJABLE:** confirmació de la reordenació o fusió de TFE3 en tumors reordenats per TFE3.

- **TUMOR DE CÈL·LULES EPITELIOIDES PERIVASCULARS MALIGNE (PEComa maligne) DE L'ÚTER**

(OMS 2020), DE 9 x 5 x 5 cm.

- Grau d'atípia citològica: MODERADA
  - Localització: TUMOR LIMITAT A LÚTER (subserós a fundus uterí).
  - Marge de creixement: INFILTRATIU.
  - Índex mitòtic: 8 figures de mitosi / 12 mm<sup>2</sup>.
  - Necrosi: PRESENT.
  - Invasió vascular: NO S'OBSERVA.
- 
- Afectació cervical: NO.
  - Afectació tubàrica: NO.
  - Marge de resecció cervical: LLIURE.
  - Marges de resecció laterals: LLIURE.
  - Ovaris i trompes uterines: SENSE ALTERACIONS HISTOLÒGICS SIGNIFICATIVES.
  - Endometri: ATRÒFIC SENSE ATÍPIA.
  - Altres lesions:
    - .DOS LIOMIOMES UTERINS, EL MÉS GRAN DE 2 CM.

CONCLUSIÓ: **pT1b pNx (UICC 2017) FIGO IB (FIGO 2018) .**

**COMITÈ  
MULTIDISCIPLINARI:**

SEGUIMENT C/4  
MESOS + ESTUDIS  
TOMOGRÀFICS  
ANUALS.

# SOUVENIRS

- Els PEComes són tumors **poc freqüents**.
- El principal diagnòstic diferencial es amb **tumors de múscul llis**.
- Els criteris de malignitat dels PEComes de parts toves i ginecològics són diferents.
  - **Índex mitòtic, necrosi, invasió limfovascular, atípia citològica i grandària tumoral.**
- Malgrat que no es troben alteracions moleculars típiques dels PEComes, es pot realitzar/suggerir aquest diagnòstic.

EL ÁVILA

(CARACAS, VENEZUELA)





!!!NOUS PATÒLEGS!!!







## Risk stratification and outcomes in 210 gynecologic perivascular epithelioid cell tumors (PEComas) cases

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**Table 2** Analysis of clinicopathologic features related to gynecological PEComas recurrence and/or metastasis

Variable	Chi-square test				Uni-variate Cox analysis							Multi-variate Cox analysis			
	R/M rate*	Risk ratio	95% CI	P value	R/M rate after surgery	Risk ratio	95% CI	P value	death rate	Risk ratio	95% CI	P value	Risk ratio	95% CI	P value
<i>Primary location</i>															
< 2	7/12 (58.3%)	1			5/11 (45.5%)	1			8/137 (5.8%)	1					
≥ 2	46/144 (31.9%)	1.826	1.070– 3.116	0.124	30/138 (21.7%)	2.091	1.017– 4.298	0.233	2/10 (20.0%)	3.425	0.836– 14.032	0.055			
<i>Tumor size</i>															
< 5 cm	8/56 (14.3%)	1			6/55 (10.9%)	1			1/55 (1.8%)	1		0.055	1		
≥ 5 cm	43/96 (44.8%)	3.135	1.590– 6.182	< 0.001	29/90 (32.2%)	2.954	1.311– 6.656	0.01	8/87 (9.2%)	5.057	0.650– 39.335	0.835	0.129– 5.388	0.850	
<i>Infiltrative pattern</i>															
No	7/60 (11.7%)	1			5/58 (8.6%)	1			2/56 (3.4%)	1		1			
Yes	39/82 (47.6%)	4.077	1.960– 8.480	< 0.001	27/77 (35.1%)	4.068	1.668– 9.918	< 0.001	5/75 (6.7%)	1.933	0.389– 9.611	0.333	0.976	0.145– 6.571	0.980
<i>Mitotic rate</i>															
Low	7/70 (10.0%)	1			3/68 (4.4%)	1			0/68 (0%)	1		1			
High	40/71 (56.3%)	5.634	2.710– 11.714	< 0.001	28/65 (43.1%)	9.764	3.119– 30.563	< 0.001	9/64 (14.1%)	–	–	< 0.001	13.170	0.918– 188.840	0.058
<i>Grade</i>															
Mild to moderate	19/87 (21.8%)	1			8/82 (9.8%)	1			2/82 (2.4%)	1		1			
Severe	28/51 (54.9%)	2.514	1.573– 4.018	< 0.001	24/49 (49.0%)	5.02	2.449– 10.290	< 0.001	7/47 (14.9%)	6.106	1.322– 28.201	0.005	12.05	1.245– 116.599	0.032
<i>Necrosis</i>															
Absent	10/77 (13.0%)	1			5/73 (6.8%)	1			1/74 (1.4%)	1		1			
Present	38/65 (58.5%)	4.502	2.438– 8.313	< 0.001	25/56 (44.6%)	6.518	2.664– 15.949	< 0.001	5/59 (15.3%)	11.288	1.472– 86.593	0.001	0.726	0.156– 3.371	0.682
<i>Vascular invasion</i>															
Absent	14/59 (23.7%)	1			10/60 (16.7%)	1			2/61 (3.3%)	1		1			
Present	19/25 (76.0%)	3.203	1.928– 5.322	< 0.001	15/25 (60.0%)	3.6	1.879– 6.896	0.001	5/23 (21.7%)	6.63	1.382– 31.811	0.25	0.709	0.173– 2.908	0.633

R/M rate metastasis rate before surgery and recurrence and/or metastasis rate after surgery were included, CI confidence interval