

# **CASO 3**

## **Patología de la Médula ósea**

Autores: Irene Salazar Saura, Josep Marí Alexandre, Sara Alemán Sánchez, José Joaquín Llopis Linares, Lara Navarro Cerveró, Esther Roselló Sastre

**Consorcio Hospital General Universitario de Valencia**



- Paciente varón de 16 años.
- Cuadro poliadenopático a estudio, con astenia y palidez cutánea desde hace un mes.

- Masa en hipogastrio izquierdo, conglomerado adenopático inguinal.
- Adenopatías cervicales.
- Varios nódulos cutáneos.



- Anemia normocítica con trombopenia y elevación de reticulocitos.
- Elevación de Dímero D.
- Elevación de LDH.



# Estudios de imagen

## Ecografía

- Varias **adenopatías de tamaño patológico** en región cervical, inguinal y pélvica.
- Ecografía de **testículos: múltiples LOEs sólidas** bilaterales.

## TC-TAP

- **Nódulo subpleural** en LSD.
- Masa heterogénea **paravesical** izquierda.
- Múltiples **adenomegalias** inguinales
- **Lesión ósea** sacra lítica.



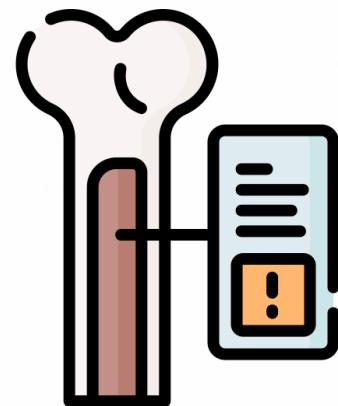
**Sustitución de la médula ósea grasa** por otro tejido que afecta a todas las estructuras óseas visualizadas.

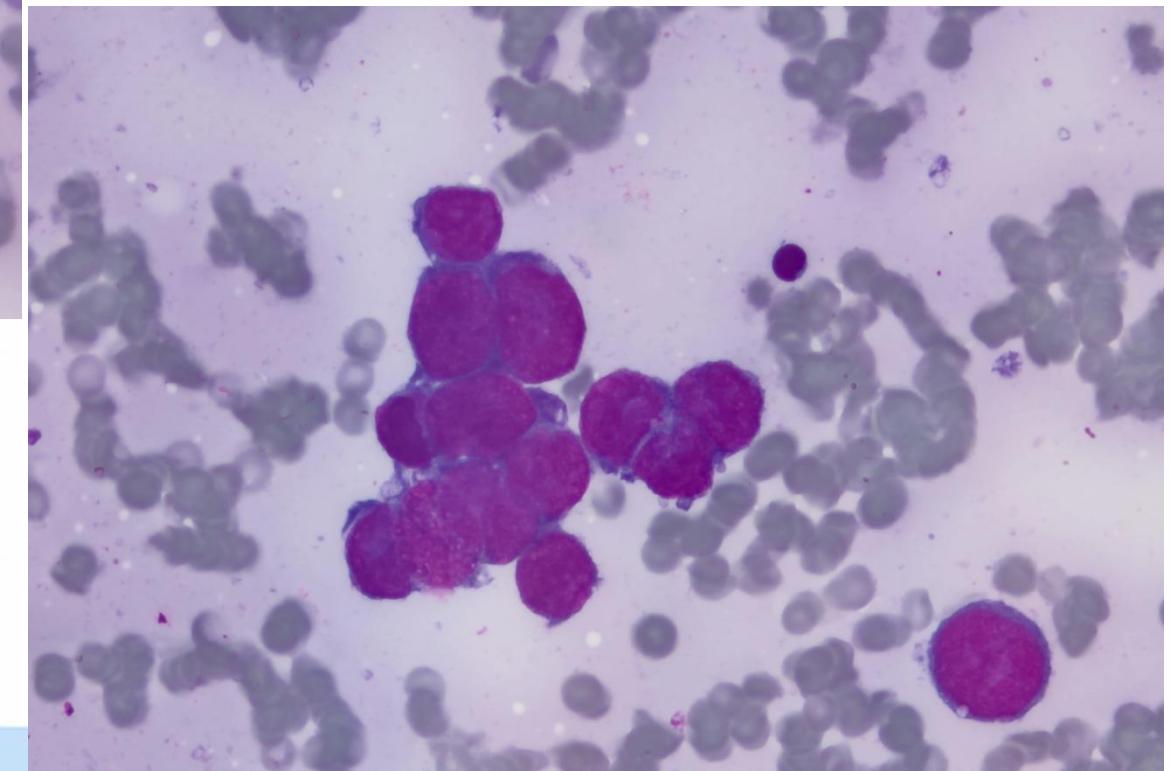
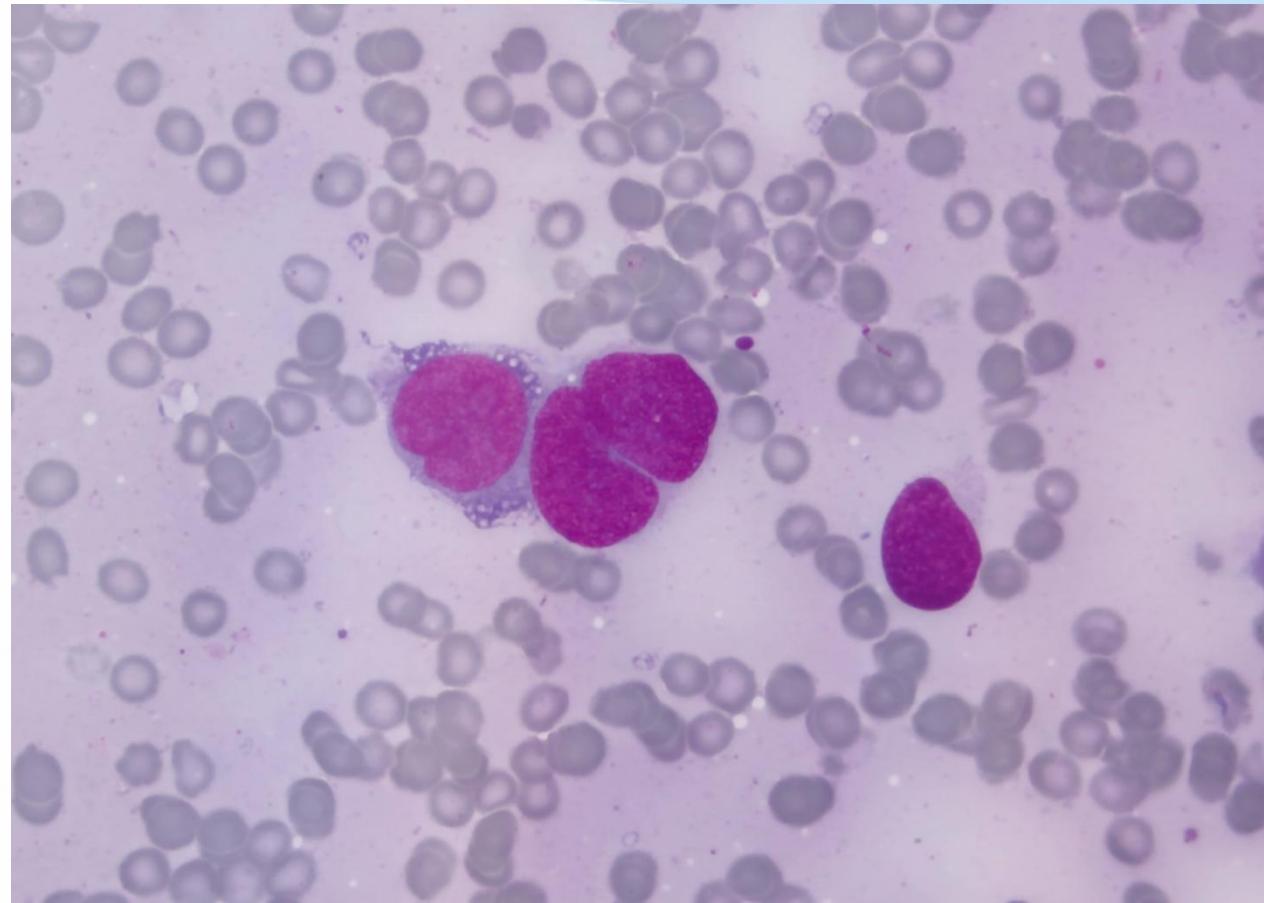
## RMN



## Aspirado de médula ósea

Hematíes, ausencia de megacariocitos y moderada celularidad nucleada, destacando la presencia de **40% de células de aspecto blástico**. En inmunofenotipo, los blastos son 0.5% positivos a CD45 y débil.





A.M.O.

## Aspirado de médula ósea

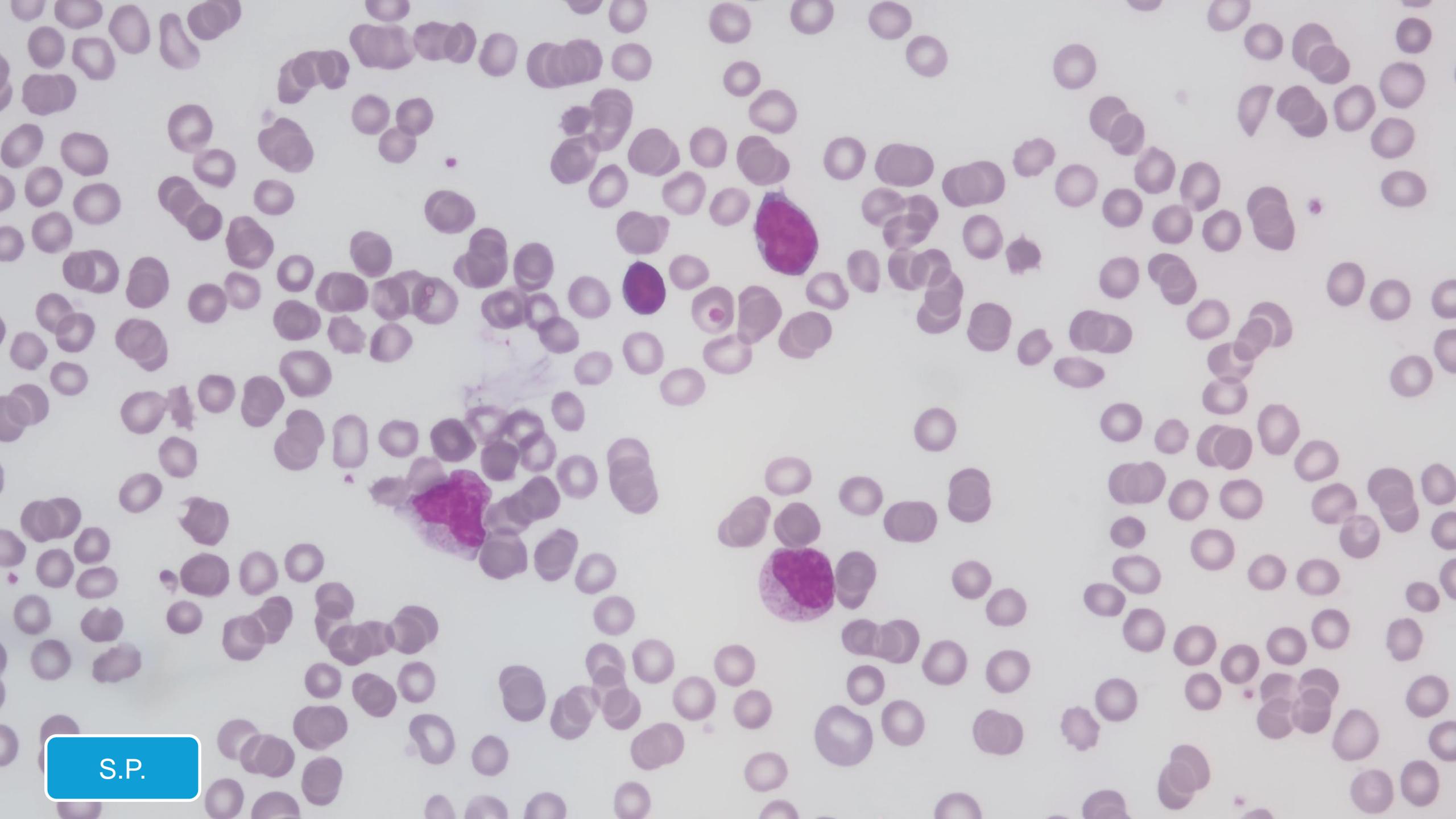
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## Sangre periférica

- Bicitopenia
- Síndrome leucoeritroblástico

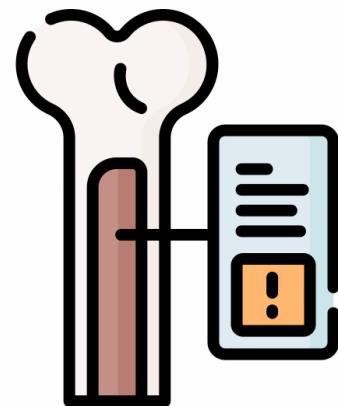




S.P.

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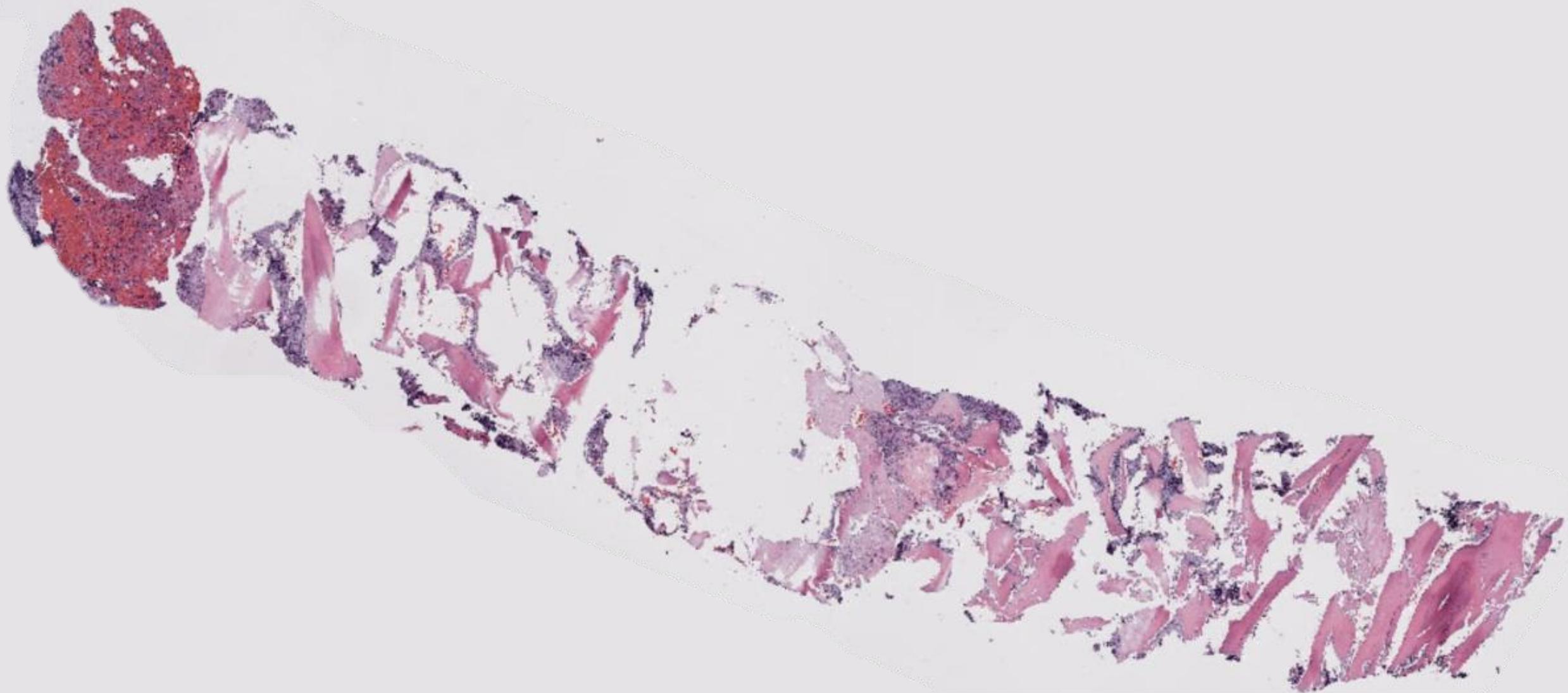


## Sangre periférica

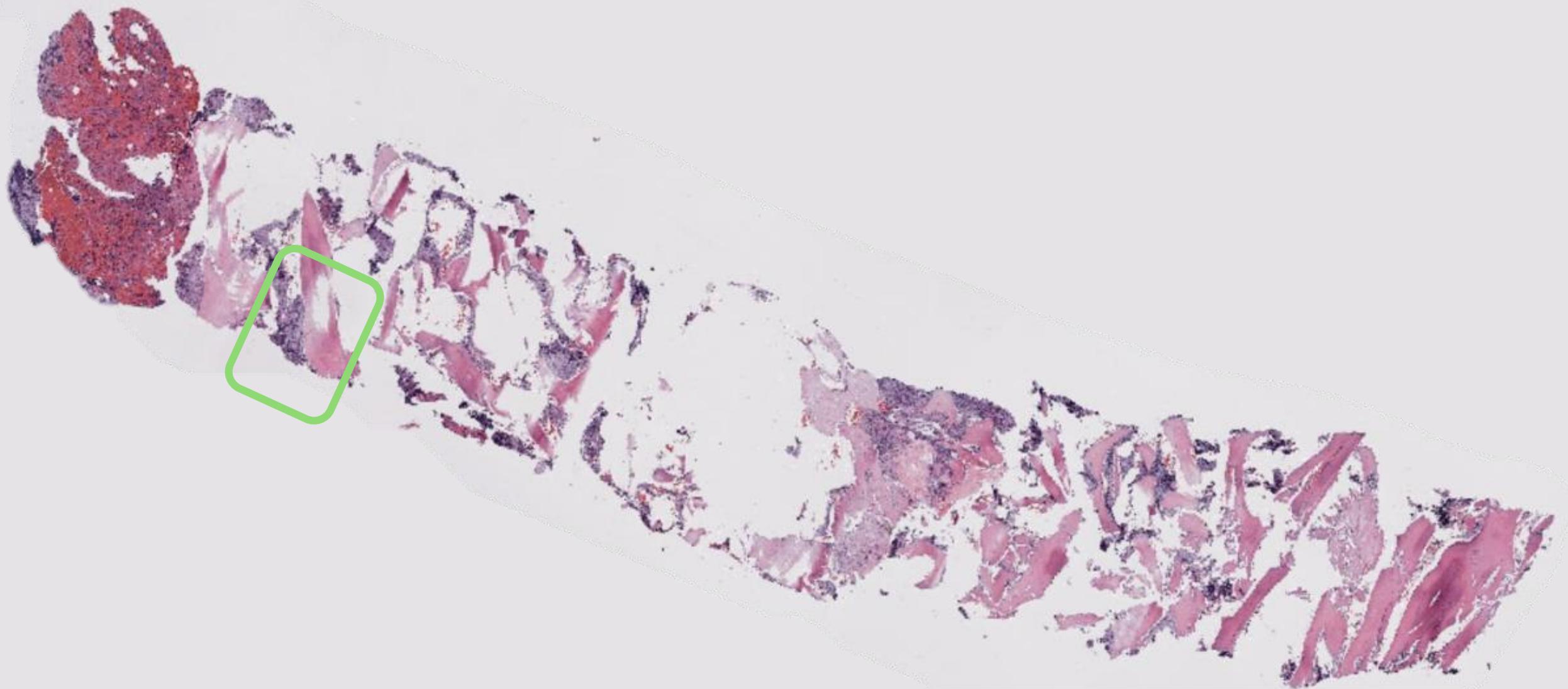
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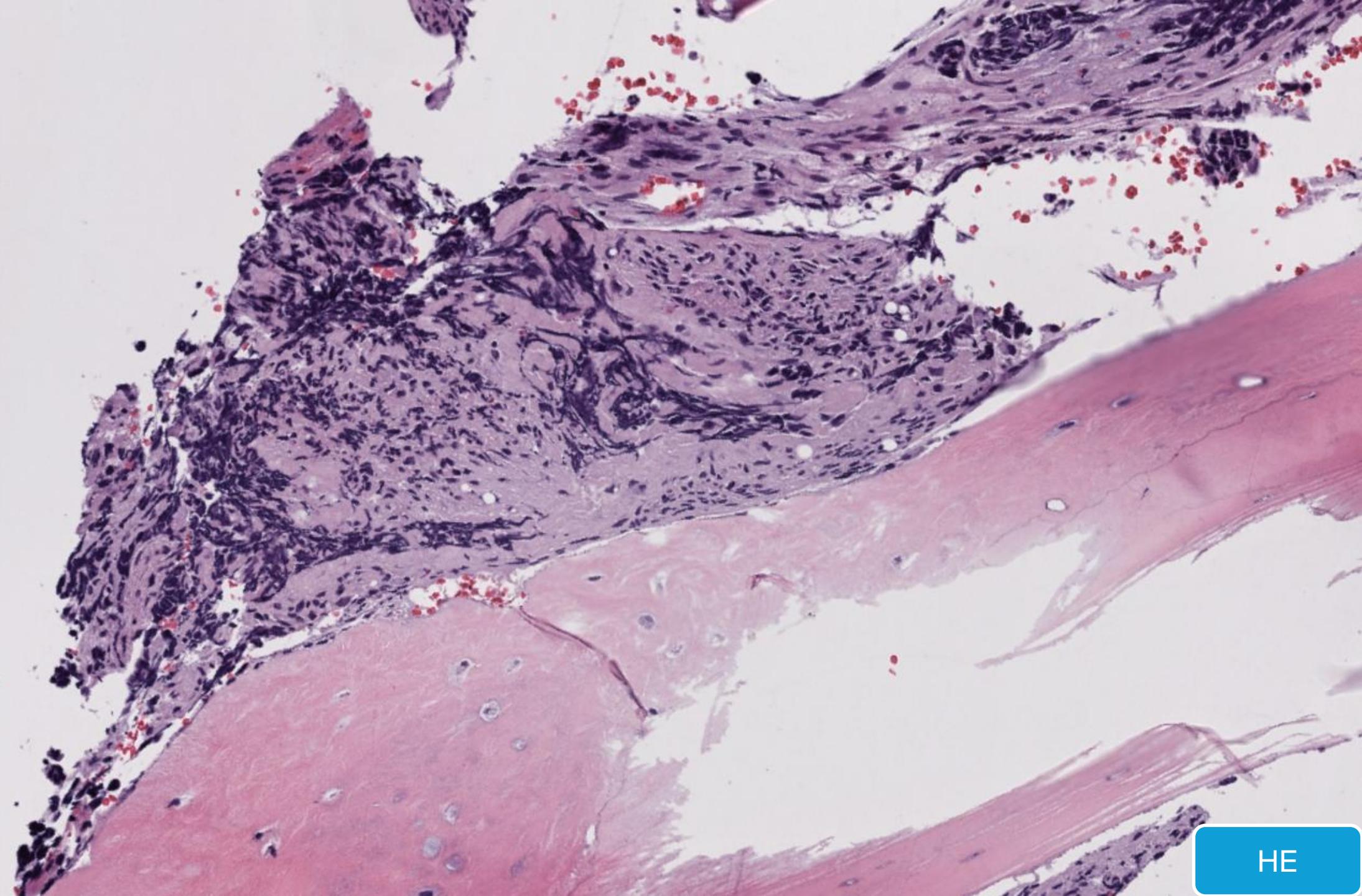
Hallazgos que sugieren **infiltración de la médula ósea por un proceso neoplásico**. Nos remiten **biopsia de médula ósea** para valoración urgente.



HE

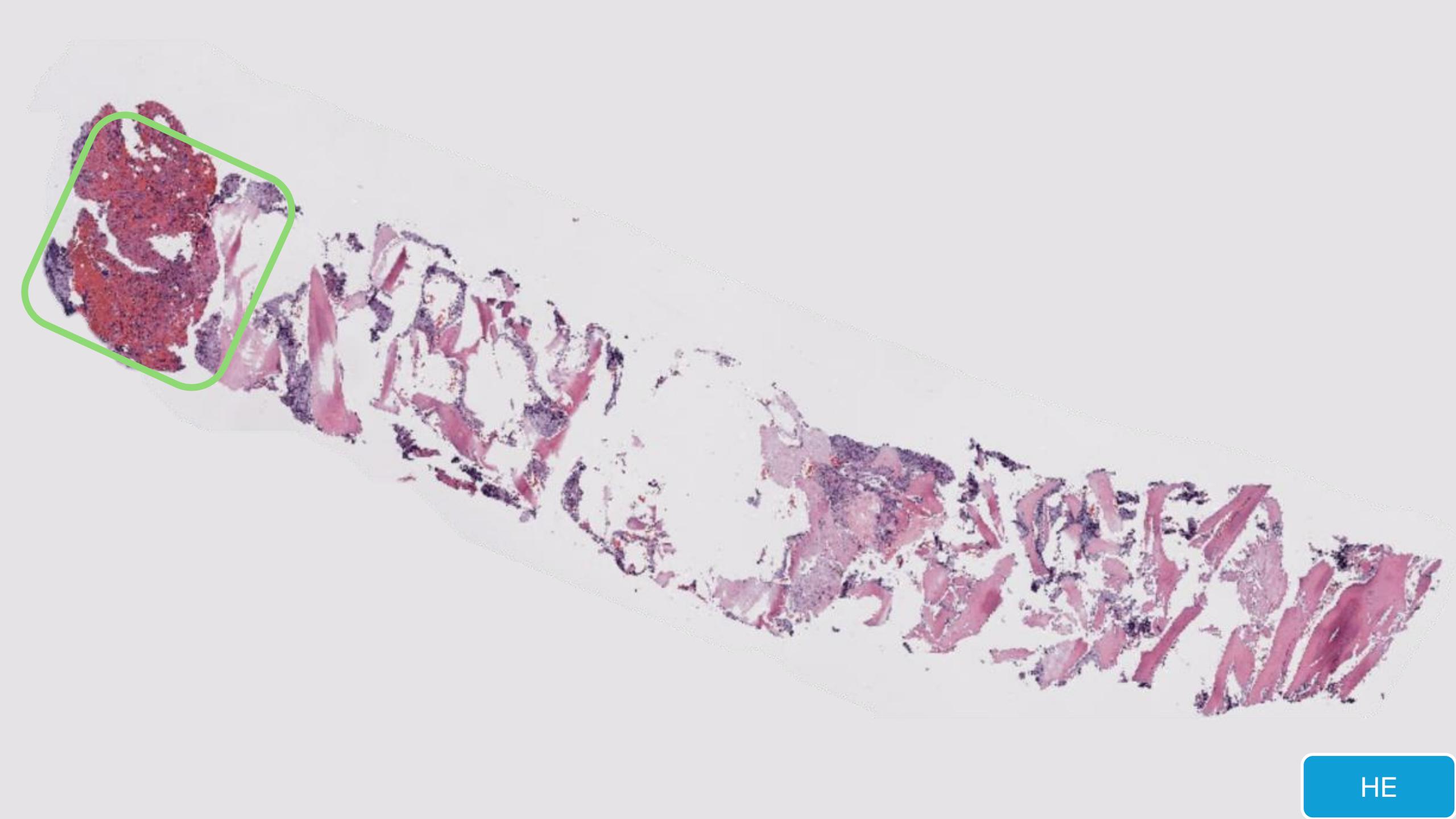


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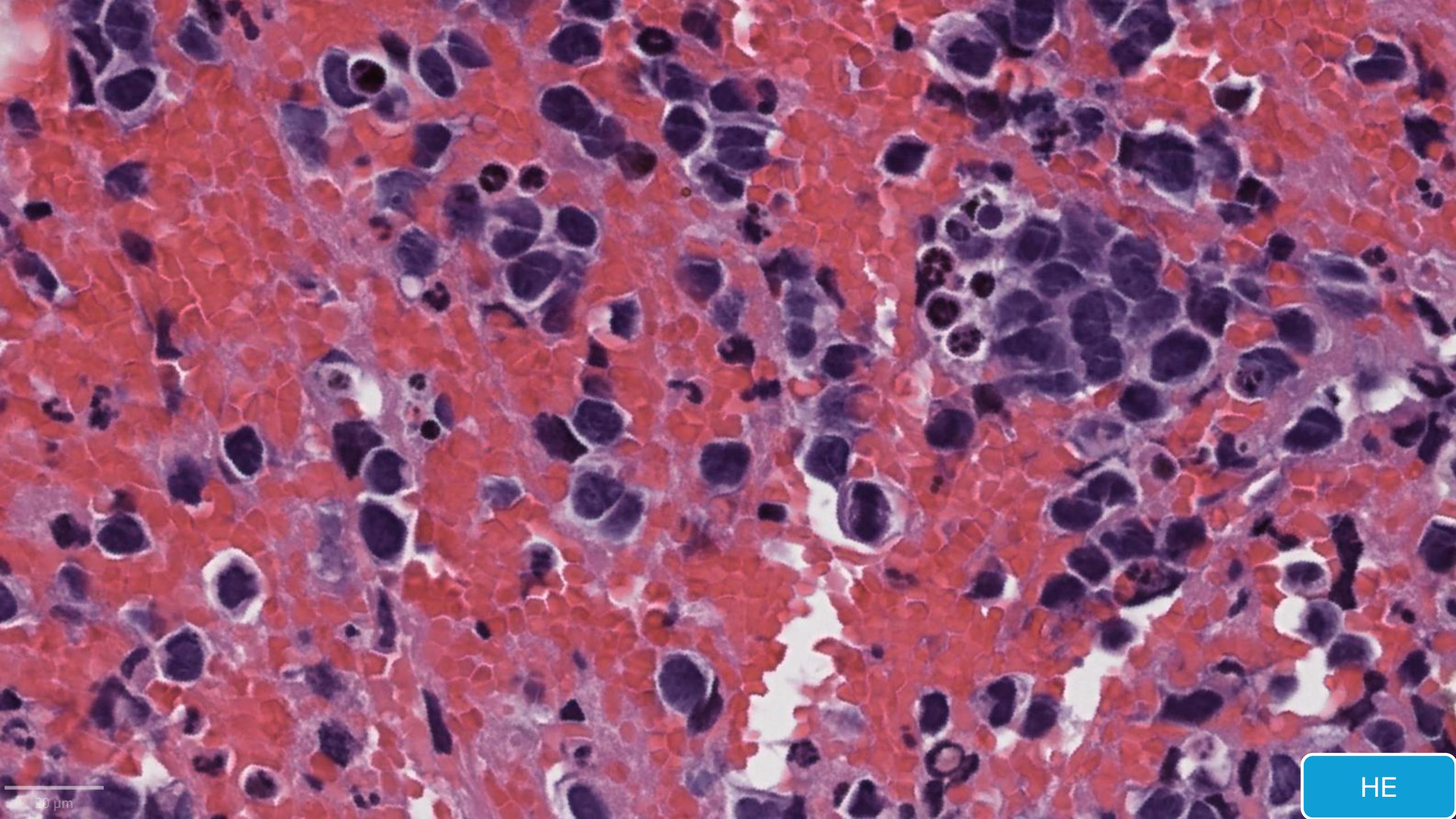


100  $\mu$ m

HE

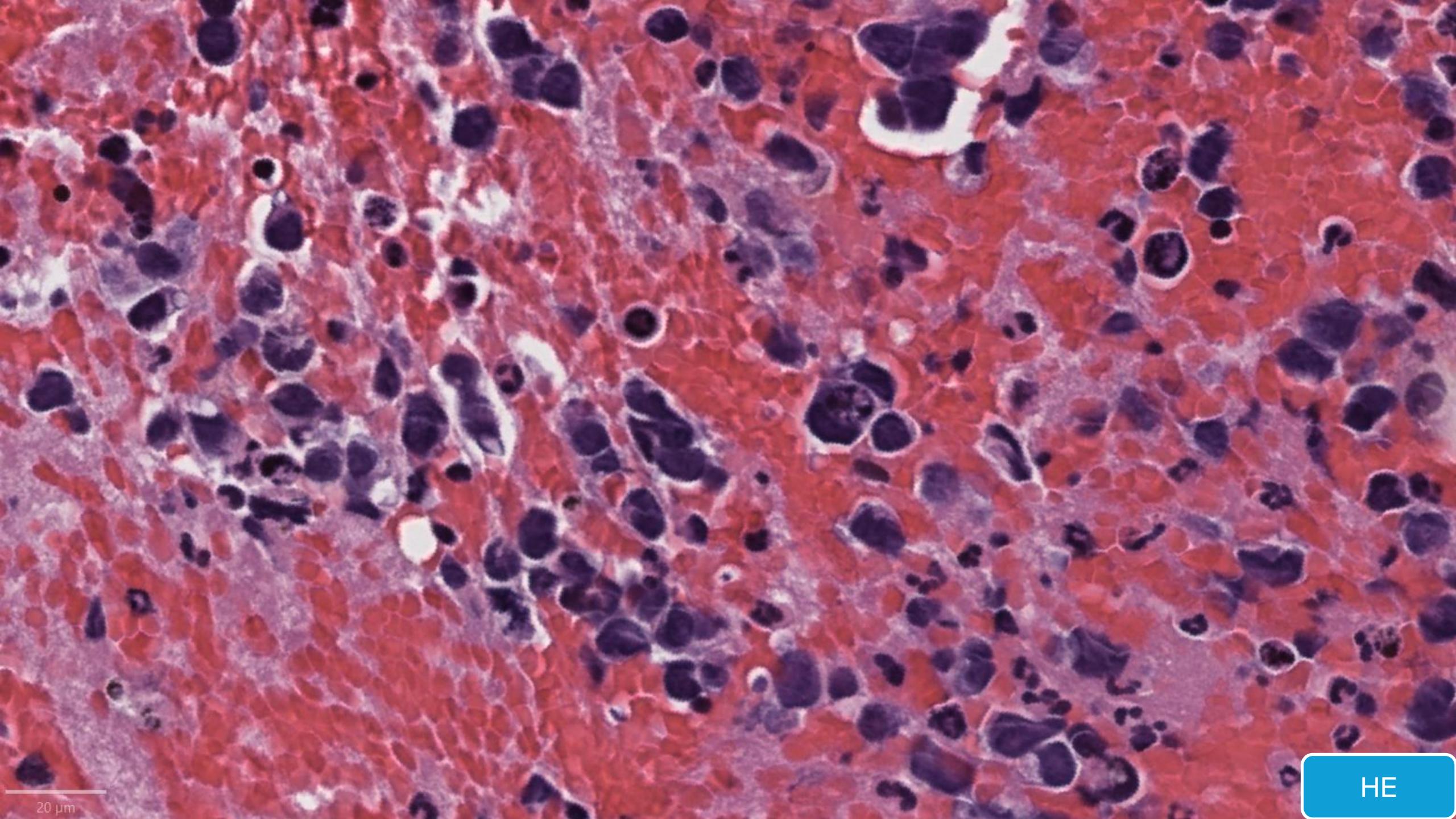


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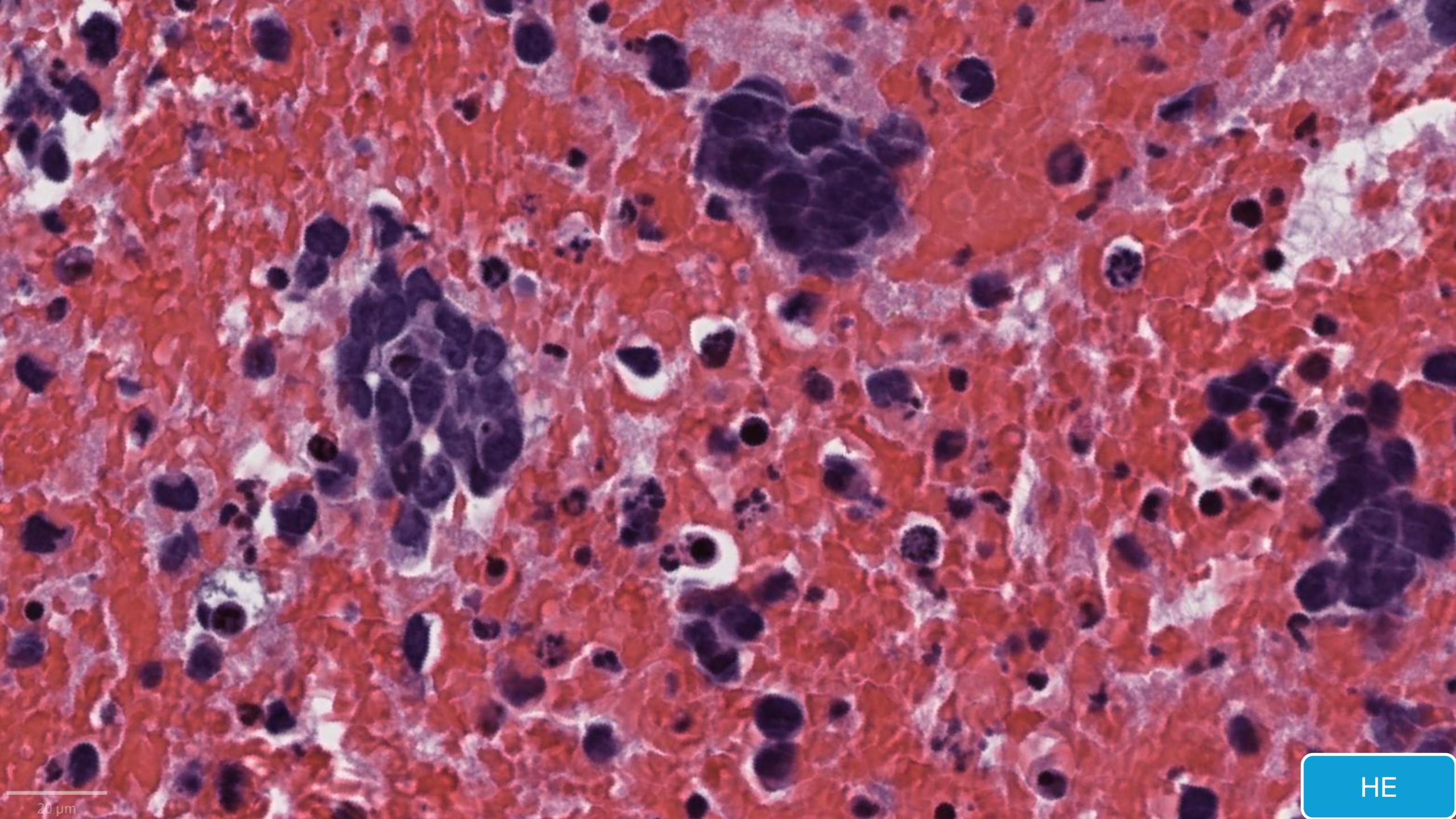
20  $\mu$ m

HE



20  $\mu$ m

HE



20  $\mu$ m

HE

# Diagnóstico diferencial

## Tumores de célula redonda pequeña

- Origen epitelial / carcinomas.
- Linfomas.
- Sarcoma de Ewing.
- Sarcomas con reordenamiento BCOR.
- Sarcomas con reordenamiento en CIC.
- Condrosarcoma mesenquimal.
- Osteosarcoma de células pequeñas.
- Rabdomiosarcoma.

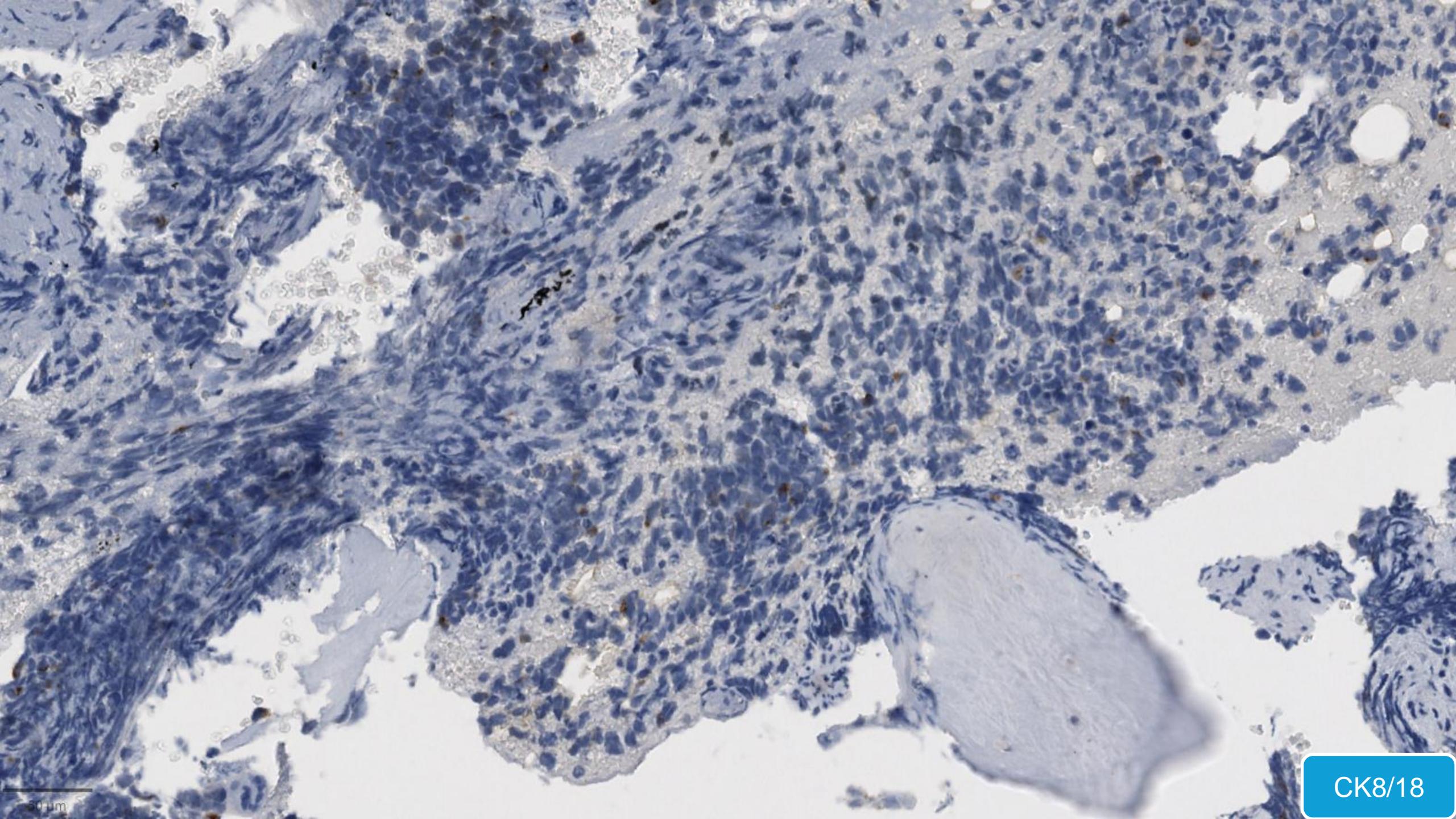


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CK8/18

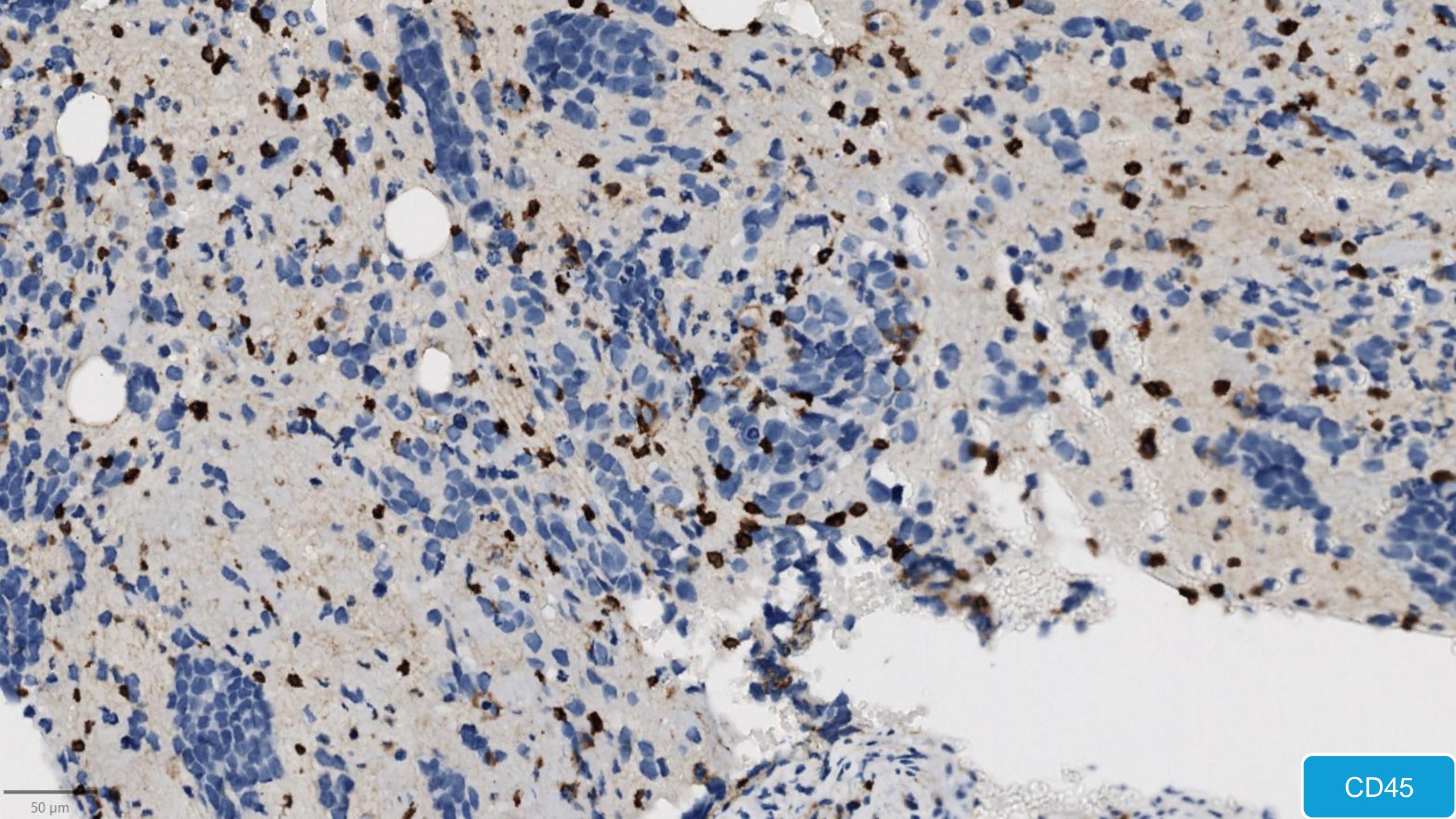
50  $\mu$ m

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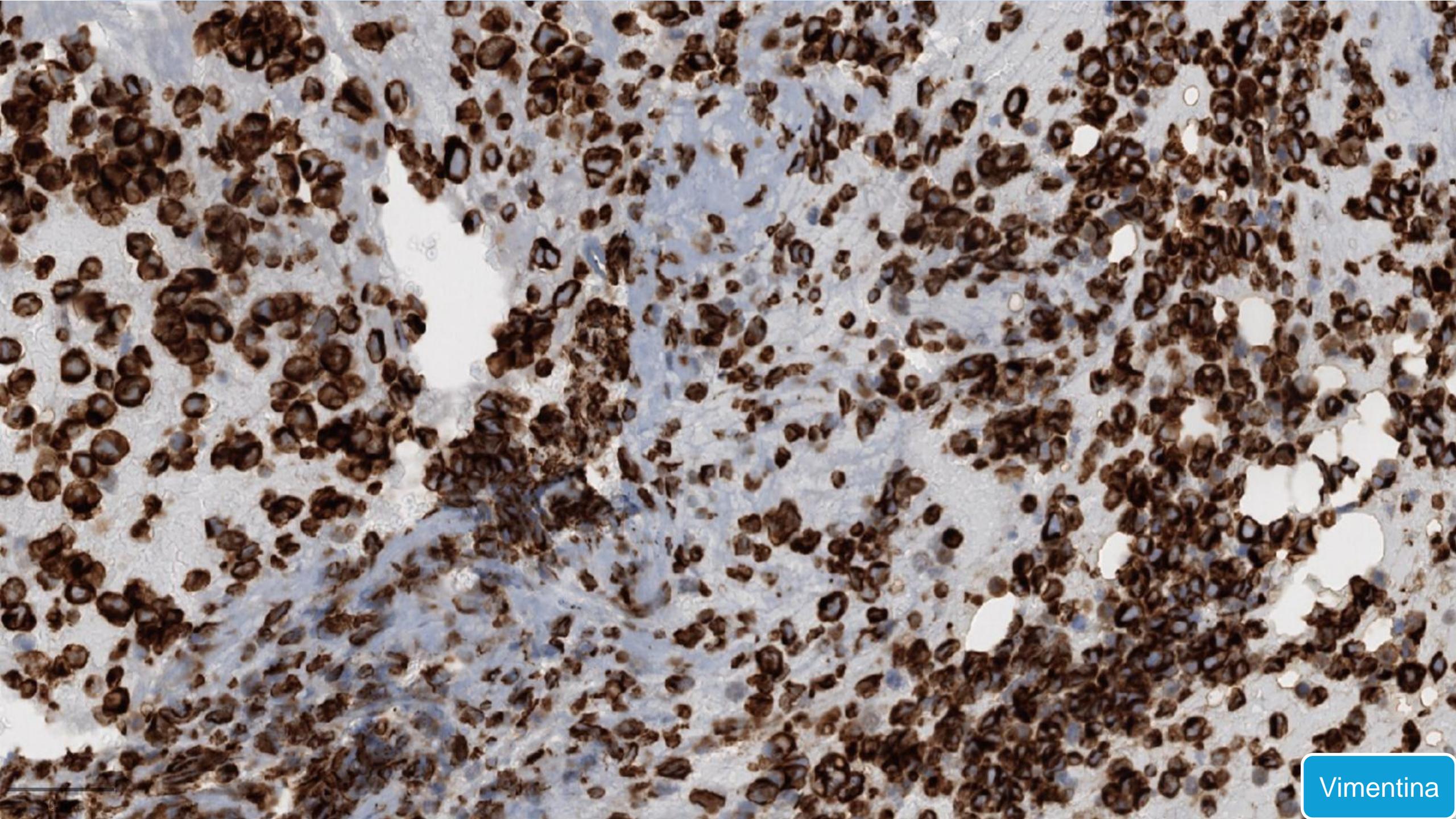
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50  $\mu$ m

CD45



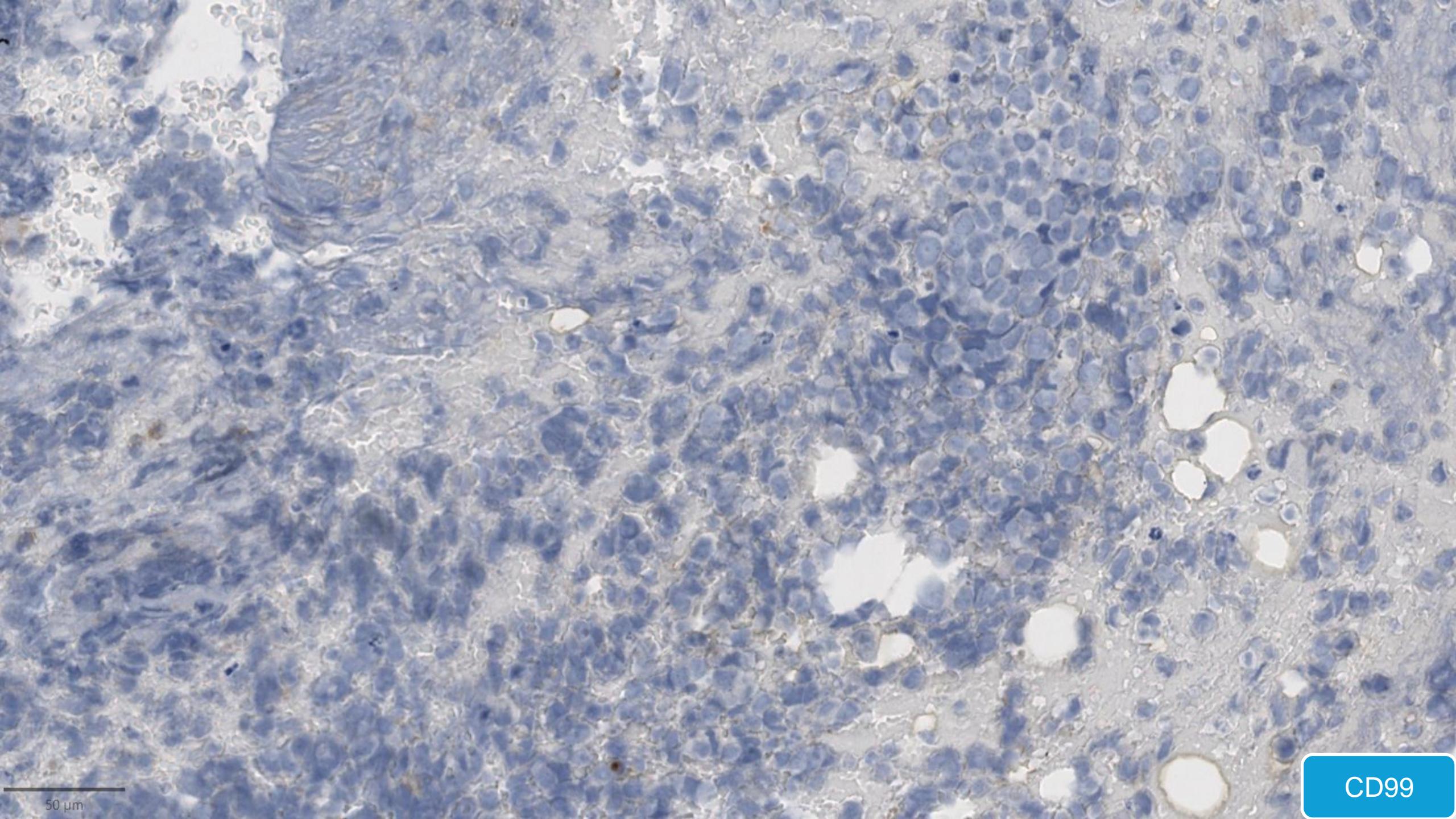
Vimentina

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50  $\mu$ m

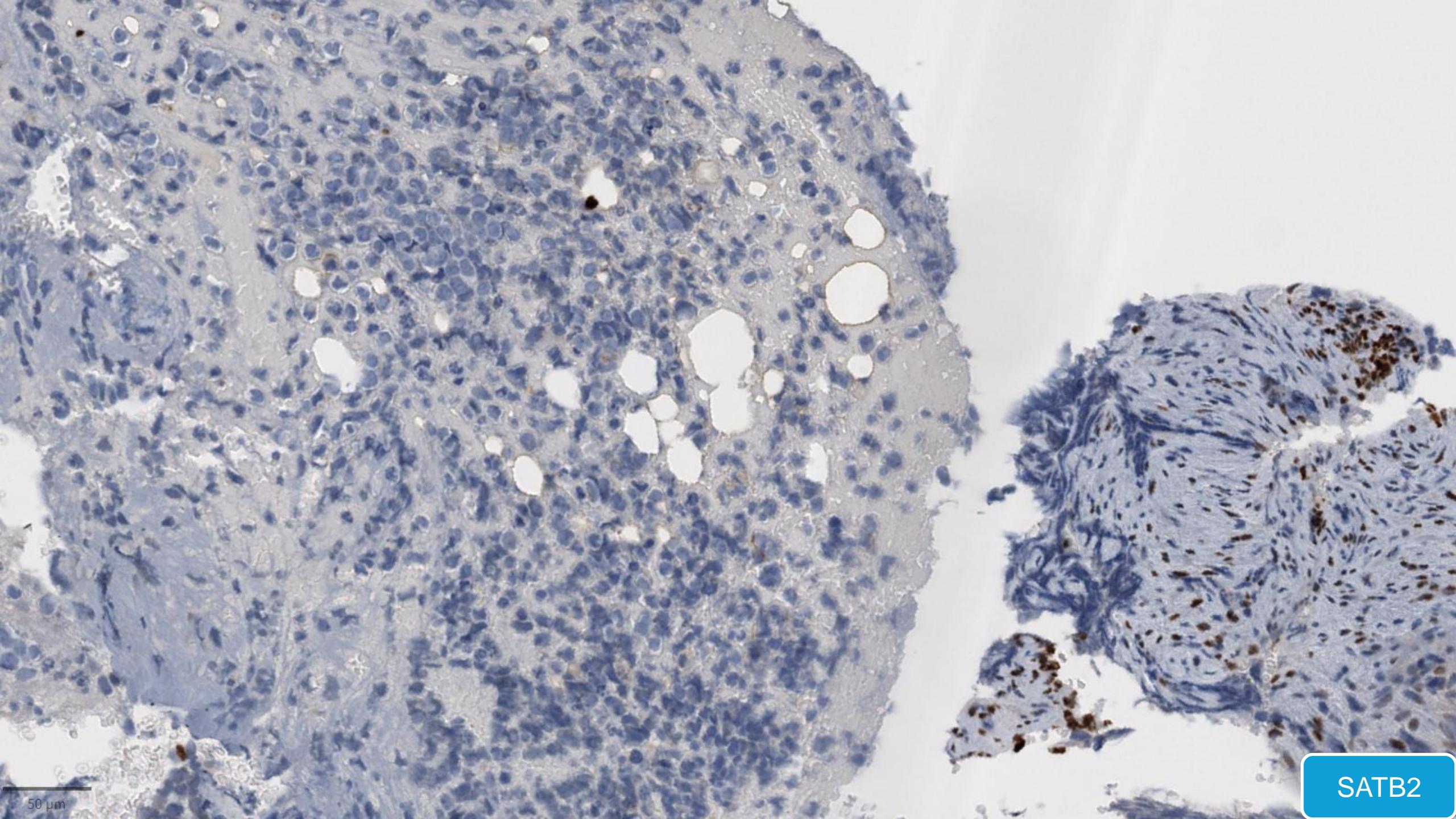
CD99

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SATB2

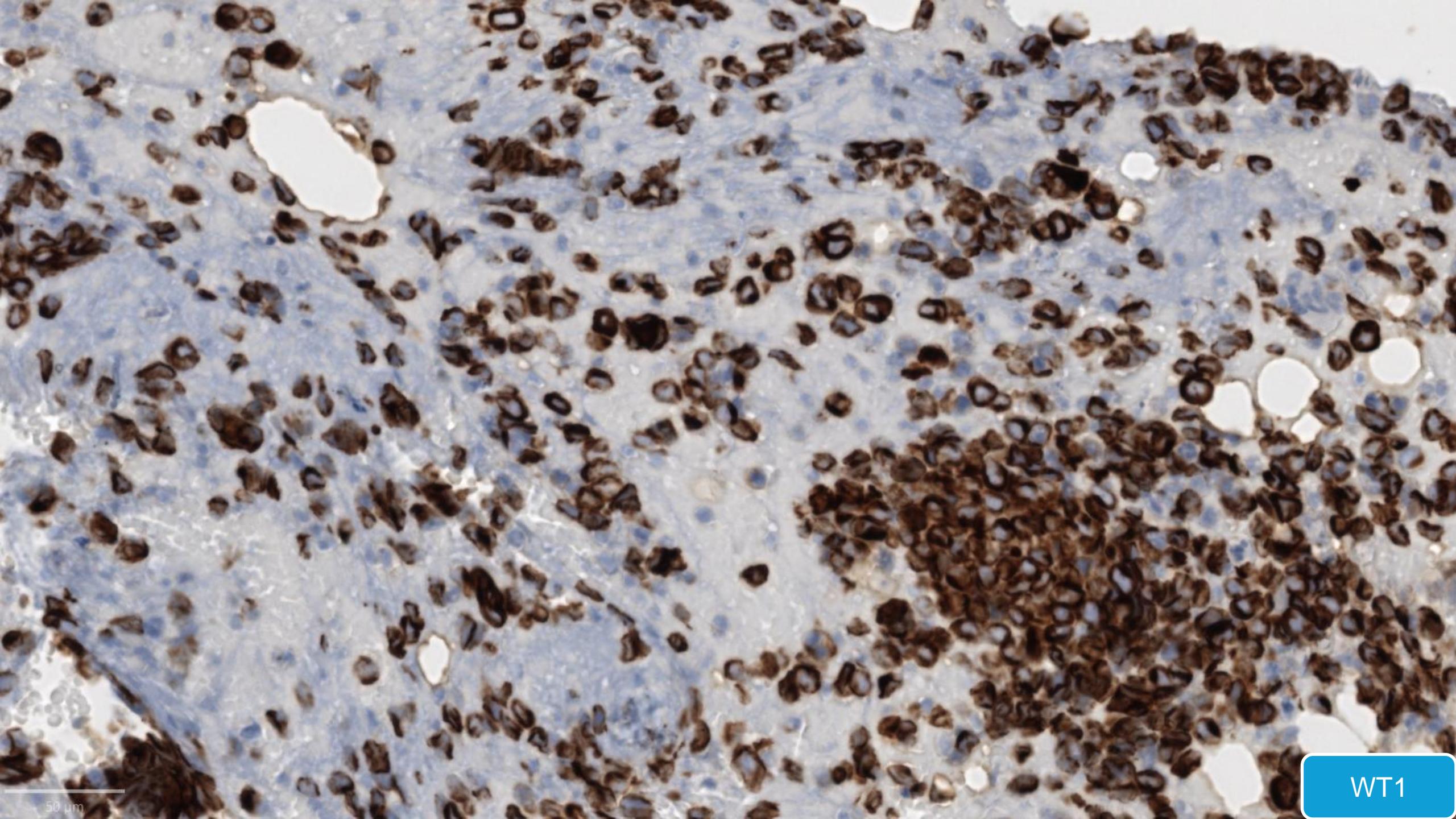
50 µm

# Diagnóstico diferencial

## Tumores de célula redonda pequeña

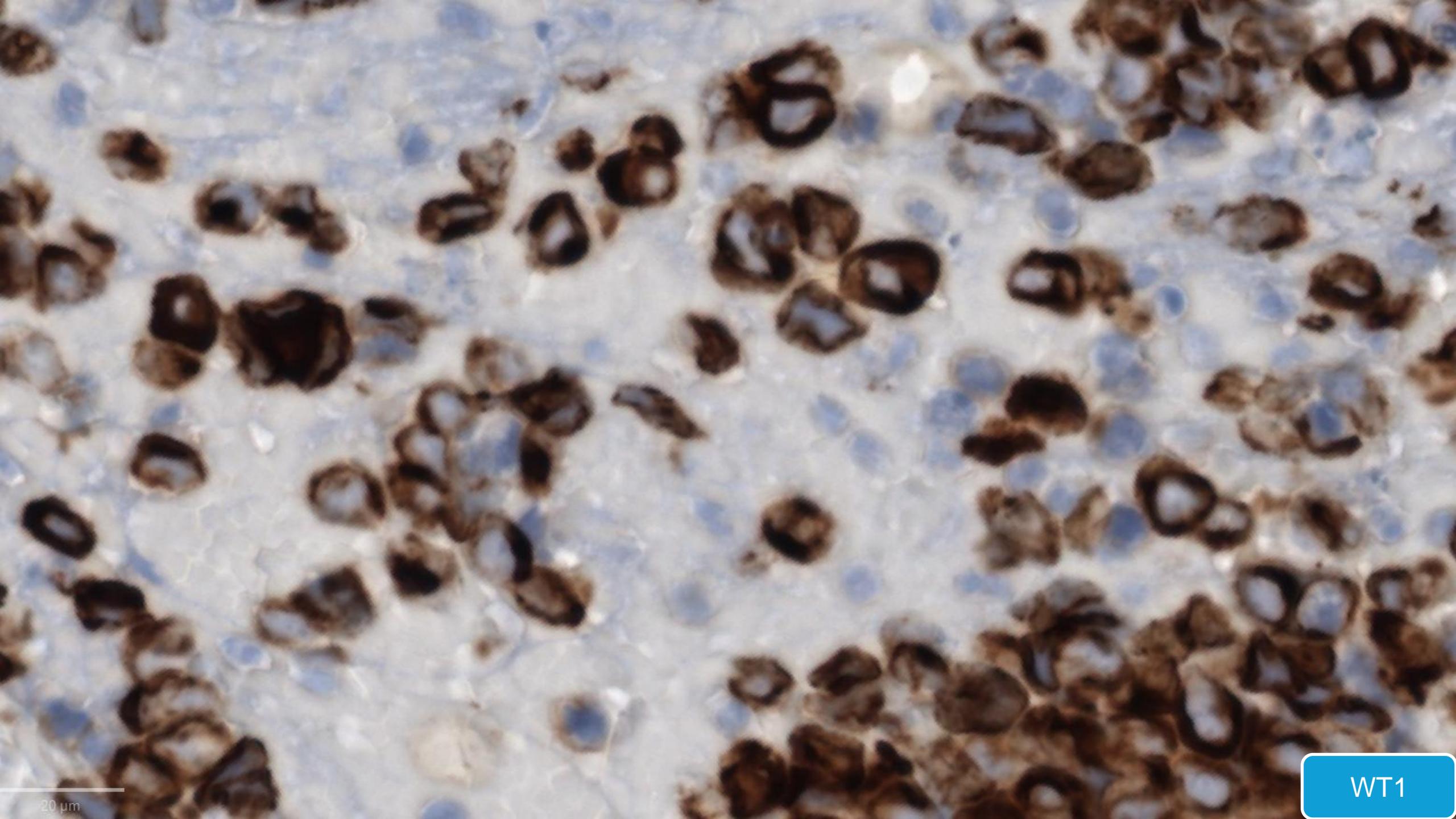
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WT1

50  $\mu$ m



20  $\mu$ m

WT1

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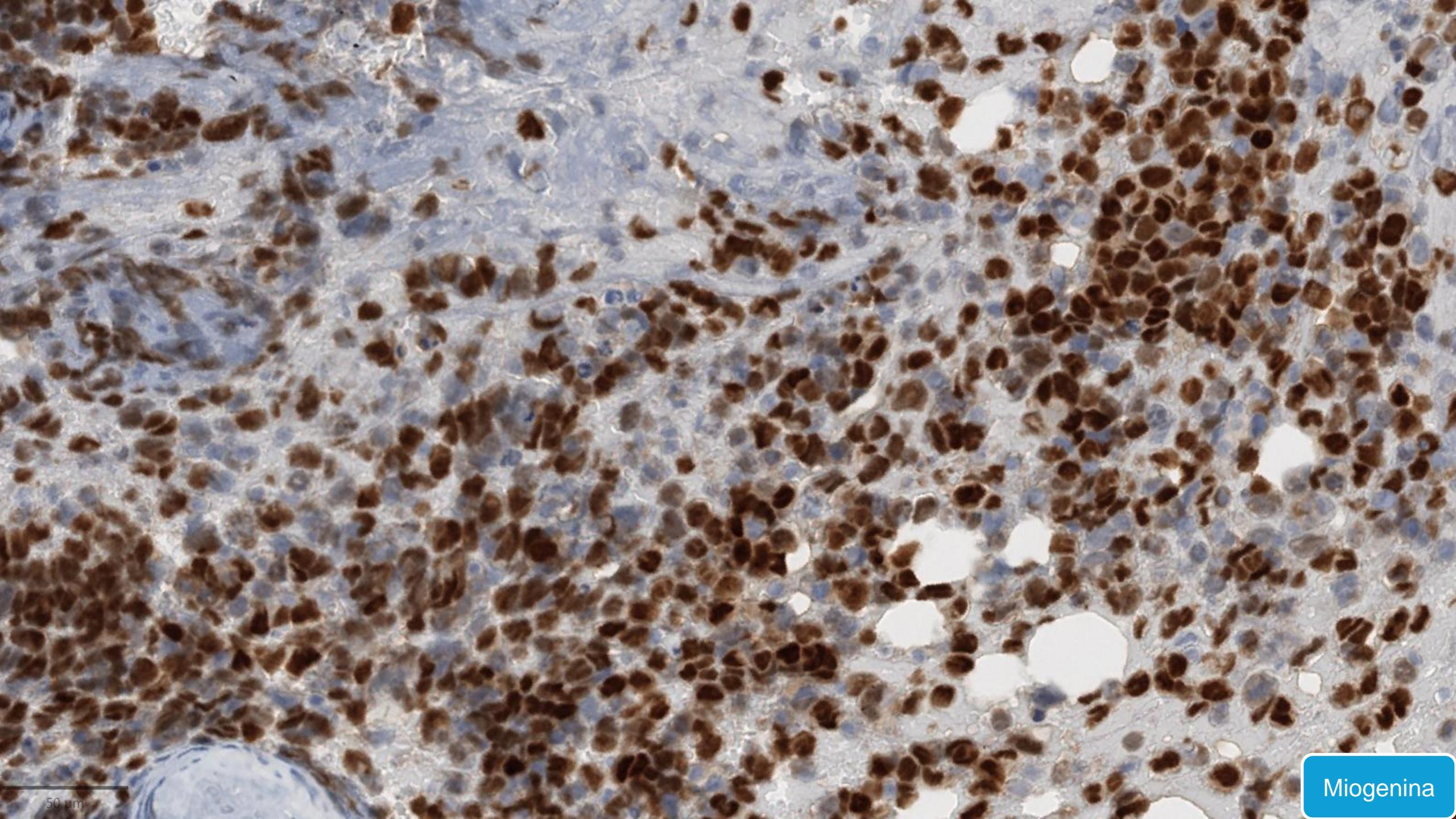


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50  $\mu$ m

Miogenina



# Oncofetal expression of Wilms' tumor 1 (WT1) protein in human fetal, adult and neoplastic skeletal muscle tissues

Gaetano Magro <sup>a,\*</sup>, Lucia Salvatorelli <sup>a</sup>, Lidia Puzzo <sup>a</sup>, Giuseppe Musumeci <sup>b</sup>,  
Michele Bisceglia <sup>c</sup>, Rosalba Parenti <sup>d</sup>

<sup>a</sup> Department of Medical and Surgical Sciences and Advanced Technologies, G.F. Ingrassia, AziendaOspedaliero-Universitaria "Policlinico-Vittorio Emanuele", Anatomic Pathology Section, School of Medicine, University of Catania, Catania, Italy

<sup>b</sup> Department of Biomedical and Biotechnological Sciences, Human Anatomy and Histology Section, School of Medicine, University of Catania, Catania, Italy

<sup>c</sup> School of Biomedical Sciences, Etromapmacs Pole, Lesina, FG, Italy

<sup>d</sup> Department of Biomedical and Biotechnological Sciences, Section of Physiology, School of Medicine, University of Catania, Catania, Italy

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## ARTICLE INFO

### Article history:

Received 28 December 2014

Received in revised form 10 February 2015

Accepted 21 February 2015

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### Keywords:

WT1

Human embryo and fetus

Myogenesis

Rhabdomyosarcoma

Immunohistochemistry

## ABSTRACT

There is increasing evidence that WT1 protein expression is found not only at nuclear, but also at cytoplasmic, level in several developing and neoplastic tissues. In order to better understand the possible role of WT1 protein in human skeletal myogenesis and oncogenesis of rhabdomyosarcoma, we assessed immunohistochemically its comparative expression in a large series of human developing, adult and neoplastic skeletal muscle tissues. The present study shows that WT1 protein is developmentally expressed in the cytoplasm of human myoblasts from the 6 weeks of gestational age. This expression was maintained in the myotubes of developing muscles of the trunk, head, neck, and extremities, while it was down-regulated in fetal skeletal fibers from 20 weeks of gestational age as well as in adult normal skeletal muscle. Notably, WT1 immunostaining disappeared from rhabdomyomas, whereas it was strongly and diffusely re-expressed in all cases (27/27) of embryonal and alveolar rhabdomyosarcoma. The comparative evaluation of the immunohistochemical findings revealed that WT1 cytoplasmic expression in rhabdomyosarcoma may represent an ontogenetic reversal, and this nuclear transcription factor can also be considered an oncofetal protein which can be exploitable as an additional, highly sensitive immunomarker, together with desmin, myogenin and MyoD1, of this tumor. Moreover, our observations support the rationale for the use of WT1 protein-based target therapy in high risk rhabdomyosarcomas in children and adolescents.

# Rabdomiosarcoma

Comprende principalmente cuatro entidades:

- Rabdomiosarcoma embrionario (ERMS)
- Rabdomiosarcoma alveolar (ARMS)
- Rabdomiosarcoma de células fusiformes/esclerosante (SCSRMS)
- Rabdomiosarcoma pleomórfico.



- Sarcoma de tejidos blandos más **común en niños**.
- El subtipo más frecuente: ERMS.
- ARMS es el segundo en frecuencia, pero más frecuentemente diagnosticado en **adolescentes**.

**Presentación clínica variada:**  
infiltración difusa de la MO.  
6-23% de los casos

## Rhabdomyosarcoma With Diffuse Bone Marrow Metastases

Daniel Huang <sup>1</sup>, Pankaj Watal <sup>2</sup>, Dennis Drehner <sup>3</sup>, Deeksha Dhar <sup>4</sup>, Tushar Chandra <sup>5</sup>

Affiliations + expand

PMID: 35265406 PMCID: PMC8897967 DOI: 10.7759/cureus.21863

## Rhabdomyosarcoma debut masquerading as acute lymphoblastic leukemia: A case report and review of the literature

Bernardo López-Andrade <sup>1</sup>, María Antonia Duran <sup>1</sup>, Lourdes Torres <sup>2</sup>, Marta García-Recio <sup>1</sup>, Laura Lo Riso <sup>1</sup>, Alejandro Formica <sup>1</sup>, Rafael F Ramos <sup>3</sup>, Niurka Cerdá <sup>4</sup>, Antonia Sampol <sup>1</sup>

Affiliations + expand

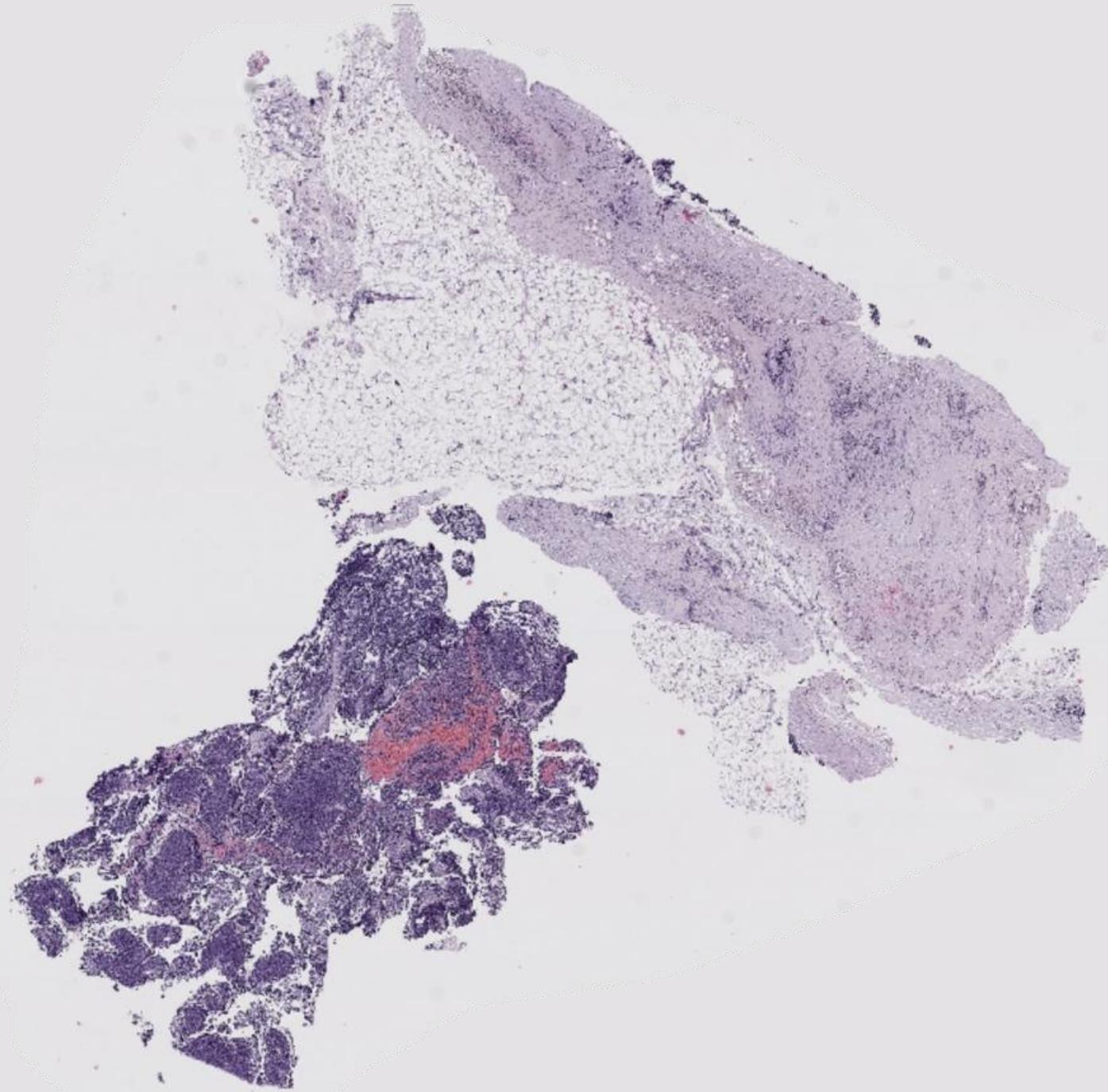
PMID: 31428386 PMCID: PMC6693075 DOI: 10.1002/ccr3.2284

## Complete mimicry: a case of alveolar rhabdomyosarcoma masquerading as acute leukemia

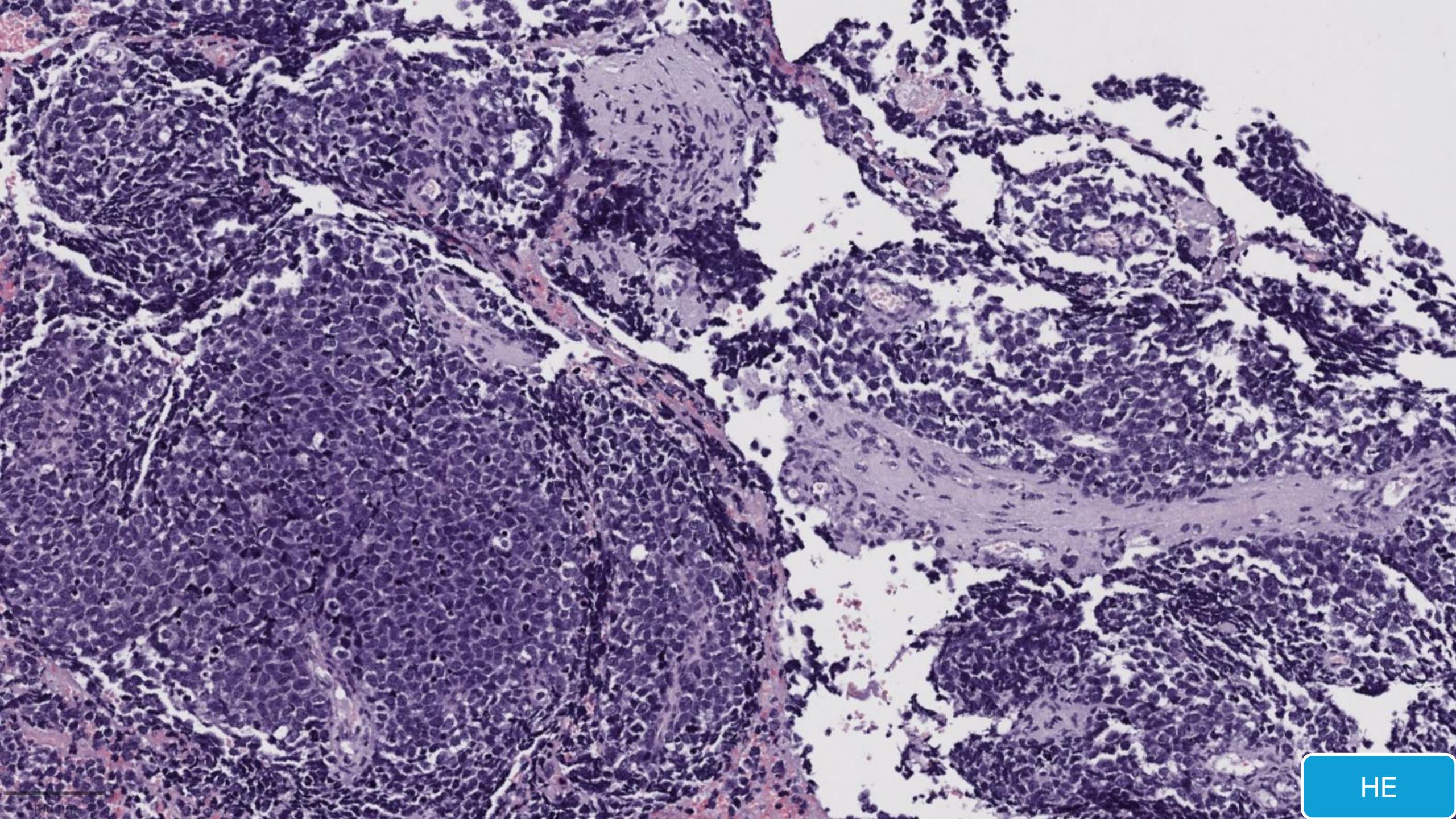
Osamu Imataki <sup>1,2</sup>, Makiko Uemura <sup>3</sup>, Shumpei Uchida <sup>3</sup>, Shigeyuki Yokokura <sup>3</sup>, Akihiro Takeuchi <sup>4</sup>, Ryo Ishikawa <sup>5</sup>, Akihiro Kondo <sup>4</sup>, Kayoko Seo <sup>4</sup>, Norimitsu Kadowaki <sup>3</sup>

Affiliations + expand

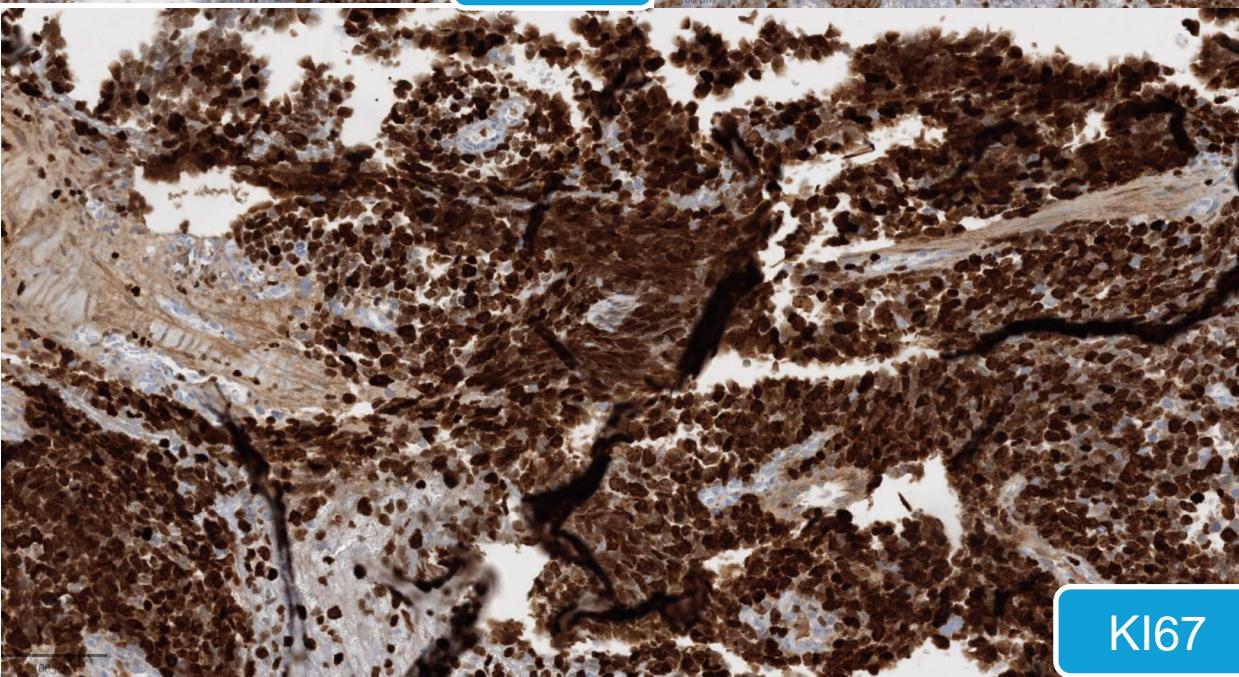
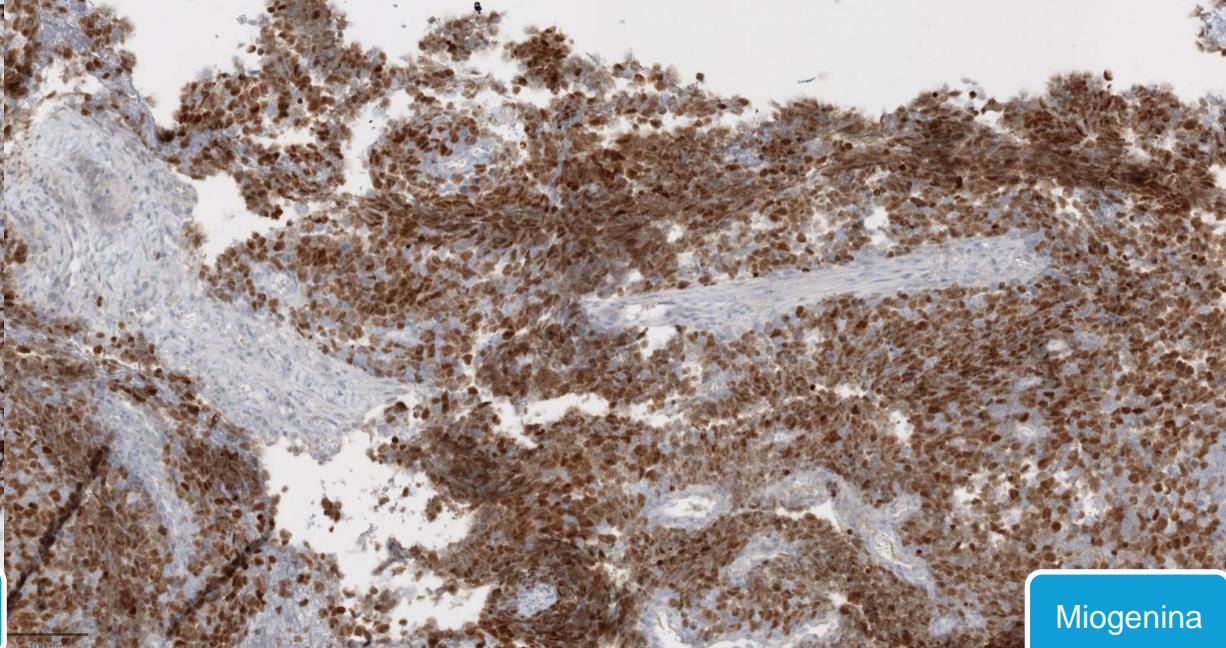
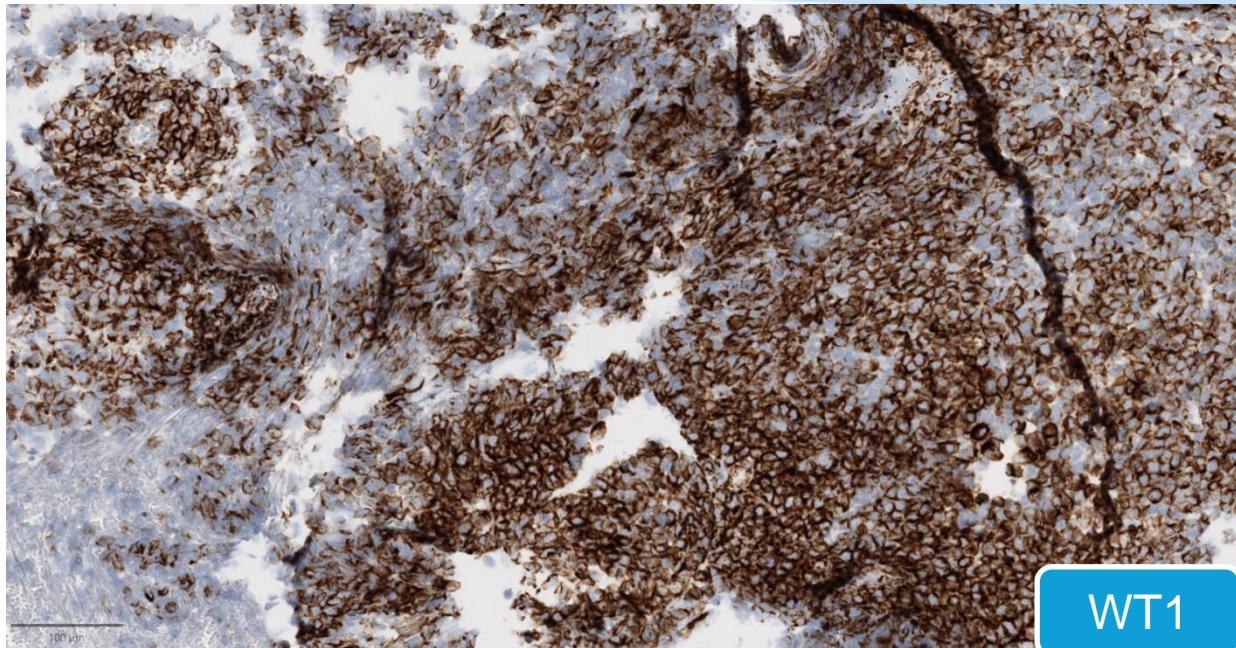
PMID: 29096655 PMCID: PMC5669030 DOI: 10.1186/s13000-017-0667-7



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CONSORCI  
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VALÈNCIA

## Estudios moleculares



Fusión génica que implica FOXO1

## RABDOMIOSARCOMA ALVEOLAR

Agradecimientos al H. la Fe de Valencia



**¡MUCHAS GRACIAS!**