



Hospital Clínic  
Universitari de València

**SéAP-IAP**  
[Sociedad Española de Anatomía Patológica]  
[International Academy of Pathology]

# **TODO ESTÁ EN LOS LIBROS**

## **(Y EN LOS MASTOCITOS)**

Yassin El Bouzaydy El Gueddari  
Antonio Ferrández Izquierdo - María Carolina Martínez Ciarpaglini

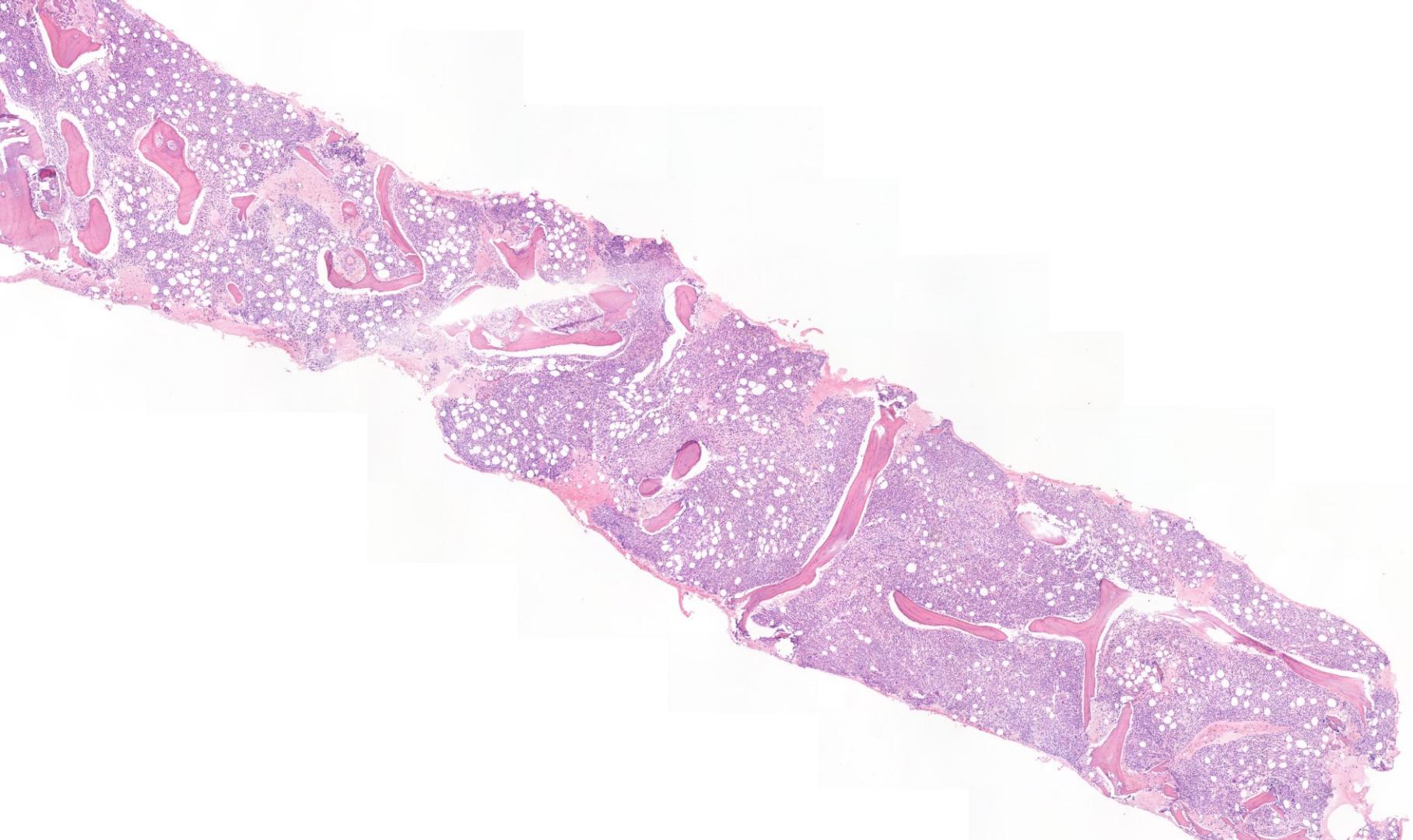
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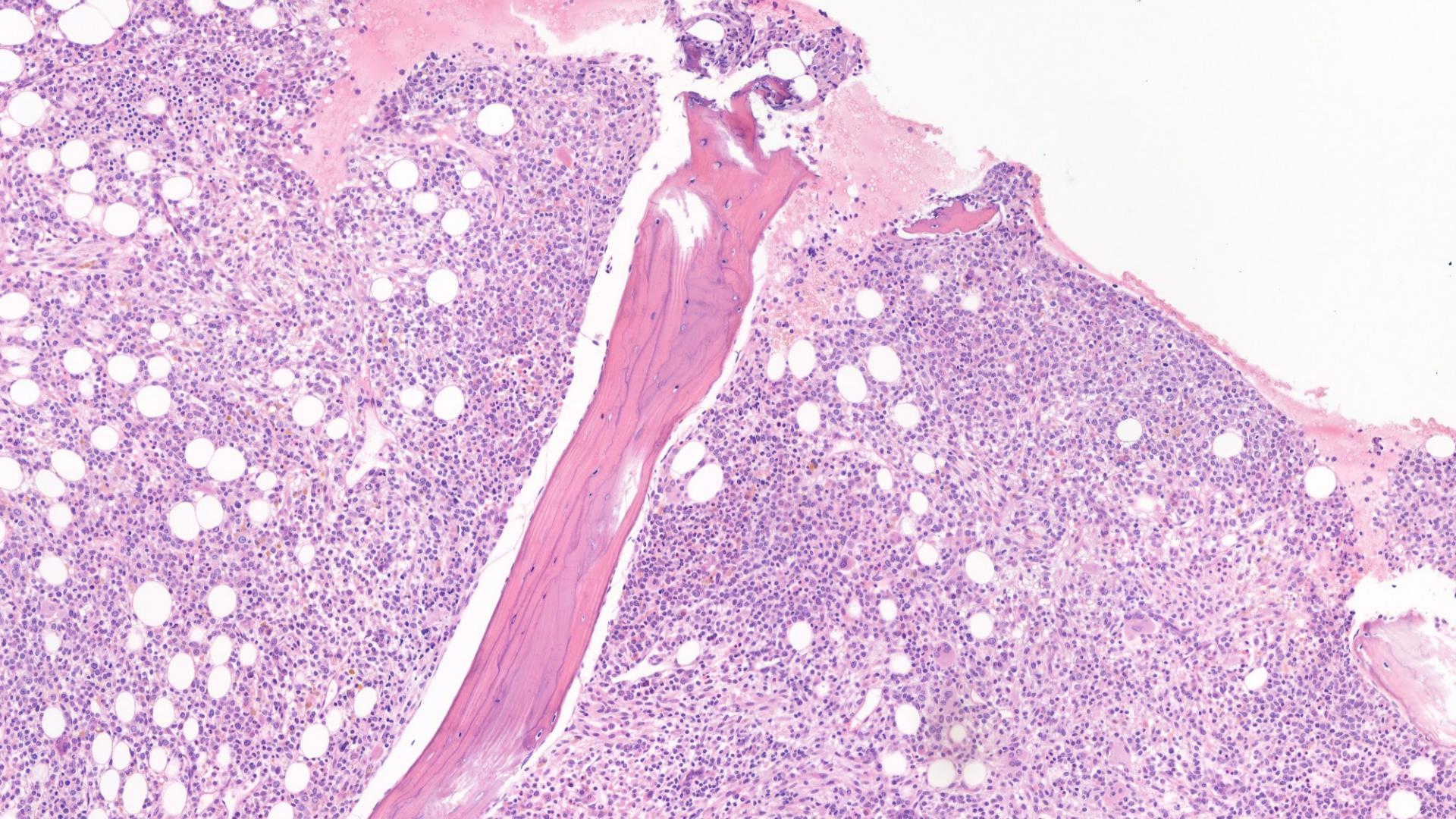


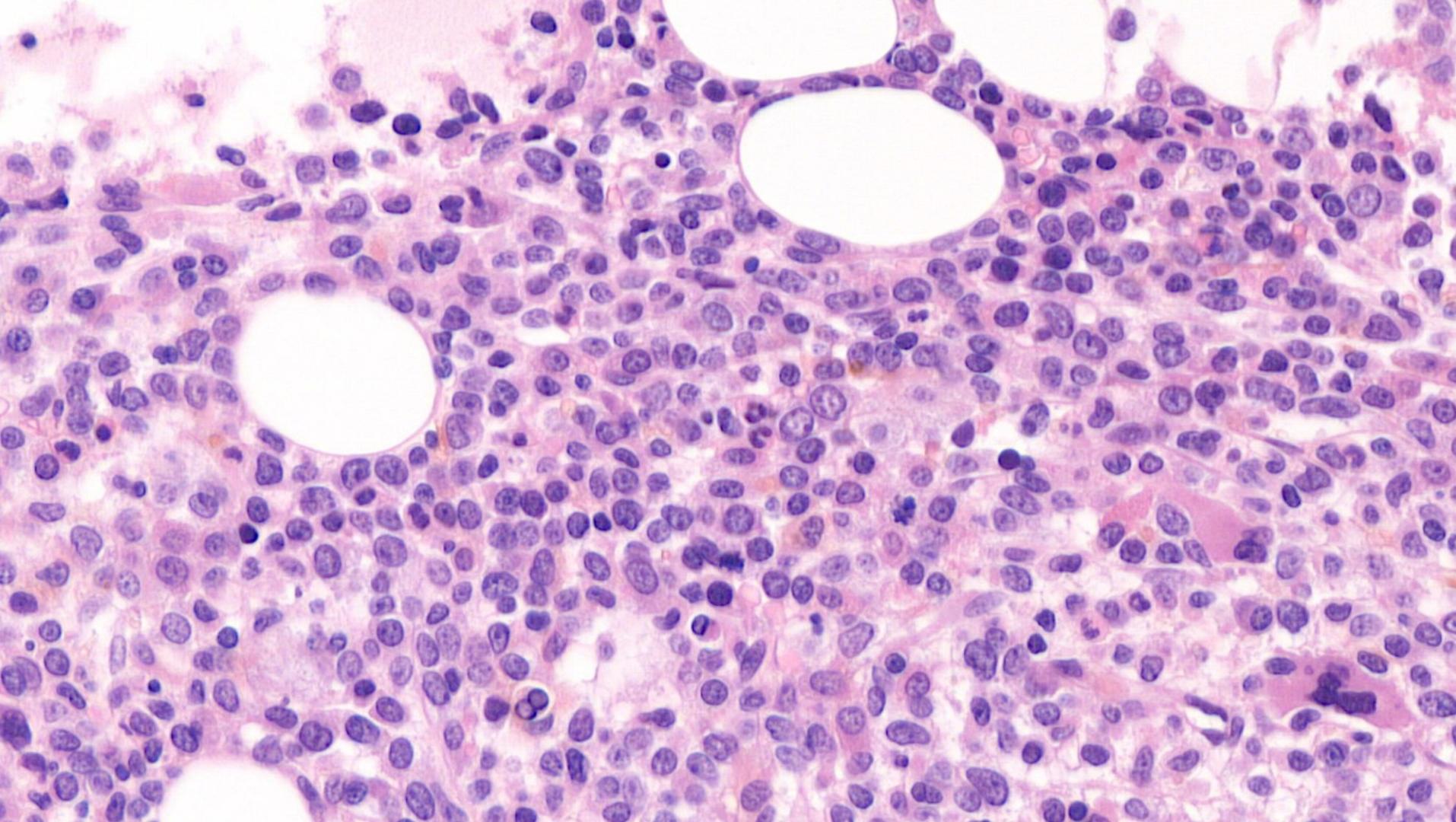
# Nuestra paciente

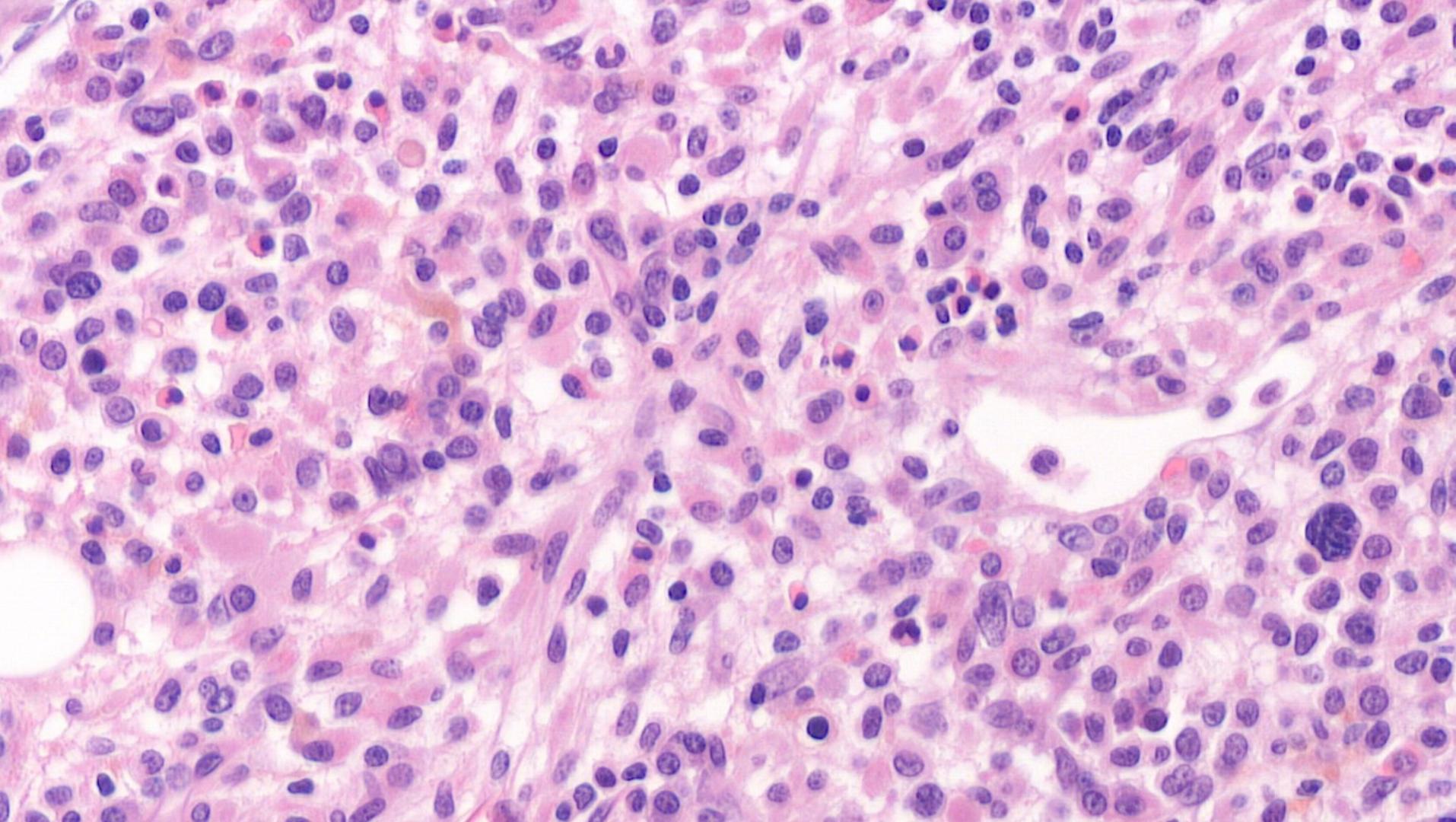
- Mujer de 68 años
- Fumadora. Mastocitosis cutánea diagnosticada hace 4 años
- Astenia y pérdida de peso
- Anemia, trombopenia y leucocitosis

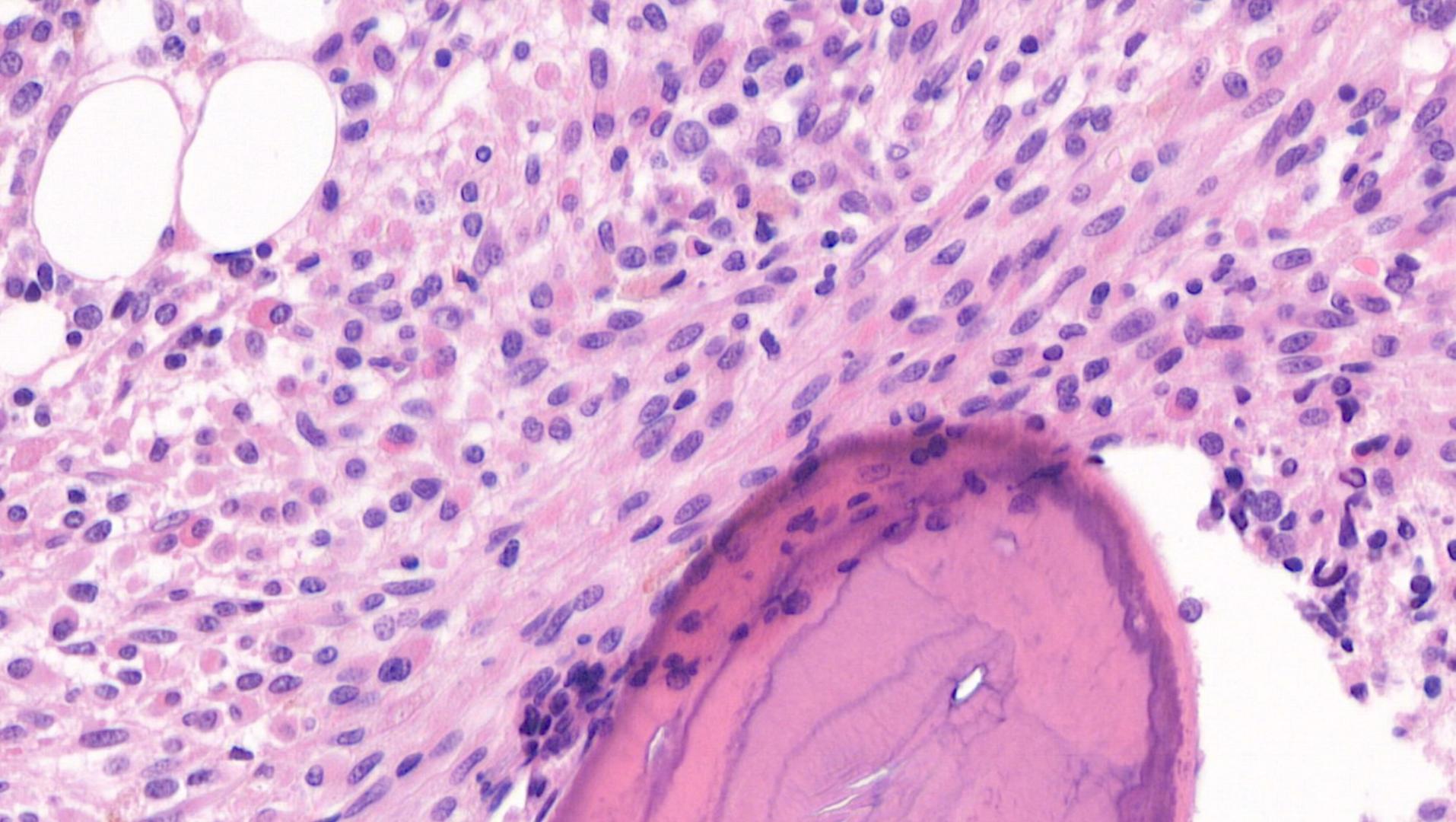


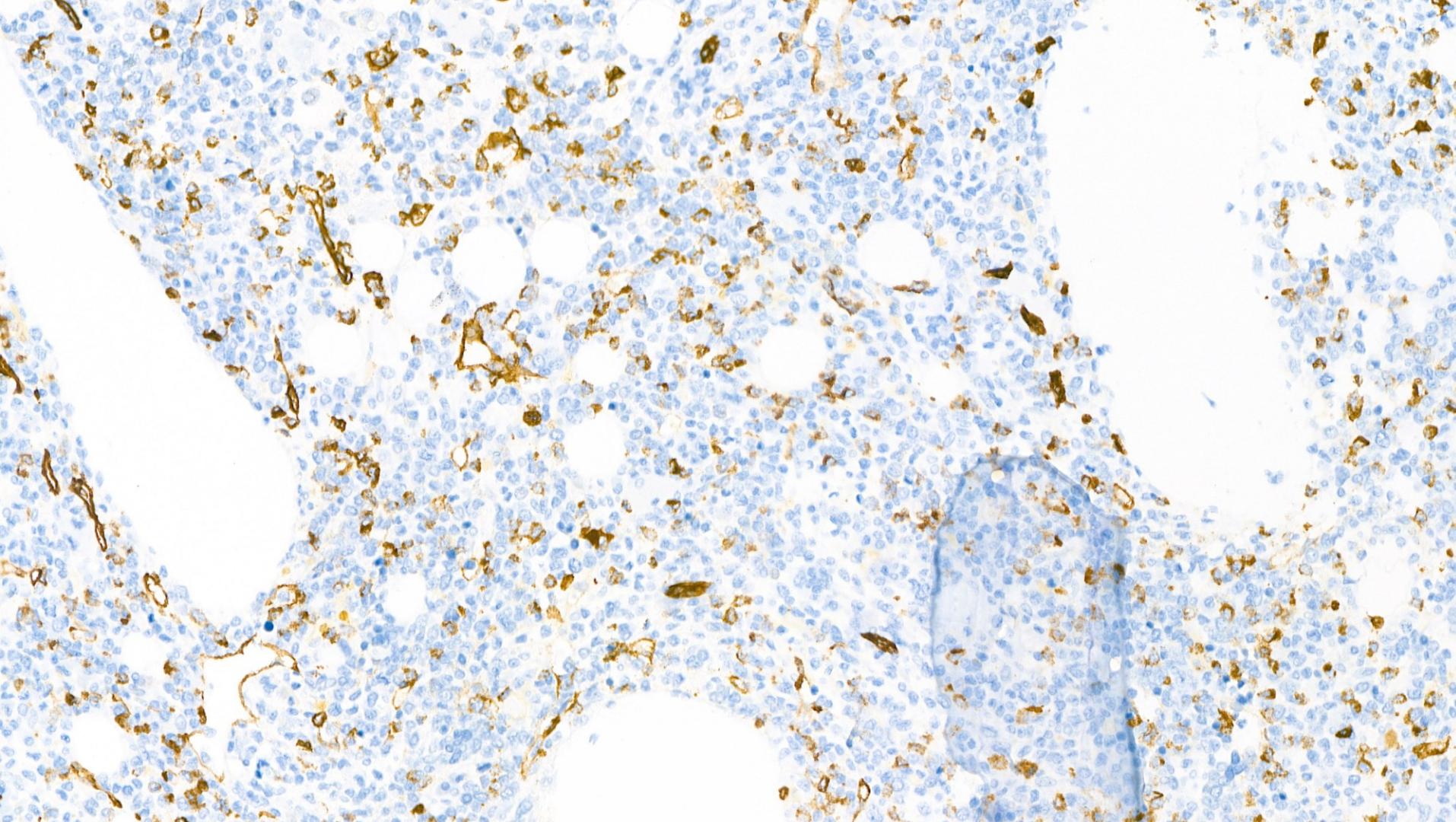


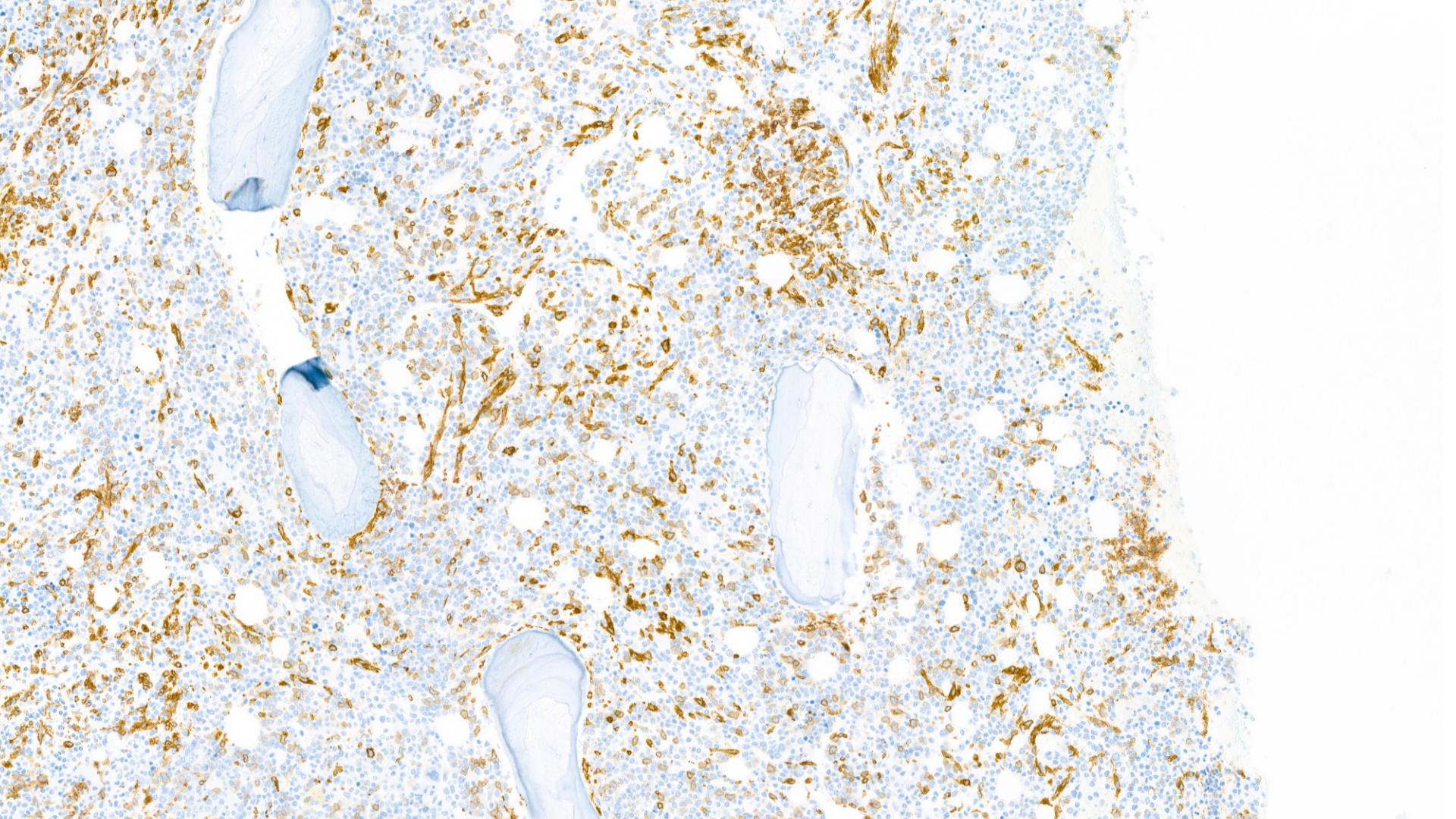


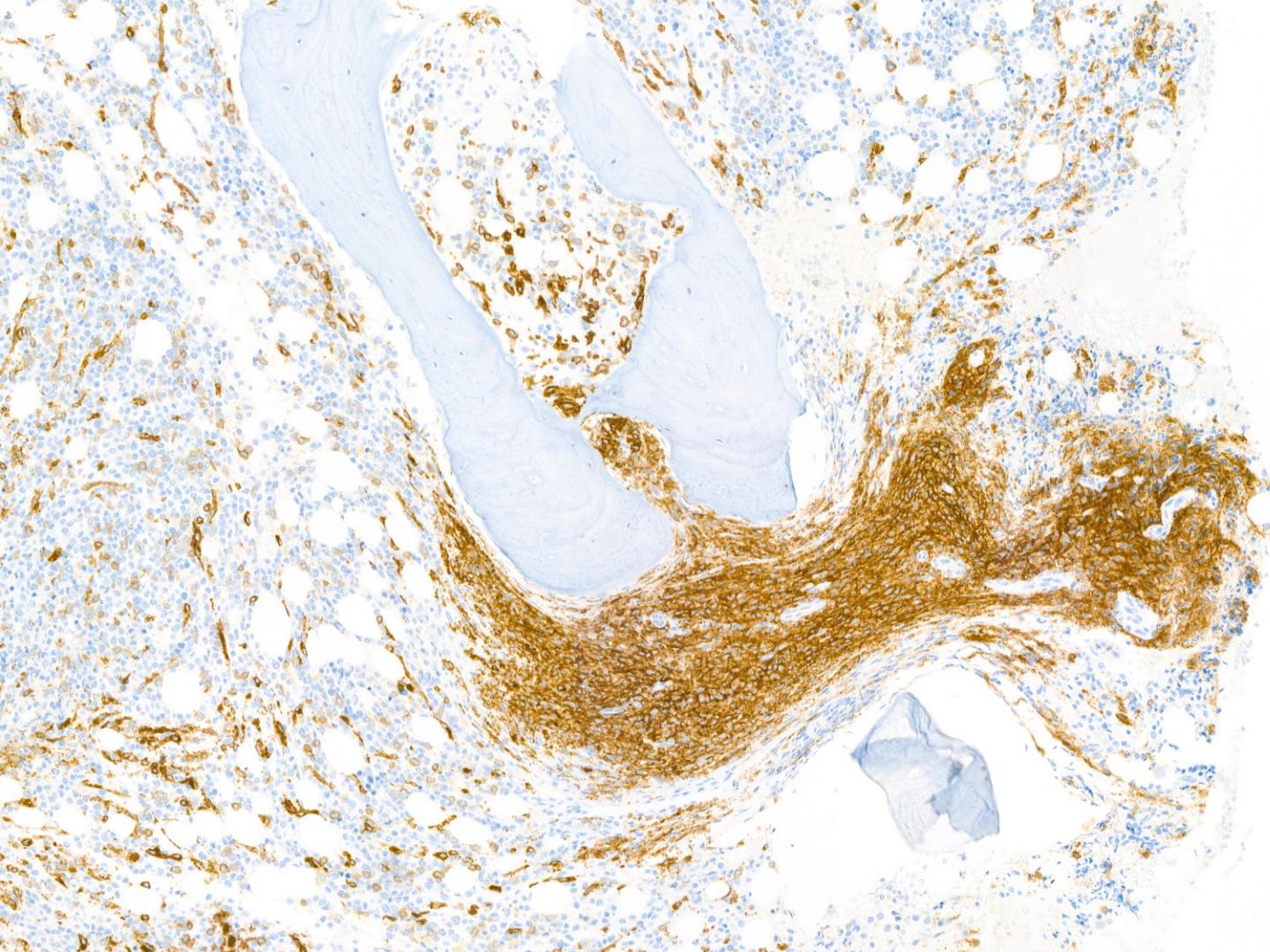


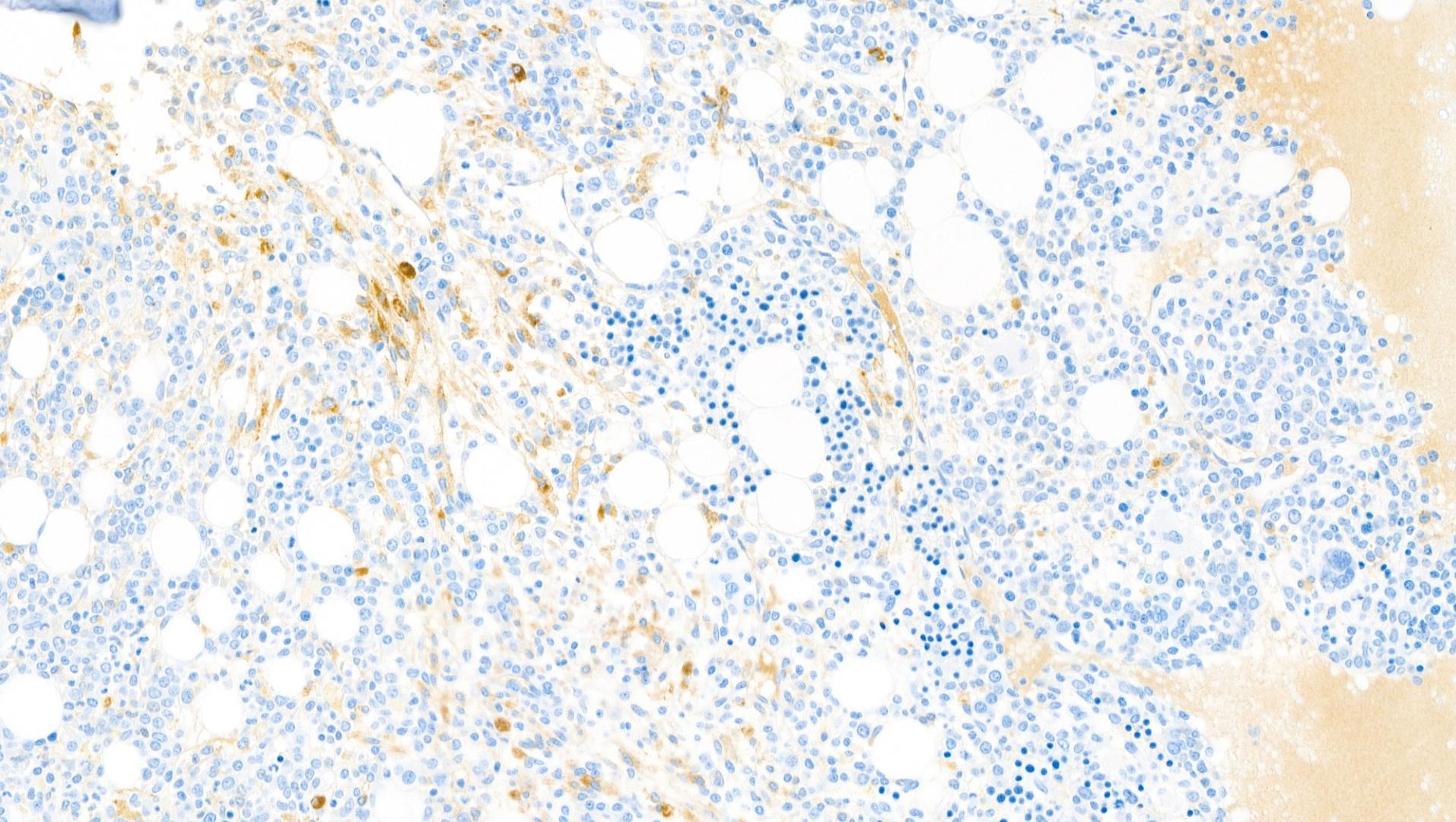


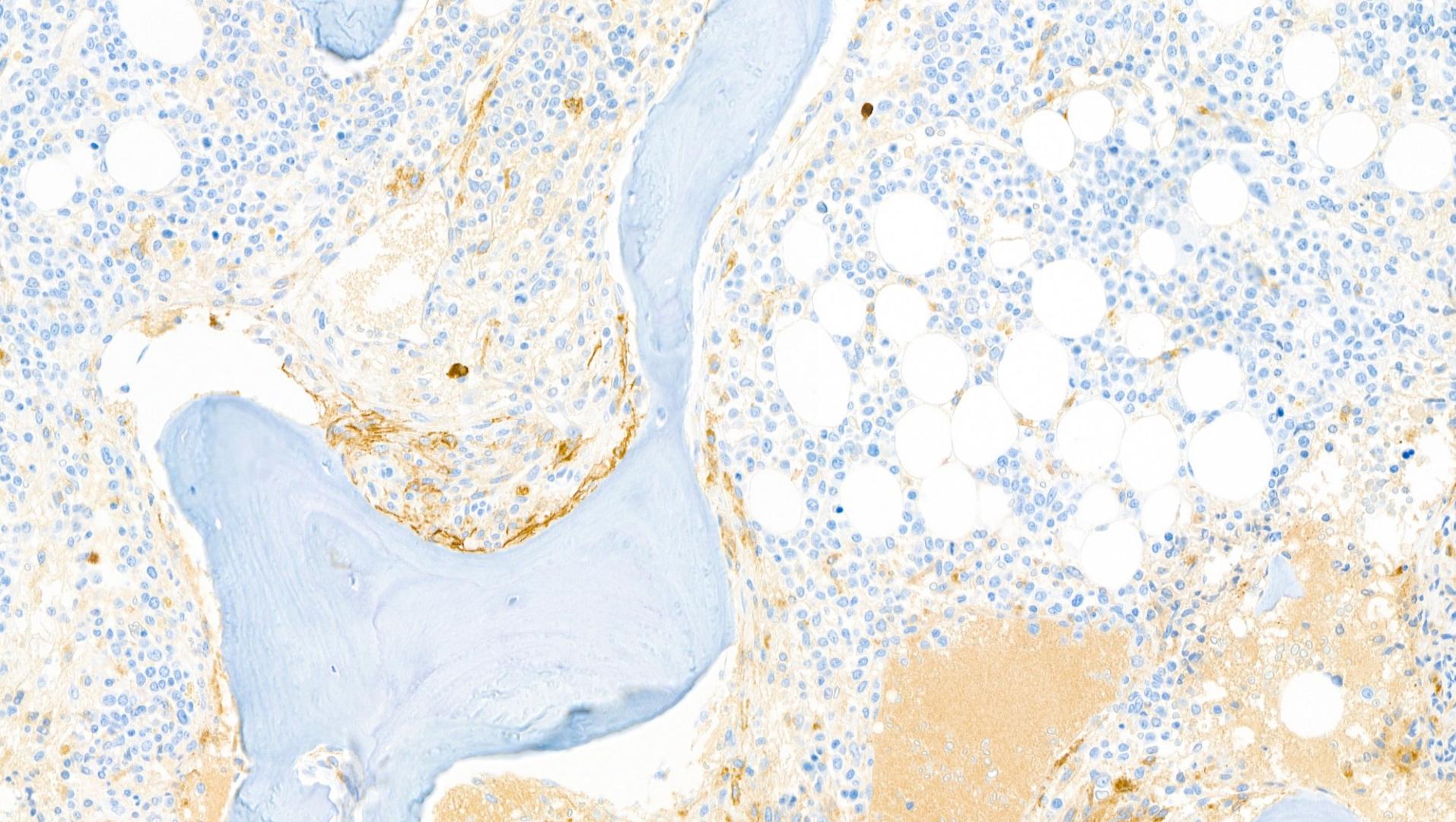












# **MASTOCITOSIS SISTÉMICA CON NEOPLASIA HEMATOLÓGICA / MIELOIDE ASOCIADA**



# Diferentes visiones

ICC 2022	Proposed WHO 5th Edition
<ul style="list-style-type: none"><li>• Cutaneous mastocytosis</li><li>• Urticaria pigmentosa/maculopapular cutaneous mastocytosis</li><li>• Diffuse cutaneous mastocytosis</li><li>• Mastocytoma of skin</li><li>• Systemic mastocytosis</li><li>• Indolent systemic mastocytosis (includes bone marrow mastocytosis)</li><li>• Smoldering systemic mastocytosis</li><li>• Aggressive systemic mastocytosis</li><li>• Mast cell leukemia</li><li>• Systemic mastocytosis with an associated myeloid neoplasm</li><li>• Mast cell sarcoma</li></ul>	<ul style="list-style-type: none"><li>• Cutaneous mastocytosis</li><li>• Urticaria pigmentosa/maculopapular cutaneous mastocytosis<ul style="list-style-type: none"><li>• Monomorphic</li><li>• Polymorphic</li></ul></li><li>• Diffuse cutaneous mastocytosis</li><li>• Cutaneous mastocytoma<ul style="list-style-type: none"><li>• Isolated mastocytoma</li><li>• Multilocalized mastocytoma</li></ul></li><li>• Systemic mastocytosis</li><li>• Bone marrow mastocytosis</li><li>• Indolent systemic mastocytosis</li><li>• Smoldering systemic mastocytosis</li><li>• Aggressive systemic mastocytosis</li><li>• Systemic mastocytosis with an associated hematologic neoplasm</li><li>• Mast cell leukemia</li><li>• Mast cell sarcoma</li></ul>



# The international consensus classification of mastocytosis and related entities

Review | Published: 10 October 2022

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**Table 4** Diagnostic criteria for systemic mastocytosis with an associated myeloid neoplasm (SM-AMN)

- 
- Meets the diagnostic criteria for systemic mastocytosis
  - Meets the criteria for an associated myeloid neoplasm, e.g., chronic myelomonocytic leukemia (CMML) or other myelodysplastic/myeloproliferative neoplasm (MDS/MPN), myelodysplastic syndrome (MDS), myeloproliferative neoplasm (MPN), acute myeloid leukemia (AML), or other myeloid neoplasm<sup>a</sup>
  - The associated myeloid neoplasm should be fully classified according to established criteria<sup>b</sup>
-

# The international consensus classification of mastocytosis and related entities

**Table 1** Systemic mastocytosis: diagnostic criteria

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Major criterion

- Multifocal dense infiltrates of tryptase- and/or CD117-positive mast cells ( $\geq 15$  mast cells in aggregates) detected in sections of bone marrow and/or other extracutaneous organ(s)<sup>a</sup>

In the absence of the major criterion, at least three of the following four minor criteria must be present

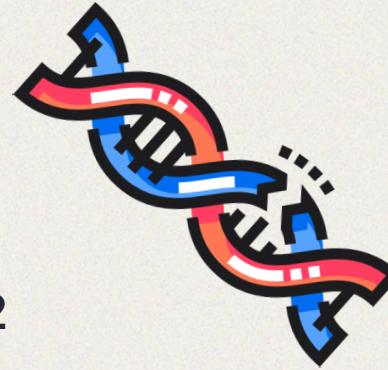
- In bone marrow biopsy or in section of other extracutaneous organs,  $> 25\%$  of mast cells are spindle shaped or have an atypical immature morphology<sup>b</sup>
- Mast cells in bone marrow, peripheral blood, or other extracutaneous organs express CD25, CD2, and/or CD30, in addition to mast cell markers
- *KIT* D816V mutation or other activating *KIT* mutation detected in bone marrow, peripheral blood, or other extracutaneous organs<sup>a/c</sup>
- Elevated serum tryptase level, persistently  $> 20$  ng/mL. In cases of SM-AMN, an elevated tryptase does not count as a SM minor criterion



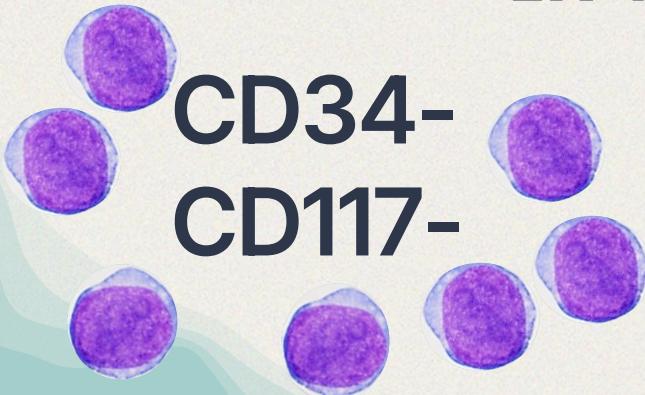
RUNX1

TET2

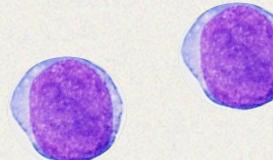
SF3B1 DNMT3A GATA2



## LMA CON MUTACIONES RELACIONADAS CON LA MIELODISPLASIA



CD34+  
CD117+



# ¿Diagnosticado?

**Table 4** Diagnostic criteria for systemic mastocytosis with an associated myeloid neoplasm (SM-AMN)

- ✓ • Meets the diagnostic criteria for systemic mastocytosis
- ✓ • Meets the criteria for an associated myeloid neoplasm, e.g., chronic myelomonocytic leukemia (CMML) or other myelodysplastic/myeloproliferative neoplasm (MDS/MPN), myelodysplastic syndrome (MDS), myeloproliferative neoplasm (MPN), acute myeloid leukemia (AML), or other myeloid neoplasm<sup>a</sup>
- ✓ • The associated myeloid neoplasm should be fully classified according to established criteria<sup>b</sup>



# **Systemic mastocytosis with an associated myeloid neoplasm**

**Report from the XVI European Bone Marrow Working Group Course and Workshop, Barcelona 2023**

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# Diagnóstico diferencial

LEUCEMIA MIELOMASTOCÍTICA	LEUCEMIA BASOFÍLICA
No cumple criterios de SM	CD117-
No hay mutación KIT D816V	CD123+



## KEY POINTS

- The presence of an AMN in a patient with SM may be difficult to identify. Flow cytometry results and the presence of non-*KIT* mutations may support a diagnosis of SM-AMN.
- The SM may be obscured by an AMN, especially in AML (“occult” SM). Immunostaining for CD117 and tryptase is recommended in all patients with a myeloid neoplasm associated with a *KIT* mutation.
- It may be impossible to distinguish ASM from ISM within a case of SM-AMN, unless the observed “C” findings can be attributed to the SM.



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